AN INDEX OF SYNDROMES AND THEIR ANAESTHETIC IMPLICATIONS

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PATIENTS often present for anaesthesia and surgery having been "labelled" with a diagnosis of some eponymous or rare syndrome. This index is an attempt to catalogue as many syndromes as possible which have anaesthetic implications and to give an indication of their main features.

It is hoped that it will be useful as a ready reference especially for those involved in paediatric anaesthesia, as many of these patients present in infancy or childhood.

The information is presented in tabular form with a cross index of alternative names. Where possible anaesthetic references are given. Where no anaesthetic references exist those given refer to descriptive literature.

SYNDROMES

<i>Name</i>	<i>Description</i>	<i>Anaesthetic Implications</i>
Adrenogenital syndrome	Inability to synthesize hydrocortisone. Virilization of female	All need hydrocortisone even if not salt-losing. Check electrolytes (1)
Albers-Schönberg disease (marble bone disease or osteopetrosis)	Brittle bones, pathological fractures	Anaemia from marrow sclerosis. Hepatosplenomegaly. Care in positioning and restraint (2)
Albright-Butler syndrome	Renal tubular acidosis hypokalaemia. Renal calculi	Correct electrolytes to within normal limits. Renal impairment (3)
Albright's osteodystrophy (pseudohypoparathyroidism)	Ectopic bone formation. Mental retardation	Hypocalcaemia – possible ECG conduction defects, neuromuscular problems. Convulsions (4)
Alport syndrome	Nephritis and nerve deafness. Renal pathology variable	Renal failure in 2nd–3rd decade. Care with renally excreted drugs (6)
Alström syndrome	Obesity, blindness by seven years. Hearing loss. Diabetes after puberty – glomerulosclerosis	Renal impairment. Management of diabetes and obesity
Amyotonia congenita (infantile muscular atrophy)	Anterior horn cell degeneration	Sensitive to thiopentone (reduced muscle mass) and respiratory depressants. Care with muscle relaxants (7–10)

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<i>Name</i>	<i>Description</i>	<i>Anaesthetic Implications</i>
Amyotrophic lateral sclerosis	Degeneration of motor neurones	Avoid succinylcholine. Possible K ⁺ release and cardiac arrest. Minimal thiopentone and curare. Avoid respiratory depressants (11)
Analbuminaemia	Almost absent albumin 4–100 mg%	Very sensitive to protein bound drugs including thiopentone, coumarin anticoagulants, curare
Andersen's disease (Glycogen storage disease IV)	Debrancher enzyme deficiency	Possibility of hypoglycaemia under anaesthesia
Aperts syndrome (acrocephalosyndactyly)	Craniosynostosis	Difficult intubation. Possibly raised intracranial pressure, associated congenital heart disease (13)
Arthrogryposis multiplex	Multiple congenital contractures	Ten per cent have congenital heart disease. Minimal thiopentone required – muscles replaced by fat. Possible airway problem with mandible (14)
Asplenia syndrome	Absent spleen, bilateral visceral right sidedness	Very complex cardiovascular anomalies, present with cyanosis and heart failure (15)
Ataxia-telangiectasia	Cerebellar ataxia. Skin and conjunctival telangiectasia. Decreased serum IgA and IgE. 10 per cent develop reticulo-endothelial malignancy	Defective immunity – recurrent chest and sinus infections. Bronchiectasis (16)
Beckwith syndrome (infantile gigantism)	Birth weight > 4,000 gm macroglossia and exomphalos	Persistent severe neonatal hypoglycaemia. Airway problems (17, 18)
Blackfan-Diamond syndrome	Congenital idiopathic red cell aplasia	Liver and spleen enlarged. Hypersplenism thrombocytopenia. Steroid therapy required (19)
Bowen's syndrome (cerebrohepatorenal syndrome)	Hepatomegaly and neonatal jaundice. Polycystic kidneys. Associated congenital heart disease. Muscular hypotonia	Hypoprothrombinaemia. Care with renally excreted drugs and muscle relaxants (20)
Carpenter's syndrome	Cranial synostosis. Associated congenital heart disease	Hypoplastic mandible. Possibly difficult intubation (13)
Central core disease	Muscular dystrophy	See amyotonia congenita (7–10)
Chediak-Higashi syndrome	Partial albinism immunodeficiency hepatosplenomegaly	Steroid therapy. Recurrent chest infection. Thrombocytopenia – may require platelets (21)

<i>Name</i>	<i>Description</i>	<i>Anaesthetic Implications</i>
Cherubism	Tumourous lesion of mandibles and maxillae with intraoral masses. May cause respiratory distress	Intubation may be extremely difficult. May require tracheostomy for acute respiratory distress. Profuse bleeding at surgery (22)
Chotzen syndrome	Craniosynostosis	May be difficult intubation. Associated renal anomalies and possible impaired renal excretion of drugs (13)
Christ-Siemens Touraine syndrome (anhidrotic ectodermal dysplasia)	Absent sweating – heat intolerance	Cannot control temperature by sweating. Persistent upper respiratory and chest infection due to poor mucus formation (23)
Chronic granulomatous disease	Inherited disorder of leucocyte function. Recurrent infections with non-pathogenic organisms	Hepatomegaly in 95 per cent. Poor pulmonary function. Avoid infection-strict asepsis (24)
Collagen diseases: dermatomyositis, rheumatoid arthritis, systemic lupus erythematosus, polyarteritis nodosa	Systemic connective diseases frequently treated with steroids. Osteoporosis and fatty infiltration of muscle. Variable systemic involvement	Often have pulmonary infiltration or fibrosis. May have temporomandibular or cricoarytenoid arthritis causing airway and intubation difficulties. Anaemia common. Risk of fat embolism after osteotomy, fracture or minor trauma (5, 86, 87)
Conradi's syndrome	Chondrodystrophy with contractures, saddle nose, mental retardation. Associated congenital heart disease and renal anomalies	Problems are those of associated renal and cardiac disease (25)
Cretinism (congenital hypothyroidism)	Absent thyroid tissue or defective synthesis thyroxine and goitre	Airway problems – large tongue, goitre. Respiratory centre very sensitive to depression. CO ₂ retention common. Hypoglycaemia, hyponatraemia, hypotension. Low cardiac output. Transfusion poorly tolerated (26)
Cri-Du-Chat syndrome	Chromosome 5-P abnormal. Abnormal cry, microcephaly micrognathia. Congenital heart disease	Airway problems – stridor, laryngomalacia. Possibly difficult intubation (27)
Crouzon disease	Craniosynostosis	Possibly difficult intubation. Severe blood loss with cranial operation (13)
Cutis laxa	Elastic fibre degeneration pendulous skin, frequent hernias. Emphysema and cor-pulmonale. Arterial fragility	Pulmonary infection, emphysema and cor-pulmonale. Poor tissues – I.V. difficult to maintain. Excess of soft tissues around larynx may lead to respiratory obstruction (28, 29)

<i>Name</i>	<i>Description</i>	<i>Anaesthetic Implications</i>
Di George's syndrome (3rd & 4th Arch syndrome)	Absent thymus and parathyroids. Immune deficiency. Susceptibility to fungal and viral infections. Treated by foetal thymic transplants	Recurrent chest infections. Hypoparathyroidism. Low Ca^{++} and tetany. Stridor. Aortic arch abnormalities and reduced cardiac output. Donor blood must be irradiated with 3000 rads to prevent graft-versus-host reaction (30)
Down's syndrome (mongolism)	Microcephaly. Small nasopharynx. Hyptonia. Sixty per cent have congenital heart disease. Duodenal atresia in some. Cervical spine abnormalities	Difficult airway – large tongue, small mouth. Risk of laryngeal spasm especially on extubation. Problems of cardiac anomalies (31, 27)
Duchenne muscular dystrophy	Muscular dystrophy with frequent cardiac muscle involvement. Usually die in 2nd decade	As for Amyotonia congenita plus cardiac involvement. Minimal drug dosage. Avoid respiratory depressants, muscle relaxants. Post-operative I.P.P.V. may be required (7-10)
Edward's syndrome (Trisomy 18(E))	Congenital heart disease in 95 per cent. Micrognathia in 80 per cent. Renal malformations 50-80 per cent. Usually die in infancy	Possible difficult intubation. Care with renally excreted drugs (27)
Ehlers-Danlos syndrome	Collagen abnormality with hyperelasticity and fragile tissues. Dissecting aneurysm of aorta. Fragility of other blood vessels. Bleeding diathesis ? cause	CVS – spontaneous rupture of vessels. Angiogram 1 per cent mortality. ECG conduction abnormalities. I.V. difficult to maintain – haematoma. Poor tissues and clotting defect lead to haemorrhage especially G.I. tract. Spontaneous pneumothorax (28, 29)
Ellis-Van-Creveld syndrome (Chondroectodermal dysplasia)	Ectodermal defects, skeletal anomalies. 50 per cent have congenital heart disease, usually septal	Chest wall anomalies lead to poor lung function. May have abnormal maxilla and upper lip, hepatosplenomegaly (32)
Epidermolysis bullosa	Erosions and blisters from minor skin trauma	Airway – oral lesions, adhesion of tongue. Ketamine is recommended or use small oral tube to avoid laryngeal trauma. Avoid skin trauma from tapes. History of steroid therapy. Check for porphyria. (Similar skin lesions) (33)
Fabry's disease	Lipid storage disease	CVS – hypertension, myocardial ischaemia (before 3rd or 4th decade). Renal failure – care with renally excreted drugs
Familial periodic paralysis	Muscle disease. Hypokalaemia, attacks of quadriplegia	Monitor serum K^{+} . Limit use of dextrose Monitor ECG. Avoid relaxants (35)

<i>Name</i>	<i>Description</i>	<i>Anaesthetic Implications</i>
Fanconi syndrome (renal tubular acidosis)	Usually 2° to other disease. Proximal tubular defect. Acidosis K ⁺ loss, dehydration	Impaired renal function. Treat electrolyte and acid-base abnormalities. Look for 1° disease (galactosaemia, cystinosis, etc.) (3, 36)
Farber's disease (lipogranulomatosis)	Sphingomyelin deposition. Widespread visceral lipogranulomas especially in the central nervous system	Deposits in larynx – careful intubation. Generalized systemic involvement leading to cardiac, renal failure (37, 29)
Favism	G-6-P-D deficiency. Haemolytic anaemia	Haemolysis following oxidant drugs, (e.g. A.S.A. and sulphas). Anaemia – transfuse if necessary (37)
Freidreich's ataxia	Degeneration of cerebellum, lateral and posterior column of spinal cord. Scoliosis. Myocardial degeneration and fibrosis	Heart failure and arrhythmias. Care with cardiac depressant drugs
Gardner's syndrome	Multiple polyposis, bony tumours, sebaceous cysts, fibromas	No anaesthetic problems described (38)
Gaucher's disease	Cerebroside accumulation in CNS, liver, spleen, etc.	Pulmonary disease from aspiration (pseudobulbar palsy). Hepatosplenomegaly, hypersplenism may cause platelet deficiency (34)
Glanzmann's disease (thrombasthenia)	Platelet ADP. reduced – abnormal function	No specific therapy for bleeding. Platelet transfusion disappointing. History of steroids (39)
Goldenhar syndrome (oculo-auriculo-vertebral syndrome)	Unilateral facial hypoplasia. Congenital heart disease in 20 per cent. Sixty per cent mandibular hypoplasia	Difficult airway and intubation. Problems of associated cardiac disease (91)
Goltz-Gorlin syndrome (focal dermal hypoplasia)	Herniae, prolapse, etc. Congenital heart disease – A.S. or A.S.D. Renal anomalies	Asymmetry of head – difficult airway (40)
Gorlin syndrome	Basal cell naevi, skeletal anomalies	No anaesthetic problem described (40)
Groenblad-Strandberg (pseudoxanthoma elasticum)	Degeneration elastic tissue in skin, eye and cardiovascular system	Rupture of arteries, especially G.I. tract, hypertension, arterial calcification. Occlusion cerebral, coronary, arteries. Difficult maintenance of I.V. cannula (29)
Guillain-Barré syndrome (acute idiopathic polyneuritis)	Acute polyneuropathy. Progressive peripheral neuritis often involving cranial nerves, bulbar palsy with hypoventilation and hypotension	Avoid succinylcholine for at least 3/12 – K ⁺ release. May require tracheostomy and IPPV, support of blood pressure (41, 42)

<i>Name</i>	<i>Description</i>	<i>Anaesthetic Implications</i>
Hand-Schüller Christian (histiocytosis X)	Histiocytic granulomata in bones and viscera – larynx, lungs, liver and spleen	Laryngeal fibrosis. Pulmonary – diffuse or hilar infiltration. Respiratory failure, cor pulmonale. Hypersplenism and pancytopenia. Liver involvement. Diabetes insipidus if sella turcica involved. History of steroids (43)
Hermansky syndrome	Albinism, bleeding diathesis	Platelet abnormality. Try platelet transfusion. No specific treatment
Holt-Oram syndrome (heart-hand syndrome)	Upper limb abnormalities. Congenital heart disease normally ASD. Possibly sudden death from pulmonary embolus, coronary occlusion	Problems of cardiac defect. No other anaesthetic problem (44)
Homocystinuria	Inborn error of metabolism. Thromboembolic phenomena due to intimal thickening. Ectopia lentis, osteoporosis, kyphoscoliosis	Dextran-70 to reduce viscosity and platelet adhesiveness, increase peripheral perfusion. Angiography may precipitate thrombosis especially cerebral (45, 46, 29)
Hunter syndrome (mucopolysaccharidosis II)	Stiff joints, dwarfing, hepatosplenomegaly. Pectus excavatum and kyphoscoliosis. Valvular and coronary heart disease	Upper airway obstruction due to infiltration of lymphoid tissue and larynx. Pneumonias. Possible hypersplenism. Cardiac failure (29, 37)
Hurler syndrome (mucopolysaccharidosis I)	Pulmonary hypertension. Usually die before ten years from respiratory and cardiac failure	As for Hunter, but more severe. Frequent upper respiratory infection. Abnormal tracheobronchial cartilages. Severe coronary artery disease at early age, valvular and myocardial involvement (29, 37)
Jervell-Nielson syndrome (cardio-auditory syndrome)	Cardiac conduction defects. Deafness	Syncope, arrhythmias. ECG-large T waves prolonged QT. May need digoxin and/or propranolol or pacemaker (47)
Kartagener's syndrome	Dextrocardia, sinusitis and bronchiectasis. Abnormal immunity. See Asplenia syndrome	Chronic respiratory infection (48)
Kasabach-Merritt syndrome	Haemangioma and thrombocytopenia. Average age at death five weeks	Haemangioma suddenly increases in size with associated severe thrombocytopenia and haemorrhage. Replace blood loss, transfuse platelets. Splenectomy. Steroids (49)
Klinefelter syndrome (gonosomal aneuploidy)	Tall stature, reduced intelligence. Vertebral collapse due to osteoporosis	No described anaesthetic problem. Care in positioning (49)

<i>Name</i>	<i>Description</i>	<i>Anaesthetic Implications</i>
Klippel-Feil syndrome	Congenital fusion two or more cervical vertebrae leading to neck rigidity	Difficult airway and intubation
Klippel-Trenaunay syndrome (angio-osteohypertrophy)	Usually unilateral. Arteriovenous fistulae thrombocytopenia.	Arteriovenous fistulae and anaemia lead to high cardiac output state. Thrombocytopenia in visceral haemangiomata (40)
Laurence-Moon-Biedl syndrome	Obesity, retinitis pigmentosa. Polydactyly. Mental retardation	May be associated with cardiac defects, renal disease and occasionally diabetes insipidus (50)
Larsens syndrome	Multiple congenital dislocations. Connective tissue defect. Poor cartilage in rib cage, epiglottis, arytenoids	Possible difficult intubation. Chronic respiratory problems (28, 29)
Leopard syndrome	Multiple large freckles. Congenital heart disease. ECG anomalies – aberrant conduction. Hypertelorism	Ninety-five per cent pulmonary stenosis
Leprechaunism	Failure to thrive, endocrine disorders, severe mental retardation	Hypoglycaemia due to hyperinsulinism from hyperplastic Islets of Langerhans. Renal tubular defects – impaired renal function (51)
Lesch-Nyhan syndrome	Hyperuricaemia and mental retardation. Renal failure by age ten	High serum uric acid leads to red cell damage and renal stones. Care with renally excreted drugs (52)
Letterer-Siwe disease (histiocytosis X)	Histiocytic granulomata in viscera and bones. Clinical course similar to acute leukaemia	Pancytopenia-anaemia and purpura haemorrhage. Pulmonary infiltration. Hepatic involvement. Gingival inflammation and necrosis, loss of teeth (43)
Lipodystrophy (total lipodystrophy)	Generalized loss all body fat. Fatty, fibrotic liver. Portal hypertension and splenomegaly. Nephropathy. Diabetes	Liver failure – avoid halothane and drugs metabolised by liver. Hypersplenism – anaemia and thrombocytopenia. Possible renal failure. Usual diabetic precautions (34)
Lowe syndrome (oculocerebrorenal syndrome)	Male only. Cataract, glaucoma, Mental Retardation. Hypotonia. Renal acidosis, proteinuria, osteoporosis and rickets	Check electrolyte and acid-base balance. Check serum Ca^{++} (treated with Vit. D and Ca^{++}) Care with renally excreted drugs (3, 53)
Maffucci syndrome	Enchondromatosis and haemangiomata with malignant change	Pathological fractures. G.I. bleeding from haemangiomata. Orthostatic hypotension. May be sensitive to vasodilator drugs
Maple-Syrup urine disease (branched chain ketonuria)	Amino acid disturbance treated by diet only. Severe neurological damage and respiratory disturbances	General supportive measures (54)

<i>Name</i>	<i>Description</i>	<i>Anaesthetic Implications</i>
Marfan's syndrome (arachnodactyly)	Connective tissue disorder. Dilatation aortic root leads to AI. Aortic, thoracic or abdominal aneurysm. Pulmonary artery, mitral valve involved. Kyphoscoliosis, pectus excavatum, lung cysts. Joint instability and dislocation	Care with myocardial depressant drugs. Beware possible dissection of aorta. Lung function poor. Possible pneumothorax. Care in positioning – easily dislocated joints (28, 29)
Maroteaux-Lamy syndrome (mucopolysaccharidosis IV)	Myocardial involvement. Kyphoscoliosis and chest infection. Hepatosplenomegaly	Heart failure by age 20. Care with cardiac depressant drugs. Chronic respiratory infection with poor lung reserve. Hypersplenism, anaemia, thrombocytopenia (29, 37)
Meckel's syndrome	Microcephaly, micrognathia and cleft epiglottis. Congenital heart disease. Renal dysplasia	Intubation may be difficult. Cardiac problems. Renal failure in infancy – care with renally excreted drugs
Median cleft face syndrome	Varying degrees of cleft face. Frontal lipomas, dermoids	Cleft nose, lip and palate may cause intubation difficulties
Möbius syndrome (congenital facial diplegia)	Associated limb deformities, micrognathia	Feeding difficulties and aspiration may cause chronic pulmonary problems. May be difficult to intubate
Morquio-Ullrich syndrome (mucopolysaccharidosis IV)	Severe dwarfing. Aortic incompetence. Thoracic deformities. Unstable atlanto-axial joint	Cardiorespiratory symptoms by 2 ^o decade. Severe kyphoscoliosis with poor lung function. All develop spinal cord damage from atlanto-occipital subluxation (29, 37, 88)
Moschkowitz disease (thrombotic thrombocytopenic purpura)	Haemolytic anaemia and thrombocytopenia. Small vessel disease. Neurological damage and renal disease. Treatment splenectomy and steroids	History of steroid therapy. Care with renally excreted drugs if kidneys affected (39)
Myasthenia congenita	Like adult myasthenia gravis	Avoid respiratory depressants, muscle relaxants. May require post-op IPPV. Problems of anticholinesterase therapy pre- and post-op. Possibility of cholinergic crisis (55–57)
Myositis ossificans (fibrodysplasia ossificans)	Bony infiltration of tendons, fascia, aponeuroses, and muscle	Airway and intubation problems if neck rigid. Thoracic involvement leads to aspiration and asphyxia, lung pathology and grossly reduced thoracic compliance (29)
Myotonia congenita (Thomsens disease)	Decreased ability to relax muscles after contraction. Diffuse hypertrophy of muscle	Avoid relaxants and depressants, as for Myotonic Dystrophy, though this is a more benign disease, non-progressive (7–10)

<i>Name</i>	<i>Description</i>	<i>Anaesthetic Implications</i>
Myotonic dystrophy (myotonia dystrophica)	Weakness and myotonia Ptosis, cataracts, partial baldness and gonadal atrophy. Cardiac conduc- tion defects and arrhyth- mias. Impaired ventila- tion	Avoid succinylcholine which causes myotonia in 50 per cent. Nondepola- rizing drugs do not relax myotonia. Neostigmine induces myotonia. Monitor ECG. Extremely sensitive to respira- tory depressants, use regional or inhala- tional agents, IPPV post-op if necessary. Halothane may cause post-op shivering and myotonia. Pulmonary complications due to poor cough (7-10, 89)
McArdle disease	Glycogen storage Disease V	Muscles affected including cardiac muscle: care with cardiac depressant drugs (12)
Neonatal hypoglycaemia (idiopathic)	Symptomatic hypo- glycaemia in infancy - convulsions, lethargy and mental retardation if un- treated. No ketosis. Therapy subtotal pan- createctomy	Extreme care in monitoring blood glu- cose. Steroids, diazoxide and glucagon as required. After pancreatectomy may require insulin and glucose to maintain blood sugar in normal limits (17)
Niemann-Pick disease	Sphingomyelin and cho- lesterol accumulation in CNS, marrow, liver and spleen. Diffuse infiltra- tion of lungs. Epilepsy, ataxia and mental retar- dation	Anaemia and thrombocytopenia due to marrow and spleen involvement. Pul- monary insufficiency, pneumonia (34)
Noack's syndrome	Craniosynostosis and digital anomalies. Obesity	May be difficult to intubate because of skull deformity (13)
Noonan syndrome (male Turner syndrome)	Short stature, mental retardation, congenital heart disease, microgna- thia. Hydronephrosis or hypoplasia of kidneys	Usually pulmonary stenosis or PDA, tetralogy, ASD. Care with renally ex- creted drugs if kidneys affected (29)
Olliers syndrome (enchondromatosis)	Multiple chondromata within bones, usually unilateral. Pathological fractures	With cavernous haemangioma is de- scribed as Maffucci syndrome. Care with positioning
Oro-facial-digital syndrome	Cleft lip and palate, lobed tongue. Hypoplas- tic mandible and maxilla. Digital anomalies. Hydrocephalus, poly- cystic kidneys	Difficult airway and intubation. Possible renal failure - care with renally excreted drugs (40)
Osler-Weber-Rendu syndrome (haemorrhagic telangiectasia)	No coagulation abnor- malities. Associated pul- monary A-V fistula	Blood loss may be impossible to control. I.V. may be difficult to maintain due to poor tissues. More than 90 per cent have recurrent chest infection, dyspnoea, cyanosis, clubbing by age 60 (39)

<i>Name</i>	<i>Description</i>	<i>Anaesthetic Implications</i>
Osteogenesis imperfecta (fragilitas ossium)	I. Congenita – stillborn or rapidly fatal. II. Tarda – Pathological fractures. Osteoporosis leads to kyphoscoliosis	Chest deformity leads to lung pathology. Fragile vessels lead to subcutaneous haemorrhage. Dentin deficiency causes carious and easily broken teeth. Extreme care in positioning (58)
Paramyotonia congenita (Eulenberg)	Myotonia on exposure to cold. Paroxysmal weakness. Serum K ⁺ may be high or low	Anaesthesia as for myotonic Dystrophy. Also care with K ⁺ -level (7–10)
Patau syndrome (trisomy 13)	Mental retardation 100 per cent. Microcephaly, micrognathia and/or dextracardia. Cleft lip or palate. Congenital heart disease. Usually fatal by three years	Difficult intubation. Usually V.S.D. (27)
Pendred syndrome	Deafness and goitre. Incomplete block of thyroxine production	May be euthyroid or hypothyroid. Make sure euthyroid pre-op. Otherwise as for cretinism (26, 59)
Phenyl ketonuria	Phenyl alanine hydroxylase deficiency. Vomiting, irritability, mental retardation, Hypertonia, convulsions. Very sensitive to narcotics and other CNS depressants	Inhalation induction and maintenance. Continue anti-epileptic drugs. Tendency to hypoglycaemia – 100 per cent Dextrose infusion (26)
Pierre Robin syndrome	Cleft palate, micrognathia, glossoptosis. Associated congenital heart disease may occur	Newborn may asphyxiate – nurse prone on frame. May require tongue suture, intubation or tracheostomy. May be very difficult to intubate. Awake intubation (61)
Polycystic kidneys	One-third have associated cysts in liver, pancreas, spleen, lungs, bladder, thyroid. Cerebral aneurysm in 15 per cent	Care with renally excreted drugs. Beware lung cysts: may lead to pneumothorax. Avoid hypertension because of possible cerebral aneurysm (62)
Polycystic liver	Familial – 60 per cent have polycystic kidneys, lung and pancreas	Usually liver function not impaired until late – fibrosis, splenomegaly, oesophageal varices, etc. Beware possible renal failure, lung cysts (62)
Polysplenia	Bilateral visceral left sidedness (see also asplenia – converse)	Complex cardiac anomalies common: ASD and endocardial cushion defects. Usually not so complex as in asplenia (25)
Pompe's disease (glycogen storage II)	Muscle deposits – severe hypotonicity. Massive cardiomegaly. Death before two years of age	Extreme care, avoidance respiratory depressants, muscle relaxants, cardiac depressants. Large tongue may cause airway problem (12)

<i>Name</i>	<i>Description</i>	<i>Anaesthetic Implications</i>
Porphyria	Paralyses, psychiatric disorder. Autonomic imbalance – hypertension, tachycardia. Abdominal pain precipitated by drugs, infections, etc.	Avoid barbiturates including thiopentone. Sedatives – meprobamate, librium, glutethimide, carbromal. Hydroxydione (steroid anaesthetic) Nitethamide. Hydantoin derivatives. Sulphonamides. Antipyretics. Hypoglycaemic agents. <i>Have been used safely:</i> chlorpromazine, promazine, promethazine, chloral, propanidid, morphine, pethidine, Procaine, N ₂ O, Ether, Succinylcholine, 4-tubocurarine, gallamine. Atropine and neostigmine. Pentolinium (37)
Prader-Willi syndrome	Neonate – hypotonia, poor feeding, absent reflexes. <i>Second phase</i> – hyperactive, uncontrollable polyphagia, mental retardation	Obesity of extreme proportions leading to cardio-pulmonary failure (63)
Progeria (Hutchinson-Gilford syndrome)	Premature ageing starts 6 months–3 years. Cardiac disease – ischaemia, hypertension, cardiomegaly	Anaesthesia as for adults with myocardial ischaemia (64)
Prune Belly syndrome	Agenesis of abdominal musculature with renal anomalies	Poor cough – respiratory infections. Respiration requires use of accessory muscles. Treat as full stomach. Intubate and assist or control ventilation. Avoid muscle relaxants. Beware possible renal failure (65)
Pyle's disease (metaphyseal dysplasia)	Craniofacial abnormalities. Enlarged mandible. Cranial N. paralyses	No described anaesthetic problem (66)
Rieger syndrome	Myotonic dystrophy and other myopathies. Hypoplasia of maxilla, abnormal teeth, mental retardation. Occasional imperforate anus	Anaesthetic requirements dictated by associated muscle disease – see amyotonia congenita, myotonic dystrophy (7–10)
Riley-Day syndrome (familial dysautonomia)	Deficiency of dopamine hydroxylase. Hyper- and hypotensive attacks, absent lacrimation, abnormal sweating. Insensitive to pain. Poor suckling and swallowing	Emotional lability. Recurrent aspiration, pneumonia and chronic lung disease. Labile blood pressure – care with halothane etc. Sensitive to adrenergic and cholinergic drugs. Pre-medication – atropine and chlorpromazine. Respiratory centre insensitive to CO ₂ – need IPPV. Avoid respiratory depressants (67, 90)
Rubinstein syndrome	Mental retardation, microcephaly. Frequent chest infections. Swallowing abnormality. Congenital heart disease	Repeated aspiration leads to pneumonia and chronic lung disease (68)

<i>Name</i>	<i>Description</i>	<i>Anaesthetic Implications</i>
San-Filippo syndrome (mucopolysaccharidosis III)	CNS malfunction in childhood progresses to mental retardation and dementia. No hepatosplenomegaly or cardiac problems	No anaesthetic problems described. Emotional disturbance, agitation and dementia (29, 37)
Scheie disease (mucopolysaccharidosis V)	Corneal clouding, hernias. Joint stiffness especially hands and feet. Aortic valve involvement	Aortic incompetence by third decade. Joint stiffness – care in positioning (29, 37)
Scleroderma	Diffuse cutaneous stiffening. Plastic surgery required for contractures and constrictions	Scarring face and mouth – difficult airway and intubation. Chest restriction – poor compliance. Diffuse pulmonary fibrosis – hypoxia. Veins – often invisible and impalpable. Cardiac fibrosis or cor pulmonale. History of steroid therapy (69)
Sebaceous naevi (linear)	Linear naevi from forehead to nose. Hydrocephalus, mental retardation, associated with coarctation and hypoplasia of aorta	Cardiovascular complications (40)
Shy-Drager syndrome	Orthostatic hypotension. Diffuse degeneration of C.N.S., and autonomic NS. Decreased sweating. Hypersensitive to angiotensin and epinephrine	Labile pulse and blood pressure possibly due to defective baroreceptor response – avoid methoxyflurane, cyclopropane and ether. Cautious use of halothane. Treat hypotension with infusion Neosynephrine (70)
Silver syndrome	Short stature, skeletal asymmetry. Micrognathia. Abnormal sexual development	Possibly difficult intubation (91)
Sipple syndrome (multiple endocrine adenomatosis type II)	Phaeochromocytoma and medullary thyroid carcinoma. Parathyroid adenoma, CNS tumours. Schwannoma of mediastinum associated. Cushing's disease	Management of phaeochromocytoma. (75 per cent bilateral). Problems of multiple endocrine disorders (71)
Smith-Lemli-Opitz syndrome	Mental retardation. Genital and skeletal anomalies – micrognathia. Thymic hypoplasia	Airway and intubation problems. Pneumonia, possible increased susceptibility to infection (91)
Sotos syndrome (cerebral gigantism)	Acromegalic features. Dilated ventricles but normal intracranial pressure	All features non-progressive. Possible airway problems due to acromegalic skull. No other described problems (91)

<i>Name</i>	<i>Description</i>	<i>Anaesthetic Implications</i>
Stevens-Johnson syndrome	Erythema multiforme, urticarial lesions and erosions of mouth, eyes genitalia. Possibly hypersensitivity to exogenous agents. (Drugs, infections, etc.)	Oral lesions – avoid intubation, oesophageal stethoscope. Monitoring difficult because of skin lesions but essential. ECG – fibrillation, myocarditis, pericarditis occur. Temperature control – febrile episodes. Intravenous infusion – essential but avoid cut-down, because of infection. Ketamine probably best anaesthetic. Pleural blebs and pneumothorax may occur (72)
Sturge-Weber syndrome	Cavernous angioma over 1–3 divisions. Vth cranial nerve. Intracranial calcification, convulsions and maybe progressive neurological deficit	No specific anaesthetic problems (73)
Supravalvular aortic stenosis syndrome (idiopathic infantile hypercalcaemia) (William's syndrome)	Hypercalcaemia and mental retardation. Abnormal facies. Cardiac – dyspnoea, angina. Therapy, low calcium diet, steroids. Cardiac surgery	Fixed cardiac output and ischaemia. History of steroids. Monitor serum Ca++ (74)
Tangier disease (alphalipoproteinaemia)	Low serum cholesterol. Orange tonsils and rectal mucosa. Splenomegaly. 50 per cent neurological abnormality. Premature coronary disease	Anaemia and thrombocytopenia due to hypersplenism. Abnormal EMG – care with muscle relaxants. Beware premature ischaemic heart disease (34)
Tay-Sach's disease	Gangliosidosis. Blindness and progressive dementia and degeneration of central nervous system	No described anaesthetic hazard. Progressive neurological loss leads to respiratory complications. Supportive measures only treatment (34)
Thomsen's disease	See Myotonia congenita	
Thrombocytopenia with absent radius	Episodic thrombocytopenia precipitated by stress, infection, surgery, etc. Low platelets improve to normal by adulthood. Congenital heart disease in 30 per cent	Platelet transfusion for surgery or bleeding (35–40 per cent die in first year of intracranial haemorrhage). Avoid elective surgery in first year (75)
Treacher Collins syndrome (mandibulo-facial dysostosis)	Micrognathia and aplastic zygomatic arches. Microstomia, choanal atresia. Congenital heart disease may occur	Possible airway and intubation difficulties. Less severe than Pierre Robin deformity (61)
Tuberous sclerosis	Adenoma sebaceum of skin, epilepsy and mental retardation. Intracranial calcification in 50 per cent. Hamartomas in lungs, kidneys, heart	Kidneys – pyelonephritis and renal failure. Care with renally excreted drugs. Lungs – possible rupture of lung cysts. Possible cardiac arrhythmia (76)

<i>Name</i>	<i>Description</i>	<i>Anaesthetic Implications</i>
Turner's syndrome	XO chromosome. Micrognathia, Coarctation, dissecting aneurysm of aorta or pulmonary stenosis. Renal anomalies in more than 50 per cent	Possibly difficult intubation. Cardiovascular abnormality. Possible renal disease – care with renally excreted drugs (77)
Urbach-Wiethe disease (cutaneous-mucosal hyalinosis)	Type of histiocytosis (see Hand-Schüller-Christian disease). Hoarseness or aphonia. Hyaline deposits in larynx and pharynx	Cautious intubation (43)
Von Gierke's disease	Glycogen storage disease I. Hepatomegaly, enlarged kidneys, severe attacks of hypoglycaemia	Monitor blood sugar and acid-base balance (I.V. glucose infusion). Diazoxide for hypoglycaemia (12)
von-Hippel-Lindau syndrome	Retinal or CNS haemangioblastoma. (Posterior fossa or spinal cord). Associated with pheochromocytoma, renal, pancreatic, or hepatic cysts	Problems those of associated pheochromocytoma, renal and hepatic pathology (71)
von-Recklinghausen disease (neurofibromatosis)	Café-au-lait spots. Tumours all parts CNS. Peripheral tumours associated with nerve trunks. Increased incidence pheochromocytoma. Fifty per cent Kyphoscoliosis. Honeycomb cystic lung changes. Renal artery dysplasia and hypertension	Screen for pheochromocytoma (urinary VMA). Should be investigated for lung function. Tumours may occur in the larynx and right ventricle outflow tract. Care with renally excreted drugs if kidneys involved. (78, 79)
von Willebrand's disease (pseudohaemophilia)	Prolonged bleeding time (decreased factor VIII activity) due to defective platelet adhesiveness. ? Capillary abnormality	Bleeding can be controlled by fresh or fresh frozen plasma, cryoprecipitate. Avoid salicylate therapy (effect on platelets, possible GI bleeding) (39)
Weber Christian disease (chronic non-suppurative panniculitis)	Necrosis of fat, in any situation including retroperitoneal, pericardial, meningeal	Involvement of retroperitoneal tissues may cause acute or chronic adrenal insufficiency, of pericardium leads to restrictive pericarditis, of meninges causes convulsions. Avoid trauma to fat by heat, cold or pressure (80)
Welander muscular atrophy	Initial involvement peripheral muscles. Prognosis good for life, poor for ambulation	May require spinal fusion. Extreme care with thiopentone, muscle relaxants. Avoid respiratory depressant drugs (7-10)

<i>Name</i>	<i>Description</i>	<i>Anaesthetic Implications</i>
Werdnig-Hoffman disease	Infantile muscular atrophy more severe than Werdnig. Feeding difficulties, aspiration, usually death before puberty	Chronic respiratory problems. Minimal anaesthesia required. Avoid muscle relaxants and respiratory depressant drugs. Ventilatory support may be required and weaning may be difficult (7-10)
Werner syndrome	Premature ageing, diabetes. Early cataracts. Mental retardation in 50 per cent. Bony lesions like osteomyelitis. Cardiac infarction and failure	Anaesthesia as for adult with myocardial ischaemia (29)
Wermer syndrome (multiple endocrine adenomatosis type I)	Hyperparathyroidism. Tumours of pituitary and pancreatic islet cells. Gastric ulcer. Occasionally have carcinoid tumours of bronchial tree	Renal failure due to stones. Hypoglycaemia from hyperinsulinism (81)
Wilson's disease (hepatolenticular degeneration)	Decreased ceruloplasmin causes abnormal copper deposits especially in liver, and CNS motor nuclei. Renal tubular acidosis	Hepatic failure due to fibrosis. Thiopentone may be used in small doses. Muscle relaxants – succinylcholine apnoea rare despite pseudocholinesterase reduction. d-Tubocurarine – short action due to globin binding. Care with renally excreted drugs (37, 82)
Wilson-Mikity syndrome	Prematurity < 1500 gm birth weight. Severe chronic lung disease leading to fibrosis and cystic areas aetiology – ? oxygen toxicity	Right heart failure. Repeated chest infection and aspiration. Use of steroids to prevent pulmonary fibrosis (83, 84)
Wiskott-Aldrich syndrome	Immunodeficiency with thrombocytopenia. One hundred per cent have low platelets, absent iso-haemagglutinins. High IgA, low IgM. Eczema, asthma	Blood transfusion and platelets may be required. Bone marrow transplantation has been used. All blood products must be irradiated with 3,000 rads. to prevent graft-vs.-host reaction. Avoid contamination – often die from generalized herpes or infection by not-pathogenic organisms (39)
Wolff-Parkinson-White syndrome	ECG abnormality – Short P.R., prolonged QRS with phasic variation in 40 per cent. Associated with many cardiac defects. Anomalous conduction path between atria and ventricles	Hyoscine preferred to atropine as drying agent. Tachycardia due to atropine or apprehension may change ECG and suggest infarction, with ST segment depression. Paroxysmal SVT on induction of anaesthesia or during cardiac surgery has been reported. Should be treated with digitalis, propranolol, pacemaker if necessary. Neostigmine may accentuate W-P-W- pattern (85)

<i>Name</i>	<i>Description</i>	<i>Anaesthetic Implications</i>
Wolman's disease (familial xanthomatosis)	Adrenal calcification. Resembles Niemann- Pick disease with hepato- splenomegaly and hyper- splenism. Involvement other tissues from foam cells, including myocar- dium	Anaemia, thrombocytopenia. Platelet transfusion only successful post- splenectomy (34)

Acrocephalosyndactyly – Apert S.
 Albinism – Chediak-Higashi S., Hermansky S.
 Amaurosis congenita – Leber S.
 Anhidrotic ectodermal dysplasia – Christ-Siemens-Touraine
 Bournville's disease – Tuberous sclerosis
 Branched chain Ketamuria – Maple-Syrup urine D.
 Cardio-Auditory S. – Jervell-Nielson S.
 Cerebrohepatorenal S. – Bowens S.
 Chondrodystrophies – Conradi S., Ellis-van-Creveld S.
 Connective tissue disorders – Cutis Laxa, Ehlers Danlos S., Groenblad-Strandberg S., Marfans S.
 Cranial synostoses – Apert S., Carpenters S., Chotzen S., Crouzon S., Noack S.
 Enchondromatosis – Maffucci S., Olliers S.
 Eulenberg disease – Paramyotonia congenita
 Familial dysautonomia – Riley Day S.
 Fragilitas Ossium – Osteogenesis imperfecta
 Glycogen Storage Diseases – I Von Gierke, II Pompe, III Forbes, IV Andersen, V McArdle, VI
 Hers, VII Lewis
 Goitre – Cretinism, Pendred's S.
 Heart and Hand S. – Hold-Oram S.
 Heredity haemorrhagic telangiectasia – Osler-Weber-Rendu S.
 Heptaolenticular degeneration – Wilsons D.
 Histiocytosis X Syndromes – Hand Schüller-Christian, Letterer-Siwe, Urbach-Wiethe
 Homogentisic-Aciduria – Alkaptonuria
 Hutchinson-Gilford S. – Progeria
 Idiopathic Infantile Hypercalcaemia – See Supravalvular aortic stenosis syndrome
 Immunodeficiency Syndromes – Ataxia – telangiectasia, Chediak-Higashi, Chronic granulomatous
 disease, Wiskott-Aldrich S.
 Lipid storage diseases – Fabry's disease, Gauchers disease, Niemann-Pick disease, Tay-Sacks
 disease, Wolman's disease
 Lipogranulomatosis – Farber's disease
 Louis-Bar disease – Ataxia Telangiectasia
 Mandibulo-Facial dysostosis – Treacher-Collins Syndrome
 Marble-bone disease – Albers Schönberg
 Mongolism – Down's syndrome
 Mucopolysaccharidoses – I Hurler, II Hunter, III San Filippo, IV Morquio-Ullrich, V Scheie, VI
 Maroteaux-Lamy
 Multiple Endocrine Adenomatosis – I Wermer Syndrome, II Sipple Syndrome
 Muscular atrophy – amyotonia congenita, Welanders muscular atrophy, Werdnig-Hoffman
 disease
 Muscular dystrophy – Duchenne, Central core (Shy Magee), Reigers S.
 Myotonic syndromes – Myotonia congenita, dystrophia myotonica, Reigers S.
 Neonatal hypocalcaemia – Beckwith syndrome, Idiopathic, Glycogen-storage disease esp. Pompe's
 Neurofibromatosis –
 Oculoauriculovertebral S. – Goldenhar S.
 Oculocerebrorenal S. – Lowe S.
 Phaeochromocytoma – Sipple S., von-Hippel-Lindau S., von Recklinghausen S.
 Pseudohypoparathyroidism – Albright's Osteodystrophy
 Renal Tubular Acidosis – Albright Butler S., Fanconi S., Lowes S.
 Shy-Magee – Central core disease
 Total lipoatrophy – Lipodystrophy

Telangiectasia – Ataxia telangiectasis, Osler-Weber-Rendu S.
 Thrombasthenia – Glanzmann's disease
 Thrombotic thrombocytopenic purpura – Moschkowitz disease

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