A RATIONALE FOR THE BRAIN

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A guide to the diagnosis of diseases that may cause neurological symptoms.
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A chart that leads the user through the headache symptoms to possible diagnoses.

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The symptoms, signs, investigation and treatment of medical conditions that may cause neurological symptoms.

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INTRODUCTION

This book is designed for both the medical student and the doctor who is not a specialist in neurology.

It will take the user through a logical rationale in order to diagnose, and then treat, virtually every neurological condition likely to be encountered outside a specialist practice.

There are two ways to reach a diagnosis, using the chart in Section One, or the Diagnostic Algorithms in Section Two.

In Section One, the chart will guide the user through headache symptoms to a selection of possible diagnoses.

In Section Two the algorithms will indicate the diagnoses possible with a variety of neurological presenting symptoms.

Once a diagnosis has, or number of differential diagnoses have been made, a detailed explanation of the various diagnoses can be found in the largest part of the book, Section Three. This has been written in a style that should be easy to understand by even junior medical students, with technical terms explained in each monograph, but should still be useful to the non-specialist doctor. The symptoms, signs, investigations and treatment of a very wide range of appropriate conditions are explained, along with pictures of the more common conditions.

I trust that you will find it useful.

Warwick Carter
Brisbane

OTHER BOOKS IN THIS SERIES

A Rationale for Rashes
A Rationale for Eyes
A Rationale for the Abdomen
A Rationale for the Chest
Section ONE

HEADACHE
DIAGNOSTIC CHART
A RATIONALE FOR THE BRAIN

HEADACHE

HEAD INJURY

PENDS UNEQUAL

No

Yes

DROWSY OR CONFUSED

No

Yes

NAUSEA OR VOMITING

No

Yes

OVER 3 YOMITS

No

Yes

CONCUSSION

POSSIBLE INTRACRANIAL HAEMORRHAGE

ADULT

WORST EVER HEADACHE

No

Yes

CHILD

DROWSY OR CONFUSED

No

Yes

NECK STIFF

NEUROLOGICAL SYMPTOMS

No

Yes

SIGNS OF INFECTION

TENDER IMMEDIATELY ABOVE OR BELOW EYES

SIGNS OF INFECTION

BILATERAL HEADACHE

FEMALE

Yes

No

PREMENSTRUAL

Yes

No

HORMONAL HEADACHE

TENDER FOREHEAD, TEMPLES OR OCCIPUT

Yes

No

MUSCLE TENSION HEADACHE

ANALGESIC OVERUSE

Yes

No

CLUSTER HEADACHE

ANALGESIC NEUROPATHY

CHRONIC DAILY HEADACHE

SINUSITIS

INFECTION OR FEVER HEADACHE

SINISTER HEADACHE

e.g. meningitis, encephalitis, CVA, space occupying lesion, intracranial haemorrhage

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Section Two

DIAGNOSTIC ALGORITHMS FOR NEUROLOGICAL SYMPTOMS
# Diagnostic Algorithms for Neurological Symptoms and Signs

## Format

**Presenting Symptom**

*(Alternate Name)*

Explanation of terminology

System or other group of symptoms

Diagnoses that may present with this symptom [alternate name of diagnosis] (other symptoms of each diagnosis, or a discussion of the diagnosis)

*Other entries to consider*

**Clinical Sign**

*(Alternate Name) [Abbreviation]*

**Exp:** An explanation of the sign, with its methodology described in sufficient detail to enable the practitioner to perform the test.

**Int:** The interpretation of the sign.

(+): The diseases, syndromes etc. that should be considered if the test is positive

(++) The interpretation of an exaggerated or grossly positive test

(−): Ditto for a negative test result

(AB): Ditto for an abnormal test result

**Phys:** The pathophysiology of the sign to enable its significance to be better understood

*Other entries to consider*

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### Acromegaly

See Head, Large

### Agnosia

**Sensory suppression**

Parietal lobe lesions (astereognosis, personality changes)

Gerstmann syndrome (agraphia, unable to calculate)

### Agraphia

**Inability to write**

Cerebrovascular accident (hemiparesis, neurological signs)

Cerebral disease (e.g., Gerstmann syndrome) (R to L disorientation, agnosia, constructional disorders)

Psychiatric disturbances

Tremors or muscular weakness from any cause

Dyslexia

Visual problems

### Amnesia

**Total or partial loss of memory**

Epilepsy (convulsions, faint, paroxysmal)

Cerebrovascular accident (headaches, confusion, weak)

Cerebral haemorrhage

Transient global amnesia (temporary, compete recovery, familial)

Hysteria

Cerebral trauma

Menopause (flushes, sweating, menstrual changes)

Post-traumatic stress syndrome (nightmares, depression)

Hypothermia (shiver, confusion, arrhythmias)

Cerebral tumours (headache, nausea, neurological signs)

Viral encephalitis

Meningitis (cerebral irritation)

Hydrocephalus (papilloedema)

Wernicke's encephalopathy

Alzheimer disease (irrational)

Anoxia and hypoxia

Psychiatric disorders (e.g., fugue states)

Hypoglycaemia

Hyponatraemia

Alcohol and other drugs

*See also Memory Disturbance*
Ankle Clonus

Exp: Sharp dorsiflexion of foot causes repeated spasm of calf muscles and foot plantar flexion.

Int: (+) Upper motor neurone lesions and disease, functional nervous disorders, epilepsy, tuberous sclerosis, Wilson's disease, uraemia, encephalitis, multiple sclerosis, CVA, cirrhosis, Creutzfeldt-Jakob disease, encephalitis lethargica, alcohol withdrawal, drugs, benign idiopathic

Phys: With an upper motor neurone lesion there is no inhibition of the tendon stretch reflex
See also Ankle Jerk

Ankle Jerk

Exp: With patient kneeling on a chair, or supine, the Achilles tendon is struck firmly, causing spasm of the calf muscle

Int: (–) Peripheral neuropathies, polio, tabes dorsalis, posterior root tumour, spinal tumours, subacute combined degeneration of the cord, syringomyelia, multiple sclerosis, hypothyroidism, diabetes mellitus, beriberi, often absent in elderly
(+++) Upper motor neurone lesions, encephalitis, anxiety, tetanus, hyperthyroidism, cord transection

Phys: Reflex absence due to interruption of the reflex arc or muscular disease Exaggeration due to lack of suppression of reflex by cortical centres. Acts at L2, L3, L4
See also other signs listed under Reflexes

Anosmia

Loss of the sense of smell
Nasal infections (discharge, fever, headache)
Hypothyroidism (fatigue, dry skin, cold intolerance)
Frontal lobe tumour (amnesia, fits, aphasia)
Brain abscess (preceding infection, neurological changes)
Fracture of skull involving olfactory plate
Kallmann syndrome (hypogonadism)
Drugs (eg. phenol, chromium)

Aphasia and Dysarthria

Loss of the power of speech, or difficulty in speaking
Cerebrovascular accident (confusion, neurological changes)
Transient ischaemic attack (brief, confusion, drop attack)
Cerebral tumour (headaches, nausea, amnesia)
Multiple sclerosis (diverse neurological anomalies)
Sydenham's chorea (involuntary jerks, weak)
Myasthenia gravis (weak, diplopia, ptosis)
Hypothyroidism (fatigue, cold intolerance, dry skin)
Phaeochromocytoma (hypertension, sweating, headache)
Motor neurone disease (weak, myalgia)
Creutzfeldt-Jakob disease (Parkinsonian-like effects)
Landau-Kleffner syndrome (acquired aphasia, epilepsy)
Machado-Joseph syndrome (rigidity, limb weakness)
Moebius syndrome (ptosis, fixed facies)
Shy-Drager syndrome (tremor, vertigo)
Guillain-Barré syndrome (weak, dysphagia)
Alcohol and other drugs

Asterixis

See Tremor, Flapping

Ataxia

Exp: Lack of proper coordination. An unsteady, uncontrolled gait and/or a clumsy nose-finger test


Phys: Sensory (posterior column) and motor (cerebellar) forms. Former may be compensated for by ocular impressions (poorer coordination with eyes shut)
See also Heel-Knee Test; Nose-Finger Test; Romberg’s Sign

Athetosis

(Torsion Dystonia)
Involuntary, slow, writhing movements
Cerebral palsy
Hemiplegia from any cause
Cerebrovascular accident (aphasia, headache, confusion)
Wilson's disease (copper poisoning)
Lesch-Nyhan syndrome (gout, retarded, mutilation)
Juvenile Huntington's chorea
Dystonic juvenile movement disorders
Louis-Bar syndrome (telangiectasia of face and flexures)
Extrapyramidal lesions
Basal ganglia lesions
Encephalitis lethargica
Kernicterus (jaundice, infant)
Hallervorden-Spatz disease (iron deposits in brain)
Idiopathic and paroxysmal dystonias
Drugs (eg. phenothiazines, diazoxide)
See also Choreiform Movements

Aura
A premonition or sensation preceding a paroxysmal disorder
Epilepsy (twitching, loss of consciousness)
Anaphylaxis (oedema, itch, rash)
Migraine (headache, visual disturbances)
Transient ischaemic attack (various CNS symptoms)

Babinski's Sign
(Plantar Reflex)
Exp: Normally, slow plantar flexion of the great toe and fanning of the other toes occurs when the lateral side of the sole is stroked firmly with a pointed object, from the heel towards the toes. Positive with extension of the great toe
Int: (+) Corticospinal tract lesions, coma, post epileptic seizure, upper motor neurone lesions, multiple sclerosis, CVA, subacute combined degeneration of cord, paraplegia, normal infants
Phys: Acts at S1, S2 level

Balance, Disturbed
Vestibulitis (inner ear dysfunction)
Benign paroxysmal positional vertigo
Blocked eustachian tube (nasal congestion)
Hypotension (faints)
Parkinson's disease (abnormal gait, tremor)
Cerebrovascular accident (paralysis, dysarthria)
Cerebellar disease
Frontal lobe tumour (personality changes)

Dementia (inappropriate responses)
Hypothyroidism (constipated, weak)
Drugs (eg. sedatives, tranquillizers)
See also Vertigo

Ballism
See Chorea

Behavioural Problems
See Mental Retardation; Overactive; Swearing, Uncontrolled

Blackout
See Syncope

Bradykinesia
Abnormally slow movement
Parkinson's disease (tremor, rigidity, disturbed balance)
Cerebrovascular accident (mental changes, paralysis)
Cerebellar disease (disturbed balance)
Hypothyroidism (constipation, dry skin, bradycardia)
Arthritis (joint pain)
Frontal lobe tumour (personality changes, disturbed balance)
Depression (early waking, loss of interest)
Dementia (mental changes)
Drugs (eg. sedatives, tranquillizers)

Brudzinski's Sign
Exp: The head is flexed on the chest, causing the lower limbs to be drawn up
Int: (+) Meningeal irritation, meningitis, cerebral abscess, subdural empyema, subarachnoid haemorrhage, other cerebral haemorrhages, typhus, leptospirosis
Phys: Traction of the inflamed meninges on spinal nerves causes a protective flexor reflex
See also Neck Stiffness; Kernig's Sign

Cat Cry
Cri-du-chat syndrome (mental retardation, ocular hypertelorism)
Cheyne-Stokes Respiration

Exp: Respirations that gradually decrease in frequency until a temporary cessation occurs. Respiration then restarts and the frequency builds to a maximum before the cycle repeats itself.

Int: (+) CVA, meningitis, uraemia, narcotic or barbiturate overdose, advanced cardiac disease (eg. left ventricular failure), terminal stage of many chronic diseases, cerebral tumours, raised intracranial pressure.

Phys: Damage to cerebral respiratory centre. One full cycle equates to twice the circulation time.

See also Papilloedema

Chorea and Ballism

Continual, nonrepetitive, purposeless limb jerks

Rheumatic fever (arthralgia, rash, fever)
Huntington's chorea (hereditary, mental deterioration)
Sydenham's chorea (weakness, aphasia)
Hyperthyroidism (tachycardia, sweating)
Chorea gravidarum (pregnancy or oral contraceptives)
Systemic lupus erythematosus [SLE]
Senility
Polycythemia rubra vera
Encephalitis (headache, fever)
Cerebral tumours
Basal ganglia disease
Oral contraceptive sensitivity
Kernicterus (infant, jaundice)
Hemiballismus
Lesch-Nyhan syndrome (retarded, gout, mutilation)
Carbon monoxide poisoning
Drugs (eg. phenytoin, amphetamines, oral contraceptives, phenothiazines)

Clumsiness

Cranial
Alcohol intake
Epilepsy (absences, convulsions)
Migraine (pain, visual disturbances, photophobia)
Parkinson's disease (tremor, dysarthria, stiff gait)
Cerebrovascular accident (paralysis, speech defect)
Transient ischaemic attack (brief, weak, headache)
Alzheimer's disease (elderly, dementia)
Cerebral space occupying lesion (eg. tumour, haemorrhage)
Multiple sclerosis (bizarre neurological signs)
Birth trauma
Hydrocephalus
Chorea (see separate entry)

Other
Hypothyroidism (cold intolerance, dry skin, fatigue)
Cervical myelopathy
Spinal cord tumour or compression
Motor neurone disease (incoordination, paralysis)
Peripheral neuropathy
Muscular dystrophy
Myopathies
Gerstmann-Straussler-Scheinker syndrome

Cogwheel Rigidity

Exp: Resistance to passive movement diminishes in jerky steps

Int: (+) Extrapyramidal lesions, athetosis, paralysis agitans (parkinsonism), cerebral palsy

Phys: Static tremor masked by rigidity

See also Gait, Abnormal; Lead-Pipe Rigidity; Clasp-Knife Rigidity

Coma

Unrousable loss of consciousness

Cerebral
Head injuries and other severe trauma
Cerebrovascular accident (paralysis, neurological changes)
Cerebral haematoma
Epilepsy (convulsions, auras, headaches)
CNS infections (fever, convulsions)
CNS tumours (neurological changes)
Degenerative CNS disease
Increased intracranial pressure
Cerebral abscess
Hysteria (conversion reaction)

Endocrine
Diabetes mellitus (vomiting, dehydrated, tachycardia)
Hypothyroidism (dry skin, alopecia, anaemia)
Hypoglycaemia (flaccid, cold, sweating)
Addisonian crisis (vomiting, diarrhoea, pigmentation)

Other
Hypotension or shock (eg. myocardial infarct, haemorrhage)
Uraemia (oliguria, vomiting, fetid breath)
Hepatic coma (tremor, hyperreflexia, hyperventilation)
Anaphylaxis (shock, convulsions, dilated pupils)
Severe systemic infection (eg. cholera, septicaemia)
AIDS (rash, adenitis, fever)
Eclampsia (pregnant, hypertension, convulsions)
Porphyria (tachycardia, sweating, convulsions)
Hypothermia (pale, bradycardia)
Heat stroke (vomiting, convulsions)
Dehydration from any cause
Anoxia or hypoxia
Respiratory failure
Cardiac failure (dyspnoea, peripheral oedema)
Hypertensive encephalopathy
Hypothyroidism (tired, constipation, dry skin)
Malaria (fever, jaundice)
Cardiac arrhythmia or arrest
Typhoid fever (fever, diarrhoea)
Waterhouse-Friderichsen syndrome
Carbon monoxide poisoning (flushed)
Drug overdosage (eg. alcohol, barbiturates)
Poisons (ingested, snake bite, insect bite, spider bite, etc.)
See also Syncope

Confusional Reflex
Exp: Light shone into one eye causes the pupils of both eyes to contract
Int: (–) (only opposite eye contracts)
Phys: Afferent path of reflex arc is interrupted, but efferent path remains intact

Convulsions
(Fits, Seizures)
Cerebral
Epilepsy (blackouts, paroxysmal, amnesia)
Hysteria (erratic, precipitating cause)
Cerebrovascular accident (neurological signs, aphasia)
Cerebral tumour (headache, nausea, neurological signs)
Subdural haematoma (trauma, vomiting, pupil changes)
Migraine (headache, vomiting, visual disturbance)
Meningitis (fever, vomiting, neck stiffness)
Encephalitis (fever, nausea, sore throat)
Tuberculosis [TB]
Sporotrichosis (microphrophagia)
Raised intracranial pressure from other causes
Syncope

Syndromes
Behçet syndrome (uveitis, arthritis, mouth ulcers)
A RATIONALE FOR THE BRAIN

Catatonic syndrome (hypertonia)
Creutzfeldt-Jakob syndrome (dementia, paralysis)
Fragile X syndrome (subnormal intelligence, overactive, facial dysmorphism)
Gilles de la Tourette syndrome [Tourette syndrome] (foul language)
Landau-Kleffner syndrome (acquired aphasia)
Leigh syndrome (encephalopathy)
Lennox-Gestaut syndrome (mental retardation)
Lissencephaly syndrome (hypotonia, jaundice)
Organic brain syndrome (psychiatric changes)
Rett syndrome (female, autistic, hyperventilation)
Sandifer syndrome (reflux oesophagitis)
Stokes-Adams syndrome (syncpe, bradycardia)
Sturge-Weber syndrome (port wine stain, mental retardation)

Other
Severe bacterial, viral, rickettsial or protozoal infections (eg. toxoplasma, cytomegalovirus)
Fever from any cause (common in children)
Hypertension (essential or secondary)
Electrolyte disturbances
Shock (hypovolaemia)
Hypoglycaemia (?insulin overdose)
Anoxia (physical or pathological causes)
Severe trauma (coma, paralysis, pupil signs)
Dehydration (from dehydration or water deprivation)
Menke syndrome (low serum copper, kinky hair)
SLE (butterfly rash, arthritis)
Hypothyroidism and cretinism
Chromosomal abnormalities (often severe and prolonged)
Hypoparathyroidism (tetany, wheeze, stridor)
Eclampsia (pregnancy, hypertension)
Uraemia (fatigue, headache, pruritus)
Tetany (hyperventilation, hypocalcaemia, tetanic hand)
Behavioural problem
Breath holding (child)
Tetanus (rigid jaw, muscle spasms, trauma)
Porphyria (tachycardia, sweating, vomiting)
Niemann-Pick disease (mental retardation, hepatosplenomegaly)
Rabies (animal bite, hydrophobia, paraesthesia)
Drugs (eg. strychnine, atropine, cyanide, nicotine)

Corneal Reflex
Exp: Lightly touching one cornea with a piece of cotton wool produces blinking in both eyes
Int: (– bilateral) Coma, general anaesthesia, death
(– unilateral) Lesion of 7th or ophthalmic division of 5th nerve
Phys: Tests for anaesthesia of cornea, or interruption of reflex arc

Déjà Vu
Feeling of intense familiarity
Epilepsy (convulsions, amnesia, absences)
Emotional trauma or stress
Psychiatric disorders
Cerebrovascular accident (neurological changes)

Dementia
Alzheimer's disease
Cerebrovascular disease (eg. stroke)
Alcoholism
Senile dementia
Pick's disease (lobar atrophy, lack of insight)
Cerebral trauma (eg. subdural haematoma)
Cerebral abscess or tumour
Meningitis (nuchal rigidity)
Encephalitis (fever, malaise)
Parkinson's disease (advanced stages only)
Hypothyroidism (constipation, fatigue, dry skin)
Hepatic failure
Neurosyphilis (Argyll-Robertson pupils)
Pernicious anaemia (lethargy, sore tongue)
Pellagra (anaemia, rash, diarrhoea)
Metastatic carcinoma
Uraemia (fatigue, pruritus, thirst)
Hypocalcaemia
Hypopituitarism
Hypercalcaemia
Hydrocephalus
Huntington's disease (tremor, ataxia)
Subacute sclerosing panencephalitis
Anoxia
Creutzfeldt-Jakob disease
Lysosomal storage diseases
Hyperuricaemia
Vitamin B deficiency
Poisons (eg. glue sniffing, organophosphates, lead)
Drugs (eg. bromides, alcohol, barbiturates, amphetamines)

Syndromes
AIDS (abnormal infections)
Cushing syndrome (obese, striae, moon face)
Punch drunk syndrome

Corneal Anaesthesia
Loss of corneal sensation to light touch
Riley-Day syndrome
Dole-Richardson-Olszewski syndrome
( parkinsonian effects)
Wernicke-Korsakoff syndrome (alcohol dependent)
*See also Confusion*

**Diplopia**

**Double vision**
Paralytic squint due to 3rd, 4th or 6th cranial nerve palsy (limited movement of one eye)
Cerebrovascular accident (neurological changes)
Concussion (trauma, headache, nausea)
Orbital trauma
Migraine (nausea, headache, photophobia)
Botulism (dry mouth, dysphagia, paralysis)
Cerebral tumours (headache, neurological signs)
Myasthenia gravis (weakness, ptosis)
Thyroid diseases
Multiple sclerosis (weakness, abnormal sensation)
Gradenigo syndrome (headache, facial pain)
Wernicke-Korsakoff syndrome (ataxia, demented)

**Dizziness**
*See Vertigo*

**Doll's Head Manoeuvre**

*Exp*: In a comatose patient, rolling the head to one side causes counter-rolling of the eyes to the other side
*Int*: (+) Brain stem intact
(–) Brain stem damage
*Phys*: Used in assessment of comatose patient. Should not be used if there is any possibility of neck injury

**Drop Attack**

**Sudden brief loss of consciousness without warning**

**Cardiovascular**
Atrioventricular conduction block
Ventricular tachycardia
Atrial fibrillation onset
Carotid sinus syncope
Aortic or mitral stenosis (bruit)
Pulmonary embolism (chest pain, dyspnoea)
Severe pulmonary hypertension
Atrial myxoma

**Enophthalmos (Enophthalmia)**
Recession of eyeballs within sockets

**Other**
Severe chronic obstructive airways disease (cough)
Transient ischaemic attack
Epileptic seizure (incontinence)
Hypoglycaemia
Psychological attention seeking
Drugs (eg. glyceryl trinitrate, antihypertensives)
*See also Syncope (Faint)*
A RATIONALE FOR THE BRAIN

Dehydration
Cachexia
Malnutrition
Advanced carcinoma
Other wasting diseases
Lacrimal gland tumour
Horner syndrome (myosis, exophthalmos, anhydrosis)

Epilepsy
See Convulsions

Exophthalmos
(Proptosis)
Protrusion of eyeballs within sockets.
Hyperthyroidism (tachycardia, sweating)
Cerebral tumour
Optic or orbital tumour
Cushing disease (hirsute, obese, ecchymoses)
Cavernous sinus thrombosis
Hand-Schueller-Christian disease (diabetes insipidus, lung damage)
Pituitary tumours
Osteomas
Neurofibromatosis
Wegener’s granulomatosis (fever, weakness, sinusitis)
Metastatic carcinoma
Xanthomas
Malignant hypertension
Uraemia (fatigue, pruritus, thirst)
Cellulitis (erythema, fever, warm skin)
Vascular malformation
Lacrimal tumours
Mucocoele (fluid filled cyst)
Rhabdomyosarcoma (muscle tumour)
Apert syndrome (premature skull bone fusion)
Crouzon syndrome (abnormal facies, squint)
Sturge-Weber syndrome (port wine stain, convulsions, intellectual disability)

Fasciculation, Muscular
(Twitching)
Exp: Fibrillary twitching of voluntary muscles visible through the skin. Exacerbated by tapping muscle bundles
Int: (+) Depolarising drugs, muscular dystrophies, amyotrophic lateral sclerosis, lower motor neurone lesions, motor neurone disease, poliomyelitis, Guillain-Barré syndrome, syringomyelia, hypocalcaemia, severe viral diseases, thyrotoxicosis, polymyositis
Phys: May occur without neurological cause. Due to uncoordinated depolarisation of muscle fibres
See also Tremor, Intention; Tremor, Postural; Tremor, Resting; Myotonia

Facial Weakness
Bell's palsy (unilateral, temporary)
Cerebrovascular disease (other neurological defects common)
Cerebral neoplasm
Multiple sclerosis (slow onset)
Motor neurone disease (bilateral)
Parkinson's disease (tremor, gait abnormal)
Parotid tumours (unilateral)
Brain stem encephalitis
Sarcoidosis

Poliomyelitis
Herpeszoster infection (vesicular rash, pain)
Mumps (facial sialedenititis)
Other serious viral infections
Tetanus (pain, paralysis)
Lyme disease (fever)
Brucellosis (fever, arthralgia, fatigue)
Other serious bacterial infections
Myasthenia gravis (ptosis, diplopia, dysarthria)
Guillain-Barré syndrome (progressive, dysphagia, polynueurtis)
Muscular dystrophies
Trauma
Emotional and psychogenic

Faint
See Syncope; Drop Attack

Familiarity
See Déjà Vu

Festination
See Gait, Abnormal; Cogwheel Rigidity

Fits
See Convulsions

Flaccid Paralysis
See Paralysis, Flaccid
Flapping Tremor
(Asterixis)
See Tremor, Flapping

Flashes, Visual
Migraine (headache, photophobia, nausea)
Vitreous haemorrhage
Posterior vitreous detachment

Floppy Baby
Generalised muscle hypotonia in infancy
Viral or bacterial infection
Cerebral palsy (ataxia)
Starvation/Malnutrition
Werdnig-Hoffman disease (familial, progressive, muscular dystrophy)
Myasthenic syndromes
Glycogen storage disease
Duchenne muscular dystrophy
Cerebral palsy (ataxia)
Starvation/Malnutrition
Werdnig-Hoffman disease (familial, progressive, muscular dystrophy)
Myasthenic syndromes
Glycogen storage disease
Duchenne muscular dystrophy
Congenital muscular dystrophy
Intellectual deficits
Aminoaciduria
Osteogenesis imperfecta (fracture bones easily)
Rickets
Other neuromuscular diseases

Gait, Abnormal
Exp: Ataxic gait – Unsteady, unbalanced, lack of confidence
Extrapyramidal gait – Slow, rigid gait, no arm swinging
Festination – Quick, shuffling, trunk bent
“Marche-a-petit-pas” – ‘Gait of little steps', jerky, unbalanced, muscle spasm, feet ‘stick' to floor on turning or starting, asymmetrical brisk reflexes
Scissors gait – Legs cross left to right and vice versa when walking
Spastic ataxic gait – Muscle spasm, jerky, unsteady, unbalanced
Waddling gait – Exaggerated elevation of the hip on the stepping side and abnormal yielding of the hip on the grounded side, giving excessive lateral movement to the trunk

Int: Ataxic gait – (+) Drug induced cerebellar ataxia, alcoholic cerebellar degeneration, hypothyroidism, transient ischaemic attack, Gerstmann-Straussler-Scheinker syndrome, other cerebellar disease
Extrapyramidal gait – (+) Parkinson's disease, drug induced parkinsonism (eg. prochlorperazine, metoclopramide)
Festination – (+) Parkinson's disease, extrapyramidal lesions
“Marche-a-petit-pas” – (+) Multiple lacunar strokes
Scissors gait – (+) Cerebral diplegia, diseases of hip joint, spastic paraplegia
Spastic ataxic gait – (+) Multiple strokes, transient ischaemic attacks, cerebral palsy, vitamin B12 deficiency, cervical myelopathy, spinocerebellar degeneration
Waddling gait – (+) Progressive muscular dystrophy, congenital dislocation of hips, Huntington's chorea, pseudohypertrophic muscular paralysis, inclusion body myositis

Formication
Sensation of insects crawling on skin
Neurological sensory disorders
Psychiatric syndromes
Alcoholism withdrawal
Drugs

Foster Kennedy Sign
Exp: Unilateral papilloedema with contralateral optic atrophy
Int: (+) Cerebral tumour adjacent to optic nerve of atrophied eye
Phys: Pressure on one optic nerve causes optic atrophy while increasing intracranial pressure to cause papilloedema of the other eye

Gag Reflex
See Pharyngeal Reflex

**Scissors gait** – Acute adduction of both hips from hip joint disease or adductor muscle contracture.

**Spastic ataxic gait** – May be associated with incontinence and cognitive disturbances in elderly.

**Waddling gait** – Weakness of gluteal muscles.

See also Cogwheel Rigidity; Parkinsonism

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### Giddiness

See Vertigo

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### Grasp Reflex

**Exp:** Automatic grasping of objects placed against palm of hand

**Int:** (+) Normal neonate, upper prefrontal lobe lesions and tumours

**Phys:** Prefrontal lobes concerned with intellect and the origination of motor movements. Patient is unable to prevent a primitive reflex with lesions to this area.

See also Nystagmus

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### Hallpike Test

(Dix-Hallpike Test)

**Exp:** The patient is carefully seated on an examination couch with legs on the couch. There should be no pillow and no wall or other obstruction for 50 cm beyond the head of the couch. The patient's head is turned at 45° to one side. The patient is then quickly lowered, with the support of the examiner, so that the head is hanging over the edge of the table and extended to 30° still in a 45° orientation. The test is then repeated with the head turned 45° in the opposite direction, and with the head in the direct anteroposterior position. Immediately after each test, any vertigo is recorded and the patient's eyes are checked for nystagmus, its character and duration being noted.

**Int:** (+ marked vertigo, delayed nystagmus with rapid movement away from lower ear that fatigues easily) - Benign positional vertigo, common in elderly

(+ mild vertigo, immediate nystagmus with variable direction, and prolonged) - Central positional nystagmus, cerebellopontine or brain stem lesions

See also Nystagmus

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### Hallucinations

Schizophrenia (withdrawn, delusions, loss of ego)

Affective disorders

Other organic mental states

Metabolic encephalopathy

Encephalitis (fever, malaise, neck stiffness)

Epilepsy (auras, absences, convulsions)

Delerium tremens (alcohol abuse)

Alcohol dependence withdrawal

Posthypnotic

Charles Bonnet syndrome (elderly, affective disorder)

Iatrogenic (eg. psychoactive drugs)

Drugs (eg. LSD, heroin, marijuana, amphetamines)

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### Headache

General

Bacterial, viral, rickettsial and protozoal infections of any organ but particularly CNS

Fever of any cause

Ocular disorders (eg. refraction error, glaucoma)

Dental causes (eg. root abscess)

Muscular spasm (stress, occipital or temporal)

Trauma

Fatigue (acute or chronic)

Drugs (eg. methyldopa)
Infections
- Sinusitis (frontal, fever, rhinorrhoea, face ache)
- Mastoiditis (tender mastoid, earache)
- Meningitis (fever, neck stiffness, confusion)
- Otitis media (fever, ear pain)
- Encephalitis (fever, malaise, neck stiffness)
- Brucellosis (fever, arthralgia, fatigue)
- Cerebral abscess (preceding infection, neurological signs)
- Syphilis (varied symptoms)
- Dengue fever (retro-orbital pain)

Central Nervous System
- Cerebral tumour (neurological signs)
- Migraine (nausea, photophobia, vertigo)
- Cerebrovascular accident (neurological signs)
- Cerebral haemorrhage (subdural or subarachnoid)
- Trigeminal neuralgia (face, frontal, unilateral)
- Cluster headaches (sweating, lacrimation, unilateral)
- Neuralgia (sharp localised pain)
- Prolactinoma (visual defect, cranial nerve palsy)
- Psychiatric disorders (eg. phobia, anxiety)

Endocrine
- Adrenal insufficiency (fatigue, nausea, diarrhoea)
- Acromegaly (gigantism, vision loss, amenorrhoea)
- Diabetes insipidus (polyuria, polydipsia)
- Hypothyroidism (dry skin, fatigue, cold intolerance)
- Phaeochromocytoma (hypertension, sweating, abdominal pain)
- Menopause (flushes, lightheaded, amenorrhoea)

Chinese restaurant syndrome (nausea, facial pressure)
- Chronic fatigue syndrome (fever, poor exercise tolerance)
- Cushing syndrome (hirsute, obese, ecchymoses, plethora)
- Gradenigo syndrome (diplopia, facial pain)
- Post-traumatic cerebral syndrome (vertigo, mental changes)
- Premenstrual tension syndrome (mastalgia, nausea)
- Scapulocostal syndrome (neck and arm pain)
- Sick building syndrome (fatigue, malaise)
- Sticky platelet syndrome (strokes, migraines)
- SUNCT syndrome (very brief, unilateral)
- Toxic shock syndrome (vomiting, diarrhoea, fever)
- Vogt-Koyanagi-Harada syndrome (blind, fever)

Other
- Hypertension (fatigue, tinnitus)
- Anaemia (fatigue, palpitations)
- Polycythaemia vera (malaise, pruritus)
- Uraemia (fatigue, pruritus, thirst)
- Glomerulonephritis (oliguria, malaise, oedema)
- Temporomandibular joint dysfunction
- Vascular anomalies
- Cervical osteoarthritis
- Temporal arteritis (unilateral, visual disturbances)
- Glaucoma (visual disturbance)
- Pre-eclampsia (pregnancy, hypertension)
- SLE (rash, arthralgia)
- Carotidynia (neck pain, unilateral, tender carotid artery)
- Paget's disease (bone pain)
- Anaphylaxis

See also Headache Diagnosis Chart in section One

Head, Large
- Acromegaly (large jaw, large hands)
- Hydrocephalus
- Dandy-Walker syndrome (vomiting, nystagmus, cleft palate)
- Proteus syndrome (gross disfigurement)
- Stewart-Morgagni-Morel syndrome (hyperostosis frontalis)

Heel-Knee Test
Exp: While supine, the patient is told to touch one knee with the opposite heel, and then to run the heel down the shin
Int:(+) Ataxia (eg. tabes dorsalis, posterior column lesions, cerebellar lesions, Friedreich's ataxia), poliomyelitis
Phys: Loss of integrity of sensory pathway along posterior columns to cerebellum

*See also Ataxia; Romberg's Sign; Nose-Finger Test*

**Hemianopia**

Loss of half visual field in eye
- Cerebral abscess (preceding infection, headache)
- Migraine (transient, headache)
- Cerebral tumour (neurological signs)
- Temporal arteritis (pain, unilateral)
- Cerebrovascular accident (neurological signs)
- Acromegaly (headache, gigantism, amenorrhoea)
- Pituitary tumour (headache)

**Hemiplegia**

See Paralysis and Muscular Weakness; Paraplegia and Quadriplegia

**Hydrocephalus**

Exp: Accumulation of excess CSF under pressure to cause skull enlargement in children and cerebral damage in adults

Int: (+) Congenital malformations, ventricular colloid cyst, pineal tumours, intraventricular haemorrhage, aqueduct stenosis, pituitary tumours, cranioopharyngioma, ependymoma, choroid plexus disease or tumour, astrocytoma of cerebellum, medulloblastoma, Arnold-Chiari malformation

Phys: Any obstruction of the CSF circulation will lead to hydrocephalus

**Hypertonicity**

Involuntary resistance to passive movement
- Upper motor neurone lesions
- Extrapyramidal system lesions
- Cerebrovascular accident [CVA]
- Parkinsonism (tremor, rigidity)
- Cord transection
- Postasphyxia
- Kernicterus (jaundice, infant)
- Premature infant
- Encephalitis (headache, fever)
- Cerebral oedema
- Trauma
- Meningitis
- Hydrocephalus
- Cerebral space occupying lesion
- Cerebral palsy

*See also Myotonia; R rigidity, Muscular

**Hypotonia**

Reduced resistance to passive movement
- Lower motor neurone lesion
- Cerebellar lesions
- Grossly debilitating diseases
- Poliomyelitis
- Tabes dorsalis
- Spinal shock
- Myopathies
- Sydenham's chorea
- Premature infant

*See also Paralysis and Muscular Weakness*

**Incoordination**

See Clumsiness; Dysdiadochokinesia

**Insomnia**

Inability to sleep
- Anxiety and pain
- Behaviour (eg. day time naps)
- Evening exercise
- Elderly (depression, irritable)
- Psychiatric disorders (eg. schizophrenia, affective disorders)
- Menopause (flush, sweating, mood changes)
- Depression (anxiety, phobia)
- Postpartum depression
- Chorea (involuntary jerks, aphasia, weakness)
- Sleep apnoea (snoring)
- Alcohol withdrawal
- Drugs (eg. anorectics, caffeine, pseudoephedrine)

** Syndromes**

- Cushing syndrome (obese, striae, weakness)
- Post-traumatic stress syndrome (after catastrophe, psychiatric problems)
- Premenstrual syndrome (headache, irritable)
- Restless legs syndrome (hyperkinesia)

**Intellectual Disability**

Genetic or Chromosomal
- Cretinism
- Lysosomal storage disease
- Niemann-Pick disease
- Duchenne muscular dystrophy
- Gaucher's disease
- Galactosaemia
- Phenylketonuria
- Tuberous sclerosis
- Microcephaly
A RATIONALE FOR THE BRAIN

Environmental
Lead and other toxins
Drugs
Toxoplasmosis
Rubella
Cytomegalovirus
Other viral and bacterial diseases
Congenital syphilis
Sclerosing panencephalitis
Wilson's disease

Psychological
Psychoses
Autism
Stimulus deprivation etc.

Other
Cerebral space occupying lesion (eg. tumours, abscess, haematoma)
Diabetes mellitus (polydipsia, polyuria)
Chronic epilepsy
Birth trauma
Brain trauma
Cerebral palsy
Multiple sclerosis (slow onset)
Hydrocephalus
Deafness may mimic subnormality

 Syndromes
Angelman syndrome (mute, laughter, ataxic)
Blue diaper syndrome (neurocalcinosis, hypercalcaemia)
Cockayne syndrome (dwarf, optic atrophy, deaf)
Coffin-Lowry syndrome (prominent lips, coarse facies)
Coffin-Siris syndrome (hypoplastic toe nails)
Conrad syndrome (deaf, cataracts)
Cri-du-chat syndrome (cat cry, ocular hypertelorism)
De Lange syndrome (microcephaly, “Amsterdam dwarf”)
Down syndrome (typical facies, reduced growth)
Dubowitz syndrome (micrognathia, ptosis)
Fetal alcohol syndrome (hirsute, reduced growth)
Fragile X syndrome (hyperactive, autistic, epilepsy, large)
Hunter syndrome (gross facies, arthralgia, hepatomegaly)
Hurler syndrome (dwarf, gross facies, arthralgia)
Johanson-Blizzard syndrome (deaf, anal atresia)
Klinefelter syndrome (XXY, male, hypoplastic genitals)
Laurence-Moon-Biedl syndrome (retinitis pigmentosa)
Lennox-Gastaut syndrome (convulsions)
Lesch-Nyhan syndrome (gout, athetosis, mutilation)
Louis-Bar syndrome (telangiectasia of face and eye)
Loweyer syndrome (cataracts, epicanthal folds)
Marinesco-Sjögren syndrome (ataxia, cataracts)
Miller-Dieker syndrome (abnormal facies)
Neu-Lexova syndrome (face anomalies, oedema)
Patau syndrome (cleft lip, polydactyly)
Pompe syndrome (hypotonia, cardiac anomalies)
Prader-Willi syndrome (hypotonia, male, obese)
Rett syndrome (female, hyperventilation, seizures)
Richner-Hanhart syndrome (uveitis, keratoderma)
Sanfilippo syndrome (gross facies, hepatosplenomegaly)
Savant syndrome (specific talent)
Seckel syndrome (hip and elbow dislocation)
Smith-Lemli-Opitz syndrome (ptosis, hypoplasias)
Sturge-Weber syndrome (port wine stain, convulsions)
WAGR syndrome (Wilms’ tumour, iris abnormality)
Wolf-Herschorn syndrome (cleft lip)

Intention Tremor
See Tremor, Intention

Jaw Jerk
Exp: With the jaw relaxed and the mouth hanging open, a finger is placed across the front of the jaw and struck with a rubber hammer.
Contraction of masseters may result
Int: (+) Lesion of both corticospinal tracts
Phys: Acts at the level of the pons

Jerks
See Tremor; Chorea

Kernig's Sign
Exp: One hip of supine patient is fully flexed. Any subsequent attempt to straighten the knee results in painful spasm of the hamstrings when positive
Int: (+) Meningeal irritation, bacterial and aseptic meningitis, cerebral or spinal cord abscess, subdural empyema, subarachnoid haemorrhage, encephalitis, typhus, leptospirosis, other cerebral haemorrhages
Phys: Activation of protective flexor reflexes which shorten and immobilise the spine
See also Neck Stiffness; Brudzinski's Sign
**Lead-Pipe Rigidity**

Exp: Heavy passive stiffness of limb throughout range
Int: (+) Extrapyramidal lesions, parkinsonism, basal ganglia disease
Phys: Chronic overactive lengthening reaction
*See also Cogwheel Rigidity; Clasp-Knife Rigidity*

**Lightheadedness**

Vertigo (see separate entry)
Postural hypotension
Lack of sleep
Stress and anxiety
Malnutrition/starvation
Excess alcohol intake
Migraine (headache, visual disturbances, nausea)
Hyperventilation (tetany, vertigo, paraesthesiae)
Cervical spondylosis
Transient ischaemic attacks (various CNS symptoms)
Visual deterioration (eg. cataracts)
Viral or bacterial infection
Fever of any cause
Impaired vestibular function
Deafness
Peripheral neuropathy
Psychiatric conditions
Drugs (eg. antihistamines, tranquillizers, antihypertensives)

**Memory Disturbance**

Cerebral trauma or tumour
Alzheimer's disease
Acute brain syndrome (elderly, psychotic, disorientation)
Amnestic syndrome (alcohol dependence, thiamine deficiency)
Organic brain syndrome (multiple psychiatric changes)
Transient global amnesia (temporary, compete recovery, familial)
*See also Amnesia*

**Memory Loss**

See Amnesia; Memory Disturbance

**Meningism**

See Neck Stiffness; Kernig's Sign; Brudzinski's Sign; Opisthotonos

**Mental Retardation**

See Intellectual Disability

**Microcephaly**

Small head
Angelman syndrome (mental retardation, ataxic, laughter)
De Lange syndrome (mental retardation, low hair line)
Langer-Giedion syndrome (sparse hair, exostoses)
Craniostenosis (premature fusion of sutures)

**Miosis**

Abnormally contracted pupils
Bright light
Convergence
Syphilis (tabes dorsalis)
Sympathetic nerve paralysis
Pontine lesions
Congenital
Hysteria
Horner syndrome (myosis, exophthalmos, anhydrosis)
Holmes-Adie syndrome
Insecticide poisoning
Drugs (eg. pilocarpine, physostigmine, narcotics, beta-blockers)

**Moro Reflex**

(Startle Reflex)
Exp: While supporting an infant prone, allow the head to fall back sharply for a short distance. Results in extension of trunk and extension and abduction of limbs, followed by flexion and adduction of limbs
Int: (+) Normal infant up to 4 months
A RATIONALE FOR THE BRAIN

(→) Bilateral - Over 4 months age, severe brain damage, premature infant
(←) Unilateral - Limb fracture, peripheral neuropathy, Erb's palsy

Movement, Abnormal
See Athetosis; Bradykinesia; Chorea; Dyskinesia, Tardive; Gait, Abnormal; Tremor, Flapping; Tremor, Intention; Tremor, Postural; Tremor, Resting

Muscle Spasm
See Chorea; Cramps, Muscular; Hypertonicity; Myoclonus; Myotonia; Tremor

Muscular Weakness
See Paralysis and Muscular Weakness; Fatigue, Abnormal

Myoclonus
Brief sudden muscle jerks
Hypnic jerks (occur while falling asleep)
Startle reflex (sudden loud noise)
Drug or alcohol intoxication
Focal epilepsy
Spinocerebellar degenerations
Lesions of cortex or brainstem
Spinal lesions (segmental, localised)
Post-anoxic
Demyelinating diseases
Metabolic encephalopathy
Baltic myoclonus
Hereditary
Lafora body disease (child to teens, dementia)
See also Ankle clonus

Myosis
See Miosis

Myotonia
Exp: Delayed muscular relaxation following forceful muscular contraction (eg. repeated muscular fasciculation after tap with rubber hammer)
Int: (+) Myotonic dystrophy, myotonia congenita, hyperkalaemia, Talma's disease
Phys: Usually reduced by repeated activity. Often worse in cold
See also Hypertonicity; Fasciculation, Muscular

Neck Stiffness
Undue tautness of neck muscles on passive flexion
Meningeal irritation (eg. aseptic or bacterial meningitis, encephalitis, cerebral abscess, subdural empyma, typhus, leptospirosis, subarachnoid or other cerebral haemorrhage, migraine)
Cervical arthritis
Torticollis and other neck muscular spasms and strains
Adenitis
See also Brudzinski's Sign; Kernig's Sign; Opisthotonos

Nose-Finger Test
Exp: Using the index finger, patient touches own nose then examiner's finger, back and forth as rapidly as possible. Examiner changes position of finger between touches. Positive if clumsy
Int: See Ataxia
Phys: See Ataxia
See also Heel-Knee Test

Nuchal Rigidity
See Neck Stiffness

Numbness
See Anaesthesia; Paraesthesia; Formication

Nystagmus
Exp: Involuntary rhythmic movement of eyeball. Two types: Pendular (oscillating) – with regular movements
Jerk (rhythmic) – with movement faster in one direction than the other
Int: (+) Normal with acute lateral vision and watching a moving object, barbiturates, labyrinthine and vestibular disease, brain stem lesions (often vertical nystagmus), demyelinating diseases (eg. multiple sclerosis), during epileptic fit (eg. petit mal), brain tumours, syringobulbia, Dandy-Walker syndrome, Parinaud syndrome, diencephalic syndrome, pinealoma, central vision loss (eg. albinism, retinal disease), other visual disturbances, cerebral abscess,
coma, Friedreich’s ataxia, congenital, alcohol, some normal infants
Phys: Jerk form more common and is neurological in aetiology. Pendular is due to a visual defect. Direction of nystagmus can give further clue to localise lesion
See also Hall Pike Test

Oculogyric Crisis
Exp: Varies from mild cases with abnormal uncontrolled random eye movements, to severe cases with fixed elevated gaze associated with painful extension of the neck which may be so severe that the occiput nearly touches the thoracic vertebrae and the airway may be compromised
Int: (+) Rare side effect of prochlorperazine, encephalitis or Parkinson’s disease
Phys: Effect rapidly reversed by IV benztpine

Opisthotonos
Acute rigid arching of the body due to spasm of the back muscles
Tetanus
Spinal meningitis
Infantile meningitis
Cerebellar lesions
Other causes of meningeal irritation
See also Neck Stiffness; Kernig’s Sign

Optic Disc, Abnormal
Exp: Characteristics noted during ophthalmoscopic examination of retina
Int: (+) Extra vascularity – Optic neuritis (+)Papilloedema (bulging of disc) – Increased intracranial pressure (+) Atrophy (reduced vascularity) – Optic nerve disease (eg. optic atrophy) (+) Pale disc – Central retinal artery occlusion (+) Haemorrhages – Central retinal vein occlusion

Paraesthesia
(Pins and Needles)
Psychiatric and emotional causes
Nerve compression (eg. bruising, inflammation, joint overuse)
Transient ischaemic attack (temporary, clumsiness, confusion)
Atypical migraine (visual disturbances, intermittent, nausea)
Pernicious anaemia (pallor, anorexia, dyspepsia)
Diabetes mellitus (polyuria, blurred vision, fatigue)
Ischaemic limb (pallor, pain, pulseless)
Hyperventilation (tetany)
Chronic renal failure (nausea, pruritus)
Menopause (usually palms and soles)
Multiple sclerosis (muscle weakness)
Posterolateral sclerosis (weakness, sensory loss)
Beriberi (leg cramps, oedema, anorexia)
Tetanus (muscle spasm, dysarthria, wound)
Rabies (hydrophobia, animal bite)
Leprosy (macular rash, anaesthesia)
Acromegaly (psychic changes, coarse facies)
Ciguatera poisoning (tropical fish ingestion)
Other poisons

Syndromes
Carpal tunnel syndrome (arm pain, burning)
Cervical rib syndrome (arm pain and weakness)
Conn syndrome (weak, hypertension)
A RATIONALE FOR THE BRAIN

Painful bruising syndrome (female, spontaneous bruises)
Raynaud's phenomenon (finger pain, pallor and cyanosis)
Refsum syndrome (distal polyneuropathy)
Restless legs syndrome (leg movement)
Strachan syndrome (amblyopia, orogenital dermatitis)

Paralysis and Muscular Weakness
Loss of motor power
Cerebral
Cerebrovascular accident (confusion, aphasia, anaesthesia)
Head trauma (headache, ocular signs)
Bell's palsy (face involved, spontaneous onset and recovery)
Myasthenia gravis (ptosis, diplopia, dysarthria)
Transient ischaemic attack (clumsiness, temporary, confusion)
Transverse myelitis
Cerebral or spinal abscess

Nervous System
Zoster paresis (follows shingles)
Multiple sclerosis (diffuse neurological symptoms)
Polyneuritis (pain, anaesthesia, limbs)
Motor neurone disease (progressive, several forms)
Familial periodic paralysis (intermittent)
Poliomyelitis (stiff neck, sore throat, flaccid)
Subacute combined degeneration of the cord (vitamin B12 deficiency)
Charcot-Marie-Tooth disease (familial)
Peroneal muscular dystrophy (familial, onset in teens)

Musculoskeletal
Vertebral disc herniation (back pain, hyporeflexia)
Muscular dystrophy (child, proximal weakness)
Rheumatoid arthritis (joint pain, nodules)
Ischaemic limb (pain, pallor)
 Dermatomyositis (proximal weakness, rash)
Polymyositis (muscle weakness)
Osteomalacia and rickets (bone bowing, fatigue)
 Duchenne muscular dystrophy
Inclusion body myositis (elderly, white, male, peripheral)

Syndromes
Bartter syndrome (polyuria, weakness, polydipsia, short)
Behcet syndrome (uveitis, arthritis, ulcer)
Bell's palsy (facial, unilateral)
Brown-Sequard syndrome (hemisection of cord)

Carpal tunnel syndrome (hand pain and weakness)
Cervical rib syndrome (arm involved, pain)
Conn syndrome (weak, hypertension)
Cori syndrome (glycogen storage disease)
Creutzfeldt-Jakob syndrome (jerks, seizures)
Cushing syndrome (obese, ecchymoses, hirsute)
Eaton-Lambert syndrome (myasthenic symptoms)
Erb-Duchenne palsy (shoulder girdle paralysis)
Floppy baby syndrome (partial widespread muscular paralysis)
Guillain-Barré syndrome (progressive, dysphagia, polyneuritis)
Klumpke's palsy (neonate, limp wrist)
Kugelberg-Welander syndrome (shoulder girdle weakness)
Locked-in syndrome (total paralysis of body)
McArdle syndrome (cramps, myopathy)
Parkinsonism (tremor, rigidity, reduced strength)
Parsonage-Turner syndrome (brachial plexus disturbance)
Pompe syndrome (hypotonia, mental retardation)
Post-polio syndrome (fatigue, arthralgia, myalgia)
Potassium wastage syndrome (polyuria, weak, dilute urine)
Prader-Willi syndrome (hypotonia, obese, mental retardation)
Refsum syndrome (distal sensorimotor polyneuropathy)
Roussy-Levy syndrome (hypotonia, ataxia, kyphoscoliosis)
Shy-Drager syndrome (hypotension, vertigo, tremor)
Sicard syndrome (paralysis of cranial nerves 9, 10, 11, 12)
Sturge-Weber syndrome (hemiplegia, port wine stain)
Uveoparotid syndrome (facial paralysis, uveitis)
Werdnig-Hoffman syndrome (progressive dystrophy, neonate)

Other
Psychological stress
Lymphoma
HIV
Hyperparathyroidism (polyuria, polydipsia, bone pain)
Tick bite (child more common)
Hyperaldosteronism (tetany, headache, polyuria)
Hyperthyroidism (sweaty, proximal myopathy)
Porphyria (nausea, abdominal colic, sweating)
Botulism (dry mouth, diplopia, dysphagia)
Syphilis (rash, sexually transmitted, various symptoms)
See also Facial Weakness; Floppy Baby; Paraplegia and Quadriplegia


**Paralysis, Flaccid**  
Relaxed muscles that cannot be moved voluntarily  
Poliomyelitis  
Infantile muscular atrophy  
Cerebral atonic diplegia  
Amyotonia congenita  
Glycogen storage diseases (weak rather than paralysed muscles)  
Plexus palsies  
Multiple sclerosis (slow onset)  
Hyperkalaemia  
Conn syndrome (weak, hypertension)  
Spinal cord shock  
Botulism  
Poisons (eg. organophosphates)  
Neuropathies  
Motor neurone disease  
Encephalitis  
Other lower motor neurone lesions  
Guillain-Barré syndrome  
Myasthenia gravis  
Hysteria  
Drugs (eg. curare derivatives)  

**Paralysis of Upward Gaze**  
Unable to look upwards  
Pinealoma

**Paralysis, Spastic**  
Involuntary spasm of muscles  
Spinal cord transection  
Cerebral palsy  
Cerebral or cord tumours  
Vascular accidents of cerebrum or cord  
Cerebral or cord infections  
Upper motor neurone lesions  
Multiple sclerosis

**Paraplegia and Quadriplegia**  
Paralysis of the lower or all four limbs  
Trauma to vertebral column and spinal cord  
Vertebral disc herniation (pain, hyporeflexia)  
Cord tumour or vascular malformation  
Multiple sclerosis (intermittent, variable)  
Infections of spinal cord (fever, backache)  
Meningitis (neck stiffness, headache, fever)  
Devic syndrome (visual loss)  
See also Paralysis and Muscular Weakness

**Paresis**  
See Paralysis and Muscular Weakness

**Parkinsonian Facies**  
Exp: Rigidity of facial muscles that gives a characteristic loss of facial expressiveness  
Int: (+) Parkinson's disease (paralysis agitans), encephalitis lethargica, cerebral arteriosclerosis, Wilson's disease, phenothiazines, manganese poisoning  
Phys: Degeneration of or damage to the basal ganglia results in muscular hypertonicity

**Parkinsonism**  
Exp: Hypokinesia, tremor and rigidity  
Int: (+) Parkinson's disease, familial, encephalitis lethargica, syphilitic mesencephalitis, tuberculoma, brain stem tumours, drugs (eg. phenothiazines, butyrophenones, tetrabenazine, rauwolfia alkaloids)  
Phys: Parkinson's disease and iatrogenic causes most common. Caused by degeneration of the substantia nigra and locus ceruleus  
See also Cogwheel Rigidity; Festination; Gait, Abnormal; Lead-Pipe Rigidity; Parkinsonian Facies; Tremor, Resting

**Personality Change**  
Psychiatric  
Paranoid personality disorder (eccentric)  
Schizophrenia (antisocial, unemotional)  
Histrionics (demands attention)  
Narcissistic personality disorder (reacts badly to criticism)  
Obsessive compulsive disorder  
Antisocial personality (conduct disorder)  
Borderline personality disorder (unstable)  
Phobic disorders (avoidance behaviour)  
Manic depressive psychosis (mood swings)  
Stress reaction  

Other  
Frontal lobe tumours  
Cerebral space occupying lesions  
Alzheimer's disease (confused, irrational)  
Arteriosclerosis
Pharyngeal Reflex

Exp: A spatula lightly touched against pharynx causes contraction of pharyngeal muscles and gagging

Int: (–) 9th cranial nerve lesions, hysteria, anaesthesia

Phys: Innervation of the pharynx is via a reflex arc involving the glossopharyngeal (9th) nerve

Plantar Reflex

See Babinski's Sign

Precocious Puberty

(Sexual Precocity)

Exp: Premature genital, axillary and facial hair; breast, clitoris or penis enlargement; voice changes; or menstruation

Int: (+) Adrenal cortical hyperplasia, cerebral or pineal tumours, Albright's syndrome, hypotalamic cysts or tumours, gonadal tumours, postencephalitic or postmeningitic lesions, hypothalamus, constitutional

Phys: Premature release of gonadotrophic hormones from the hypothalamus occurs with various forms of stimulation of that part of the cerebrum

Proptosis

See Exophthalmos

Ptosis

Drooping eyelid(s)
Bell's palsy (unilateral, spontaneous, painless)
Myasthenia gravis (generalised weakness)
Third cranial nerve palsy from any cause
Pseudoptosis (fat deposits in lid)

 Syndromes
Dubowitz syndrome (reduced growth, mental retardation)
Eaton-Lambert syndrome (myasthenic sympotms)
Guillain-Barré syndrome (progressive palsy)
Horner syndrome (myosis, exophthalmos, anhydrosis)
Marcus Gunn syndrome (lid twitch with jaw movement)
Möbius syndrome (ophthalmoplegia, dysphagia, drool)

Puberty

See Precocious Puberty

Puff Reflex

See Santmyer Swallow

Quadriplegia

See Paraplegia and Quadriplegia

Queckenstedt's Test

Exp: Pressure on either or both jugular veins during lumbar puncture normally produces a rise in the CSF manometric pressure, and then a drop when pressure on veins released

Int: (–) Block of spinal subarachnoid space or blocking of CSF escape from cerebral cavity (eg. vertebral fracture, TB, tumours of cord or vertebrae, haematomas)

Phys: Physical block of fluid connection via jugular veins, cerebral CSF and spinal CSF causes test to be negative

Radial Jerk

Exp: With the elbow flexed at 90° and the forearm in neutral position, the styloid process of the radius is tapped with a rubber hammer. Normal result is flexion of elbow and supination of forearm

Int: (–) Lower motor neurone lesion, peripheral neuropathy, polio, tabes dorsalis, posterior root tumours, syringomyelia, muscular dystrophies, subacute combined degeneration of cord, coma

(++) Upper motor neurone lesion, tetanus, CVA

Phys: Due to contraction of brachioradialis. Acts at C7, C8 level

See also other signs listed under Reflexes
Reflexes
See Ankle Jerk; Babinski's Sign; Biceps Jerk; Consensual Reflex; Corneal Reflex; Cremasteric Reflex; Grasp Reflex; Hering-Breuer Reflexes; Knee Jerk; Mass Reflex; Moro Reflex; Pharyngeal Reflex; Radial Jerk; Santmyer Swallow; Triceps Jerk

Retarded Mentality
See Intellectual Disability

Rigidity, Muscular
Wilson's disease  (cirrhosis, green-brown cornea) Frontal lobe disease Upper motor neurone lesions (hyperreflexia) Extrapyramidal lesions (cogwheel rigidity) Parkinson's disease (tremor, weakness, fixed facies) Multiple sclerosis Severe hypothyroidism (bradycardia, weak)

Syndromes
Machado-Joseph syndrome (dysarthria, weakness) Neuroleptic malignant syndrome (fever, tranquillizer use) Shy-Drager syndrome (tremor, vertigo, hypotension) Steele-Richardson-Olszewski syndrome (dementia, gaze paralysis) Stiff-man syndrome (idiopathic toxic muscular rigidity) See also Clasp-Knife Rigidity; Cogwheel Rigidity; Hypertonicity; Lead-Pipe Rigidity; Neck Stiffness; Opisthotonus

Rigor
See Convulsions; Shivering and Rigors

Romberg's Sign
Exp: Patient stands at attention with heels close together and then shuts eyes. Positive when severe swaying or falling occurs
Int: (+) Tabes dorsalis, posterior column lesion, subacute combined degeneration of cord, intoxication
Phys: Due to loss of proprioceptive sensation. Negative in cerebellar disease
See also Ataxia; Proprioceptive Sense, Loss of; Heel-Knee Test; Tandem Romberg Test

Santmyer Swallow
(Puff Reflex)
Exp: Blowing gently on the face of an infant induces a reflex swallow
Int: Useful procedure for easing the passage of a nasogastric tube or medication. Developmental assessment test – should have disappeared by 24 months of age

Scanning Speech
(Staccato Speech)
Exp: A speech form in which there are marked, and sometimes rhythmic, pauses between syllables and/or words
Int: (+) Cerebellar ataxia, multiple sclerosis
See also Slurred Speech

Scissors Gait
See Gait, Abnormal

Seizure
See Convulsions

Sensation, Loss of
See Anaesthesia

Sensory Suppression
See Agnosia

Shivering and Rigors
Fever from any cause  (eg. infection, malignancy) Hypothermia (environmental cold) Fear Malaria (jaundice, cyclical, hepatomegaly) Addictive drug withdrawal
See also Convulsions

Sleep Disturbance
See Insomnia; Sleep, Excess

Sleep, Excess
Cerebral space occupying lesion Head injury Narcolepsy  (sudden onset of sleep)
Other forms of epilepsy
Kleine-Levin syndrome (hungry, sexually overactive)
Psychiatric conditions
Drugs (eg. sedatives, antihistamines, antidepressants)

**Slow Movement**
See Bradykinesia; Rigidity, Muscular

**Slurred Speech**
Exp: Marked difficulty or trembling in attempts to pronounce words of several syllables
Int: (+) Drugs (eg. alcohol, sedatives), general paralysis of insane, Friedreich's ataxia, bulbar palsy
See also Scanning Speech

**Spasm, Muscular**
See Chorea; Convulsions; Cramps, Hypertonicity; Muscular; Myotonia; Rigidity, Muscular; Tetany; Tremor

**Spastic Paralysis**
See Paralysis, Spastic

**Speech**
See Aphasia and Dysarthria; Scanning Speech; Slurred Speech

**Staccato Speech**
See Scanning Speech

**Startle Reflex**
See Moro Reflex

**Subnormal Mentality**
See Intellectual Disability

**Swearing, Uncontrolled**
Attention deficit hyperactivity disorder
Gilles de la Tourette syndrome (convulsions, spasms)

**Syncope**
(Faint)

**Cardiovascular**
Stokes-Adams attack (heart block, bradycardia)
Myocardial infarct (chest pain, hypotension)
Hypotension from any cause (eg. blood loss, shock, postural)
Aortic stenosis (bruit, poor pulses)
Paroxysmal tachycardia (rapid irregular pulse, shock)
Pulmonary hypertension
Pulmonary artery stenosis
Atrioventricular block
Bradycardia (eg. heart block, infarct)
Vasovagal syncope (emotional, trauma)
Atrial fibrillation (irregularly irregular pulse)
Ventricular tachycardia
Carotid sinus syndrome (after head turning)
Mitral stenosis (murmur)
Anaemia (fatigue, pallor, dyspnoea)
Arteriosclerosis (mental deterioration)
Atrial myxoma
Cardiac tamponade
Hypertrophic cardiomyopathy

**Pulmonary**
Pulmonary embolism (shock, chest pain, cough)
Hypoxia from any cause (cyanosis, bradycardia)
Pulmonary hypertension (hepatomegaly)
Severe coughing fit
Hyperventilation due to tetany or fright

**Cerebral**
Emotional states, fear or pain
Cerebrovascular accident (headache, confusion, convulsion)
Transient ischaemic attack (brief, headache, confusion)
Migraine (headache, nausea, photophobia)
Cerebral tumours (neurological anomalies)
Cerebral abscess or cyst
Narcolepsy (excessive sleep)
Epilepsy (convulsions, amnesia, paroxysmal)
Vertebrobasilar disease (vertigo)
Parkinson’s disease (tremor, rigidity)
Meningitis (headache, fever)
Carotid ischaemia

**Syndromes**
Dumping syndrome (postgastrectomy, postprandial)
Jervell-Lange-Nielsen syndrome (deaf)
Sick sinus syndrome (variable heart rate)
Subclavian steal syndrome (arm claudication)
Wolff-Parkinson-White syndrome (loud first heart sound)
**A RATIONALE FOR THE BRAIN**

**Other**
- Severe infections (e.g. myocarditis)
- Hypoglycaemia (blurred vision, weakness, convulsions)
- Micturition syncope
- Addison's disease (weak, nausea, diarrhoea)
- Pregnancy (causes postural hypotension)
- Acidosis (hyperventilation, confusion)
- Autonomic peripheral neuropathy
- Anaphylaxis
- Dehydration (primary, or secondary to excess diuretics)
- Psychogenic (attention seeking)
- Alcohol dependence
- Drugs (e.g. hypotensives, tricyclics, phenothiazines, narcotics, hypnotics)

See also Coma; Drop Attack

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**Tandem Romberg Test**

Exp: Patient stands with one foot in front of the other (toe to heel in tandem), then closes eyes. Positive if patient sways or is unsteady within 60 seconds

Int: (+) Labyrinth, posterior column lesions, tabes dorsalis, subacute combined degeneration of cord, intoxication

Phys: More sensitive than standard Romberg's sign

See also Romberg's Sign

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**Tardive Dyskinesia**

See Dyskinesia, Tardive

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**Tics**

Repetitive muscular movement
- Emotional (habit, stress induced)
- Degenerative nervous disease
- Epilepsy (amnesia, variation in mental state)
- Basal ganglia disease
- Chorea (see separate entry)
- Psychoses (other psychiatric symptoms)

See Paraesthesia

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**Tone, Increased Muscular**

See Hypertonicity

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**Torsion Dystonia**

See Athetosis

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**Torticollis**

Exp: Spasm of the muscles in one side of the neck which causes the head to twist to that side. Straightening of the head usually causes pain

Int: (+) Muscle trauma, hysteria, rheumatoid arthritis, corpus striatum disease, adenitis of neck glands, neck abscess, ocular diseases, 11th cranial nerve disease, fibrositis, thermal trauma to neck, habitual tic, labyrinthine disease, tissue scarring, congenital

Phys: Sternomastoid muscle most commonly involved

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**Tremor**

Physiological tremor (minimal, arms, postural)
- Essential [benign familial] tremor (postural, arms, legs, head)
- Orthostatic tremor (14-16 Hz.,axial, unsteady)
- Anxiety and stress
- Muscular fatigue
- Cerebrovascular accident (intention tremor)
- Cerebral dysfunction (intention tremor)
- Cerebral trauma (tremor at rest)
- Cerebellar lesions (intention tremor)
- Cerebral tumour
- Multiple sclerosis (neurological signs, young adult)
- Wilson's disease (cirrhosis, rigidity, brown cornea)
- Chorea (involuntary jerks, speech impaired, weakness)
- Delirium tremens (alcohol, anxiety)
- Thyrotoxicosis (weak, sweating, weight loss)
- Peripheral neuropathy
- Neurosyphilis
- Hypoglycaemia
- Trypanosomiasis (apathy, neurological signs)
- Phaeochromocytoma (hypertension)
- Mercury or arsenic poisoning
- Addictive drug withdrawal
- Drugs (e.g. alcohol, caffeine, salbutamol, lithium, phenytoin)

**Syndromes**
- Diencephalic syndrome (cachexia, pallor, vomiting)
Neuroleptic malignant syndrome (tranquillizer use, fever)
Parkinsonism (rigidity, worse at rest, limbs, head, jaw)
Punch drunk syndrome (gait abnormal, boxer)
Serotonin synd. (tremor, antidepressant drugs)
Shy-Drager syndrome (hypotension, dysarthria, vertigo)

See also Chorea; Tremor, Flapping; Tremor, Intention; Tremor, Postural; Tremor, Resting

**Tremor, Flapping**
(Asterixis)
Exp: With arms, hands and fingers outstretched, the patient dorsiflexes the wrists and spreads the fingers. An irregular but synchronous tremor of a flapping nature is seen, with maximum activity at the wrist and metacarpophalangeal joints. May also involve feet and tongue
Int: (+) Encephalopathy, liver failure, metabolic diseases, subdural haematomas, cerebral infarcts in diencephalon region
Phys: Distortion of proprioceptive cerebral pathways and inappropriate motor stimulation

**Tremor, Intention**
(Dysmetria)
Exp: Attempt to touch nose with finger or perform any other definite movement causes increasing shaking of hand until movement is completed. Precision movement lost
Int: (+) Cerebellar lesions (eg. tumours, CVA, abscess), multiple sclerosis, brain stem disease, Friedreich's ataxia, spinocerebellar degenerations, mercury poisoning
Phys: Loss of appreciation of the force and rate of muscular contraction necessary for a movement

See also Dysdiadochokinesia

**Tremor, Postural**
Exp: Tremor obvious in certain postures such as outstretched hands
Int: (+) Anxiety, thyrotoxicosis, alcohol, drugs (eg. bronchodilators, tricyclics), heavy metal poisoning, Wilson's disease, neurosyphilis, cerebellar lesions, familial
Phys: Loss of fine control of flexor/extensor reflex

**Tremor, Resting**
Exp: Rhythmic sinusoidal movement of limbs and/or head at rest
Int: (+) Parkinson's disease (paralysis agitans), postencephalitic, cerebral tumours, drugs (eg. reserpine, phenothiazines)
Phys: Degeneration of substantia nigra and associated pathways
See also Cogwheel Rigidity; Gait, Abnormal

**Triceps Jerk**
Exp: With the elbows at 90° and relaxed, the triceps tendon is tapped just above the point of the elbow. This results in extension of the elbow
Int: (−) Poliomyelitis, peripheral neuropathies, posterior root disease, tabes dorsalis, spinal cord tumours or degenerations, spinal shock, syringomyelia, muscle dystrophies, coma
(+++) Upper motor neurone lesion, tetanus, hyperthyroidism, anxiety, cord transection
Phys: Acts at C6, C7, C8 level. Due to contraction of triceps brachii muscle
See also other signs listed under Reflexes

**Twitching**
See Fasciculation, Muscular; Tremor

**Unconscious**
See Coma; Drop Attack; Syncope (Faint)

**Vertigo**

Dizziness/giddiness

**COMMON CAUSES OF VERTIGO**

<table>
<thead>
<tr>
<th>DISEASE</th>
<th>INCIDENCE</th>
<th>DURATION</th>
</tr>
</thead>
<tbody>
<tr>
<td>Benign positional vertigo</td>
<td>16%</td>
<td>Seconds</td>
</tr>
<tr>
<td>Cerebrovascular ischaemia</td>
<td>6%</td>
<td>Minutes</td>
</tr>
<tr>
<td>Menière's disease</td>
<td>5%</td>
<td>Hours</td>
</tr>
<tr>
<td>Labyrinthitis</td>
<td>9%</td>
<td>Days</td>
</tr>
</tbody>
</table>

**Otic**
Menière's disease (deaf, tinnitus, nausea)
Labyrinthitis (nystagmus, stagger, tinnitus)
Eighth cranial nerve damage (eg. neuroma)
Otitis media (pain, fever)
Eustachian tube blockage (ear discomfort, deaf)
Vestibular neuronitis (nystagmus, resting vertigo)
Mastoiditis (pain, mastoid tender, fever)
Perilymphatic fistula
Cardiovascular and Circulatory
Myocardial infarct (shock, chest pain)
Postural hypotension
Vertebralbasilar insufficiency (diplopia, dysarthria, faint)
Myocarditis (asthenia, dyspnoea, nausea)
Arteriosclerosis (mental deterioration and confusion)
Anaemia (pallor, fatigue, palpitations)
Hypertension (headache)
Cardiac arrhythmias

Central Nervous System
Motion sickness
Temporal lobe epilepsy (aura, faint)
Cerebellar lesions (incoordination, headache, nausea)
Psychiatric disorders
Cerebrovascular accident (confusion, headache, aphasia)
Transient ischaemic attack (brief, confusion, headache)
Migraine (headache, nausea, photophobia)
Multiple sclerosis (weakness, abnormal sensation)
Parkinson's disease (tremor, stiffness)
Subdural haematoma
Cerebral tumours

Syndromes
Cogan syndrome (tinnitus, deaf)
Dandy syndrome (bilateral loss of vestibular function, loss of foveation)
Post-traumatic cerebral syndrome (headache)
Ramsay Hunt syndrome (earache, Herpes zoster)
Shy-Drager syndrome (hypotension, tremor, ataxia)

Other
Hyperventilation from any cause
Cervical spine osteophytes (neck pain and stiffness)
Thyrotoxicosis (sweating, heat intolerance)
Hypothyroidism (cold intolerance, dry skin)
Diabetes mellitus (paraesthesiae, blurred vision, polyuria)
Hypoglycaemia (insulin overdosage)
Benign paroxysmal positional vertigo (worse lying)
Addison's disease (weak, pigmentation)
Acromegaly (coarse facies, psychoses, back pain)
Pre-eclampsia (pregnancy, hypertension)
Syphilis (various neurological symptoms)
Trauma (eg. fractured temporal bone)
Altitude sickness (headache, drowsiness, nausea)
Drugs (eg. phenytoin, benzodiazepines, phenobarbitone, salicylates, gentamicin, streptomycin)

Vision, Double
See Diplopia

Voice, Abnormal
See Aphasia and Dysarthria

Waiter's Hand
Exp: The arm is adducted and extended at the elbow and the forearm is pronated, with the palm facing backwards as though accepting a surreptitious tip
Int: (+) Erb-Duchenne paralysis
Phys: Damage (usually at birth) to the 5th cervical anterior nerve root and subsequent paralysis of deltoid, brachioradialis and biceps

Wasting, Muscular
Reduction in muscle bulk
Localised
Paralysis of muscle bundle (eg. paraplegia, motor neurone disease, plexus palsy)
Congenital

Generalised
Diabetes mellitus (thirst, polyuria, fatigue)
Thyrotoxicosis (tachycardia, sweats, proptosis)
Addison's disease (weak, nausea, diarrhoea)
Phaeochromocytoma (severe hypertension)
Occult carcinoma
Hypopituitarism
Anorexia nervosa
Fad diets

Weakness, Muscular
See Paralysis and Muscular Weakness; Fatigue, Abnormal; Floppy Baby; Facial Weakness

Wrist Drop
Exp: Inability to extend wrist
Int: (+) Radial nerve lesion, peripheral neuropathy, muscular dystrophy, lead poisoning
Phys: Lower motor neurone lesion of wrist extensors
Section Three

MEDICAL CONDITIONS WITH NEUROLOGICAL SYMPTOMS
ABETALIPOPROTEINAEMIA
Abetalipoproteinaemia (Bassen-Kornzweig syndrome) is a rare abnormality of body fat metabolism (chemistry). The cause is a congenital birth defect that prevents absorption of fat and vitamin E from food and prevents the production of necessary body fats in the liver.

The patient is a child with foul smelling fatty diarrhoea, low body weight, retarded growth, poor co-ordination, abnormal sensations and a pigmented retina at the back of the eye. Permanent brain damage is a common complication if not diagnosed early.

Abnormal red blood cells and very low levels of fats (cholesterol and triglycerides) are found in blood tests.

The only treatment is a fat restricted diet, special triglyceride dietary supplements and vitamin E but the prognosis is generally poor.

ACROCEPHALOSYNDACTYLY OF APERT
The acrocephalosyndactyly of Apert (Apert syndrome) is a very rare familial disorder of hand and head development that results from premature fusion of the bones that make up the skull and abnormal development of the fingers. The head rises to a point, eyes protrude, the cheeks are sunken and two or more fingers are fused together. Intellectual disability may also occur. Surgery can separate the fingers and open up the suture lines between the skull bones but there is no cure.

ACROMEGALY
Acromegaly is excess growth in specific parts of the body due to over production of growth hormone in the pituitary gland, which sits underneath the brain. Growth hormone is required for the normal growth of a child, but if it is produced inappropriately later in life, acromegaly results. The most common reason for this is a tumour in the pituitary gland, but occasionally tumours elsewhere can secrete the hormone. Some sportsmen and body builders take growth hormone supplements inappropriately and develop acromegaly.

Patients have excessive growth of the hands, feet, jaw (macrognathia), face, tongue and internal organs. They also suffer headaches, sweating, weakness, and loss of vision. A woman’s menstrual periods will stop, and diabetes insipidus is a common complication of the disease and its treatment.

Blood tests can be used to prove the diagnosis, and X-rays and CT scans of the skull can detect the tumour. Specialised microsurgery is performed through the nose, and up into the base of the brain, to remove the tumour. Occasionally irradiation of the tumour may be performed. Usually hormone supplements must be taken long term to replace those normally produced by the destroyed pituitary gland. Treatment is very successful, particularly in younger adults.

ACUTE BRAIN SYNDROME
The acute brain syndrome (acute confusional syndrome) is common in suddenly hospitalised or relocated elderly patients due to disorientation.

There is an abrupt onset of confusion in an elderly person who is psychotic, has clouding of their consciousness, impaired thought processes, poor short-term memory, strange illusions, misinterpretations, anxiety, irrational fears, loss of interest in life, restlessness and apathy. Some patients experience hallucinations that involve the sensations of vision, touch, hearing and smell. Sedatives may aggravate the confusion.

It is diagnosed by psychiatric assessment and treated by careful nursing, psychotherapy and medications (eg. haloperidol). It often persists but may settle spontaneously with time.

See also ALZHEIMER DISEASE; DEMENTIA; PSYCHOSIS

ACUTE HEPATIC PORPHYRIA
Acute hepatic porphyria is one of a number of different uncommon types of porphyria, which is a liver disease.
It is an inherited disease that passes from one generation to the next but causes symptoms in only 10% of those affected.

Symptoms develop at the time of puberty with vague abdominal pains, nausea, vomiting and abnormal sensations. As the disease progresses, the abdominal pains may become severe, but nothing abnormal can be found in the abdomen. In advanced cases, nerve pain, paralysis, personality changes and fits may occur. The urine turns a dark purple colour, then brown, if left standing. Some patients may have the otherwise quiescent disease triggered by severe infections, starvation, some drugs or steroids. Complications include liver damage, which may progress to liver failure or liver cancer (hepatoma), and nerve damage which may cause varying forms of paralysis.

It is diagnosed by special blood tests. Treatment involves careful genetic counselling of families and avoiding factors (eg. crash diets, emotional stress, alcohol, certain drugs) that may precipitate an attack, and it is controlled by the use of a complex drug regime.

Acute hepatic porphyria may be controlled, but not cured, and death may occur due to the paralysis of the muscles of breathing.

See also PORPHYRIA; PORPHYRIA CUTANEA TARDA

ADDISON DISEASE

Addison disease is also known as adrenocortical Insufficiency and chronic hypoadrenocorticism. It is a rare underactivity of the outer layer (cortex) of the adrenal glands that sit on top of each kidney, and produce hormones (chemical messengers) such as cortisol that control the levels of vital elements in the body and regulate the breakdown of food. In most cases, the reason for adrenal gland failure is unknown, but tuberculosis is a possible cause.

The symptoms include weakness, lack of appetite, diarrhoea and vomiting, skin pigmentation, mental instability, low blood pressure, loss of body hair and absence of sweating. Complications include diabetes, thyroid disease, anaemia, and eventual death. A sudden onset of disease is known as an Addisonian or adrenal crisis, which may be rapidly fatal.

It is diagnosed by special blood tests that measure the body’s response to stimulation of the adrenal gland (synacthen stimulation test).

Treatment involves a combination of medications (eg. steroids such as cortisol) to replace the missing hormones, and dosages vary greatly from one patient to another. Frequent small meals high in carbohydrate and protein are eaten, and infections must be treated rapidly. Patients must carry an emergency supply of injectable cortisone with them at all times. Treatment can give most patients a long and useful life, but they cannot react to stress (both physical and mental) adequately, and additional treatment must be given in these situations. The ultimate outcome depends greatly on the patient’s ability to strictly follow all treatment regimes.

The condition is named after the English physician Thomas Addison (1793-1860).

ADDISONIAN CRISIS

See ADDISON DISEASE

AFFECTIVE DISORDER

See DEPRESSION

ALBRIGHT SYNDROME

Albright syndrome (fibrous dysplasia or McCune-Albright syndrome) is characterised by the abnormal formation of bone and body chemistry abnormalities. It is a congenital condition and usually occurs in young females who have early puberty and abnormal bone formation in fibrous tissue. Some patients have an overactive thyroid gland and acromegaly.

The abnormal bone deposits show on X-rays and specific blood and urine tests are also abnormal.

Surgical correction of bony deformities and steroids are the treatments available, but there are variable results from treatment.

ALCOHOLISM

Alcohol abuse may be a temporary problem as a reaction to anxiety or stress, but it may lead to the chronic medical condition of alcoholism (sometimes referred to by the slang term “dipsomania”).
A RATIONALE FOR THE BRAIN

Alcoholism affects up to 3% of the adult population in developed countries. It is a disease in the same way that infections and cancer are diseases. It does no good to tell an alcoholic to “pull yourself together” or “stop drinking before it kills you”. They need professional counselling and treatment. The biggest problem faced by families and doctors is the denial by so many alcoholics that they have a problem.

When alcohol is swallowed, it is absorbed very rapidly from the stomach, and commences its actions on the brain and other organs. This of course is one of the attractions of alcohol - it can make you very happy very quickly, and this can lead to addiction in some people. The children of alcoholics are more likely themselves to become alcoholics, and should be very wary when using alcohol.

Blood tests on liver function and alcohol levels may confirm diagnosis, and an ultrasound scan of the liver may show damage (cirrhosis).

Alcoholism has two stages of development - problem drinking, and alcohol addiction. Problem drinking is the use of alcohol intermittently to ease tension and anxiety. It may be associated with the use of prescription drugs to control emotional problems. Alcohol addiction is more serious.

An alcoholic is someone who has three or more of the following symptoms or signs:-
- drinks alone
- tries to hide drinking habits from others
- continues to drink despite convincing evidence that it is damaging their health
- disrupts work or social life because of alcohol
- craves alcohol when none is available
- appears to tolerate the effects of alcohol well
- blacks out for no apparent reason
- binges on alcohol
- averages six standard alcoholic drinks a day
- has abnormal liver function blood tests.

The social complications of alcohol are obvious and vary from the disruption of family life to poor performance at work and the risks of drink-driving.

The medical effects of alcoholism can be serious to the point where they can significantly alter the quality of life and shorten the life of the alcoholic. They include:-
- Cirrhosis. In this, the soft normal liver tissue is replaced by firm scar tissue that is unable to process the waste products of the body adequately. The other vital actions of the liver in converting and storing food products and producing chemicals essential to the body are also inhibited.
- Wernicke-Korsakoff psychosis. This syndrome causes brain damage with symptoms of depression, irrational behaviour and insanity. These conditions are related to vitamin deficiencies caused by an inadequate diet while on alcoholic binges.
- Degeneration of the cerebellum (the part of the brain that is at the back of the head) caused by alcoholism can cause permanent incoordination, difficulties in walking and performing simple tasks.
- Peripheral neuropathy is damage to the nerves supplying the body. It causes muscle cramps, pins and needles sensations and muscle pains.

Treatment involves counselling, professional treatment programs in hospital, supportive groups (eg. Alcoholics Anonymous), medications to ease withdrawal and prevent relapses (eg. acamprosate, calcium carbimide, disulfiram, naltrexone). Withdrawal from alcohol may cause delirium tremens.

The medical effects of alcoholism can be serious to the point where they can significantly alter the quality of life and shorten the life of the alcoholic, and adversely impact on other members of the family.

See also WERNICKE-KORSAKOFF PSYCHOSIS

ALTITUDE SICKNESS

Altitude or mountain sickness (called soroche in Peru and Bolivia) is caused by lack of oxygen from ascending rapidly to heights over 3000m. A slow ascent is less likely to cause problems than a rapid one. It is impossible to predict who will be affected, how rapidly or at what altitude. Ascending at a rate of no more than 300m a day at altitudes over 3000m is less likely to result in problems. An extra rest day with every 1000m above 2500m is also useful.

Symptoms starts with a headache, shortness of breath, and excessive tiredness, followed by inability to sleep, nausea, vomiting, diarrhoea, abdominal pains and a fever. Fluid fills the lungs, patients start coughing up
A RATONALE FOR THE BRAIN

blood, the heart races, and they may eventually drown as blood fills the lungs. Permanent lung and other organ damage may result from a severe attack.

A rapid descent to a lower altitude is the only effective treatment for severe cases, although mild cases may recover with rest at high altitude. Fluid removing drugs (diuretics) may be used in an emergency to remove fluid from the lungs, and acetazolamide (125 mg. twice a day from the day before ascent for three days after ascent) or dexamethasone (4 mg. twice a day from the day before ascent for three days after ascent, then 2 mg a day for two days) may be given during the climb for prevention. Oxygen in cylinders is used by very high altitude climbers.

The condition may be life-threatening unless a lower altitude can be reached.

ALZHEIMER DISEASE

Alzheimer disease (or senile dementia) use to be called second childhood, or the person was described as eccentric. Today it is recognised as the most common form of dementia in the elderly, but it may start as early as the mid-fifties.

It is named after the Wroclaw (Poland) neurologist Alois Alzheimer, who was born in 1864, and first described the disease in the medical literature.

The cause is a faster than normal loss of nerve cells in the brain, the exact cause of which is unknown, but studies suggest specific genes may predispose a person to the disease, and there is a familial tendency (runs in families from one generation to the next).

Initially it causes loss of recent memory, loss of initiative, reduced physical activity, confusion and loss of orientation (confused about place and time), then progresses to loss of speech, difficulty in swallowing which causes drooling, stiff muscles, incontinence of both faeces and urine, a bedridden state and eventually the patient is totally unaware of themselves or anything that is happening around them. Some patients may not deteriorate for some time, then drop to a lower level of activity quite suddenly. Admission to a nursing home or hospital is eventually necessary.

Reduced brain volume and wasting may show on a CT scan, but the diagnosis is primarily a clinical one made by a doctor after excluding all other forms of dementia by blood tests, X-rays, electroencephalogram (EEG) and sometimes taking a sample of the spinal fluid. The progress of the disease can be followed by tests of skill, general knowledge, simple maths, etc.

Medication is useful for restlessness and insomnia, and a number of medications (eg. rivastigmine, tacrine, memantine) are now being used to slow the progression of the disease. In women, hormone replacement therapy after menopause reduces the incidence of Alzheimer disease, and slows its progress. Visits by the family general practitioner, physiotherapists, occupational therapists, home nursing care and health visitors are the main forms of management. Many claims have been made for various herbal remedies, but none have proved to be beneficial.

There is no cure, and treatments are aimed at keeping the patient content. Medications in the anticholinesterase class (eg. donepezil) have been shown to slow the progression of the disease in some patients. From diagnosis to eventually death takes seven years on average.

See also DEMENTIA; ORGANIC BRAIN SYNDROME

AMNESTIC SYNDROME

The amnestic syndrome is a complication of alcoholism involving the brain. The cause is a lack of thiamine (vitamin B3) caused by alcoholism. Patients have a sudden onset of short term memory disturbances and permanent loss of memory of periods when affected. The other complications of alcoholism are also common. Blood tests show low levels of thiamine, liver damage and presence of alcohol. The prognosis is poor as the alcoholism is well advanced by this stage.

AMYOTROPHIC LATERAL SCLEROSIS

Amyotrophic lateral sclerosis is a rare form of motor neurone disease that affects the nerves that supply the muscles of the body. It is sometimes known as Lou Gehrig disease after a 1930s American baseballer who developed the condition.

The absolute cause unknown, but it may run in families, and results in a steadily progressive degeneration of the motor nerves in the body. Symptoms may include muscle weakness that usually starts in the hands or feet, muscle cramps and twitches, difficulty in swallowing and talking, drooling of saliva, inability to cough effectively, reduced tongue movement, and progressive weakness up the arms and legs. Eventually the muscles used for
breathing are involved and lung infections such as pneumonia develop, and often lead to death within a few years of diagnosis. Electrical tests of the motor nerves are used to determine how well they are functioning, and a nerve biopsy is diagnostic.

No cure is available, and treatment is aimed at relieving muscle spasm, assisting feeding, preventing infections, aiding breathing and making the patient as comfortable as possible. The medication riluzole is now being used in some countries to slow the progress of the disease. Physiotherapy on a very regular basis is essential.

See also MOTOR NEURONE DISEASE

ANGELMAN SYNDROME
The Angelman or happy puppet syndrome is a rare congenital cause of intellectual impairment due to damage on chromosome 15. One case occurs in every 25,000 children who have severe intellectual disability, inability to talk, abnormal walk, intractable seizures, inappropriate laughter, a small head and abnormal facial structure. It is diagnosed by chromosome analysis. No treatment is available and patients have a shorter than normal life span.

See also INTELLECTUAL DISABILITY

APERT SYNDROME
See ACROCEPHALOSYNDACTYLY OF APERT

ARNOLD-CHIARI MALFORMATION
See SYRINGOMYELIA

ASTROCYTOMA
An astrocytoma is a slow growing, low-grade malignant or benign tumour arising in the connective cells of the brain. The cause is unknown.

The symptoms are often very mild and confusing in the early stages, and as a result the tumour may be quite large before it is detected. Common symptoms include visual disturbances, abnormal pituitary gland function, paralysis of facial muscles, incoordination and difficulty in walking. Rarely it may become aggressively malignant and progress rapidly as a glioblastoma.

The tumour can be visualised by CT and MRI scans, but a biopsy is required for final diagnosis. Treatment involves surgery to remove growth, and any parts that cannot be removed are treated with drugs (eg. temozolomide). Because of their size when diagnosed, they sometimes cannot be completely removed. They are often cured if complete surgical excision is possible, and the outcome is often better in children.

See also GLIOMA

ATRIAL FIBRILLATION
The heart has two small chambers (atria) which receive blood from the lungs and body through large veins, and two large chambers (ventricles) which pump blood out through arteries to the lungs and body. Atrial fibrillation occurs if the atria beat in a rapid uncoordinated manner, and as a result the ventricles (main pumping chambers of the heart) will receive only an intermittent blood supply from the atria, and will beat in a very irregular rhythm. If the atria beat rapidly, but not fast enough to cause irregular contractions by the ventricles, the condition is atrial flutter.

It may occur in normal people at times of stress, but more commonly as a reaction to heart damage such as a heart attack or infection. Other causes include an overactive thyroid gland, heart valve damage, severe high blood pressure, lung damage that restricts blood flow (eg. emphysema), or because of imbalances in body chemistry.

Patients have a very irregular pulse, tiredness due to low blood pressure, palpitations and sometimes chest pains, shortness of breath and fainting. An embolism (blood clot) that may cause a stroke or death if it travels through arteries to the brain may occur due to the formation of a clot in the heart with the irregular pressure patterns caused by the fibrillation. There is also an increased risk of heart failure and heart attack.

It is diagnosed with an electrocardiograph (ECG), but doctors can usually make the diagnosis by analysing the irregular heart beat rhythm.
Numerous medications or electric shock treatments to the heart (electrocardioversion) are available to control the heart rhythm. If the atrial fibrillation remains uncontrolled, an anticoagulant (eg. warfarin) should be used to prevent an embolism.

Most cases can be controlled by medication, but if persistent there is a small mortality rate due to complications.

**ATTENTION DEFICIT HYPERACTIVITY DISORDER**

Attention deficit hyperactivity disorder (ADHD) is a very complex behaviour problem. A subtype is attention deficit disorder (ADD or minimal brain dysfunction) in which there is no excess activity.

Patients are most commonly males who inherit the disorder from their father’s side of the family, and it affects between 3 to 8% of primary school students. Boys show more aggressive and impulsive symptoms, while girls seem to have a lack of attention due to daydreaming.

These children are often fidgeting, unable to remain seated for long, unable to play quietly, easily distracted, unable to sustain attention, always impatient, have difficulty in following instructions, often move from one incomplete task to the next, talk excessively, often interrupt or intrude, do not seem to listen, have poor short term memory, often lose items and engage in physically dangerous activities. Most are average or above average in intelligence, but due to their genuine inability to pay attention and control their impulsiveness often do not take in all of the information in school. 30% have a reading disorder and 10-15% have other academic disabilities. It may lead to criminal activity in the teens and early adult life.

Treatment involves behaviour modification with the assistance of a psychologist and the cooperation of the parents, social skills training, family counselling, psychostimulant medication (eg. methylphenidate) and occupational therapy. Many professionals have conflicting ideas about the best form of treatment, and it is a process of trial and error to find the best treatment for an individual. Diet modification is commonly thought to be useful, but there is no evidence to support this. Every individual will respond differently to treatment, but most grow out of the problem in their mid-teens.

**AUTISM**

Autism is one of the autism spectrum disorders that include Asperger syndrome and Rett syndrome as well as autism. It is a social developmental disorder that may be an abnormality in the development of the brain due to damage during growth as a foetus, at birth, or in the first years of life. The absolute cause is not known, but the incidence is increasing and it now affects about one in every 200 children and is four times more common in boys than girls and appears to be more common in developed countries.

The child fails to develop normal social skills, language skills and communication skills. They are often excessively preoccupied with a particular type of behaviour and very resistant to change or education. Repetitive habits are common. Occasionally they have exceptional talents in a particular area (eg. maths or music - the idiot savant syndrome). Epilepsy occurs in up to 30% of cases.

The main difference between autism and Asperger syndrome is that in the latter there is no major delay in speech development and intellectual ability is relatively normal.

There has been a lot of speculation about the cause of autism, but none has been proven. There is no evidence that immunisations or intestinal disturbances play any roll in its development.

There are no diagnostic tests, but CT brain scans sometimes show non-specific abnormalities. Electroencephalograms (EEG) are usually normal, except in those who develop epilepsy.

Treatment is difficult and prolonged, and primarily involves education and behaviour therapy. Many patients remain mentally below normal and most require care throughout their life, but life expectancy is close to normal. Treatments that have been tried include many different forms of intensive behaviour and physical therapy, and medications such as risperidone, amphetamines (eg. methylphenidate), clonidine, antidepressants (eg. fluoxetine) and anticonvulsants.

Because treatment is difficult and time consuming many parents seek alternative treatments that vary from chelation to megadose vitamin supplements. Unfortunately there is no evidence that any of these treatments work, and they may have a significant deleterious effect on the parent’s bank balance.

**AUTOIMMUNE DISEASES**

Autoimmune diseases are a large group of diverse conditions that cause the body to inappropriately reject some specific types of its own tissue in the same way that transplanted organs or tissue may be rejected. The
reaction is due to disorders of the immune system. For example, in rheumatoid arthritis, the synovial membrane lining a joint is rejected.

Antigens in the body's own tissue inappropriately stimulate the production of antibodies (autoantibodies) against the tissue. Almost any tissue type in the body may become involved in an autoimmune reaction, and the symptoms (and disease diagnosis) depend upon which tissues are involved.

The absolute cause of autoimmune diseases is unknown, but they often follow physical or emotional stress, or viral infections.

Examples of autoimmune diseases (and the affected tissue) include Devic disease (optic nerve and spine), halo naevus (skin moles), Hashimoto thyroiditis and some forms of hyperthyroidism (thyroid gland), idiopathic pulmonary fibrosis (lungs), the Koebner phenomenon, pemphigoid, pemphigus, pyoderma gangrenosum, scleroderma and psoriasis (different layers within the skin), relapsing polychondritis and rheumatoid arthritis (joints), scleritis (the eye), temporal arteritis (arteries), thrombocytopenia (blood cells), transverse myelitis (spinal cord), polymyalgia rheumatica and myasthenia gravis (muscles), the nephrotic syndrome (kidneys), polyglandular autoimmune syndromes (glands throughout the body), Schmidt syndrome (adrenal and thyroid glands), collagenous colitis (large bowel) and the more generalised rejection of connective tissue that occurs with mixed connective tissue diseases, Sjögren syndrome, systemic lupus erythematosus and Churg-Strauss syndrome.

**BARTTER SYNDROME**

Bartter syndrome is a rare inherited kidney condition that is far more common in females than males. It is caused by failure of the kidney to conserve adequate potassium in the blood, giving the body chemistry effects of Conn syndrome without the high blood pressure.

The condition presents as a child or young adult with short stature, who frequently passes large quantities of urine day and night (often causing bed wetting) and has muscle weakness. Some patients develop muscle spasms and cramps, a craving for salt, and in advanced stages vomiting and constipation occur. Numerous blood and urine tests are abnormal.

Indomethacin is used to reduce kidney inflammation, medication (eg. spironolactone) is given to reduce potassium loss, and potassium supplements are taken. There is no cure.

**BASSEN-KORNZWEIG SYNDROME**

See ABETALIPOPROTEINAEMIA

**BATTEN DISEASE**

Named after the English ophthalmologist Frederick batten (1865-1918), batten disease is a progressive brain disorder (encephalopathy) of children between five and ten years of age. These children are unable to metabolise (break down) polyunsaturated fatty acids due to the lack of an enzyme, and these fatty acids accumulate in the brain to cause sudden blindness and progressive mental deterioration.

**BEHÇET SYNDROME**

Behçet syndrome is a serious condition of unknown cause that results in widespread apparently unconnected symptoms such as recurrent severe mouth and genital ulcers, inflammation of the eye, arthritis and brain abnormalities such as convulsions, mental disturbances, partial paralysis and brain inflammation. Other symptoms may include rashes (eg. erythema nodosum), skin ulcers, inflamed veins and blindness.

Treatment is often unsatisfactory. Steroids and immune suppressant medications are used, but the condition usually follows a long course with spontaneous temporary remissions. It is often seriously disabling and sometimes fatal.

**BELL’S PALSY**

Facial muscles are controlled by the facial nerve, which comes out of a hole in the skull just below and in front of the ear. From there, it spreads like a fan across the face to each of the tiny muscles that control facial expressions. Inflammation of the nerve at the point where it leaves the skull causes the facial muscles to stop working. The exact reason for this inflammation is unknown.

Patients with Bell's palsy (idiopathic facial paralysis) experience a sudden paralysis of the facial muscles on one side only. They can no longer smile or close the eye properly. There may be some mild to moderate pain at
the point where the nerve leaves the skull beside the ear, but this settles after a few days. There may also be a
disturbance to taste sensation.

No treatment is necessary for most patients, but in the elderly, if the paralysis is total, or if there is severe
pain, treatment with high doses of prednisone (a steroid) may be tried, provided it is started within five days of
onset.

10% of patients are significantly affected long term by facial paralysis, but two thirds of patients recover
completely within a few weeks with no treatment. Most of the others obtain almost complete recovery.

**BENIGN PAROXYSMAL POSITIONAL VERTIGO**

Benign paroxysmal positional vertigo (BPPV) is sometimes shortened to benign positional vertigo. It is an
annoying but harmless cause of dizziness due to loose particles floating in the semicircular canals (vestibular
apparatus) in the inner ear that control balance. The cause is usually unknown, but it may follow a head injury.

Patients experience sudden, severe, brief, episodes of dizziness that are worse lying down. Attacks are
triggered by any movement of the head. Some patients find that a particular head movement (eg. looking up)
starts an attack.

The Hallpike test is diagnostic. This involves lying the patient down with the head hanging down off the top of
a bed. The head is then rotated and abnormal eye movements (nystagmus) are noted if the test is positive.

A series of head manoeuvres (Brandt Daroff exercises or Epley manoeuvres) is carried out under the
direction of a specialist doctor or physiotherapist to remove the loose particles from the vestibular apparatus.
Surgery is a treatment of last resort, but drugs are not helpful.

Treatment is moderately successful, but the condition usually settles spontaneously after many months or
years.

See also MÉNIÈRE’S DISEASE; VESTIBULITIS

**BERIBERI**

Beriberi is caused by a lack of thiamine (vitamin B1) in the diet of those who are malnourished, have food
idiosyncrasies, overcook their food, in alcoholics (who obtain nutrition from alcohol and neglect normal food) and
in those who require abnormally large amounts of thiamine due to an overactive thyroid gland or prolonged fever.

In early stages patients experience a multitude of vague complaints including tiredness, loss of appetite,
twitching, and muscle cramps and pains. In later stages swollen joints, shooting pains, paralysis of feet and
hands, and heart abnormalities occur.

Thiamine supplements (initially by injection in severe cases) and a well-balanced diet rapidly control the
condition, but permanent organ damage is possible in advanced cases.

See also ALCOHOLISM

**BLUE DIAPER SYNDROME**

The blue diaper syndrome is a rare metabolic (body chemistry) defect due to defect in absorption of an amino
acid (tryptophan) from the gut. This is degraded by the body to other substances (indoles), which are passed out
in the urine and react with napkin starch to produce a blue stain.

Affected children have an intellectual disability, deposition of calcium in tissues throughout the body and
formation of calcium based kidney stones. High levels of calcium are found in the blood. No treatment or cure is
available.

**BORDERLINE PERSONALITY**

A borderline personality is one in which the person has unstable mood patterns, variable self-image,
interpersonal relationships that fluctuate significantly and an inability to make significant decisions. Many
affected people believe that all things are either good or bad, and are unable to see subtle differences or shades
of a decision or discussion.

**BOTULISM**

Botulism is an extremely severe form of food poisoning. Home-preserved fruits and vegetables, and very
rarely commercially canned foods, may be responsible for harbouring the bacterium *Clostridium botulinum*,
which is capable of producing an extremely potent poison (toxin) that attacks the nervous system.
A RATIONALE FOR THE BRAIN

Twelve to 36 hours after eating inadequately preserved food, the patient develops double vision, difficulty in swallowing and talking, a dry mouth, nausea and vomiting. The muscles become weak, and breathing becomes steadily more difficult. The patient must be hospitalised immediately and put upon an artificial breathing machine (ventilator) to maintain lung function once the paralysis occurs. An antitoxin is also available for injection.

Death occurs in about 70% of patients unless adequate medical treatment in a major hospital is readily available. In the best circumstances, up to 25% of patients will still die.

BRACHIAL AMYOTROPHY

Brachial amyotrophy (acute brachial neuritis, neuralgic amyotrophy or the Parsonage-Turner syndrome) is a rare failure of some or all of the nerves in the brachial plexus to function correctly. The brachial plexus is a network of nerves in the armpit that connects the nerves coming from the spinal cord in the neck to the nerves that run down the arm to supply sensation and cause muscle movement. The condition is more common in men than women. The absolute cause is unknown but it often follows major injury, surgery or childbirth.

The patient experiences disturbances in sensation and muscle weakness in the affected arm. A permanent loss of function or sensation in the arm is possible. Specific electrical tests on nerve function in the arm can confirm the diagnosis. No specific treatment is available but physiotherapy may be useful in preserving arm function and most patients recover slowly over several years.

See also ERB-DUCHENNE PALSY

BRAIN ABSCESS

A brain (cerebral) abscess is a collection of pus within or on the brain due to localised infection in the brain or its surrounding structures (e.g., skull). The most common cause is a penetrating wound into the brain that allows infection to enter directly. Treatment involves antibiotics against the original focus of infection, surgical drainage or excision of the abscess cavity and irrigation of abscess cavity with antibiotic solution. If the patient is debilitated, the spread of infection may be controlled by intensive intravenous treatment with appropriate antibiotics. The prognosis is reasonable if the condition is aggressively treated, but it is fatal if left untreated.

BRAIN CANCER

Cancer of the brain is uncommon, and most general practitioners would on average see a case only once every five years or so. There are many different types of cell in the brain, including nerve cells, membrane cells, glandular cells, and cells that secrete the fluid that surrounds the brain. Each of these different types of cells can develop one or more different cancers. Metastatic cancers can also be deposited and grow in the brain after migrating through the blood stream from other parts of the body including the lung, kidney and breast.

More than half of all brain cancers are gliomas. These develop from the support cells that surround and separate the nerve cells in the brain and spinal cord.

The symptoms of brain cancers and tumours (abnormal growths that are not necessarily cancer) are very varied, depending on the type, size, and position in the brain. Symptoms that may occur by themselves or in combinations include convulsions, twitching, personality changes, nausea and vomiting, intellectual decline, strange sensations, loss of speech or sight, confusion and headaches. Contrary to popular belief, in only 20% of all cases is headache the first symptom noted by patients. Later in the development of the condition, the patient may become paralysed, unconscious and have difficulty in breathing.

When a brain cancer is suspected, investigation will include blood tests, X-rays (sometimes involving the injection of dye into the arteries supplying the brain), measuring the brain waves electrically (electroencephalogram), CT and MRI scans, taking samples of the fluid around the brain (spinal tap) and injecting safe radioactive material into the blood stream and using scanners to see how it is concentrated in the brain. A biopsy is often necessary to confirm the diagnosis.

Treatment also varies depending on the type and position of the cancer. Surgery to remove the cancer is obviously the prime choice, but sometimes it is not possible because of the position of the cancer or the part of the brain involved. Irradiation and cancer-killing drugs (cytotoxics) are often used alone or in combination with surgery. Steroids are often given to reduce the swelling of the brain that occurs around the tumour. The result of treatment varies dramatically between patients.

Brain cancers include gliomas, astrocytomas, medulloblastomas, ependymomas, pinealomas, neurinomas (neuromas) and menigiomas.

See also ASTROCYTOMA; CRANIOPHARYNGIOMA; DIENCEPHALIC SYNDROME; EPENDYMOMA; GLIOMA; MEDULLOBLASTOMA; MENINGIOMA; PINEALOMA
BROWN-SÉQUARD SYNDROME

The Brown-Séquard syndrome is an uncommon spinal cord condition caused by injury, transverse myelitis, overgrowth of arteries (haemangioma), tumours or compression affecting the spinal cord. The symptoms include muscle spasm and loss of position sense on one side of the body, loss of pain and temperature sense on the opposite side, with light touch sensation preserved and muscle strength lost on both sides. The cause is treated if possible, but often there is no cure.

The condition is named after the French physiologist Charles-Edouard Brown-Séquard (1817-1894).

See also TRANSVERSE MYELITIS

BRUCELLOSIS

Undulant fever or brucellosis is a bacterial infection of cattle, goats and pigs, which can spread to man, and most commonly infects meat workers, veterinarians and farmers. The bacteria Brucella abortus, Brucella melitensis or other species of Brucella, enter a human through a cut or graze in the skin, or is swallowed. It is found in raw meat and unprocessed milk.

Patients initially experience a fever, tiredness and intermittent sweats. After several weeks, further symptoms of headache, swollen painful joints, loss of appetite and abdominal pains (from a large spleen and/or liver) develop. The fever may come and go for many months in a low-grade chronic form of the disease (thus undulant fever).

Specific antibody blood tests are used to make the diagnosis.

Treatment involves taking antibiotics (eg. tetracycline) and rest until all symptoms have settled. Animals may be vaccinated to prevent them from catching the disease, but not humans. Occasionally the infection may spread to involve the lung, brain and heart, causing specific problems in those areas. Long-term complications include arthritis, and bone weakness.

There is a good response to treatment, but symptoms may recur over several years and require further courses of treatment.

CARBON MONOXIDE POISONING

Poisoning can occur by inhalation of carbon monoxide (CO), which is a colourless and odourless gas produced by the burning of carbon containing material. Carbon monoxide binds to haemoglobin in the blood to form carboxyhaemoglobin, which prevents the haemoglobin from carrying oxygen to all tissues in the body.

CO may be inhaled by accidental or deliberate (suicide) inhalation of automobile exhaust gas, smoke inhalation in a fire, or unvented combustion heater.

Initially symptoms include headache, dizziness, belly pain, nausea and shortness of breath. Later symptoms include red skin, confusion, fainting, coma, convulsions and death. Permanent brain damage is possible in survivors of severe poisoning. The level of carbon monoxide in the blood can be measured to determine the severity of poisoning.

The patient should be immediately removed from the gas source, and artificial respiration should be applied if necessary. In hospital or an ambulance, oxygen 100% is given using a close fitting mask to flush the carbon monoxide out of the blood. The prognosis depends on the level of exposure, but is usually good if the patient is resuscitated without coma or convulsions occurring.

CARDIAC FAILURE

Heart failure (cardiac failure) occurs when the heart is no longer capable of pumping blood around the body effectively. Many conditions can cause heart failure including heart attacks (which damage the heart muscle), endocarditis (heart infection), narrowing or leaking of heart valves, high blood pressure, narrowing of the aorta (aortic stenosis), irregular heart rhythm, alcoholic heart damage, severe anaemia (inadequate oxygen being transported by the blood) and an overactive thyroid gland (increased rate of body activity results in greater demands for blood).

Patients with cardiac failure complain of being short of breath when exercising or climbing stairs, but as the condition worsens they are constantly out of breath, particularly when lying down at night. Other symptoms include a hard dry cough, having to get out of bed to pass urine at night, general tiredness and weakness, a rapid heart rate, chest and abdominal discomfort and swelling of the feet, ankles and hands. These patients may...
be noticed by others to be losing weight, unable to speak a full sentence without taking a breath, and in advanced cases a blue tinge develops on and around the lips.

The diagnosis can often be made by a doctor without resorting to any sophisticated tests, but it is important to discover the cause of the heart failure, particularly in younger patients, and this may involve extensive investigations over a considerable period. Blood tests (e.g. brain natriuretic peptide), chest X-rays and electrocardiograms are the main tests performed. Further tests may be indicated after the results of these are obtained. Echocardiograms and cardiac catheterisation (passing a tube through a vein into the heart) are sometimes undertaken if surgical treatment of a heart defect is being contemplated.

Treatment involves correction of any specific cause for the heart failure if possible. If the thyroid is overactive or the patient is anaemic, these can be treated and the heart failure may disappear. Correction of high blood pressure, controlling an irregular heart rhythm and treating heart infections are other methods of dealing with a specific cause of heart failure. Sometimes surgical correction of a heart valve deformity is also possible.

A diet low in salt, and avoiding strenuous exercise can often be beneficial. Medications to remove excess fluid from the body (diuretics) and to strengthen the action of the heart (e.g. digoxin, ACE inhibitors, beta-blockers, proscillaridin) are in common use. Digoxin is derived from digitalis, which in turn is found in the foxglove plant. The foxglove has been used as a folk remedy for heart disease since the Middle Ages. A number of more sophisticated drugs are available for use in resistant or difficult cases. Oxygen may be supplied to seriously ill patients.

In many elderly patients, there can be a multitude of causes, or no specific cause at all for the heart failure. In these cases, the condition is treated as a disease in itself.

Unless an underlying correctable cause can be found, heart failure cannot be cured, only controlled. As the years pass, the condition usually slowly worsens and becomes steadily harder to control. Some patients succumb to heart attacks, while others become more and more incapacitated so that they cannot leave a bed, and develop pneumonia. The actual outcome in any individual is very difficult to predict, but it may take many years before serious incapacitation or death occurs.

**CARDIAC TAMPONADE**

The heart is contained within a firm fibrous sac, the pericardium. If fluid (e.g. blood) enters the pericardial sac and gradually increases in volume, it puts pressure on the heart, preventing the ventricles (main chambers of the heart) from filling with blood between each contraction. The heart starts to beat inefficiently, resulting in a poor blood supply to the rest of the body. This condition is called cardiac tamponade.

Common causes include pericarditis (inflammation of the pericardium), bleeding into the pericardium during heart surgery, uraemia (kidney failure) and a cancer of the heart or pericardium. It may also be a side effect of anticoagulant (e.g. warfarin) overdose, and investigative procedures (e.g. biopsy) on the heart or pericardium.

Cardiac tamponade is a very serious condition that may be rapidly fatal if not adequately treated. Its symptoms include a low and falling blood pressure, increasing venous blood pressure (jugular venous pressure), pulsus paradoxus (a pulse that becomes weaker while taking a deep breath), shortness of breath and faint heart sounds. Often these signs come on very slowly and subtly and are difficult to detect.

The diagnosis is confirmed by echocardiography (ultrasound of the heart), and treatment involves surgical drainage of the fluid from inside the pericardium, usually by passing a catheter into the accumulated fluid. The cause of the cardiac tamponade must also be treated.

**CAROTID SINUS SYNDROME**

Carotid sinus syndrome is a disease affecting the carotid body, a tiny pressure sensitive structure at both sides of the front of the neck at the point where the main artery to the head (carotid artery) divides into internal and external branches. Excessive sensitivity of the carotid body results in inappropriate nerve signals being sent to the heart and blood vessels. It is more common in the elderly.

Symptoms include low blood pressure, slow heart rate, dizziness and fainting. Massaging the carotid body can trigger the symptoms.

A pacemaker is inserted to regulate heart rate, and most, but not all, patients are controlled by this treatment. Many are not diagnosed until serious falls from fainting have occurred.

**CAROTIDYNIA**

Carotidynia is a syndrome characterised by blood vessel spasm and inflammation that affects one side of the face and neck. It is a migraine-like reaction y the carotid artery in the neck. Those affected experience discomfort
to intense pain in and behind angle of the jaw that spreads across the face and temple, tender carotid artery in
the neck, stiff neck muscles and swelling of the neck (oedema). Some patients develop a runny nose, Horner
syndrome (drooping eyelid, contracted pupil and a sunken eye, associated with reduced sweating) and
hoarseness.

There is no diagnostic test, but blood tests and x-rays need to be done to exclude other possible causes of
symptoms (eg. temporal arteritis, dental abscess, jaw joint arthritis).

Medications such as pizotifen and propranolol can prevent most attacks. Acute attacks settle spontaneously
in a few hours to days.

**CATATONIC SYNDROME**

Catatonia (catatonic syndrome) is a psychiatric or neurological (brain damage) disorder that affects muscle
control. It is usually associated with schizophrenia, but may be due to brain tumours or inflammation, strokes,
drugs, poisons and body chemistry disorders.

Patients develop increased muscle tone at rest that disappears during movement, sudden impulsive
movements and excitement. Multiple investigations (eg. blood tests, CT scan) must be carried out to determine
the cause. Treatment involves hospitalisation, medications (eg. haloperidol, fluphenazine, thiothixene),
psychotherapy, behavioural and social therapy.

See also SCHIZOPHRENIA

**CAVERNOUS SINUS SYNDROME**

A clot (thrombus) forming in the cavernous sinus causes the cavernous sinus syndrome. This is
characterised by swelling of the whites of the eye (conjunctival oedema), the upper eyelid and base of the nose,
and paralysis of some eye movements. The thrombosis is usually due to an infection spreading into the
cavernous sinus from the nose or skin of the central face. Treatment involves anticoagulants and antibiotics, but
the prognosis is often poor.

**CEREBRAL HAEMORRHAGE**

A cerebral haemorrhage is a bleed into the brain from a head injury or a ruptured artery (eg. cerebral
aneurysm) or vein. the result is indistinguishable from a stroke except by a CT or MRI scan. The haemorrhage
may occur within the brain, or between the various layers of the meninges that surround the brain. The
symptoms can vary dramatically depending on the site and amount of bleeding. Surgery may be necessary to
control continued bleeding within the brain. Otherwise treatment is as for a stroke.

See also CEREBROVASCULAR ACCIDENT; SUBARACHNOID HAEMORRHAGE; SUBDURAL
HAEMATOMA

**CEREBRAL PALSY**

Cerebral palsy (spasticity) is a brain condition causing abnormal uncontrolled muscle spasms. It is usually
due to abnormal development of the brain before birth, viral infections in the mother during pregnancy, a poor
blood supply from the placenta during pregnancy, very premature birth or uncommonly to brain damage around
the time of birth because the baby is deprived of oxygen for several minutes. About two in every thousand births
result in a child with cerebral palsy.

Symptoms vary dramatically from one patient to another depending on the area of brain damaged. Some
have slight difficulty in controlling one limb, others may be unable to talk clearly, yet others may be totally unable
to care for themselves in any way. Mental functioning may be completely normal, or there may be significant
intellectual disability.

Electroencephalograms (EEG) which measure brain waves, electromyelograms (EMG) which measure the
electrical conduction in muscles, and CT scans of the brain and spine may be abnormal, but blood tests are
usually normal.

Most help comes from teams of nurses, physiotherapists, occupational therapists, social workers and
volunteers. Medications may be used to treat skin, intestinal and arthritic complications. Operations to correct
defeormities and release spasm in limbs can complement medications, which reduce the uncontrollable twitching
that may occur. Paramedical staff can teach the patient how to control an unwilling body, and emotional and
psychiatric support is often necessary.
Cerebral palsy is often associated with epilepsy due to brain damage, and there is a high incidence of arthritis, pressure ulcers, chest infections, peptic ulcers, and emotional and social problems.

Although there is no cure, some symptoms can be eased, but life expectancy is slightly less than average.

**CEREBROVASCULAR ACCIDENT**

A stroke is an accident involving the blood vessels in the brain, and is technically known as a cerebral infarct or cerebrovascular accident (CVA). If a clot, or piece of material from elsewhere in the body, blocks an artery in the brain (cerebral thrombosis), or if an artery bursts in the brain, a stroke may occur. The risk of stroke is higher in those who smoke, have high blood pressure, high cholesterol levels, are diabetic, and drink alcohol to excess.

Any blood vessel in the brain may be involved, so any part of the brain may be damaged, and the area damaged determines the effects on that person's body. The symptoms can therefore be very varied. If a motor area of the brain, which controls movement is affected, the patient becomes paralysed down the opposite side of the body because the nerves supplying the body cross over to the opposite side at the base of the brain (the right side of the brain controls the left arm and leg). Other patients may lose their memory, power of speech, become uncoordinated, unbalanced, start fitting, have strange smells, hear abnormal noises or any of dozens of other possibilities. The area of the brain affected may increase as a blood clot extends along an artery, or bleeding into the brain continues.

The cause of the stroke can be determined by using special X-rays, CT scans, MRI (magnetic resonance imaging), blood tests, tests on the fluid around the brain, and measuring the brain waves electrically (EEG).

A wait-and-watch attitude is adopted in most cases, with medication given to prevent the stroke from worsening and to protect other organs. Surgery to a bleeding or blocked artery in the brain may be appropriate in some cases. Physiotherapists, speech pathologists and occupational therapists will assist in recovery. Further strokes can often be prevented by the long-term use of low-dose aspirin or warfarin, which prevent blood clots. Patients who are at a high risk can also use these medications.

It will be several days or even weeks before doctors can give an accurate prognosis. The brain does not repair itself, but it can often find different ways of doing a task and bypassing damaged areas. Most improvement occurs in the first week, but full recovery may take months. Patients who become unconscious during a stroke generally have a poorer outcome than those who do not. Strokes are the third major cause of death in developed countries after heart disease and cancer.

See also **TRANSIENT ISCHAEMIC ATTACK**

**CFS**

See **CHRONIC FATIGUE SYNDROME**

**CHARCOT-MARIE-TOOTH DISEASE**

Charcot-Marie-Tooth disease is a gradually progressive degeneration of nerves that supply the arms and legs, caused by a familial (runs in families) genetic abnormality. Patients initially have an abnormal gait (way of walking) and foot deformities in late childhood or early adult life. The gait gradual worsens over several years with weakness and loss of sensation in the legs, and later the arms. Paralysis of both arms and legs may be the final result. The diagnosis is confirmed by a nerve biopsy. There is no treatment and no cure.

**CHARLES BONNET SYNDROME**

The Charles Bonnet syndrome is a psychological disturbance associated with isolation. It is often found in elderly widows with poor vision and depression who are deprived of any company or outside contact. Those affected develop vivid, elaborate and recurrent hallucinations. Hallucinations of smell, taste and sounds may also occur.

Psychological counselling and support, and psychiatric treatments are given, but most patients end up in an institution.

See also **DEPRESSION; PSYCHOSES**

**CHINESE RESTAURANT SYNDROME**

The Chinese restaurant syndrome is an abnormal reaction to food preservatives caused by the overuse of monosodium glutamate in food and aggravated by alcohol. Victims develop facial pressure, a headache, nausea,
chest pain and a burning sensation of the head and chest. It settles spontaneously after 12 to 48 hours, while the symptoms may be eased by aspirin or paracetamol.

CHOREA
Chorea is the presence in a person of uncoordinated, purposeless, rapid jerky movements. It particularly affects the hands and face, but any part of the body may be involved. It is a symptom of a degeneration of the brain that may occur with Huntington’s chorea, Sydenham’s chorea or a very rare complication of pregnancy (chorea gravidarum).

The term is derived from the Greek word for dance, *choreia*.
See also HUNTINGTON’S CHOREA; SYDENHAM’S CHOREA

CHRONIC FATIGUE SYNDROME
Chronic fatigue syndrome (CSF) has been given many different names in various places, including myalgic encephalomyelitis (ME), postviral syndrome, Royal Free disease (named after a London hospital), and Tapanui flu (in New Zealand). It is characterised by a persistent tiredness and easy fatigue that persists for many months for no obvious reason.

The condition is may be caused by a virus, but some patients find that certain foods aggravate the condition. It is possible that it is actually several diseases that overlap with their symptoms, and may be due to a combination of infection, immune deficiencies, autoimmune type condition (where the body rejects its own tissue), chronic inflammation, stress and psychiatric disturbances. It is a matter of debate whether the distressing symptoms cause the psychological problems, or vice versa.

The diagnosis can only be confirmed if in the following list both major criteria are met, plus six symptoms and two signs from the minor criteria

**MAJOR CRITERIA**
- New persistent or intermittent, debilitating fatigue severe enough to reduce or impair average daily activity below 50% of normal activity for a period of more than 6 months
- Exclusion of all other causes by thorough clinical evaluation, and blood tests

**MINOR CRITERIA - SYMPTOMS**
- Generalised fatigue lasting more than 24 hours following levels of exertion that would have been easily tolerated previously
- Vague headache
- Unexplained general muscle weakness
- Muscle pains
- Arthritis that moves from joint to joint without any apparent damage to the joint
- One or more of the following problems :-
  - avoidance of bright lights
  - forgetfulness
  - irritability
  - confusion
  - poor concentration
  - depression
  - intermittent visual disturbances
  - difficulty thinking
- Inability to sleep, or excessive sleepiness
- Rapid onset over hours or days of major criteria

**MINOR CRITERIA - SIGNS - documented by a physician on at least two occasions at least a month apart**
- Mild fever greater than 38.6°C
- Sore throat with no pus present
- Tender enlarged lymph nodes in neck or arm pit

There are no specific diagnostic tests, but numerous blood tests may show minor abnormalities. Tests are always performed to exclude any other possible cause.

There is no specific treatment available, but patients can benefit by having an understanding doctor who may use antidepressants, anti-inflammatory medication, steroids and other drugs that may be helpful. Although there is no cure, with time, most cases slowly improve, but some patients are left with long term tiredness so severe that they are unable to return to work or undertake normal daily activities.
CIGUATERA POISONING

This is a form of seafood poisoning caused by eating reef fish that contain the ciguatera toxin. The fish itself is not affected, and there are no tests for differentiating safe from toxic fish. Generally the larger the fish, the more likely it is to be toxic. The poison is produced at certain seasons by a microscopic animal (Dinoflagellida) that proliferates on tropical reefs. This is eaten by very small fish, who are then eaten by bigger fish, who are then eaten by still bigger fish. There may be a dozen steps along this chain, with the poison being steadily concentrated in the fish tissue at every step. Ciguatera is present in a low concentration in most reef fish, but only when it exceeds a certain concentration does it cause problems in humans. There are far higher concentrations in the gut, liver, head and roe of reef fish, which should never be eaten or used to make fish soup. The toxin cannot be destroyed by heat or cooking.

Symptoms vary dramatically from one patient to another, depending on the amount of toxin eaten, the size of the victim, and the individual reaction. They may include unusual skin sensations and tingling, diarrhoea, nausea, abnormal sensation, headaches and irregular heartbeats. Unusual tingling sensations may persist for years, and subsequent serious attacks may be triggered by eating tiny amounts of ciguatera that may be present in fish that others can eat without adverse effects.

There are no diagnostic tests, and no specific treatment or antidote, but medication may be used to control symptoms.

Patients with a mild reaction usually recover in a few days as the toxin is naturally eliminated from the body, but severe attacks may cause symptoms for a couple of months. Death is rare, but possible, usually occurs within 36 hours of the onset of the attack, and is caused by the effects of the toxin on the heart and blood vessels.

CIRRHOSIS

Cirrhosis is a slowly developing form of damage to the liver, which results in it becoming hard and enlarged, as normal tissue is replaced by fibrous scar tissue. There are many possible causes including recurrent attacks of hepatitis A, one attack of hepatitis B, other liver infections, excess alcohol intake, gall stones, a number of rare diseases that affect the liver (eg. haemochromatosis, Wilson's disease, Gaucher's disease), toxins, poisons (eg. arsenic) and drugs (eg. methotrexate, isoniazid). The diagnosis is confirmed by blood tests, and ultrasound and/or CT scans of the liver.

The symptoms may include itchy skin without a rash, jaundice (yellow skin), diarrhoea and abdominal discomfort.

Other than a liver transplant, there is no cure for cirrhosis. Patients must stop all further alcohol intake to reduce further damage. Vitamin supplements and nutritious diets are recommended, and medication can be prescribed to slow the progress of the disease (eg. ursodeoxycholic acid) and ease some symptoms. The liver tends to become steadily more damaged until it ceases to function completely.

See also ALCOHOLISM

CJD

See CREUTZFELDT-JAKOB DISEASE

CLUSTER HEADACHE

A cluster headache is a severe, intermittent one-sided headache that occurs in clusters lasting from days to weeks. Attacks may be triggered by alcohol, stress, exercise, certain foods and glare. They are more common in middle-aged men.

Patients experience severe, one-sided pain around the eye that occurs daily for weeks and then subsides, only to flare again months later. The pain may be quite disabling, and are often accompanied by a congested nostril on the same side as the headache, a watery red eye and weakness on the affected side of the face. Unfortunately, there are no specific diagnostic tests available, and the diagnosis rests on the clinical acumen of the doctor.

Once present, these headaches are very difficult to control. Normally it is a matter of trial and error to determine the most effective treatment regime in any individual. The inhalation of pure oxygen may settle an otherwise intractable attack in a few minutes. Prevention is far better than cure, and medications such as
propranolol, ergotamine, lithium and amitriptyline can be used on a regular basis to prevent further attacks. In severe cases prednisone is prescribed.

See also MIGRAINE

**COCKAYNE SYNDROME**
The very rare Cockayne syndrome is a congenital condition characterised by dwarfism, blindness from failure of the retina to develop, deafness, intellectual disability, light sensitive skin and jaw abnormalities.

**COFFIN-LOWRY SYNDROME**
The Coffin-Lowry syndrome is a developmental abnormality affecting multiple organs that has sex-linked inheritance, and affects only boys, but females act as carriers.

Patients have prominent lips, coarse facial features, tapering fingers, reverse eye slant, intellectual disability and excess curvature of the spine (kyphosis and scoliosis).

There are no specific diagnostic tests, no treatment is available, and there is no cure.

**COFFIN-SIRIS SYNDROME**
The Coffin-Siris syndrome is a familial (runs in families) developmental abnormality in which both parents must be carriers. Patients have poorly developed toenails, are very hairy at birth but have sparse hair in later life, suffer intellectual disability and coarse facial features.

There are no specific diagnostic tests, and no treatment is available, but life expectancy is reasonable.

**COGAN SYNDROME**
Cogan syndrome is a rare ear, eye and other organ inflammatory condition with rapid onset in young adults. The cause is unknown.

Patients develop eye surface inflammation, ringing in the ears (tinnitus), dizziness and deafness. Some patients develop a fever, enlarged tender lymph nodes, joint aches and pains, belly pain, enlarged spleen, black faeces from blood leaking into the bowel and heart abnormalities (eg. aortic valve disease).

Non-specific blood tests showing inflammation are positive, but there is no specific diagnostic test.

Medications such as corticosteroids and cyclophosphamide are used in treatment, although the disease course is variable and self-limiting.

**COLLOID CYST**
A colloid filled cyst may form in the thyroid gland, where it may affect the function of the gland, or in the third ventricle (a fluid filled space) in the brain, which may cause hydrocephalus by blocking the circulation of cerebrospinal fluid.

See also HYDROCEPHALUS

**coma**
A coma is an unrousable loss of consciousness.

The severity and depth of a coma is measured by the Glasgow Coma Scale.

Many causes of a coma relate to the brain but other diseases and conditions may be responsible. A wide range of blood, urine, x-ray, CT and ultrasound scan, and cerebrospinal fluid (CSF- the fluid around the brain) tests will be performed in order to determine the cause.

The brain is suspended in cerebrospinal fluid inside the bony skull. Any blow to the head will make the brain rattle around inside the skull, and it may be bruised or damaged. A fractured skull may cause a laceration of the brain. This damage can cause unconsciousness that may persist for seconds, minutes, occasionally hours and rarely for months.

In a stroke (cerebrovascular accident) various parts of the brain may be affected by having its blood supply cut off by a blockage in an artery, or a blood vessel in the brain may burst causing bleeding and damage to part of the brain. The onset is almost instantaneous, may be associated with a wide variety of symptoms from paralysis and headache to weakness, loss of sensation, anaesthesia, confusion and coma. Tumours, cancers, an abscess or cyst in the brain can have a similar effect to a stroke.

The brain is supported and completely surrounded by a three layered membrane (the meninges) which contain the cerebrospinal fluid. If these meninges are infected by a virus or bacteria (meningitis) the patient may
experience headache, fever, fits, neck stiffness and in severe cases may become comatose. Encephalitis is an infection of the brain that may be confused with meningitis. The symptoms include headache, intolerance of bright lights, fever, stiff neck, lethargy, nausea, vomiting, sore throat, tremors, confusion, convulsions, stiffness and paralysis. This can progress to coma, and sometimes to death.

If too much cerebrospinal fluid is produced, or insufficient is absorbed, the pressure of this fluid in and around the brain will gradually increase (hydrocephalus). The resultant pressure on the brain will affect its function and result in headaches, personality changes, memory loss, reduced intelligence, convulsions and coma.

Epilepsy is a condition that causes recurrent seizures (fits). Fits can vary from very mild absences in which people just seem to lose concentration for a few seconds, to uncontrolled bizarre movements of an arm or leg, to the grand mal convulsion in which an epileptic can thrash around quite violently and lose control of bladder and bowel. A prolonged period of confusion or coma may follow a fit.

Degeneration of the brain may occur steadily in various forms of dementia, including Alzheimer disease. Confusion, forgetfulness, poor coordination, incontinence and irritability will gradually worsen. The final stage of these degenerative conditions is coma.

Patients with an intense form of hysteria as a reaction to severe fright, fear, shock or some psychiatric conditions, may appear to be in a coma, and act completely as though they are unconscious, although there is no physical cause.

Overdoses of alcohol, illegal drugs (eg. heroin) and many different medications may cause a life threatening coma. Numerous poisons, attempted suicide with a car exhaust (carbon monoxide poisoning), snake and venomous spider bites may also cause a loss of consciousness.

Dehydration from lack of water or excessive sweating in a hot climate, or due to severe diarrhoea with cholera or other bowel infections, may have coma as an end result.

Very low blood pressure (hypotension) caused by loss of blood (eg. into the body or from a deep wound), heart attack or other serious disease, will lead to collapse and coma that may lead to death.

An irregular heart beat (palpitation) may prevent sufficient blood from reaching the brain.

Diabetes is caused by a lack of insulin production in the pancreas (type 1 or juvenile diabetes), or is due to the cells of the body becoming resistant to insulin and preventing it from transporting sugar from the blood and into the cell (type 2 or maturity onset diabetes). The early symptoms are unusual tiredness, increased thirst and hunger, excess passing of urine, weight loss despite a large food intake, itchy rashes, recurrent vaginal thrush infections, pins and needles and blurred vision. An excess of insulin or diabetic tablets can lower blood sugar levels excessively (hypoglycaemia) to cause a coma. Conversely, uncontrolled diabetes may cause a coma due to excessive blood sugar levels (hyperglycaemia).

Severe bacterial infections, particularly septicaemia when the blood itself is infected, can cause a persistent high fever that affects the brain and other organs, leading to confusion, convulsions and coma.

Less common causes of coma include suffocation, lack of oxygen at high altitude, a near drowning experience, severe asthma, pneumothorax, pneumonia, heart failure, heart attack, failure of the liver or kidney, severe hypothyroidism (underactive thyroid gland), severe allergy reaction (anaphylaxis), very high blood pressure (malignant hypertension), serious tropical diseases that have associated high fevers (eg. typhoid fever, yellow fever) and Addison disease.

A coma is always serious and needs immediate medical treatment, but correct treatment will depend on identifying the cause. The prognosis depends on the cause and is very unpredictable. Some comas last for years while others may be quite brief.

See also CONCUSSION; FAINT

CONCUSSION

Concussion is due to bruising of part of the brain, from a moderate to severe blow on the head (generally at the back) or a severe shake of the body. Symptoms can vary in severity from mere giddiness and a headache for an hour or two, to a complete loss of consciousness, sometimes lasting for weeks. The range of symptoms includes temporary, partial or complete loss of consciousness, “seeing stars”, shallow breathing, nausea and vomiting, paleness, coldness and clamminess of the skin, blurred or double vision, and possibly loss of memory.

A skull x-ray and CT scan may be performed to exclude fracture or other complications, but do not specifically diagnose concussion.

The first-aider should lie the patient down, keep them warm and comfortable, apply cold compresses applied to the brow or the site of injury, and not give anything to eat or drink for the first few hours after the injury. Medical attention should be sought. Paracetamol may be used for pain, but aspirin should be avoided. Keep the victim under observation for at least 24 hours for signs of more serious injury.
There may be slow bleeding into the brain, which can cause problems hours or days later. The symptoms are a worsening headache, continued vomiting, drowsiness, stupor, deliriousness or other mental changes, collapse, fits, blackouts, giddiness, clear or bloodstained fluid draining from the nose or ears. If any of these symptoms occur, get medical advice immediately.

In most cases, complete recovery within a few hours or days is normal. See also COMA; PUNCH DRUNK SYNDROME; SUBDURAL HAEMATOMA

**CONN SYNDROME**

Conn syndrome is also known as aldosteronism and hyperaldosteronism. It is a rare disease due to overactivity of the adrenal glands, which sit on top of each kidney.

A tumour in one of the adrenal glands, or other even rarer diseases, causes excessive amounts of the hormone aldosterone to be produced by the gland. Aldosterone controls the amount of salt in the body. If the level of salt in the blood drops, more aldosterone is secreted by the adrenal gland, and it acts on the kidney to reduce the amount being lost in the urine. The tumour is not a cancer, but the increased aldosterone production causes excess salt to be retained in the body.

Excess salt causes high blood pressure, increased urine production, muscle weakness, pins and needles sensations, headache and thirst. The syndrome may be the cause of high blood pressure that does not respond to normal treatments.

The diagnosis is confirmed by blood tests, and a CT or MRI scan. Radioactive substances that concentrate in the abnormal adrenal gland may also be given to a patient in whom the disease is suspected, and the degree of concentration of the substance in each adrenal gland can then be measured.

The tumour of the adrenal gland can be removed surgically, but medications may be required to control the symptoms and high blood pressure before the operation. Most cases can be cured by surgery, while the others can be controlled by medication (eg. trilostane).

**CONRAD SYNDROME**

The Conrad syndrome is a congenital developmental abnormality. It varied symptoms include cataracts in the eyes, limb contractures, deafness and intellectual disability. Abnormal long bone ends (stippled epiphyses) are seen on x-ray. Surgery can be performed for the cataract and limb contractures. Although there is no cure, life expectancy is reasonable. See also INTELLECTUAL DISABILITY

**CORI SYNDROME**

See GLYCOGEN STORAGE DISEASES

**COR PULMONALE**

Cor pulmonale is also known as pulmonary hypertension or right heart failure. It results in enlargement of the right side of the heart (which pumps blood through the lungs), and increased blood pressure in the lungs. It is primarily a sign of obstructed circulation of blood through the lungs, and not heart disease, although congenital heart disease and left heart disease must be excluded.

Lungs damaged by emphysema, smoking, inhaled coal dust or asbestos, recurrent lung infections or a number of rarer lung diseases may be so abnormal that the blood has difficulty in passing through them. The right side of the heart must work harder to force the blood through the damaged lungs, which causes a significant rise in the blood pressure in the right heart and lungs. This causes further damage to arteries and worsens the disease. The heart muscle thickens and enlarges, and because of the lung damage, inadequate oxygen enters the blood, which further compounds the problem.

Patients have a cough that produces clear or bloodstained phlegm, a wheeze, shortness of breath with any exertion and general weakness. In advanced cases the ankles may be swollen, nausea and indigestion may occur, and the liver enlarges. Patients are more susceptible to lung infections such as bronchitis and pneumonia.

The diagnosis can be made by a chest X-ray and an electrocardiogram (ECG). Other investigations include cardiac catheterisation (passing a tube through a vein into the heart to measure the blood pressure), echocardiography and angiography.
Medication (eg. bosentan) can be prescribed to strengthen the heart, open the lungs and cure any lung infection. Physiotherapy can help drain phlegm from the lungs, and oxygen may be used to relieve the shortness of breath.

Unfortunately, no cure is possible and patients steadily deteriorate over many years to eventually die from heart attacks, pneumonia or other complications of the disease.

See also CARDIAC FAILURE

CRANIOPHARYNGIOMA

Craniopharyngiomas are uncommon slowly growing brain tumours that occur in the centre of the brain, and put pressure on the optic (vision) nerve. They are more common in children and young adults, but their cause is unknown.

Patients develop partial or total blindness in one or both eyes, headache, vomiting, personality and mental changes. Short stature, failure of puberty in children and other hormonal abnormalities may occur due to pressure on the pituitary gland. It can be diagnosed by a CT or MRI scan.

Treatment involves irradiation and sometimes surgery, but is generally unsatisfactory.

CRANIOSTENOSIS

Craniostenosis is a congenital deformity of the head in which the skull fails to grow and expand due to premature fusion of the joints (sutures) between the bones that make up the skull. This results in brain damage, intellectual disability, convulsions and a small, deformed skull. The closed sutures can be seen on x-ray.

Sometimes surgical splitting of sutures is attempted, but generally no treatment is available and life expectancy is significantly shortened.

CRETINISM

Cretinism is hypothyroidism (under active thyroid gland) in a child, a condition that occurs in one in every 4000 births.

The thyroid gland in the front of the neck is responsible for producing a substance called thyroxine, which acts on every cell in the body to control the rate at which it works. In cretinism, the thyroid gland fails to function correctly from birth to cause impaired brain development and intellectual disability (which may be severe). It is diagnosed by specific blood tests that are routinely performed on all babies at birth.

Thyroxine tablets or mixture control the problem, but there may be some degree of brain damage due to lack of thyroxine in the foetus before birth. Most cretins can function normally in society with their intelligence and functional capacity only slightly below average. No further deterioration occurs once treatment is started.

See also HYPOTHYROIDISM

CREUTZFELDT-JAKOB DISEASE

Creutzfeldt-Jakob disease (CJD) or spongieform encephalitis is a prion (virus like) infection of the brain.

Rare spontaneous cases have occurred where the source of infection is unknown, but in more recent times it has been spread by the use of growth hormone extracted from the pituitary gland of corpses, and given to children who have inadequate growth. Rarely, transmission has also occurred through some corneal and meninges organ transplants. In the United Kingdom during the late 1980s and early 1990s, cattle were fed a protein supplement that included the ground up carcasses of sheep. Some of these sheep suffered from a disease called scrapie, which was caused by the same prion as CJD. These cattle developed mad cow disease, and when slaughtered for consumption by humans, passed on the prion to cause CJD.

There may be no symptoms for years after the prion enters the body, before it activates and attacks the brain. Progressive dementia, tremors, incoordination, drowsiness, emotional instability and speech difficulties then occur.

The diagnosis is difficult and no treatment is available. The infection leads inevitably to death, often within a year of diagnosis.

The condition is named after the German psychiatrists Hans Creutzfeldt (1885-1964) and Alfons Jakob (1884-1931).
Cri du chat syndrome is a rare congenital brain development abnormality that causes intellectual disability, wide apart eyes, a small head, round face, low set ears, and a catlike cry (“cri du chat”) as a newborn. The cat cry is due to a temporary poor development of the larynx that corrects itself with age. Diagnostic chromosome studies show deletion of the short arm of the 5th chromosome. There is no treatment or cure.

See also INTELLECTUAL DISABILITY

The Crouzon syndrome is a familial (runs in families) developmental abnormality of the face. Those affected have abnormal growth and shape of their face and skull, protruding eyes, a squint and loss of vision in one or both eyes. From the side, the face appears flattened. A skull X-ray is abnormal. There is no cure, but reasonable results are obtained from major facial surgery, depending on the severity of deformities.

Cushing syndrome is also known as adrenocortical hyperfunction and hyperadrenocorticism. It is a syndrome resulting from excessive amounts of steroids in the blood.

The hypothalamus is the part of the brain that decides how much natural steroid is required. It sends nerve messages to the pituitary gland, which sits under the centre of the brain and it in turn sends a chemical message to the adrenal glands that sit on top of each kidney. The adrenals produce the steroids required by the body. Tumours or overactivity (may be triggered by pregnancy or stress) in the hypothalamus, pituitary gland or adrenal gland can result in the overproduction of steroids.

Cushing syndrome may also be due to taking excessive amounts of steroids (eg. prednisone) for medical reasons.

Patients develop a fat face (moon face), fatty deposits on the upper back (called a buffalo hump), obesity of the abdomen and chest with thin arms and legs, high blood pressure, impotence, cessation of menstrual periods, skin infections and pimples, headaches, backache, excess hair growth on the face and body, mood changes, excessive bruising, thinning of the bones (osteoarthritis - which can cause bones to fracture easily), stretch marks on the breasts and abdomen, kidney stones, and generalised weakness. Strokes, heart attacks, broken bones, diabetes, increased susceptibility to infections (particularly of the skin and urine), and psychiatric diseases may be complications.

Complex blood and urine tests can confirm the diagnosis, but finding the cause of the syndrome can be very difficult, and CT scans and magnetic resonance imaging (MRI) may be used to find very small tumours.

If a tumour can be found in the adrenal or pituitary gland, it is surgically removed. Other treatments include irradiation of the pituitary gland, or removal of both adrenal glands. Drug treatment is generally unsuccessful, but if both adrenal glands are removed it is necessary to supply steroids and other hormones by taking tablets or having injections regularly. If the Cushing syndrome is due to taking steroid medication, the dosage of this should be reduced if possible. Nelson syndrome (skin and tongue pigmentation, and enlargement of the pituitary gland under the brain) is a complication of treatment.

The prognosis depends on the cause. Some tumours of the adrenal or pituitary glands are very aggressive and spread to other areas to continue the syndrome, and these patients have a poor life expectancy. In others, a lifelong cure may be obtained by removing a localised tumour. If caused by excessive steroid medication, the syndrome is cured by stopping the medication, but the patient may require the steroids for control of asthma, rheumatoid arthritis or other diseases, and they must tread a very narrow path between the side effects of the medication and the necessary treatment of a disease.

Harvey Cushing (1869-1939) was an American neurosurgeon and physiologist.

See also HYPOPITUITARISM

The Dandy syndrome is a very rare abnormality of the inner ear caused by damage to the vestibular apparatus (balance and position mechanism) in both inner ears. The patient suffers a total loss of balance,
dizziness and the horizon bounces up and down as the patient walks. A CT or MRI scan of inner ear may be abnormal.

No treatment or cure is available but the dizziness may be eased by medication.

**DANDY-WALKER SYNDROME**

The Dandy-Walker syndrome is a rare brain developmental abnormality, in which there is failure of the central portion of the cerebellum (lower back part of brain) to develop.

Children with the condition have a very large head, vomiting, irritability, poor head control, cleft palate, and abnormal side-to-side eye movements (nystagmus). Some patients have an abnormal way of walking (gait), headaches, multiple cysts in the kidneys, abnormal lumbar vertebrae, subnormal mentality and delayed muscle control.

MRI and CT scans of the skull are abnormal and can be used to confirm the diagnosis.

Brain surgery may prevent further deterioration of symptoms, but there is no cure.

**de CLÉRAMBAULT SYNDROME**

Also known as erotomania, de Clérambault syndrome takes the form of a fixed single-minded psychiatric delusion (monomania). Patients usually have an inadequate dependent personality, often after a period of real dependency on a person (eg. doctor during pregnancy and labour). They have a fixed delusional conviction that another is in love with them despite minimal contact. The majority of patients are female and may persecute or stalk the victim, who is often a doctor, film star, sportsman or other famous person.

There are no diagnostic tests, and the condition is diagnosed by psychiatric assessment.

Psychoanalysis, and drugs such as phenothiazines and clomipramine are used in treatment.

Legal action may need to be taken to stop stalking, and the person who is the object of affection is sometimes attacked and harmed if advances are rejected.

Treatment is extremely difficult and often unsuccessful, and certification is occasionally necessary.

**DEHYDRATION**

Dehydration is a lack of water in the body. As the human body is almost 70% (7/10ths) water, even a small drop in the total amount of water in the body can have significant effects.

Patients who lose less than 5% (1/20th) of their body water will feel thirsty, have a dry mouth, but few other symptoms.

More severe dehydration resulting in a loss of 5% to 10% (1/10th) of the body water will cause sunken eyes, loose skin (reduced turgor), rapid heart rate, minimal passing of urine, and depression of the soft spot at the front of a baby’s skull.

Dehydration in excess of 10% may be life threatening, particularly in children. Symptoms include altered mood, poor concentration, drowsiness, irritability, weak pulse, cold white hands and feet, loose folds of skin and eventually loss of consciousness.

Dehydration may be caused by loss of fluid in diarrhoea, copious vomiting, excessive sweating (eg. exercise, heat) which also causes a loss of sodium in salt, passing excess urine (eg. taking too many fluid tablets, diabetes insipidus and other diseases); or by lack of fluid intake, usually when fluids are not readily available.

Blood tests can accurately determine the degree of dehydration.

Treatment involves giving a solution of water and electrolytes (vital elements) by mouth if possible, or intravenously. In an emergency, a mixture containing a level teaspoon of salt and eight level teaspoons of sugar or glucose into a litre of boiled water may be given by mouth. Plain water should not be given as it will pass straight through the body. Because of their lower body weight, children will dehydrate far more rapidly than adults.

A decrease of 5% in water volume can cause significant disease, and a 10% loss may be fatal in children. Fortunately there is a very good response to correct treatment.

See also HEAT STROKE

**de LANGE SYNDROME**

The de Lange syndrome is an uncommon congenital cause of dwarfism that is also known as Amsterdam dwarfism, and Brachmann-de Lange syndrome.
These children have a small head, severe intellectual disability, bushy eyebrows that meet in centre, low birth weight and failure to thrive, low hair line, and excess hair on the skin. There are no specific diagnostic tests and no treatment is available. Patients rarely survive beyond 10 years.

It is named after the Dutch paediatrician Cornelia de Lange (1871-1950).

DEMENTIA

Dementia is an mental disorder in which the patient develops confusion, wanders aimlessly, irrational behaviour, inappropriate reactions, poor or jumbled speech patterns, hallucinations (both visual and auditory), and loss of short term memory. Some patients become uninhibited in their language and habits, and may act in a socially unacceptable manner. Symptoms are often worse at night. It is a permanent condition, as opposed to confusion, which may be temporary.

By far the most common cause of dementia in the elderly is Alzheimer disease (senile dementia or second childhood), but unfortunately it may strike as early as the mid-fifties and cause extreme distress to spouses, family and friends. It is characterised by loss of recent memory, loss of initiative, reduced physical activity, confusion, loss of orientation (patients become confused about where they are and dates), and then it gradually progresses to loss of speech, difficulty in swallowing (drooling results), stiff muscles, incontinence of both faeces and urine, and a bedridden state in which the patient is totally unaware of themselves or anything that is happening around them. It is caused by a faster than normal loss of nerve cells in the brain.

Damage to the brain from an injury (eg. fractured skull), lack of oxygen (eg. near drowning), essential surgery, abscess, tumour or cancer may affect brain function to cause dementia at any age.

In a stroke (cerebrovascular accident) part of the brain is affected by having its blood supply cut off by a blockage in an artery, or a blood vessel in the brain may burst causing bleeding and damage to part of the brain including dementia of sudden onset. Multi-infarct dementia occurs if multiple small strokes occur.

The brain is supported and completely surrounded by a three-layered membrane (the meninges) which contain the cerebrospinal fluid. If these meninges are infected by a virus or bacteria (meningitis) the patient may experience headache, fever, fits, neck stiffness and in severe cases may suffer permanent brain damage. Encephalitis is an infection of the brain tissue itself that may be confused with meningitis as it has similar symptoms and consequences.

Less common causes of dementia include Binswanger disease, hydrocephalus (increased pressure of the fluid within the brain), pernicious anaemia, failure of the kidneys (uraemia), liver failure, AIDS, Pick disease, Wernicke-Korsakoff syndrome (often secondary to alcoholism), punch drunk syndrome (from boxing), and a number of poisons such as organophosphates (in insecticides), glue sniffing, and drugs (eg. amphetamines, barbiturates) may cause permanent damage to the brain.

There are many other rare causes of dementia.

There are no diagnostic blood or other tests, but in advanced stages, a CT scan of the brain will show abnormalities.

The patient should be kept in a pleasant, safe, non-threatening environment with adequate medical, nursing, physiotherapy, occupational therapy and general support services. Medications may be given for irrational behaviour, hallucinations and violent tempers, but do not affect the disease process.

Unfortunately there is no cure for most causes, and patients progressively deteriorate.

See also ALZHEIMER DISEASE; DIFFUSE LEWY BODY DISEASE; DIOGENES SYNDROME; ORGANIC BRAIN SYNDROME

DENGUE FEVER

Dengue (or breakbone) fever is a generalised viral infection that is very common in many tropical countries of the world. The responsible Togavirus spreads from person to person by the bite of the Aedes mosquito, and the incubation period is usually three to seven days, but may stretch out to two weeks.

Symptoms include the sudden onset of a high fever, chills, and a severe aching of the back, head and legs (thus breakbone fever). Over the next few days a sore throat, blotchy skin and depression develop. These symptoms then totally cease for a day or two, before the second phase of the disease commences. This is similar to the first phase, but generally milder, and is usually accompanied by a rash that starts on the hands and feet and spreads to cover the entire body with the exception of the face. In severe cases, skin bleeding, and bleeding into the gut with accompanying diarrhoea can occur.
A RATIONALE FOR THE BRAIN

The diagnosis can be confirmed by a specific antibody blood test, but there is no cure and no vaccine. Aspirin and anti-inflammatories (eg. ibuprofen) are given for fevers and pains, and prolonged rest is required. Patients may become dehydrated because of diarrhoea, and require fluid replacement.

Eventually complete recovery occurs, but may take several months.

DEPRESSION

Depression is also known as an affective disorder, melancholia, hypothyrmia or a nervous breakdown. It is a medical condition, not just a state of mind, that affects 30% of people at some time in their life. Patients are not able to pull themselves together and overcome the depression without medical aid, although a determination to improve the situation certainly helps the outcome.

Depression may be a symptom (having a bad day and feeling sad), personality type (inherited with the genes), reaction (depressed because of loss of job, death in family etc.) or a disease (depression due to chemical imbalances in the brain). It is usually a mixture of several of these.

There are two main types of depression, endogenous and reactive, with very different causes.

Endogenous depression has no obvious reason for the constant unhappiness, and patients slowly become sadder and sadder, more irritable, unable to sleep, lose appetite and weight, and feel there is no purpose in living. They may feel unnecessarily guilty, have a very poor opinion of themselves, feel life is hopeless and find it difficult to think or concentrate. After several months they usually improve, but sometimes it can take years. It is due to an imbalance of the chemicals (neurotransmitters) that normally occur in the brain to control mood. The neurotransmitters include serotonin, noradrenaline and dopamine. If too little of any one is produced, the patient becomes depressed - if too much, the patient may become manic.

Endogenous depression can be further subdivided, depending on the combination of neurotransmitters that are too low. The subtypes are:-

<table>
<thead>
<tr>
<th>Type</th>
<th>Neurotransmitter level too low</th>
<th>Characteristics</th>
</tr>
</thead>
<tbody>
<tr>
<td>Non-melancholic depression</td>
<td>Serotonin</td>
<td>Obsession, panic, compulsions, anxiety</td>
</tr>
<tr>
<td>Non-psychotic melancholia</td>
<td>Serotonin, noradrenaline</td>
<td>+ lack of energy, tired</td>
</tr>
<tr>
<td>Psychotic melancholia</td>
<td>Serotonin, noradrenaline, dopamine</td>
<td>+ unmotivated, no pleasures, lack of concentration, no insight.</td>
</tr>
</tbody>
</table>

Those patients with endogenous depression are not able to pull themselves together and overcome the depression without medical aid, but doctors can alter the abnormal chemical balance by giving antidepressant medications. When they do start to improve, some patients with depression go too far the other way and become over-happy or manic. These patients are said to be manic depressive, have bipolar personality (generally severe swings of mood) or cyclothymic disorder (milder mood changes).

Reactive depression is the sadness that occurs after a death in the family, loss of a job, a marriage break-up or other disaster. Patients are depressed for a definite reason, and with time, will be often be able to cope with the situation, although some patients do require medical help.

Patients who are severely affected by depression are described as having a major depressive disorder.
There are no diagnostic blood tests or brain scans to prove these diagnoses, and the final diagnosis depends on the clinical acumen of the doctor.

There are many other causes of depression that overlap between the two types above or have totally independent causes.

The elderly often become depressed because they are confused, ill, unable to sleep as well as they would like, in discomfort, have no pleasure in life and can see no future. A change in attitude, environment and a bit of medication may often change their outlook dramatically.

The hormonal changes associated with pregnancy and menopause are often triggers for significant clinical depression. Postnatal depression usually occurs just after the birth of a child, with the sudden drop in the level of hormones. The mother feels inadequate, helpless and unable to cope. Urgent medical assistance is vital. In the menopause, the varying hormone levels may cause wide variations in mood that can be corrected by hormone replacement therapy.

Many women find that the normal sex hormone variations during the month will also cause mood changes, with depression and irritability being particularly common just before a menstrual period (premenstrual tension - PMT).

Many other diseases may have depression as a component, but doctors must be careful to differentiate between depression caused by the disease process itself, and depression in the patient because they are upset at having the disease.

Possible medical causes for depression include a tumour, cyst, abscess, cancer or infection of the brain; a stroke (cerebrovascular accident); hypothyroidism (a lack of thyroxine); Parkinson disease (a degeneration of part of the brain that co-ordinates muscle movement); serious viral infections (eg. AIDS, hepatitis, influenza, glandular fever); pernicious anaemia; systemic lupus erythematosus (an autoimmune disease); multiple sclerosis (a nerve disease that can affect any nerve in the body in a random and intermittent way); and abnormalities in the levels of potassium, sodium, bicarbonate and chloride (electrolytes) in the blood due to kidney or other diseases.

A number of medications, including cortisone, methylldopa (used for high blood pressure), beta blockers (used for heart disease) and various hormones (including the contraceptive pill) may have depression as a side effect.

There are many rarer medical causes of depression.

Numerous medications (antidepressants) that control the production or activity of the depressing chemicals in the brain are available to treat depression, but most antidepressant drugs work slowly over several weeks. Hospitalisation in order to use high doses of drugs or other treatments, and to protect the patient from the possibility of suicide, is sometimes necessary when the disease is first diagnosed. The other form of treatment used is shock therapy (electroconvulsive therapy - ECT), which is a safe and often very effective method of giving relief to patients with severe chronic depression.

Untreated depression may lead to attempted or actual suicide, which can be seen as a desperate plea for help.

Depression is not a diagnosis that patients should fear, as medication and counselling by a general practitioner, psychologist or psychiatrist will cure or control the vast majority of cases.
DERMATOMYOSITIS

Dermatomyositis is a rare disease that combines a persistent rash with muscle weakness. When it occurs without the rash (which is present in only 40% of cases) it is called polymyositis. The cause is unknown, but it commonly attacks those in late middle-age.

Patients experience a gradually progressive weakness and pain of the muscles in the neck, upper arms, shoulder, buttocks and thighs. Patients may also develop a dusky red rash on the cheeks and nose, shoulders and upper chest and back. The eyelids are often swollen and appear bruised. Unusual symptoms include redness and bleeding under the nails, cold hands, and a scaly rash over the knuckles.

The condition is diagnosed by blood tests, muscle biopsy and by measuring the muscle's electrical activity.

Drugs such as steroids, methotrexate and azathioprine are commonly used in treatment. Para-amino benzoic acid is sometimes used on the skin.

One in ten patients risk developing cancer, but there is no cure. Most patients can lead a relatively normal life, although a minority are disabled by muscle weakness.

See also POLYMYOSITIS

DEVIC DISEASE

Devic disease or syndrome (neuromyelitis optica) is an autoimmune inflammatory condition of the spinal cord and optic (eye) nerve that usually occurs in Asians. It is often associated with a recent viral or bacterial infection.

Patients develop eye pain and vision disturbances, neck or back pain, followed by altered sensations (eg. pins and needles, loss of sense of touch) and muscle weakness in the body below the area of back pain. The condition is diagnosed by an MRI scan.

Steroids may be tried as a treatment, but are often ineffective. Patients may have some spontaneous recovery, but the effects are usually permanent.

It was named after the French physician Eugène Devic (1869-1930).

See also AUTOIMMUNE DISEASES; TRANSVERSE MYELITIS

DIABETES INSIPIDUS

Diabetes insipidus is an uncommon type of pituitary gland failure. This gland lies under the centre of the brain and controls all other glands in the body. The condition may be triggered by a head injury, or develop slowly over many months because of a brain infection, tumour or stroke. It occurs when the pituitary gland fails to produce the hormone vasopressin that controls the rate at which the kidney produces urine. Without this hormone, the kidney constantly produces large amounts of dilute urine.

Patients have a huge urine output, are constantly thirsty, lose weight, develop headaches and muscle pains, become easily dehydrated, and may have an irregular heart beat. The diagnosis can be confirmed by a series of ingenious blood and urine tests after exposing the patient to varying degrees of water intake.

It can be controlled by regular injections of vasopressin which last from one to three days. Milder cases can be treated with a nasal spray containing a synthetic form of vasopressin, but this only lasts for a few hours.

Although diabetes insipidus cannot be cured, it is usually well controlled. Some cases do settle spontaneously, but most patients require life long treatment.

DIABETES MELLITUS TYPE ONE

Diabetes mellitus type one is also known as insulin dependent diabetes mellitus (IDDM) or juvenile diabetes, and causes excessive levels of glucose in the blood.

Glucose is used as fuel by every cell in the body. When glucose is eaten, it is absorbed into the blood from the small intestine. Once it reaches a cell, it must cross the fine membrane that forms its outer skin. This is normally impermeable to all substances, but insulin has the ability to combine with glucose and transport it across the membrane from the blood into the interior of the cell. Insulin is made by cells in the Islets of Langerhans in the pancreas, which sits in the centre of the abdomen.

Only 10% of diabetes suffer from this form of diabetes mellitus which is caused by a lack of insulin production by the pancreas. Most people develop this type as a child or in early adult life.

Symptoms include excessive tiredness, thirst, excess passing of urine, weight loss despite a large food intake, itchy rashes, recurrent vaginal thrush infections, pins and needles and blurred vision. Patients become steadily weaker because their muscles and other organs cannot work properly.
A RATIONALE FOR THE BRAIN

The diagnosis can be confirmed by blood and urine glucose levels and a glucose tolerance test (GTT). By measuring the amount of glucose in certain blood cells, the average blood glucose level over the past three months can also be determined. The level of insulin can also be measured in blood.

Daily self-testing is advisable to ensure that disease control is adequate. Both blood and urine tests for glucose are available, but the blood tests are far superior.

Diet is an essential part of treatment because the amount of glucose eaten is not normally constant, and diabetics lack the means of adjusting the amount of glucose in their blood with insulin. The diet must restrict the number of kilojoules (calories) being eaten, and sugar in all its forms should be eaten only with great caution. Fat should not account for more than a third of the total calories, and cholesterol intake should be restricted. Protein should be obtained more from poultry and fish than red meats. Carbohydrates other than sugar can be consumed freely. Grains and cereals with a high fibre content should be the main part of the diet. Artificial sweeteners such as aspartame (NutraSweet) can be used to flavour food and drinks. Fat cells can react abnormally to insulin very easily, and so overweight diabetics must lose weight. Exercise is encouraged on a regular daily basis. Patients should carry glucose sweets with them at all times to use if their blood sugar levels drop too low.

When first diagnosed, patients are often quite ill, and most are hospitalised for a few days to stabilise their condition. Insulin injections must be given regularly several times a day for the rest of their life. Initially derived from pigs and cattle, human insulin has now been produced by genetic engineering techniques. Insulin cannot be taken by mouth as it is destroyed by acid in the stomach, but can be injected into any part of the body covered by loose skin, although the same site should not be used repeatedly. The newer pen-style delivery systems enable diabetics to easily dial the required dose and inject as necessary with minimal inconvenience. There are many different types of insulin that vary in their speed of onset and duration of action.

The complications of type one diabetes mellitus include an increased risk of both bacterial and fungal skin and vaginal infections, the premature development of cataracts in the eye, microscopic haemorrhages and exudates that destroy the retina at the back of the eye, damage to the kidneys that prevents them from filtering blood effectively, poor circulation to the extremities (hands and feet) that may cause chronic ulcers and even gangrene to the feet, the development of brown skin spots on the shins, and sensory nerve damage (diabetic neuropathy) that alters the patient's perception of vibration, pain and temperature.

There are also complications associated with treatment such as a “hypo” in which too much insulin is given, excess exercise undertaken or not enough food is eaten, and blood glucose levels drop (hypoglycaemia) to an unacceptably low level. The patient becomes light-headed, sweats, develops a rapid heart beat and tremor, becomes hungry, then nauseated before finally collapsing unconscious. Glucose drinks or sweets given before collapse can reverse the process, but after collapse, an injection of glucose is essential. In an emergency, a sugary syrup or honey introduced through the anus into the rectum may allow a diabetic to recover sufficiently to take further sugar by mouth. Rarer complications of treatment are adverse reactions to pork or beef insulin, and damage to the fat under the skin if the same injection site is used too frequently. Diabetic ketoacidosis is the most severe complication.

On the other hand, with the correct treatment and careful control, patients should live a near-normal life, with a near-normal life span.

See also DIABETES MELLITUS TYPE TWO; DIABETIC KETOACIDOSIS

DIABETES MELLITUS TYPE TWO

Diabetes mellitus type two is also known as maturity onset diabetes or non-insulin dependent diabetes mellitus (NIDDM).

Glucose is used as fuel by every cell in the body. When glucose is eaten, it is absorbed into the blood from the small intestine. Once it reaches a cell, it must cross the fine membrane that forms its outer skin. This is normally impermeable to all substances, but insulin has the ability to combine with glucose and transport it across the membrane from the blood into the interior of the cell. Insulin is made in the pancreas, which sits in the centre of the abdomen.

Nine out of ten of diabetics suffer from the maturity onset form of diabetes mellitus, which is far more common in obese patients. There is adequate insulin production, but cells throughout the body fail to respond to the insulin, so glucose cannot enter the cell.

The symptoms include excessive tiredness, thirst, excess passing of urine, visual problems, skin infections and sensory nerve problems. Many patients are totally without symptoms when the diagnosis is discovered on a routine blood or urine test.
A RATIONALE FOR THE BRAIN

Blood and urine glucose levels are high in untreated or inadequately treated patients. A blood glucose tolerance test (GTT) is performed to confirm the diagnosis and determine its severity.

By measuring the amount of glucose in certain blood cells, the average blood glucose level over the past three months can also be determined.

Regular blood testing of glucose levels is also necessary, but normally on a weekly rather than daily basis. Urine tests are often inaccurate in the elderly, as their kidney function may be reduced to the point where glucose cannot enter the urine.

Diet is essential part of the diet because the amount of glucose eaten is not normally constant, but the medication levels do not normally vary from day to day. The diet must restrict the number of kilojoules (calories) being eaten, and sugar in all its forms should be eaten only with great caution. Fat should not account for more than a third of the total calories, and cholesterol intake should be restricted. Protein should be obtained more from poultry and fish than red meats. Carbohydrates other than sugar can be consumed freely. Grains and cereals with a high fibre content should be the main part of the diet. Artificial sweeteners such as aspartame (NutraSweet) can be used to flavour food and drinks. Fat cells can react abnormally to insulin very easily, and so overweight diabetics must lose weight. Exercise is encouraged on a regular daily basis.

Education of patients with diabetes is very important, so that they understand what they can and cannot eat and drink. Older people who develop diabetes can often have the disease controlled by diet alone or a combination of tablets and diet. Tablets (eg. metformin, glimepride, tolbutamide, chlorpropamide, glibenclamide, glipizide) make the cell membrane respond to insulin again. Weight loss is a vital part of treatment because if normal weight levels can be maintained, the disease may disappear.

Diabetics have an increased risk of both bacterial and fungal skin and vaginal infections, the premature development of cataracts in the eye, microscopic haemorrhages and exudates that destroy the retina at the back of the eye, damage to the kidneys that prevents them from filtering blood effectively, poor circulation to the extremities (hands and feet) that may cause chronic ulcers and even gangrene to the feet, the development of brown skin spots on the shins, and sensory nerve damage (diabetic neuropathy) that alters the patient's perception of vibration, pain and temperature. High blood pressure is more common than in the average person of their age.

There are also complications associated with treatment such as a "hypo" which is usually due to excessive medication. As a result blood glucose levels drop (hypoglycaemia) to an unacceptably low level. The patient becomes light-headed, sweats, develops a rapid heart beat and tremor, becomes hungry, then nauseated before finally collapsing unconscious. Glucose drinks or sweets given before collapse can reverse the process, but after collapse, an injection of glucose is essential. Diabetic ketoacidosis is the most severe complication.

With the correct treatment and careful control, patients should live a near-normal life, with a near-normal life span.

See also DIABETES MELLITUS TYPE ONE; DIABETIC KETOACIDOSIS

DIABETIC KETOACIDOSIS

Ketoacidosis is a severe complication or initial presentation of diabetes mellitus. It is due to a build-up of waste products and glucose in the bloodstream because of untreated or under-treated diabetes. Patients who are careless about their treatment, diet and self-testing may be affected. Almost invariably, it is the juvenile insulin dependent diabetics that develop this complication.

The symptoms include mental stupor, nausea, vomiting, shortness of breath and eventually coma. Blood sugar levels are very high and other blood and urine tests are abnormal.

Treatment involves the emergency injections of insulin, but urgent hospital treatment is necessary to control the situation adequately. If left untreated, death will occur due to kidney, heart or brain damage.

The prognosis is good with prompt medical care, but permanent organ damage may occur if treatment is delayed.

See also DIABETES MELLITUS TYPE ONE; DIABETES MELLITUS TYPE TWO

DIENCEPHALIC SYNDROME

The diencephalic syndrome is an abnormal reaction to a brain tumour in the part of the brain known as the anterior hypothalamus.

Those affected have significant weight loss, a pale complexion, vomiting and abnormal side to side eye movements (nystagmus). Patients may also develop wasting of the nerves to the eyes (optic atrophy) and
blindness, tremor, sweats, low blood sugar and pass urine frequently. The condition is diagnosed by a CT or MRI scan.

Sometimes surgery is possible, but the tumour is often inoperable and the prognosis is poor. 
See also BRAIN CANCER

**DIFFUSE LEWY BODY DISEASE**
Diffuse Lewy body disease can be best thought of as a mixture of dementia and Parkinson disease. It is a rare condition in which patients become progressively more confused, their attention span is reduced, they have hallucinations, delusions, unreasonable fears (phobias), uncontrolled movements of the limbs, muscle rigidity and they become very agitated. The cause is the presence of Lewy bodies scattered throughout the brain. Lewy bodies are tiny abnormal spheres found in nerve cells in the brain which alter the function of the cell. They can be seen when affected cells are examined under a microscope. The disease has no cure and progresses steadily, but some symptoms may be temporarily relieved by appropriate medications.

See also DEMENTIA; PARKINSON DISEASE

**DOWN SYNDROME**
Down syndrome is also known as mongolism and trisomy 21. It is a genetic defect due to the presence of three copies of chromosome 21 instead of two (normally one from each parent), causing various body and organ malformations. It occurs in one in every 800 births overall, but the incidence rises with the age of the mother.

<table>
<thead>
<tr>
<th>AGE OF MOTHER</th>
<th>INCIDENCE OF DOWN SYNDROME</th>
</tr>
</thead>
<tbody>
<tr>
<td>20</td>
<td>1:1520</td>
</tr>
<tr>
<td>30</td>
<td>1:900</td>
</tr>
<tr>
<td>35</td>
<td>1:350</td>
</tr>
<tr>
<td>40</td>
<td>1:100</td>
</tr>
<tr>
<td>45</td>
<td>1:25</td>
</tr>
</tbody>
</table>

The symptoms vary considerably between individuals. Common characteristics include poor muscle tone, joints that move further than normal, slanted eyes, a flattened facial appearance (accounting for the former name of “mongolism”), small stature, some measure of intellectual disability, small nose, a short broad hand and finger prints that have a whorl with the loop on the thumb side of the finger tip and other abnormal features. Other characteristics may include a fissured protruding tongue, short neck, widely spaced first and second toes, dry skin, sparse hair, small genitals, small ears, poorly formed teeth, keartoconus (protruding eye surface) and a squint.

It is usually easily recognised and diagnosed at birth. Nuchal translucency scanning and the triple blood test performed between 11 and 14 weeks of pregnancy can assess the risk a woman has of having a foetus affected by Down syndrome. A definite diagnosis before birth is possible from the fifteenth week of pregnancy by amniocentesis (taking a sample of fluid from around the foetus) or chorionic villus (placenta) biopsy.

There is no specific treatment, but plastic surgery may help some deformities. Patients require more than average medical attention for ear, nose and throat infections. More severe health problems include abnormal heart formation, abnormal formation of the intestines (especially the duodenum), a clouded lens in the eye, infertility and a higher than normal incidence of leukaemia may occur.

There is no cure, but provided there are no serious heart abnormalities, the life expectancy is close to normal. Good education and physiotherapy are effective in helping patients achieve a relatively normal life. People with this syndrome do have one very positive benefit though; the rate at which they develop cancer is dramatically lower than that of unaffected people.

**DUBOWITZ SYNDROME**
A rare developmental abnormality, that is familial, but both parents must be carriers.

The Dubowitz syndrome is characterised by a low birth weight baby with drooping eyelids (ptosis), small jaw, sparse hair, short stature, mild intellectual disability and eczema. No treatment is available, and there is no cure.

See also INTELLECTUAL DISABILITY

**DUCHENNE MUSCULAR DYSTROPHY**
Duchenne muscular dystrophy is a progressive and permanent gender linked inherited condition affecting only males. Females can be carriers, and statistically half the sons of a carrier are affected.

The symptoms start in infancy or early childhood, and progresses rapidly with worsening weakness of the pelvic, shoulder, arm and leg muscles resulting in inability to walk by 12 years of age. Eventually the muscles essential for breathing are affected. The condition can be diagnosed by specific blood tests, electrical studies of muscle action, and muscle biopsy.

There is no effective treatment and no cure is available, but physiotherapy is beneficial. Curvature of the spine (kyphoscoliosis) is the main complication. It eventually leads to death in the twenties or thirties.

It is named after the French neurologist Guillaume Duchenne (1806-1875).

See also MUSCULAR DYSTROPHY

**EATON-LAMBERT SYNDROME**

The Eaton-Lambert Syndrome (myasthenic syndrome) is a rare syndrome that affects motor nerve function. It is caused by an autoimmune reaction (inappropriate rejection of the body’s own nerve tissue) similar to myasthenia gravis, and results in muscle weakness that affects varying groups of muscles. Treatment involves the use of immunosuppressive drugs.

Patients have an increased risk of lung cancer, and the prognosis is poorer than for myasthenia gravis.

See also MYASTHENIA GRAVIS

**ECLAMPSIA**

See PRE-ECLAMPSIA AND ECLAMPSIA

**EHLERS-DANLOS SYNDROME**

Ehlers-Danlos syndrome (cutis hyperelastica or the elastic skin syndrome) is a congenital abnormality of skin development with eight clinically and genetically different variants. Patients have over extendable joints, excessively elastic and fragile skin, scarring of skin and growths on their knees and elbows, and premature arthritis may occur. It is diagnosed by a skin biopsy. There is no treatment or cure available, but life expectancy is normal.

The syndrome is named after Danish dermatologist Edvard Ehlers (1863-1937) and French dermatologist Henri Danlos (1844-1912).

**ENCEPHALITIS**

Encephalitis is any infection (eg. Japanese encephalitis, Murray Valley encephalitis) or inflammation (eg. Creutzfeldt-Jakob disease) of the brain tissue.

See also CREUTZFELDT-JAKOB DISEASE; JAPANESE ENCEPHALITIS; MURRAY VALLEY ENCEPHALITIS; RABIES; ST.LOUIS ENCEPHALITIS; SUBACUTE SCLEROSING PANENCEPHALITIS; WEST NILE ENCEPHALITIS

**ENCEPHALOPATHY**

An encephalopathy is any degenerative condition affecting the brain.

See also BATTEN DISEASE; WERNICKE-KORSAKOFF PSYCHOSIS

**EPENDYMOMA**

An ependymoma is a rare form of brain tumour that in most cases is benign. They tend to occur at the base of the brain and may extend into the spinal cord. It arises from the lining (ependyma) of the spaces (ventricles) within the brain that are filled with cerebrospinal fluid.

See also BRAIN CANCER

**EPILEPSY**

Epilepsy is a brain condition causing recurrent seizures (fits). It may be congenital or acquired later in life after a brain infection, tumour, injury or with brain degeneration in the elderly. Chemical imbalances in the body, kidney failure and removing alcohol from an alcoholic or heroin from an addict may also trigger the condition. Fits are caused by a short-circuit in the brain after very minor and localised damage. This then stimulates another
part of the brain, and then another, causing a seizure. Triggers such as flickering lights, shimmering televisions, certain foods, emotional upsets, infections or stress can start fits in some patients.

The symptoms vary depending on the type of epilepsy. There are several different types of epilepsy that are specifically described:

- **Petit mal absences.** The mildest form of epilepsy is petit mal or an absence seizure. These may vary from stopping in mid-sentence for a second or two and loss of concentration, to fluttering of the eyelids or other milder unusual muscle movements. There is no loss of consciousness or collapse. They are more common in children.
- **Abnormal absences.** These are similar to petit mal, but may be associated with partial seizures or a fugue state.
- **Simple partial seizures** (also known as focal seizures). Start with abnormal activity within one nerve cell in the brain, and the symptoms depend on the area of brain affected. There may be spasm of an arm or leg, strange smells, hallucinations or other phenomena. There is no loss of consciousness.
- **Complex partial seizures** (also known as temporal lobe or psychomotor seizures). These are the same as simple partial seizures, but there is some change in consciousness or loss of awareness.
- **Jacksonian epilepsy.** This is a simple partial seizure in which there is a progression in the muscle spasm or seizure from one area to another (eg. a muscle spasm may spread up the arm from the fingers to the shoulder). Occasionally there may be loss of power in the affected muscles (Todd paralysis) for hours or days afterwards.
- **Myoclonic seizures.** Sudden, short lasting muscle contractions involving just a single muscle, a limb, or sometimes the whole body. The patient may fall, but there is no loss of consciousness, and recovery is immediate, but the problem may recur.
- **Atonic seizures** (also known as drop attacks). A brief loss of all muscle strength, and loss of consciousness. Usually occur in children.
- **Secondary seizures.** These occur when a partial seizure progresses to become a generalised seizure.
- **Tonic-clonic seizures** (also known as grand mal seizures). Sudden onset of generalised muscle spasm, rigidity, loss of consciousness and collapse which last for a minute or two. The patient may go blue due to the cessation of breathing for the duration of the attack. The patient usually urinates and may pass faeces during the attack. After recovering from a grand mal fit, the patient has no memory of the event, is confused, drowsy, disoriented and may have a severe headache, nausea and muscle aches.
- **Status epilepticus.** A condition where one grand mal attack follows another without the patient regaining consciousness between attacks.
- **Tonic seizures.** These are a milder and briefer form of tonic-clonic seizures that last only a few seconds.

Many patients with epilepsy develop warning auras before an attack, which can be a particular type of headache, change in mood, tingling, light-headedness or twitching.

Epilepsy can be investigated by an EEG (electroencephalogram) to measure the brain waves, blood tests to exclude other diseases and a CT scan of the brain to find any structural abnormality.

Many different anti-epileptic drug combinations in tablet or mixture form are used to control epilepsy, and regular blood tests ensure that the dosage is adequate. Medication must be continued long term, but after several years without fits, a trial without medication may be undertaken. Epileptics must not put themselves in a position where they can injure themselves or others.

There are a large number of other medical conditions that may cause epilepsy as one of their symptoms. These include cerebral palsy, epilepsy partialis continua, Lafora body disease, Landau-Kleffner syndrome, lissencephaly syndrome, Sturge-Weber syndrome, Todd paralysis, Moyamoya disease, Unverricht disease etc.

**ERB-DUCHENNE PALSY**

Erb-Duchenne palsy is a rare complication of pulling too hard on the head during a difficult delivery of a baby, which stretches and damages the nerves running from the neck across the top of the shoulder to the muscles in the upper arm. The muscles are not damaged, but without a nerve supply, they cannot function.
Patients are unable to move the upper arm away from the body, fully bend the elbow, or turn the hand so that the palm faces backwards when the arm is beside the body. The arm hangs limply by the side, and if treatment is unsuccessful, it appears withered and wasted as the child grows older.

Splinting the shoulder in a position that allows the nerves to grow back and recover is necessary immediately after birth. More than nine out of ten cases recover with adequate treatment, usually within a month, but sometimes it takes six months.

It is named after German neurologist Wilhelm Erb (1840-1921) and French neurologist Guillaume Duchenne (1806-1875).

**ERB MUSCULAR DYSTROPHY**

Erb muscular dystrophy (limb-girdle dystrophy) is a rare gradually progressive muscle-wasting disease that may occur in the one family in successive generations, or appear for no apparent reason. It affects the muscles around the shoulder and pelvis, and progresses at a variable rate from its onset between ten and thirty years of age to cause severe disability in mid-life. A muscle biopsy is diagnostic, but no treatment is available other than physiotherapy and occupational therapy. The severity varies significantly between patients, some being only moderately inconvenienced while others are severely disabled and unable to care for themselves.

It is named after German neurologist Wilhelm Erb (1840-1921).

See also MUSCULAR DYSTROPHY

**ERB PALSY**

See ERB-DUCHENNE PALSY

**ESSENTIAL TREMOR**

An essential or familial tremor is an inherited muscle tremor that usually commences in the twenties. It causes a tremor of the hands at rest that worsens with emotional upsets and slowly deteriorates with advancing age. Attempts must be made to find a cause using investigations such as a CT scan of the brain, but all are negative. Numerous medications to reduce tremor are available, and although there is no cure, reasonable control is usually possible.

**EXTRAPYRAMIDAL LESION**

The extrapyramidal tracts of the brain and spinal cord are responsible for nerve signals that control movement of the body. They include motor nerves, the motor cortex of the brain and the nerve connections through the brain and spinal cord connecting these. An extrapyramidal lesion is damage to one of these areas and tracts of the brain and spinal cord. A stroke or spinal injury may be described as an extrapyramidal lesion if the pyramidal tracts are not involved.

**FAINT**

A faint (syncope) is a sudden, unexpected loss of consciousness that may be preceded for a few seconds by a feeling of light headedness. If a person has fainted, they should be made to lie flat with their legs raised to increase the flow of blood to the brain. Tilt the head backwards and make sure the airways are clear. Loosen any tight clothing. The person should regain consciousness within a few minutes. If the victim does not recover spontaneously within a short period, turn them on their side in the coma position and get medical help. Recovery usually occurs within a minute or two, and it is not associated with any convulsion or passing of urine or faeces.

Low blood pressure (hypotension) and poor blood supply to the brain are the absolute causes of a faint, and these in turn may be due to a number of conditions including stress, anxiety, fright, over exertion, lack of sleep, lack of food, heat, dehydration, lack of ventilation, prolonged standing and hormonal fluctuations.

A significant infection of any sort, from a bad dose of influenza to pneumonia or gastroenteritis may lead to a faint, particularly if the patient is trying to push on and not rest.

Stokes-Adams attacks are caused by a sudden change in the heart rate, with the heart slowing down markedly for a few seconds or minutes, and then recovering. It is due to a problem with the conduction of electrical impulses through the heart muscle.

Other causes of a faint may include the vasovagal syndrome (response to stress), a heart attack (myocardial infarct), a pulmonary thrombosis (blood clot in the lung), a stroke (cerebrovascular accident), sudden changes in emotional state, transient ischaemic attacks (temporary blocking of a small artery in the brain by a blood clot),
low blood sugar (hypoglycaemia from starvation, or overuse of insulin or sugar lowering tablets in a diabetic),
micturition syncope (faint that occurs when urine passed), pregnancy, hardening of the arteries (arteriosclerosis),
severe anaemia, dehydration, alcohol intoxication, and the effects of many drugs (eg. those that lower blood
glucose, narcotics, sleeping tablets, anxiety relieving medications).

There are many less common causes of fainting including narrowing (stenosis) of the main artery from the
heart to the body (the aorta), sudden episodes of irregular heart beat, high blood pressure (may sometimes
cause a faint as the increased pressure on the brain prevents it from working properly), migraines, epilepsy (may
be mistaken for a faint), a severe allergy reaction (anaphylaxis), the Shy-Drager syndrome and the Wolff-
Parkinson-White syndrome (peculiar abnormality of the electrical conduction system in the heart).

Some psychiatric patients may fake a faint as an attention seeking device.

No investigations are usually necessary, but if repeat attacks occur, the more serious diseases above that
may cause this condition (eg. low blood sugar, low blood pressure, irregular heart beat, infections, anaemia)
must be excluded by blood tests and electrocardiographs.

The patient usually recovers quickly once lying down, but should only rise slowly and when completely well.

See also COMA

FOETAL ALCOHOL SYNDROME
Excessive alcohol consumption by the mother for a prolonged period during pregnancy results in damage to
the foetus before birth and numerous deformities collectively known as the foetal alcohol syndrome. The baby is
small at delivery, and has excessive body hair, small finger nails, underdevelopment of the central part of the
face and mild intellectual disability. Less common symptoms include a small head and small jaw, poor
coordination, poor concentration and reduced muscle tone. It is a clinical diagnosis made on history from mother
and observation.

No treatment is available for the baby, and if recovery to normal developmental levels does not occur by one
year of age, further improvement is unlikely.

Mothers should be advised as strongly as possible not to drink alcohol to excess during pregnancy.

See also ALCOHOLISM

FRAGILE X SYNDROME
The fragile X syndrome was first described in 1969 and is the most common cause of intellectual disability. It
is an inherited condition affecting mainly males caused by an abnormal gene carried on the X chromosome. Men
have only one X sex chromosome matched to a small Y sex chromosome, while women have two X sex
chromosomes and the faulty gene's activity can be replaced by the one on the good X chromosome. Women
carry the abnormal gene from one generation to the next and sometimes have a minor intellectual disability.

The symptoms may include intellectual disability, behaviour problems, excess activity, epilepsy and autism.
They are often large babies, with smooth skin, large prominent ears, high forehead and oversized jaw. A squint,
visual abnormalities, poor speech, enlarged testes, flat feet and slack joints are other possible
complications. Affected boys are particularly susceptible to middle ear infections (otitis media).

It is diagnosed by examination of the genes in a cell sample taken from the blood after birth, or by a sample
from the placenta taken before birth.

There is no cure for this life long disability, but treatment can greatly assist these people to manage their
disability. It involves the use of appropriate support services (eg. occupational therapy), special education and
medication to reduce the excitement and excess activity, while genetic counselling of families is essential. Their
life expectancy is normal.

See also AUTISM; EPILEPSY; INTELLECTUAL DISABILITY

FRIEDREICH'S ATAXIA
Friedreich's ataxia (spinocerebellar degeneration) is an inherited condition causing degeneration of nerves in
the cerebellum (lower back part of the brain) and spinal cord, which usually starts in childhood or early adult life.
Patients develop an abnormal way of walking, incoordination, clumsiness, weakness and have abnormal
sensations. Heart inflammation and degeneration is a complication.

Nerve conduction studies, CT and MRI scans are used to confirm the diagnosis. No treatment is available
and death in early adult life is normal.
A RATIONALE FOR THE BRAIN

FUGUE
A fugue is a psychological condition in which a person may appear and act as though conscious, but is actually not aware of their surroundings, and acts in a way that may not fit with their normal personality. The person has no memory of their acts while in the fugue state. It may be associated with some unusual forms of epilepsy or psychiatric disorders.
See also EPILEPSY; SCHIZOPHRENIA

GALACTOASAEMIA
Galactosaemia is a congenital lack of the liver enzyme galactokinase that is responsible for the metabolism (break down) of galactose, which is one of the sugars in milk. These infants vomit, fail to thrive, and develop liver disease, intellectual disability, frequent infections and eye cataracts. A specific diagnostic test can be performed on red blood cells. A strict dietary avoidance of all milk products will prevent all the symptoms except intellectual disability.
See also INTELLECTUAL DISABILITY

GAUCHER DISEASE
Gaucher disease is an inherited condition that is more common in eastern Europeans and Jews, that causes the excess accumulation of fat in cells throughout the body due to the lack of a liver enzyme. The symptoms include enlargement of the spleen, anaemia, damage to bones in the back and thigh, and bone pain. Fractures of softened bones and poor liver function may occur, and sometimes there is lung involvement. It is diagnosed by a specific blood tests and tissue biopsy, but no treatment is available. Patients can expect a reasonable life expectancy unless the lungs become damaged.

GERSTMANN SYNDROME
Gerstmann syndrome is the loss of ability to remember previously known objects or learned skills. It is caused by damage to the dominant side of the brain by a stroke, tumour or other injury.
The patient is unable to recognise objects or people, write, calculate simple maths or distinguish left from right. CT and MRI scans of the brain are performed to determine the site of brain injury, and blood tests are done to exclude other diseases. If possible, the cause of the brain damage is corrected, but the prognosis is usually poor.
See also ASTROCYTOMA; CEREBROVASCULAR ACCIDENT; CRANIOPHARYNGIOMA; GLIOMA; MENINGIOMA

GERSTMANN-STRAUSSLER-SCHEINKER SYNDROME
The Gerstmann-Straussler-Scheinker syndrome is a rare inherited degeneration of the spine and posterior part of the brain (cerebellum) caused by a prion. Patients become progressively more unsteady, clumsy and uncoordinated in mid-life, and their sight and hearing may also be affected. There is no treatment available.

GILLES DE LA TOURETTE SYNDROME
See TOURETTE SYNDROME

GLAUCOMA
Glucoma is an increase in the pressure of the half-set jelly-like fluid inside the eyeball that damages the eye and affects the vision. The eye is filled with a thick clear fluid (aqueous humour) that is slowly secreted by special cells within the eye, while in another part of the eye the fluid is removed, allowing a slow but steady renewal. If there is a blockage to the drainage of the fluid from the eye while new fluid continues to be secreted, the pressure inside the eye increases, and damage occurs to the light-sensitive retina at the back of the eye. Other conditions may also cause glucoma including eye tumours, infections, injury, and in rare cases drugs (eg. steroids) may be responsible.
Three types of glucoma occur - chronic, acute and congenital:-
- Chronic glucoma (open-angle glucoma) is the most common type with a slow onset over years. It usually occurs in both eyes simultaneously and runs in families. Initially it affects the peripheral vision, which is how far can be seen to the sides and up and down while looking
straight ahead, and patients develop tunnel vision. One in every 75 people over 40 years have this type of glaucoma.

- **Acute glaucoma** (angle-closure glaucoma) is the worst type, as it develops in a few hours or days, but usually involves only one eye. There is a rapid deterioration in vision, severe pain, rainbow-coloured halos around lights, nausea and vomiting. It may start after a blow to the eye, or for no discernible reason. Immediate treatment of acute glaucoma is essential if the sight of the eye is to be saved, but even with good treatment, permanent blindness can occur.

- **Congenital glaucoma** (buphthalmos) occurs in babies who are born with the condition. The earliest sign is the continual overflow of tears from the eye, and the baby turns away from lights rather than towards them as a normal.

Glaucoma is diagnosed in most cases by measuring the pressure of the fluid within the eye. This can be done by anaesthetising the eye surface with eye drops and then resting a pressure measuring instrument (tonometer) on the surface of the eye while the patient is lying down, or by using a machine that directs a puff of air onto the eye to measure the pressure. Glaucoma may also be detected by measuring deterioration in peripheral vision using a computerised device, charts or by following a white dot on a large black screen. More complex tests include examining the eye through a microscope to determine the nature and seriousness of the glaucoma.

The excessive pressure in the eye caused by glaucoma can be reduced by eye drops, which are usually beta-blockers, and/or tablets that remove some fluid from the eye. Apraclonidine may be used in severe cases.

Beta-blocker eye drops include betaxolol (Betoptic), bimatoprost (Lumigan), carteolol, latanoprost (Xalantan), levobunolol (Betagan), metipranolol and timolol. Their side effects may include blurred vision, headache and a small pupil. They should be used with caution in asthma and heart disease. The other commonly used eye drops for glaucoma are brimonidine, brinzolamide, carbachol pilocarpine (Pilopt), dipivefrine, dorzolamide and travoprost.

The tablet used to treat glaucoma is acetazolamide (Diamox). Side effects may include pins and needles, excess urination and a poor appetite. It must be used with caution in pregnancy, and not in patients with liver disease.

In serious cases, laser microsurgery to the tiny drainage canals in the front of the eye is necessary. Congenital glaucoma always requires surgical treatment.

Without treatment, glaucoma progresses inexorably to total blindness, but if the disease is detected early, glaucoma in most patients can be successfully controlled but not cured.

**GLIOBLASTOMA**

See ASTROCYTOMA; GLIOMA

**GLIOMA**

A glioma (glioblastoma or malignant astrocytoma) is a form of brain cancer arising from the cells that support and surround the nerve cells in the brain. The cause is unknown, but there is a higher incidence in some families and occupations (eg. petroleum processing). The very variable symptoms may include personality changes, seizures, weakness in some areas and abnormal sensations. Bleeding into the tumour may suddenly worsen the symptoms. An optic glioma is one that develops on the optic nerve supplying the eye, and symptoms can include visual disturbances.

CT and MRI scans can usually identify the location of the tumour, but a biopsy is necessary to make a definitive diagnosis. Brain surgery is then performed to remove the tumour, followed by radiation or cytotoxic (eg. temozolomide) therapy. Unfortunately the prognosis for these aggressive tumours is very poor.

See also ASTROCYTOMA; BRAIN CANCER; CRANIOPHARYNGIOMA; MEDULLOBLASTOMA; MENINGIOMA; PINEALOMA; RETINOBLASTOMA

**GLYCOGEN STORAGE DISEASES**

The glycogen storage diseases are a number of rare inherited or congenital conditions including McArdle syndrome, von Gierke syndrome, Cori syndrome, Hers syndrome, Pompe syndrome, Tarui disease and Andersen syndrome. The enzymes and other natural chemicals in the liver that are necessary to process carbohydrates in food into glycogen and make it available to the body as a form of energy fuel, are missing or inactive in all these conditions.

The different diseases vary in their symptoms and effects depending upon which enzymes and chemicals are affected. Most patients have low blood sugar, large livers, short stature, subnormal mentality, and may bleed
excessively and fail in their development to progress through puberty. Other forms cause muscle pain and weakness.

The diagnosis can be confirmed by appropriate blood tests and a liver biopsy. Special diets and liver transplant sometimes assist in relieving the symptoms. The prognosis varies from minimal effects throughout life to death in infancy, depending on which type of glycogen storage disease is present.

**GRADENIGO SYNDROME**

The Gradenigo syndrome (also known as petrositis) is a bony abnormality of the skull that causes nerve compression.

Infection or a tumour of the petrous bone in the skull damages the 6th cranial nerve (abducent nerve) to cause a headache, double vision, facial pain and a middle ear infection. An infection may spread to the brain, ear or other areas. A skull X-ray or CT scan shows the abnormal bone.

Treatment involves potent antibiotics and surgery on the skull to drain any bone abscess or remove a tumour. The prognosis is good if diagnosed early, but permanent nerve damage may result.

**GUILLAIN-BARRÉ SYNDROME**

The Guillain-Barré syndrome (Landry-Guillain-Barré syndrome) is a rare inflammation of nerves (neuropathy) that usually follows a viral infection, injury, surgery, vaccination or a period of stress. The nerves in the legs are most commonly involved and the muscles that they supply become paralysed, and sensation to the affected areas may also be lost. No specific diagnostic investigations are available.

Steroids are used in treatment, but their use is controversial, and up to 20% of patients are left with a permanent disability. Most patients slowly recover, but in rare cases the muscles of breathing may be paralysed, which can result in death.

The disease is named after the French neurologists George Guillain (1876-1961) and Jean Barré (1880-1967).

**HALLERVORDEN-SPATZ SYNDROME**

The Hallervorden-Spatz syndrome is a rare progressive nerve disease that affects children and is caused by abnormal deposits of iron in specific parts of the brain. Affected children have athetosis (uncontrolled writhing movements), rigid muscles and dementia. Some medications used to treat Parkinson disease may be useful but there is no cure.

See also PARKINSON DISEASE

**HAND-SCHUELLER-CHRISTIAN DISEASE**

Hand-Schueллер-Christian disease is one of a group of congenital diseases known as histiocytosis X or Langerhans cell histiocytosis, that occur in children. The disease causes diabetes insipidus, patchy bony absorption in the skull, protruding eyes, lung damage and outer ear inflammation. Other symptoms may include skin rashes and gum inflammation and in advanced cases numerous organs may fail.

The diagnosis can be made by skull x-rays, biopsy of skin rashes, bone marrow biopsy and chest x-rays, then treatment involves irradiation of the bone lesions, very potent medications to prevent further bone and lung damage and conventional management of the diabetes insipidus, but no cure is possible.

**HEART FAILURE**

See CARDIAC FAILURE

**HEAT STROKE**

Heat stroke is a failure of temperature regulation by the brain due to overloading the body’s cooling mechanisms due to a combination of a hot environment, exertion and dehydration. It is a common problem in endurance athletes, soldiers and the elderly.

Victims develop a fever over 41°C, sweating stops, they becomes dizzy, weak, nauseated, confused, start vomiting and eventually becomes comatose and may convulse. Kidney failure, muscle breakdown, irregular heart beat, heart attack, liver failure and blood clots (thromboses) may also occur in extreme cases, and blood tests show abnormal biochemistry and high levels of white blood cells.
First aid is cool (not cold) water immersion or water spray and fan, followed by intravenous fluids, and medication to control shivering and prevent fitting. Regular blood tests are performed to monitor progress. It may be fatal if untreated, but responds well to appropriate management, although permanent organ damage is possible.

See also DEHYDRATION

HEERFORDT SYNDROME
Heerfordt syndrome (also known as Heerfordt-Waldenström syndrome and uveoparotid fever) is a complication of sarcoidosis involving the face, eyes and salivary glands. The cause is unknown.

The symptoms include painful inflammation of the salivary glands around the jaw, partial paralysis of one side of the face, inflammation of the eye (iritis) and sarcoidosis.

No treatment is necessary except for the sarcoidosis as the syndrome is a self-limiting condition that settles with time and rest, but the sarcoidosis may have serious consequences.

It is named after the Danish optician Christian Heerfordt (1872-1953).

HEMIBALLISMUS
Hemiballismus is a medical term describing uncoordinated and sudden movements (dyskinesia) that occur on only one side of the body. The usual cause is a tumour, abscess or infection involving a specific part of the thalamus in the brain.

HOLMES-ADIE SYNDROME
The Holmes-Adie syndrome (Adie’s pupil) is a congenital abnormality of the pupils that is more common in women. The pupils in the eyes are different sizes, respond poorly to light stimulation and tendon reflexes in the arms and legs may be slower than normal. Patients may be adversely affected by bright light, or find it difficult to see in dim light. It is a harmless condition and no treatment is necessary.

HORNER SYNDROME
Horner syndrome is a bizarre combination of symptoms involving the eye and sweat glands caused by compression of a special network of nerves (autonomic nervous system) in the chest due to lung cancer or pneumothorax, or in the brain due to a tumour. It may be the first sign of a quite advanced lung cancer. The syndrome is characterised by a drooping eyelid, contracted pupil and a sunken eye, associated with reduced sweating.

Numerous investigations must be undertaken to find the cause, including x-rays, CT and MRI scans. It is necessary to correct the underlying cause of the nerve compression, usually by surgery.

It is named after the Swiss ophthalmologist Johann Horner (1831-1866).

HUNTER SYNDROME
The Hunter syndrome (mucopolysaccharidosis type II) is a rare inherited abnormality of the metabolic system in which patients are unable to eliminate certain substances (mucopolysaccharides) from the body. Stiff joints, grotesque facial appearance, enlarged spleen and liver, heart abnormalities and mild intellectual disability are the usual characteristics, while deafness may also be a problem. It is diagnosed by specific blood and bone marrow tests and x-rays. Surgery may be performed for heart and facial abnormalities, but no cure is possible, although patients have a reasonable life expectancy. It is named after the Canadian physician Charles Hunter (1873-1955).

See also HURLER SYNDROME

HUNTINGTON CHOREA
Huntington’s chorea is a distressing, congenital condition that affects muscle function and coordination and is passed to half the children of a patient, but because the symptoms do not become apparent until between 30 and 50 years of age, it has often already been passed to the next generation before diagnosis. It is likely that all cases in existence can be traced back to previous sufferers.

In mid-life, patients develop irregular, random movements of the arms, legs and face; irritability, mood changes, antisocial behaviour, restlessness, fidgeting, mental deterioration, premature senility, and rigid
muscles. The symptoms develop very slowly over many years and serious psychiatric disturbances may also occur.

The chromosomal location of the gene that carries the condition from one generation to the next has been identified and the children of a patient can now decide if they wish to know if they are carrying the abnormal gene. This decision will obviously have dramatic effects upon their future lifestyle.

No effective treatment is available, but some psychiatric drugs can control mood changes, and muscle relaxants may ease the abnormal movements. The inevitable progression cannot be halted and death within 10 to 20 years of symptoms developing is usual.

The condition is named after American physician George Huntington (1850-1916).

See also SYDENHAM’S CHOREA

HURLER SYNDROME

Hurler syndrome (mucopolysaccharidosis type 1) is a rare inherited abnormality of the metabolic system in which patients are unable to eliminate certain substances (mucopolysaccharides) from the body. These children have a grotesque facial appearance, short stature, stiff joints, spinal deformities, intellectual disability, heart abnormalities, blindness and enlarged liver and spleen. Heart failure often occurs early in life. It is diagnosed by specific blood and bone marrow tests and x-rays. Surgery is possible for heart and facial abnormalities, but no cure is possible. It is similar to, but more serious than, Hunter syndrome, and patients usually die in childhood. It is named after the Austrian paediatrician Gertrud Hurler (1889-1965).

See also HUNTER SYNDROME

HYDROCEPHALUS

The brain and spinal cord are surrounded by cerebrospinal fluid (CSF). In the brain are a number of cavities, one of which contains a network of veins (the choroid plexus) that secretes the CSF, which passes through small ducts to the outside of the brain. From there it flows down and around the spinal cord in the back, from where it is absorbed into the blood. Hydrocephalus occurs when excess CSF accumulates in or around the brain.

There are three types of hydrocephalus:
- Obstructive hydrocephalus occurs if CSF cannot escape from the cavities within the brain due to a blockage in the draining tubes, and the brain is blown up by the fluid it contains.
- Communicating hydrocephalus occurs when there is a blockage of the circulation down the spinal cord and the fluid cannot be absorbed back into the bloodstream.
- Normal pressure hydrocephalus may occur in the elderly after a stroke, bleed into the brain or changes in brain structure with dementia.

The first two types are usually caused by a developmental abnormality of the foetus, or may develop in later life because of brain infections, tumours in the brain or skull, a colloid or other form of cyst in the brain, blood clots and other rarer conditions.

In babies with hydrocephalus, the soft skull is grossly dilated by the excess fluid. In older children or adults, a severe headache, personality changes, partial paralysis and loss of consciousness may be symptoms, as the harder skull is unable to expand. Other symptoms will depend upon the effect of the increased fluid pressure on the brain. It is diagnosed by a CT or MRI scan.

Treatment involves inserting a tube (ventriculoperitoneal shunt) into the skull to drain away the excess CSF. The tube has a one way valve allowing the CSF to escape, but preventing other fluids or infection from entering the brain. The far end of the tube is inserted into a vein in the neck or chest, or is run all the way through the chest, and allowed to drain into the abdominal cavity. Shunts can become blocked and require replacement or clearing occasionally. Any brain or intellectual damage that occurs before the condition is treated may be permanent.

Treatment is usually very successful in controlling the condition and allows the patient to lead a normal life with minimal impairment of body function or intelligence.

HYPERALDOSTERONISM

See CONN SYNDROME
HYPERKINETIC SYNDROME
The hyperkinetic syndrome is a developmental form of minimal brain dysfunction that starts before birth. These children have an early onset of personality problems, overactivity, poor coordination of limbs, learning disorders, and antisocial behaviour. It may be associated with epilepsy, and criminal activity common in early adult life. An EEG (electroencephalogram) may be abnormal.

Treatments available include behaviour modification techniques, drugs such as methylphenidate and dexamphetamine, special education programs, and environmental modification. Although the condition persists, satisfactory control is usually possible.
See also ATTENTION DEFICIT HYPERACTIVITY DISORDER

HYPERTHYROIDISM
Hyperthyroidism is overactivity of the thyroid gland, which sits in the front of the neck and is responsible for secreting a hormone called thyroxine. More severe forms may be called thyrotoxicosis or Grave’s disease or Basedow’s goitre.

Thyroxine acts as the accelerator for every cell in the body. If the level of thyroxine is high, the cells function at an increased rate - if the level of thyroxine is low, the cells function at a less than normal rate. The most common cause is an autoimmune disease, in which antibodies attack the thyroid gland and over stimulate it, but there are numerous other rarer causes.

Patients sweat excessively, lose weight, are nervous, tired, cannot tolerate hot weather and have a mild diarrhoea. Other effects include a rapid heart rate, slightly protruding eyes, warm skin, and a slight tremor. Patients also tend to fidget, dart quickly in their activity, and speak rapidly. The thyroid gland may be grossly enlarged (a goitre) or normal size.

The complications are serious. The weight loss and muscle wasting may become permanent, liver damage and heart failure may be fatal, psychiatric disturbances may lead to hospitalisation, eye scarring may lead to blindness, and infertility may occur.

The level of thyroxine and gland activity can be measured by blood tests, antibodies (eg. TRAB) can be detected in the patient’s blood and abnormalities may also be seen on an electrocardiogram (ECG).

The overactivity can only temporarily be controlled by medication (eg. propylthiouracil), but a cure can be obtained by surgically removing most of the thyroid gland or destroying it by giving the patient radioactive iodine (sodium iodide or iodine 131), which concentrates in the gland as it is an essential component of thyroxine. Because there is usually insufficient thyroid gland left behind after these procedures to produce adequate amounts of thyroxine, it is necessary for most patients to take thyroxine tablets on a daily basis.

If treated early, the prognosis is excellent, but if treatment is delayed until complications occur the outcome is far less favourable.

HYPOPARATHYROIDISM
The four parathyroid glands behind the thyroid gland in the neck regulate the amount of calcium in the blood and bones. In hypoparathyroidism, the glands secrete inadequate amounts of the hormone calcitonin, which results in excessive amounts of calcium to be taken from the blood and into the bones. It is a rare condition that may occur after thyroid gland surgery, or may be spontaneous for no apparent reason.

Symptoms may include spasms of the small muscles in the hands and feet, tingling lips, tiredness, wheezing, muscle cramps, fungal infections, abdominal pains, anxiety attacks, and behavioural alterations. If present for some time, the nails will become thin and brittle, the teeth will be deformed, cataracts may develop in the eyes, and the skin becomes dry and scaly. Untreated, it may cause irregular heartbeat, reduced growth in children, anaemia and intellectual disability.

The diagnosis is confirmed by measuring the amount of calcium in the blood, and x-rays show very dense bones and calcium deposits in abnormal areas (eg. brain).

Treatment may be an emergency in serious cases. Calcium injections and tablets, and vitamin D tablets are used, and once stabilised on treatment, the long-term outlook is good, but damage already done to eyes, teeth and other tissues may be irreversible. Very regular blood tests, follow-up visits, and lifelong medication are essential.
HYPOPITUITARISM

The pituitary gland sits in the centre of the brain and secretes hormones into the bloodstream that control every other hormone-producing gland in the body (e.g., thyroid gland, adrenal gland, ovary, testes). Hypopituitarism (Simmonds disease) occurs if the pituitary fails to produce the appropriate regulating hormones. If the gland fails to produce all the possible hormones, it is called panhypopituitarism. The causes include a tumor or abscess of the pituitary gland, head injury, stroke, swollen blood vessels, malnutrition, or other rare and complex reasons.

When the pituitary gland is underactive, every other gland it controls will also become underactive and the patient will have reduced sexual desire and activity, lose pubic hair, men will stop growing a beard and women will stop having monthly periods. Infertility may be a problem in both sexes. Other problems include weakness, tiredness, poor resistance to infections, low blood pressure, vision defects, and becoming stressed easily. Sometimes only part of the pituitary gland is underactive, so only some glands will malfunction. The diagnosis can be confirmed by blood tests, CT, and MRI scans.

Treatment involves correcting the cause, if possible, by surgery or irradiation, and giving hormone supplements by tablet or injection. The prognosis depends on the cause, but in most cases, the condition can be well controlled.

See also CUSHING SYNDROME

HYPOTHERMIA

Hypothermia or exposure is an abnormally low body temperature below 35°C due to being in cold conditions without adequate protection. Cold air alone can cause hypothermia, but if combined with wind, hypothermia occurs more rapidly. Cold water is the most serious cause, and death may occur in as little as a few minutes in icy water. An inadequately clad person may suffer hypothermia after only half a day in a climate where the temperature does not drop below 20°C. Even in tropical waters, shipwreck victims may die from hypothermia.

Alcoholics may neglect themselves and even in relatively mild conditions suffer from hypothermia due to inadequate clothing, shelter, and nutrition.

The blood vessels to the skin contract so that victims feel far colder to the touch than expected. Other symptoms are weakness, drowsiness, irritability, irrational behaviour and poor coordination. As the temperature drops further, delirium, coma, and death from an irregular heartbeat occur. The temperature of the patient must be measured using a rectal (through the anus) thermometer, as the skin temperature and mouth temperature are often inaccurate.

The treatment depends on the severity of the hypothermia. Mild cases respond well to good warm clothing, warm bed and rest. Shared body heat may be appropriate.

Moderate to severe cases will require hospitalisation for warmed air or oxygen, warm drinks, and warm fluids through a drip into a vein to heat the core of the body. This may be followed by immersion in a lukewarm bath that may have its temperature increased slowly over several hours. Heated blankets may also be used.

Patients who appear to have died because of hypothermia must be given mouth-to-mouth resuscitation and external heart massage for several hours while continuing to warm the body. Recovery may not occur until the body temperature rises to 32°C or more. Patients (particularly children) have been known to recover fully after prolonged periods of immersion or apparent death, with appropriate resuscitation, as the low temperature protect
the brain and body from damage. Rapid warming, and warming the surface of the body only (which may cause premature dilation of the arteries in the skin), can cause heart irregularities and death.

The prognosis depends upon the severity, prior health and age, but children recover far better than the elderly.

**HYPOTHYROIDISM**

Hypothyroidism is underactivity of the thyroid gland, which sits in the front of the neck and is responsible for secreting a hormone called thyroxine. This acts as the accelerator for every cell in the body. If the level of thyroxine is high, the cells function at an increased rate - if the level of thyroxine is low, the cells function at a less than normal rate. In children, hypothyroidism causes cretinism. The thyroid gland tends to fail with advancing age, particularly in women, it may be associated with an enlarged thyroid gland (goitre), and less commonly cysts or tumours may destroy the gland tissue.

Tiredness, weakness, muscle cramps, constipation, dry skin, headaches, nervousness, intolerance to cold weather and a hoarse voice are the most common symptoms. In more severe cases additional symptoms may include thinning of the hair, skin thickening, brittle nails, weight gain, shortness of breath, a thick tongue and a slow heart rate. The symptoms are referred to as myxoedema. The drop in thyroxine levels is usually gradual over many years and the symptoms may be overlooked until the disease is quite advanced. It is diagnosed by blood tests that measure the amount of thyroxine, and other thyroid-related substances.

Thyroxine or liothyronine tablets are taken long term to replace that not being produced by the gland. Patients usually notice a remarkable improvement in their quality of life as the thyroxine replacement tablets start to work. With adequate treatment the patient should lead a normal active life, but untreated, there is an increased risk of developing severe infections and heart failure, and premature death will occur.

**HYPOXIA**

Hypoxia is a lack of oxygen in the blood. It may be due to a very wide range of conditions from anaemia and suffocation to heart failure and lung diseases. It causes disorientation, headache, nausea, rapid rate of breathing, incoordination, bluish skin, chest pain, high blood pressure and a false happiness (euphoria). If not corrected it may lead to convulsions, sweating, vomiting, coma, irregular pulse, weakness and possibly death.

**HYSTERIA**

Hysteria is a psychological phenomenon in which the patient develops a subconscious and uncontrolled alteration to their normal functioning that falsely indicates the presence of a significant bodily malfunction. This may be paralysis of a limb, abnormal skin sensations, breath holding, collapse, hearing voices, seeing visions, shouting or screaming, vomiting or almost any other physical symptom. It may be triggered by anxiety, fear, frustration, pain or the behaviour of others around them (mass hysteria).

**IDIOT SAVANT SYNDROME**

The cause of the idiot savant syndrome is unknown, but it may be due to one area of the brain over developing before birth at the expense of other areas. The patient is usually subnormal in all areas of mental activity except one narrow field (eg. maths, music) in which they are extraordinarily talented. Mental tests are used to differentiate areas of skill from areas of below normal function. Intensive education is necessary to develop life skills and areas of subnormal ability.

See also AUTISM

**INCLUSION BODY MYOSITIS**

Inclusion body myositis is a rare form of progressive muscle wasting and weakness in the elderly with a very slow insidious onset and no known cause. Patients experience gradually worsening muscle weakness, which starts in the hands and feet and moves towards the trunk. Involvement of the muscles in the oesophagus may make swallowing difficult. It is diagnosed by muscle biopsy and tests on the electrical activity within muscles (electromyography - EMG). No specific treatment is available, but progression of the disease may be slowed in some patients by medication.

See also MUSCULAR DYSTROPHY
INTELLECTUAL DISABILITY

An intellectual disability or mental retardation is an inability to function mentally at a level compatible with living independently in the community. It may be present from infancy, or may develop later in life, and may be due to brain injury, genetic (present since conception), environmental, metabolic (body chemistry) or psychological causes.

Be warned that deafness and partial or complete blindness may mimic intellectual disability.

Any injury to the brain, from a blow, tumour, abscess, cancer, stroke (cerebrovascular accident), bleeding blood vessel or repeated convulsions (status epilepticus) may result in intellectual disability.

Birth injury, when the head is compressed for a prolonged period in the birth canal (vagina), sudden release of pressure at the moment of birth resulting in bleeding into the brain, premature separation of the placenta (which supplies all oxygen and nutrition to the baby) from the wall of the womb (uterus), or compression of the umbilical cord for a long time to reduce the flow of blood and oxygen to the baby, may all result in intellectual disability.

Cerebral palsy (spasticity) is a form of brain damage that occurs during the pregnancy, usually for no obvious reason. A poorly functioning placenta, and a foetus who takes nutrition from its twin, are possibilities.

If too much cerebrospinal fluid (the fluid around the brain) is produced, or insufficient is absorbed, the pressure of this fluid in and around the brain will gradually increase (hydrocephalus). The resultant pressure on the brain will affect its function and result in headaches, reduced intelligence, personality changes, memory loss and convulsions. In babies, the head will enlarge dramatically unless the excess fluid is drained.

Hundreds of different rare syndromes are known to cause intellectual disability. Some of these are inherited (passed from one generation to the next) while others are the result of the faulty joining of chromosomes at the moment of conception when an egg (ova) and sperm fuse. Some of the more common examples include:-
- Down syndrome (mongolism), which is due to the presence of three copies of chromosome 21 instead of two. Other features are flattened facial features, short stature, low set ears, thick tongue, broad hands with only a single transverse crease and slanted eyes. It occurs more commonly in children of older mothers.
- The fragile X syndrome is believed to cause a quarter of the intellectual disability in males. All men have only one X chromosome paired with a Y chromosome. Females, who have two X chromosomes, may be carriers from one generation to the next. The X and Y chromosomes determine the sex of every individual. Other symptoms include over activity, epilepsy, large build, large jaw and testes, and short sightedness.
- Klinefelter syndrome also only affects males. They have additional X chromosomes matched with a single Y chromosome. Their genetic make up is therefore XXY or XXXY instead of the normal XY. Other features are delayed puberty, tall slim build, emotional disturbances and underdeveloped genitalia.
- Savants (idiot savant syndrome) have extraordinary talents in one narrow area (eg: music, maths), but are otherwise subnormal. It may be due to overdevelopment of one area of the brain at the expense of others.

Rarer syndromes that may be responsible for intellectual disability include Angelman syndrome (abnormal walk, convulsions, inappropriate laughter and abnormal facial appearance), cat eye syndrome (vertical slit shaped pupil), Coffin-Lowry syndrome (males with prominent lips, coarse facial features, slanted eyes and curved back), Conrad syndrome (cataracts and limb contractures), cri-du-chat (cat cry) syndrome, de Lange syndrome (dwarfism), Dubowitz syndrome (droopy eyelids), galactosaemia (a disturbance of the breakdown of the sugar galactose in milk), Gaucher disease (fat storage disorder), Louis-Bar syndrome, Miller-Dieker syndrome, moyamoya disease (affects Japanese), Neu-Lexova syndrome, Niemann-Pick disease (excess storage of certain fats), Prader-Willi syndrome (small at birth, have poor muscle tone, eat compulsively to become obese and have small genitals), Rett syndrome (rapid breathing, autism), Richner-Hanhart syndrome (excess tyrosine in blood), Rubenstein-Taybi syndrome, Seckel syndrome, Soto syndrome (enlargement of the forehead, abnormal eye shape, increased physical growth in childhood), Sturge-Weber syndrome (dark red birth mark on the face, convulsions, paralysis of one side of the body and eye abnormalities), Tay-Sachs disease (disease of Ashkenazi Jews), trisomy 8, tuberous sclerosis (red nodules on the face, deformed nails and eye damage) and Williams syndrome.

Various toxins and infections may adversely affect the brain and its mental capacity. Examples include foetal alcohol syndrome (caused by the mother drinking excessive amounts of alcohol during pregnancy), lead or copper poisoning, and overdoses of prescribed drugs or addiction to illegal drugs (eg. amphetamines, cocaine).
Infections of a pregnant woman may affect her unborn child. Examples include German measles (rubella), toxoplasmosis (caught from cats), cytomegalovirus (CMV - which is usually harmless to adults) and syphilis (a sexually transmitted bacteria).

Disturbances to the body’s chemistry may also damage the brain and prevent it from functioning to its full potential. Examples of conditions that can cause this are phenylketonuria (PKU), malnutrition and untreated or poorly controlled diabetes mellitus.

Psychological conditions may also be responsible for intellectual disability. Examples include autism, psychoses, and isolation and stimulus deprivation of a child.

**INTRACRANIAL HAEMORRHAGE**
See SUBARACHNOID HAEMORRHAGE; SUBDURAL HAEMATOMA

**JAPANESE ENCEPHALITIS**
Japanese encephalitis is a viral infection of the brain that is transmitted from animals such as pigs and horses to man by a mosquito bite. It is found in areas where rice paddies occur throughout east and south Asia and on some Pacific islands. Symptoms include a fever, belly pains, dizziness, sore throat, cough, headache, neck stiffness, nausea, vomiting, tiredness, disturbed mental functions, disorientation, coma and sometimes death. It may cause permanent brain damage in up to half of survivors with personality changes, fatigue and inability to concentrate.

Tests on blood and the fluid around the brain are abnormal, and can confirm the diagnosis, but no specific treatment is available, although it can be prevented by a vaccine.

Acute attack lasts for one to three weeks, but complete recovery may take months. The overall mortality rate is up to 50%, but worsens in children.

A vaccine is available and can be given from one year of age. Three vaccinations at weekly intervals give at least five years protection, but the vaccine takes at least 10 days to become effective. The vaccine may be used with caution in pregnancy, breastfeeding and children, and with a history of urticaria, asthma, hay fever and allergies. Avoid alcohol for 2 days after vaccination. Side effects to the vaccine may include allergy reaction, swelling of tissue, asthma-like reaction, fever, headache and tiredness, and less commonly a rash, dizziness, muscle pain, nausea and vomiting.

See also ENCEPHALITIS

**JERVELL-LANGE-NIELSEN SYNDROME**
The Jervell-Lange-Nielsen syndrome is a rare inherited disorder of electrical conduction within the heart. Both parents must carry the gene for an infant to be affected. The child is deaf and has episodes of abnormal heart contraction, which cause a sudden collapse. Abnormal ECG (electrocardiograph) tracings are diagnostic. No treatment is available and sudden death possible.

**JOHANSON-BLIZZARD SYNDROME**
The rare Johanson-Blizzard syndrome is an inherited abnormality of connective tissue development, but both parents must carry the gene for an infant to be affected. The baby has abnormal nose structure, failure of the anus to form, underdeveloped and poorly functioning skin, deafness, mild intellectual disability, and failure to gain weight and height. Plastic surgery may be used to correct some deformities, but no cure is available.

See also INTELLECTUAL DISABILITY

**KALLMANN SYNDROME**
Kallmann syndrome is a familial (passes from one generation to the next) sex hormone abnormality that causes the part of the brain (hypothalamus) that controls hormone production to malfunction. The symptoms include poorly developed genitals in a boy, loss of sense of smell, long arms and legs, and in a male a female body shape.

The blood level of testosterone is very low and male hormone replacement therapy is used as a form of treatment. Although there is no cure, good control is usually possible.
A RATIONALE FOR THE BRAIN

KERNICTERUS
Excess bilirubin in infants may not only cause jaundice (yellow skin) but may also be deposited in the brain and spinal cord to cause kernicterus. Untreated, it may cause permanent brain and spinal cord damage.

KIDNEY FAILURE, CHRONIC
See RENAL FAILURE, CHRONIC

KLEINE-LEVIN SYNDROME
The Kleine-Levin syndrome is a sleep and mood disorder of no known cause in which patients experience recurrent periods of severe excessive sleepiness, increased appetite, mood disturbances, increased sexual activity, disorientation, hallucinations and memory loss. There may be no symptoms for months between attacks and it is more common in men, with onset from 12 to 20 years of age. It is treated with stimulants, and is self-limiting by about 40 years of age.

KLINEFELTER SYNDROME
The Klinefelter syndrome (XXY syndrome) is a congenital sexual abnormality that affects one in every 500 males. The chromosomes from the mother and father of these men combine incorrectly with two X chromosomes and one Y being present (XXY) instead of one of each (XY). Patients have very small testes and penis, small breasts develop, they have scanty body hair, and are impotent and sterile. It is diagnosed by chromosomal analysis of blood sample.
Testosterone (male hormone) tablets or injections can be given to improve the body shape and impotence and plastic surgery to remove the breasts is sometimes necessary, but the infertility cannot be corrected and no cure is possible.

KLUMPKE PALSY
The Klumpke (Déjerine-Klumpke) palsy is a form of arm damage to an infant that occurs during birth due to stretching of the lower nerves in the armpit during a forceps or difficult delivery. The infant’s hand is limp below the wrist. Nerve conduction studies sometimes performed to confirm the diagnosis, and the wrist is then splinted in neutral position. Most babies fully recover.
See also ERB-DUCHENNE PALSY

KUGELBERG-WELANDER SYNDROME
The Kugelberg-Welander syndrome is a familial (runs in families) nerve disease that affects muscle function. It is due to an abnormality of the point where a nerve joins to a muscle.
Patients develop muscular weakness of the shoulder girdle and pelvis and an abnormal gait. Some patients develop over large calf muscles and tongue weakness. It is diagnosed by abnormalities in blood tests, nerve conduction studies and muscle biopsy. Reflexes involving affected muscles are also abnormal. Physiotherapy may help, but there is no specific treatment available. Patients have a relatively normal life span, but the disease process is slowly progressive.
See also MOTOR NEURONE DISEASE; MUSCULAR DYSTROPHY

LABYRINTHITIS
Labyrinthitis is an inflammation, viral infection, or rarely bacterial infection, of the semicircular canals (labyrinth) in the inner ear that control balance. Sometimes toxins or ear damage may be responsible. Severe dizziness occurs along with abnormal eye movements, and non-existent noises may be heard. Rarely, permanent damage to the balance mechanism may occur. Caloric tests (alternating heat and cold in the outer ear canal) induce worse dizziness and confirm the diagnosis.
Medications can be used to reduce inflammation and dizziness, and antibiotics are sometimes necessary. Most cases settle in a few days, some persist for weeks.
See also BENIGN PAROXYSMAL POSITIONAL VERTIGO
LA FORA BODY DISEASE

Lafora body disease is a rare progressive form of epilepsy affecting children and teenagers. There is a familial (inherited) tendency caused by a recessive gene on chromosome 6. Steadily worsening seizures, muscle spasms, poor coordination and dementia occur.

It is diagnosed by finding characteristic changes on EEG (electroencephalogram), and specific inclusion bodies are found in brain, liver and skin cells. Antiepileptic medications sometimes assist, but death usual within a decade of onset.

See also EPILEPSY

LANDAU-KLEFFNER SYNDROME

The Landau-Kleffner syndrome is a rare form of epilepsy of no known cause. The patient has muscular seizures, followed by a slow failure of the ability to speak or understand language, and behavioural disorders. The syndrome usually starts between 4 and 9 years of age and is treated with anticonvulsant medications and speech therapy. 50% recover, but the prognosis is poor with early onset.

See also EPILEPSY

LANDRY-GUILLAIN-BARRÉ SYNDROME

See GUILLAIN-BARRÉ SYNDROME

LANGER-GIEDION SYNDROME

Langer-Giedion syndrome is a rare childhood deformity caused by a congenital chromosome abnormality. Affected children develop sparse hair, bulbous nose, a small head and multiple bony overgrowths. It is diagnosed by an abnormal chromosomal analysis, and X-rays show extra bone formation. Plastic surgery may be used for deformities, but there is no cure.

LAURENCE-MOON-BIEDL SYNDROME

The Laurence-Moon-Biedl syndrome is an inherited condition of the eye, brain and genitals that causes night blindness due to excessive amounts of pigment in the retina at the back of the eye, obesity, intellectual disability, extra fingers and toes and underdeveloped genitals. Examination of the back of the eye through an ophthalmoscope (magnifying light) will show the excess pigment. There is no treatment available and no cure.

LEAD POISONING

Lead (Pb) has been widely used in industry including batteries, paints (particularly dangerous in flaking old paint), crystal glass, ceramics, old plumbing fixtures, leaded petrol and some old-fashioned medications. Swallowing or inhaling lead compounds may lead to lead poisoning (plumbism) which causes belly pains, irritability, tiredness, loss of appetite, anaemia, poor coordination, slurred speech, convulsions, coma and death. Permanent damage to nerves (neuropathy), brain (encephalopathy) and kidneys is possible in survivors.

Lead can be detected in the body by specific blood tests. The normal range is less than 1.2 µmol/L. 95% of whole population should have levels less than 0.8 µmol/L. Higher results should be interpreted thus:

- 1.3 to 1.9 µmol/L Retest adults three monthly, risk of mental damage in children
- 1.9 to 2.4 µmol/L Retest adults monthly, check sources of exposure.
- Greater than 2.4 µmol/L Remove adults from exposure, treat children with aggressive elimination therapy.

Lead can also be detected in a urine sample. The normal range is 5 to 105 µg/24 hours (less than 0.5 µmol/L or less than 0.25 µmol/day).

If lead has been recently swallowed, induce vomiting and give activated charcoal. In chronic cases, medications can slowly remove the lead from the body. The prognosis depends on age, health and weight of the patient, and the dose of lead.

LEIGH DISEASE

Leigh disease (subacute necrotising encephalomyelopathy) is a rare disease of brain metabolism that affects children between 6 months and 6 years of age. It is caused by an inherited inability of the body to deal with specific waste products in the brain, that slowly build up until the brain becomes inflamed and symptoms occur.
The child appears normal until s/he suddenly starts vomiting, losing weight, fitting, becomes mentally unstable, blind and then dies. This can occur in a matter of weeks, or may progress slowly over several very distressing months.

Blood tests show abnormal levels of waste products, but no treatment available, and the condition is invariably fatal.

LENNOX-GESTAUT SYNDROME
The Lennox-Gestaut syndrome is a very severe form of epilepsy of no known cause in which patients have multiple variable types of seizures, intellectual disability, sudden drop attacks, and intractable epilepsy. Self-injury or death from persistent fitting is possible. An abnormal EEG (electroencephalogram - measures electrical activity in brain) confirms the diagnosis.

Combinations of potent anti-epilepsy drugs are used in treatment, but brain surgery may need to be performed in resistant cases. There is no cure, and control of seizures is usually very difficult.

See also EPILEPSY

LEPROSY
Leprosy (Hansen’s disease) is a very slowly progressive infection caused by the bacterium Mycobacterium leprae that damages the skin and nerves (neuropathy). It is spread from one person to another by prolonged close contact, most commonly in childhood.

Pale, thick patches of skin on the hands and feet are the first sign, followed by slowly enlarging nodules, then nerves supplying the affected areas of skin become involved and sensation is lost. The cooler parts of the body, furthest from the heart, are affected first. As the disease progresses, a pins and needles sensation may be felt, ulcers form, and bones in the fingers and toes begin to disintegrate. There is rarely any pain. Damage and deformity is due to unintentional burns and injuries to totally numb tissue. In severe cases, fingers and toes do fall off, but this is very rare. The diagnosis can be confirmed by microscopic examination of a skin biopsy.

A number of antileprotic drugs (eg. dapsone, clofazimine, rifampicin, ethionamide) are available that will slowly cure leprosy over several years. Any existing deformities must be treated with plastic surgery. With good management, cure is possible and patients can live normally in the community. Untreated, the disease progresses to death over 10 to 20 years.

Curiously, the armadillo is the only animal other than humans that can catch leprosy.

LEPTOSPIROSIS
Leptospirosis is a bacterial infection of the liver and other organs caught from infected cattle and pigs by abattoir workers, veterinarians and farmers. In third-world countries, dogs and rats may also be carriers. The spirochete bacteria Leptospira interrogans enter through minor abrasions or by being swallowed. The incubation period varies from three days to three weeks.

Patients develop a sudden high fever, headache, stomach pain, muscle aches and inflamed eyes. After a couple of days, these symptoms disappear, and the second stage of the infection commences which lasts for one to four weeks, and the patient complains of swollen lymph nodes, a generalised rash, eye pain, and in severe cases yellowing of the skin (jaundice). The second stage may cause permanent liver damage and Weil Syndrome. The diagnosis is confirmed by a specific immunoglobulin found on a blood test.

Antibiotics such as penicillin are prescribed as treatment, but sometimes they have remarkably little effect. Careful nursing is important. The disease can usually be prevented by taking a doxycycline antibiotic tablet once a week.

It is usually cured by correct treatment, but if jaundice develops, the death rate may be as high as 10%.

Leptospirosis is thought to be the oldest disease caused by civilisation as it probably first affected humans at the same time that they domesticated cattle and pigs about 5000 BC.

LESCH-NYHAN SYNDROME
The Lesch-Nyhan syndrome is a rare X-linked inherited genetic error of metabolism involving uric acid, which is produced by the breakdown of protein in the diet. It passes through the female line but only affects males.

Symptoms include mental deficiency, severe gout, self-mutilation, and abnormal uncontrolled writhing movements of arms and legs. Severe arthritis may be a complication.
Blood and urine tests show very high levels of uric acid, and medications to remove uric acid are prescribed. There is no cure and control is often poor.

**LISSENCEPHALY SYNDROME**
The lissencephaly syndrome is a rare brain and developmental disorder. It is an inherited condition, but only if both parents are carriers of a defective gene. Epileptic seizures, poor muscle tone, jaundice, cataracts in the eye, and wrinkled forehead skin are the symptoms.
No treatment is available, and seizures are difficult to control with normal epilepsy medication. An early death is normal.
See also EPILEPSY

**LOCKED-IN SYNDROME**
The locked-In syndrome is an horrendous complication of certain types of brain damage caused by a stroke, tumour or injury to particular parts of the brain, or late stage multiple sclerosis. The patient has total paralysis of limbs and facial nerves, but normal consciousness, and is able to communicate only by eye movements.
No treatment is available, and death from pneumonia due to lack of movement and poor function of muscles of breathing is usually a blessed release.
See also MULTIPLE SCLEROSIS

**LOUIS-BAR SYNDROME**
The Louis-Bar syndrome is a rare form of rapidly progressive brain deterioration due to degeneration of the cerebellum (lower back portion of brain) and spinal cord. The symptoms include dilated capillaries on the whites of the eyes, the face and areas of skin flexion (eg. arm pit, behind knee); intellectual disability; recurrent infections of lungs and ears, and poor coordination that steadily worsens. Late symptoms include twitching movements of the eyes and abnormal writhing movements of the arms and legs. There is an above average incidence of cancer.
Numerous blood tests are abnormal, including very low immunoglobulin levels.
No treatment is available and death in teenage years is usual.
See also INTELLECTUAL DISABILITY

**LOWE SYNDROME**
The Lowe syndrome (oculocerebrorenal syndrome of Lowe) is a rare, inherited, body chemistry disorder that is passed to males only through the female side of the family. These boys have intellectual disability, eye cataracts, clouding of the cornea (outer surface of eye), abnormal skin folds beside the eyes, and abnormal eye socket shape. Some patients have rickets and Fanconi syndrome. Abnormal levels of amino acids (protein breakdown products) found in urine. There is no cure, but they have a reasonable life span.
See also RICKETS

**LYME DISEASE**
Lyme disease is a relatively common blood infection caused by the bacterium *Borrelia burgdorferi* that occurs in the northeast United States. It is spread by the bite of the tick *Ixodes* from infected mice or deer to humans. The tick may lie dormant for up to a year before passing on the infection with a bite.
The disease has three stages:-
- in stage one the patient has a flat or slightly raised red patchy rash, fever, muscle aches and headache.
- stage two comes two to four weeks later with a stiff neck, severe headache, meningitis (inflammation of the membrane around the brain) and possibly Bell's palsy.
- in stage three, which may come three to twelve months later, the patient has muscle pains, and most seriously a long lasting severe form of arthritis that may move from joint to joint. Persistent crippling arthritis sometimes occurs.
The diagnosis is confirmed by specific immunoglobulin blood tests, then a prolonged course of antibiotics is prescribed.
Long term, one third of patients may suffer from continuing muscle and joint pains, while a smaller percentage have after effects of the meningitis.
See also BELL'S PALSY; MENINGITIS
MACHADO-JOSEPH SYNDROME
Machado-Joseph syndrome (or Azorean disease) is a progressive degeneration of the lower part of the brain (cerebellum) and the adjacent spinal cord. It is a congenital condition that passes readily from one generation to the next. First discovered in the Azores Islands in the North Atlantic.
Muscle spasticity and rigidity, poor coordination, inability to speak clearly, limb weakness and a fixed facial expression are the common symptoms. Tremor, eye movement paralysis and eye twitching may also occur. Joint damage from muscle spasms may be a complication. There is an abnormal brain appearance on a CT scan.
Drugs such as levodopa and baclofen help ease rigidity and spasticity, but there is no cure, and the condition is slowly progressive.

MANIC-DEPRESSIVE
The manic-depressive disorder, which is also known as the bipolar affective disorder or cyclothymia, results in severe swings in mood that start most commonly around 30 years of age and almost never after 60.
The patient suffers dramatic changes in mood from very depressed to manic and vice versa. When depressed the patient is sad, loses interest in pleasure, loses weight, wakes through night and cannot get back to sleep, becomes restless, fatigues easily, feels worthless, cannot concentrate and may think of death. In the manic stage which may follow days, or even hours after the depression, the patient has an inflated self esteem, decreased need for sleep, is very talkative, has sudden changes in thoughts, poor attention span, increased sexual activity including promiscuity, spends excessively and takes risks.
A diagnosis can only be made after careful assessment by a doctor - there are no specific diagnostic tests. Once diagnosed, specific medications to control mood swings (eg. lithium, valproic acid, carbamazepine) can be prescribed.
There is generally a good response to treatment, but there is a risk of suicide in the depressive phase, and of self injury from dangerous activities in the manic phase.
See also DEPRESSION

MARCUS GUNN SYNDROME
The Marcus Gunn or jaw winking syndrome is a rare cause of a drooping eyelid that is present from birth. When the person looks straight ahead the eyelid droops (ptosis) but looking downwards does not cause a further drop in the eyelid. When the jaw is opened or moved from side to side, the eyelid rises in time with the jaw movement. It is caused by abnormalities in the development of the tiny muscles in the eyelid and abnormal nerve supply to these muscles. The condition can be improved by surgery.

MARFAN SYNDROME
Marfan syndrome is an uncommon inherited condition that affects the skeleton, heart and eye, and occurs in all races but only in one out of every 20,000 people.
Its characteristics include very long thin bones in the arms, legs, fingers and toes (arachnodactyly), a tall skull, excessive joint movement, a high foot arch and a humped back. Half the patients have an eye lens that is in the wrong position, and they may develop keratoconus (protruding eye surface) and a detached retina (the light-sensitive area at the back of the eyes), which results in partial or total blindness. An abnormality in the elastic tissue of the heart valves and major arteries causes these to fail and the pumping of the heart to be inefficient. The main artery of the body, the aorta, becomes overly dilated and distorted and may eventually rupture, and heart infections (endocarditis) are common. Most patients do not have all these symptoms, as there is great variation between them. Some may be totally unaware that they are affected and just appear to be very tall and thin.
It is diagnosed by the characteristic appearance of the long bones on X-ray, and by assessing the heart abnormalities with echocardiograms. The problems in the heart and aorta are controlled and corrected by both medication and surgery, but death in middle age is common unless corrective surgery is successful.

MARINESCO-SJÖGREN SYNDROME
The Marinesco-Sjögren syndrome is a rare progressive degeneration of the cerebellum (lower back part of brain) that starts in early childhood. It is a familial (inherited) condition, but both parents must be carriers. Sufferers have poor coordination (ataxia), eye cataracts, intellectual disability, multiple bony abnormalities and underdeveloped testes or ovaries. Medication can be used to control ataxia, but there is no cure, and the symptoms steadily worsen to death.

See also FRIEDREICH'S ATAXIA

Mastoiditis
Mastoiditis is a bacterial infection of the mastoid bone, which is a small bump of bone at the bottom of the skull immediately behind the ear that contains a microscopic honeycomb of air filled spaces. The infection almost invariably occurs as a result of infection spreading from the middle ear in patients who have recurrent or severe attacks of middle ear infection (otitis media).

Severe pain and tenderness occurs behind the ear, as well as fever, and redness over the mastoid bone. In some cases the infection will eat away the bone at the back of the ear canal and allow the pus to escape into the ear. The hole between the ear canal and the mastoid air cells, and the cavity in the mastoid bone that results from the infection, are permanent. If left untreated, the infection may spread into the brain.

An X-ray of the mastoid shows the air spaces within it to be destroyed and replaced by pus (an abscess). Potent antibiotics are then prescribed, which are sometimes given by injection. If these fail, an operation to drain the pus out of the mastoid bone will be necessary (a mastoidectomy). The prognosis is good with appropriate treatment.

McARDLE SYNDROME
See GLYCOGEN STORAGE DISEASES

MEDULLOBLASTOMA
A medulloblastoma is a cancer of the nerve cells in the brain. The cause is unknown but they are more common in children.

The tumour increases the pressure in the brain to cause nausea and headaches. The tumour is diagnosed by CT and MRI scans, but a biopsy required for definitive diagnosis. Treatment involves a combination of surgery, irradiation and drugs. The tumours are usually very sensitive to irradiation, but they may suddenly spread rapidly through the brain. Unfortunately only one in three can be cured.

See also ASTROCYTOMA; CRANIOPHARYNGIOMA; GERSTMANN SYNDROME; GLIOMA; MENINGIOMA; PINEALOMA; RETINOBLASTOMA

MÉNIÈRE'S DISEASE
Ménière’s disease is a syndrome causing dizziness, deafness and a constant noise in the ears.

The inner ear contains both the hearing and balance mechanisms. The latter consists of three tiny semicircular canals full of fluid. The exact cause of the disease is unknown, but there is a build-up of pressure inside the hearing and balance mechanisms, and it may occur after a head injury or ear infection. It is more common in men and with advancing age, but avoiding prolonged episodes of loud noise (eg. jet engines, rock bands) reduces the incidence.

The most distressing symptom is a constant high-pitched ringing noise (tinnitus) in the ear. Patients also have attacks of dizziness and nausea that come and go for no apparent reason, and a slowly progressive and permanent deafness. Other symptoms may include sweating, nausea and vomiting. There are no specific tests that can diagnose the disease.

Reducing the amount of salt in the diet may help in early stages. Numerous medications may be tried including betahistine (increases the blood supply to the inner ear), antihistamines, diuretics, prochlorperazine, amitriptyline and chlorpromazine, but none have more than a 50-50 chance of success. A tinnitus masker is a hearing-aid device that emits a constant tone that counteracts the noise already heard in the ear. Microsurgical techniques involve draining the high-pressure fluid from the affected parts of the inner ear, or as a last resort destroying the auditory nerve, leaving the patient deaf in that ear but without the distressing buzz saw noise. There is no cure and treatment is not very satisfactory, but some cases settle spontaneously.
A MENINGIOMA
A meningioma is a slowly progressive, mildly malignant cancer of the meninges, the membranes that surround and support the brain. The cause is unknown, but they are more common in women and older people and often grow to a large size before symptoms develop. The tumour compresses the brain, causing symptoms that relate to the part of the brain compressed. For example, if the area of the brain controlling the arm is compressed, the arm may become weak or paralysed. Seizures are common. The tumour can be visualised by CT and MRI scans, but a biopsy is required for definitive diagnosis. Surgical removal of the tumour is the only treatment, and most cases can be cured.

See also BRAIN CANCER

A MENINGITIS
Meningitis is a viral (aseptic) or bacterial (septic) infection of the meninges, membranes that wrap all the way around the brain and spinal cord, and act to contain the cerebrospinal fluid in which the brain is supported.

The diagnosis of both types of meningitis is confirmed by taking a sample of cerebrospinal fluid from the lower end of the spine (which is an extension of the brain) and examining it under a microscope for the presence of certain cells and it can be cultured to find the responsible bacteria. Blood tests also show abnormalities.

A VIRAL MENINGITIS
Viral (aseptic) meningitis is a relatively benign condition that may be caught by close contact with someone who has a viral infection, or it may be a complication of diseases such as mumps, glandular fever and Herpes. It causes a fever, headache, nausea and vomiting, tiredness and sometimes muscle weakness or paralysis, and neck stiffness may be present. No specific treatment or prevention available, but bed rest, good nursing, paracetamol, and sometimes medication for vomiting are prescribed. It is rare for there to be any after-effects and patients usually recover in one or two weeks.

A BACTERIAL MENINGITIS
Bacterial (septic) meningitis is caught from people who are carriers of the bacteria, but the victims are usually weak, ill, under stress or have their ability to resist infection reduced in some way. The most common forms of bacterial meningitis is caused by Haemophilus influenzae B (HiB), while the most serious is meningococcal meningitis (caused by Neisseria meningitidis ). It is a much more serious condition, with the severity and symptoms varying depending upon which type of bacteria is responsible. Common symptoms include severe headaches, vomiting, confusion, high fevers, patients become delirious, unconscious and may convulse. Neck stiffness is quite obvious, and patients may lie with their neck constantly extended as though they are looking up. Meningococcal meningitis is accompanied by a bruise-like rash on the skin and inside the mouth. Complications include permanent deafness in one or both ears, damage to different parts of the brain, heart or kidney damage, arthritis and the excess production of cerebrospinal fluid which can put pressure on the brain (hydrocephalus). The worst complication is intravascular coagulation, which involves the blood clotting within the arteries and blocking them.

The treatment of bacterial meningitis involves antibiotics in high doses, usually by injection or a continuous drip into a vein, and patients always require hospitalisation. Patients can deteriorate very rapidly and most deaths occur within the first 24 hours. The overall mortality rate is about 20%, although it is higher in children and with the Meningococcal form. Both common causes of bacterial meningitis can be prevented by a vaccine. The HiB vaccine is routine in childhood, but the meningococcal vaccine is an optional extra childhood vaccine or may be given during epidemics to close contacts of victims. Other forms of bacterial and viral meningitis cannot be prevented.

See also MENINGOCOCCAL MENINGITIS

A MENINGOCOCCAL MENINGITIS
Meningococcal meningitis is an uncommon, serious bacterial infection of the meninges (membranes around the brain) and blood stream (septicaemia). Sporadic outbreaks occur worldwide, usually in winter, but up to 40% of the population carry the responsible bacteria in their nose and throat without any symptoms. Infection is more common in closed communities such as military camps and boarding schools. It affects about one person in every 100,000 every year.

The infection is caused by the bacteria Neisseria meningitidis, which occur in 5 common strains (forms), and several dozen uncommon strains. The C strain is the most serious, while strains M, W and Y are probably next
in severity, but this varies between patients. It is spread by prolonged close contact with a person who has the disease by inhaling their sputum or phlegm in coughs and sneezes.

Symptoms include a high fever, severe headache, vomiting, neck and back stiffness, limb pains, confusion, convulsions and a rapidly spreading bruise like rash that starts on the arms and legs. The rash does not go white with pressure under a glass slide, a symptom that is critical in differentiating Meningococcal infections from other rashes, although there are some other infective rashes that do the same thing. In terminal stages the patient becomes delirious, and goes into a coma. Rarely, abscesses may form in the brain, and pneumonia may develop.

Cultures of blood and/or spinal fluid from the lower back can confirm the presence of the responsible bacteria, then penicillin, or more potent antibiotics, are given by injection as soon as the diagnosis is suspected. The patient should be admitted to hospital for confirmation of the diagnosis, and continuation of antibiotics given through a drip into a vein. Life support in an intensive care unit may be necessary. The infection may be rapidly progressive causing death within hours, but overall 80 to 90% of all cases survive, with only 5% of survivors developing long-term consequences such as epilepsy.

Two vaccines are available. One is against strain C only, but lasts long term, the other prevents four strains of the bacteria, but lasts for only two years. The former can be given to infants, and is now part of most routine vaccination schedules.

See also MENINGITIS; WATERHOUSE-FRIDERICHSEN SYNDROME

MENKE KINKY HAIR SYNDROME
Menke kinky hair syndrome is an inherited disorder of copper absorption into in the body from the gut resulting in sparse kinky hair, brain degeneration, reduced growth and early death. Symptoms may be relieved or delayed by copper injections.

MENTAL RETARDATION
See INTELLECTUAL DISABILITY

MERCURY POISONING
Mercury poisoning (Minamata or pink disease) can be caused by swallowing mercury (Hg) or mercury-containing compounds, or inhaling mercury vapour. Mercury is used in industry in thermometers, batteries, thermostats, dental fillings and chemical processing. Contamination of the food chain (particularly fish) from mercury containing industrial wastes may lead to poisoning in humans, and a serious series of poisonings occurred in Minamata, Japan by this route. Mercury used to be present in medications such as teething powders until the 1950s.

If a large amount of mercury is swallowed at one time symptoms include a metallic taste, thirst, burning in the throat, excessive saliva formation, belly pain, vomiting, bloody diarrhoea, collapse and kidney failure. Inhaling mercury vapour can cause an intractable form of pneumonia. Chronic mercury poisoning, in which small amounts of mercury are swallowed over a long period of time, causes “pink disease” with red and swollen hands and feet, irritability, fever, hair loss and damaged nails. Later problems include tremor, convulsions, brain damage and death. Mercury may cause birth defects in the children of mothers with chronic lead poisoning (Minamata disease).

The presence of mercury can be detected by specific blood and urine tests.

If mercury is swallowed, induce vomiting, give egg whites and milk, then take the patient to hospital for stomach wash out. Mercury can also slowly be removed from the body by some medications in a process known as chelation.

In the eighteenth century mercury was widely used as a medication to treat everything from colic and leprosy to rheumatism and syphilis. It was not particularly efficacious.

MICTURITION SYNCOPE
The medical term for fainting that occurs while passing urine is micturition syncope. It most commonly occurs with a sudden drop in blood pressure with the emptying of the bladder (particularly if arising in the middle of the night) or it may be a sign of excess use of alcohol or a poor blood supply to the brain, and has been associated with the onset of a heart attack, stroke or transient ischaemic attack (TIA).

See also STROKE; TRANSIENT ISCHAEMIC ATTACK
MIGRAINE

Migraine is a form of headache that is usually associated with other significant symptoms. They may occur once in a person's life, or three times a week; may cause a relatively mild head pain, or may totally disable the patient.

They are caused by the contraction of an artery in the brain, which may give the patient an unusual sensation and warning of an attack (aura), followed within a few seconds or minutes by an over-dilation of the artery. Excess blood passes to the part of the brain that the artery supplies and it is unable to function properly. The patient feels intense pressure, pain and other symptoms. The artery dilation may occur for no apparent reason, or be triggered by certain foods, anxiety and stress, hormonal changes, allergies, loud noises or flashing lights. The frequency and severity of migraines tends to decrease with age, an initial attack over the age of 40 is unusual, and they may cease in old age.

The effects vary dramatically from one patient to another, depending on the part of the brain involved. As well as intense head pain, most patients suffer nausea and vomiting and loud noises or bright lights aggravate the pain. Other symptoms may include partial blindness, personality changes, loss of hearing, noises in the ears, paralysis, numbness, and violence. Migraines are rarely serious, but patient may be disabled for some hours or days.

There are no specific diagnostic tests, but doctors can sometimes diagnose a migraine by their visual pattern. If the patient closes their eyes, patterns can be seen on the back of the eyelids which are actually the random activity of the nerves in the light sensitive retina at the back of the eye and in the visual centre of the brain. In normal people, a swirling smooth pattern will be seen, but a patient with a migraine will see flashes of light, bright colours and jagged patterns.

Migraines may be prevented by regular medication, or treated when they occur.

Many different drugs can be taken regularly to prevent migraines including propranolol, methysergide, clonidine, sodium valproate, ketoprofen and pizotifen. It is often a matter of trial and error to find the most effective one.

The longer a migraine has been present, the more difficult it is to treat. They can be rapidly cured in most patients by nose sprays, tablets or injections containing naratriptan, rizatripan, sumatriptan or zolmitriptan. The more often these medications are used, the more effective they become. Other treatments include tablets that may be placed under the tongue or swallowed (eg. ergotamine, isometheptene), or normal pain killers (eg. paracetamol, aspirin), antihistamines, mild sedatives and anti-vomiting medications (eg. promethazine). Tolfenamic acid is an unusual nonsteroidal anti-inflammatory drug that was introduced to some countries in 2002 to treat migraine. Strong narcotic pain killers should be avoided if possible. Resting in a cool, dark room is also helpful.

Most cases can be prevented or effectively treated, but a small number are resistant to all medications.

See also CLUSTER HEADACHE

MILLER-DIEKER SYNDROME

The Miller-Dieker syndrome is a congenital birth defect of the head and brain due to an abnormality localised to chromosome 17. Affected children have abnormal head shape, abnormal facial structure, mental subnormality, growth retardation and low birth weight. Specific analysis of chromosomes is abnormal, as is a CT scan of the brain. It can be diagnosed before birth by taking a sample from the placenta (chorionic villus sampling). There is no treatment or cure, but reasonable life expectancy.

See also INTELLECTUAL DISABILITY

MÖBIUS SYNDROME

Möbius (Moebius) syndrome is a developmental abnormality of cranial nerves. Two nerves from the brain (numbers 6 and 7) fail to develop properly before birth. These children are born with drooping eyelids, inability to move their eyes normally, faces that cannot show expressions, difficulty in swallowing and speaking, constant drooling, but normal intelligence. There is no treatment or cure, but the lifespan is normal. Paul Möbius (1853-1902) was the German neurologist who first described the syndrome.
MOTOR NEURONE DISEASE

Motor neurone disease is a horribly insidious disease of no known cause that affects the nerves that supply the muscles of the body.

Nerves are divided into two main groups - sensory nerves that feel heat, cold, touch and pain; and motor nerves that take the signals from the brain to the muscles and instruct the muscles to contract or relax. Motor neurone (neurone means nerve) disease is a steadily progressive degeneration of the motor nerves in the body, or the areas in the brain that control motor nerves. It normally affects adults between 35 and 70 years of age.

Muscles in various parts of the body become steadily weaker until complete paralysis results, but the muscles affected, and therefore the symptoms, vary between patients. Common symptoms include difficulty in swallowing and talking, drooling of saliva, inability to cough effectively, reduced tongue movement, and weakness of the arms and legs. As the disease progresses, weakness of the muscles required for breathing cause severe shortness of breath occur and lung infections such as pneumonia develop, and often lead to death. Some muscles may go into spasms that cause jerking movements and speech.

Electrical tests of the motor nerves to determine how well they are functioning, and a nerve biopsy are used to confirm the diagnosis.

No cure is available, and treatment is aimed at relieving muscle spasm, assisting feeding, preventing infections, aiding breathing and making the patient as comfortable as possible. The medication riluzole is now being used in some countries to slow the progress of the disease. Physiotherapy on a very regular basis is essential. It is steadily progressive to death within three to five years.

See also AMYOTROPHIC LATERAL SCLEROSIS; WERDING-HOFFMAN SYNDROME

MULTIPLE SCLEROSIS

Multiple sclerosis (MS) or disseminated sclerosis, is an uncommon disease of the brain and spinal cord that interferes with the brain’s ability to control the body.

The cause is not known precisely, but there are several theories. It is possibly due to an unidentified virus, it may be that the body becomes allergic to itself and starts attacking its own cells in an immune response, it may be related to low vitamin D levels in the mother during pregnancy, or it may start as a transverse myelitis. Scattered parts of the brain and spinal cord are damaged at random, the affected areas fail to function properly, and nerve messages from the brain to the muscles do not flow smoothly. Sometimes the message cannot get through at all, and paralysis results, while at other times the message may go to the wrong place, causing abnormal movement or a tremor. MS often attacks people in the prime of life rather than old age, it is more common in women with western European ancestry, and is rare in the tropics between 30°S and 30°N. It is not contagious or preventable.

Symptoms vary greatly from one patient to another, but usually include vision problems, unusual forms of paralysis, tremor, loss of balance, poor coordination, general tiredness and numbness. Patients may experience difficulty in controlling an arm or leg, cannot talk, or may have periods of blindness. Symptoms also change in a patient because damaged tissue can repair itself and start functioning again, while other nerves becomes damaged, causing yet another set of symptoms. Pressure to skin areas and bacterial infections of various organs may occur due to lack of movement.

The diagnosis can be difficult to confirm. Electroencephalograms (EEG), electromyograms (EMG), blood tests (no specific test is diagnostic) and magnetic resonance imaging (MRI) are all used.

There is no effective treatment available, but some medications (eg: beta interferon, steroids) can slow its progress, control acute attacks and bring about remissions. Tizanidine is a muscle relaxant medication used as a tablet to treat muscle spasm caused by multiple sclerosis. Physiotherapists, speech therapists and occupational therapists are also used.

The disease goes through a series of attacks and remissions, and periods of good health between attacks can last for months or years. Most patients can lead independent, active and satisfying lives and take care of their own needs for many years after the diagnosis is made. The life span of victims is not significantly altered.

See also TRANSVERSE MYELITIS

MURRAY VALLEY ENCEPHALITIS

Murray Valley encephalitis (MVE) is a viral infection of the brain spread from water birds to man by a mosquito bite. It is found in the Murray River valley of Victoria and NSW in Australia, and epidemics occur every few years after flooding when both bird life and mosquitoes multiply. The incubation period is three to six days.
Fever and a rash are often the only symptoms, but in severe cases it may cause eye pain, enlarged tender lymph nodes, dizziness, sore throat, joint pains, headache, neck stiffness, nausea, vomiting, tiredness, disturbed mental functions and disorientation, although it rarely causes any long-term problems. Death is rare.

No specific treatment or vaccine is available, and acute attack lasts for one to three weeks.

See also ENCEPHALITIS

MUSCULAR DYSTROPHY

The muscular dystrophies are a diffuse group of diseases, all of which cause some degree of muscle weakness and wasting. Many are inherited, while others may be due to a viral infection.

See also AMYOTROPHIC LATERAL SCLEROSIS; DUCHENNE MUSCULAR DYSTROPHY; ERB MUSCULAR DYSTROPHY; KUGELBERG-WELANDER SYNDROME; INCLUSION BODY MYOSITIS; MOTOR NEURONE DISEASE; MULTIPLE SCLEROSIS; MYASTHENIA GRAVIS; WERDNIK-HOFFMAN SYNDROME

MYASTHENIA GRAVIS

Myasthenia gravis is an uncommon condition characterised by varying weakness of the muscles that control the eyelids, the movement of the eyes and swallowing. Signals from the nerves that supply affected muscles are blocked, for which there may be an immunological cause when antibodies that normally fight off infection, actually attack nerve tissue (autoimmune response). It may occur at any age, but is most common in young women and may be associated with rheumatoid arthritis, systemic lupus erythematosus, thymus and thyroid disease.

Drooping eyelids (ptosis), double vision and difficulty in swallowing are the main symptoms. In severe cases the muscles used in breathing and walking are also affected. Muscle weakness may vary in severity during the day and can disappear entirely for days or weeks before recurring, but over a period of months or years, the attacks become more severe. Unless adequate treatment is obtained, death eventually results from breathing difficulties.

The diagnosis is confirmed by the patient's reaction to an anticholinergic drug, which immediately reverses all the muscle weakness or measurement of the anti-skeletal muscle antibodies or anti-acetylcholine receptor (Anti-AChR) antibody titre.

Treatment involves surgically removing the thymus gland, which is the source of most of the antibodies in the blood, and using anticholinesterase drugs (eg. distigmine, neostigmine) on a regular basis to control the muscle weakness. Steroids can be used in patients who respond poorly to other treatments.

There is no cure and patients require treatment for the rest of their lives, but some have lengthy periods when the disease is inactive, during which they may be able to cease their medication.

See also AUTOIMMUNE DISEASES; EATON-LAMBERT SYNDROME; SYSTEMIC LUPUS ERYTHEMATOSUS

MYXOMA

A cardiac myxoma is the most common form of benign tumour occurring inside the heart. The cause is unknown in most cases, but it may be familial (run in families). The symptoms are very variable depending upon position within the heart, its size and the effect it has on heart function. Non-specific symptoms may include a fever, weight loss, tiredness and loss of appetite, and it can be diagnosed by echocardiography (ultrasound scan). Sudden death due to blockage of a heart valve or irregular heart beat (ventricular fibrillation) is possible. Surgical removal of the tumour is necessary and gives good results if diagnosed early enough.

Myxomas may also occur in other areas of connective tissue including under the skin, in bones and at the back of the abdomen under the peritoneum (retroperitoneal myxoma).

NARCOLEPSY

Narcolepsy is an unusual disorder of the brain's electrical activity that is characterised by sudden uncontrolled episodes of sleep. The cause is unknown, but patients go from wakefulness almost immediately into the deepest type of sleep, known as REM (rapid eye movement) sleep, without passing through the normal intermediate stages. They have sudden periods of sleeping for 5 to 30 minutes several times a day, sudden muscle weakness, hallucinations before and during sleep periods, and paralysis immediately before and during sleep. Patients suddenly fall asleep, sometimes in the middle of a sentence, or when halfway across a pedestrian
crossing. There is a wide range of severity from those who merely appear to sleep excessively, to those who are barely able to function or care for themselves.

The diagnosis is confirmed by an electroencephalogram (EEG) and by observing the patient in a sleep laboratory.

Stimulants such as dexamphetamine or methylphenidate must be used on a regular basis, and patients must not be allowed to drive, swim or operate machinery until they have been well controlled for a long time, as there is the obvious danger that the patient may accidentally harm themselves or others. In many patients, good control of symptoms is quite difficult to achieve.

**NEU-LEXOVA SYNDROME**

The Neu-Lexova syndrome is a rare familial (runs in families) abnormality, but occurs only if both parents carry the abnormal gene. It is characterised by absent eyelids, underdeveloped nose and jaw, abnormal skin structure and texture, swelling of feet and hands, multiple skin contractures and intellectual disability. No treatment is available and the prognosis is poor.

See also INTELLECTUAL DISABILITY

**NEURALgia**

Neuralgia is a pain in any nerve. It may be due to pinching of a nerve between other tissues (eg. between ribs with prolonged coughing), a reduced blood supply to a nerve (eg. migraine, diabetes), an infection of the nerve (eg. shingles), an injury to the nerve, or joint arthritis (eg. back arthritis causing sciatica).

Patients experience sudden, severe, shooting and often brief stabs of pain that may occur anywhere, but the chest, face and arms are more frequently affected. Permanent damage to the nerve may cause loss of sensation to the affected area.

The cause must be investigated by x-rays, and sometimes blood tests. The treatment will depend on the cause. Anti-inflammatory drugs can reduce inflammation associated with muscle strain and arthritis, and steroids can be given as injections into the damaged area. Physiotherapy is often useful.

See also TRIGEMINAL NEURALGIA

**NEURITIS**

Any form of nerve inflammation may be referred to as neuritis. It is often associated with neuralgia and/or paraesthesia (a pins and needles sensation).

**NEUROFIBROMA**

A neurofibroma is a benign fibrous tumour that arises from nerve tissue. They form hard lumps anywhere on or in the body that can be surgically removed if they cause symptoms or are unsightly.

**NEUROLEPTIC MALIGNANT SYNDROME**

The neuroleptic malignant syndrome is a very rare (under 200 cases reported) complication of medication used for severe psychiatric disturbances. It may occur early or late after starting to use major tranquillisers (eg. phenothiazines, butyrophones, thioxanthenes, loxapine) at normal dosages.

Patients develop a very high fever, muscular rigidity, tremor, abnormal sweating, confusion and loss of consciousness. Rhabdomyolysis (muscle meltdown) may also occur. Some blood tests may be abnormal, but none are specifically diagnostic.

The only treatment is ceasing the medication and support in a hospital intensive care unit. Numerous medications have been tested to help patients, but none proven effective. Death occurs in 20% of patients, but early treatment improves mortality.

**NEUROSYPHILIS**

See SYPHILIS
NIEMANN-PICK DISEASE
Niemann-Pick disease is a rare congenital (present from birth) disease is caused by an inability of the body to process some types of fat due to the lack of an enzyme in the liver. It causes enlargement of the liver, failure to gain weight after birth, and retarded mental development.

There are several different forms of the disease, which vary in their severity, depending on the precise enzyme deficiency. The disease can be diagnosed by specific blood and bone marrow tests, but there is no treatment available.

See also INTELLECTUAL DISABILITY

OPTIC ATROPHY
Optic atrophy (wasting) can be seen on ophthalmoscopic examination of the retina at the back of the eye. A pale optic disc with blurred margins is noted. Disc cupping may also occur, and the patient may complain of reduced visual acuity and reduced visual field (width of vision).

The causes of optic atrophy include glaucoma, arteriosclerosis, retinal ischaemia, optic neuritis, Paget's disease, tumour pressing on optic nerve, retinitis pigmentosa, vitamin B deficiency and methanol poisoning. These conditions all cause a poor arterial blood supply to, or chronic inflammation of, the optic disc.

ORGANIC BRAIN SYNDROME
The organic brain syndrome is deterioration in the higher functions in the brain, which is always due to a specific cause such as alcoholism, drugs (prescribed and illegal), cancer or infection anywhere in the body, body chemistry or hormone disorders, autoimmune diseases or epilepsy.

The symptoms may include emotional disturbances (eg. anxiety, depression), poor memory and logic, disorientation, inappropriate sensations, an inability to recognise their own body parts and behavioural changes (eg. exhibitionism, aggression). Extensive investigations may be necessary to determine the cause.

The cause is treated when found, and the patient is given medications to control symptoms, nursed in a supportive environment, and given counselling and behavioural therapy. The symptoms may be persistent unless the cause is treated and reversed early.

See also ALCOHOLISM; ALZHEIMER DISEASE; DEMENTIA

OSTEOGENESIS IMPERFECTA
Osteogenesis imperfecta (Adair-Dighton syndrome) is a rare disease, commonly known as brittle bone disease in which a child is born with fragile, brittle bones that break easily and heal poorly. The condition tends to run in families in an irregular pattern, and varying degrees of severity are possible, with some patients being far more severely affected than others.

These children suffer multiple painful fractures that take months to heal and often leave a permanent deformity. Other characteristics include deafness, a blue colour to the whites of the eyes, spinal deformities and teeth defects. It is diagnosed by X-rays, which show a typical appearance in the long bones of the legs and arms.

No treatment available and many patients die before puberty, but if they survive to adult life, the disease tends to become less severe, and a relatively normal life expectancy is possible.

OTITIS MEDIA
Otitis media is a bacterial infection of the middle ear.

The middle ear is a cavity that contains three tiny bones that transmit the vibrations of the eardrum to the hearing mechanism in the inner ear. There is a small tube (the Eustachian tube) connecting the middle ear to the back of the nose, and infection can enter the middle ear from there. Infection can also spread from the outer ear to the middle ear. Children are more commonly affected than adults.

Patients experience a sudden onset of severe pain, often at night, and a fever. Pressure on the outside of the ear causes additional pain and relative deafness. Antibiotics and medications are prescribed to dry up phlegm, but it is sometimes necessary to perform a small operation on the eardrum to relieve the pressure. If left untreated or there is rapid worsening of the infection, the bulging eardrum may burst, and blood and pus will ooze out of the ear canal. The pain may be relieved by rupture of the eardrum, but treatment with antibiotics is essential to ensure that the eardrum repairs itself. If the hole in the eardrum fails to heal after several months, it
may be necessary to have an operation to repair it. Rarer complications include a spread of the infection into the surrounding bone (mastoiditis), or into the bloodstream or brain.

Patients get very good results with appropriate treatment, and a ruptured eardrum usually heals in one or two weeks.

**PAGET’S DISEASE OF BONE**

Sir James Paget (b.1814) was a London surgeon after whom a number of diseases, tests, tissues and structures were named. In Paget's disease of bone (osteitis deformans), bone in scattered parts of the body becomes thickened and soft, causing compression of nerves and collapse of those bones that support weight. There is no known cause, but it is unusual under 60 years of age.

The disease has a very insidious onset, and may be quite advanced before diagnosis, and can vary from very mild to rapidly progressive. The skull, thighbone (femur) and shinbone (tibia) are often involved, giving a characteristic head appearance and bowing of the legs as they bend under the body's weight. Skull enlargement causes pressure on nerves and constant headaches, while fractures may occur in long bones with only slight injury, and the back becomes bent, painful and deformed.

The diagnosis made on the characteristic X-ray appearance, and blood tests show specific chemical imbalances, including excess calcium. The excess calcium may cause kidney damage, extra blood flow to the bones can cause circulatory and heart problems, and a small number of patients develop a form of bone cancer.

Medications such as disodium etidronate, disodium pamidronate or tiludronate are taken regularly for the rest of the patient's life to control the disease. The injection salcatonin may also be used in some patients. Drugs slow the disease progress, but there is no cure, and the earlier it occurs in life, the more likely it is to be severe.

**PANCOAST SYNDROME**

Pancoast syndrome is a complication of lung cancer that occurs in patients with cancer in the top part of the lung, and is a sign that the cancer is progressing very rapidly. As with other forms of lung cancer, smoking is by far the most common cause, but asbestos exposure and toxic fumes may also be responsible. A chest x-rays may show the cancer, but it may be hidden by the shoulder blades, and a CT scan is often necessary to reveal the tumour.

The syndrome symptoms consist of shoulder, arm and chest pain with an associated Horner syndrome (a drooping eyelid, lack of sweating on one side of the face and a contracted pupil in the eye). Death usually follows soon after the syndrome is diagnosed as the diagnosis is usually made late.

**PARINAUD SYNDROME**

The most common initial presentation of patients with the Parinaud or pretectal syndrome is that they are unable to look upwards without tilting their head back - the muscles that pull the eyes upwards are paralysed. Other symptoms include abnormal reaction of the pupils to light and other abnormalities of eye function.

The cause is a tumour of the pineal gland (in the front of the brain) or brain that puts pressure on one of the nerves that control eye movement, or a stroke that affects the part of the brain that controls this nerve.
PARKINSON DISEASE

Parkinson disease, which is also known as paralysis agitans, Parkinsonism and shaking palsy, is one of the more common causes of a tremor in elderly people. It is named after the English physician James Parkinson (b.1755) who first described the condition in the medical literature.

The cause is the presence of Lewy bodies which are tiny abnormal spheres found in nerve cells in the brain. They alter the function of the cell and can be seen when affected cells are examined under a microscope. Why they occur in nerve cells is not known, but what happens in the brain to cause the symptoms is understood. When a muscle contracts, the opposite muscle must relax. For example, when you bend your finger, the muscles on the palm side of the finger contract, while those on the back of the finger must relax. This coordination occurs in the brain. In Parkinsonism, the brain cells that control this coordination have degenerated so that smooth control of movement is lost.

Early signs of the disease are a failure to swing the arm when walking, deterioration in handwriting, and poor balance. Later symptoms are a constant tremor, general body stiffness, loss of facial expression, a stiff way of walking and lack of coordination. The intelligence and mental powers of victims are not affected in the early stages of the disease, and this causes great frustration, particularly when speech may be impaired. Patients may become depressed, anxious and emotionally disturbed.

No blood or other test is diagnostic. CT scans (special x-rays) may reveal changes in certain parts of the brain, as may electroencephalograms (EEG), which measure the electrical brain waves. Magnetic resonance imaging (MRI) and positron emission tomography (PET) scans are now being used in some centres.

A number of drugs (eg. amantadine, levodopa, pergolide) are available to control the symptoms and slow the progression of the disease, but it is a matter of trial and error to determine which medications will help any particular patient. None of them cure Parkinson disease - they aim to control it. Levodopa acts to replace the missing chemical in the brain that causes the disease. It is sometimes combined with other medications that increase the effectiveness of the levodopa and reduce side effects (eg. pergolide). Bromocriptine also acts to control Parkinson disease. It is, strangely enough, also used to stop the production of breast milk and treat a bone overgrowth disease called acromegaly. Side effects include a reduced tolerance to alcohol. Other medications that may be used in treatment include pramipexole, ropinirole, selegiline and procyclidine.

Physiotherapy is also very important. In rare cases, brain surgery, in which part of the brain is destroyed in an attempt to block nerve pathways that cause the constant tremor, is performed.

There is no cure, but medications allow some patients to lead normal lives. The disease process progresses steadily over many years, rarely causing death, but causing otherwise normal people to become invalids, totally dependent on others for everyday tasks.

See also STEELE-RICHARDSON-OLSZEWSKI SYNDROME

PAROTID TUMOUR

The parotid glands sit under the angle of the jaw on each side to secrete saliva into the back of the mouth. Several different types of tumour can develop in this gland, and the other salivary glands (submandibular and submental glands) that sit under the chin. The most common form is called a mixed parotid tumour, which is not malignant (cancerous).

The tumour appears as a slowly enlarging painless lump at the angle of the jaw, but as it enlarges the tumour puts pressure on surrounding tissue and nerves to eventually cause discomfort and pain. The diagnosis is often difficult, but can be made by a CT or MRI scan and needle biopsy (sticking a needle into the gland to take a tissue sample).

Extremely intricate surgery is necessary to remove the enlarged gland and the tumour it contains. If the tumour is cancerous, irradiation of the area to prevent a recurrence is necessary. The nerve supplying the face runs through the parotid gland, and it is very easy to damage this during surgery, leaving the face numb and paralysed. The prognosis is generally very good, but if the tumour is cancerous it is far more serious.

PAROXYSMAL VENTRICULAR TACHYCARDIA

A paroxysmal ventricular tachycardia is a sudden very rapid heart rate, often due to a heart attack. It is accompanied by chest tightness, palpitations, shortness of breath and collapse. It may progress to ventricular fibrillation, which is usually fatal.

An ECG (electrocardiograph) will show the abnormal rhythm. Medications (eg. lignocaine) are injected into a vein followed by cardioversion (electric shock), stabilisation in hospital, and then tablets to prevent a recurrence. The outcome depends on cause and severity.
PARSONAGE-TURNER SYNDROME
See BRACHIAL AMYOTROPHY

PATAU SYNDROME
Patau syndrome (trisomy 13-15) is a rare congenital defect affecting numerous parts of the body, caused by the presence of three copies of chromosomes 13 and 15 instead of two. The infant has extra fingers and toes, abnormal heart structure, cleft lip and palate, small eyes and brain malformations. Tests are performed on heart and brain function (eg. CT scan, echocardiogram) to confirm the diagnosis. No treatment is available, and the prognosis is poor.

The incidence of Patau syndrome increases with the age of the mother.

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PELLAGRA
Pellagra is caused by a lack of nicotinic acid (niacin or vitamin B3) in the diet. Nicotinic acid is essential for the normal functioning of the body, and is found in many foods including rice, meats, vegetables and dairy products. Pellagra occurs in countries where the diet is primarily corn, which has minimal amounts of niacin.

In the early stages patients have a poor appetite, general weakness, irritability, sore mouth, fissured tongue and weight loss. More advanced cases develop dermatitis, diarrhoea, and become demented to the point where severe psychiatric disturbances occur in advanced cases.

The diagnosis can be confirmed by blood tests, then the disease is easily cured by vitamin B supplements given by mouth. If left untreated, death will eventually occur.

PERIPHERAL NEUROPATHY
Peripheral neuropathy is abnormal nerve function in the arms and legs. There are many causes including toxins, poisons, alcoholism, poor blood supply, poorly controlled diabetes mellitus, kidney failure, vitamin deficiencies, liver failure (eg. cirrhosis) and an underactive thyroid gland.

Patients feel tingling and burning sensations in the hands and feet, other abnormal sensations, and have abnormal reflexes. Clumsiness and difficulty in walking, muscle wasting and weakness may occur.

Electromyography (EMG) measures the electrical activity and function of muscles and can be used to confirm the diagnosis. Numerous clinical tests can also be performed to check sensation, and blood and other tests are undertaken to find the cause. Appropriately treating the cause may slow the progress of the disease, or reverse its affects.

The prognosis depends on the cause, its severity and response to treatment.
See also ALCOHOLISM; CIRRHOSIS; WERNICKE-KORSAKOFF PSYCHOSES

PERNICIOUS ANAEMIA
Pernicious (megaloblastic or macrocytic anaemia) is due to a lack of vitamin B12 (cyanocobalamin), which is essential for the formation of haemoglobin, the red oxygen-carrying substance in red blood cells. For vitamin B12 to be absorbed from the stomach and into the blood a substance called intrinsic factor is required. Patients with pernicious anaemia lack intrinsic factor and therefore develop a lack of vitamin B12, which in turn leads to an inability to produce haemoglobin. It is common in middle-aged women and is given its name because of its very gradual and “pernicious” onset over many years.

In addition to tiredness and pallor, patients have a smooth and sore tongue, indigestion, lack of appetite, and occasionally jaundice (yellow skin). Atrophic gastritis may be a complication.

It is diagnosed by seeing characteristic large red blood cells (megaloblasts or macrocytes) in a blood film, measuring the level of vitamin B12 in a blood sample and the presence of parietal cell autoantibodies.
Regular injections of vitamin B12 are necessary for the rest of the patient's life. The injections may be weekly at first, then reduced to one every two or three months. Although pernicious anaemia cannot be cured, it can be very effectively controlled. Untreated, the disease is fatal.

**PERONEAL MUSCULAR ATROPHY**
Peroneal muscular atrophy (or dystrophy) is an inherited condition characterised by weakening of the muscles in the outer part of the lower leg that control the shape and position of the foot. The cause is a progressive deterioration of the nerve supply to the affected muscles that often starts in the teenage years. The arch of the foot becomes raised (claw foot), control of the ankle joint is weak and the patient has an awkward high stepping gait. Leg braces and corrective surgery may be necessary in the latter stages of the disease.

**PHAEOMOCYTOMA**
A phaeochromocytoma is a rare black-celled tumour in the adrenal glands (which sit on top of each kidney), which releases a substance into the blood stream that causes very high blood pressure (hypertension). It is sometimes a hereditary tendency, but most arise for no apparent reason.
Patients have extremely high blood pressure, severe headaches, palpitations of the heart, abnormal sweating, nausea and vomiting, abdominal pains, blurred vision, and brain damage that may result in loss of speech, blindness or unconsciousness. Other symptoms may include increased appetite, nervousness and irritability, shortness of breath, weight loss, light-headedness and chest pain (angina). Some patients have multiple tumours in other parts of the body, and an unexplained sudden death may be due to a heart attack caused by an undiagnosed tumour. Some forms are associated with cancer, but a phaeochromocytoma is not a cancer itself.

The diagnosis is confirmed by special blood tests that measure excessive levels of catecholamines (the chemical released by the tumour). A CT scan or a magnetic resonance imaging scan (MRI) is performed to locate the tumour.

Controlling the high blood pressure with medication is the initial aim of treatment, and then surgically removing the tumour. Long-term management with medication, but without surgery, is not practical.

The prognosis depends on the damage caused by the high blood pressure before diagnosis, and how many tumours are present. If the tumour is removed early, a complete recovery is expected. Without treatment, the disease is invariably fatal, and even in the best medical centres, a small percentage of patients will die from complications of the disease or the surgery.

**PHENYLKETONURIA**
Phenylketonuria (PKU) is an uncommon congenital (present from birth) metabolic (body chemistry) disorder that may have serious consequences. The disease that runs in families from one generation to the next, and is more common in people of Scottish or Irish descent (1 in 5000 children), but extremely rare in Negroes (1 in 300,000).

A baby with PKU cannot tolerate foodstuffs that contain the amino acid phenylalanine, and a build-up of this in the blood causes brain damage, intellectual disability, epilepsy, behaviour problems and eczema. As a result, every baby in western countries is routinely tested a couple of days after birth using a single drop of blood taken from a heel prick. If the condition is diagnosed, further blood tests are performed regularly to ensure that the amount of phenylalanine does not rise above normal.

A protein free diet from before two months of age until the patient is at least eight years old is essential. Phenylalanine is found in most proteins, but other amino acids (the building blocks of protein) can be provided in a special milk formula. A correct diet started early will prevent the disease from damaging the brain, and the child will grow up to be a normal adult with normal intelligence.

See also INTELLECTUAL DISABILITY

**PICK DISEASE**
Pick disease is similar to Alzheimer disease, except that only one lobe of the brain (usually the frontal or temporal lobe) wastes away (atrophies) rather than the brain as a whole. The cause is unknown, but it may be familial (passed from one generation to the next) in some cases.
Patients experience slowly progressive dementia, excessive use of the mouth in a baby-like manner, deliberate vomiting after meals, disturbances in speech and use of words, loss of normal social inhibitions,
irritability and persistent aimless wandering. A CT scan shows degeneration of part of the brain. No treatment is available and there is a slow inevitable progression.
See ALZHEIMER DISEASE; DEMENTIA

PINEALOMA
A pinealoma is rare benign or cancerous tumour of the pineal gland, which acts as the body’s clock and sits underneath and between the frontal lobes of the brain. Patients develop a headache, difficulty in looking upwards, personality changes, nausea and tiredness. An increase in the pressure of cerebrospinal fluid that surrounds the brain due to the tumour may cause epileptic fits. The growth is diagnosed by CT and MRI scans, and treatment involves surgical removal or irradiation of the tumour. The prognosis is good if the tumour is benign, but poor if cancerous.

PINEAL TUMOUR
See PINEALOMA

PITUITARY TUMOUR
Tumours of the pituitary gland are usually adenomas. They cause abnormalities in the production of the numerous controlling hormones released by the pituitary gland, and the symptoms depend on the hormonal imbalance caused by the tumour. Many very small incidental tumours without symptoms may be found in older people using MRI or CT scans - these require no treatment.

Pituitary tumours may also put pressure on adjacent structures as they enlarge. The optic chiasma is a point at which the nerve fibres carrying the visual signals from the eye to the brain cross over, and this lies just in front of the pituitary gland. Pressure on these nerves can cause abnormalities in the field of vision that can be another presenting symptom of a patient. Other effects can be paralysis of eye movement, abnormal body temperature regulation and headaches with pressure on the nerves and brain beside the pituitary gland.

A sudden bleed into a pituitary tumour can cause a rapid increase in its size and a condition known as pituitary apoplexy. This has symptoms of severe sudden onset headache, vomiting and coma.

Pituitary tumours can be treated by medications such as bromocriptine to correct some hormonal imbalances, but are better treated by surgery to remove the tumour. Interestingly this can be performed relatively easily through the nose as the pituitary gland is situated just behind the upper part of the nasal cavity. Special instruments are used to penetrate the bone between the nasal cavity and the pituitary gland and then to remove the tumour and sometimes the entire gland. Irradiation of the tumour is another option.

After treatment the patient may suffer from hypopituitarism and it is usually necessary to take replacement hormones on a regular basis by tablet, injection or both.
See also HYPOPITUITARISM

POLIO
Polio (the full name is poliomyelitis) is a generalised viral infection, which passes from one person to another through droplets in the breath or by touch, and attacks muscles. It has been eradicated in developed countries, but still occurs in some poorer countries.

The symptoms are severe muscle spasm followed by paralysis and muscle contractures. If the muscles of breathing or the heart are affected, the patient may die or remain on a respirator for life. It is diagnosed by specific blood tests.

No treatment is available other than general physical support and muscle relaxants, and the overall prognosis is poor. Many patients recover, but most of them have significant disabilities.

Oral (Sabin) and injected vaccines are available. They are extremely effective, safe, and have no significant side effects.

POLYCYTHAEMIA RUBRA VERA
Polycythæmia rubra vera (the "rubra" is sometimes omitted) is an excessive production of red blood cells that is most common in middle-aged to elderly, overweight men, but may occur in both sexes. It is rare under 40 years of age.

Red blood cells are made in the spleen and bone marrow, primarily of the breastbone (sternum), pelvis and thighbone (femur). If the marrow becomes overactive, excessive numbers of cells may be produced, and the
patient develops a headache, dizziness, tiredness, blurred vision, generalised itching, noises in the ears, high blood pressure and an enlarged spleen.

Blood clots may occur in vital organs (eg. brain to cause a stroke), and some patients develop a form of chronic leukaemia.

The diagnosis can be confirmed by finding excess red blood cells in a blood test, and further tests on bone marrow determine the severity of the disease.

The disease can be controlled, but not cured, by draining large quantities of blood out of a vein initially, and smaller amounts on a regular basis long term. Medications (eg. busulfan) to reduce the activity of the bone marrow may also be used. The average survival time after diagnosis is twelve years.

**POLYMYOSITIS**

Polymyositis is the inflammation of many muscles in varied parts of the body. It is usually accompanied by swelling (oedema), pain, muscle tenderness, weakness and sweating. The cause is often never found, but it may be due to a viral infection, autoimmune response or metastatic cancer. If the skin is also involved it is called dermatomyositis.

See also AUTOIMMUNE DISEASES; DERMATOMYOSITIS

**POMPE SYNDROME**

See GLYCOGEN STORAGE DISEASES

**PORPHYRIA**

See ACUTE HEPATIC PORPHYRIA; PORPHYRIA CUTANEA TARDA

**PORPHYRIA CUTANEA TARDA**

Porphyria cutanea tarda (latent hepatic porphyria) is the most common form of porphyria, which are a group of liver diseases. It is usually inherited, but may be triggered by some poisons (eg. dioxin), and occurs in all races but is more common amongst the Bantu tribes of Africa.

Patients have skin that is very sensitive to sunlight, with skin thickening and pigmentation occurring in sun exposed areas such as the face and forearms. The urine has a strange characteristic in that it turns a dark purple colour, then brown, if left standing. Liver damage may occur, and may progress to liver failure or liver cancer (hepatoma). It occurs in varying degrees of severity from so mild that it is undetected to a rapidly fatal form.

It is diagnosed by special blood tests, and treatment involves careful genetic counselling of families and the use of a complex drug regime. It may be controlled, but not cured, and is rarely fatal.

See also PORPHYRIA

**POSTERIOR VITREOUS DETACHMENT**

The ball of the eye behind the lens and in front of the retina is filled with a jelly-like substance called vitreous humour. It is this that gives the eyeball its firmness and develops its spherical shape during foetal growth. The vitreous is attached to the retina at the back of the eye.

With ageing the structure of the gel that constitutes the vitreous humour slowly degenerates and the vitreous may detach from the retina at the back of the eye. Patients experience minimal symptoms in most cases but may see flashing lights at the edge of their vision or may see a “floater” in their vision. A floater is a collection of cells or protein in the thick fluid that fills the eyeball, which casts a shadow on the light-sensitive retina at the back of the eye. These symptoms usually settle in a few weeks but it is essential for patients to be assessed by an ophthalmologist to assess them for damage to the retina.

As the vitreous detaches from its adhesion to the retina it is possible in about 10% of patients for the retina to tear and also separate from the back of the eyeball. This causes much more significant visual disturbances that require urgent medical management.

If a posterior vitreous detachment is detected by examining the eye through a slit lamp or ophthalmoscope, and there is no sign of a tear in the retina or retinal detachment, and over the next few weeks the flashes and floaters disappear, then no treatment is necessary as it is a normal part of the ageing process and it is unlikely that there will be any further visual problems. If the floaters or flashes worsen or recur, further specialist assessment is necessary.
POSTNATAL DEPRESSION

Postnatal depression (PND, postpartum depression or the baby blues) is a spontaneous form of depression that occurs in some women just before, or soon after childbirth, and is a response to the effect on the brain of sudden changes in hormone levels.

In its mildest form most women have some feelings of up and down emotions with teary episodes in the first week after delivery. This is normal and passes within a few days.

Women with true postnatal depression experience constant unhappiness for which there is no reason. They are unable to sleep, lose appetite and weight, and feel there is no purpose in living. They may feel unnecessarily guilty, have a very poor opinion of themselves, feel life is hopeless, find it difficult to think or concentrate, worry excessively about their infant or neglect the child. Rarely it may lead to attempted or actual suicide. It is diagnosed after careful psychiatric assessment.

Emotional and practical support from the partner, family and friends are vital in assisting an affected woman in her recovery. Counselling and support groups may also be beneficial. If necessary medications are prescribed to control the production of depressing chemicals in the brain (eg. fluvoxamine, moclobemide, nefazodone, paroxetine, venlafaxine) while hospitalised or given intensive home support. About one in every 500 mothers are hospitalised for postnatal depression. Shock therapy (electroconvulsive therapy - ECT) may be used as a last resort for those women whose depression is prolonged and life threatening. Virtually all cases settle with support and medication in a few weeks.

See also DEPRESSION

POSTPARTUM DEPRESSION
See POSTNATAL DEPRESSION

POST-POLIO SYNDROME

The post-polio syndrome occurs in some survivors of polio, 30 to 40 years after recovery from the infection, and may be due to the overuse of muscles previously damaged by polio. They develop unaccustomed tiredness, joint and muscle pain and muscle weakness. Because of muscle weakness, more stress is put on joints, and arthritis of major joints and in the back may also occur. In a small number of severely affected patients, difficulty in breathing may lead to sleep apnoea and the necessity for breathing aids. There is no cure, and the condition persists long term, but anti-inflammatory medications and steroids may help.

See also POLIO

POST-TRAUMATIC STRESS DISORDER

PTSD is not new, as a century ago the post-traumatic stress disorder was known as shell shock. It occurs after experiencing a situation that causes extreme stress and a feeling of horror and helplessness (eg. armed hold-up, serious accident, war violence, being assaulted or raped, observing atrocities etc.). Most symptoms start between two weeks and three months of the triggering catastrophe, but may start as late as six months.

There are no specific diagnostic tests, but patients must have at least one symptom from each of the following categories to be diagnosed:-

General:-
- Symptom duration more than one month, with significant distress or inability to function normally in society.
- Re-experiencing phenomena
- Experience intrusive recollections, nightmares, flashbacks as if the event was recurring, psychological distress on exposure to cues that may trigger memories, or physiological effects (eg. rapid pulse, rapid breathing) on exposure to cues

Avoidance behaviour:-
- Avoiding thoughts, feelings or conversations about the incident
- Avoiding places, people or activities connected with the incident
- Selective amnesia about the traumatic event
- Reduced interest in everyday activities or detachment from others
- Unable to look forward to future events with pleasure
- Abnormal personality compared to before the incident
Excessively aroused:-
- Insomnia, irritability, anger, poor concentration, increased vigilance or increased startle response to frights.

Treatment involves psychological counselling and debriefing immediately after the event, and a trained counsellor should follow up the victim for at least six months. Normal work and activities should be resumed as soon as possible. Referral to a psychiatrist is necessary if the patient does not appear to recover within six months, or deteriorates sooner, when medication may be necessary.

PTSD may become chronic and lead to recurrent minor illnesses, poor physical health, and in extreme cases, suicide, but in most cases it usually settles within 3 to 6 months.

**POSTURAL HYPOTENSION**

Postural or positional hypotension is a sudden, brief drop in blood pressure (hypotension) caused by the altered relative positions of the brain and the heart when the patient moves from lying to sitting or standing. The heart must pump harder, and the arteries must contract more to maintain blood flow by means of the blood pressure to a brain that is 30 cm above the heart, rather than one that is at the same level as the heart. It takes a few seconds for this adjustment to be made, particularly in patients who have hardening of the arteries (atherosclerosis).

There are many causes including medications (eg. diuretics, sedatives, levodopa, barbiturates, antidepressants), heart failure, heart valve disease, Parkinson disease, a stroke, diabetes, atherosclerosis (hardening of the arteries), connective tissue disorders, amyloidosis, alcoholism, and of course medications that lower blood pressure. Many of these problems are associated with advancing age.

The patient experiences light-headedness and dizziness, and sometimes blackouts or faints, when rising quickly from lying or sitting to a standing position. Fainting and falling may cause serious injuries.

The condition is diagnosed by measuring the blood pressure when the patient is lying, and again immediately after standing. A drop in blood pressure of at least 20 mm Hg is necessary for the patient to experience symptoms. There is usually a rise in the heart rate as the blood pressure drops, but if the heart rate remains the same, the symptoms are worse and the cause is more likely to be due to nerve or brain damage.

The doctor should advise elderly patients to be slow in changing position, to slow down activities, avoid being overheated (eg. hot bath), avoid alcohol, elevate the head of the bed about 10 cm., and wear support stockings to prevent blood pooling in the legs. Medications should be reviewed to detect any that may be responsible. Drinking two cups of coffee in the morning may be helpful. In severe cases the medication fludrocortisone may be used.

The prognosis depends on the cause, but the problem may be difficult to control.

See also FAINT

**POTASSIUM WASTAGE SYNDROME**

The potassium wastage syndrome is a serious abnormality of blood and urine chemistry. It may be due to Fanconi syndrome, Conn syndrome, Cushing syndrome, chronic kidney (renal) failure and rarely a side effect of medication. Sometimes no specific cause can be found. Patients pass a large amount of very dilute urine and have muscle weakness. Heart rhythm abnormalities and heart attack may also occur. The urine levels of potassium are very high while blood levels are very low, and the blood is abnormally alkaline. It is necessary to treat the underlying cause and give potassium supplements. It may be fatal if left untreated but there is a good prognosis if the cause can be identified and adequately treated.

**PRADER-WILLI SYNDROME**

The Prader-Willi syndrome is a rare congenital (present since birth) brain condition caused by a chromosomal defect that affects only boys.

The syndrome is characterised by a small infant who develops into an obese child due to compulsive overeating from an abnormality in the part of the brain that controls hunger.

The child is usually short, has underdeveloped genitals, is mentally retarded, muscles are weak and have very poor tone, and the belly is very flabby. There is no effective treatment, and they tend to develop diabetes later in life.

See also INTELLECTUAL DISABILITY
PRE-ECLAMPSIA AND ECLAMPSIA

Eclampsia (toxaemia of pregnancy) is a rare but very serious disease that occurs only in pregnancy. In developed countries it is very uncommon, because most women undertake regular antenatal visits and checks. Pre-eclampsia is a condition that precedes eclampsia, and this is detected in about 10% of all pregnant women. The correct treatment of pre-eclampsia prevents eclampsia.

The exact cause of pre-eclampsia is unknown, but it is thought to be due to the production of abnormal quantities of hormones by the placenta. It is more common in first pregnancies, twins and diabetes. Pre-eclampsia normally develops in the last three months of pregnancy, but may not develop until labour commences, when it may progress rapidly to eclampsia if not detected.

The early detection of pre-eclampsia is essential for the good health of both mother and baby. Doctors diagnose the condition by noting high blood pressure, swollen ankles, abnormalities (excess protein) in the urine and excessive weight gain (fluid retention). Not until the condition is well established does the patient develop the symptoms of headache, nausea, vomiting, abdominal pain and disturbances of vision.

If no treatment is given, the mother may develop eclampsia. This causes convulsions, coma, strokes, heart attacks, death of the baby and possibly death of the mother.

Pre-eclampsia is treated by strict rest (which can be very effective), drugs to lower blood pressure and remove excess fluid, sedatives, and in severe cases, early delivery of the baby. The correct treatment of pre-eclampsia prevents eclampsia, and the prognosis is very good if detected early and treated correctly.

PRETECTAL SYNDROME

See PARINAUD SYNDROME

PROTEUS SYNDROME

Proteus syndrome is a rare congenital bone and tissue growth abnormality that is also know as the elephant man deformity, after Joseph Merrick, who suffered from this condition in the 19th Century.

Excessive and unequal bone growth in face, arms, legs and hands results in severe facial and body disfigurement, bony growths on the skull that cause a great increase in skull circumference, and wrinkled bumps on feet, face and hands. X-rays show the abnormal bone growth.

Plastic surgery helps some deformities, and there is near normal life expectancy, but deformities persist.

Proteus was one of the ancient Greek gods who had the ability to change his form.

PSYCHOGENIC

The term psychogenic refers to any symptoms that are created by abnormal emotional conditions of the mind (psyche) rather than having a physical cause (eg. psychogenic pain is pain that has no physical explanation).

PSYCHOSES

Psychiatric conditions are divided into two broad classes, neuroses and psychoses. In a psychosis, the patient has no insight into the fact that they are mentally disturbed.

The most severe form of psychosis is schizophrenia, while others include paranoid disorders and manic-depressive states. A patient with a psychosis is described as being psychotic.

See also SCHIZOPHRENIA

PULMONARY EMBOLISM

A pulmonary embolism occurs when a blood clot or other substance (embolus - eg. fatty plaque from high cholesterol levels in the blood) travels through bloodstream and the pulmonary artery to a small artery in the lung, which it then blocks. The tissue beyond the blockage dies (pulmonary infarct).

Blood clots may occur in the veins of leg muscles (deep venous thrombosis), but may also arise in other parts of the body. They travel through veins to the right side of the heart, and then into the lungs where they cut off the blood supply to a segment of lung, which will collapse and die. Emboli are more common after major surgery, in patients who are bedridden for long periods, and in the elderly.

The symptoms may include chest pain, shortness of breath, coughing of blood, fainting, heart rate increases, and a fever. Increased back pressure of blood on the heart may lead to right heart failure (cor pulmonale), and an extending clot can cut off more arteries in the lung, and destroy a larger area of lung tissue.
The condition is diagnosed by a chest X-ray, ventilation-perfusion (V/Q) scan, CT scan or angiography. Blood tests can show signs of clotting within the body, and an electrocardiogram (ECG) shows strain on the heart. Specialised tests of lung function are sometimes necessary, and an X-ray in which dye is injected into the veins and can be seen moving through the arteries in the lung may be performed in cases of doubt.

Treatment must start as soon as possible to prevent extension of the clot and further damage to the lung. Anticoagulants drugs that prevent blood clotting (eg. heparin) are initially given as an injection, and later as tablets (eg. warfarin, aspirin). Regular blood tests are performed throughout treatment with anticoagulants to check the dosage required. Anticoagulant therapy is continued for some months after the attack, but in high-risk patients it may be continued for life. In severe cases thrombolytics (clot dissolving drugs) are injected directly into the involved veins. In rare circumstances, surgery to remove the clot from the lungs or leg is undertaken, or a filter is inserted surgically into the main vein of the body leading from the legs to the heart, to filter out any blood clots that may form in the future. Blood clots in the legs can be prevented by using pressure stockings during long operations, early mobilisation after surgery, physiotherapy to keep leg muscles active, and elevation of the legs in bed-bound patients.

Rapid death occurs in 10% of patients who have a large area of lung involved, but the majority of patients recover provided appropriate treatment is given quickly.

**PULMONARY HYPERTENSION**

See COR PULMONALE

**PUNCH DRUNK SYNDROME**

Recurrent blows to the head over a period of months or years can result in repeated episodes of concussion. The brain is unable to repair itself, and eventually damaged brain tissue is replaced by scar tissue, and the patient's brain is then unable to function normally. Symptoms of the punch drunk syndrome include an unsteady walk, tremors, personality changes, poor coordination and deterioration in mental ability. Boxers are almost the only people who suffer from this condition, although others who suffer repeated accidental head injuries (eg. soccer players) may also be affected. The diagnosis is confirmed by CT scans and electroencephalograms (EEG). There is no cure, but some of the worst symptoms can sometimes be controlled by medication.

See also CONCUSSION

**RABIES**

Rabies is an invariably fatal viral infection spread by animal bites (eg. dog, cat, bat, monkey, rat). It is found throughout Asia, Europe (except Spain, Italy, Scandinavia and Britain), Africa and the Americas, but is not present in Australia and the Pacific. Other forms of the rabies virus have been implicated in rare infections, including a fatal encephalitis that can be caught from infected bats in north-eastern Australia.

The virus responsible is the *Lyssavirus*, which infects the salivary glands of animals, so that any bite causes the injection of the virus into the victim's wound. The incubation period after a bite is three to seven weeks. If possible, the animal causing the bite should be isolated and observed to see if it is affected. The diagnosis can be confirmed by a specific Lyssavirus antibody blood test.

The classic symptom is fear of water (hydrophobia), which is due to the severe pain that swallowing any food or liquid causes as a result of muscle spasm in the throat. Further symptoms include skin pain and tingling, generalised muscle spasms, convulsions, the production of copious amounts of thick saliva and eventually muscle paralysis.

First aid after a bite from a possibly infected animal is thorough washing of the wound with soap and water, then drenching the bite in antiseptic (eg. Betadine). If rabies is suspected it is essential for the patient to receive a rabies vaccine because no treatment is available once symptoms appear, and death occurs within two or three days of symptoms appearing.

The rabies vaccine can be given routinely in those working with animals in rabies areas or immediately after a bite from a rabid animal. For prevention two injections are given one month apart, and repeated annually. After a suspect animal bite a series of frequent injections must be given as determined by a doctor. The vaccine is not designed for use in pregnancy but must be used if the mother has been exposed to a bite from a rabid animal. It may be used in breast feeding and children. Use rabies vaccine with caution in immune deficiency and history of allergy. It must be injected into muscle and not injected subcutaneously (under skin) or in to vein. Side effects may include local redness, soreness and hardness at injection site. Uncommonly a fever, muscle pains, nausea and diarrhoea occur.
It is routinely given only to veterinarians and others working with animals in areas affected by rabies, but is given after any bite by an animal in an area affected by rabies. Once symptoms of rabies occur, it is inevitably fatal. An inadvertent additional injection is unlikely to have any serious adverse effects. Rabies does not occur in Australia, New Zealand or the United Kingdom, but does occur in most other areas of the world.

RAMSAY-HUNT SYNDROME
The Ramsay-Hunt syndrome is the infection of a facial nerve with the virus Herpes zoster, which also causes chickenpox and shingles.

Shingles may affect any nerve leading out from the brain or spinal cord, but if the nerve affected (the geniculate ganglion) is the one supplying the ear and face, the patient will develop this syndrome.

It causes severe earache, dizziness, and a painful blistering rash across the upper face and ear. No investigations are normally necessary, but if required the diagnosis can be confirmed by taking special swabs from a sore.

Antiviral medication (eg. aciclovir, valaciclovir) must be taken as soon as the shingles starts to prevent its spread. Steroids may also be used to reduce complications, but permanent deafness and dizziness can result if treatment starts too late.

It is named after the American neurologist James Ramsay-Hunt (1872-1937).

RAYNAUD’S PHENOMENON
Raynaud’s phenomenon is a distressing spasm of small arteries, almost invariably affecting women. Attacks are usually triggered by cold conditions, such as entering an air-conditioned building or a cold climate. Other triggers may be hormonal changes, stress and anxiety, exercise and some foods. Raynaud's disease is the most common cause, but in most cases no specific cause can be found, although it may be associated with rheumatoid arthritis, CREST syndrome and scleroderma.

The hands go white then blue, swell and become very painful episodically. It usually starts in the teenage years or early twenties, may remain lifelong, and affects one in every five women, but often eases after the menopause.

Patients should keep their hands warm, and alcohol in low doses may be useful. A wide range of tablets (eg. alpha-blockers) and ointments can be used to dilate the tiny arteries in the fingers. As a last resort, operations to cut the nerves that cause the artery spasm can be performed.

REFSUM DISEASE
Refsum disease or syndrome is a rare congenital (inherited) disorder of nerve metabolism (chemistry) that results in the accumulation of damaging phytanate in nerve tissue. The onset is usually between 5 and 10 years
of age. Affected children develop poor sensation and muscle control of the hands, forearms, feet and lower legs that affects the legs more than the arms. Other symptoms include nerve deafness, thickened skin and pigmentation of the light sensitive retina at the back of the eye. Heart failure and nerve defects in the heart are late complications. The diagnosis is confirmed by a nerve biopsy, which shows a characteristic nerve deformity. No treatment is available.

**RENAL FAILURE, CHRONIC**

Chronic kidney (renal) failure, or uraemia, is a slow, gradual failure of kidney function. Old age is the most common cause, but it may also be due to many other conditions including a damaged blood supply to the kidney from hardened arteries (arteriosclerosis), poisons, infections, the body trying to reject the kidney in autoimmune conditions such as systemic lupus erythematosus, and many rarer diseases.

Because of its slow onset, patients may not present to a doctor until the condition is well advanced, by which time they have weakness, tiredness, lack of appetite, weight loss, nausea, headaches, passing urine frequently and at night, and in advanced cases itchy skin, vomiting, high blood pressure and anaemia. Abnormal blood and urine tests are diagnostic, but further investigations are carried out to discover any specific cause.

It is necessary to treat any cause of the condition if possible, followed by a strict diet (low in protein), and control of all fluids that are drunk. Unless the cause can be corrected, long-term treatment with an artificial kidney machine (dialysis), or a kidney transplant operation is necessary. Patients must also be very careful with medications, as they are likely to be far more effective, last longer in the body than normal, have more side effects and may be toxic.

Kidney transplants have an 80% cure rate, while dialysis can be continued for many years if necessary.

**RESTLESS LEGS SYNDROME**

Patient’s with the restless legs or Wittmaack-Ekbom syndrome have legs that feel as though they want to exercise when the body is trying to rest. The cause is unknown, but it is more common in women, made worse by pregnancy and heat, and is sometimes aggravated by antihistamine medications. It is not related to previous exercise.

When the patient goes to bed, or is resting, they can't get to sleep because they feel that they have to keep moving their legs. It can be helped by keeping the legs cooler than the body, and using paracetamol, the medication ropinirole or a small dose of a mild muscle relaxant such as diazepam (Valium). Unfortunately, getting out of bed and going for a run doesn't help.

It is a distressing but not serious problem that often occurs episodically for years, but is usually well controlled by treatment.

**RETINOBLASTOMA**

A retinoblastoma is cancer of the retina (light sensitive cells at the back of the eye) that usually occurs in children under three years of age. There is a familial (inherited tendency) in 40% of cases, but cause in others is unknown.

The pupil becomes white, a squint is noticed, the affected eye bulges forward, becomes reddened and the vision is affected. The cancer may spread (metastasise) from the eye along the optic (vision) nerve to the brain.

It is diagnosed by examining the eye with a magnifying light (ophthalmoscope) and a CT scan.

Small tumours may be treated by laser or chemotherapy (medication), but most are not diagnosed until large, and the eye must be removed. The prognosis depends on the size of the tumour at time of diagnosis. Survival rate with no spread is 85%, but this drops dramatically if cancer cells are found in the optic nerve. The overall five year survival rate is about 70%.

**RET T SYNDROME**

About one in every 10,000 women in has Rett syndrome, which is a congenital (present from birth) condition characterised by episodes of rapid over breathing (hyperventilation), seizures, subnormal mentality, autism, constipation and repetitive movement disorders of the hands. The symptoms become apparent at 12 to 18 months of age, and the only treatment is medication to control the seizures. It is named after the Austrian paediatrician Andreus Rett (b.1924).

See also AUTISM; INTELLECTUAL DISABILITY
A RATIONALE FOR THE BRAIN

RHABDOMYOSARCOMA
A rhabdomyosarcoma is an extremely aggressive malignancy (sarcoma) of muscle. It is found most commonly in the neck muscles, but may occur anywhere in the body. The symptoms depend on the location of the tumour and its affects on surrounding structures and organs, but a painful lump is usually felt at the site of the sarcoma.

They can occur at any age, including in a foetus before birth.

Surgical removal is often not possible due to their insidious spread through surrounding tissues, but amputation of a limb may be curative. More centrally located tumours are treated by radiotherapy and cytotoxic medications such as vincristine, actinomycin D and cyclophosphamide.

RHEUMATIC FEVER
Rheumatic fever is a damaging inflammation of the heart valves that follows some types of bacterial infections and was common before antibiotics were readily available, but is now rare in developed countries.

Patients have two or more of a number of widely different symptoms, so every case is completely different. Symptoms include inflammation of the heart and its valves, a rapid pulse, irregular heart beat, irregular shaped red patches and rings on the skin, chorea (uncontrolled twitching of the arms, legs and face), fever, and arthritis that moves from one large joint to another. In 70% of patients it causes permanent damage to heart valves that leak and fail in later life, and are susceptible to infection (endocarditis). All patients who have had rheumatic fever must take antibiotics whenever they have any dental treatment or operation.

The diagnosis is confirmed by blood tests (eg. anti-deoxyribonuclease-B titre - Anti-DNAse-B), and an electrocardiogram (ECG), then antibiotics (commonly penicillin) are given to remove any remaining bacterial infection, aspirin to reduce fever and joint pains, and strict bed rest is ordered for several weeks or months.

The condition may last a few weeks to months, with children taking far longer to recover than adults. A significant number of patients have recurrences for years afterwards. 98% of patients recover from the first attack, but multiple repeat attacks may lead to death from heart damage.

See also SYDENHAM'S CHOREA

RICHNER-HANHART SYNDROME
The Richner-Hanhart syndrome is a congenital condition caused by excessive levels of the protein tyrosine in the blood because the liver lacks the necessary enzymes to break it down. The affected child suffers from eye damage, intellectual disability, convulsions and skin damage. Once diagnosed, the offending proteins can be removed from the diet, and no further deterioration of the patient’s condition should occur. Early diagnosis of this inherited syndrome is therefore vital, but may be very difficult due to the subtle nature and onset of symptoms in an infant.

See also INTELLECTUAL DISABILITY

RICKETS
Rickets is a rare disease in developed countries, but growing children in poorer countries may develop rickets if they have an inadequate intake of vitamin D. This vitamin is essential for the body to absorb calcium, which is the main constituent of bone. Vitamin D is obtained from dairy products (milk, cheese, yoghurt, etc.), eggs and fish, and can also be formed in the body by the action of sunlight on certain substances in the skin. In adults the same condition is known as osteomalacia.

Children with rickets have soft bones and grow slowly. The legs tend to bow outwards because of walking on the soft long bones of the legs, and there are abnormalities in the growth of the ribs, and excessive enlargement of the forehead may occur. Patients may also be “double-jointed”, with slack ligaments around the joints, and may have weak muscles. Any bone deformity that occurs may become permanent, resulting in premature arthritis.

Measurement of calcium and vitamin D levels in the blood, and x-rays of long bones can be used to confirm the diagnosis. it is easily treated by supplying adequate amounts of vitamin D in the diet. No further damage is likely once a good diet is started.

RILEY-DAY SYNDROME
Riley-Day syndrome (dysautonomia) is an uncommon familial (runs in families from one generation to the next) syndrome that occurs in Jews of Middle Eastern extraction. Its symptoms include a lack of tears in the
eyes, excessive sweating, intermittent fevers and episodes of low body temperature, blood pressure swings between being too high and too low, the surface of the eye may feel no pain, and generally patients feel only the most severe pains. As a result they may have fractures and other injuries of which they have no knowledge. Less commonly, they may have poor coordination, difficulty in swallowing, difficulty in talking, and extreme mood swings. They may suffer serious personal injury, particularly to the eye resulting in blindness. No treatment is available.

**ROUSSY-LEVY SYNDROME**

The Roussy-Levy syndrome or disease is a familial (inherited) developmental defect of nerves in the arms and legs. Sufferers have poor coordination, curvature of the spine and hump back (kyphosis), thin arms and legs with very weak muscles. The tendon reflexes in the arms and legs are absent, no treatment is available and the condition is slowly progressive.

**SANDIFER SYNDROME**

Sudden spasms of the neck muscles with neck extension and muscle stiffening in an infant, caused by reflux oesophagitis from the backflow of concentrated hydrochloric acid from the stomach into the oesophagus resulting in sudden severe pain, is described as Sandifer syndrome. It may be confused with an epileptic seizure.

It is treated by antacids, anti-reflux (eg. cisapride) and anti-ulcer (eg. ranitidine) medications. A trial of treatment is usually both diagnostic and successful, but in cases of doubt a barium swallow x-ray may be performed.

**SANFILIPPO SYNDROME**

Sanfilippo syndrome or mucopolysaccharidosis type III is a rare inherited abnormality of the metabolic system in which patients are unable to eliminate certain substances (mucopolysaccharides) from the body. The effects include severe intellectual disability starting at age 5 years or later, excess body hair, abnormal face structure, and enlarged liver and spleen. It may be diagnosed by urine tests, which show excess levels of heparitin sulfate. No treatment is available.

See also HUNTER SYNDROME; HURLER SYNDROME

**SARCOIDOSIS**

Sarcoidosis is an uncommon disease that causes damage and inflammation to a wide range of organs within the body, but most commonly to the lungs. The cause is unknown, but women are more commonly affected than men, and the usual age of onset is 40 to 60 years.

The symptoms can be very varied and sometimes bizarre because almost any part of the body may be involved as affected tissues fail to function correctly. Patients may have a fever, tiredness, shortness of breath, rashes, enlarged glands, liver or spleen enlargement, pain, arthritis, pins and needles sensation and heart failure. Gradual destruction of the lungs and other organs may occur.

Blood tests and lung x-rays show abnormalities, but cannot specifically diagnose sarcoidosis. A definitive diagnosis requires the microscopic examination of a biopsy (Kveim test).

Steroids are used to reduce the inflammation, but the disease is slowly progressive and cannot be cured, although control is usually sufficient to give the victim a relatively long life.

**SAVANT SYNDROME**

See IDIOT SAVANT SYNDROME

**SCHIZOPHRENIA**

Schizophrenia is a psychotic mental illness, which causes abnormal behaviour and perceptions. The idea that schizophrenics suffer from a “split personality” is not true though. That idea comes from the origin of the word schizophrenia, which comes from the Greek word meaning “to split”, and rather than meaning “split personality” it actually means a “split from reality”.

The exact cause unknown, but there is a family tendency and the environment in which the patient is raised, and emotional stress, may trigger a person to escape into schizophrenia. Marijuana abuse is often a trigger in young people.
Patients have a distorted view of the world because of delusions and hallucinations. They often change the topic of conversation for no apparent reason, may not look after themselves, become dishevelled in appearance, withdrawn, and fail to communicate properly with others. Their mood and behaviour changes seem bizarre and they often believe that others are persecuting them. They may hear unfriendly voices, or have frightening hallucinations.

**SCHIZOPHRENIA INHERITANCE**

<table>
<thead>
<tr>
<th>Relative</th>
<th>Risk of Developing Schizophrenia (%)</th>
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</thead>
<tbody>
<tr>
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<td>1%</td>
</tr>
<tr>
<td>Identical twin</td>
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<tr>
<td>Non-identical twin</td>
<td>15%</td>
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<tr>
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<td>9%</td>
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<tr>
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<tr>
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<tr>
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</tr>
<tr>
<td>Uncle</td>
<td>2%</td>
</tr>
<tr>
<td>Niece/nephew</td>
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</tr>
</tbody>
</table>

A lot of people think that schizophrenics are much more violent and dangerous than people who don't have schizophrenia, but in fact crime and violent behaviour is no more common amongst schizophrenics than it is other people. They can become violent, but generally speaking, people with schizophrenia are more of a danger to themselves than to others as suicide is possible. Many patients refuse medication and treatment because they lack insight into their condition.

There are no diagnostic blood or other tests, and the diagnosis rests on the ability of the doctor to recognise a characteristic form of behaviour.

There is a wide range of effective medications (antipsychotics) available to treat schizophrenia, including clozapine, fluphenazine, olanzapine, pimozide, promazine, risperidone, sulpride, thioridazine, thiothixene, zuclopenthixol and trifluoperazine. They can be given as tablets or injections to control the disease. Just as important are a supportive environment and psychological counselling for the patient and their family.

A permanent cure is not usually possible, but the condition can be well controlled if the patient remains on long-term medication.

See also CATATONIC SYNDROME; FUGUE; PSYCHOSES

**SECKEL SYNDROME**

The Seckel syndrome is a rare inherited genetic abnormality, but both parents must be carriers for it to affect their children. Affected children have reduced growth as a foetus before birth, low set ears, hip and elbow dislocation, intellectual disability and a beaked nose. No treatment is available.

See also INTELLECTUAL DISABILITY

**SEPTICAEMIA**

Septicaemia, or blood poisoning, is a bacterial infection of the blood. The infection usually starts in another part of the body, such as the lungs, tonsils (quinsy) or after childbirth (now very rare), but in some cases the origin of the infection may never be found. Many different bacterial infections have septicaemia as a complication.

Patients are usually very ill, with a high fever, prostration and generalised aches and pains. A small number will have an overwhelming infection with resistant bacteria, which leads to death.

Many different bacteria may be responsible for the infection, and it is important to identify them by blood tests before antibiotic treatment commences. Potent antibiotics are given by mouth, injection or drip infusion in hospital to cure the infection. The original site of infection must also be treated if possible.

Provided an appropriate antibiotic can be found, most patients can be cured.

**SEROTONIN SYNDROME**

The serotonin syndrome is a rare complication of antidepressant medication use caused by interaction between different antidepressants after inadequate rest period between stopping one medication and starting another, or interaction between antidepressants and other drugs (eg. pethidine, pseudoephedrine, dextromethorphan - the last two are found in cold remedies).
The symptoms include agitation, vomiting, tremor, muscle spasm, dizziness, incoordination, rapid heart rate, overactive reflexes and abnormal eye movements (nystagmus). Some patients develop a very high fever, flushing and diarrhoea.

Once diagnosed, all antidepressant medication must be ceased, and medication (eg. cyproheptadine, diazepam) is given to reverse the adverse effects. The prognosis is good.

See also NEUROLEPTIC MALIGNANT SYNDROME

SHY-DRAGER SYNDROME

The Shy-Drager syndrome is a severe form of multiple system failure of no known cause.

Patients have low blood pressure, reduced sweating, slight tremor, difficulty in speaking, rigidity, poor coordination, impotence, dizziness, varying muscle paralysis and incontinence. Fainting may occur with changes in position. An MRI scan of the brain may be abnormal.

Treatment is unsatisfactory. Patients must take care with postural changes, wear elastic stockings and a support girdle, and take medication (eg. fludrocortisone, ephedrine sulfate). Unfortunately, it is usually progressive to death in 5-7 years.

See also FAINT

SICARD SYNDROME

The Sicard (Collet-Sicard) syndrome is due to damage to nerves from brain that control muscles in the head and neck caused by a tumour (eg. of the parotid salivary gland) or injury involving the cranial nerves 9, 10, 11 and 12. Patients suffer paralysis of the muscles in the larynx, tongue, pharynx, palate and neck on one side. A CT or MRI scan is used to detect the tumour, which is treated if possible. The prognosis depends on the cause, but in most cases there is no cure.

SICK BUILDING SYNDROME

The sick building syndrome is a condition that affects workers in air-conditioned buildings.

In order to conserve cold air and therefore energy, some large air-conditioned buildings allow minimal amounts of fresh air into circulation with each cycle. This allows organic solvents (eg. from photocopiers, glues, paints), fungal spores, pollens, dusts and other contaminants to recirculate in increasing concentrations, particularly if the air conditioning filters are poorly maintained.

Residents and workers in the building develop snuffly noses, eye irritation, dry skin, headaches and tiredness. The longer they spend in the building, the worse the symptoms become. Increasing the amount of fresh air entering building and carefully maintaining filters solves the problem.

SICK SINUS SYNDROME

The sick sinus syndrome (SSS) is an abnormality of heart rhythm, due to failure of the heart pacemaker (sinus node) or nerve conduction within the heart. Patients have a variable heart rate from brief standstill or very slow (sinus bradycardia) to markedly rapid beat or atrial fibrillation and resultant fainting or collapse. Heart attack and death may rarely occur. A continuous ECG reading (Holter monitor) for 24 hours will be abnormal and can be used to make the diagnosis.

Treatment involves an artificial pacemaker and drugs to stabilise heart rhythm, which give good control once diagnosed.

SLE

See SYSTEMIC LUPUS ERYTHEMATOSUS

SLEEP APNOEA

A cessation of breathing (apnoea) during sleep most commonly occurs in overweight middle-aged men due to a complete relaxation of the small muscles at the back of the throat. The throat tissue becomes very soft, flabby and collapses as the patient breathes in, closing off the throat and preventing breathing. Snoring is caused in the same way. In elderly men with high blood pressure there may be a suppression of the urge to breathe by the brain during very deep sleep.

In sufferers breathing stops for periods from 10 to 60 seconds on many occasions during the night while asleep, resulting in tiredness during the day, morning headaches, personality changes, poor concentration, bed-
wetting and impotence. The sleeping partner complains about the patient’s loud snoring and thrashing restless sleep. Minor brain damage may occur with every episode of apnoea, and this eventually leads to a noticeable deficit in brain function.

The diagnosis is best made in a sleep laboratory, where the patient's sleep and breathing pattern can be monitored through an entire night.

Treatment involves weight loss, and avoiding alcohol, sedatives and smoking. In persistent cases a small mask is fitted to the patient's nose, and air is blown up the nose at a slightly increased pressure with a small electrically driven blower (continuous positive airway pressure - CPAP). In severe cases surgery to the back of the throat and nose to remove the uvula and part of the soft palate opens the airway. A significant deterioration in the quality of life may occur unless successfully treated.

**SMITH-LEMLI-OPITZ SYNDROME**

The Smith-Lemli-Opitz syndrome is an inherited genetic abnormality causing varied deformities. Both parents must be carriers for the syndrome to develop. The symptoms may include droopy eyelids, narrow forehead, intellectual disability, abnormal penis, malformed nostrils and webbed toes. Surgical correction of the deformities is possible, but there is no cure.

See also INTELLECTUAL DISABILITY

**SPINOCEBERELLAR DEGENERATION**

See FRIEDREICH’S ATAXIA

**STEELE-RICHARDSON-OLSZEWSKI SYNDROME**

The Steele-Richardson-Olszewski syndrome (progressive supranuclear palsy) is a deterioration of the part of the brain responsible for muscle control. Its cause is unknown, but it occurs twice as often in males as females, and is often confused with Parkinson disease.

The typical patient is an elderly person whose back is rigidly extended, falls backwards easily, is demented, eyes are unable to look down, reflexes are increased and has difficulty swallowing.

Physiotherapy, occupational and speech therapy, and medications to ease muscle rigidity and spasm are used in treatment, but the condition is slowly progressive.

See also PARKINSON DISEASE

**STEWART-MORGAGNI-MOREL SYNDROME**

Stewart-Morgagni-Morel syndrome is a disorder of the pituitary gland and adjacent part of the brain (hypothalamus) that controls the gland, resulting in excess production of androgenic (male sex) hormones. Patients develop acromegaly (enlarged head and major bones) and a very prominent forehead. CT and MRI scans are used to investigate the condition and any cause.

Treatment involves surgery or irradiation to the pituitary gland to reduce androgen production, but this causes hypopituitarism as a side effect. The prognosis is reasonable, but depends on the success of treatment.

**STICKY PLATELET SYNDROME**

The sticky platelet syndrome is an uncommon cause of abnormal blood clots (thromboses) in young patients. Platelets are the cells in blood that stick together to form a clot. The excess stickiness of the platelets causes migraines and thromboses, but the symptoms vary markedly depending on the organs affected. Stroke may occur in the brain in severe cases. The cause is unknown.

Medications that reduce platelet stickiness such as aspirin and dipyridamole are taken long term, and good control is possible, but serious permanent organ damage may occur before the condition is diagnosed.

**STIFF-MAN SYNDROME**

The stiff-man syndrome is a rare cause of muscle stiffness and spasm. Patients have rigid limbs with severely painful muscle cramps. Bones may develop stress fractures and joints may partially dislocate (subluxate) due to the constant muscle spasm. The cause is unknown but patients have an abnormal electromyogram (EMG), which measures electrical activity in muscles. Medications (eg. diazepam, suxamethonium) are taken to relax the muscles and injections or surgery is used to block or destroy the nerves to muscles. There is no cure and management is difficult.
St.Louis Encephalitis

St.Louis encephalitis is a viral infection (Flavivirus) of the brain that spreads from birds to man by the bite of a mosquito. It occurs in the central and western United States, and is more common in the warmer months and the elderly. The incubation period is one to three weeks.

Patients develop symptoms that include a fever, belly pains, dizziness, sore throat, cough, headache, neck stiffness, nausea, vomiting, tiredness, disturbed mental functions, disorientation, coma, and in up to 5%, death. It commonly causes permanent brain damage in elderly survivors with paralysis, epilepsy, tremor and personality changes. By contrast, in children the symptoms may be minimal. There is no specific diagnostic test, but tests on blood and the cerebrospinal fluid around the brain are abnormal.

No specific treatment or vaccine is available. Acute attack lasts for one to three weeks, but complete recovery may take months. The mortality rate is worse in the elderly.

See also ENCEPHALITIS

Stokes-Adams Attack

A Stokes-Adams attack (Adams-Stokes syndrome) is a sudden slowing of the heart rate due to an electrical problem within the heart, resulting in inadequate blood supply to the brain. It often occurs after a heart attack, may have no apparent cause, or may be due to a side-effect of medication.

The patient faints suddenly, convulses, and has a slow heart rate or multiple missed heart beats. Facial flushing occurs for a minute or so on recovery. Rarely an attack may cause permanent damage to heart leading to further heart complications.

The diagnosis of a series of unexplained faints can be made by monitoring the heart electrical activity for a prolonged period (Holter monitor), while maintaining a normal lifestyle, until an attack occurs, and the heart electrical problem can be identified. Once diagnosed, appropriate medications can be prescribed to regulate the heart rhythm or surgical implantation of an electrical pacemaker can be performed.

Recovery from a sudden attack is usually rapid, but recurrences may be frequent. Attacks are usually well controlled by appropriate treatment once cause identified, and patients get good result from a pacemaker.

See also FAINT

Strachan Syndrome

Strachan syndrome (also known as Jamaican neuritis) causes widespread nerve and skin damage due to poor nutrition, particularly a lack of vitamin B. It often occurs in alcoholics and with starvation, and is worse in smokers.

The symptoms include amblyopia (dim vision), dermatitis around the mouth and genitals, and painful and excessively sensitive areas of skin. Some patients have muscle weakness or spasms and permanent vision damage occurs if the syndrome is left untreated for a prolonged period. Abnormalities of the retina (light sensitive area at back of eye) can be seen when examined through an ophthalmoscope (magnifying light), and there are blood test abnormalities, and over active reflexes.

Vitamin supplements and a good diet cure the condition.

Stroke

See CEREBROVASCULAR ACCIDENT

Sturge-Weber Syndrome

The Sturge-Weber syndrome is a congenital developmental disorder of the brain and skin that causes intellectual disability, a red stain across part of the face, convulsions, paralysis down one side of the body and eye abnormalities.

Medication can control the convulsions, surgery can correct the cosmetic deformities, and a combination of drugs and surgery are used for the eye abnormalities. Although there is no cure, reasonable control of symptoms is possible.

See also INTELLECTUAL DISABILITY
SUBACUTE COMBINED DEGENERATION OF THE CORD
Subacute combined degeneration of the cord (or combined system disease) is a complication of untreated pernicious anaemia and a long-term lack of vitamin B12. There is a slow and progressive degeneration of the spinal cord and the nerves radiating out from it to the body resulting in weakness, loss of some sensations (eg. vibration, position sense) and eventually loss of control of the limbs and an inability to walk.

It can be successfully treated with regular vitamin B12 injections, but the damage already present cannot be significantly reversed.

SUBACUTE SCLEROSING PANENCEPHALITIS
Subacute sclerosing panencephalitis is a widespread inflammation of the brain in children (more commonly boys) that is thought to be caused by the measles virus. It causes poor coordination, muscle spasms, intellectual damage, blindness due to brain damage and muscle wasting. The disease is usually slowly and steadily progressive to death.

See also ENCEPHALITIS

SUBARACHNOID HAEMORRHAGE
A subarachnoid or intracerebral haemorrhage is a bleed into the substance of the brain. The arachnoid mater is the middle of the three meninges (membranes) that surround and support the brain, so by definition a subarachnoid haemorrhage is a bleed within the arachnoid membrane. The innermost membrane is the pia mater.

Rupture of a blood vessel in the brain may be due to high blood pressure (stroke), the rupture of an aneurysm (ballooning on the side of an artery), bleeding disorders (eg. thrombocytopenia, leukaemia), brain tumours, head injury or as a side effect of medication (eg. warfarin).

Patients experience a sudden loss of consciousness or confusion, vomiting, dizziness, headache and abnormal brain function (eg. partial paralysis, strange sensations) depending on the position of the bleed within the brain. A CT scan or angiography (x-ray of arteries after injecting a dye) is used to find site of bleed.

Treatment will depend on the cause of the bleed, and can vary from time and rest to surgery to stop continued bleeding. The prognosis is very variable depending on the position and severity of the bleed. Some patients recover rapidly, while other may lapse into a long term coma or die, or develop permanent brain damage and epilepsy.

See also CEREBRAL HAEMORRHAGE; SUBDURAL HAEMATOMA

SUBCLAVIAN STEAL SYNDROME
The subclavian steal syndrome restricts the blood supply to the left arm and head. It is caused by pressure from one of the thoracic outlet syndromes (eg. Naffziger syndrome, scapulocostal syndrome) on the left subclavian artery or innominate artery, which supply blood to the left side of the head and the left arm. It eventually results in a restricted blood supply to the brain. Significant brain damage may occur if the correct diagnosis is not made.

Patients have arm pain, particularly with exercise, and varying brain symptoms similar to a transient ischaemic attack. Angiography (x-ray of arteries after injection of a dye) is abnormal and confirms the diagnosis. Surgical bypass of the affected artery is necessary when there are significant symptoms.

See also TRANSIENT ISCHAEMIC ATTACK

SUBDURAL HAEMATOMA
A subdural haematoma is a collection of blood between the brain and the skull that puts pressure on the brain and affects its function. The dura mater is the outermost of the three meninges (membranes) that surround and support the brain, so by definition this is a bleed between the dura mater and the arachnoid mater, which is the middle membrane. The innermost membrane is the pia mater.

The bleeding is usually due to a significant head injury, but sometimes may be due to the rupture of a blood vessel affected by arteriosclerosis (hardening of the artery), high blood pressure or for no obvious reason. The onset may be sudden, or may be delayed for some weeks after a head injury if the bleed and build up of pressure is very gradual.
A RATIONALE FOR THE BRAIN

The symptoms may include confusion, vomiting, dizziness, headache and abnormal brain function (eg. partial paralysis, strange sensations) depending on the position of the blood collection and the pressure it applies to the brain.

A CT or MRI scan is used to find blood collection, and this is followed by urgent surgical removal of the blood collection.

It may be fatal or cause permanent disability if left untreated, but there are good results from surgical treatment.

See also SUBARACHNOID HAEMORRHAGE

SUNCT SYNDROME

The SUNCT syndrome is a variant form of cluster headache whose name is an acronym of its major symptoms (short lasting, unilateral, neuralgiform, conjunctival injection, tears). It may be triggered by alcohol, stress, exercise, certain foods and glare.

The symptoms are severe headaches that are short lasting (seconds to minutes), one sided (unilateral) and piercing (neuralgiform) with associated red eyes (conjunctival injection) and excess tear production. They may occur regularly or spasmodically.

Medication such as sumatriptan and ergotamine, nasal capsaicin spray, and inhaling pure oxygen are used as treatments. The headaches are very annoying but not serious.

See also CLUSTER HEADACHE

SYDENHAM’S CHOREA

Sydenham’s chorea used to be called St.Vitus dance. It is a complication of rheumatic fever or other bacterial infections, and now a rare condition, but was more common before the development of antibiotics.

Patients have irregular jerky movements of a limb or the body, with a complete loss of muscle tone between each movement. The underlying infection must be treated with antibiotics (eg. penicillin).

SYNCOPE

See FAINT

SYPHILIS

In the 18th. and 19th. Centuries, syphilis was called the French pox by the English, and the English pox by the French.

Syphilis is an infection that is usually sexually transmitted, and which passes through three main stages over many months or years. It is relatively uncommon in developed countries, but still widespread in poorer societies.

The cause is the spirochete bacterium Treponema pallidum, which is transmitted by heterosexual or homosexual contact, sharing injecting needles, blood transfusions, or from a mother to her child during pregnancy (congenital syphilis). The same bacteria also cause yaws and bejel, which are transmitted by close body contact, but not necessarily sexual contact.

The symptoms are totally different in each of the three stages:-

- **First stage** syphilis causes a painless sore (chancre) on the penis, the female genitals, or around the anus of homosexuals, which heals after three to six weeks. There may be painless enlarged lymph nodes in the armpit and groin that also disappear.

- **Second stage** syphilis starts a few weeks or months later with a widespread rash, mouth and vaginal ulcers, and a slight fever. The patient is highly infectious but will usually recover and enter a latent period that may last many years.

- **Third (tertiary) stage** syphilis develops years later with tumours (gumma) in the liver, major arteries, bones, brain, spinal cord (tabes dorsalis), skin and other organs. Symptoms vary depending on organs involved but may include arthritis, bone weakness, severe bone pain, paralysis, strokes, heart attacks, internal bleeding from aneurysms, blindness, headaches, jaundice (liver failure), muscle spasms, skin ulcers, scars, nodules in the larynx and lungs, vomiting, confusion, insanity and death.

- **Congenital syphilis** occurs in newborn infant who have teeth abnormalities, deafness, misshapen bones, deformed saddle nose, pneumonia, and intellectual disability.
A RATIONALE FOR THE BRAIN

It can be diagnosed at all stages by specific blood tests (eg. fluorescent treponemal antibodies, syphilis IgG enzyme immunoassay), or by finding the responsible bacteria on a swab taken from a genital sore in the first stage of the disease. All pregnant women should be routinely tested.

The first stage and second stages are treated by antibiotics such as penicillin (often as an injection), tetracycline or erythromycin. In the third (tertiary) stage antibiotics are also used, but can merely prevent further deterioration, as organ damage is irreversible. A child suffering from congenital syphilis is infectious when born and is treated with antibiotics.

There are many complications associated with a syphilis infection. In the first stage there are usually none, but in second stage syphilis there may be spread of the infection to involve the joints, brain, liver and kidney which may be severely damaged. In the third (tertiary) stage almost any organ can be seriously damaged. Infants with congenital syphilis may develop more serious problems if the condition is not treated aggressively.

Neurosyphilis is a serious complication of third stage syphilis that affects the brain to cause varying forms of paralysis and abnormal brain function depending upon which parts of the brain are infected.

A course of antibiotics for a few weeks almost invariably cures the disease in its first two stages, but there is no cure for tertiary or congenital syphilis. Plastic surgery may correct the more obvious congenital deformities.

See also TABES DORSALIS

SYRINGOMYELIA

Syringomyelia is an expansion of a cavity within the spinal cord that places pressure on the nerves passing this point, and affects nerve function and circulation of the surrounding cerebrospinal fluid below it. It most commonly occurs in the neck, and may be a developmental disorder (congenital), or due to tumours, injury or inflammation. Sometimes it is associated with the Arnold-Chiari malformation, in which the lowest part of the brain (cerebellum) slips down into the spinal canal.

Patients experience a gradually worsening reduction in sensation and muscle weakness below the affected point of the spine. It is diagnosed by CT and MRI scans, and then surgical drainage of the cavity in the spinal cord, and removal of any tumour is performed. Untreated, quadriplegia or paraplegia may occur, but good results are obtained from treatment.

SYSTEMIC LUPUS ERYTHEMATOSUS

Systemic (or disseminated) lupus erythematosus (SLE) is a relatively common inflammatory condition affecting joints, skin, liver, and kidney most commonly, but almost any tissue in the body may be involved. 85% of cases occur in women (usually young), and it is more common in Negroes than Caucasians.

Lupus is an autoimmune disorder in which the body inappropriately rejects normal tissue for no known reason. Attacks may be precipitated by stress, some medications or chemicals. There is also a familial tendency.

Common symptoms are arthritis of several joints, a red scaly rash across both cheeks and the bridge of the nose (“butterfly rash”), rashes on other areas that are exposed to sunlight, mouth ulcers, poorly functioning kidneys and anaemia. Additional symptoms may include a fever, loss of appetite, tiredness, weight loss, damaged nails, loss of hair and painfully cold fingers. Less common complaints include conjunctivitis, blurred vision, chest pain, pneumonia, heart failure, belly pain, constipation, depression and convulsions. The symptoms vary significantly from one patient to another, and none will have them all. Many patients are free of symptoms for months before a recurrence. After each attack, there is slightly more permanent liver, kidney or heart damage, and eventually these problems accumulate to the point where the disease becomes life threatening. In rare cases it proceeds relentlessly to death within a relatively short time.

Specific blood tests can diagnose the condition (eg. lupus anticoagulant antibody, ANCA, anti-Smith antibodies, anti-DNA, anti-dsDNA).

Treatment depends upon the severity of the disease, and with mild symptoms, no treatment is required. Sun exposure should be avoided, and all non-essential medications ceased. In severe cases, a wide range of drugs, including steroids, cytotoxics, immunosuppressives and antimalarials may all be used. Regular blood tests follow the course of the condition, which is very variable, from a mild arthritic complaint to a rapidly progressive disease. There is no cure, but with careful management, compliance with treatment, and regular check-ups, 90% of patients are alive more than ten years after the diagnosis is made.

See also AUTOIMMUNE DISEASES
TABES DORSALIS
Tabes dorsalis is a rare complication of third stage syphilis due to the development of a syphilitic deposit in the spinal cord. Patients experience repetitive, brief, severe pain in the legs, back, chest, and sometimes arms and face. Some patients develop poor coordination of the legs and difficulty in walking, loss of bladder control, vomiting, abdominal pain, and abnormal sensations.
Antibiotics can treat the syphilis and prevent progression of the disease, but there is no cure for existing symptoms.
The disease name comes from the Latin words *tabes* (wasting) and *dorsalis* (back).
See also SYPHILIS

TALMA SYNDROME
Talma syndrome (or disease) is a condition that causes prolonged contraction and spasm of muscles when movements are initiated, a decrease in the degree of relaxation even when the muscle is at rest. Patients have normal mechanical and electrical excitability of the motor nerves but increased excitability of muscles. It usually develops in adult life after trauma, acute infection, or intoxication.

TEMPORAL ARTERITIS
Temporal or giant cell arteritis, is an inflammation of medium to large arteries throughout the body, but most commonly the arteries in the temples at the side of the head. The cause is unknown but it may an autoimmune disease, and often follows a significant viral infection.
Involved arteries become extremely tender and swollen. Symptoms depend on which arteries are inflamed, but may include headache, scalp tenderness, pain in the jaw with chewing, throat pain and vision disturbances. Less commonly a cough, shoulder pain, weakness and a fever occur. Blindness due to involvement of the arteries in the eye, and aneurysms (dilations) of arteries are complications. About half of all patients also have polymyalgia rheumatica.
Blood tests are usually performed to detect the inflammation, and a biopsy of an artery will reveal the presence of characteristic giant cells. Treatment involves taking steroid tablets (eg: prednisone) for several months. It is usually well controlled and eventually cured, but recurrences when medication is ceased are common.
See also AUTOIMMUNE DISEASES

TETANUS
Tetanus (lockjaw) is a very serious worldwide disease that attacks muscles. The bacterium *Clostridium tetani*, which lives harmlessly in the gut of many animals, particularly horses, is responsible. When it passes out in faeces it forms a hard microscopic cyst, which contaminates soil. It can remain inactive for many years until it enters a cut or wound where it starts multiplying and produces a chemical (toxin), which spreads throughout the body in the blood.
Deep wounds, such as treading on a nail, are particularly susceptible to a tetanus infection.
The toxin attacks the small muscles used for chewing making it difficult to open the mouth (thus the common name of lockjaw). Larger and larger muscles are then attacked, irritating them and causing severe spasm. Excruciating pain from widespread muscle spasms may be triggered by the slightest noise. The patient remains conscious, but eventually the muscles that control breathing and the heart are affected.
There is no effective treatment other than muscle relaxants and mechanical ventilation. Although the bacteria may be killed by antibiotics, the toxin remains in the body. Death occurs in about 50% of patients, even in good hospitals.
A vaccine (tetanus toxoid) is available, but it does not give lifelong protection, and revaccination is necessary every ten years until age 50, or after five years with a deep wound.

TETANY
Tetany is a form of muscle spasm that is totally different to a tetanus infection. The small muscles in the hand go into spasm with the wrist bent and fingers and thumb bunched together and pointed towards the wrist (a phenomenon known as main d'accoucheur or obstetric hand). Sometimes muscles in the forearms and feet also go into a firm spasm.
Tetany may be due to low levels of calcium, potassium or magnesium in the blood, but is most commonly caused by rapid over breathing (hyperventilation) after a shock, surprise, injury or vigourous exercise.

Rapid shallow breathing causes the amount of carbon dioxide in the lungs to increase. This dissolves in the blood to make it more alkaline (high ph) than normal, and small muscles in the hand and elsewhere are sensitive to this change in blood chemistry.

Breathing into a paper bag for a few minutes slows the breathing, lowers the level of carbon dioxide in the lungs, and eases the spasm. It is also assisted by giving repeated reassurance to the patient.

Tetany is not serious, and settles spontaneously eventually, but sometimes not until after the patient has collapsed. There is quick recovery with treatment.

**TICK BITE**

Ticks are from the same family as spiders (arachnids) but are modified so much that they look nothing like their distant cousins. They have a large black body from which mouth parts protrude and grasp the skin. The tick does not have a head as such. A tube-like mouth part pierces the victim's skin to suck up blood. When the tick is full of blood, it drops off and waits for its next victim, which may be almost any warm-blooded animal, although some species preferentially attack certain animals. A full feed of blood may last the tick for a year or more.

Ticks are most active in the spring and summer. Bush ticks, which are the only dangerous form of tick, live on the eastern coastal strip of Australia. The most common victims of ticks are children playing in the bush, and golfers, as ticks shelter in foliage and drop on a victim and burrow in as they engorge themselves on blood. Ticks are usually found on the head, burrowing in amongst the hair, or in body crevices.

Ticks generally cause painful irritation and a raised lump on the skin. Uncommonly they may lead to paralysis if left untreated, especially in children.

Symptoms of tick bite are irritation and pins and needles at the site of the bite, nausea, double vision, unsteadiness, and eventually weakness and difficulty in moving first the lower limbs, then the upper limbs, and finally the face and breathing apparatus.

Do NOT attempt to pull the tick off or cut it out or squeeze it - this forces more venom into the system.

To remove a tick, wash it and the surrounding skin with an alcohol solution, such as methylated spirits. Place a pair of tweezers flat on the skin so that the jaws are on either side of the tick. Grasp the tick firmly, as close to the skin as possible, twist through 90 degrees, and then lift off. The tick will come away easily with minimal pain. Some tiny black marks, the mouth parts, may be left behind, but these rarely cause any trouble. Place some antiseptic cream or lotion on the bite and leave it alone to heal over the next couple of days. If the area becomes red and angry, it may have become infected, and a doctor should be consulted.

Ticks are responsible for the transmission of a wide range of infections including Lyme disease, typhus and Dutton relapsing fever.

**TOLOSA-HUNT SYNDROME**

In this syndrome, an aneurysm (ballooning) on the side of the internal carotid artery at the base of the skull puts pressure on nerves to cause a painful paralysis of one eye.

The diagnosis is made by angiography (x-ray of an artery after injection of a dye), CT or MRI scan. Surgical treatment of the cause, which may be difficult to find, usually cures the problem.

**TORTICOLLIS**

Torticollis is a severe spasm of a muscle in the neck. The sternocleidomastoid muscle that runs diagonally from the mastoid process at the base of the skull behind the ear across the neck to the top of the breast bone (sternum) is most commonly involved.

It is usually caused by repeated turning of the head (eg. watching a tennis match) or sleeping heavily (eg. after excess alcohol) and awkwardly (eg. in a chair), but rarely may be due to a tumour of the brain affecting the nerves to the involved muscles.

The patient has a very painful spasm of muscles, usually on only one side of the neck, that limits neck movement and causes the head to be held at an abnormal angle. No investigations are normally necessary unless a brain cause is suspected. Treatment involves muscle relaxants (eg. diazepam - Valium) and powerful pain killers (eg. pethidine), often by injection, with pain relieving tablets to follow. Heat and physiotherapy are also useful.

Symptoms usually settle in a day or so with treatment.
TOURETTE SYNDROME

Gilles de la Tourette syndrome (to give the condition its full title) is a severe behaviour disorder of children. The cause is unknown, but it is possibly associated with the Crigler-Najjar syndrome.

Patients suffer seizures, other forms of uncontrollable body movements, and often swear and shout excessively. No investigations can specifically diagnose the condition.

Medications are available to control the more serious symptoms, although the syndrome usually persists long term, but often fades in adult life.

The syndrome is named after the French neurologist Georges Gilles de Tourette (1857-1904) who became psychotic himself and was finally restricted to a lunatic asylum after being shot in the head by a patient.

TOXOPLASMOSIS

Toxoplasma gondii is a single-celled animal that is found world-wide as a parasite of cats, other animals and birds, from whom it may spread to humans. The eggs pass out in the faeces of the animal and may then enter a human mouth (eg. after careless handling of cat litters or soil contamination of fingers or food). Once in the gut, the microscopic egg hatches and multiplies into millions of single-celled animals.

In many patients, the symptoms are so mild that they are ignored, but in severe cases the patient complains of a low-grade fever, tiredness, muscle aches, joint pains, headache, sore throat, a mild rash and enlarged glands. In the rare severe cases, the liver, spleen, lungs, eye, heart and brain may be involved.

Patients usually recover without treatment in four to eight weeks. If symptoms are significant or complications develop, medications are available (e.g. pyrimethamine) to destroy the infection.

The worst complication of toxoplasmosis occurs in women who are pregnant. The infection may cause miscarriages, still birth, and deformities in the baby (eg. small head, hydrocephalus, mental retardation, fits, blindness). The disease can be detected by a specific immunoglobulin blood test, and this test is often routinely performed during antenatal blood examinations. If toxoplasmosis is detected in pregnancy, treatment will be given to cure the disease. Unfortunately, because the disease has already occurred, there may still be some damage to the foetus.

There is no vaccination or other form of prevention available. Pregnant women should not associate closely with cats.

TRANSIENT GLOBAL AMNESIA

Amnesia is a loss of memory and transient global amnesia is a total loss of all memory for a specific period in the recent past. It may last from a few minutes to a few days, but usually lasts a few hours. The affected person functions relatively normally during the attack as they retain long term and immediate memory, but cannot remember anything for more than a minute or two, and as a result repeatedly ask the same question over and over. The memory of what happened during the period of amnesia is permanently lost, but an otherwise complete recovery is normal.

Transient global amnesia tends to run in families, but usually only occurs once or twice in a lifetime. The cause is unknown, but may be related to a migraine, or physical or emotional stress. All affected patients should have a thorough medical examination to exclude any more sinister causes (eg. brain injury or tumour).

TRANSIENT ISCHAEMIC ATTACK

A transient ischaemic attack (TIA) may be the cause for a type of funny turn or drop attack in elderly people due to a temporary miniature stroke. The usual cause is hardening and narrowing of arteries (arteriosclerosis) in the neck and brain by excessive deposition of cholesterol that causes small blood clots to form. A clot may break off from the artery wall and travel through the arteries into the brain, where it may briefly obstruct an artery, causing temporary damage to the brain tissue beyond the blockage. Spasms of arteries caused by stress, toxins or allergies, and Fabry disease may also be responsible.

The patient feels strange and acts peculiarly. There may be weakness in one arm or leg, abnormal sensations (eg. pins and needles, numbness), disturbances in vision, abnormally slurred speech, dizziness, confusion, tremor and blackouts. The symptoms may last for a few seconds or several hours. A TIA may be an early warning of narrowed arteries in the brain, and can forewarn of strokes.

All patients experiencing a TIA needs to be fully investigated by blood tests, ultrasound examination of arteries in the neck, special x-rays of arteries in the brain, and CT scans of the brain to determine the cause.
There is no specific treatment, but aspirin or warfarin taken long term in low doses prevent most TIAs, and often prevent strokes too, by preventing blood clots. The patient usually returns to normal within 24 hours.
See also CEREBROVASCULAR ACCIDENT

TRANSVERSE MYELITIS
Transverse myelitis is an inflammatory autoimmune disorder of the spinal cord that is often associated with a recent viral (eg. influenza, measles, mumps), or bacterial (eg. Mycoplasma) infection; or very rarely it is a reaction to a vaccination.
Patients develop neck or back pain, followed by altered sensations (eg. pins and needles, loss of sense of touch) and muscle weakness in the body below the area of pain, and reflexes are abnormal. It may progress to complete paraplegia or quadriplegia.
It is diagnosed by an MRI scan. Steroid injections into the spinal cord may be tried as a treatment, but they are often ineffective. There may be some spontaneous recovery, but effects are usually permanent.
See also AUTOIMMUNE DISEASES; BROWN-SÉQUARD SYNDROME; DEVIC DISEASE; MULTIPLE SCLEROSIS

TRIGEMINAL NEURALGIA
Trigeminal neuralgia (tic doloureux) is an inflammation of the trigeminal nerve, which leaves the brain and passes through a hole in the skull just beside the ear. It fans out across the face, to receive sensations from the skin of the face, and to give movement instructions to the muscles in the face. Occasionally it may be caused by a brain or nerve disease such as multiple sclerosis, or a tumour that presses on the nerve, but usually there is no specific cause
Patients develop a sudden severe pain in the face which often arises beside the mouth and spreads almost instantly up to the eye, down to the jaw, and across to the ear. The pain may last a few seconds or several minutes and only one side of the face is affected. Attacks may be started by cold winds, eating, yawning, or touching the face, and they tend to come in episodes, with attacks coming every few minutes for a few days or weeks, and then disappearing for a time. Unfortunately, each successive attack tends to last longer than the preceding one, and the pain-free periods become shorter. No specific tests are available to prove the diagnosis.
Painkillers are not particularly effective, but anti-epileptic drugs such as carbamazepine and phenytoin are quite successful treatments. If these medications prove unsuccessful, surgical exploration of the nerve may find an area of compression or abnormality as a cause of the pain. As a last resort, the nerve may be destroyed to give relief from intractable pain, but this leaves the face permanently numb.
Control is usually reasonable, but cure difficult, although spontaneous, permanent cures do occur.
See also MULTIPLE SCLEROSIS; NEURALGIA

TUBEROUS SCLEROSIS
Tuberous sclerosis (epiloia) is an uncommon nodule formation in organs of young children. It is a congenital condition (present since birth) that may occur in successive generations in the one family or develop randomly.
Repeated convulsions occur in infancy due to the presence of brain nodules. Later in childhood, intellectual disability is noted and a rash consisting of red nodules (small lumps) appears on the face and neck. Other unusual rashes may develop elsewhere on the body, and lumps may form under the nails. Eye damage, cysts in the heart, bone and lungs, and nodules in the bowel, may also develop.
No curative treatment is available, but medication is given to control convulsions, and surgery is performed to remove some of the more serious nodules. Mental retardation usually steadily worsens with age.
See also INTELLECTUAL DISABILITY

TYPHUS
Typhus is a worldwide infection caused by various types of the primitive bacteria Rickettsia, that causes a significant generalised illness. The bacteria pass to humans from rats and mice through a lice, tick or flea bite, and the incubation period is 3 days to two weeks.
It is more common around the Mediterranean Sea, in the Middle East and in East Africa, and different names are given to the infection in different parts of the world, and to infections caused by different species of Rickettsia. These include:-
- Rickettsia typhi - Almost world wide, but often in urban areas - Murine typhus.
A RATIONALE FOR THE BRAIN

- *Rickettsia australis* - Tropical Australia - Queensland Tick Typhus.
- *Rickettsia japonica* - Japan - Oriental Spotted Fever.
- *Rickettsia honei* - Flinders Island, Tasmania - Flinders Island Spotted Fever.

All forms have similar symptoms and treatment, but some are milder (eg. Siberian tick typhus) than others.

Symptoms include a black spot on the skin at the site of the tick bite, swelling of skin, a widespread red large spot rash, fever, generalised aches and pains, tiredness, headache, joint pains, loss of appetite and enlarged lymph nodes. Liver damage and skin ulceration may be complications.

It is diagnosed by specific blood tests (eg. Weil-Felix test) and/or a skin biopsy.

Treatment involves antibiotics such as tetracycline and ciprofloxacin, which settle the infection in a few days.

Without treatment, and with no complications, symptoms settle in three or four weeks.

The prognosis is usually good in western countries with appropriate treatment, but up to 10% of patients die from liver damage without medical attention.

The disease name comes from the Greek word for fever, *typhos*.

**UPPER MOTOR NEURONE LESION**

An upper motor neurone lesion is damage to the motor nerves in the brain or spinal cord above the point at which the nerves leave the spinal cord between each vertebra in the back. It results in paralysis of the muscles supplied by that nerve. There is increased muscle tone and an increase in any reflexes involving that nerve or the muscles it supplies.

**URAEMIA**

Uraemia (azotaemia) is the presence of excessive amounts of urea in the blood due to failure of the kidneys to clear this waste product of protein into the urine.

See also RENAL FAILURE, CHRONIC

**UVEOPAROTID FEVER**

See HEERFORDT SYNDROME

**VASOVAGAL SYNDROME**

The vasovagal syndrome may be the cause of repeated fainting attacks. Triggers may include stress, anxiety, or significant emotional or physical upset. Past stresses may cause an attack by the recall of a memory at times when there is another minor stress or possibility of fainting.

Patients experience recurrent episodes of fainting, low blood pressure, a pale complexion and slow heart rate. There is always the possibility of injury from falling during an attack.

Doctors may detect low blood pressure, a slow pulse and abnormal ECG (electrocardiograph) during an attack.

Patients should avoid precipitating causes (eg. prolonged standing), lie down or bend forward with the start of symptoms, and use aromatic inhalations.

See also FAINT; POST-TRAUMATIC STRESS DISORDER

**VENTRICULAR TACHYCARDIA**

See PAROXYSMAL VENTRICULAR TACHYCARDIA

**VESTIBULITIS**

Vestibulitis (vestibular neuritis) is a disturbance to the function of the three semicircular canals that form the balance mechanism in the inner ear. It may be due to inflammation or viral infection of the nerve endings supplying the balance mechanism (vestibular apparatus), and may be associated with almost any viral infection, including influenza. Excess fluid accumulates in the balance mechanism to make it malfunction.
The patient suffers constant dizziness and unsteadiness, often at rest, but certainly with any movement. It may be associated with nausea, vomiting and a ringing noise in the ears, and abnormal side-to-side eye movements (nystagmus) may be present.

There is no cure but the disturbance settles slowly with time, and medications such as prochlorperazine and diuretics) ease the symptoms.

See also BENIGN PAROXYSMAL POSITIONAL VERTIGO

VITREOUS DETACHMENT
See POSTERIOR VITREOUS DETACHMENT

VOGT-KOYANAGI-HARADA SYNDROME
The Vogt-Koyanagi-Harada syndrome is a form of inflammation of the brain, eyes and ears of unknown cause.

Patients develop recurrent attacks of encephalitis and meningitis, with uveitis (eye inflammation), detachment of the light sensitive retina from the back of the eye (causes patches of blindness), fever, headache and dizziness. It may be associated with a white patch of hair and skin, hair loss (alopecia), cataracts and glaucoma in the eyes, and deafness and ringing in the ears (tinnitus). One or both eyes may be affected.

There is no specific diagnostic test and no treatment is available. Most cases settle spontaneously with time, but permanent eye damage often occurs.

See also ENCEPHALITIS; MENINGITIS

WAGR SYNDROME
The WAGR syndrome is a genetic developmental abnormality caused by damage to chromosome 11. Its name is an acronym for the principal features that are a Wilms tumour of the kidney, no iris in the eyes (Aniridia), ovarian or testicular tumours (Gonadoblastomas) and intellectual disability (Retarded). The diagnosis can be confirmed by a chromosome analysis.

The Wilms’ and gonad tumours can be treated but the prognosis is very poor.

See also INTELLECTUAL DISABILITY

WEGENER GRANULOMATOSIS
Wegener granulomatosis is a rare condition of no known cause that causes inflammation of the blood vessels and kidneys, and ulcerating sores in the lungs, larynx, nose and sinuses. The diagnosis is often difficult but it may be detected on a chest X-ray, or by taking a biopsy (sample) of one of the sores.

The symptoms include a fever, weakness, sinusitis, shortness of breath, cough, chest pain, coughing up blood and joint pain. Permanent damage to the involved organs may occur.

Cyclophosphamide is the drug of choice in treatment. The condition is invariably fatal without treatment, but good results have been obtained with treatment.

WERDNIG-HOFFMAN SYNDROME
Werdnig-Hoffmann syndrome is an inherited progressive and permanent weakening and wasting of muscles. Patients develop muscle weakness, which attacks the muscles of the trunk more than the limbs and makes breathing more and more difficult to the point where pneumonia may occur. It is diagnosed by electrical studies of muscle action, and muscle biopsy.

No effective treatment is available, and there is no cure, but physiotherapy may be beneficial. Most patients die before the age of five years.

See also MOTOR NEURONE DISEASE

WERNICKE ENCEPHALOPATHY
See WERNICKE-KORSAKOFF PSYCHOSIS
WERNICKE-KORSAKOFF PSYCHOSIS

Wernicke-Korsakoff psychosis, or encephalopathy, is damage to the brain caused by a deficiency in vitamin B, particularly thiamine (vitamin B1). It occurs most commonly in alcoholics who neglect their diet and elderly people who are malnourished. Blood tests can confirm the low levels of thiamine in the blood.

The symptoms may include a tremor, poor coordination, confusion, sudden eye movements, double vision, and pins and needles in the hands and feet. At this stage the condition is called Wernicke encephalopathy.

If alcoholics are not treated early, permanent brain damage may result. These patients become psychotic and demented. The condition is then referred to as Wernicke-Korsakoff psychosis or Korsakoff syndrome.

For treatment thiamine is initially given by injection, and later by tablet, to replace that which is missing. A good well-balanced diet and strictly no alcohol must follow. Good treatment and patient cooperation brings most back to a reasonable lifestyle provided there has been no permanent brain damage.

Karl Wernicke (1848-1905) was a Polish neurologist and Sergei Korsakoff (1854-1900) was a Russian psychiatrist.

See also ALCOHOLISM

WEST NILE ENCEPHALITIS

West Nile encephalitis is a viral infection of the brain that spread from birds to man by a mosquito. It is found throughout North Africa, the Middle East and southeast Europe and is more common in children and the elderly. The incubation period is three to six days.

A fever and a rash are often the only symptoms, but in severe cases it may cause eye pain, enlarged tender lymph nodes, dizziness, sore throat, joint pains, headache, neck stiffness, nausea, vomiting, tiredness, disturbed mental functions and disorientation.

Tests on blood and the fluid around the brain are abnormal and can be used to confirm the diagnosis. No specific treatment or vaccine is available but it rarely causes any long-term problems. Acute attack lasts for one to two weeks and death is rare.

See also ENCEPHALITIS

WILSON DISEASE

Wilson disease (hepatolenticular degeneration) is a rare inherited disorder of copper metabolism with symptoms relating to the brain, the liver or both, that occurs in both sexes and is usually diagnosed between 10 and 30 years of age. It results in the excessive deposition of copper in the liver and brain.

Excess copper in the brain may cause psychiatric disorders, rigid muscles and a tremor. Liver disease symptoms include jaundice (yellow skin), an enlarged liver and/or spleen, anaemia and hepatitis. A brown/green ring (Kayser-Fleischer ring) around the iris (coloured part) in the eye is easily visible. The diagnosis confirmed by blood tests that detect the excessive copper.

Copper can be removed by a number of drugs (eg. penicillamine), and a diet low in copper (eg. avoiding shellfish, beans and offal) must be followed. Lifelong treatment is necessary to keep copper levels low. Any damage to the brain or liver caused before the treatment is started cannot usually be reversed, but the long-term outlook is normally good.

Samuel Wilson (1878-1937) was an English neurologist.

WITTMAACK-EK BOM SYNDROME

See RESTLESS LEGS SYNDROME

WOLFF-PARKINSON-WHITE SYNDROME

The Wolff-Parkinson-White (WPW), ventricular pre-excitation or the accelerated conduction syndrome, may be congenital or develop later in life. It is due to an abnormal nerve pathway in the heart, which allows a short circuit between the upper (atria) and lower (ventricles) chambers of the heart.

Patients have a very abnormal heart rhythm and distressing palpitations, and an electrocardiograph (ECG) shows a typical abnormal pattern. Rarely, sudden death may occur.

Acute attacks are treated by medications (eg. verapamil, propranolol, procainamide) injected into a vein and cardioversion (electric shock to heart). Long-term prevention of further attacks involves the regular use of medication and sometimes surgery on the heart to cut abnormal nerve pathways.
WOLF-HERSCHORN SYNDROME
The Wolf-Herschorn syndrome is a congenital developmental abnormality of the face caused by damage to chromosome 4.
The symptoms include intellectual disability, slow growth, an abnormal nasal bridge ("Greek helmet" appearance), cleft lip and abnormally short distance between the nose and upper lip. The diagnosis is confirmed by a chromosome analysis. Plastic surgery is the only treatment.

XANTHOMATOSIS
Xanthomatosis is a complication of excess cholesterol in the blood that settles in the skin. Small, fatty, yellow lumps appear that are almost on top of the skin. They most commonly develop around the eyes, on the knees, elbows and buttocks. Diet and medication can lower blood cholesterol levels, but skin lumps must be destroyed by cautery (burning) or removed surgically as they persist after cholesterol levels are controlled.
APPENDICES

MINI MENTAL TEST
GLASGOW COMA SCALE
MINI MENTAL TEST

The mini mental test (or mini mental state examination - MMSE) is a test performed by doctors to determine the mental capacity of patients. It is particularly used as a test for dementia. The test consists of a series of questions and tasks, each of which is allocated a score. A perfect result gives a score of 30. Levels below this show varying degrees of dementia and reduced mental capacity. The standard test and scores are as follows:-

<table>
<thead>
<tr>
<th>QUESTION/TASK</th>
<th>SCORE</th>
</tr>
</thead>
<tbody>
<tr>
<td>Name the day of the week, date, month, season and year</td>
<td>5</td>
</tr>
<tr>
<td>Name the place (building), suburb, city, state and country</td>
<td>5</td>
</tr>
<tr>
<td>The examiner names shows three objects (eg. spectacles, cup, book), and asks the patient to learn them</td>
<td>3</td>
</tr>
<tr>
<td>Serial 7s. The patient is asked to take 7 from 100 and then 7 from the result for a total of five correct answers (ie. 93, 86, 79, 72, 65)</td>
<td>5</td>
</tr>
<tr>
<td>Recall the three items previously learnt.</td>
<td>3</td>
</tr>
<tr>
<td>The examiner shows the patient a watch and asks what it does.</td>
<td>1</td>
</tr>
<tr>
<td>The examiner shows the patient a pencil and asks what it is used for.</td>
<td>1</td>
</tr>
<tr>
<td>Repeat the phrase “No ifs, and or buts”.</td>
<td>1</td>
</tr>
<tr>
<td>Follow the three commands raise your arm, point your finger, touch your nose.</td>
<td>3</td>
</tr>
<tr>
<td>Show the patient the written command “Close your eyes”</td>
<td>1</td>
</tr>
<tr>
<td>Write a sentence that makes sense. Spelling is ignored.</td>
<td>1</td>
</tr>
<tr>
<td>Copy a design shown by the examiner of two intersecting pentagons. All ten angles must be present, and they must intersect, but distortion and tremor are ignored.</td>
<td>1</td>
</tr>
<tr>
<td>TOTAL</td>
<td>30</td>
</tr>
</tbody>
</table>

GLASGOW COMA SCALE

The depth of a patient’s coma may be assessed by the Glasgow coma scale. The score is derived from the following observations and points:-

<table>
<thead>
<tr>
<th>EYE OPENING (E)</th>
<th>POINT SCORE</th>
</tr>
</thead>
<tbody>
<tr>
<td>• Spontaneous opening</td>
<td>4</td>
</tr>
<tr>
<td>• Open to verbal command</td>
<td>3</td>
</tr>
<tr>
<td>• Open to pain</td>
<td>2</td>
</tr>
<tr>
<td>• No response</td>
<td>1</td>
</tr>
</tbody>
</table>

<table>
<thead>
<tr>
<th>MOTOR RESPONSES (M)</th>
<th></th>
</tr>
</thead>
<tbody>
<tr>
<td>• Obeys verbal command</td>
<td>6</td>
</tr>
<tr>
<td>• Responds to painful stimuli by:</td>
<td></td>
</tr>
<tr>
<td>- localises pain</td>
<td>5</td>
</tr>
<tr>
<td>- withdraws from painful stimulus</td>
<td>4</td>
</tr>
<tr>
<td>- abnormal flexion</td>
<td>3</td>
</tr>
<tr>
<td>- extensor response</td>
<td>2</td>
</tr>
<tr>
<td>- no response</td>
<td>1</td>
</tr>
</tbody>
</table>

<table>
<thead>
<tr>
<th>VERBAL RESPONSES (V)</th>
<th></th>
</tr>
</thead>
<tbody>
<tr>
<td>• Oriented and converses</td>
<td>5</td>
</tr>
<tr>
<td>• Disoriented and converses</td>
<td>4</td>
</tr>
<tr>
<td>• Inappropriate words</td>
<td>3</td>
</tr>
<tr>
<td>• Incomprehensible sounds</td>
<td>2</td>
</tr>
<tr>
<td>• No response</td>
<td>1</td>
</tr>
</tbody>
</table>

COMA SCORE = E + M + V

3 = very deeply comatose  15 = completely conscious and alert