

Skull/cranial anomalies	
	Basillary impression/invagination/cranio-spinal malformation/platybasia
	Bifid skull
	Broad forehead
	Craniostenosis/craniosynostosis/sutural synostosis
	Brachycephaly/flat occiput
	Cloverleaf skull
	Dolichocephaly/scaphocephaly
	Plagiocephaly
	Trigonocephaly
	Dense/thickened skull/calvarium/cranial/facial hyperostosis
	Frontal bossing/prominent forehead
	High forehead
	Large fontanelle/delayed fontanelle closure
	Low hair line-front
	Macrocephaly/macrocrania/megalocephaly/megacephaly
	Microcephaly
	Narrow forehead
	Poorly ossified skull/calvarium
	Prominent metopic suture
	Prominent occiput/occipital bossing
	Scalp/skull defect
	Sloping forehead
	Turricephaly/oxycephaly/acrocephaly
	Wormian bones
Face/facial anomalies	
	Facial cleft
	Lateral facial cleft
	Midline facial cleft
	Facial dysmorphism
	Broad cheeks/cherub-like/cherubin face
	Claudication/pain on mastication/while chewing
	Coarse face
	Deepset eyes/enophthalmos
	Dental malocclusion
	Depressed premaxillary region/midface

Face/facial anomalies (continued)	
	Facial dysmorphism (continued)
	Downslanted palpebral fissures/anti-mongoloid slanting palpebral fissures
	Enlargment of jaw/large jaw
	Expressionless face/amimia
	Facial palsy
	Facial structural asymmetry/facial hemiatrophy/facial hemihypertrophy
	Fine/elfin-like face
	Flat cheek bones/malar hypoplasia
	Flat face
	Flat supraorbital ridge
	Grooved/dimple chin
	High cheek bones
	Hypertelorism
	Hypoplastic mandibula/partial absence of the mandibula
	Hypoplastic maxillary bones/zygomatic bones/maxillary hypoplasia
	Hypotelorism
	Large face
	Limited opening of the mouth
	Long face
	Long midface
	Micrognathia/retrognathia/micrognathism/retrognathism
	Mid-facial hypoplasia/short/small midface
	Narrow face
	Pointed chin
	Prognathism/prognathia
	Prominent premaxillary region/midface
	Prominent supraorbital ridge
	Proptosis/exophthalmos
	Round face
	Sharp-shaped face
	Tags on cheeks
	Triangular face
	Upslanted palpebral fissures/mongoloid slanting palpebral fissures

Anomalies of eyes and vision	
	Abnormal ERG/electroretinogram/electroretinography
	Abnormal eye movements/oculomotor disorder
	Nystagmus
	Oculomotor apraxia/dyspraxia
	Ophthalmoplegia/ophthalmoparesis/oculomotor palsy
	Strabismus/squint
	Abnormal VEP/Visual evoked potential
	Abnormal visual field/hemianopsia/hemianopia/scotoma/visual peripheral rim
	Achromatopsia/dyschromatopsia/daltonism/impaired colour vision
	Aniridia/iris hypoplasia
	Anophthalmos/anophthalmia/microphthalmos/microphthalmia
	Anterior chamber anomaly
	Aphakia/microphakia/spherophakia/biphakia/absence of lens/lenticone/lentiglobus
	Astigmatism
	Blue sclerae
	Brushfield spots
	Buphthalmos
	Cataract/lens opacification
	Choroidal anomalies/atrophy/choroideremia
	Chronic uveitis/blepharitis/episcleritis/scleritis/conjunctivitis/keratitis
	Coloboma of iris
	Coloboma of the lens
	Congenital alacrimia
	Conjunctival injection/chemosis/red eye
	Conjunctival telangiectasia
	Conjunctival tumor/epibulbar dermoid
	Corneal clouding/opacity/vascularisation
	Corneal dystrophy
	Corneal ulceration/perforation
	Cyclopia
	Diplopia/double vision
	Embryotoxon
	Flat cornea
	Flitting flies/myiodesopsia/myodesopsia
	Fragility of the eye globe
	Glaucoma

Anomalies of eyes and vision (continued)	
	Heterochromia/mixed colouring of iris
	Hypermetropia
	Iridocele
	Iridocorneal dysgenesis/iridogoniodysgenesis
	Iridocorneal/iridolenticular synechiae
	Iris albinism/ocular albinism
	Iris melanosis/ocular melanosis
	Keratoconus/keratoglobus
	Lens dislocation/luxation/subluxation/ectopia lentis
	Lisch nodules/iris hamartomas
	Macular coloboma
	Macular pigmentary anomaly/cherry-red spot
	Megalocornea
	Microcornea
	Mild visual loss/impaired visual acuity
	Myopia
	Night blindness/hemeralopia
	Optic nerve anomaly/optic atrophy/anomaly of the papilla
	Coloboma of the optic nerve
	Photophobia
	Pterygion
	Pupillary anomalies/mydriasis/myosis/tonic pupil
	Retinal albinism
	Retinal detachment
	Retinal hamartoma
	Retinal vascular anomalies/retinal telangiectasia
	Retinopathy
	Macular dystrophy/absence/hypoplasia of the macula
	Retinal/chorioretinal dysplasia/dystrophy
	Retinitis pigmentosa/retinal pigmentary changes
	Retinoschisis/retinal/chorioretinal coloboma
	Sclerocornea
	Transient amaurosis/acute visual trouble
	Visual loss/blindness/amblyopia
	Vitreous anomalies/hyalitis/persistent vitreous vascularisation
	Xerophthalmia/dry eyes

Eyebrows anomalies

- Absent/decreased/thin eyebrows
- Darker than expected eyebrows
- Flared eyebrows
- High arched eyebrows
- Lateral thinning of eyebrows
- Synophris/synophrys
- Thick/bushy eyebrows

Anomalies of eyelids,eyelashes and lacrimal system

- Absent/decreased lashes
- Blepharitis/eyelid inflammation
- Blepharophimosis/short palpebral fissures
- Coloboma of the eyelid
- Cryptophthalmia/ankyloblepharon/synblepharon
- Defect/anomaly of lacrymal system
- Dystichiasis/double row of lashes
- Ectropion/entropion/eyelid eversion
- Epicanthic folds
- Euryblepharon/wide palpebral fissures
- Lagophthalmia/partial palpebral occlusion
- Long/thick/curved lashes/trichomegaly/polytrichia
- Microblepharon/ablepharon/ablephara/eyelid absence
- Palpebral edema/periorbital edema
- Ptosis
- Puffy eyelids
- Telecanthus/canthal dystopy

Anomalies of nose and olfaction

- Absent nasal cartilage/hypermobile nose
- Anosmia/cacosmia/hypogeusia/hyposmia/olfactory bulb hypoplasia
- Anteverted nares/nostrils
- Beaked nose
- Bifid tip/cleft nose/supernumerary nose
- Broad nasal root
- Broad nose/nasal bridge
- Choanal atresia
- Depressed nasal bridge
- Flared/thick ala nasi

Anomalies of nose and olfaction (continued)

Flattened nose
Frontal sinus agenesis/anomaly
High nasal bridge
Long/large/bulbous nose
Narrow nasal bridge
Narrow nasal root
Nasal atrophy/hypoplasia/arhinia/rudimentary nose
Nasal chondritis/nose chondritis
Nasal congestion/sinusitis/rhinitis/rhinorrhea
Nasal polyposis
Nasal septum deviation
Nasal/sinus obstruction/apertura pyriformis hypoplasia/stenosis
Short columella/depressed nasal tip
Short/small nose
Single nare/abouphalia
Small/triangular nares/nostrils
Thick columella
Thin/hypoplastic ala nasi

Anomalies of mouth, lip and philtrum

Cleft lip
Cleft lip and palate
Lateral cleft lip/gingival cleft/paramedian nasal cleft
Median cleft lip
Downturned mouth
Everted lower lip
Exaggerated cupid bows
Fusionned lips
Lip hyperpigmentation
Lip pits/fistulae
Long philtrum
Macrostomia/big mouth
Microstomia/little mouth
Mouth dryness/xerostomia
Mouth held open
Oral mucosa disease/cheilitis
Philtrum deeply grooved
Philtrum flat/large/featureless/absent cupidon bows

Anomalies of mouth, lip and philtrum (continued)	
	Protruding lips
	Short philtrum
	Tented upper lip
	Thick lips
	Thin/retracted lips
Anomalies of tongue, gingiva and oral mucosa	
	Abnormal pigmentation of the oral mucosa/gingivae
	Ankyloglossia/lingual synechiae
	Broad alveolar ridge
	Cleft/notched/bifid tongue
	Fissured/scrotal tongue
	Gingivitis
	Glossitis
	Glossoptosis
	Hamartoma/tumefaction of the tongue/gingivae/oral mucosa
	Macroglossia/tongue protrusion/proeminent/hypertrophic
	Microglossia/aglossia/hypoglossia/tongue hypoplasia
	Oral synechiae / abnormal frenulae
	Thickened/hypertrophic/fibromatous gingivae
Palate anomalies	
	Cleft palate without cleft lip/submucosal cleft palate/bifid uvula
	High vaulted/narrow palate
	Hypoplastic/aplastic uvula
	Immobile soft palate/soft palate anomaly
	Palate exostoses/torus palatinus
	Short palate
Anomalies of teeth and dentition	
	Abnormal dentition/dental position/implantation/unerupted/dental ankylosis
	Abnormal erosion/resorption of teeth/odontolysis
	Alveolysis/paraodontitis
	Anodontia/oligodontia/hypodontia
	Cementum anomaly/hypercementosis
	Complete/partial macrodontia
	Complete/partial microdontia
	Delayed dentition/eruption of teeth/lack of eruption of teeth
	Dental cysts/tumours

Anomalies of teeth and dentition (continued)	
	Dental staining anomaly/spotted teeth/erythrodonia
	Dentine anomaly
	Enamel anomaly
	Multiple caries
	Premature eruption of teeth/natal teeth
	Premature lost of deciduous teeth
	Solitary median incisor
	Supernumerary teeth/polyodontia
	Taurodontia
	Tooth shape anomaly
Anomalies of ear and hearing	
	Branchial archs anomalies
	Branchial/posterior auricular/preauricular/cheek cysts/fistulae
	Preauricular/branchial tags/appendages
	Chronic/relapsing otitis
	External ear anomalies
	Antihelix anomaly
	Antitragus abnormal
	Bifid/cleft ear lobe/ear lobe pits
	Calcified/rigid ear cartilage
	Ear cartilage deficiency
	Ear lobe/pinnae telangiectasia
	External auditory canal atresia/stenosis/agenesis
	Folded helix
	Helix thickened/sculpted
	Helix/crux helix absent/abnormal/adherent/crux cymbae/posterior helix pits
	Long/large ear
	Low set ears/posteriorly rotated ears
	Microtia/cryptomicrotia/antia/external auditory canal/pinnae aplasia/hypoplasia
	Pointed helix/Elfin ear
	Polyotia
	Prominent/bat ears
	Round ear
	Small/hypoplastic/adherent/absent ear lobe
	Synotia

Anomalies of ear and hearing (continued)	
	External ear anomalies (continued)
	Thick/wide ear lobe
	Tragus abnormal/absent
	Functional anomalies of the inner ear
	Dizziness
	Hyperacusia
	Tinnitus
	Hearing loss/hypoacusia/deafness
	Central deafness/hearing loss
	Conductive deafness/hearing loss
	Sensorineural deafness/hearing loss
	Pinnae chondritis
	Structural anomalies of inner ear/cochlea/vestible/semicircular canals
	Structural anomalies of middle ear/ossicles/tympanic cavity
Anomalies of the neck	
	Branchial fistulae of the neck
	Congenital torticollis
	Cystic hygroma
	Excess nuchal skin without pterygium colli
	Short neck
	Webbed neck/pterygium colli
Anomalies of chest/thorax/trunk	
	Abnormally placed nipples
	Absent pectoral muscles
	Anomalies of the ribs
	Rib number anomalies
	Rib structure anomalies
	Asymmetric rib cage/thorax
	Breast tissue/mammary gland absence/aplasia
	Clavicle absent/abnormal
	Flared chest/bell-shaped thorax/shield chest
	Gynecomastia/breast/mammary gland enlargement/hyperplasia
	Hypoplastic/absent nipples
	Long rib cage/thorax
	Narrow rib cage/thorax
	Narrow/sloping shoulders

Anomalies of chest/thorax/trunk (continued)	
	Short rib cage/thorax
	Sternal/sternum anomalies
	Pectus carinatum
	Pectus excavatum
	Supernumerary nipples/polythelia
	Wide rib cage/thorax
Anomalies of spine, vertebrae and pelvis	
	Abnormal vertebral size/shape
	Epiphyseal vertebral anomaly
	Lack/delayed ossification of spine/vertebrae
	Odontoid hypoplasia
	Platyspondyly
	Punctated vertebrae
	Camptocormia
	Coxa profunda/acetabular protrusion
	Intervertebral disk anomaly
	Intervertebral disk calcification
	Intervertebral disk herniation
	Kipphosis
	Lordosis
	Pelvis anomaly/Narrow/broad iliac wings/pubis abnormality
	Rachidian/spine canal stenosis
	Rigid spine
	Sacrococcygeal teratoma
	Sacro-coccyx/sacrum anomaly
	Sacral sinus/dimple
	Sacrococcyx agenesis
	Sacrum/sacral duplication
	Sacroiliac joints anomalies
	Scoliosis
	Spina bifida occulta
	Straight spine
	Vertebral segmentation anomaly/hemivertebrae

Anomalies of the abdominal wall	
	Absent/hypotonic/flaccid abdominal wall muscles
	Celosomia
	Gastroschisis
	Herniae
	Inguinal/inguinoscrotal/crural hernia
	Umbilical hernia
	Omphalocele/exomphalos
	Patent urachus/fistula of the urachus
Global upper and lower limbs anomalies	
	Amelia/tetramelia/hemiamelia
	Arthrogryposis
	Cutaneous/amniotic bands/webbing of joints
	Popliteal web
	Ectromelia/absence of one or more limb segments
	Aphalangia/hands and feet phalangeal bones absence/hypoplasia/aplasia
	Phocomelia
	Limb duplication/dimelia/ulnar/fibular dimelia/diplopodia
	Long limbs/dolichostenomelia
	Short limbs/micromelia/brachymelia
	Mesomelic micromelia
	Rhizomelic micromelia
Upper limb segmental anomalies	
	Anomalies of hands
	Abnormal dermatoglyphics
	Absence of palmar creases
	Camptodactyly of fingers
	Camptodactyly of some fingers
	Complete claw hand/camptodactyly of all fingers
	Claw hand/retracted fingers
	Clinodactyly of fifth finger
	Clinodactyly of fingers 1,2,3,4/overlapping fingers
	Deep palmar creases
	Flat palm
	Hand agenesis/absence
	Irregular length/shape of fingers
	Large hand

Upper limb segmental anomalies (continued)	
Anomalies of hands (continued)	
	Long hand/arachnodactyly
	Macroactyly/fingers hypertrophy/megalodactyly (hand)
	Metacarpal anomalies/Archibald's sign
	Oligodactyly/ectrodactyly of fingers
	Radial club hand
	Short hand/brachydactyly
	Simian crease/transverse/unique palmar crease
	Small hand/acromicria
	Spindle shaped fingers
	Symphalangy of fingers
	Syndactyly of fingers/interdigital palm
	Tapered fingers
	Terminal/third phalangeal bone of fingers broadened/deviated
	Terminal/third phalangeal bone of fingers hypoplasia
	Thick/dowel fingers
	Thumb anomalies (excluding hypoplasia)
	Broad/bifid thumb
	Clasp thumb/thumb adduction/distal thumb phalangeal bone deviated/large
	Fingerlike/triphalangeal thumb
	Proximally set thumb
	Thumb ankylosis
	Thumb duplication/distal bifid thumb phalangeal bone
	Thumb hypoplasia/aplasia/absence
	Trident hand/split hand/abnormal median ray
	Ulnar deviation of fingers
	Upper limb polydactyly/hexadactyly
	Postaxial polydactyly (hand)
	Preaxial polydactyly (hand)
Elbow anomalies(excluding luxation)	
	Cubitus valgus
	Humeroradial fusion
	Humeroulnar joint anomaly
	Radioulnar synostosis
Humerus anomaly/absence/agenesis/hypoplasia/congenital humerus varus	
Radius anomaly/absence/agenesis/hypoplasia/abnormal radial ray	

Upper limb segmental anomalies (continued)	
	Scapula structural/position anomaly/congenital elevation/Sprengel anomaly
	Ulnar/cubital anomaly/absence/agenesis/hypoplasia/abnormal ulnar/cubital ray
	Upper limb asymmetry/hemiatrophy/hemihypertrophy
	Upper limb transverse anomaly (excluding hand)
	Wrist/carpal anomalies
	Carpal bones fusion/synostosis
	Madelung's deformity
Lower limb segmental anomalies	
	Ankle anomalies
	Talipes-valgus
	Talipes-varus/metatarsal varus
	Femur anomaly/absence/agenesis/hypoplasia/bifurcation
	Fibula anomaly (excluding short)/absence/agenesis/hypoplasia/fibular ray anomaly
	Foot anomalies
	Abnormal sole/deep creases
	Big toe anomaly (excluding absence)
	Broad/bifid big toe
	Dorsiflexed great toe
	Hallux valgus
	Long/prehensile big toe
	Metatarsus adductus
	Short big toe
	Broad foot
	Claw toes/retracted toes
	Clinodactyly of toes
	Distal phalangeal bones of toes hypoplasia/absence
	Flat foot
	Flexion deformity of toes (excluding big toe)/hammer toe/camptodactyly of toes
	Foot absent/apodia/adactyly/acheiropodia
	Foot hypoplasia/hypotrophy
	Irregular length/shape of toes
	Long foot/arachnodactyly of toes
	Macroactyly/megalodactyly of toes
	Malimplantation of toes
	Median ray abnormal/split foot

Lower limb segmental anomalies (continued)	
	Foot anomalies (continued)
	Oligodactyly/ectrodactyly of toes
	Pes cavus
	Pes talus
	Polydactyly of toes
	Postaxial polydactyly of toes/fifth supernumerary toe
	Preaxial polydactyly of toes/big toe duplication
	Short foot/brachydactyly of toes
	Small foot
	Syndactyly of toes
	Terminal broadening/clubbing of toes
	Wide space between 1st-2nd toes
	Knee anomalies (excluding patella)
	Genu recurvatum
	Genu valgum
	Genu varum
	Wind-swept deformity of knees
	Lower limb asymmetry/hemiatrophy/hemihypertrophy
	Lower limb transverse anomalies (excluding feet)
	Patella absent/abnormal (excluding luxation)
	Sirenomelia/mermaid/lower limb fusion
	Tarsal anomaly/fusion/synostosis
	Tibia anomaly (excluding short)/absence/agenesis/hypoplasia/tibial ray anomaly
	Tibiofibular diastasis
Anomalies of skin, subcutaneous tissue and mucosae	
	Abnormal pigmentary skin changes/skin pigmentation anomalies
	Acanthosis nigricans
	Cafe-au-lait spot
	Diffuse/generalised skin hyperpigmentation/melanoderma
	Diffuse/generalised skin hypopigmentation/cutaneous albinism
	Excessive freckling
	Irregular/in bands/reticular skin hyperpigmentation
	Irregular/patchy skin hypopigmentation
	Mongoloid spot
	Pigmented naevi/naevus pigmentosus/lentigo

Anomalies of skin, subcutaneous tissue and mucosae (continued)

Abnormal scarring/cheloids/hypertrophic scars
Acne/acnea
Anetoderma/dermal atrophy
Cellulitis/panniculitis/pseudocellulitis/inflammation of subcutaneous tissue
Chronic skin infection/ulcerations/ulcers/cancrum
Cutaneous edema
Cutaneous inflammation
Cutaneous rash
Eczema
Erysipela
Erythema/erythematous lesions/erythroderma/polymorphous erythema
Psoriasis
Urticaria
Cutis marmorata/marbled skin/livedo
Dermal/subcutaneous infiltration/induration
Dermoid sinus/dimple/pit (excluding sacral)
Dry/squally skin/exfoliation
Enanthema/aphtosa/aphta/leukoplakia
Follicular/erythematous/edematous papules/milium
Follicular/vermiculata atrophoderma
Freezing lesions
Furuncle/cutaneous abscess/hidrosadenitis suppurativa
Hairy patch
Hyperelastic skin/cutaneous hyperlaxity
Hyperhidrosis/increased sweating
Hyperkeratosis/ainhum/hyperkeratotic skin fissures
Hypohidrosis/decreased sweating/thermoregulation disorder/heat intolerance
Ichthyosis/ichthyosiform dermatitis
Lichen
Lipoatrophy
Loose skin/skin relaxation/excess skin/creases
Macules
Palmoplantar hyperkeratosis/keratoderma
Palmoplantar pustulosis
Pruritus/itching
Pustula/pustulosis
Rippled skin

Anomalies of skin, subcutaneous tissue and mucosae (continued)

	Seborrhea/hyperseborrhea/seborrheic dermatitis
	Shagreen patch
	Skin hypoplasia/aplasia/atrophy
	Skin photosensitivity
	Skin tumours/lumps/epidermal cysts
	Adenoma sebaceum
	Cutaneous neurofibromas/facial angiofibromas/Koenen tumors
	Follicular/conjunctival hamartomas
	Pedunculated skin lesions
	Subcutaneous nodules/lipomas/tumefaction/swelling
	Warts/papillomas
	Xanthomas/lipomas
	Striae
	Thick skin/pachydermia/orange skin
	Thin skin
	Tight skin/lack of elasticity
	Unusual body odour
	Vascular anomalies of skin/mucosae
	Capillary hemangioma/nevus/naevus flammeus/port-wine stain
	Cavernous/tuberous hemangioma
	Ecchymoses
	Purpura/petichiae
	Telangiectasiae of mucosae
	Telangiectasiae of the skin
	Telangiectasic erythema/poikiloderma
	Vesicles/bullous/exsudative lesions/bullous/cutaneous/mucosal detachment
	Negative Nikolski's sign
	Positive Nikolski's sign/achantolysis

Hair and scalp anomalies

	Abnormal fall of hair
	Abnormal hair texture/hair dysplasia
	Brittle hair/distrix/trichorrhexis
	Coarse/thick hair
	Fine hair
	Pili torti
	Woolly/frizzy hair

Hair and scalp anomalies (continued)

Abnormal implantation of hair
High hair line (front)/widow peak
Low hair line (back)
Albinism (hair)
Alopecia
Decreased body hair/axillar/pubic hairlessness
Decreased hair pigmentation/hypopigmentation of hair
Dense scalp hair
Hirsutism/hypertrichosis/Increased body hair
Hypersensibility of scalp/scalp tenderness
Hypotrichosis/atrichia/atrichiasis/scalp hairlessness
Lanugo
Premature greying of hair
Scalp cyst/giant nevus
slow growth of the hair
Thick scalp/scalp dysplasia
White forelock/piebaldism

Nails anomalies

Abnormal fingernails
Absent/small fingernails/anonychia of hands
Dysplastic/thick/grooved fingernails
Thin/hypoplastic/hyperconvex fingernails
Ungual/paraungual fibromas (fingernails)
Abnormal nails colour/leukonychia/melanonychia
Abnormal toenails
Absent/small toenails/anonychia of feet
Dysplastic/thick/grooved toenails
Thin/hypoplastic toenails
Ungual/parungual fibromas (toenails)
Nail tumors
Onycholysis
Onyxia/paronychia/ungual inflammation
Punctate nails

Structural anomalies of the digestive tract

Anomaly of the pharynx/pharyngeal anomaly
Anomaly/ectopia/hypoplasia/atresia of salivary glands/salivary duct

Structural anomalies of the digestive tract (continued)	
	Anus/rectum anomalies
	Anus ectopia/anteponition/malposition
	Imperforate anus/rectum atresia/agenesis/recto-vaginal/vesical/perineal fistula
	Persistence of the cloaca
	Ascitis
	Esophageal anomaly
	Endobrachyesophagus/Barrett esophagus
	Esophageal diverticulum/pouch
	Esophageal duplication
	Esophageal webs
	Megaesophagus/cardiospasm/congenital dilation of the esophagus/achalasia
	Tracheo-esophageal fistula/esophageal atresia/stenosis
	Intestinal/colonic anomaly
	Colonic atresia/stenosis/absence/agenesis/hypoplasia/microcolon
	Colonic/intestinal volvulus
	Colonic/intestinal/bowel diverticulosis/diverticulitis
	Dolichocolon/megacolon/megadolichocolon/Hirschsprung's disease
	Duodenal atresia/stenosis/megaduodenum
	Intestinal atresia/stenosis/absence/agenesis/hypoplasia (excludes duodenum)
	Intestinal/bowel fistulae
	Intestinal/gut/bowel duplication
	Intestinal/gut/bowel malrotation
	Meckel diverticulum
	Mesenterium anomaly/common mesentery/mesenteric adhesion
	Polyposis of the bowel/colon/intestine
	Short bowel
	Short colon
	Situs inversus visceralis/colon/intestine trasposition/heterotaxia
	Pharyngitis
	Respiratory-digestive intersection/aero-digestive cross-roads anomaly
	Stomach/gastric anomaly
	Congenital wallet stomach/pseudo hour-glass stomach
	Gastric diverticulum

Structural anomalies of the digestive tract (continued)	
	Stomach/gastric anomaly (continued)
	Gastric duplication
	Gastric ectopia/malposition
	Gastric volvulus
	Gastric/pyloric stenosis
	Megalogastria
	Microgastria
	Structural anomaly of the peritoneum
	Tonsil anomaly/hypertrophy/adenoiditis
Functional anomalies of the digestive system	
	Acute abdominal pain/colic
	Acute diarrhea
	Constipation
	Encopresis/anal incontinence
	Epigastralgia/heartburn/gastric/duodenal ulcer/gastritis
	Eructation/aerophagia
	Esophageal varices
	Feeding disorder/dysphagia/swallowing/sucking disorder/esophageal dyskinesia
	Functional colopathy/irritable bowel syndrome
	Gastroesophageal reflux/pyrosis/esophagitis/hiatal hernia/gastroparesia
	Gastrointestinal bleeding/haemorrhage/hematemesis/melena/rectorrhagia
	Halitosis/bad breath
	Hyperperistalsis/hypoperistalsis
	Hypersialorrhea
	Hyposialorrhea
	Inflammatory intestinal disease
	Intestinal obstruction/ileus
	Intestinal perforation
	Intestinal transit disorder
	Malabsorption/chronic diarrhea/steatorrhea
	Mucous stools/acholic stools
	Nausea/vomiting/regurgitation/mercyism/hyperemesis
	Peritoneal diseases
	Peritonitis/peritoneal abscess

Structural anomalies of the liver and the biliary tract	
	Abnormal liver lobulation
	Congenital hepatic fibrosis
	Extrahepatic biliary tract atresia/obstruction
	Gallbladder/common bile duct anomalies
	Intrahepatic biliary tract atresia/obstruction
	Liver/hepatic absence/agenesis
	Polycystic liver disease/hepatic cysts
	Supernumerary liver/midline liver/liver/hepatic transposition
Functional anomalies of the liver and the biliary tract	
	Abnormal hepatic enzymes/transaminases
	Biliary/gallbladder stones/lithiasis/cholecystitis
	Cirrhosis
	Hemobilia
	Hepatitis/icterus/cholestasis
	Hepatocellular liver disease/hepatic failure
	Acute hepatic failure
	Chronic hepatic failure
	Hepatomegalia/liver enlargement (excluding storage disease)
	Liver/hepatic abscess
	Liver/hepatic steatosis
	Portal hypertension
	Storage liver disease
Structural anomalies of the pancreas	
	Annular pancreas
	Congenital absence/agenesis/aplasia/hypoplasia of the pancreas
	Congenital pancreatic cyst
	Pancreatic failure/exocrine pancreas disease
	Pancreatic fibrosis
	Pancreatic fistula
	Pancreatitis
	Supernumerary/aberrant/accesory/ectopic pancreas
Structural and functional anomalies of the spleen	
	Asplenia/polysplenia/spleen lobulation/accesory spleen
	Hypersplenism
	Splenomegaly

Structural anomalies of the respiratory system and diaphragm

Abnormal pleura/hydrothorax/pleuresia/pleural effusion/chylothorax
Absent lobe/lung/segmentation defect/abnormal lobulation/sequestration
Arterioveinous pulmonary fistulae
Diaphragmatic hernia/defect/agenesis
Diaphragmatic palsy
Hypoplastic lungs/pulmonary hypoplasia/agenesis
Laryngomalacia
Larynx/laryngeal agenesis
Larynx/laryngeal palsy
Larynx/laryngeal stenosis/atresia
Larynx/laryngeal/epiglottic/subglottic cysts
Lung fibrosis
Mediastinal anomalies
Mediastinal/hilar adenopathies
Pulmonary edema
Radiologic lung abnormalities/changes
Ground-glass/reticular/alveolar/interstitial lung/pulmonary opacity
Lung/pulmonary infiltrates
Tracheal atresia/stenosis
Tracheomalacia/tracheobronchomalacia
Tracheomegalia/tracheobronchomegalia

Functional anomalies of the respiratory system and diaphragm

Abnormal cry/voice/phonation disorder/nasal speech
Apnea/sleep apnea
Asthma/bronchospasm
Atelectasia/pulmonary collapse
Bronchial dilation/dilatation/bronchiectasia
Bronchiolitis
Bronchogenic cyst
Chronic hiccup
Chronic obstructive pulmonary disease/COPD/obstructive respiratory syndrome
Cough
Emphysema
Hemoptysis
Interstitial/restrictive pneumopathy/restrictive respiratory syndrome
Lung/pulmonary/alveolar hemorrhage/hematoma

Functional anomalies of the respiratory system and diaphragm (continued)

	Pulmonary hypertension
	Repeat respiratory infections
	Respiratory distress/dyspnea/respiratory failure/lung volume reduction
	Respiratory rhythm disorder
	Thoracic/chest pain

Structural anomalies of the cardio-circulatory system

	Aortic arches anomalies																		
	Aortic dilatation/dilation																		
	Aortic dissection																		
	Aortic root dilatation/dilation/aneurysm																		
	Arteriovenous malformations/vascular malformations (excluding port-wine stains)																		
	Cardiac diverticulum																		
	Cardiac valvulopathy																		
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	Carotid artery anomalies																		
	Congenital cardiac anomaly/malformation/cardiopathy																		
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Structural anomalies of the cardio-circulatory system (continued)

Congenital cardiac anomaly/malformation/cardiopathy (continued)	
	Hypoplastic left heart/ventricle
	Patent ductus arteriosus
	Pulmonary artery stenosis/absence/hypoplasia of the pulmonary branches
	Tetralogy of Fallot/trilogy of Fallot
	Transposition of great vessels
Dextrocardia/abnormal heart position/cardiac heterotaxia/situs inversus	
Endocardium anomalies/fibroelastosis/endocarditis	
Myocardium anomalies/myocarditis	
Pericardium anomalies/pericarditis/absence/cysts/pericardial effusion	
Peripheral arteriovenous fistula	
Pulmonary veins anomalies	
Single umbilical artery	
Visceral angiomas (excluding skin)	

Functional anomalies of the cardio-circulatory system

Acrocyanosis/Raynaud's phenomenon/vasomotor disorders	
Acute arterial hypertension/hypertensive crisis	
Acute ischemic syndrome	
	Acute ischemia of the lower limbs
	Gangrena/necrosis
	Mesenteric/intestinal infarction
Angor pectoris/myocardial infarction	
Arterial aneurism (excluding aorta)	
Arterial atheroma/precocious atherosclerosis/arteriosclerosis	
Arterial embolism/thrombosis	
Arterial pulse abolition	
Arterial rupture	
Arterial stenosis/occlusion	
Bruisability	
Cardiac conduction defect/sinoauricular/heart/auriculoventricular/branch block	
Cardiac rythm disorder/arythmia	
Chronic arterial hypertension	
Collapse/sudden death/cardiac arrest/cardiorespiratory arrest	
Edema of the legs/lower limbs	
Heart/cardiac failure	
Hypotension	

Functional anomalies of the cardio-circulatory system (continued)

	Pulmonary thromboembolism
	Varices/varicous veins/venous insufficiency
	Vascularitis/vasculitides/arteritis
	Vein of Galien dilatation/dilation
	Venous stenosis
	Venous thrombosis/phlebitis/thrombophlebitis

Anomalies of the lymphatic system

	Lymphadenopathy/polyadenopathies
	Lymphangioma/lymphatic malformations
	Lymphoedema
	Thymic aplasia/hypoplasia
	Thymic hyperplasia

Structural anomalies of the kidney and the urinary tract

	Bladder and ureter anomalies	
		Bladder/cloacal exstrophy
		Bladder/vesical diverticulum
		Cystocele
		Megaureter/hydronephrosis/pyeloureteral junction syndrome
		Ureter/calyx/pelvis duplication/bifid/retrocava/retroiliac ureter
		Ureteral stenosis/narrowing
		Ureterocele/cystic dilation/dilatation of the ureter/ureterovascular stenosis
		Urethral anomalies/stenosis/posterior urethral valves/megalocystis
		Urogenital fistulae
		Vesicorenal/vesicoureteral reflux
	Renal/kidney anomalies	
		Agensis/hypoplasia/aplasia of kidneys
		Ectopic/horseshoe/fused kidneys
		Megacalycosis
		Multicystic kidney/renal dysplasia
		Nephrosclerosis
		Polycystic kidneys
		Renal cyst (single)
		Supernumerary kidney

Functional anomalies of the kidney and the urinary tract

Abnormal colour of the urine/cholic/dark urines
Abnormal odour of the urine
Aseptic leucocyturia
Bladder inflammation/cystitis/painful/irritable bladder
Bladder/vesical lesions/glomerulations/petechiae
Cystalgia with clear urine
Hematuria/microhematuria
Nephrotic syndrome
Pollakiuria/polyuria/dysuria/anuria/acute urine retention/oliguria
Prostatic inflammation/prostatitis
Proteinuria
Recurrent urinary infections
Renal disease/nephropathy
Interstitial nephropathy
Renal failure
Renal glomerular defect/glomerulopathy
Renal tubular defect/tubulopathy
Renal/kidney calcifications
Renovascular hypertension
Urinary/renal lithiasis/kidney stones/nephrocalcinosis/nephritic colic

Structural anomalies of the genital system

Abnormal/polycystic ovaries
Ambiguous genitalia
Female pseudohermaphrodisism/virilisation/clitoridomegaly
Male pseudohermaphrodisism/lack of virilisation
Mixed gonadal dysgenesis
True hermaphrodisism
Clitoris/labia majora/labia minora/female external genitalia hypoplasia
Male internal genitalia anomaly/deferent ducts agenesis/ectopy/duplication
Penis anomalies
Corpus cavernosum sclerosis
Hypospadias/epispadias/bent penis
Macropenis/megapenis/large penis
Micropenis/small penis/agenesis
Penis duplication/diphallia/penoscrotal transposition
Phimosis/preputial adhesions/paraphimosis
Prostate atrophy/hypotrophy

Structural anomalies of the genital system (continued)	
	Scrotum abnormalities
	Bifid scrotum
	Horizontal folds on scrotum
	Hypoplastic scrotum/hemiscrotum/scrotal ridges
	Shawl scrotum
	Spermatocele/epididymal/funicular/spermatic/vaginal/albugineal cyst
	Testis anomalies
	Funicular/testicular torsion/absent gubernaculum testis
	Macroorchidism/macrotestes
	Small/atrophic/hypoplastic testes/monorchidia/macrorchidia
	Undescended/ectopic testes/cryptorchidia/unfixed testes
	Urogenital sinus
	Uterine/uterus/vaginal anomalies
	Vagina anomalies/atresia/hydrometrocolpos/hymen imperforation
	Vaginal hydrocele/hematocele/pyocele/lymphocele/varicocele
Functional anomalies of the genital system	
	Dyspareunia/coital pain/vaginal dryness
	Genital prolapse
	Impotence/painful erection/priapism/erection troubles
	Metrorrhagia/menorrhagia/hemorrhagic cycles/hyper/poly/spanio/dysmenorrhea
	Penile induration
	Sterility/hypofertility
	Azoospermia/oligospermia/asthenospermia
	Testes inflammation/orchitis
	Uterine rupture
	Vaginal spotting
Anomalies of the endocrine glands	
	Adrenal glands anomalies
	Cortico-adrenal hyperplasia/hypersecretion
	Cortico-adrenal hypoplasia/insufficiency
	Hyperaldosteronism
	Hypoaldosteronism

Anomalies of the endocrine glands (continued)	
	Anomaly of pancreatic hormones
	Diabetes mellitus
	Hyperinsulinism/hyperinsulinemia
	Insulin-dependent/type 1 diabetes
	Insulin-independent/type 2 diabetes
	Resistance to insuline
	Hypoglycemia
	Galactorrhea
	Hypothalamic-hypophyseal axis anomalies/hypothalamus/pituitary anomalies
	Diabetes insipidus
	Hypothalamic-hypophyseal axis functional anomalies/hypopituitarism
	Late puberty/hypogonadism/hypogenitalism
	Parathyroids anomalies
	Hyperparathyroidy
	Hypoparathyroidy
	Precocious menopause/secondary amenorrhea
	Precocious puberty
	Primary amenorrhea
	Thyroid anomalies
	Ectopic/agenesis/dysgenesis/hypoplastic thyroid
	Goiter
	Hyperthyroidy
	Hypothyroidy
	Thyroiditis
Structural anomalies of the nervous system/of the brain	
	Agyria/micro/pachy/macrogryria/lissencephaly/gyration/neuronal migration defect
	Cerebellum/cerebellar vermis anomaly/agenesis/hypoplasia
	Cerebral vascular anomalies
	Corpus callosum/septum pellucidum total/partial agenesis
	Cortical atrophy without hydrocephaly/cerebral hemiatrophy/subcortical atrophy
	Cranial nerve anomalies
	Dilated cerebral ventricles without hydrocephaly
	Dural ectasia/
	Holoprosencephaly/arhinencephaly/unique lateral ventricle
	Hydranencephaly

**Structural anomalies of the nervous system/of the brain
(continued)**

Hydrocephaly							
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Intracranial/cerebral calcifications							
Megalencephaly							
Midbrain/brainstem/pons/medulla anomalies							
Neural tube defect							
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Meningocele							
Myelomeningocele							
Spina bifida							
Porencephaly							
Schizencephaly							
Septo-optic dysplasia							
Syringomelia							
Thalamic/hypothalamic defect							

Functional anomalies of the nervous system

Alexia/agraphia/writing/reading troubles
Central neuropathy
Cranial hypertension
Cranial nerves palsy
Dysautonomia/autonomous nervous sytem anomalies
EEG anomalies
Elocution disorders/dysarthria/dysphonia
Encephalitis
Facial pain/cephalalgia/migraine
Hyperactivity/attention deficit
Incessant startle response to noise
Intellectual deficit/mental/psychomotor retardation/learning disability
Meningitis/meningeal syndrome

Functional anomalies of the nervous system (continued)	
Motor deficit/trouble	
	Abnormal gait
	Acute palsy
	Areflexia/hyporeflexia
	Ataxia/incoordination/trouble of the equilibrium
	Execution movement disorder/dysmetria/bradykinesia/akinesia/apraxia
	Extrapyramidal syndrome
	Hemi/para/di/quadruplegia/mono/para/hemiparesia/limb palsy
	Hypereflexia
	Hypertonia/spasticity/rigidity
	Hypotonia
	Movement disorder
	Chorea/athetosis/choreoathetosis/choreic syndrome
	Dystonia/torticollis/writer's cramp/blepharospasms
	Hemiballism/unilateral movement disorders
	Hyperkinesia/dyskinesia
	Myoclonus/fasciculations
	Tics/stereotypias
	Tremor
	Pyramidal syndrome
Myasthenia	
Nerve conduction abnormality	
Neuritis/polyneuritis/multineuritis	
Peripheral neuropathy	
Pseudobulbar signs/spasmodic laugh and cry	
Psychic/behavioural troubles	
	Anorexia
	Autism/autistic disorders
	Auto-aggressivity/auto-mutilation
	Bulimia/hyperphagia
	Delirium/hallucination
	Humour troubles/anxiety/depression/apathy/euphoria/irritability
	Obsessive-compulsive disorder
	Psychosis/schizophrenia/maniac disorder
Psychic/psychomotor regression/dementia/intellectual decline	
Retrolbulbar optic neuritis	
Seizures/epilepsy/absences/spasms/status epilepticus	

Functional anomalies of the nervous system (continued)	
	Sensitive trouble/deficit
	Hyperesthesia/allodynia/hyperalgia
	Insensitivity to pain
	Paresthesia/dysesthesia/hypoesthesia/anesthesia/numbness
	Sleep and vigilance disorders
	Insomnia
	Obnubilation/coma/lethargia/desorientation
	Somnolence/hypersomnia/parasomnia
	Speech troubles/aphasia/dysphasia/echolalia/mutism/logorrhea/dysprosodia
	Sphincter dysfunction
	Transient cerebral ischemia/stroke
	Trigeminal neuralgia
	Troubles of memory/amnesia/hypermnesia
	Visual/auditory/tactile agnosia/propopagnosia/simultagnosia
Muscle anomalies	
	Abnormal EMG/electromyogramm/electromyography
	Abnormal muscle biopsy/muscle enzymes/CPK/LDH/aldolase/creatin phosphokinase
	Contractures/cramps/trismus/tetania/claudication/opisthotonos
	Fasciitis
	Muscle hypertrophy
	Muscle hypotrophy/atrophy/dystrophy/agenesis/amyotrophy
	Muscle ossification
	Muscle weakness/flaccidity
	Muscular edema
	Muscular sclerosis
	Myalgias/muscular pain
	Myopathy
	Myositis
	Myotonia
Anomalies of bones/skeletal anomalies	
	Abnormal/absent ossification
	Advanced bone age
	Bone ankylosis
	Bone cyst
	Bone pain
	Bone tumefaction/swelling

Anomalies of bones/skeletal anomalies	
	Bone/osseous hyperplasia
	Bone/osseous hypoplasia
	Delayed bone age
	Diaphyseal anomaly
	Bowed diaphysis/diaphyses/long bones
	Cortical anomaly/thick bone cortical layer
	Enlarged diaphysis/diaphyses
	Narrowed/gracile diaphysis/diaphyses/long bones/dolichostenomelia
	Dysostosis/chondrodysplasia/osteodysplasia/osteochondrosis/skeletal dysplasia
	Enchondroses
	Epiphyseal anomaly
	Cone epiphyses/epiphysis
	Punctate epiphyses/epiphysis
	Exostoses
	Fibromatosis/bone fibroma
	Hyperostosis
	Metaphyseal anomaly
	Mutiple fractures/bone fragility
	Osteochondrite/ostechondrosis
	Osteolysis/osteoclasia/bone destruction/erosions
	Osteomyelitis/osteitis/periostitis/spondylodisciitis
	Osteonecrosis/bone infarction
	Osteosclerosis/osteopetrosis/bone condensation
	Ostoporosis/osteopenia/demineralisation/osteomalacia/rickets
	Rough trabeculation of bone
	Synostosis
Anomalies of cartilages, joints and periarticular tissue	
	Arthritis/synovitis/synovial proliferation
	Arthrosis
	Articular click
	Articular/joint pain/arthralgia
	Calcification of cartilages/intraarticular calcification
	Cartilage destruction/chondrolysis
	Chondritis/cartilage inflammation
	Enthesis anomaly/inflammation/enthesitis/enthesopathy
	Hydrarthrosis/articular/joint effusion

Anomalies of cartilages, joints and periarticular tissue (continued)

	Hyperextensible joints/articular hyperlaxity
	Joint dislocation/subluxation
	Elbow dislocation
	Hip dislocation/dysplasia/coxa valga/coxa vara/coxa plana
	Patella dislocation
	Shoulder dislocation
	Joint/articular deformation
	Musculo-tendinous retractions
	Periarticular tissue anomaly/extraarticular calcifications
	Restricted joint mobility/joint stiffness/ankylosis
	Sprain/twisted joint
	Tendon rupture/tendinitis/bursitis/tenosynovitis

Anomalies of the immunitary system

	Autoimmunity/autoimmune reaction/autoantibodies
	Cryoglobulinemia
	Hypergammaglobulinemia
	Immunodeficiency/increased susceptibility to infections/recurrent infections
	Agammaglobulinemia/hypogammaglobulinemia/B-cell deficiency
	Anomalies of complement
	Severe combined immune deficiency syndrome/SCID
	T-cell deficiency/cellular immunity deficiency
	Monoclonal immunoglobulins/gammapathy/dysglobulinemia
	Severe allergic reaction/atopy

Anomalies of the hematological system

	Bone marrow anomalies
	Bone marrow failure/pancytopenia
	Bone marrow/medullar infiltration
	Clotting/hemostasis disorders
	Blood hyperviscosity/hypercoagulability
	Hemorrhage/hemorrhagic syndrome/excessive/long-lasting bleeding
	Epistaxis/nose bleeding
	Gingivorrhagia/gingival bleeding
	Hemarthrosis
	Hematomas
	Internal hemorrhage
	Intracranial/cerebral/meningeal hemorrhage
	Mucosal/cutaneous hemorrhage

Anomalies of the hematological system (continued)																											
	Platelet disorders/thrombopathies																										
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	Carbohydrates metabolism anomalies																										
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	Hyperferritinemia/iron overload																										
	Hyperkalemia																										
	Hypernatremia																										
	Hypoammonemia																										

Metabolic anomalies (continued)	
	Hypokalemia
	Hyponatremia
	Hypoproteinemia
	Lipid metabolism anomalies
	Hyperlipidemia/hypercholesterolemia/hypertriglyceridemia
	Lipidosis/sulfatidosis
	Metabolic decompensation
	Oligoelements metabolism anomalies
	Organic acid metabolism anomalies
	Phosphocalcic metabolism anomalies
	Hypercalcemia
	Hypercalciuria
	Hyperphosphatemia
	Hypocalcemia
	Hypocalciuria
	Hypophosphatemia
	Porphyryns metabolism anomalies/porphyria
	Purine and pyrimidine metabolism anomalies
	Hyperuricemia
	Respiratory chain /mitochondrial anomalies
	Vitamin deficiency
	Vitamin A deficiency
	Vitamin B1 deficiency
	Vitamin B12 deficiency
	Vitamin B2 deficiency
	Vitamin B3/PP deficiency
	Vitamin B5 deficiency
	Vitamin B6 deficiency
	Vitamin B8 deficiency
	Vitamin B9 deficiency
	Vitamin C deficiency
	Vitamin D deficiency
	Vitamin E deficiency
	Vitamin K/K1/K2 deficiency

Neoplasms/tumors	
	Benign tumor of the brain/nervous system
	Breast neoplasm/tumor/carcinoma/cancer
	Central nervous system/peripheral nerves neoplasm/tumor/carcinoma/cancer
	Glial tumor/glioblastoma
	Medulloblastoma
	Meningioma
	Neuroblastoma
	Choriocarcinoma
	Digestive neoplasm/tumor/carcinoma/cancer
	Colon neoplasm/tumor/carcinoma/cancer
	Esophageal neoplasm/tumor/carcinoma/cancer
	Estomach/gastric neoplasm/tumor/carcinoma/cancer
	Rectum/rectalneoplasm/tumor/carcinoma/cancer
	Small bowel neoplasm/tumor/carcinoma/cancer
	Endocrine tumor
	Adrenal neoplasm/tumor/carcinoma/cancer
	Cortical adrenal neoplasm/tumor/carcinoma/cancer
	Medullar adrenal neoplasm/tumor/carcinoma/cancer
	Pheochromocytoma/paraganglioma
	Neuroendocrine tumor/apudoma/carcinoid
	Parathyroid neoplasm/tumor/carcinoma/cancer
	Pituitary/hypophyseal neoplasm/tumor/carcinoma/cancer
	Thyroid neoplasm/tumor/carcinoma/cancer
	Extrahepatic biliary tract/gallbladder neoplasm/tumor/carcinoma/cancer
	Eye neoplasm/tumor/carcinoma/cancer
	Retinoblastoma
	Germ cell malignancy/germinoma/teratoma (excluding ovary/testis)
	Head and neck neoplasm/tumor/carcinoma/cancer
	Laryngeal neoplasm/tumor/carcinoma/cancer
	Lip neoplasm/tumor/carcinoma/cancer
	Nasopharynx neoplasm/tumor/carcinoma/cancer
	Nose/nasal sinus neoplasm/tumor/carcinoma/cancer
	Odontogenic neoplasm/tumor/carcinoma/cancer
	Oral cavity/tongue neoplasm/tumor/carcinoma/cancer
	Oropharyngeal neoplasm/tumor/carcinoma/cancer
	Salivary gland neoplasm/tumor/carcinoma/cancer
	Heart/cardiac tumor

Neoplasms/tumors (continued)	
Hematologic/blood/lymphatic cancer	
	Acute leukemia
	Histiocytosis
	Lymphoma
	Macroglobulinemia
	Multiple myeloma
	Mastocytosis
	Myelodysplastic syndrome
	Myeloproliferative syndrome/chronic leukemia
Hepatic/liver neoplasm/tumor/carcinoma/cancer	
	Hepatoblastoma
Kidney/renal neoplasm/tumor/carcinoma/cancer	
	Nephroblastoma/Wilms tumor
Lung/bronchopulmonary neoplasm/tumor/carcinoma/cancer	
	Pleuropulmonary blastoma
Melanoma	
Mesothelioma	
Middle ear neoplasm/tumor/carcinoma/cancer	
Ovary/Fallopian tube neoplasm/tumor/carcinoma/cancer (excl. teratoma/germinoma)	
Ovary/ovarian teratoma/germinoma	
Pancreatic/pancreas neoplasm/tumor/carcinoma/cancer	
	Pancreatoblastoma
Penis/scrotum neoplasm/tumor/carcinoma/cancer	
Prostate/prostatic neoplasm/tumor/carcinoma/cancer	
Sarcoma	
	Bone/osseous neoplasm/tumor/carcinoma/cancer
	Gastrointestinal stromal tumor
	Kaposi's sarcoma
	Soft tissue sarcoma/cancer/tumor/liposarcoma/myosarcoma
Skin/cutaneous neoplasm/tumor/carcinoma/cancer (excluding melanoma)	
Testicular teratoma/germinoma	
Testicular/seminal neoplasm/tumor/carcinoma/cancer (excl. teratoma/germinoma)	
Thymus/thymic neoplasm/tumor/carcinoma/cancer/thymoma	
Tracheal neoplasm/tumor/carcinoma/cancer	
Ureteral/urethral/vesical/bladder neoplasm/tumor/carcinoma/cancer	

Neoplasms/tumors (continued)	
	Uterus/uterine/cervix/endometrium neoplasm/tumor/carcinoma/cancer
	Vaginal/vulvar neoplasm/tumor/carcinoma/cancer
	Vascular malignancy/tumor
Chromosomal or genetic anomaly	
	Autosomal dominant inheritance
	Autosomal recessive inheritance
	Chromosome breakage
	Fragile chromosomal site (other than Xq28)
	Fragile chromosome X site
	Genomic imprinting anomaly
	Interstitial/subtelomeric microdeletion/deletion
	Polygenic/multifactorial inheritance
	Total/partial monosomy/deletion
	Total/partial trisomy/duplication
	X-linked dominant inheritance
	X-linked recessive inheritance
Build/stature/longevity anomalies	
	Abnormal fat distribution/lipodystrophy
	Acromegaly
	Asymmetry of the body/hemiatrophy/hemihyperthrophy
	Cushingoid morphotype
	Early death/lethality
	Death in infancy
	Early death in adulthood
	Stillbirth/neonatal death
	Eunochoid morphotype
	Failure to thrive/difficulties for feeding in infancy/growth delay
	Generalized obesity
	Marfanoid morphotype
	Premature ageing
	Short stature/dwarfism/nanism
	Tall stature/gigantism/growth acceleration
	Truncal obesity
	Wasted (excluding lipodystrophy)/poorly muscled build/cachexy

Health status anomalies	
	Angioedema/angioneurotic edema
	Asthenia/fatigue/weakness
	Biological inflammatory syndrome/increased erythrocyte sedimentation rate/CRP
	Dehydration/hydroelectrolytic loss
	Fever/chilling
	Hot flushes/sensation of cold
	Hypothermia
	Malignant hyperthermia
	Pallor
	Sepsis severe/septicemia
	Thirst
	Weight loss/loss of appetite/break in weight curve/general health alteration
Antenatal period/pregnancy anomalies	
	Antenatal exposure : AIDS
	Antenatal exposure : alcohol
	Antenatal exposure : antibiotics
	Antenatal exposure : anticoagulatives
	Antenatal exposure : anticonvulsants
	Antenatal exposure : antimetotics
	Antenatal exposure : chemical product
	Antenatal exposure : cocaine/LSD
	Antenatal exposure : cytomegalovirus
	Antenatal exposure : hepatitis
	Antenatal exposure : hormones
	Antenatal exposure : influenza
	Antenatal exposure : insuline
	Antenatal exposure : iodures
	Antenatal exposure : ionizing radiations
	Antenatal exposure : rubella
	Antenatal exposure : sedatives
	Antenatal exposure : syphilis
	Antenatal exposure : toxoplasmosis
	Antenatal exposure : varicella
	Antenatal exposure to infectious agent
	Conjoined twinning
	Fetal immobility/abnormal fetal movements

Antenatal period/pregnancy anomalies (continued)	
	Hydrops fetalis
	Increased nuchal translucency
	Intrauterine growth retardation
	Maternal diabetes
	Maternal hyperphenylalaninemia
	Maternal hypertension/eclampsia/preeclampsia/gravidic toxemia
	Maternal seizures
	Oligoamnios
	Polyhydramnios
	Prematurity
	Spontaneous abortions
	Twinning (type unknown)
	Dizygotic twinning
	Monozygotic twinning