



Winter-Baraitser Dysmorphology Database

A list of features & definitions



Acknowledgments

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Introduction

This document has been prepared for publication on the DYSCERNE website by the DYSCERNE Coordinating Centre based at the University of Manchester, UK. DYSCERNE is a European Commission funded project which aims to improve the diagnosis, clinical management and information dissemination for rare dysmorphic diseases. More information about DYSCERNE can be found at www.dyscerne.org

This document contains the list of features and definitions from the Winter-Baraitser Dysmorphology Database. DYSCERNE have edited some of these definitions and added definitions where none exist for completeness. Features are sorted alphabetically by Body System, which are all listed in the Contents page. Bookmarks to the side of this Pdf will take you directly to the desired Body System. To search for a particular feature please use the 'find' function in your pdf reader.

Contents

ABDOMEN.....	4	LOWER LIMBS.....	50
BACK AND SPINE.....	8	MOUTH.....	52
BLOOD VESSELS.....	10	MUSCLES.....	54
BUILD.....	11	NAILS.....	55
CRANIUM.....	12	NECK.....	56
EARS.....	16	NEUROLOGY.....	57
ENDOCRINE.....	20	NOSE.....	60
EYES. ASSOCIATED STRUCTURES.....	23	ORAL REGION.....	63
EYES, GLOBES.....	26	PELVIS.....	65
FACE.....	31	SKELETAL.....	66
FEET.....	33	SKIN.....	69
FOREHEAD.....	37	STATURE.....	74
GENITALIA.....	38	TEETH.....	75
HAEMATOL/IMMUNOLOGY.....	41	THORAX.....	76
HAIR.....	42	UPPER LIMBS.....	84
HANDS.....	44	URINARY SYSTEM.....	87
JOINTS.....	49	VOICE.....	90

Body System (and feature path)	Feature	Useful comments aiding choice of feature
ABDOMEN		
Abdomen, general abnormalities	Abdominal muscle hypoplasia/aplasia	A generalised deficiency of muscle. It includes 'prune belly'.
	Ascites	Fluid in the abdomen (peritoneal cavity).
	Common mesentery	An abnormality in mesenteric attachment
	Constipation	Sometimes a manifestation of Hirschprung disease, or secondary to gross hypotonia. Use with caution.
	Diarrhea	Frequent & watery bowel movements. Many underlying causes.
	Diastasis recti	A separation of the midline abdominal muscles, leaving a gap in between.
	Dysphagia	Difficulty with swallowing due to pain or incoordination. Note there is a separate entry for cranial nerve palsies and when in doubt use both together.
	Feeding problems in infants	A symptom rather than a sign!
	Gastrointestinal tumour/polyp/haemangioma	Any tumour.
	Gastroschisis	The protruding loops of bowel are not covered by a membrane. It is mostly para-umbilical. Should be distinguished from omphalocele/exomphalos where the intestines are covered by a membrane.
	Inguinal hernia	The presence of abdominal viscera in the inguinal canal (the open communication of the peritoneal cavity with the diverticulum known as the processus vaginalis (the inguinal canal).
Intestinal duplication	These are cystic, tubular structures lying alongside the intestine, mostly partially incorporated into it. Most frequently at the ileum, but they might occur at any level.	

Body System (and feature path)	Feature	Useful comments aiding choice of feature
ABDOMEN (continued)		
Abdomen, general abnormalities (continued)	Intestinal malrotation	The normal rotation and fixation of the gut does not take place. The caecum might end up in the epigastrium to the left of the duodenum. Volvulus might occur.
	Lymphangiectasia of intestine	There are abnormally dilated lymphatic vessels in the lamina propria, with leakage of lymph into the gut. Congenital types have in addition extra-intestinal lymphatic abnormalities.
	Malabsorption	Includes steatorrhea, or any chronic absorption problem.
	Meckel diverticulum	This is a vitelline duct (omphalo-mesenteric duct) remnant. It normally disappears by 10 weeks of intrauterine life. The diverticulum results from incomplete obliteration.
	Omphalocele/exomphalos	Results from the failure of the intestine to return to the body cavity during development. It is midline, at the point of insertion of the umbilical cord. The contents are enclosed in a membranous sac. Intestine, liver pancreas, spleen may be present in the sac.
	Protuberant abdomen	Protuberant as in "blown out", like a balloon. Note the separate feature abdominal muscle hypoplasia . Use together if that is the cause.
	Situs inversus-abdominal	Lateral transposition of the abdominal organs. Spleen and liver on the wrong side, for instance. Can be complete or incomplete.
	Small bowel atresia/absence/obstruction/short	Includes multiple obstructions or ileal or jejunal atresias. There is a separate feature for duodenal atresia .
	Telangiectasia of intestine	Superficial, abnormal, tangles of small vessels. Like spider legs.
	Umbilical hernia	Differs from an omphalocele in that at most it contains a mid-gut loop but no viscera. Umbilical herniae are often seen in those of African origin, and are of no significance.
Umbilicus, abnormal shape or position	Includes redundant skin.	

Body System (and feature path)	Feature	Useful comments aiding choice of feature
ABDOMEN (continued)		
Anus, general abnormalities	Abnormal placement of anus	Also includes an ectopic anus. In most instances there is an anterior displacement, with, in females, possible communication with the genito-urinary tract.
	Anal atresia/stenosis	Also includes "covered" anus and imperforate anus.
	Anal fistula	Fistulae to the genito-urinary tract or perineum (seen in imperforate anus)
Colon, general abnormalities	Colon/caecum atresia	If part of multiple atresias, add the others in separate boxes. Colon not as frequent as small bowel.
	Colonic tumours	Any tumour.
	Diverticulosis of colon	Mostly at the sigmoid flexure. Usually multiple diverticulae (sac-like pouches).
	Megacolon or Hirschsprung syndrome	Typically there is a dilated hypertrophic segment, a cone and a narrow zone. There must be, histologically, a complete depletion of ganglion cells from myenteric, mucosal and submucosal plexuses.
Duodenum, general abnormalities	Duodenal atresia	The commonest has the pancreas occupying the blind ends between the atresia. Accounts for 23% of bowel atresias. Use with duodenal stenosis as this might be a spectrum.
	Duodenal stenosis	Narrowing. Use with duodenal atresia as this might be a spectrum.
	Duodenal ulcers	Seldom congenital, but well documented in infants and children.
Liver/biliary system, general abnormalities	Abnormal liver (including function)	Enlarged liver is a separate feature. Use together when appropriate.
	Absent/abnormal gallbladder	Hypoplasia, duplication, and abnormal position are all included.
	Biliary atresia/stenosis	This includes intra- and extrahepatic occlusion.
	Choledochal cyst	congenital anomaly associated with benign cystic dilatation of the bile duct

Body System (and feature path)	Feature	Useful comments aiding choice of feature
ABDOMEN (continued)		
Liver/biliary system, general abnormalities (continued)	Enlarged liver	Use with abnormal liver function .
	Jaundice	Any type, except physiological jaundice.
	Liver cysts/tumours/angiomas	Any mass lesion.
	Plasma ammonia abnormal	Beware of false positives in elevated levels
Oesophagus, general abnormalities	Abnormal motility of oesophagus/achalasia	Achalasia is due to the failure of the lower end of the oesophagus to relax (cardiospasm).
	Oesophageal atresia or stenosis	Most atresias (95%) are associated with a fistula. The commonest type ends in a pouch at T2. It is caused by a failure of the ventral diverticula of the foregut, which will form the trachea, to separate from the oesophagus.
	Tracheo-oesophageal fistula	The oesophagus ends as a pouch, and a fistula connects the oesophagus and trachea. The commonest is type 1 (there are 6 types) where the fistula enters the trachea (often stenosed at entry site) from the non-blind ending lower part of the oesophagus.
Pancreas (exocrine), general abnormalities	Annular pancreas	The pancreas might surround the second part of the duodenum, due to the persistence of the ventral part. Seen in atresia of the second part of the duodenum, but also alone.
	Ectopic/additional pancreas tissue	Nodules of pancreas can occur throughout the gastrointestinal tract.
	Malformation of the pancreas	Congenital hypoplasia, complete aplasia.
	Pancreatic cysts/tumours	Of any type
	Pancreatic insufficiency	Diminished excretion of lipase, amylase and trypsin, which manifests clinically in failure to thrive, and steatorrhea.
Spleen, general abnormalities	Absent or hypoplastic spleen	Agensis, asplenia are two other words used.
	Ectopic/accessory spleen	Use when appropriate with situs inversus if the extra spleen(s) are on the right side.

Body System (and feature path)	Feature	Useful comments aiding choice of feature
ABDOMEN (continued)		
Spleen, general abnormalities (continued)	Large spleen	By palpation or imaging.
	Polysplenia	Accessory spleens on the left side.
	Splenic abnormalities, unspecified	Better to use spleen, general abnormalities
Stomach, general abnormalities	Pyloric stenosis	A tumour-like mass of pale muscle, 3-5cm long, situated at the pylorus. It seems to derive from the circular layer of fibers.
	Small stomach/microgastria	Just what it says.
	Stomach tumours	Any tumour.
	Stomach ulcers	These are peptic ulcers, the diagnosis of which is based on history, imaging and endoscopy.
BACK AND SPINE		
Back and spine, general abnormalities	Gibbus	In Latin a "hump". A flexion angulation of the spine.
	Kyphosis	A bent spine. Flexion angulation of the spine over many segments.
	Lordosis	Literally meaning "bent backwards". Mostly seen in the lumbar region and it is an exaggeration of the normal hollowed out configuration in that area.
	Meningocele/meningo-myelocele	A defect in the closure of the neural tube resulting in the herniation of the meninges through the gap. These lesions may also be described as open spina bifida.
	Pilonidal cyst/sinus/dimple	Any hole other than a sacral dimple or sinus which are features found under Sacrum .
	Rigid spine	A stiff, immobile spine. Could be the result of pathology of muscle, joints, ligaments or bone.
	Scoliosis	A lateral curvature of the spine.

Body System (and feature path)	Feature	Useful comments aiding choice of feature
BACK AND SPINE (continued)		
Back and spine, general abnormalities (continued)	Short trunk	Can use in conjunction with short stature -short trunk found under 'build - short stature'.
	Spinal dysraphism	Use for midline hairy patches/lipomas/diastematomyelia. For open spina bifida, use ' meningocele/meningo-myelocele '. See under 'vertebrae' for spina bifida occulta .
	Spinal tumours	All types.
	Syringomyelia	A fluid containing, cystic swelling in the central canal, which expands the cord.
Sacrum, general abnormalities	Abnormal sacrum	Can be used with absent sacrum , if it is very small and malformed.
	Absent sacrum	Must be confirmed radiologically. Includes sacral agenesis. If very small (not quite absent) can use together with abnormal sacrum .
	Caudal appendage	A tail or other protuberance.
	Sacral dimple/sinus	Dimples, channels, canals overlying the sacrum.
	Sacral teratoma/tumour	Pre-sacral and sacral tumours. If the sacrum is in addition small or absent, can use sacral teratoma and absent sacrum together as they can be variable manifestations of the same condition.
Vertebrae, general abnormalities	Atlanto/axial/occipital abnormalities	Fusion, missing arches, occipitalisation, bony defects.
	Beaked/wedged vertebrae	The body of the vertebrae slopes forwards. It looks like a triangular wedge (from the side).
	Coronal clefts of vertebrae	Seen on a lateral radiograph, coronal clefts are normal in the newborn and represent delay in fusion of ventral and dorsal ossification centers of the vertebral body. It consists of cartilage. Later it is pathological.
	Fusion of vertebrae	Mostly fusion of the body, but other parts (the spinous processes and arch) might be included. Block vertebrae.
	Hemivertebrae	Half the vertebra fails to develop, and with it the costal element.

Body System (and feature path)	Feature	Useful comments aiding choice of feature
BACK AND SPINE (continued)		
Vertebrae, general abnormalities (continued)	Irregular end-plates to vertebrae	An irregularity of the articular surfaces between the vertebral bodies.
	Odontoid hypoplasia/dysplasia	A small or absent odontoid peg, as seen radiologically. There is a separate feature for platybasia which might accompany defects of the odontoid.
	Platyspondyly	The vertebral bodies are flattened.
	Segmentation defects of spine	Extra or missing vertebrae.
	Spina bifida occulta	A closed spina bifida. One with a skin covering. Note separate features for sacral dimple and spinal dysraphism . If appropriate can use together as this is a continuum.
	Spondylo-epiphyseal dysplasia	Abnormal epiphyses (the growing articular end of long bones) and flattened vertebrae. Use with epiphyseal dysplasia if uncertain.
	Spondylolisthesis	Slipping forward of one vertebra on the next. Occurs most commonly at the lumbosacral junction.
	Tall vertebrae	Tall vertebral bodies. The height on a lateral radiograph is greater than the width.
	Vertebrae, unossified	Unossified, or poorly ossified, as seen radiologically.
	Vertebral interpedicular distance, narrow	Might be an indication (seen on lateral radiograph) of spinal cord stenosis.
BLOOD VESSELS		
Blood vessels, general abnormalities	Aneurysms	Saccular expansion a blood vessel, mostly due to weakness of the vessel wall. Includes berry aneurysm and cirroid aneurysms.
	Aplasia of blood vessels	Defective or retarded development of blood vessels
	Arterial stenosis	Narrowing of the artery.
	Arterio-venous malformations	A tangled mass of vessels due to direct communication between artery and vein. There is no intervening capillary bed.

Body System (and feature path)	Feature	Useful comments aiding choice of feature
BLOOD VESSELS		
Blood vessels, general abnormalities (continued)	Arteritis	Diseases characterised by inflammation of arterial vessels. Includes vasculitis.
	Calcification of arteries	A radiological feature. Subcutaneous calcification or muscle calcification are separate features.
	Carotid aplasia	A rare anomaly of the cerebral vessels. May be associated with aneurysm or abnormal collateral channels.
	Cholesterol/lipids, abnormal	Any abnormality of cholesterol, lipids or triglycerides.
	Dissection of the aorta	A splitting (degeneration) lets blood into the layers of the wall of the vessel, causing further stripping of one layer from another.
	Hypertension	Raised blood pressure for any reason.
	Peripheral gangrene	The necrotic end result of poor arterial supply
	Premature atherosclerosis	Evidence of extensive vascular disease from atheroma in young adulthood or below. Might be part of premature ageing.
	Tortuosity of arteries	The arteries are twisted, sometimes like a corkscrew.
	Varicose veins	Enlarged, tortuous veins. May only be significant in young patients.
Vascular ring	A congenital defect in which there is an abnormal formation of the aorta and/or its surrounding blood vessels. The trachea and esophagus are completely encircled and sometimes compressed by a "ring" formed by these vessels, which can lead to breathing and digestive difficulties.	
BUILD		
Muscular build, general abnormalities	Muscular build	Refers to the impression of a muscular build as seen in the lipodystrophies where there is a deficiency of subcutaneous tissue. Also a true muscular build as seen in myotonia congenita.

Body System (and feature path)	Feature	Useful comments aiding choice of feature
BUILD (continued)		
Obesity, general abnormalities	Generalized obesity	An increase of bodily fat out of proportion to the family pattern, resulting in an increase of weight above the 97th centile for age
	High birth weight (> 90th centile)	As judged by gestational age. Advanced bone-age is a separate feature which might be present.
	Truncal obesity	Where the abundant fat distribution involves the trunk alone and spares the face.
Thin or slender build, general abnormalities	Low birthweight (< 3rd centile)	Below the 3rd centile for gestational age.
	Thin	Difficult to judge when anyone is abnormally thin. Use with caution and take the family pattern into account.
CRANIUM		
Brain, general abnormalities	Agenesis/hypoplasia of corpus callosum	The 8cm long interhemispheric structure joining the two halves of the brain. Includes absence or small.
	Anencephaly/craniorachischisis	An absence of development of the cerebral hemispheres plus the skull vault.
	Anterior encephalocele/meningocele	An encephalocele protruding from the forehead, between the eyes, or through the ethmoid.
	Aqueduct stenosis	This is narrowing of the aqueduct of Sylvius, the passage between the third and fourth ventricles.
	Arnold-Chiari malformation	Includes type 1 (the adult form) and type 2 (the infantile type). There is a caudal displacement of the brainstem, cerebellum and more. There might be impaction in the foramen magnum.
	Cerebellar abnormalities (structural)	A small or malformed cerebellum. Includes vermis aplasia or hypoplasia.
	Cerebral atrophy/myelin abnormality	Any white matter abnormality of the brain or cerebral atrophy.

Body System (and feature path)	Feature	Useful comments aiding choice of feature
CRANIUM (continued)		
Brain, general abnormalities (continued)	Dandy-Walker malformation	A fourth ventricle malformation characterised by atresia of the foramina of Luschka and Magendi and a cystic malformation of the midline cerebellar structures.
	Hamartoma of brain	Any non-malignant nodule. Mostly seen in tuberous sclerosis.
	Holoprosencephaly/arhinencephaly	An undivided frontal lobe. Might be alobar (a single ventricle, small cerebellum and no lobes), semilobar (the cortex is partially divided in the midline) or even milder, i.e. lobar. Agenesis of the corpus callosum is a separate feature. Arhinencephaly is the absence of the olfactory tracts.
	Hydranencephaly/porencephaly/arachnoid cyst	The almost complete replacement of the hemispheres by a filled sac. Includes porencephaly which is a more localised, fluid filled sac. Can be multiple.
	Hydrocephaly/large ventricles, non-specific	Any enlargement of the ventricles. See also Dandy-Walker malformation , Arnold-Chiari malformation and aqueduct stenosis .
	Intra-cranial calcification	Includes all intracranial calcification.
	Intra-cranial lipoma	These can sometimes be recognised on imaging because of their density and signal characteristics. They arise from the skull and spinal canal and are often midline. They contain adipose material.
	Lissencephaly/pachygyria/polymicrogyria	Lissencephaly means a smooth brain or agyria (an absence of fissures). The underlying defect is one of neuronal migration. Pachygyria is also a migration defect causing thickened and wide gyri. Polymicrogyria means an increased number of small gyri, usually caused by an insult to the brain at 12-20 weeks gestation.
	Neuronal migration abnormality/heterotopia	A diverse group of conditions resulting from abnormal movement of neurons from their origin to final position in the brain
	Pons/medulla/basal ganglia, abnormal	Any malformation of the brainstem excluding atrophy, which is a separate feature brainstem atrophy
Posterior encephalocele/meningocele	An encephalocele at the occiput.	

Body System (and feature path)	Feature	Useful comments aiding choice of feature
CRANIUM (continued)		
Brain, general abnormalities (continued)	Schizencephaly	A split brain, the result of a neuronal migration defect causing the cerebral mantle not to form. Mostly in the region of the Sylvian fissure and especially of importance to geneticists if bilateral.
	Septum pellucidum/falx, abnormal	Any abnormality. See also corpus callosum .
	Tumours/cysts	Any type
	Vascular malformations/haemorrhage of brain	Includes aneurysms, fistulae and congenital defects.
Cranial bones, general abnormalities	Advanced pneumatization of cranial sinuses	The air filled spaces (sinuses) are far larger and extensive than usual.
	Ossification defects of skull	Localised 'holes' in the skull.
	Parietal foramina	These are holes (lucencies) in the skull vault over the parietal area. See also ossification defects of skull .
	Sclerosis of skull	Increased density of skull bones.
	Thick calvarium	A thick cranial vault.
	Thin calvarium	A thin cranial vault.
	Under-development of cranial sinus	The air filled spaces (sinuses) are less extensive and are encroached upon by bone.
	Undermineralization of skull	Poor bone mineral density of the skull
	Wormian bones	Small areas of bone, like islands, in the cranial sutures (especially posteriorly). There are normally fewer than 11, but they are difficult to count.
Cranial sutures, general abnormalities	Cranial sutures, ridged	Ridged sutures are often seen as a feature of premature fusion. If so use with craniosynostosis. Also called 'beaking' and referred to as bony bridging or poor skull mineralization.

Body System (and feature path)	Feature	Useful comments aiding choice of feature
CRANIUM (continued)		
Cranial sutures, general abnormalities (continued)	Cranial sutures, wide	Delayed closure for many reasons, or raised intracranial pressure, will cause this.
	Craniosynostosis	Premature fusion of the cranial sutures.
	Fontanelles of skull, extra	A third fontanelle (or more).
	Fontanelles, delayed closure/large	The anterior fontanelle is usually closed by 18 months.
	Fontanelles, early closure	The anterior fontanelle measures 2.5 cm by 2.5 cm at birth and usually closes by 18 months. Early closure might be a sign of one of the craniosynostoses or if there is microcephaly, the early closure might be related to the small brain size.
	Kleeblattschadel/Clover-leaf skull	The result of early and rapid closure of all the sutures. The head has a trilobular configuration.
Cranium, general abnormalities	Acrocephaly/turricephaly	A tower-shaped pointed, steep, skull. When due to premature fusion of the coronal sutures, the skull often also shows brachycephaly .
	Brachycephaly	A short head from back to front. If there is a craniosynostosis, it is mostly bi-coronal.
	Dolichocephaly/scaphocephaly	A long head in the anterior-posterior plane (mostly due to premature fusion of the sagittal suture). In scaphocephaly the calvaria is long and narrow. Like the keel of an up-turned boat.
	Macrocephaly	A head circumference greater than 2 SD above the mean for age. Parental measurements must be taken into account.
	Microcephaly	A reduction in head circumference of greater than 2SD below the mean. Parental measurements must be taken into account.
	Plagiocephaly/asymmetrical skull	An asymmetrical skull shape. Often, but not always, caused by unilateral fusion of one coronal suture

Body System (and feature path)	Feature	Useful comments aiding choice of feature
CRANIUM (continued)		
Cranium, general abnormalities (continued)	Platybasia	An increase (more than 150 degrees) of the angle between the plane of the sphenoid and the clivus.
	Trigonocephaly	A triangular shape of the skull (especially of the forehead when viewed from above). See also metopic ridge .
Occipital region, general abnormalities	Flat occiput	Flat at the back. Seen in floppy children who lie continuously on their back. And in lambdoid craniosynostosis (bilateral), in which most of the posterior fossa is under-developed.
	Occipital projection	A prominence of the occipital bone
	Prominent occiput	A bulging (posteriorly) of the occiput.
Scalp, general abnormalities	Cutis gyrata of scalp	This is often 'cutis verticis gyrata' or vertical grooves. These are scalp folds or furrows which give the scalp a corrugated appearance.
	Scalp defects	Aplasia cutis congenita of the scalp. There is another category of patchy aplasia under SKIN which can be used with this feature.
	Scalp tumours	Not only true tumours, but also nodules, cysts, hamartomas or other elevated lesions.
Sella turcica, general abnormalities	Sella turcica, J-shaped	Seen in normals and in the mucopolysaccharidoses. It looks like a J on its back.
	Sella turcica, large	A sella is large in tumours, hydrocephalus and in some of the mucopolysaccharidoses (and NF1).
	Sella turcica, small	The sella might be absent in congenital aplasia of the pituitary and small in hypoplasia.
EARS		
Anti-helix, general abnormalities	Abnormal anti-helix	The anti-helix has two crura forming the triangular fossa between the arms. Any abnormality other than prominent anti-helix which is a separate feature.

Body System (and feature path)	Feature	Useful comments aiding choice of feature
EARS (contined)		
Anti-helix, general abnormalities (continued)	Prominent anti-helix	The anti-helix stands out and is larger than normal.
Anti-tragus, general abnormalities	Abnormal anti-tragus	Not a very helpful sign, but it refers to any abnormality in shape or size of that area opposite the tragus. The tragus is just below the meatus.
Auditory canal, general abnormalities	Absent auditory canal	Can use this in conjunction with narrow/atretic auditory canal as they might be gradations of the same underlying abnormality.
	Duplication of external canal	As described
	Narrow/atretic auditory canal	The external opening is narrow, or almost shut.
Deafness, general abnormalities	Deafness, conductive	Used synonymously with middle ear deafness.
	Deafness, congenital	Mostly refers to sensorineural deafness which is a separate feature.
	Deafness, non-specific	Deafness general abnormalities would describe all types.
	Deafness, sensorineural	The nerve itself is involved.
	Deafness, unilateral	Use with caution as sensorineural deafness or conductive deafness might be more appropriate.
Ear crus, general abnormalities	Horizontal ear crus	The crus normally slopes down from medial to lateral.
	Prominent ear crus	Occasionally the crus stands out and extends further laterally than normal.
	Small ear crus	Not a useful feature. The crus usually fades out about halfway across the ear.
Ear helix, general abnormalities	Crumpled ear helix	As if someone had crunched it between their thumb and fingers and the folds had stayed in place.
	Notched ear helix	Notches (V-shaped indentations) or slits (focal deficiencies).
	Over-folded ear helix, lop ear	An exaggeration of the normal fold. A lop ear is a limp, droopy often anteverted ear. A lop ear is sometimes cup shaped.

Body System (and feature path)	Feature	Useful comments aiding choice of feature
EARS (contined)		
Ear helix, general abnormalities (continued)	Pits of ear helix	Deep or shallow indentations, like bite marks. Look both front and back.
	Prominent ear helix	Not a good sign. Prominent means it projects out. See also prominent ears .
	Thin ear helix	A deficiency in the width of the helix.
Ear lobule, general abnormalities	Absent or hypoplastic ear lobule	Not present or notably small. There is quite a lot of variation in normals.
	Attached ear lobule	Where the lobule is attached posteriorly to the skin behind the ear.
	Crease of ear lobule	Single or tramline creases overlying the lobule.
	Large ear lobule	An ear lobe which looks disproportionately large considering the size of the ear.
	Uplift of ear lobule	The lobule is tilted upwards rather than hanging down.
External ears, general abnormalities	Absent ears	There is a continuum between absent ears and severe microtia. Microtia is a separate feature.
	Asymmetric ears	A difference in size between the two sides. If one is significantly smaller, microtia might be a better description.
	Auricular pits/fistulas	These are pits, sinuses or fistulae in front of the ear. Pits of ear helix is a separate feature.
	Auricular tags	These are pre-auricular hillocks, or nubbins of tissue. They are vestigial remnants of the developing ear.
	Calcification of ear cartilage	A nodule might be felt, but this mostly needs X-ray confirmation.
	Cystic ear pinna	Similar to the cysts seen in professional boxers after continual trauma ("cauliflower ears"). The cysts seem to arise from the triangular fossa.
	Deficient ear cartilage	On palpation the ear is soft and floppy.

Body System (and feature path)	Feature	Useful comments aiding choice of feature
EARS (contined)		
External ears, general abnormalities (continued)	Dysplastic ears	A malformed ear. A small ear (microtia) might be dysplastic in which case can use both features together
	Large ears	These are measured from top to bottom. Strictly speaking they should be above the 97th centile on the chart. Note that ears get larger with age.
	Low-set ears	The topmost (superior) insertion of the ear is below an imaginary line drawn horizontally between the canthi of the eyes. The head must be in an upright position. A much over-diagnosed sign.
	Posteriorly rotated ears	Viewed with the head in an upright position, the ears are rotated backwards (posteriorly).
	Preauricular dermal aplasia	Localised skin dysplasia in front of ear
	Prominent ears (anteverted)	Most ears that appear prominent are large and rotated forwards (anteverted).
	Rigid ears	On palpation they seem more cartilaginous than usual. There is a separate category for calcification of ear if this is suspected
	Simple ears	Ears without the normal "geography" especially in the triangular fossa between the helix and the crus of the anti-helix. The ear is often 'cup' shaped (smooth and uninteresting).
	Small ears/microtia	Small normal looking ears, but used by some for small dysplastic ears .
	Telangiectasia of ears	Prominent spider-web vessels often on the helix but also in the triangular fossa.
	Thickened ears	Thickened, swollen, coarse looking ears.
Inner ear, general abnormalities	Cochlear/saccular abnormalities	Any abnormality seen mostly on imaging, or on electrophysiology.
	Temporal bone aplasia/dysplasia	A radiological feature.
	Vestibular apparatus abnormalities	Abnormal on testing. Can cause vertigo. Nystagmus (a separate feature) may also be present.

Body System (and feature path)	Feature	Useful comments aiding choice of feature
EARS (contined)		
Mastoids, general abnormalities	Advanced pneumatization of mastoids	An X-ray observation of the air filled spaces which in childhood seem to be more prominent and extensive than usual.
	Under-development of mastoids	Poorly developed air spaces in the temporal bone behind the ear.
Middle ear, general abnormalities	Abnormal auditory ossicles	Fusion or other abnormality (except absence).
	Absent auditory ossicles	An X-ray finding of the absence of the bones of the inner ear.
Tragus, general abnormalities	Tragus, hypoplastic or absent	Degree of prominence of the tragus appears very small, underdeveloped or absent. Note this finding is highly variable.
ENDOCRINE		
Adrenals, general abnormalities	Adrenal calcification	Seen radiologically.
	Adrenal hyperplasia	Anatomically, there is considerable nodular hyperplasia, the cortical cells being filled with cholesterol and other lipids. The hyperplasia is the result of chronic ACTH stimulation. The adrenal androgen causes the virilization in females. Use, when appropriate with other features under female genitalia .
	Adrenal hypoplasia/insufficiency	Primary adrenocortical failure. Hypoadrenalism. It may be complete or selective.
	Adrenal tumours (excluding neuroblastoma)	Adenomas, carcinomas.
	Neuroblastoma	One of the tumours of neural crest origin.
Endocrine, general abnormalities	Multiple adenomatosis	Also called 'multiple endocrine adenomatosis' (MEA). There are 3 types, MEA1, MEA2a, MEA2b. The thyroid, parathyroid, pancreas, pituitary and adrenals are involved, depending on the type.
	Multiple hypo-endocrinism	A general category for multiple deficiencies. Might be better to describe with separate deficiencies of endocrine organs.

Body System (and feature path)	Feature	Useful comments aiding choice of feature
ENDOCRINE (continued)		
Pancreas (endocrine), general abnormalities	Diabetes mellitus/hyperglycaemia	Note that there is a separate feature for glycosuria .
	Hypoglycaemia	Less than 2.2mmol/l in the older child, and less than 2.8mmol/l in the well neonate.
Parathyroids/calcium, general abnormalities	Hypercalcaemia	Raised serum calcium. There is another feature hypercalciuria under urinary system.
	Hypocalcaemia	Low serum calcium. Normal after the neonatal period is 2.1mmol/l to 2.85mmol/l.
	Hypophosphataemia	Phosphate depletion for what-ever reason.
	Parathyroid tumours/hyperplasia	Diffuse, asymmetrical or nodular. Might be part of multiple endocrine adenomatosis .
	Parathyroid, absent/hypoparathyroidism	Hypoparathyroidism might be central (deficient PTH secretion) or end-organ. Glands might be ectopic rather than absent.
Pituitary, general abnormalities	Absent pituitary	Might use this with hypo-pituitarism
	Ectopic pituitary	Pituitary tissue located elsewhere from normal pituitary site
	Hyper-pituitarism	Mostly the result of adenomas. Consider, when appropriate, to use with pituitary tumours . Clinically associated with an acromegalic type of appearance.
	Hypo-pituitarism	The gland may be aplastic or hypoplastic or be secondarily destroyed by tumour or radiation.
	Isolated growth hormone deficiency	Mostly a relative deficiency in response to provocation. Presents commonly with mild short stature and slow rate of growth.
	Pituitary - diabetes insipidus	Manifests with polyuria and polydipsia and varying degrees of plasma hypertonicity. Mostly associated with vasopressin deficiency, or poor action.

Body System (and feature path)	Feature	Useful comments aiding choice of feature
ENDOCRINE (continued)		
Pituitary, general abnormalities (continued)	Pituitary calcification	Might be indicative of a craniopharyngioma or other tumours. Use with pituitary tumours , when indicated.
	Pituitary duplication	A rare malformation occurring most commonly with complex midline abnormalities and skull base anomalies.
	Pituitary tumours	Of all types. Include suprasellar region tumours. Gliomas, craniopharyngiomas, adenomas, germ-cell tumours and other developmental masses. See also brain tumours/cysts .
Thymus, general abnormalities	Absent/hypoplastic thymus	Can be ectopic, rather than absent, and it can shrink in any stressful situation. An important, but on occasions difficult feature, as it might appear absent on a plain radiograph, but will be shown to be present on CT and MRI. Hypoplasia is even more difficult in view of the reaction to stress.
	Enlarged thymus	It can enlarge as a rebound phenomenon after stress and in cysts and tumours (thymomas, lymphomas, etc.).
Thyroid, general abnormalities	Ectopic/enlarged thyroid	Includes a lingual thyroid (at the root of the tongue), sublingual and elsewhere. Use together with goitre for enlarged thyroid.
	Hyperthyroidism	This includes hyperthyroidism due to an overactive thyroid. If pituitary in origin use hyper-pituitarism .
	Hypothyroidism/small/absent thyroid	Mostly detected on neonatal screening.
	Thyroglossal cyst	A sinus or cyst. The thyroglossal duct extends from the base of the tongue to the gland. A cyst is situated anteriorly in the midline of the neck. A rupture of the cyst leads to a sinus in the same area.
	Thyroid tumours	Benign and malignant (medullary carcinoma, follicular adenoma and carcinoma, papillary carcinoma etc). See also parathyroid tumours .

Body System (and feature path)	Feature	Useful comments aiding choice of feature
EYES, ASSOCIATED STRUCTURES		
Eyebrows, general abnormalities	Absent eyebrows	This might be variable. Use when necessary with sparse/decreased eyebrows.
	Arched eyebrows	In the form of a half-moon.
	Double eyebrows	A second row of eyebrows (often imperfectly formed), one above the other.
	Eyebrows extending to upper eyelid	A streak of hair extends from the lateral border of the eyebrows to reach the upper lid.
	Lateral eyebrow flare	lateral aspect of the eyebrow spreads outwards
	Lateral hypoplasia of eyebrows	A lateral deficiency (note that eyebrow growth can be very variable).
	Medial eyebrow flare	An upward, fan shaped sweep of the medial part of the eyebrows.
	Sparse/decreased eyebrows	Difficult to quantify. Sometimes seen in conjunction with sparse scalp hair.
	Synophrys	Eyebrows that meet in the middle.. Not always pathological.
	Thick eyebrows	Mostly thick and bushy. Not always pathological.
Eyelashes, general abnormalities	Absent or sparse eyelashes	As seen in the ectodermal dysplasias. When they curve in and irritate, they may be absent because they are pulled out.
	Curly/curved eyelashes	They mostly curve in and irritate. Must be significantly outside the norm for the family.
	Double eyelashes	Also called distichiasis. This is a second row of lashes, coming out of the meibomian gland orifices. They might irritate and be pulled out
	Long/prominent eyelashes	Trichomegaly. Unusually long lashes which turn up.
Eyelids, general abnormalities	Absent eyelids	Ablepharon. Not always totally absent.
	Chronic blepharitis	Chronic inflammation of the eyelids.

Body System (and feature path)	Feature	Useful comments aiding choice of feature
EYES, ASSOCIATED STRUCTURES (continued)		
Eyelids, general abnormalities (continued)	Coloboma of eyelids	Slits (clefts) of either upper or lower lid. There might be multiple clefts in the same lid. Most common in the nasal half of the upper lid, or lateral part of the lower lid.
	Cryptophthalmos/absent palpebral fissures	A rare anomaly where the skin is continuous over the eyeball with absence of eyelids
	Ectropion of eyelids	An outward rotation of the rim/margin of the lid (either upper or lower lid).
	Entropion of eyelids	An inward turning of the lid margin.
	Flame nevus of eyelids	Nevus flammeus. Like a stork mark at the nape of the neck.
	Nodular eyelids	These could be granulomas, or small tumours, warts or hamartomas.
	Ptosis of eyelids	Congenital or acquired. Use cranial nerve palsies for III nerve palsy.
	Synechia/fused eyelids/ankyloblepharon	In ankyloblepharon the eyelid margins are partially or completely fused. Synechia are strands of tissue which, in this case, join the lids together.
	Telangiectasia of eyelids	Spidery network of dilated vessels.
Nasolacrimal duct, general abnormalities	Blocked/absent nasolacrimal duct	There is a failure of the lacrimal system to connect to the nasal mucosa. The duct might be blocked, or is absent.
	Nasolacrimal duct, abnormal position	These are often fistulae (a false passage). Their main position is just below the medial canthus.
Palpebral fissures, general abnormalities	Almond shaped palpebral fissures	In the shape of an almond nut. Said to be typical of Prader-Willi syndrome, but common in any type of obesity and it is very non-specific.
	Blepharophimosis/blepharospasm	Small eye openings. The aperture is reduced along all planes. In blepharospasm there is forceful, bilateral eye closure.
	Palpebral fissures slant down	An 'anti-mongoloid' slant to the palpebral fissure, which is normally horizontal.

Body System (and feature path)	Feature	Useful comments aiding choice of feature
EYES, ASSOCIATED STRUCTURES (continued)		
Palpebral fissures, general abnormalities (continued)	Palpebral fissures slant up	A 'mongoloid slant' to the palpebral fissure.
	Short palpebral fissures	The fissure is short in the horizontal plane i.e. from side to side.
	Wide/long palpebral fissures	Wide from side to side. The fissure appears to disappear around the corner of the face.
Periorbital skin, general abnormalities	Epicanthic folds	These are folds which extend from the upper eyelid towards the medial canthi. They are common in neonates (epicanthus palpebralis) and Asians (epicanthus tarsalis). This includes epicanthus inversus which is an epicanthus extending from the medial canthi towards the upper eyelid.
	Fullness of peri-orbital region	Swelling or redundant tissue.
	Infra-orbital creases	Often seen in normals. The groove runs medially to laterally in a downward sweep.
	Peri-orbital tumours/cysts	Any cyst or tumour around the orbit. Might occur together with orbital cyst
	Pigmented peri-orbital skin	Dark pigmentation, below or encircling the eye.
	Sagging peri-orbital skin/blepharochalasis	Blepharochalasis must not be confused with ptosis. There is excessive wrinkling and atrophy of subcutaneous tissue giving rise to sagging of the lid.
Tears, general abnormalities	Absent or sparse tears	Includes abnormalities of lacrimal gland (alacrima) and disturbances of autonomic nervous system.
	Increased tear production	Mostly an overflow phenomenon - as in facial nerve palsies and obstruction of nasolacrimal duct. Use with caution.

Body System (and feature path)	Feature	Useful comments aiding choice of feature
EYES, GLOBES		
Anterior chamber, general abnormalities	Anterior chamber abnormalities, unspecified	This mostly concerns abnormalities of development of the anterior segment causing it to be shallow. Glaucoma , iris and cornea abnormalities are separate features
	Peters' anomaly	A congenital central corneal opacity
	Rieger anomaly	Hypoplasia of the anterior iris stroma, accompanied by strands which run from the iris to Schwalbe's line (the posterior limit of Descemet membrane) and posterior embryotoxin (abnormal thickening of Schwalbe's line).
Conjunctiva, general abnormalities	Conjunctival nodules	These nodules could be papillomas, granulomas or any raised, circumscribed lesion.
	Conjunctival telangiectasia	Telangiectatic, tortuous vessels especially temporally.
	Conjunctivitis	Inflammation of the conjunctiva (a discharging, red, swollen eye). Used to describe both allergic and infected eyes.
	Symblepharon	The adhesion of one or both eyelids to the eyeball via the cornea
Cornea, general abnormalities	Asymmetric corneae	One usually smaller than the other. Often indicative of a small eye (use microphthalmia when appropriate).
	Cloudy corneae/sclerocornea	A white, hazy, cloudy, unclear cornea
	Epibulbar dermoid	These are a combination of fat, hair follicles, and sebaceous gland, occurring at the limbus.
	Keratoconus	A central thinning of the cornea, causing an irregular anterior bulging (protrusion).
	Macrocornea/megalocornea	In adulthood the diameter should be 11.7mm and this is attained by 2 years of age. Macrocornea is a diameter of > 13mm.
	Microcornea	A corneal diameter of > 2SD below the mean.

Body System (and feature path)	Feature	Useful comments aiding choice of feature
EYES, GLOBES (continued)		
Cornea, general abnormalities (continued)	Ulceration of cornea	Or corneal erosions. Sometimes the result of corneal anaesthesia, infection, exposure, or weak tissue.
	Visible nerve fibres on cornea	These may be visible in the periphery of normal people, but may be abnormally prominent.
Eyes, general abnormalities (including spacing)	Asymmetric eyes	Mostly asymmetric in position rather than in structure. Often seen in the context of asymmetric face .
	Cyclopia	A central, single eye, situated where the root of the nose normally occurs. Indicates holoprosencephaly (undivided frontal lobes).
	Deep-set eyes	A subjective observation of eyes that seem sunken in the skull.
	Dystopia canthorum (telecanthus)	Lateral displacement of the inner canthi, giving a false impression of hypertelorism.
	Hypertelorism	Wide-set eyes. Measured as an interpupillary distance greater than 2SD of the mean.
	Hypotelorism	Eyes too closely set. Measured as an interpupillary distance less than 2SD of the mean.
	Prominent eyes/proptosis	Bulging, prominent, exophthalmic eyes. Eyes which appear too big for the socket.
Globes, general abnormalities	Anophthalmia	An absent eye although there is often a tiny cystic remnant. There might be a spectrum between anophthalmia and microphthalmia .
	Buphthalmos	A large eye resulting from congenital glaucoma.
	Glaucoma	Raised intraocular pressure. When congenital use in conjunction with buphthalmos .
	Microphthalmia	Defined as an ultrasonic measurement of axial length of less than 19mm in a child of 1 year, and less than 21mm in an adult (minus 2SD). Dysmorphologists usually diagnose this by inspection.

Body System (and feature path)	Feature	Useful comments aiding choice of feature
EYES, GLOBES (continued)		
Globes, general abnormalities (continued)	Orbital cyst	This includes dermoid cysts, sinus mucocoeles, congenital eyeball cysts.
	Spontaneous rupture of globe	Usually the end result of a severe collagen defect.
Iris, general abnormalities	Aniridia	An absent or very hypoplastic iris.
	Brushfield spots	These are white iris spots found in normal individuals as well as in Down syndrome.
	Coloboma of iris	A failure of closure of the fetal fissure leaving a slit. 'Typical' refers to those in the fetal fissure (inferior medial, keyhole type). Those in other places are 'atypical'.
	Depigmentation of iris	Use in conjunction with pigmentary abnormalities of iris or even Iris atrophy .
	Heterochromia of iris	A difference in iris colour on the two sides. The abnormality might be a darker or lighter pigmentation in one eye.
	Iris atrophy/dysplasia	Stromal atrophy or poorly formed stroma.
	Pigmentary abnormalities of iris	This excludes heterochromia of iris . Mostly, focal areas of pigmentary change, unilateral or bilateral.
Lens, general abnormalities	Cataract	Lens opacities of all types
	Dislocation of lens	Both up and down
	Size or shape of lens abnormal	Includes: Absent lens - aphakia Small lens - microspherophakia Abnormality of posterior surface - lentiglobus Abnormality of anterior surface - lenticonus
Macula, general abnormalities	Macular degeneration	This includes any of the progressive macula dystrophies.
	Pigmentary abnormality of macula	Use with caution, as it might be a manifestation of one of the macular dystrophies in which case use with macular degeneration , or part of a pigmentary retinopathy (see under retina).

Body System (and feature path)	Feature	Useful comments aiding choice of feature
EYES, GLOBES (continued)		
Optic disc and nerve, general abnormalities	Coloboma involving optic nerve	A slit in the disc/nerve.
	Hypoplasia/dysplasia of optic nerve	A small disc/nerve. Must not be confused with very early optic atrophy , which is much rarer in infancy.
	Optic atrophy	Pallor of the optic disc, with fewer than normal vessels on the surface. The pallor might initially be segmental (temporal) before becoming diffuse.
	Papilloedema/optic neuritis	Swelling of the disc secondary to raised intracranial pressure or inflammation of the nerve.
Pupil, general abnormalities	Asymmetric pupils	Includes anisocoria or unequal pupils.
	Ectopic pupils	A displacement of the pupil from its usual position.
	Persistent pupillary membrane	The membrane floats free or is attached to the anterior surface of the lens. It might represent an incomplete resorption of the tunica vasculosa of the anterior surface of the lens.
	Size or movement of pupil abnormal	Large or pinpoint (microcoria). Includes abnormal response to light. Includes Horner syndrome (the pupil part at least). Consider describing with cranial nerve palsies if IIIrd nerve palsy.
Retina, general abnormalities	Aplasia or dysplasia of retina	Maldevelopment of the retina - usually congenital and stationary.
	Coloboma of retina/choroid	A slit resulting from the failure of closure of the fetal fissure.
	Detached retina	Includes rhegmatogenous detachment (as in high myopia), tractional detachment (as in vitreoretinal dystrophies), exudative detachment (as in retinitis pigmentosa) and solid detachment (as in tumours).
	Macular red spot/cherry red spot	A round ball of red which is the fovea shining through a ring of lipid laden neurons.
	Phakoma/pseudoglioma of retina	These are hamartomas (a tumour mass rising from anomalous tissue).
	Punched-out lesions of the retina	These are clear-cut lacunar defects in the retinal pigment epithelium.

Body System (and feature path)	Feature	Useful comments aiding choice of feature
EYES, GLOBES (continued)		
Retina, general abnormalities (continued)	Retinitis pigmentosa/pig retinop/chorioretinitis	This includes 'bone spicule' type and 'salt and pepper' changes. It also includes retinitis pigmentosa sine pigment.
	Retinoblastoma	The most common malignant tumour of childhood.
	Vascular abnormalities of retina	Includes capillary haemangiomas, vessel tortuosity, Coats disease, A-V malformations.
Sclera, general abnormalities	Blue sclera	An important marker for collagen disorders, but deceptive in young babies and easy to falsely diagnose if the baby is wearing blue. Take the baby to the window to view.
	Nevus of Ota	A localised area of blue pigmentation.
Vision, general abnormalities	Blindness	It is better to describe using a feature which indicates the cause - if known.
	Duane anomaly	A congenital inability to abduct the eye, with widening of the fissure on attempted abduction. In addition there is impairment of abduction with narrowing of the palpebral fissure in the opposite eye.
	Hypermetropia	Long sighted. Might be incidental - ask family.
	Myopia	Significant short-sightedness. Take family history because it might be unrelated to the syndrome under consideration.
	Night blindness	Nyctalopia. Decreased visual acuity in the dark.
	Nystagmus	Rhythmic oscillation about one or more axes. This includes cerebellar nystagmus and that due to a visual defect.
	Photophobia	Sensitivity to normal light causing discomfort (the eyes might be closed).
	Strabismus/gaze palsy	Squint. Check cranial nerves for III, IV and VI nerve palsies.
	Vision, non-specific impairment	A better description might be Vision - general abnormalities .

Body System (and feature path)	Feature	Useful comments aiding choice of feature
EYES, GLOBES (continued)		
Vitreous, general abnormalities	Persistent hyaloid artery	Includes persistent hyperplastic primary vitreous in which there might be a retrolental plaque and prominent vessels on the iris.
	Vitreous, abnormal	Unspecified abnormality. A better description might be Vitreous - general abnormalities
FACE		
Cheeks, general abnormalities	Full cheeks	Prominent or puffy cheeks. A 'jowly' face.
	Sunken cheeks	A concavity in the profile of the cheek.
Chin, general abnormalities	Dimpled or grooved chin	A mid-line groove
	Pointed chin	Not a good sign. Use when pronounced.
Face, general abnormalities	Asymmetric face	One side of the face is smaller or larger than the other. There are other descriptions if the asymmetry is more specific eg asymmetric eyes , asymmetric ears , asymmetric mouth , asymmetric mandible .
	Broad face	Broad from side to side. Not a good feature. Note separate feature wide forehead
	Coarse facial features	A heavy look to the face because of sagging cheeks, thick lips, heavy brow.
	Expressionless/dull face	An immobile face as in Parkinson's disease or other extrapyramidal disorders. Also called a 'poker' face. Seen also in the muscular dystrophies with facial involvement.
	Facial cleft	Often extensions of clefts that arise from the upper lip (cleft upper lip) or the side of the nose (clefting of nose). Includes all clefts as classified by Tessier.
	Facial haemangiomas	Blood vessel tumours or birthmarks.
	Facial hirsutism	An increase in facial hair alone. If more extensive use generalised hirsutism

Body System (and feature path)	Feature	Useful comments aiding choice of feature
FACE (continued)		
Face, general abnormalities (continued)	Facial tumours	Any tumour.
	Facial weakness	Mostly a lower motor neuron palsy. Most of the upper motor neuron lesions will be part of a hemiplegia. This includes facial weakness due to muscle disease.
	Flat face	There are 3 features which overlap and can be used together:1) Mid-face hypoplasia 2) Flat face 3) Flat malar region . The term 'dished out' face is also used in the literature.
	Mid-face hypoplasia (excluding malar region)	There are 3 features which overlap and can be used together:1) Mid-face hypoplasia 2) Flat face 3) Flat malar region .
	Premature ageing	Mostly the appearance of aging (a wizened face) in infancy or childhood. The skin is thin, atrophic, shiny or wrinkly. The hair growth is poor, blood vessels are prominent and tortuous.
	Round face	Mostly seen in generalised obesity . A moon face.
	Small face	Small, but not triangular.
	Telangiectasia	Small, tangled, superficial vessels. There are separate features for telangiectasia of eyelids and nasal telangiectasia . All can be used together if necessary.
	Thin/long face	Thin from side to side and long from top to bottom.
	Triangular face	A broad forehead, and a small chin, with, the two sides of the triangle creating a small, delicate looking face. The head looks large (pseudo-hydrocephalus).
Malar region, general abnormalities	Flat malar region	There are 3 features which overlap and can be used together:1) Mid-face hypoplasia 2) Flat face 3) Flat malar region
	Prominent malar region	The zygomatic bones (situated in the upper and lateral parts of the face) form the prominences of the cheek. The malar surface is slightly convex (directed laterally and forward). This area might be prominent.

Body System (and feature path)	Feature	Useful comments aiding choice of feature
FACE (continued)		
Mandible, general abnormalities	Absent mandible	Agnathia. The ears seem particularly lowset. Might also use this feature to describe severe hypoplasia.
	Asymmetric mandible	Mostly due to hypoplasia of the ramus on one side.
	Bifid condyle	bifid mandibular condyle is a rare anomaly
	Cleft mandible	Mostly a midline cleft. May overlap with dimpled or grooved chin .
	Prominent mandible/prognathism	Might clinically present with prognathism. A jutting jaw.
	Small mandible/micrognathia	A subjective impression of a small jaw. Difficult in infancy and early childhood. Family background must be taken into account. The jaw often grows 'out' with age.
	Temporo-mandibular joint anomaly	Mostly a fusion or inflammation at the joint.
Maxilla, general abnormalities	Accesory maxilla	A rare anomaly
	Agenesis of the pre-maxilla	The anterior part of the bony palate (in the mid-line) is deficient.
	Asymmetrical maxilla	Use with asymmetric face , if more than just the maxilla is involved.
	Hypoplastic maxilla (excluding malar region)	Poorly developed, small maxilla. If on one side use with asymmetrical maxilla .
	Prominent maxilla	A jutting upper jaw. Might create an over-bite.
	Prominent pre-maxilla	The anterior part of the bony palate is prominent.
FEET		
Feet, general abnormalities	Abnormal dermatoglyphics on feet	Not frequently examined except in Down syndrome.
	Abnormal/deep plantar creases	There are deep furrows on the sole, often between the big and second toes or on the foot pad.

Body System (and feature path)	Feature	Useful comments aiding choice of feature
FEET (continued)		
Feet, general abnormalities (continued)	Absent feet	Also called a terminal transverse defect. Reduction deformity of legs - no digits means the same and so both features can be used together
	Asymmetric feet	This usually means that one is smaller or larger than the other.
	Club foot, valgus	This is often a pes calcaneocavo valgus, in that the arch is high. The foot turns outward from the ankle downwards and is dorsiflexed and everted.
	Club foot, varus	One or both feet are in-turned from the ankle downwards. It might be fixed and structural, or postural.
	Flat arches of feet	A foot is flat when the medial side is too close to the ground, or the heel is unduly everted. Also called pes planus.
	Foot pads	Puffy pads on the under surface of the feet. Only useful if persistent
	High arches of feet (pes cavus)	The normal arch of the foot is exaggerated, giving the impression that the medial part is too raised off the ground. Later, the toes claw.
	Large feet	Feet that seem too big (shoe size is helpful).
	Long feet	Long for age, mostly as judged by shoe size!
	Narrow feet	Often in conjunction with long feet. Narrow from side to side.
	Oedema of feet	Pitting and un-pitting, swollen feet. Mostly due to fluid retention. Oedema of hands and oedema of lower limbs are separate features.
	Prominent heels	The posterior protrusion of the heel is prominent. Possibly due to a vertical talus . See also Rocker-bottom feet .
	Rocker-bottom feet (see also vertical talus)	As in the curved rails on which a rocking chair swings. If a vertical talus is also present, use that or both together.
	Small feet	Small for age (and shoe size).
Split/cleft foot/ectrodactyly	Also called a 'lobster claw' deformity.	
Wide feet	Wide from side to side. A flat foot can look wide, so use with caution.	

Body System (and feature path)	Feature	Useful comments aiding choice of feature
FEET (continued)		
Hallux, general abnormalities	Absent or hypoplastic hallux	Absent or small big toe. Might be part of tibial aplasia so check.
	Broad hallux	A broad big toe (which sometimes deviates outwards).
	Dorsi-flexed hallux	Also called a hammer toe . Caused by overflexion at the interphalangeal joint and over-extension at the metatarsophalangeal joint.
	Hallux valgus	Lateral deviation of the big toe.
	Hallux varus	Medial deviation of the big toe.
	Long hallux	Beware that this is not an impression caused by shortness of the other toes.
	Polydactyly/bifid hallux	Use together with preaxial polydactyly of toes .
	Short hallux	Can be confused with hypoplastic hallux .
Metatarsals, general abnormalities	Abnormal/absent metatarsals	Missing metatarsals. Also fusion or accessory epiphyses.
	Metatarsus valgus	The foot anterior to the navicular is abducted.
	Metatarsus varus	Also called "hook foot" and "skew foot". The foot anterior to the navicular is adducted and sometimes supinated.
Tarsals, general abnormalities	Abnormal/absent tarsals	Missing or abnormally shaped tarsals.
	Bipartite calcaneus	Two ossification centres to the calcaneum
	Osteolysis of tarsals	Resorption of tarsal bone - use osteolysis of carpals as well if appropriate.
	Tarsal synostosis	Coalition or fusion with each other or with talus. A failure of segmentation.
	Vertical talus (see also rocker-bottom feet)	With the foot placed at 90 degrees to the leg, the calcaneum is in equinus and the talus appears to continue in the vertical line of the tibia. The talus is in the shape of an hourglass. In the front, at the 'waist', articulating with the navicula, and with the beak of the calcaneum behind. The talonavicular dislocation is an essential part.

Body System (and feature path)	Feature	Useful comments aiding choice of feature
FEET (continued)		
Toes, general abnormalities	Absent toes	Might be in the spectrum of hypoplastic toes , so use both in the same box.
	Broad toes	Mostly broad, spatulate ends.
	Camptodactyly/hammer toes	An inability to extend the toes. Flexion should be normal. If not, these are joint contractures . A hammer toe, usually the 2nd, is longer than the others and becomes flexed.
	Hyper-mobile toes	Part of joint laxity in most cases. Seldom an isolated feature. Use joint laxity in preference.
	Hypoplastic toes (including phalanges)	Use with absent toes if necessary. Might be a spectrum.
	Long toes	The toe equivalent of arachnodactyly of the fingers.
	Meso-axial polydactyly of toes	The extra digit seems to be in the middle of the foot.
	Mirror image polydactyly of toes	Symmetrical seemingly duplicated toes.
	Over-riding toes (including clinodactyly)	Bent, overlapping, curly toes.
	Post-axial polydactyly of toes	An extra toe or toes on the 5th digit side of the foot. It might just be a nubbin of tissue.
	Pre-axial polydactyly of toes	An extra toe on the big toe side. Use in the same box with polydactyly of the hallux .
	Sandal gap of toes	An increased space between the first and second toes.
	Short toes	Might be difficult to differentiate from hypoplastic toes . If this is the case, use both together
	Syndactyly 2-3 of toes	Webbing of toes 2 and 3. Common in families, so use only if it isn't present in either parent.
Syndactyly of toes (not 2-3)	Webbing (joining together by skin) (and bone) of toes other than just 2-3.	
Wide-spaced toes	Increased spaces between all of the toes.	

Body System (and feature path)	Feature	Useful comments aiding choice of feature
FOREHEAD		
Forehead, prominent etc., general abnormalities	High frontal hairline	Often seen in conjunction with frontal bossing . If so use both features together The origin (roots) of the frontal hair seems posteriorly displaced.
	Hyperplastic supra-orbital ridges	The bony ridges above the eyebrows are prominent.
	Prominent forehead/frontal bossing	The term bossing means a protuberance, or round knob. A bossed forehead mostly has two prominences, divided by a shallow groove. For this feature any prominence will do (if not familial).
	Prominent glabella	The glabella is the most prominent point in the plain between the eyebrows (the word means 'smooth part').
	Ridged forehead	Vertical or horizontal grooves.
	Wide forehead	Broad from side to side.
	Glabella bone defect	An indentation (deficiency) in the area between the eyebrows.
	Hypoplastic supra-orbital ridges	The forehead is featureless, without the normal slight prominence above the eyes. Feel to make sure.
	Low frontal hairline	The frontal hairline extends on to and down, the forehead.
	Metopic ridge	Premature closing of the metopic suture results in a triangular head shape. The ridge is the point of the triangle pointing to the front. See also trigonocephaly .
	Narrow forehead/temporal narrowing	An accentuation of the normal bi-temporal hollowing.
	Short forehead	Short from top to bottom. Might be seen in a sloping forehead (use that feature in preference if appropriate). A low frontal hairline might also give this appearance.
Sloping forehead	Sloping backwards from bottom to top. Often seen in cases of microcephaly . If that is the case use both features together	

Body System (and feature path)	Feature	Useful comments aiding choice of feature
GENITALIA		
Female genitalia, general abnormalities	Abnormal labia	Labia which are too big, too small or absent.
	Absent ovaries	Mostly detected at operation, but they might be demonstrated on imaging.
	Absent uterus	Just what it says.
	Early puberty in females	This is defined by an onset of puberty before 8 years. It is also called sexual precocity. In true precocity all the events of normal puberty occur early, whereas in pseudo-precocity only some are early.
	Fused labia	Mostly a secondary phenomenon
	Late puberty in females	This is diagnosed if breast stage 2 (breast bud palpable) has not started by 13-14 years
	Malformed uterus	Any malformation.
	Ovarian cysts/tumours	Cysts, single or multiple and any type of tumour.
	Primary amenorrhea	The situation after puberty (often delayed), in which menstruation has never occurred.
	Prominent clitoris	Clitoral size is enlarged (note ambiguous genitalia is separate).
	Recto-vaginal fistula	The fistula between rectum and vagina is often associated with anorectal anomalies higher up.
	Secondary amenorrhea	There are often irregular menstrual periods before cessation.
	Short vagina	Make sure this is not the result of a vaginal septum (see separate feature).
	Streak ovaries	A term used to describe streaks of connective tissue which have replaced the ovaries. There are no primordial follicles, only fibrous stroma in whorls.
Uterine tumour/fibroid	Any tumour.	
Vaginal atresia	Includes the absence of the vaginal opening.	

Body System (and feature path)	Feature	Useful comments aiding choice of feature
GENITALIA (continued)		
Female genitalia, general abnormalities (continued)	Vaginal septum/duplication/hydrometrocolpos	There may be longitudinal midline septa, which if complete, cause a double vagina (sometimes with double cervix and uterus). The commonest form is the transverse septum (may be confused with vaginal atresia). Menstrual blood and fluid will accumulate (hydrometrocolpos)
Genitalia, general abnormalities	46, XX with Wolffian structures	i.e. remnants of a vas deferens.
	46, XY with Mullerian structures	i.e. remnants of a uterus or fallopian tubes.
	Abnormal genital pigmentation	Mostly an increase in pigmentation which could be spotty or diffuse. In males the glans penis must be inspected.
	Ambiguous/absent genitalia	A disorder of sexual differentiation. Nearly always there is a phallus-like structure, larger than a clitoris but smaller than a penis, with a urethral opening either at the base of the phallus or in the perineum. It must not be confused with severe hypospadias and cryptorchid testes .
	Female pseudo hermaphroditism	Excessive masculinization in the presence of ovaries.
	Hypogonadism	Abnormally decreased gonadal function giving rise to delayed sexual development and growth. It might be primary (at gonadal level) or secondary, (at pituitary level).
	Male pseudo hermaphroditism	Insufficient masculinization in the presence of a testis.
	True hermaphroditism	Both ovarian and testicular tissue are present. Any pattern of sex chromosomes might be present and the external genitalia can be predominantly male, or female, or ambiguous.
Male genitalia, general abnormalities	Absent testis	Diagnose with caution and if uncertain use with cryptorchid testes . If noted at operation or post-mortem use alone.
	Chordae of penis	Downward bowing of the penis.
	Cryptorchid testes	Testes that have not descended into the scrotum (an empty scrotum).

Body System (and feature path)	Feature	Useful comments aiding choice of feature
GENITALIA (continued)		
Male genitalia, general abnormalities (continued)	Early puberty in male	Onset of puberty before 9 years.
	Epispadias	This is the mildest end of the spectrum of bladder exstrophy . The meatal opening lies on the ventral surface and might be penile, balanic and penopubic. The penis is often short and has a chordee (it curves upwards).
	Hydrocele/lymphedema of testis	Fluid filled scrotal sac.
	Hyperplasia of Leydig cells	Hypertrophy or overdevelopment of the interstitial cells in the testes.
	Hypoplasia of Leydig cells	Underdevelopment of the testosterone-producing interstitial cells.
	Hypoplastic scrotum	Underdevelopment of the scrotal sac, mostly secondary to the absence of a testis. Use with absent testis or cryptorchid testis .
	Hypospadias	The urethral opening is on the underside of the penis.
	Large penis	There are measurements, but most dysmorphologists will assess visually and listen to the parents.
	Large testes	Testicular size can be measured eg using spoons and wooden balls for comparison or a ruler
	Late puberty in male	Diagnosed if genital stage 2 (penis begins to lengthen, scrotal skin thickens) has not been reached by 15-16 years.
	Oligospermia/azoospermia	Deficiency in the number of spermatogonia in the semen.
Overriding scrotum	Often referred to as a shawl scrotum. Many boys (especially the obese) seem to have a rounded fold, of no particular significance. A gothic arch is much more significant.	

Body System (and feature path)	Feature	Useful comments aiding choice of feature
GENITALIA (continued)		
Male genitalia, general abnormalities (continued)	Penis bifid	A rare congenital anomaly where the penis is partly or completely duplicated. Occurs when 2 genital tubercles develop. May be symmetrical or placed one above the other. Often associated with other urogenital anomalies or baldder exstrophy.
	Scrotum, bifid	An exaggeration of the cutaneous median raphe to form a distinct groove, thereby giving the impression of a division into two sacs.
	Small penis (including micro)	Be sure to differentiate from a concealed penis (concealed in fat). Less than 2cm at birth and less than 4cm before normal puberty.
	Small testes	A subjective measurement by palpating.
	Supernumerary testes	Also called polyorchidism. The extra testes may be in the sac or in the groin (may look like a hernia)
	Testicular tumours	Any mass in the scrotal sac.
HAEMATOL/IMMUNOLOGY		
Haematology/Immunology, general abnormalities	Anaemia/red cell abnormalities	All abnormalities of red blood cells.
	B-cell deficiency	These are marrow derived cells that produce immunoglobulins and antibodies. They may leave lymph nodes and enter the blood stream following an antigenic challenge.
	Bleeding diatheses	Bleeding not due to platelet abnormalities which is a separate feature.
	Chromosome instability/breakage	This is the only chromosomal feature on the database. It includes breakage when challenged with suitable mitogens or increased spontaneous breakage,
	Immunoglobulin abnormality	IgA, IgG etc., including any sub-class deficiency.

Body System (and feature path)	Feature	Useful comments aiding choice of feature
HAEMATOL/IMMUNOLOGY (continued)		
Haematology/Immunology, general abnormalities (continued)	Lymphadenopathy	Enlarged lymph glands from any cause.
	Lymphomas/leukaemias	Includes all kinds of leukaemias and malignant tumours of lymphatic tissue.
	Platelet abnormalities	This includes thrombocytopenia, thrombocytosis, giant platelets etc.
	Polymorph abnormalities	Any abnormality of any type of polymorphonucleocyte. See also B-cell deficiency and T-cell deficiency .
	Recurrent infections	In this context, often due to immune changes.
	SCID	Severe Combined Immuno-Deficiency.
	Storage cells/vacuolated lymphocytes	These include foamy, storage cells in the bone marrow, and vacuolated lymphocytes in peripheral blood.
	T-cell deficiency	Thymus derived cells. Responsible for delayed allergic reactions, graft rejections, a defense against many viruses, fungi and some intracellular pyogenic bacterial pathogens.
HAIR		
Hair growth pattern, general abnormalities	Abnormal secondary sexual hair	For instance, a male pattern of pubic hair, or beard growth in a female. For precocious puberty see early puberty in females . Use together when appropriate.
	Frontal upsweep/cowlick	Literally as if a cow had licked the frontal hair and it stayed in an upward sweep from the forehead.
	Generalized hirsutism/hypertrichosis	Generalized hairiness in those areas that are normally hairy, such as the scalp, eyebrows, axilla etc. Hypertrichosis (included here) strictly speaking refers to overgrowth of hair in areas not normally hairy.
	Localized hirsutism	Patchy area of hairiness. If there is an underlying pigmented, raised skin lesion use with skin pigment abnormalities when necessary.

Body System (and feature path)	Feature	Useful comments aiding choice of feature
HAIR (continued)		
Hair growth pattern, general abnormalities (continued)	Unusual hair whorl/pattern	A double whorl, or an eccentrically placed whorl. Use with caution as normal variation is great.
	Widow's peak	An accentuation of the normal V-shaped insertion of the frontal hair. The V extends onto the forehead.
Hair pigmentation, general abnormalities	Generalized depigmentation of hair	As in albinism, the hair is white and never develops its normal colour. Also includes gradual, non-congenital loss of colour. See also premature greying .
	Patchy depigmentation of hair/white forelock	The type of patchy depigmentation as seen in Waardenburg syndrome or chromosomal mosaicism. Probably involves depigmentation of the underlying skin. Includes poliosis (patches of white hair).
	Premature greying of hair	This might occur in families as an isolated finding or be a part of a syndrome such as Waardenburg syndrome.
Hair texture, general abnormalities	Brittle hair	Hair that breaks easily.
	Coarse hair	The hair feels on palpation unusually hard or rough.
	Fine hair	Fine or thin to the touch. Seen sometimes in conjunction with sparse hair or depigmentation of hair .
	Kinky/curly hair (including pili torti)	Pili torti refers to the twisting of hair through 180 degrees on its own axis. This gives a spangled or beaded appearance. This feature also includes abnormally (for the family) curly hair as in Noonan syndrome, although histologically the hair might be normal.
	Trichorrhexis nodosa	A localised loss of cuticular cells, resulting in damage to the cortical fibres. This is seen microscopically as nodular swellings of the shaft.
Hair, sparse etc., general abnormalities	Alopecia totalis	Total absence of scalp hair (universalis refers to total lack of body hair).
	Frontal balding	As in male pattern balding. Begins temporally and extends medially.
	Slow growth of hair	This refers to normal distribution of hair which grows slowly. A parent might say "I have never cut my child's hair".

Body System (and feature path)	Feature	Useful comments aiding choice of feature
HAIR (continued)		
Hair, sparse etc., general abnormalities (continued)	Sparse hair/alopecia areata	Either generally sparse, or patches of sparse hair.
HANDS		
Carpals, general abnormalities	Dislocated carpals	An unusual place to dislocate.
	Extra carpals	Accessory carpal bones, sometimes overlying other bones.
	Hypoplastic or absent carpals	Small, or absent (even dysplastic) carpal bones.
	Large carpals	For instance the capitate and hamate might be unduly large.
	Osteolysis of carpals	The carpal bones become eroded and might disappear. "Vanishing" bone.
	Synostosis	Fusion of the carpal bones.
Dermatoglyphics, general abnormalities	Abnormal dermatoglyphic patterns	Most are non-specific, hence why they are lumped together under a single feature.
	Abnormal palmar creases	Any abnormality of the larger creases of the palm. Single palmar crease is a separate feature
	Absent palmar creases	A smooth, featureless hand.
	Deep palmar creases	An increase in the depth of the normal hand creases. Seen in chromosomal mosaicism or as the consequence of clenched hands or oedema in utero.
	Single palmar crease	Note that this might be present unilaterally in 4% of the normal population and in a smaller percentage bilaterally.
Fingers, general abnormalities	Absent finger tips	A terminal distal deficiency. They seem chopped off. Might need to use with constriction rings .
	Absent fingers or oligodactyly	We have made no distinction as to what side (i.e., radial or ulnar) is involved. There might be an underlying bony defect in the forearm bones i.e., the little finger might be absent in ulnar hypoplasia/aplasia.

Body System (and feature path)	Feature	Useful comments aiding choice of feature
HANDS (continued)		
Fingers, general abnormalities (continued)	Arachnodactyly	Long, spindly, thin fingers
	Brachydactyly	Short fingers
	Camptodactyly	An inability to straighten the fingers (they can flex - if they can't, use joint contractures) If in doubt use both. See also clenched hands .
	Clinodactyly	An incurving - often seen in little finger. It may, or may not, be important.
	Clubbing of fingers (drumstick)	Note elsewhere for Wide finger tips . When in doubt use both together in the same box.
	Constriction rings of digits	Bands of tissue seem to have been tied around the fingers leaving indentations. Use when necessary in conjunction with absent finger tips , if this has additionally happened.
	Delta phalanges	A rare deformity. The bone is triangular shaped with a continuous epiphysis running from proximal to distal end of the short side.
	Fetal finger pads	These are puffy pads on the ventral surface of the finger tips. Normal in neonates and only useful as a sign if they persist (they are not always abnormal).
	Hyper-mobile/extensible fingers	They give the feeling that you can move them (even temporarily dislocate them) in all directions.
	Limited movement of fingers	Decreased mobility in the absence of joint contractures. Note separate features of clenched hands and camptodactyly and try more than one if you are unsure.
	Macroductyly	Often a large (huge) single finger, but all or some might be involved. Always compare with other hand.
Meso-axial polydactyly of fingers	The extra finger seems to be in the middle of the hand.	

Body System (and feature path)	Feature	Useful comments aiding choice of feature
HANDS (continued)		
Fingers, general abnormalities (continued)	Mirror image polydactyly of fingers	Symmetrical, seemingly duplicated, hand. If unsure use with post-axial polydactyly of fingers
	Osseous syndactyly of fingers	There is not only skin fusion between the fingers, but they are also joined by bone (need an x-ray to confirm this)
	Overlapping fingers (trisomy 18-like)	In trisomy 18, the little fingers overlap the dorsum of the 4th whereas on the radial side digit 2 overlaps digit 3. Other types of overlapping are included.
	Palmar polydactyly	Extra digit arising from palmar surface near wrist. Rare abnormality. Single case report.
	Post-axial polydactyly of fingers	The extra digits arise from the little finger side.
	Pre-axial polydactyly of fingers	The extra digits arise from the thumb side. Might be difficult to distinguish from a bifid thumb in which case use both in the same box.
	Radial deviation of fingers	The fingers are swept to the radial side.
	Skin syndactyly of fingers	There is normally a little skin between the fingers, especially when viewed from the dorsum. Here the amount of skin is exaggerated. The fingers seem 'webbed'. Also includes complete soft-tissue fusion between fingers.
	Tapering fingers	The fingers taper normally from proximal to distal aspects, but here, there is an exaggeration of this.
	Thick fingers	Thick, 'juicy', swollen-looking fingers - they may also be tapered.
	Thin fingers	Often in association with long phalanges - rather use the latter feature if present.
	Ulnar deviation of fingers	The term 'wind swept' is used to describe the deviation of the fingers (they seem blown to the ulnar side).
	Wide finger tips	The term 'spatulate' is used to denote the clubshaped expansion of the distal part of the digits. See clubbing of fingers (drumstick) . Could be confusing - use both as either/or when appropriate.

Body System (and feature path)	Feature	Useful comments aiding choice of feature
HANDS (continued)		
Hands, general abnormalities	Absent hands	Amania. Might have to use this in conjunction with reduction deformity of arms - no digits .
	Broad hands	A wide hand.
	Clenched hands	If appropriate use with camptodactyly and limited movement of fingers .
	Hypothenar hypoplasia	Those hand muscles on the ulnar side of the palm are atrophic.
	Large hands	Large, hypertrophied hands.
	Oedema of hands	Swollen hands due to fluid retention.
	Radial deviation of hand	Much rarer than ulnar deviation, the hand deviates towards the radial side. Also called radial club hand. May be associated with radial aplasia/hypoplasia.
	Small hands	Tiny, delicate hands.
	Split/cleft hand/ectrodactyly	A lobster-claw deformity. Includes monodactylous (there is a single finger bordering the cleft)
	Thenar hypoplasia	Under-development of those muscles at the base of the thumb.
	Trident hand	As seen in achondroplasia. The 4th and 5th fingers seem to deviate laterally whereas the thumb, index and forefinger deviate medially (best seen from the dorsum) leaving gaps between the splayed fingers.
Metacarpals, general abnormalities	Ulnar deviation of hand	Note that there is a feature ulnar deviation of fingers - use when appropriate together in same box.
	Absent metacarpals	Mostly in association with absence of elements of the fingers.
	Accessory metacarpals/pseudo-epiphyses	Additional metacarpals or extra ossification centres at the base.
	Long metacarpals	Often seen in arachnodactyly . Use this latter feature in preference, if present.

Body System (and feature path)	Feature	Useful comments aiding choice of feature
HANDS (continued)		
Metacarpals, general abnormalities (continued)	Proximal tapering of metacarpals	The metacarpals are normally expanded at their proximal ends (the base). Here they are pointed.
	Short/hypoplastic metacarpals	Short and sometimes 'stubby'. The term brachymetaphalangy is used when both metacarpals and phalanges are short.
	Synostosis of metacarpals	Bony fusion, either proximal or distal.
	Wide metacarpals/modelling defect	Wide and without the normal mid-shaft narrowing.
Phalanges, general abnormalities	Absent phalanges	One or more is very rudimentary, or absent.
	Accessory phalanges/pseudo-epiphyses	An extra phalanx or epiphysis, usually at the base of the proximal phalanx.
	Acro-osteolysis/acral defects	Resorption of bone of the distal phalanges.
	Cone-shaped epiphyses of phalanges	The proximal epiphysis of a phalanx has lost its normal disc shape, and is elevated centrally to form a cone.
	Delta shaped phalanges	A rare deformity. The bone is triangular shaped with a continuous epiphysis running from proximal to distal end of the short side.
	Hypoplastic phalanges	An under-developed phalanx. Use in conjunction with absent phalanges
	Long phalanges	Often long and slender as in arachnodactyly . Use together as either/or.
	Narrow phalanges	Narrow or thin.
	Phalanges, bifid	Might be indicative of polydactyly . Try using these two together if the first search fails.
	Short phalanges	Use in same box as hypoplastic phalanges , as it might difficult to distinguish between the two.
	Synostosis/symphalangism of fingers	Osseous union between two or more phalanges, obliterating the joint space.
Wide phalanges	Often shaped like a bullet, especially if wide and short.	

Body System (and feature path)	Feature	Useful comments aiding choice of feature
HANDS (continued)		
Thumbs, general abnormalities	Abducted/hitchhiker thumb	To hitch-hike one hyper-extends the thumb away from the palm
	Absent or hypoplastic thumbs	Seen alone or as part of radial aplasia.
	Adducted thumbs	A thumb which deviates over the palm of the hand. Also called a cerebral thumb.
	Broad thumbs	A broad, spatulate and sometimes deviated thumb.
	Polydactyly/bifid thumb	Please use together in the same box as pre-axial polydactyly of fingers . In the literature they are often used interchangeably.
	Proximal placement of thumb	The thumb seems short, but in essence it is inserted much too close to the radius (i.e., proximally).
	Short thumb	Difficult feature - use together with hypoplastic thumb and make sure that the so-called shortness is not due to proximal placing.
	Triphalangeal thumb	Clinically a long thumb that might look like a finger (a fingerised thumb). This might be opposable or non-opposable.
	Valgus deformity of thumb	Don't use. It is not clear what the thumb should look like.
	Varus deformity of thumb	Don't use. It is not clear what it means
JOINTS		
Joints, general abnormalities	Contractures (including arthrogryposis)	Fixed flexion or sometimes extension of multiple joints.
	Enlarged joints	Mostly a secondary phenomenon. Underlying pathology includes inflammation and structural metaphyseal changes.
	Laxity	A commonly inherited, uncomplicated feature. Check both parents before using it in a patient with a possible syndrome.
	Multiple joint dislocation	The hip usually dislocates first, but this feature needs multiple (knee, hips, ankle, elbow etc) joints to be affected.

Body System (and feature path)	Feature	Useful comments aiding choice of feature
JOINTS (continued)		
Joints, general abnormalities (continued)	Stiffness/arthritis	Joints which are stiff, but there are no true contractures.
LOWER LIMBS		
Ankle, general abnormalities	Dislocated/lax ankles	Might be part of multiple joint dislocation . See under JOINTS.
	Limited movement of ankle	Look for underlying cause, before using this feature.
Femur, general abnormalities	Absent or hypoplastic femur	An absent or small femur. There is another feature short femur . Use together.
	Bifid distal femur	A distal bifurcation, sometimes felt as an extra knob, or seen radiologically.
	Bowed femur	A bent (both acute- like kyphomelia, and gradual angles) femur.
	Coxa valga	The angle between the neck and shaft is > 140 degrees.
	Coxa vara	There is, radiologically, a decrease in the angle between the neck and the shaft, to less than 120 degrees
	Short femur	You might want to use this with hypoplastic femur .
Fibula, general abnormalities	Bowed fibula	A bent or serpentine, wavy fibula
	Duplicated fibula	Total or partial duplication (includes bifid).
	Hypoplastic or absent fibula	Small, or not present.
	Long fibula	Both proximal and distal ends might be longer than normal. An expert radiologist is needed.
	Wide fibula	A broad fibula, which is normally a slender bone
Hip, general abnormalities	Dislocation of hip	Not always a relevant feature. Use with care.
	Flexion deformity of hip	Use with multiple joint contractures if appropriate.
	Limited movement of hip	Look for a cause and use that feature - if possible!

Body System (and feature path)	Feature	Useful comments aiding choice of feature
LOWER LIMBS (continued)		
Hip, general abnormalities (continued)	Perthe's/dysplastic hip	An avascular necrosis of the capital femoral epiphyses and the femoral head. Better known as Legg-Calve-Perthe hip.
Knee, general abnormalities	Cartilage hypertrophy	Increased cartilage tissue due to enlargement of cartilage cells.
	Dislocated knee	Check for other dislocations and if present can also use multiple joint dislocation .
	Flexion deformity of knee	An inability to straighten knee. Look for cause, before using this feature.
	Genu valgum	Also called knock knees. The child stands with knees together and feet apart.
	Genu varum - bow legs	Also called "bow legs".
	Hyper-extensible knees	Part of joint laxity. If generalised, rather use laxity
	Limited movement of knee	Note another feature flexion deformity of knee . Not good features. Seek cause. Use both, if no cause is found.
	Webbing of knee	Might be part of multiple sites or it might just be a popliteal pterygium.
Lower limbs, general abnormalities	Absent lower limb	A total, or near total deficiency.
	Acromelia of lower limbs	The distal segment (ankle to toes) is short.
	Asymmetric lower limbs	Usually one limb is smaller than the other. There is a separate feature for hypertrophy of lower limb
	Constriction rings of lower limb	There is a visible indentation, as if made by the marks of a string, tightly bound around the limb. Distal to the constriction, the limb tapers.
	Hypertrophy of lower limb	Hypertrophy of subcutaneous tissue, and possibly bone
	Lower limb duplication	A rare manifestation of disorganisation, or partial twinning.
	Mesomelia of lower limbs	Middle segment (from knee to ankle) shortening.
	Oedema of lower limbs	Fluid retention (sometimes pitting) in lower limb

Body System (and feature path)	Feature	Useful comments aiding choice of feature
LOWER LIMBS (continued)		
Lower limbs, general abnormalities (continued)	Proportionate shortening of lower limb	Please use with short stature proportionate
	Reduction deformity of legs, no digits	Sometimes difficult to differentiate from constriction rings of lower limb . If so, use both together.
	Reduction deformity of legs, some digits	The limb has a terminal transverse defect, but there is an attempt to form digits.
	Rhizomelia of lower limbs	A short upper segment (from hip to knee).
	Sirenomelia	Mermaid deformity. There is fusion of the lower limbs.
Patella, general abnormalities	Absent or hypoplastic patella	Absent or small. Note that the patella is only radiologically visible after the age of 36 months. It is better to feel for it.
	Bipartite patella	The patella is divided into two halves. A duplicated patella.
	Dislocated patella	Mostly displaced laterally.
Tibia, general abnormalities	Bowed tibia	A bent or serpentine or wavy tibia
	Hypoplastic or absent tibia	Under-developed or absent tibia.
	Long tibia	An infrequent finding!
MOUTH		
Lower lip, general abnormalities	Cleft of the lower lip	Rare, but use for both lateral and median clefts
	Drooping of lower lip	It might be necessary to use with prominent/everted lower lip , even with thick lower lip .
	Pits of lower lip	Seen on either side of the centrum of the lower lip, although sometimes hidden from view in the mouth cavity. Can be very subtle. Some pits have areas that are raised above the surface.

Body System (and feature path)	Feature	Useful comments aiding choice of feature
MOUTH (continued)		
Lower lip, general abnormalities (continued)	Prominent/everted lower lip	The lower lip is prominent because it rolls downwards. Use in same box as thick lower lip and drooping of lower lip if uncertain.
	Thick lower lip	Might be confused with drooping lower lip or prominent/everted lower lip - use all three together
	Thin lower lip	Mostly seen with thin upper lip , but use with discretion!
Mouth, general abnormalities	Asymmetric mouth	Usually means that it is 'crooked,' but not caused by a facial nerve palsy. Does include 'asymmetric crying face'.
	Commissural lip pits	Lip pits situated at the angle of the mouth. Note that there is also a feature for pits of lower lip on either side of the midline.
	Cupid bow/tented upper lip	A cupid (a winged boy with a bow) has a bow which has a pronounced upward (convex) curve with a smaller concave curve in the middle.
	Down-turned corners of the mouth	The corners of the mouth normally end in the horizontal position.
	Macrostomia	A large opening. Includes a wide mouth
	Microstomia/narrow mouth	A small mouth opening.
	Open mouth appearance	The mouth droops open and tends to stay that way for long periods of time. Seen in profound hypotonia of any cause.
	Stomatitis	Irritation/inflammation especially at the corners of the mouth.
Philtrum, general abnormalities	Long philtrum	A long area between the columella of the nose and margin of the upper lip.
	Prominent/deep philtrum	A deep groove between the pillars or, prominence of the pillars.
	Short philtrum	Short area between columella and upper lip margin.
	Simple/smooth/absent philtrum	A featureless philtrum. A smooth philtrum.

Body System (and feature path)	Feature	Useful comments aiding choice of feature
MOUTH (continued)		
Philtrum, general abnormalities (continued)	Wide philtrum	The philtral pillars and the intervening groove are widely spaced.
Upper lip, general abnormalities	Cleft upper lip (non-midline)	As in cleft lip and palate.
	Midline cleft upper lip	Often a small V-shaped indentation in the mid-line.
	Prominent upper lip	A projecting upper lip. Use with thick upper lip if necessary.
	Thick upper lip	Use with prominent upper lip if unsure.
	Thin upper lip	A pencil-line upper lip
MUSCLES		
Muscles, general abnormalities	Absent muscles	Congenital absence of, for instance, trapezius or latissimus dorsi (not pectoralis).
	Calcification of muscle	There is a separate feature for subcutaneous calcification . When in doubt use together
	Lactic acidosis	Acidification of the blood due to a build up of lactic acid, arterial blood pH less than 7.35
	Malignant hyperthermia	Often, post anaesthetic, there is a rising temperature, rigidity, tachycardia, and an inability to wake up!
	Muscle atrophy	Do not use for muscle atrophy due to anterior horn cell disease or a peripheral neuropathy. There must be clear evidence of primary muscle disease.
	Muscle fasciculation	Contractions which can be seen, of the myofibrils which make up the motor unit. Nearly always seen (when pathological) in the presence of muscle atrophy. Indicative of anterior horn cell disease (mostly).
	Muscle hypertrophy	True and pseudohypertrophy. Do not use when the muscle is prominent due to overlying fat atrophy.

Body System (and feature path)	Feature	Useful comments aiding choice of feature
MUSCLES (continued)		
Muscles, general abnormalities (continued)	Muscle weakness/myopathy	Use with muscle atrophy where necessary. For this category, weakness can be at either muscle, peripheral nerve or anterior horn cell levels.
	Muscles, abnormal morphology/insertion	This is not a useful feature and should only be used for an abnormal muscle insertion.
	Myotonia	Prolonged contraction of muscle after voluntary contraction. The inability to briskly unclench the fist (do not use the handshake sign - it is too crude). Ask the patient to make a fist.
NAILS		
Nails, general abnormalities	Abnormal coloured nails	Often seen in conjunction with 'dysplastic' nails, where the nails might be yellowish.
	Absent nails	Anonychia. Might be seen in conjunction with small hypoplastic nails . Use together if a single feature search fails. We have not distinguished between absent nails on radial and ulna sides, although the difference might be significant.
	Bifid nails	This might be indicative of an underlying polydactyly (Pre-axial polydactyly of fingers , Pre-axial polydactyly of toes , Post-axial polydactyly of fingers , Post-axial polydactyly of toes , Polydactyly/Bifid hallux , Polydactyly/Bifid Thumb). X-ray to be sure. If an extra digit is present, use together as either/or (in same box).
	Broad nails	Might be seen with underlying polydactyly (Pre-axial polydactyly of fingers , Pre-axial polydactyly of toes , Post-axial polydactyly of fingers , Post-axial polydactyly of toes , Polydactyly/Bifid hallux , Polydactyly/Bifid Thumb) especially pre-axial polydactyly. If very broad, an X-ray might be appropriate.
	Concave nails	Koilonychia. An unusual feature. The nails are 'scooped out', or spoon shaped.

Body System (and feature path)	Feature	Useful comments aiding choice of feature
NAILS (continued)		
Nails, general abnormalities (continued)	Dystrophic nails	Means bad growth, but it implies a progressive (degenerative) condition of the nails.
	Hyperconvex/clubbed nails	An accentuation of the normal convexity.
	Nail pits	Discreet, small, indentations in the nail.
	Short nails	Must not be confused with nails that are bitten. Short from proximal to distal.
	Small/hypoplastic/deepset nails	Small, underdeveloped nails.
	Thickened nails	Might be a sign of dystrophic nails (use together if the case), or incidental fungal infection.
	Thin/brittle nails	Might be a sign of a dystrophic nails . If so use both in the same box. Mostly diagnosed (they break easily) on history.
	Volar nails	Nail arising from ventral surface of a digit instead of the normal dorsal aspect. Usually found on the 5 th digit in the hands. Also describes nails at any ectopic location
NECK		
Neck, general abnormalities	Branchial cleft/sinus/cysts	Pits, or skin tags seen in the neck, which are remnants of the embryological branchial clefts.
	Goitre	Enlarged thyroid gland. Use with hyperthyroidism or hypothyroidism if associated
	Long neck	A swan-like neck
	Loose skin in neck	Extra folds of skin at the nape of the neck (seen sometimes in conjunction with a short neck . See also webbed neck .)
	Low posterior/trident hairline	A trident is a 3-pronged fish spear used by Neptune. A trident hairline has 3-forks pointing down the neck. Beware it may be a normal feature.

Body System (and feature path)	Feature	Useful comments aiding choice of feature
NECK (continued)		
Neck, general abnormalities (continued)	Midline web	A midline web of the neck between undersurface of mandible and sternum. Rare. May be associated with a cleft chin or cleft sternum.
	Nuchal bleb/cystic hygroma of neck	These are fluid filled cystic swellings, mostly at the back or side of the neck, but which might encircle the neck. They might have a single space, or be divided by septa into many chambers.
	Short neck	The head seems to sit directly on the shoulders (or closer than normal to the shoulders).
	Thick/broad neck	A neck which seems overburdened with tissue.
	Torticollis, including stiff neck	A fixed, twisted position of the neck, either to the side (torticollis) or the back (retrocollis).
	Webbed neck	This is the description of a prominent fold of skin extending from the mastoid area to the acromion on the shoulder. Also called pterygium colli.
NEUROLOGY		
Behaviour, general abnormalities	ADHD	Attention deficit hyperactivity disorder: Inability to concentrate, failure to listen to instructions, fidgeting with hands and feet, talking too much, and having trouble paying attention to and responding to details
	Aggression	Physical or verbal behaviour that is intended to cause harm or pain.
	Autism/Autistic Behaviour	Severe impairment in social relationships, verbal and to some extent non-verbal communication, ritualistic and compulsive behaviour and defects of attention.
	Dementia	Loss (usually gradual) of mental abilities such as thinking, remembering, and reasoning.
	Irritability	An emotional incontinence. Easily irritated (aroused by a trivial stimulus).
	Lethargy	Drowsy inertia.

Body System (and feature path)	Feature	Useful comments aiding choice of feature
NEUROLOGY (continued)		
Behaviour, general abnormalities (continued)	Obsessive behaviour	Repetitive and persistent behaviours or mental acts that the patient feels driven to perform, possibly according to rules that must be applied rigidly.
	Psychosis	A major mental disorder of organic or emotional origin marked by derangement of personality and loss of contact with reality, with delusions and hallucinations and often with incoherent speech, disorganised and agitated behaviour, or illusions
	Self mutilation	The act of intentionally hurting oneself
	Sleep disorder	Abnormal sleep patterns e.g.dysomnias, insomnia, periodic limb movement disorder, hypersomnia, circadian rhythm sleep disorders, tooth-grinding, bedwetting, snoring.
	Stereotypic/ repetitive movement	Repetitive, non-functional motor behavior e.g. hand wringing, head tics or lip biting
Learning disabilities, general abnormalities	Developmental regression	Loss of previously learned skills
	Early developmental delay	Delayed attainment of developmental milestones e.g. sitting, crawling, walking, talking
	Expressive speech delay	A delay in the ability to generate speech
	Global speech delay	A delay in the ability to generate or to understand speech
	Mild learning disabilities	Mild disruption of functional skills including the ability to speak, listen, read, write, spell, reason and organise information, IQ of 50-70
	Moderate learning disabilities	Moderate disruption of functional skills including the ability to speak, listen, read, write, spell, reason and organise information, IQ of 35-50
	Profound learning disabilities	Profound disruption of functional skills including the ability to speak, listen, read, write, spell, reason and organise information, IQ below 20
Receptive speech delay	A delay in the ability to understand the speech of others	

Body System (and feature path)	Feature	Useful comments aiding choice of feature
NEUROLOGY (continued)		
Learning disabilities, general abnormalities (continued)	Severe learning disabilities	Severe disruption of functional skills including the ability to speak, listen, read, write, spell, reason and organise information, IQ of 20-35
Neuro, general abnormalities	Anosmia	Loss of sense of smell. Could be local in the nose or have intracranial reasons.
	Ataxia	A wide-based unsteady gait, titubation, an abnormal finger-nose test with increasing tremor as the nose or finger is reached.
	Autonomic dysfunction	This feature includes impotence, loss of control of bowel and bladder, inability to maintain blood pressure and loss of temperature control.
	Cranial nerve palsies	Excludes dysphagia , facial weakness , and anosmia . These are separate features.
	Extra-pyramidal disorder	This includes dystonia, chorea, athetosis, and rigidity.
	Hemiplegia	Weakness on one side of the body. Includes alternating hemiplegia.(first one side, then the other)
	Hypotonia	Floppiness including that due to muscle disease or peripheral nerve disease. The origin is mostly central nervous system.
	Indifference to pain	This feature includes congenital insensitivity to pain but also includes those unable to feel extremes of pain and who are yet are able to feel touch.
	Mental retardation/developmental delay	Includes mental handicap, learning disabled, but not dementia. It is not progressive and the delay should be present from birth.
	Peripheral neuropathy	Absent reflexes, peripheral weakness and loss of peripheral sensation. Should be confirmed on EMG and conduction studies.
	Seizures/abnormal EEG	This includes epilepsy, fits, drop attacks of all types.
	Spasticity/increased tendon reflex	Hyperactive tendon reflexes, often with increased tone and extensor plantar responses.
Spinal muscular atrophy/anterior horn cell abs	Disorders of the lower motor neurone resulting in weakness, wasting and fasciculation. Should be confirmed on EMG.	

Body System (and feature path)	Feature	Useful comments aiding choice of feature
NEUROLOGY (continued)		
Neuro, general abnormalities (continued)	Spontaneous pain sensation/hyeraesthesia	Pins and needles or an unpleasant, burning, stinging sensation.
	Tremors	Involuntary shaking of whole or part of the body.
NOSE		
Alae nasi, general abnormalities	Notched/hypoplastic alae nasi	The alae nasi are the wings of the nostrils. In some types of facial clefting they might be notched. Use with facial cleft when necessary.
	Thick alae nasi	The wings of the nostrils are thickened. Might be a part of coarse facial features .
Nares, general abnormalities	Anteverted nares	The nostrils normally point slightly laterally. Anteverted nares point forwards, so that you can look directly into the nasal cavity.
	Asymmetric nares	Mostly one cavity is smaller than the other. There is another feature for asymmetric face when more than the nares is involved. Use together when appropriate.
	Broad nares	A broad cavity (from side to side).
	Flared nares	The normal rounded configuration is altered, in that the nostrils 'tower' like that of a horse.
	Single nostril	A single, often centrally placed cavity (as in holoprosencephaly - use that term if that is present). It might be due to a malformation of one cavity with remaining remnants.
	Supernumerary nostrils	Additional cavities.
	Thin nares	Thin, narrow nostrils
Nasal bridge, general abnormalities	Depressed/flat nasal bridge	The bridge is that part of the nose situated between the eyes. It might be difficult to assess in infancy when it is normally quite flat. If it remains so, or is particularly flat, use this category

Body System (and feature path)	Feature	Useful comments aiding choice of feature
NOSE (continued)		
Nasal bridge, general abnormalities (continued)	High/prominent nasal bridge	A raised bridge (that part of the nose between the eyes). The nose appears particularly straight with a high take off.
	Thin nasal bridge	Thin from side to side. If the nose as a whole is pinched, use the separate feature pinched nose .
	Wide nasal bridge	That portion of the nose situated between the eyes (the bridge) seems wide.
Nasal columella, general abnormalities	Absent columella	The columella is the fleshy distal margin of the nasal septum. This appears absent.
	Columella below alae nasi	The columella is the fleshy distal margin of the nasal septum. It runs from the tip of the nose to the subnasion. The insertion of the columella inferiorly, is anteriorly displaced, when looking from the side.
	Rounded nasal columella	Normally the columella has a sharp configuration when viewed from the front. Here it is rounded.
Nasal septum, general abnormalities	Absent cartilage of nasal septum	If the cartilage is deficient, the septum feels soft.
	Broad nasal septum	The same as thickened.
	Deviated nasal septum	Deviated from its normal midline position.
	Long nasal septum	A difficult feature, mostly do not use. There is only one condition with this feature and even there, it is not essential.
	Narrow nasal septum	There are no syndromes with this feature. Unclear what this means.
	Short nasal septum	The septum terminates prematurely. Not easy to assess.
Nasal skin/mucosa, general abnormalities	Atrophic nasal skin	Thin, often sunken, white or pink skin overlying the nose. Gives the nose an aged appearance.
	Nasal haemangioma	A blood vessel tumour over, or in, the nose
	Nasal telangiectasia	A crop of dilated, tangled capillaries and other small blood vessels in or on the nose.

Body System (and feature path)	Feature	Useful comments aiding choice of feature
NOSE (continued)		
Nasal skin/mucosa, general abnormalities (continued)	Nasal tumours/polyps/papillomata	In fact any nodular lesion.
Nasal tip, general abnormalities	Bifid nasal tip	A cleft of the tip (not clefing of the nose which means the whole nose).
	Broad nasal tip	A cleft of the tip (not clefing of the nose which means the whole nose)
	Bulbous nasal tip	An accentuation of the normal round prominence at the tip. Becomes the shape of a light bulb.
	Overhanging/depressed nasal tip	An alteration in shape. Flat or curved down.
	Upturned tip	As described
Nose, general abnormalities	Broad base to nose	An increased distance between the alae nasi.
	Choanal atresia/stenosis	The choanae are the nasal passages. They might be obliterated or narrowed.
	Clefting of nose	Note use bifid nasal tip for a more localised cleft. A cleft of the nose can be midline or lateral. When midline consider using it with bifid nasal tip .
	Complete absence of nose	A rare malformation. Might be used in conjunction with hypoplasia as there might be a continuum.
	Concave profile of nose	Not an easy sign. Seen in a nose with flat nasal bridge.
	Convex/beaked profile of nose	A hooked nose. Racial and family characteristics must be taken into account.
	Flat nose	Might look squashed when viewed from above.
	Large nose	The nose is seldom measured, so this is a subjective assessment taking into account the family pattern.
	Pinched nose	Pinched from side to side. As if squashed between finger and thumb.
Small/short nose	Short from top to bottom. Measured from nasion to sub-nasion.	

Body System (and feature path)	Feature	Useful comments aiding choice of feature
NOSE (continued)		
Nose, general abnormalities (continued)	Upturned nose	An elevation of the tip of the nose.
ORAL REGION		
Alveolar ridges, general abnormalities	Cleft alveolar ridges	A cleft of the gum.
	Thick/wide alveolar ridges	The hillocks formed by the roots embedded in the bone, are thick and wide.
Gums, general abnormalities	Fusion of gums or jaws	Mostly a lateral fusion between upper and lower jaws.
	Gingivitis	Inflammation of the gums.
	Gum hypertrophy	A thickening of the gingiva, which might slowly cover the teeth.
	Periodontoclasia/periodontitis	Inflammation of the gums around the teeth causing them to become loose and fall out.
Oral region, general abnormalities	Leukoplakia	White plaques on the mucosal surface. They are slightly raised and might feel rough.
	Oral frenula (multiple)	The inner surface of the lip is connected to the gum by a fold of mucous membrane, the frenulum. If they are excessive in number or extent, use this feature.
	Oral pigmentation	Increased pigmentation (normal in those of African descent with dark skin).
	Oral synechia	Strands of tissue, often laterally placed, running between upper and lower jaws.
	Oral tumour	Any type
	Salivary glands, abnormal	Anything (usually absence) of the salivary gland.
Palate, general abnormalities	Cleft palate	A cleft (midline or otherwise) of the bony or soft roof of the oral cavity. Includes all shapes. If there is a cleft lip Cleft upper lip (non midline) , Cleft of the lower lip , Midline cleft upper lip .

Body System (and feature path)	Feature	Useful comments aiding choice of feature
ORAL REGION (continued)		
Palate, general abnormalities (continued)	Cleft uvula	Mostly a midline cleft. Might have the same significance as a cleft palate . Consider using them together in the same box.
	Fistulas of palate	A small hole, (opening) in the palate communicating with the nasal cavity. Not a cleft.
	High palate	A high, often gothic shaped palate, in which it is difficult to see the top.
	Narrow palate	Narrow from side to side. Often associated with a high palate so use together
	Prominent lateral palatine ridges	The main portion of the palate is formed by fusion of the lateral palatine processes (they are initially separated by the tongue). This entity refers to the prominent ridge formed by malfusion of the lateral with the medial processes.
	Short palate	Not a good sign. Short from front to back.
	Small or absent uvula	Known as the 'small tongue'. It is the pendent, (hangs down) fleshy part of the soft palate. It might be small or absent.
	Submucous cleft palate	A cleft covered by the mucous membrane of the soft palate
	Torus palatinus	A bony growth on the palate., usually on the midline of the hard palate, usually <2cm diameter
Tongue, general abnormalities	Absent or hypoplastic tongue	A small tongue lying posteriorly in the mouth, or an absent tongue.
	Bound tongue/ankyloglossia	A tongue with limited mobility due to it being attached (often by multiple frenulae) to the floor, or lateral margins of the oral cavity. In its simplest form, a 'tongue-tie'.
	Furrowed tongue/prominent groove	Mostly refers to a prominent mid-line groove. There are occasionally multiple grooves as in the so-called 'geographic' tongue.
	Glossoptosis	A tongue that hangs backwards in the oral cavity and might cause obstruction

Body System (and feature path)	Feature	Useful comments aiding choice of feature
ORAL REGION (continued)		
Tongue, general abnormalities (continued)	Large tongue	Macroglossia, as in Beckwith syndrome, due to an increase in bulk of the normal tongue tissue.
	Lobulated tongue (including hamartomata)	The tongue might be divided into lobules by multiple clefts or the lobules might consist of hamartomas. Use this feature for a single hamartoma as well.
	Midline cleft tongue	Mostly involving the tip of the tongue, but also in the form of a large cleft.
	Protruding tongue	The tendency to 'push' the tongue out and keep it out. This might be an involuntary extrapyramidal movement.
	Smooth tongue	A tongue devoid of any markings but particularly due to absent papillae as in the Riley-Day syndrome.
	Tongue fasciculations	As seen in anterior horn cell disease, the tongue shows signs of atrophy and involuntary muscle twitching. View with the tongue in the mouth.
PELVIS		
Acetabulum, general abnormalities	Acetabular spurs	These are bony projections arising from the borders of the acetabulum.
	Acetabulum protrusio	The acetabulum is abnormally deep.
	Horizontal acetabulum	The normal alignment of the acetabulum is forward and down - here it is unduly horizontal.
	Steep acetabulum	An increase in the slope of the acetabulum.
Ilium, general abnormalities	Flared iliac wings	This is the over-exaggerated fan-like (sometimes rounded) expansion of the upper part of the ilium (i.e. the wings).
	Hypoplastic ilia	An under-development of the ilium. Includes both inferior narrowing and small iliac wings.
	Iliac horns	These are horns (projections) arising from the middle of the iliac wings posteriorly.

Body System (and feature path)	Feature	Useful comments aiding choice of feature
PELVIS (continued)		
Ilium, general abnormalities (continued)	Iliac wings, lacy border	The crest of the iliac wings lose their regular outline and resemble the frilly edge of lace.
Ischium, general abnormalities	Ischium, ossification defect	Abnormal bone formation in the ischium
Pelvis, general abnormalities	Asymmetric pelvis	Mostly due to hypoplasia on one side.
	Large pelvis	Large basin which constitutes the pelvis (not a useful sign)
	Narrow pelvis	The whole pelvis is narrow from side to side.
	Small pelvis	The bony structure of the innominate bone (the hip bone) is in general small
Pubis, general abnormalities	Narrow pubis	The rami of the one side are too close to the other side. The distance between the sides is too small.
	Pubic ossification defect	Mostly delayed ossification, but includes deficient ossification. The rami are poorly ossified between birth and 8 weeks, but thereafter until 2-3 years they should normally ossify.
	Wide pubis	The anterior part of the pelvis (i.e. where the pubic rami are) is too wide.
Sacrosiatic notch, general abnormalities	Narrow sacrosiatic notch	A narrow, pinched notch in comparison with normals.
	Wide sacrosiatic notch	This is the deep notch bounded above by the ilium and below by the ilium and ischium. A radiological sign when viewed on the AP of the pelvis.
SKELETAL		
Skeleton, general abnormalities	Advanced bone age/large epiphyses	Must be measured by experts using standard tables. Sometimes manifests as 'disharmonious' maturation.
	Cartilaginous/bony exostoses	They appear as swellings at the ends of long bones, shoulder and pelvic girdle. Radiologically, they are cartilage-capped protuberances arising from near the epiphysis and pointing away from it.

Body System (and feature path)	Feature	Useful comments aiding choice of feature
SKELETAL (continued)		
Skeleton, general abnormalities (continued)	Delayed bone age	Diagosed on X-ray using standard tables showing the age of appearance of the carpal bones, and other ossification centres.
	Enchondromata	Multiple bony swellings. Radiologically they appear as multiple lucent defects.
	Fibrous dysplasia of bones	If limited to one bone 'monostotic' .If multiple bones are involved, 'polyostotic'. There are cystic radiolucent areas and sclerosis expanding the bone.
	Lytic lesions of bone	These are circumscribed areas of bone resorption (mostly single or few in number). Use osteolysis for more diffuse involvement.
	Multiple fractures	Non-accidental injury must be thought of, and excluded.
	Osteolysis	Disappearing bone due to resorption. Note there are 3 other features 1) osteolysis of carpals 2) osteolysis of tarsals 3) Acro-osteolysis . Never use alone.
	Osteomalacia	Delayed mineralisation of mature osteoid tissue. The adult equivalent of rickets.
	Osteomyelitis	An inflammation of bone and bone marrow (usually caused by bacterial infection, sometimes by fungal infection)
	Osteoporosis	The bone is thin, porotic, and will later bow or collapse. Radiologically there is a reduction in density.
	Osteosclerosis or osteopetrosis	Hardening of bone. Radiologically, an increase in density. Osteopetrosis is the most severe manifestation.
	Skeletal cysts or tumours	Excludes exostoses and enchondromata (see elsewhere).
Tubular bones, general abnormalities	Aseptic necrosis of epiphysis	Fragmentation of an epiphysis. Use Perthe's hip in addition (same box) if the aseptic necrosis involves the capital femoral epiphysis.

Body System (and feature path)	Feature	Useful comments aiding choice of feature
SKELETAL (continued)		
Tubular bones, general abnormalities (continued)	Bone asymmetry	One smaller or larger than the bone on the other side. Rather use specific bone asymmetry i.e., humerus or femur.
	Bowing of bones	The long bones seem bent. Individual bones are also used in the feature list e.g. bowed tibia or bowed femur or bowed humerus Combine where necessary.
	Broad bones/modelling defects	The long bones seem to have lost the differentiation into metaphysis and diaphysis and there is no normal constriction at mid-shaft level. The bone seems featureless.
	Cortical hyperostosis/thickening	The cortex is thick and dense, protruding deeper into the shaft than normal.
	Cortical thinning of bones	The cortex can usually be clearly seen. Here it is like a thin line.
	Diaphyseal dysplasia	Abnormalities in the shaft, usually sclerosis, and widening.
	Epiphyseal dysplasia	Abnormalities in shape and size of an epiphysis.
	Epiphyseal sclerosis	A dense epiphysis.
	Hemi-hypertrophy	Although a tubular bone feature, use for Hemi-hypertrophy of a limb (clinically). See also asymmetric limb (Asymmetric lower limbs , Asymmetric arms)
	Medullary space stenosis	The translucent cavity of a long bone is narrow and almost obliterated.
	Melorheostosis	Linear streaks of hyperostosis along the shaft, often likened to 'dripping candle wax'.
	Metaphyseal dysplasia	Mostly seen as a cupped or irregular metaphysis. See other feature for wide metaphysis .
	Osteopathia-striata	Linear, longitudinal striations, particularly around the knee and femoral neck.
Osteopoikilosis	A spotty appearance of long bones (multiple sclerotic areas)	
Pseudoarthrosis	There is discontinuity of bone, non-union and increased motility at the site.	

Body System (and feature path)	Feature	Useful comments aiding choice of feature
SKELETAL (continued)		
Tubular bones, general abnormalities (continued)	Pseudo-fractures	The appearance of a partial crack in a long bone not related to trauma.
	Short bones	Be careful as there are separate features for hypoplastic humerus , hypoplastic tibia , hypoplastic femur , etc. Use other features with this where appropriate.
	Slender/thin bones	The bones seem so thin, and delicate that they look as if weight-bearing will cause a fracture.
	Stippled or fragmented epiphyses	Also referred to as 'punctate stippling' of the epiphyses or extra-cartilaginous calcification. Seen in vertebral column, pelvis, knee and other joints. There are multiple foci of flocculent calcification.
	Subperiosteal new bone formation	An attempt, after irritation of different kinds, to form new bone under the periosteum.
	Wedge or cone-shaped epiphyses of long bones	The normally disc-like epiphysis has become wedge-shaped, or like a cone. See also cone-shaped epiphysis of phalanges .
	Wide metaphysis	Wide or splayed. See elsewhere for metaphyseal dysplasia
SKIN		
Patchy skin defects, general abnormalities	Acne	A disorder of the pilosebaceous apparatus resulting in nodules and cysts over the face, chest and back.
	Atrophy - patchy	Circumscribed areas of depressed, translucent, discoloured wrinkly skin.
	Bullae or vesicles	Blisters containing free fluid.
	Comedones	Commonly known as black heads or white heads, caused by the clogging of the skin pores.

Body System (and feature path)	Feature	Useful comments aiding choice of feature
SKIN (continued)		
Patchy skin defects, general abnormalities (continued)	Epidermolysis	Blistering skin after light trauma. There are many types divided into three main categories dependent on the plane of cleavage. This feature covers all types.
	Patchy aplasia/hypoplasia of skin	Circumscribed areas, of depressed, deficient of skin, sometimes like a healed scar or still raw underneath.
	Scleroderma	Includes morphea. Can be round, linear, guttate. Some lesions have a white ivory centre and a lilac coloured border.
	Striae/stretch marks	Especially seen over the lateral margins of the abdomen
Skin pigment, general abnormalities	Acanthosis nigricans	Light brown to black, raised verrucous lesions found especially on the neck, axilla and groin. Beware the lesions sometimes disappear on washing with soap and hot water.
	Axillary freckles	Freckles under the arm (in the arm pits) - an unusual place (but not so unusual that red haired people do not have them on rare occasions).
	Curvilinear hyperpigmentation	Increased pigmentation in a curved line distribution
	Diffuse depigmentation of skin	A widespread loss of normal skin pigment, as in albinism.
	Diffuse increased pigmentation of skin	Widespread darkening of normal (for the family) skin pigmentation.
	Linear sebaceous nevus	Streaky, raised, dark (initially raised and pink) lesions, commonly occurring over the head and neck but which can occur anywhere.
	Nevi or lentigines	These include epidermal nevi, melanocytic nevi and pigmented nevi. Lentigines are small, brown or black, round lesions (1-2mm in diameter) darker, with more uniform pigmentation, than freckles (ephelides).
	Patchy depigmentation of skin	Small or large (but discreet areas) of white depigmented skin.
	Patchy pigment of skin/cafe au lait spots	These are large (>2.5cm in adults), light brown pigmented areas. In neurofibromatosis they have a smooth outline, but they might be ragged, as in McCune-Albright syndrome.

Body System (and feature path)	Feature	Useful comments aiding choice of feature
SKIN (continued)		
Skin tumours, general abnormalities	Adenoma sebaceous	These are angiofibromas and are hamartomas of that tissue (they are not truly adenoma of sebaceous tissue).
	Cysts	Epidermal, epithelial, sebaceous, dermoid, mucous.
	Keloids	An exaggerated connective tissue response to injury. They are raised, smooth and rubbery.
	Lipomata	These are benign tumours consisting of mature fat cells.
	Neurofibromas/schwannomas	If in NEUROLOGY database 'neurofibroma' is under 'Tumour'.
	Other tumours of skin	Any other tumour not listed. It might be better to search using Skin tumour - general abnormalities .
	Pedunculated skin lesions/skin tags	These are small benign tumours on a stalk.
	Warts/papillomata	Skin coloured, papules.
	Xanthomas	Soft, slightly raised, yellow-orange, lipid-laden, papules.
Skin, general abnormalities	Abnormal scar formation	A scar (new formation of connective tissue after a destructive lesion of epidermis and cutis) which is out of proportion to the injury, or is too thin and breaks down (tissue-paper scars)
	Calcification, subcutaneous	Often felt rather than seen. Confirmed by X-ray or removal.
	Cutis laxa	Extra, redundant skin which tends to hang in folds.
	Cutis marmorata	A reticulated, bluish mottling of the skin which is common in the newborn and may persist for weeks or months. It is a physiological response to the cold and consists of the dilatation of capillaries and small veins.
	Deficient adipose tissue or fat/lipodystrophy	The subcutaneous fat is deficient, causing the muscle to seem hypertrophied.
	Dimples	Skin dimples are the result of tethering of the skin to underlying structures (bone) causing an indentation. Seen especially over the shoulder and in the vicinity of joints.

Body System (and feature path)	Feature	Useful comments aiding choice of feature
SKIN (continued)		
Skin, general abnormalities (continued)	Eczema/atopic dermatitis	Chronically recurrent erythema, scaling, vesicles and crusts). Eczema means "boil over". Atopic dermatitis is used synonymously and means "not confined to one place".
	Erythema/erythroderma	A red skin which looks otherwise normal.
	Excessive skin folds/grooves	Any abnormal folds or grooves of the skin. Note separate features for excessive skin wrinkling and hyperelastic skin and cutis laxa .
	Excessive skin wrinkling	Might be the end result of extra, redundant skin. If so use with cutis laxa
	Fragile skin	Skin that tears or bruises easily on minor trauma. May be associated with impaired healing.
	Hyperelastic skin	Tug it and it stretches. Also called hyperextensible. See cutis laxa for loose folds of skin.
	Hyperhidrosis	Increased sweating, the skin being moist and clammy to touch.
	Hyperkeratosis	A diffuse or localized thickening of the stratum corneum resulting in skin that is dry, thickened and pigmented with fissures. Can be most marked on the palms and soles.
	Hypohidrotic or dry skin	This might be a clinical symptom. The sweat pores are best seen on the fingertips through a magnifying instrument with bright light (otoscope) when the lack of the normal glistening drops of fluid will be noted.
	Ichthyosis	Used for all degrees of scaliness including the thickened, cracked membrane of the collodion baby.
	Itching	An unpleasant sensation evoking the desire or reflex to scratch
	Keratosis pilaris	Horny follicular papules
	Macules	A small flat lesion showing an alteration in colour.
Oedema (including hydrops)	Localized fluid retention to severe generalized retention. See also oedema of hands , oedema of feet , etc.	

Body System (and feature path)	Feature	Useful comments aiding choice of feature
SKIN (continued)		
Skin, general abnormalities (continued)	Papules	A circumscribed, elevated lesion.
	Peeling skin	As described
	Pustules/ulcers	A circumscribed lesion containing pus. An ulcer is a deep erosion of the epidermis and cutis.
	Seborrhoea	Abnormally copious secretion of sebum to give an oily appearance to the skin. May predispose to acne.
	Skin photosensitivity	Hypersensitive reaction to sun light (ultra violet rays).
	Slow skin healing	Might be used in conjunction with abnormal scar formation , if both are present.
	Soft skin	On palpation it is too soft (it might feel doughy).
	Thick/stiff skin	The skin forms a tight encasement.
	Thin skin/generalized skin atrophy	Atrophy results in depression of the epidermis/dermis. Skin markings are lost but it wrinkles easily if you compress from the side. See also Skin atrophy - patchy .
Vascular change, general abnormalities	Capillary haemangioma	This term is used for a "port wine", "salmon patch" or any cutaneous vascular lesion consisting of a dense network of capillaries.
	Cavernous haemangioma	They are deep-seated strawberry lesions with little penetration of the overlying skin. They contain large dilated spaces. They are bluish-red with an ill-defined border.
	Mongolian spot	A benign flat birthmark often with blue discolouration but may be blue-grey or blue-black.
	Multiple bruises	Use this feature if there are multiple bruises not related to significant trauma.
	Prominent vessels of skin	Seen especially in the presence of thin skin, subcutaneous atrophy (fat atrophy) or premature aging.

Body System (and feature path)	Feature	Useful comments aiding choice of feature
SKIN (continued)		
Vascular change, general abnormalities (continued)	Purpura	Includes petechiae (pinpoint haemorrhage) and ecchymosis (large confluent patches).
	Telangiectasia/angiokeratomata of skin	Localised dilatation of small vessels in the skin to give a spiderly or punctate appearance.
	Urticaria	Transient, well circumscribed wheals. They are red and itch.
STATURE		
Short stature, general abnormalities	Short stature, prenatal onset	The diagnosis of short stature is likely to be made post-natally so where appropriate use additional features to this. Use for bone dysplasias present at birth.
	Short stature, proportionate	Proportionate implies that despite the short stature the relationship between the trunk length and limb length has not altered.
	Short stature, short limbs	This is disproportionate short stature. The trunk is normal but the limbs are short. Note there are separate features of proportionate short arms and proportionate shortening of lower limbs .
	Short stature, short trunk	This is disproportionate short stature in that the limbs are of normal length but the trunk is short. Note under 'Back and spine' there is another feature of short trunk .
Tall stature, general abnormalities	Tall stature, disproportionate/dolichostenomelia	This is mostly an increase in limb length, the trunk being relatively normal.
	Tall stature, proportionate	Increase in height above 2SD, for age. The increase in height involves both the trunk and the limbs.

Body System (and feature path)	Feature	Useful comments aiding choice of feature
TEETH		
Teeth, general abnormalities	Abnormally shaped teeth	Peg-shaped, conical teeth, screwdriver shaped teeth - all are included.
	Advanced tooth eruption/development	Excludes neonatal teeth, but the teeth erupt much faster than the norm which is: 6 months - lower central incisors 7 months - upper central incisors 8 months - upper lateral incisors 9 months - lower lateral incisors 12 months - first molars 18 months - canines
	Delayed tooth eruption/development	Normal times are (for milk teeth) 6 months - lower central incisors 7 months - upper central incisors 8 months - upper lateral incisors 9 months - lower lateral incisors 1 year - first molars 18 months - canines 2 years second molars
	Dental caries	Use with care. Might or might not be significant. It usually is not.
	Dental cysts/tumours	Any type. See also Tumour or cyst of the mandible .
	Dentin abnormalities	The bulk of the tooth, consisting of avascular calcified tissue, penetrated by tiny canals. An abnormality results in pearly discolouration of the teeth.
	Enamel abnormalities	The visible tooth - the crown - is covered by very hard calcified tissue- the enamel. An abnormality results in excessive wear and possible erosion down to gum level.
	Irregular or crowded teeth	The normal symmetry of tooth eruption is distorted resulting in irregular positioning and crowding. Might be secondary to alterations in jaw structure (see under mandible general abnormalities).

Body System (and feature path)	Feature	Useful comments aiding choice of feature
TEETH (continued)		
Teeth, general abnormalities (continued)	Large/prominent teeth	Also includes taurodontia (this includes gigantic roots as seen on X-rays). Also includes prominent central incisors.
	Malocclusion of teeth	The teeth in upper and lower jaws do not close with any precision on one another.
	Neonatal teeth	Teeth present at birth, or which erupt within the neonatal period.
	Oligodontia	Absent or missing teeth.
	Premature loss of teeth	Teeth erupt normally (or there is delayed eruption) and then fall out.
	Single central incisor	The single incisor (mostly the upper incisor) is in the mid-line. If uncertain an X-ray will help.
	Small teeth	Small for age..
	Supernumerary teeth	There is sometimes a double row of teeth, or one or more teeth erupting in abnormal positions.
	Talon cusp	A pointed or rounded projection of enamel on an anterior tooth that imparts a T shape. Can occur in the normal population.
	Taurodontism	The body of the tooth and pulp chamber is enlarged
	Wide-spaced teeth	This might be seen in conjunction with small teeth (use together when necessary). Also to be used for a wide gap between the central incisors (diastema).
THORAX		
Breasts, general abnormalities	Absent or hypoplastic breasts	Small or absent.
	Asymmetric breasts	Use together with small or hypoplastic breasts when necessary, in same box.

Body System (and feature path)	Feature	Useful comments aiding choice of feature
THORAX (continued)		
Breasts, general abnormalities (continued)	Breast tumours	Any nodule, benign or malignant in the breast.
	Gynaecomastia	Increased amount of the male breast tissue.
	Hypertrophied breasts	An unusually enlarged breast. Use with asymmetric breasts when appropriate.
	Premature development of breasts	Note that there is a separate feature early puberty in females
Clavicles, general abnormalities	Absent or hypoplastic clavicles	Absent or small. Short clavicles might be felt, or seen, when they end in a knob.
	Bipartite clavicle/pseudoarthrosis	Bifid clavicle. There is another feature pseudoarthrosis under SKELETON. Use both if there is a poorly healed bony defect in the clavicle that looks like a fracture.
	Bowed/hooked clavicles	The distal (acromial) end terminates in a pronounced hook.
	Broad clavicles	A radiological feature.
	Long clavicles	A radiological feature. Some are long and hooked - see bowed/hooked clavicles and use together when necessary.
Diaphragm, general abnormalities	Absent/hypoplastic diaphragm	Absent or poorly developed.
	Congenital hernia of diaphragm	Posterior-lateral herniation (mostly through the foramen of Bochdalek) mostly without a membrane and often containing viscera.
	Eventration of diaphragm	Poor muscularisation of the diaphragm, results in a dome-shaped bulge into the thorax. The bulge is covered by peritoneum.
	Hiatus hernia	A portion of the gastric fundus enters the thoracic cavity.
Heart, general abnormalities	Anomalous venous return	The pulmonary veins might connect to the right atrium. Also called TAPVC. (Total Anomalous Pulmonary Venous Connections), if all the veins connect anomalously, and PAPVC, if partially.

Body System (and feature path)	Feature	Useful comments aiding choice of feature
THORAX (continued)		
Heart, general abnormalities (continued)	Aortic incompetence	Might occur in those with a congenital abnormality of the aortic valve, or aortic root dilatation.
	Aortic stenosis	Occurs in 3-6% of congenital heart disease. Includes congenital bicuspid valve which may become stenotic and supra valvar aortic stenosis.
	Atrial septum defect	Secundum ASD - the incomplete formation or incomplete fenestration of the septum primum covering the fossa ovalis. See elsewhere for septum primum defect .
	Atrio-ventricular septal defect	Includes the term AV-canal defects and endocardial cushion defects. Occurs in 4-5% of congenital heart disease. It might be used together with Atrial septum defect and Ventricular Septal Defect or Heart - general abnormalities . Partial defects include primum ASD, inlet VSD defects, cleft of mitral leaf
	Cardiac situs inversus/dextrocardia	Any abnormality of cardiac situs. May occur with or without situs inversus abdominal .
	Cardiomyopathy	Includes hypertrophic and dilated cardiomyopathies.
	Coarctation/interrupted aorta	Occurs simply with or without a PDA in about half the cases and with other lesions (especially VSD) in the other half. It may be a long or short segment. Interrupted means a complete separation of ascending and descending aortal segments. Type A is interrupted distal to the left subclavian artery, type C between the carotids and type B between carotid and subclavian.
	Congenital cardiac anomaly, unspecified	Rather use Heart - general abnormalities .
Cor triatriatum	In this situation the pulmonary veins enter an accessory chamber that communicates with left or right atrium.	

Body System (and feature path)	Feature	Useful comments aiding choice of feature
THORAX (continued)		
Heart, general abnormalities (continued)	Duplicated/right aortic arch	A right aortic arch is a single arch that crosses over the right mainstem bronchus and passes to the right of the trachea. Note that it occurs in many patients with Fallot tetralogy (use together). In double or duplicated aortic arch, both right and left aortic arches are present (one might be hypoplastic). They form a vascular ring around the trachea and oesophagus. Use Heart-general abnormalities liberally.
	Ebstein anomaly	The posterior and septal leaflets of the tricuspid adhere to the ventricular wall, whereas the anterior leaflet is normally positioned. The valve is usually incompetent, although it might be stenotic. Usually associated with an ASD.
	ECG abnormality/conduction defects	Any abnormality of the EEG in the absence of a structural heart defect.
	Ectopia cordis	A partial or complete displacement of the heart outside the thorax through a sternal defect. Where necessary use together with asternia or bifid sternum .
	Fallot tetralogy	Can be variable. Over-riding aorta is important, VSD, pulmonary stenosis (essential) and right ventricular hypertrophy (which could be secondary). Use with other features if the first search fails to reveal something relevant.
	Hypoplastic left heart	There is a continuum of anomalies from hypoplasia, of the aorta, aortic valve, mitral valve, to hypoplasia of the left ventricle and left atrium.
	Mitral incompetence	Might be an underlying mitral leaflet cleft. Mild incompetence is common in women.
	Mitral stenosis	Includes hypoplasia of the valve and mitral valve atresia.
	Patent ductus arteriosus	Closure occurs post-natally in two stages. Within 12 hours (after full-term), shortening, thickening and protrusion of the thickened intima and then within 2-3 weeks infolding of the endothelium. Isolated patency occurs in 1 in 2000 live births.
	Pericarditis (constrictive)	Thickened, adherent pericardium that restricts ventricular filling.
Pericarditis (non-constrictive)	See elsewhere for pericarditis- constrictive . A pericardial effusion is included here.	

Body System (and feature path)	Feature	Useful comments aiding choice of feature
THORAX (continued)		
Heart, general abnormalities (continued)	Pulmonary hypertension	An increase in blood pressure in the pulmonary artery, vein or capillaries.
	Pulmonary incompetence	Rare as a congenital heart lesion
	Pulmonary stenosis	Includes valvular and supra-valvular stenosis. Also includes peripheral pulmonary artery stenosis. Note that these lesions often occur with others. Be broad - use general features, or many individual features together
	Septum primum defect	Ostium primum ASD is really part of an atrio-ventricular septal defect (see that feature). So once again use multiple categories in same box or heart-general abnormalities . The primum defect is less common than secundum. Most primum defects are large, and are bordered by a crescent of atrial septal tissue and by mitral-tricuspid valvular tissue
	Single ventricle	This includes 3 types: 1) Single left ventricle with or without an outlet infundibular chamber 2) Single right ventricle 3) Undifferentiated ventricle See also hypoplastic left heart .
	Transposition of the great vessels	Congenital defect involving abnormal spatial arrangement of great vessels
	Tricuspid incompetence	Other than that associated with Ebstein, this is a rare congenital anomaly. There are mostly structural anomalies of the valve. There is a separate feature for Ebstein anomaly .
	Tricuspid stenosis	Includes tricuspid atresia which is a complete agenesis of the valve with no communication between atrium and ventricle.
	Truncus arteriosus	There is a single artery that arises from the base of the heart which gives rise to the pulmonary, coronary and systemic arteries. There is a single semilunar valve (unlike aortic and pulmonary artery stenosis in which there are two atretic semilunar valves).
	Tumours of the heart	Rhabdomyomas, rhabdomyosarcomas, myxomas, fibromas, hamartomas and haemangiomas.

Body System (and feature path)	Feature	Useful comments aiding choice of feature
THORAX (continued)		
Heart, general abnormalities (continued)	Vascular ring	Seen in congenital anomalies of the aorta
	Ventricular septal defect	Includes perimembranous (80%), muscular (5-20%), outlet (just below the pulmonary valve) and inlet (posterior and inferior to perimembranous defect). Don't be frightened to use Heart - general abnormalities as different defects even occur in the same family.
Lung, general abnormalities	Apnoea or tachypnoea	A period of cessation of breathing (apnoea). Can be central, obstructive or mixed. Tachypnoea is fast breathing. Often 'panting' like a dog.
	Bronchiectasis	The pathological dilatation of one or more bronchi. In infancy may be, the result of absent or deficient cartilage plates.
	Bronchomegaly	Large bronchi.
	Lung cysts	These include bronchogenic cysts (derived from the trachea or main bronchi), and alveolar cysts, or a combination.
	Lung hypoplasia/agenesis	In agenesis, there is a complete absence of bronchi, alveoli and their blood supply. Included here is lobar agenesis, and abortive growth (hypoplasia). Lung hypoplasia may also be secondary to oligohydramnios.
	Pneumothorax	Air or gas within pleural cavity. Can be a medical emergency. Can be spontaneous or secondary to trauma.
	Pulmonary segmentation defects	The result of lobes that are fused together or extra fissures causing additional lobes. Accessory lobes are included.
	Respiratory abnormality, unspecified	A general category. Might think of rather using lung-general abnormalities .
	Sequestration defect	Piece of lung tissue that does not connect to the pulmonary circulation or other lung tissue.
	Trachea or laryngeal anomalies	A general category which includes, atresia, webs, stenoses, fistulae.
Tracheal or laryngeal calcification	Mostly seen on a chest x-ray. Calcification of respiratory cartilages in children is pathological.	

Body System (and feature path)	Feature	Useful comments aiding choice of feature
THORAX (continued)		
Nipples, general abnormalities	Hypoplastic/inverted/absent nipples	Also called athelia.
	Supernumerary nipples	Extra nipples in the "milk line". Seen both normally and in some syndromes (also called polythelia)
	Wide-spaced nipples	They are usually placed on the anterior axillary line and wide-spaced implies lateral displacement towards the mid-axilla.
Pectoral/shoulder girdle, general abnormalities	Absent or hypoplastic pectorals	Either the whole muscle is missing alone, sometimes with other chest wall muscles, or it might only be the sternal head of pectoralis major as in Poland anomaly. View carefully the symmetry of the skin folds in the anterior axilla.
Ribs, general abnormalities	Absent ribs	An X-ray finding, unless accompanied by hypoplasia (or absent) muscles of the anterior thoracic cage, when the anomaly might be clinically suspected.
	Beaded/wavy/constrictions of ribs	Wavy like an S (sinuous), or with localised narrowing. If beaded this indicates multiple fractures.
	Bifid/fused ribs	A radiological diagnosis.
	Extra ribs (including cervical)	Supernumerary ribs. Cervical ribs originate from C7 and are often bilateral but asymmetrical. They join the 1st rib or sternum, distally.
	Flared ribs/anterior splaying	An expansion, mostly at their ends.
	Gaps in ribs	Are usually seen radiologically and might be easy to see when the gaps are large or multiple, but difficult when discreet.
	Short ribs	Radiologically short. The chest will usually be narrow.
	Thick/wide ribs	A judgment made by the expert radiologist!
	Thin ribs	If thin and wavy, use together with wavy ribs .
Scapulae, general abnormalities	High or Sprengel shoulder	An elevation of the scapula which might be hypoplastic. About 1/3 have a omo-vertebral bone (a bony/cartilaginous mass attaching the scapula to the spinous processes of the cervical vertebrae).

Body System (and feature path)	Feature	Useful comments aiding choice of feature
THORAX (continued)		
Scapulae, general abnormalities (continued)	Scapulae, absent	Use together with small scapulae as they might be a continuum.
	Small scapulae	Can be subtle. Discuss with your radiologist.
	Winged scapulae	Use in conjunction with features on 'muscle' disease together if necessary. Winging implies that the scapulae 'sticks out', is laterally displaced and often rotated.
Shoulder shape, general abnormalities	Broad shoulders	Not a easy feature. Broad means wide from side to side.
	Narrow shoulders	The distance from the sternal notch to the tip of the shoulder is short
	Sloping shoulders	The angle between the neck and the shoulders becomes greater than a right angle.
Thorax, general abnormalities	Asternia or bifid sternum	Also includes clefts of the sternum. In complete failure of fusion of the sternal halves, the heart might only be covered by skin. Incomplete fusion both superiorly and inferiorly are more common. There is another feature - ectopia cordis - please use together when necessary.
	Asymmetric thorax	Use with care. Might be the result of scoliosis (use together in same box) or there might be general asymmetry (many other codes including one for asymmetric breasts .)
	Broad/barrel thorax	Broad and round - just like a barrel.
	Narrow thorax/funnel chest	Narrow from side to side. If also hollowed out, use with pectus excavatum
	Pectus carinatum	A protrusion defect. The acute angulation superiorly produces the 'pigeon' chest. The more diffuse protrusion produces the 'keel' shaped chest, or 'chicken breast' chest.
	Pectus excavatum	Depression deformity of the sternum and anterior chest wall.
	Pleural effusion/chylothorax	The presence of fluid, including lipid-laden, milky chyle in the pleural cavity.

Body System (and feature path)	Feature	Useful comments aiding choice of feature
THORAX (continued)		
Thorax, general abnormalities (continued)	Short sternum	A subjective, feature (seldom measured) denoting a reduction in length from top to bottom.
	Short thorax	Short from top to bottom. Use in conjunction with short trunk where necessary
	Stridor	A harsh, noisy sound heard primarily on inspiration. Denotes large airway obstruction and can be the consequence of a floppy larynx or other pathology.
UPPER LIMBS		
Elbow, general abnormalities	Cubitus valgus	An increased carrying angle at the elbow. This is normally greater in females than in males.
	Dislocated elbow	Either clinically or radiologically. If there are bony lesions at the elbow, rather use that. There is a separate feature for multiple joint dislocation . Only use this feature for a single dislocation.
	Elbow joint/radio-humeral synostosis	Clinically, there is a reduction of extension (there is a separate feature for radio-ulnar synostosis . Check radiologically.
	Hyperextensibility at elbow	A common consequence of joint laxity. Check that a parent (or both) are not affected.
	Limited movement/flexion deformity of elbow	Check reason. If radio-ulnar synostosis , consider using together
	Webbing at elbow	Pterygium at elbow. If multiple add other sites as separate features.
Forearm, general abnormalities	Bowed radius	Some hypoplastic radii are bowed. Use both together where appropriate.
	Bowed ulna	If also hypoplastic ulna , use in combination where appropriate
	Hypoplastic or absent radii	A small, poorly formed radius.
	Hypoplastic or absent ulna	An absent, or short, malformed ulna.

Body System (and feature path)	Feature	Useful comments aiding choice of feature
UPPER LIMBS (continued)		
Forearm, general abnormalities (continued)	Long ulna	Especially the distal part relative to the radius - a radiological feature.
	Madelung deformity	There is a disproportionate growth between the radius and ulna leading to a dinner fork deformity at the wrist (there is a short bowed radius and a dorsal distal dislocation of the ulna).
	Radio-ulnar synostosis	An X-ray diagnosis of bony fusion between radius and ulna.
	Restriction of supination/pronation	The restriction is felt on rotation of the elbow in a flexed position. There might be an underlying radio-ulnar synostosis or radio-humeral synostosis , in which case add in same box or as additional code.
Humerus, general abnormalities	Bifid distal humerus	A radiological finding (occasionally it can be felt, or seen as an extra knob at the elbow). The distal end divides in two.
	Bowed humerus	If bowed (bent) use this. If bowed and short use hypoplastic humerus and possibly this feature in the same box.
	Hypoplastic or absent humerus	Small, malformed humerus.
Shoulder joint, general abnormalities	Dislocation of shoulder	Use for single dislocation. If more than one is involved use multiple joint dislocation as well .
	Glenoid hypoplasia	The glenoid fossa is poorly formed and shallow (where the proximal humerus articulates with the scapula).
	Limited movement of shoulder	Look for underlying cause before using this feature (see for example synostosis of shoulder joint)
	Synostosis of shoulder joint	Bony fusion between humerus and scapula, causing limited movement.
	Webbed axilla	Pterygium at axilla. If multiple add other sites as separate features
Upper limbs, general abnormalities	Absent upper limbs	Amelia. Total absence of a limb. Use in with reduction deformity of the arms-no digits , as the degree of the defect might be variable.

Body System (and feature path)	Feature	Useful comments aiding choice of feature
UPPER LIMBS (continued)		
Upper limbs, general abnormalities (continued)	Acromelia of upper limbs	Acromelia implies a distal segment (hand and fingers) shortening (not a reduction deformity as all the digital structures are present).
	Asymmetric arms	A difference in length or circumference between the two arms (check for other specific body part asymmetries).
	Constriction rings of arms	Constriction rings are bands of tissue causing an indentation in the arm, with rapid, distal, tapering of the limb. The limb might end beyond the ring, or continue with varying degrees of hypoplasia.
	Hypertrophy of upper limb	There is asymmetry due to an enlarged limb. The hypertrophied tissue might extend to include underlying bone.
	Mesomelia of upper limbs	A shortening of the middle segment, i.e., the area from the elbow to the wrist.
	Proportionate short arms	Use with caution as an X-ray might show a short humerus, or radius or ulna. See separate features for hypoplastic of humerus , hypoplastic of radius or hypoplastic of ulna . Use together when necessary.
	Reduction deformity of arms, no digits	Terminal transverse defects, without digits. If uncertain use with constriction rings of arms .
	Reduction deformity of arms, some digits	Will include 'phocomelia'. There is a reduction in length with an attempt to form digits.
	Rhizomelia of upper limbs	Shortening of the proximal segment (the distance between the top of the arm and the elbow) of the arms.
Wrist, general abnormalities	Dislocated wrist	Alone or in conjunction with dislocations elsewhere (in which case add other codes in separate boxes).
	Flexion deformity at wrist	The wrist is in fixed flexion - seldom without the fingers being in the same position. Might be part of multiple contractures .
	Hyperextensibility at wrist	Mostly part of joint laxity . Use the latter or the two together.

Body System (and feature path)	Feature	Useful comments aiding choice of feature
UPPER LIMBS (continued)		
Wrist, general abnormalities (continued)	Limited movement of wrist	Wrist not fixed in flexion but decreased movement in all directions. Check for underlying pathology.
URINARY SYSTEM		
Bladder, general abnormalities	Absent bladder	Agensis or absent. In severe hypoplasia of the bladder, the ureters open into the urethra.
	Bladder diverticulae	Gaps or weakness in the bladder wall through which the mucosa protrudes. Often, but not always, secondary to bladder neck obstruction.
	Bladder exstrophy	The bladder opens widely onto the surface of the abdomen. The mucosa of the pelvic urethra is exposed.
	Bladder fistulae	A communication between the bladder and a nearby organ.
	Bladder stones	Any type of calculus. There is a separate feature for renal stones . Use together if necessary.
	Exstrophy of the Cloaca	The exterior bladder (it opens onto the abdominal wall) is divided into two halves, between which there is intestinal mucosa (mostly the ileum). The colon and appendix might also be seen and the penis or clitoris are in two widely separated halves.
	Large bladder	Mostly the end result of outflow obstruction. Includes a hypertrophied bladder.
	Patent urachus	During early fetal life the urachus extends from the umbilicus to the dome of the bladder. Its persistence results in a diverticulum or fistula.
Kidneys, general abnormalities	Agensis/absent kidney	An absent (or near absent) kidney.
	Dysplastic kidneys	The formation of abnormal nephrons and mesenchymal stroma. Use for 'cystic dysplasia'

Body System (and feature path)	Feature	Useful comments aiding choice of feature
URINARY SYSTEM (continued)		
Kidneys, general abnormalities (continued)	Ectopic/supernumerary kidneys	Ectopy is an abnormal positioning of the kidney. It might be pelvic or thoracic in its displacement. Supernumerary kidneys may have a separate ureter and kidney
	Horseshoe kidneys	There is fusion in the midline (inferiorly) resulting in the horse-shoe shape. In lateral fusion one pelvicalyceal system, crosses the midline to drain a portion of both kidneys.
	Hydronephrosis	Fluid in, and expansion of, the renal pelvis.
	Large kidneys	Renal length for age is greater than 2SD above the mean. In newborn - 4-6cm is normal By 3 years 6-10cm. These are large, normal looking kidneys.
	Multiple renal cysts	In infantile polycystic disease the kidneys are enlarged. The renal cysts are visible on the surface and are formed by the ends of the ectatic collecting ducts. Do not use for 'cystic dysplastic' kidneys, or alternatively use with dysplastic kidneys in the same box. In medullary cystic disease the cysts are mainly in the medulla. This is included here.
	Nephritis or nephropathy	Glomerulonephritis, nephronophthisis, etc.
	Renal artery stenosis	Alone or as part of more widespread disease.
	Renal stones/calcification	There is a separate code for bladder stones . It might be better to use both together. Use this code for nephrocalcinosis.
	Renal tumours (including Wilms')	Any tumour including nephroblastomatosis.
	Single renal cysts	These are simple cysts, thin walled and mostly unilocular. They are usually situated in the cortex of the kidney.
	Small kidneys	Small, normal looking kidneys. There is a separate code for dysplastic kidneys .
Renal Function, general abnormalities	Abnormal urinary porphyrins	Abnormal levels of porphyrins in urine

Body System (and feature path)	Feature	Useful comments aiding choice of feature
URINARY SYSTEM (continued)		
Ureters, general abnormalities	Dilated ureters/ureteral atresia	Absent, or dilated for whatever reason.
	Double ureters	Mostly an incomplete duplication in that the kidney is drained by two renal pelvices and ureters which then unite before they reach the bladder. If in conjunction with a more extended duplication use with ectopic/supernumerary kidney
	Urinary reflux	A backward flow of urine. Not a good dysmorphic sign.
Urethra, general abnormalities	Urethral diverticulae	This may present with a swelling on the anterior part of the penis, or with urinary obstruction. A diverticulum is a sac, a herniation of the wall.
	Urethral fistulae	Communication with the rectum or the bowel. It is seen with ano-rectal obstructive lesions.
	Urethral stenosis/atresia	This includes meatal stenosis and in males congenital strictures in the prostatic urethra. It includes urethral diaphragms,
	Urethral valves	Posterior urethral valves (valvular folds of the urethral mucous membrane) leading to obstruction and marked dilatation.
Urinary tract, general abnormalities	Abnormal urinary colour	Excludes porphyrins and myoglobin.
	Abnormal urinary odour	An unusual smell - some clinicians are better at this than others.
	Aminoaciduria	Of all types. If you are unsure if it is an aminoaciduria or an organicaciduria , use both in same box.
	Glycosuria	Note also hyperglycemia under endocrine/pancreas
	Haematuria	Blood in urine - look for underlying pathology. Note that there is a separate code for myoglobinuria
	Hypercalciuria	Note also under endocrine/parathyroids is hypercalcaemia
	Mucopolysacchariduria/oligosacchariduria	Specific for these storage disorders.
Multiple urinary infections	Not a helpful feature. If there is an underlying structural defect, rather use that as a feature.	

Body System (and feature path)	Feature	Useful comments aiding choice of feature
URINARY SYSTEM (continued)		
Urinary tract, general abnormalities (continued)	Organicaciduria	If you are not sure whether the substance is an organic aciduria or an aminoaciduria , use both in same box.
	Polyuria	Increased frequency of micturation.
	Proteinuria	Use with nephritis or nephropathy or if there is no underlying cause known.
	Pyuria	An infected urine. Not very useful.
	Renal tubular acidosis	Fanconi syndrome.
	Urinary porphyrins	An increase in any of the urinary porphyrins.
VOICE		
Voice, general abnormalities	Cat cry/weak, high-pitched cry	Also called a 'cerebral cry' as it is often indicative of brain damage. The cry is high pitched and present only during infancy. See separate feature for high-pitched voice .
	High-pitched voice	Some octaves higher than expected for sex and age.
	Hoarse voice	A gruff, low-pitched, coarse voice.
	Low-pitched voice	A voice with a pitch in the lowest ranges.
	Nasal speech	The escape of air through a cleft, or an incomplete closure of the posterior pharyngeal space, causes the voice to have a nasal quality (as if talking through the nose with the mouth shut).
	Speech defect/dysarthria	Any of the following: slurring speech, staccato speech, stutter etc. It is an articulation defect.
	Speech delay	Speech delay is common in developmental delay/mental retardation of all types. Use developmental delay /mental retardation rather than speech delay if the delay is more global.



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