CHAPTER 20

Developmental anomalies

This chapter has 222 four-character categories.

Code range starts with LA00

This chapter includes conditions caused by failure of a particular body site or body system to develop correctly during the antenatal period.

Exclusions: Inborn errors of metabolism (BlockL2‑5C5)

This chapter contains the following top level blocks:

* Structural developmental anomalies primarily affecting one body system
* Multiple developmental anomalies or syndromes
* Chromosomal anomalies, excluding gene mutations

Structural developmental anomalies primarily affecting one body system (BlockL1‑LA0)

A deformation established before birth of an anatomical structure.

Structural developmental anomalies of the nervous system (BlockL2‑LA0)

Any condition caused by failure of the nervous system to correctly develop during the antenatal period.

LA00 Anencephaly or similar anomalies

A malformation of the nervous system caused by the failure of neuropore closure. Infants are born with intact spinal cord, cerebellum, and brainstem, but lack formation of neural structures above this level. The skull is only partially formed but the eyes are usually normal.

LA00.0 Anencephaly

Anencephaly is a neural tube defect, characterised by the total or partial absence of the cranial vault and the covering skin, the brain being missing or reduced to a small mass. Most cases are stillborn, although some infants have been reported to survive for a few hours. In most cases autopsy findings reveal absence of adrenal glands. Anencephaly is likely to be multifactorial, the result of gene-environment interactions. Familial cases with a seemingly autosomal recessive mode of inheritance have been described but most cases are sporadic. Folic acid and zinc deficiencies, as well as maternal obesity, have been shown to be risk factors.

LA00.00 Craniorachischisis

A condition caused by failure of the neural tube to close completely during the antenatal period. This condition is characterised by complete absence of the skull, extensive defects in the vertebrae and skin, and absence of the brain.

LA00.0Y Other specified anencephaly

LA00.0Z Anencephaly, unspecified

LA00.1 Iniencephaly

Iniencephaly is a rare form of neural tube defect in which a malformation of the cervico-occipital junction is associated with a malformation of the central nervous system. The cardinal features are occipital bone defect, partial or total absence of cervicothoracic vertebrae, fetal retroflexion of the head and characteristic absence of the neck. It is associated with malformations of the central nervous (spina bifida and/or anencephaly), gastrointestinal (omphalocele) and cardiovascular systems.

LA00.2 Acephaly

LA00.3 Amyelencephaly

Amyelencephaly is the absence of both the brain and spinal cord.

LA00.Y Other specified anencephaly or similar anomalies

LA00.Z Anencephaly or similar anomalies, unspecified

LA01 Cephalocele

A condition caused by failure of the skull to correctly close during the antenatal period. This condition is characterised by herniation of the meninges. This condition may present with herniation of brain, or developmental delay. Confirmation is through observation of herniated meninges by imaging.

LA02 Spina bifida

Spina bifida is the most common of a group of birth defects called neural tube defects. Spina bifida affects the backbone and, sometimes, the spinal cord. Aperta spina bifida defines the dorsal malclosure of vertebrae, associated with various degrees of spine defects. A pocket of skin may form, containing meninges (meningocele) or spinal cord and meninges (myelomeningocele). Different subtypes are distinguished according to the location of the defect. Consequences are paraplegia (paralysed lower limbs), hydrocephaly, Chiari malformation (result of the attached spine during life in utero), urinary and anorectal incontinence. The intensity of signs varies greatly with the level and extent of the lesion.

Inclusions: Rachischisis

Spinal dysraphism

Exclusions: Arnold-Chiari malformation type I (LA07.4)

Arnold-Chiari malformation type II (LA03)

Occult spinal dysraphism (LB73.0)

LA02.0 Spina bifida cystica

A condition caused by failure of the neural tube to correctly develop during the antenatal period. This condition is characterised by nerve damage and the presence of meningoceles on the back. This condition may present with physical or mental impairment.

LA02.00 Myelomeningocele with hydrocephalus

A condition caused by failure of the neural tube to correctly develop during the antenatal period. This condition is characterised by nerve damage and hydrocephalus. This condition may also present with syringomyelia, hip dislocation, headache, nausea, vomiting, blurry vision, balance problems, bladder control problems, meningitis, or mental impairment.

LA02.01 Myelomeningocele without hydrocephalus

A condition caused by failure of the neural tube to close completely during fetal development. This condition is characterised by nerve damage. This condition may also present with syringomyelia, hip dislocation, headache, nausea, vomiting, blurry vision, balance problems, bladder control problems, meningitis, or mental impairment.

LA02.02 Myelocystocele

A condition caused by failure of the neural tube to close completely during fetal development. The condition is characterised by skin covered lumbosacral masses, an arachnoid lined meningocele that is directly continuous with the spinal subarachnoid space, and a low lying hydromyelic spinal cord that traverses the meningocele and expands into a large terminal cyst. This condition can present with neural damage and consequent impairment of function below the site of the myelocystocele.

LA02.0Y Other specified spina bifida cystica

LA02.0Z Spina bifida cystica, unspecified

LA02.1 Spina bifida aperta

A condition caused by failure of the neural tube to correctly develop during the antenatal period. This condition is characterised by nerve damage originating from a known location in the spine, signified by the presence of a meningocele or myelomeningocele. This condition may present with physical or mental impairment.

LA02.Y Other specified spina bifida

LA02.Z Spina bifida, unspecified

LA03 Arnold-Chiari malformation type II

A condition caused by failure of the brain and spinal cord to correctly develop during the antenatal period. This condition is characterised by extension of both cerebellar and brain stem tissue into the foramen magnum. This condition may present with partial or complete absence of the cerebellar vermis, myelomeningocele, neck pain, balance problems, muscle weakness, limb numbness, dizziness, vision problems, difficulty swallowing, ringing in the ears, hearing loss, vomiting, insomnia, depression, or impairment of motor skills.

Exclusions: Arnold-Chiari malformation type I (LA07.4)

LA04 Congenital hydrocephalus

A disease caused by failure of the brain to correctly develop during the antenatal period. This condition is characterised by a rapid increase in head circumference or an unusually large head size due to excessive accumulation of cerebrospinal fluid in the brain. This condition may also present with vomiting, sleepiness, irritability, downward deviation of the eyes, or seizures. Confirmation is through observation of cerebrospinal fluid within cerebral ventricles by imaging.

Inclusions: Hydrocephalus in newborn

Exclusions: Myelomeningocele with hydrocephalus (LA02.00)

Hydrocephalus due to congenital toxoplasmosis (KA64.0)

Arnold-Chiari malformation type I (LA07.4)

Arnold-Chiari malformation type II (LA03)

LA04.0 Hydrocephalus with stenosis of the aqueduct of Sylvius

Hydrocephalus with stenosis of aqueduct of Sylvius (HSAS) or Bickers-Adams syndrome is characterised by the association of hydrocephaly, severe intellectual deficit, spasticity and adducted thumbs, and is part of the L1 syndrome (see this term).

Inclusions: Stenosis of the aqueduct of Sylvius

LA04.Y Other specified congenital hydrocephalus

LA04.Z Congenital hydrocephalus, unspecified

LA05 Cerebral structural developmental anomalies

Any condition caused by failure of the brain to correctly develop during the antenatal period.

Exclusions: Encephalocele (LA01)

LA05.0 Microcephaly

A condition caused by failure of the head to correctly develop during the antenatal period. This condition is characterised by a head size that is significantly smaller than normal for their age and sex. This condition may also present with developmental delays, difficulties with balance and coordination, short stature, hyperactivity, mental retardation, seizures, or other neurological abnormalities.

Coding Note: Code aslo the casusing condition

Inclusions: Micrencephaly

Exclusions: Syndromes with microcephaly as a major feature (LD20.2)

LA05.1 Megalencephaly

A condition caused by failure of the brain to correctly develop during the antenatal period. This condition is characterised by increased size or weight of an otherwise correctly formed brain. This condition may also present with seizures, motor deficits, mental retardation and mild cognitive impairment.

LA05.2 Holoprosencephaly

Holoprosencephaly is a brain malformation resulting from incomplete cleavage of the prosencephalon, occurring between the 18th and the 28th day of gestation and affecting both the forebrain and the face. In most of the cases, facial anomalies are observed: cyclopia, proboscis and median or bilateral cleft lip/palate in severe forms, and ocular hypotelorism or solitary median maxillary central incisor in minor forms. These latter midline defects can occur without the cerebral malformations (microforms). Children with HPE have many medical problems: developmental delay and feeding difficulties, epilepsy, and instability of temperature, heart rate and respiration. Endocrine disorders like diabetes insipidus, adrenal hypoplasia, hypogonadism, thyroid hypoplasia and growth hormone deficiency are frequent.

Coded Elsewhere: Cyclopia (LA10.Y)

LA05.3 Corpus callosum agenesis

Corpus callosum agenesis is the most common brain malformation and is characterised by total or partial absence of the main interhemispheric commissure, the corpus callosum.

LA05.4 Arrhinencephaly

A condition caused by failure of the olfactory organs to correctly develop during the antenatal period. This condition is characterised by absence of the olfactory bulbs and tracts.

LA05.5 Abnormal neuronal migration

Any condition caused by abnormal migration of neuronal cells during the antenatal period. These conditions may present with poor muscle tone and motor function, seizures, developmental delays, mental retardation, failure to grow and thrive, difficulties with feeding, swelling in the extremities or microcephaly.

Exclusions: Lissencephaly (LD20.1)

LA05.50 Polymicrogyria

Polymicrogyria (PMG) is a cerebral cortical malformation characterised by excessive cortical folding and by shallow sulci. Microscopic examination reveals abnormal cortical layering. Topographic distribution of PMG is variable, but bilateral symmetrical perisylvian PMG (BPP) is the most frequent form. PMG is manifested by mild intellectual deficit, epilepsy, and pseudobulbar palsy, which causes difficulties with speech learning and feeding. The severity of PMG is highly dependent on the location and size of the affected area.

LA05.51 Cortical dysplasia

A condition caused by failure of the cortex to correctly develop during the antenatal period, or by trauma. This condition is characterised by epileptic seizures. This condition may also present with learning impairments.

LA05.5Y Other specified abnormal neuronal migration

LA05.5Z Abnormal neuronal migration, unspecified

LA05.6 Encephaloclastic disorders

LA05.60 Porencephaly

Porencephaly is characterised by a circumscribed intracerebral cavity of variable size that may be bordered by abnormal polymicrogyric grey matter. In extreme cases, this cavity may result in a communication between the pial surface and the ventricle; this is termed schizencephaly.

LA05.61 Schizencephaly

Schizencephaly is a rare congenital cerebral malformation characterised by the presence of linear clefts in one or both hemispheres of the brain, extending from the lateral ventricles to the pial surface of the cortex, and that lead to a variety of neurological symptoms such as epilepsy, motor deficits, and psychomotor retardation.

LA05.62 Hydranencephaly

A condition caused by failure of the cerebral hemispheres to develop during the antenatal period. This condition is characterised by a lack of a forebrain upon imaging. This condition may present with visual impairment, lack of growth, deafness, blindness, spastic quadriparesis, or intellectual deficits.

LA05.6Y Other specified encephaloclastic disorders

LA05.6Z Encephaloclastic disorders, unspecified

LA05.7 Brain cystic malformations

A disease caused by expansion of the roof plate of the brain vesicle, or by extraaxial structures such as an arachnoid membrane or migrating ependymal cells. This disease is characterised by the presence of fluid filled cysts in the brain. This disease may present with asymmetry of the skull, brain compression, raised intracranial pressure, hydrocephalus, bleeding or seizures. This disease may also be asymptomatic. Confirmation is through observation of intracerebral cysts by imaging.

Exclusions: Acquired porencephalic cysts (8E40)

Dandy-Walker malformation with hydrocephalus (LA06.0)

Dandy-Walker malformation without hydrocephalus (LA06.0)

Coded Elsewhere: Intracranial arachnoid cyst (8D67)

LA05.Y Other specified cerebral structural developmental anomalies

LA05.Z Cerebral structural developmental anomalies, unspecified

LA06 Cerebellar structural developmental anomalies

Any condition caused by failure of the brain to correctly develop during the antenatal period.

Exclusions: Arnold-Chiari malformation type I (LA07.4)

Arnold-Chiari malformation type II (LA03)

LA06.0 Dandy-Walker malformation

LA06.1 Hypoplasia or agenesis of cerebellar hemispheres

Cerebellar hypoplasia corresponds to underdevelopment of cerebellar structures that can involve the vermis and/or the cerebellar hemispheres from partial to total agenesis. It has been described in the context of various clinical entities: chromosomal anomalies, in utero exposure to toxins and infectious agents, metabolic disorders (disorders of glycosylation and CoQ10 deficiencies), and a wide variety of rare genetic neurological diseases. It can be confined to the cerebellum, or affect other CNS structures: the midbrain (molar tooth syndromes), pons and medulla (ponto-cerebellar hypoplasia), cerebral cortex (lissencephaly cerebellar hypoplasia syndromes).

Exclusions: PHACE syndrome (LD2F.1)

LA06.2 Focal cerebellar dysplasia

A condition caused by failure of the cerebellum to correctly develop during the antenatal period. This condition may present with hypotonia, facial deformities, abnormalities in eyes or in ocular motricity, cognitive deficiencies, or motor dysfunction. Confirmation is through observation of a malformed cerebellum by imaging.

LA06.Y Other specified cerebellar structural developmental anomalies

LA06.Z Cerebellar structural developmental anomalies, unspecified

LA07 Structural developmental anomalies of the neurenteric canal, spinal cord or vertebral column

Any condition caused by failure of the neurenteric canal, spinal cord and vertebral column to correctly develop during the antenatal period.

Coded Elsewhere: Occult spinal dysraphism (LB73.0)

LA07.0 Primary tethered cord syndrome

A condition caused by failure of the spinal cord to correctly develop during the antenatal period. This condition is characterised by tethering of the spinal cord to the spinal canal. This condition may present with lower back skin appendages, radicular pain, weakness, asymmetric hyporeflexia, spasticity, sensory changes, bowel or bladder dysfunction, or motor dysfunction. Confirmation is through observation of a tethered spinal cord by imaging.

LA07.1 Diastematomyelia

A condition caused by failure of the of the spinal cord during the antenatal period. This condition is characterised by separation of the spinal cord into two parts by a rigid or fibrous septum. This condition may present with misformed vertebrae, pain, weakness, impaired gait, sensory changes in the legs, or sphincter disturbance. Confirmation is through observation of a septum-bifurcated spinal cord by imagine.

Inclusions: Split cord malformation

LA07.2 Amyelia

A condition caused by malformation of the spinal cord during the antenatal period. This condition is characterised by absence of sections of the spinal cord.

Inclusions: Spinal cord agenesis

LA07.3 Primary syringomyelia or hydromyelia

A condition caused by failure of the spinal canal to correctly develop during the antenatal period. This condition is characterised by a cavity within the spinal cord in which cerebrospinal fluid can accumulate. Confirmation is through observation of a fluid filled cavity within the spinal cord by imaging.

Exclusions: Syringomyelia due to certain specified cause (8D66.1)

LA07.4 Arnold-Chiari malformation type I

A condition caused by failure of the cerebellum to correctly develop during the antenatal period. This condition is characterised by extension of the cerebellar tonsils into the foramen magnum, without involving the brain stem. This condition may present as asymptomatic. Confirmation is through observation of the cerebellar tonsil extension by imaging.

Exclusions: Arnold-Chiari malformation type II (LA03)

LA07.Y Other specified structural developmental anomalies of the neurenteric canal, spinal cord or vertebral column

LA07.Z Structural developmental anomalies of the neurenteric canal, spinal cord or vertebral column, unspecified

LA0Y Other specified structural developmental anomalies of the nervous system

LA0Z Structural developmental anomalies of the nervous system, unspecified

Structural developmental anomalies of the eye, eyelid or lacrimal apparatus (BlockL2‑LA1)

Any condition caused by failure of the eye, eyelid and lacrimal apparatus to correctly develop during the antenatal period.

LA10 Structural developmental anomalies of ocular globes

Any condition caused by failure of the ocular globes to correctly develop during the antenatal period.

Exclusions: Holoprosencephaly with cyclopia or synophthalmia (LA05.2)

LA10.0 Microphthalmos

Inclusions: Dysplasia of eye

Hypoplasia of eye

Rudimentary eye

LA10.1 Clinical anophthalmos

This refers to the clinical absence of one or both eyes. Both the globe (human eye) and the ocular tissue are missing from the orbit. The absence of the eye will cause a small bony orbit, a constricted mucosal socket, short eyelids, reduced palpebral fissure and malar prominence. Genetic mutations, chromosomal abnormalities, and prenatal environment can all cause anophthalmia. Anophthalmia is an extremely rare disease and is mostly rooted in genetic abnormalities.

Inclusions: Agenesis of eye

Aplasia of eye

LA10.2 Buphthalmos

A condition characterised by enlargement of the globe of the eye.

LA10.3 Congenital macrophthalmos

A condition caused by failure of the eye to develop correctly during the antenatal period. This condition is characterised by enlargement of the globe of the eye.

Exclusions: macrophthalmos in congenital glaucoma (9C61.4)

LA10.Y Other specified structural developmental anomalies of ocular globes

LA10.Z Structural developmental anomalies of ocular globes, unspecified

LA11 Structural developmental anomalies of the anterior segment of eye

Any condition caused by failure of the anterior segment of the eye to correctly develop during the antenatal period.

Coded Elsewhere: Developmental glaucoma (9C61.4)

LA11.0 Blue sclera

A condition of the eye, characterised by transparency of the sclera such that the blue uvea is visible.

LA11.1 Structural developmental anomalies of cornea

Any condition caused by failure of the cornea to correctly develop during the antenatal period.

Coded Elsewhere: Corneal staphyloma (9A78.51)

LA11.2 Anterior segment dysgenesis

A condition caused by failure of the anterior structures of the eye to correctly develop during the antenatal period. This condition may present with iris hypoplasia, irregular and misplaced pupils, hazy corneas, or attachments of the iris to the cornea.

LA11.3 Aniridia

Aniridia is a congenital ocular malformation characterised by the complete or partial absence of the iris. It can be isolated or part of a syndrome (isolated and syndromic aniridia; see these terms).

LA11.4 Coloboma of iris

A disease of the eye, caused by trauma or congenital genetic mutation. This disease is characterised by notches or gaps in iris.

LA11.5 Congenital corneal opacity

A condition caused by failure of the cornea to correctly develop during the antenatal period. This condition is characterised by opacity of the cornea.

Coded Elsewhere: Peters anomaly (9C61.42)

Congenital hereditary endothelial dystrophy type 2 (9A70.0)

LA11.6 Structural disorders of the pupil

LA11.60 Irregular pupil of the eye

LA11.61 Iridoschisis

LA11.62 Anomalies of pupillary function

This is a group of conditions associated with pupillary function which is to regulate the amount of light that enters the eye controlled by the muscular structures of the iris.

Coded Elsewhere: Congenital mydriasis (9B01.3)

LA11.6Y Other specified structural disorders of the pupil

LA11.6Z Structural disorders of the pupil, unspecified

LA11.Y Other specified structural developmental anomalies of the anterior segment of eye

LA11.Z Structural developmental anomalies of the anterior segment of eye, unspecified

LA12 Structural developmental anomalies of lens or zonula

Any condition caused by failure of the lens and zonula to correctly develop during the antenatal period.

LA12.0 Coloboma of lens

LA12.1 Congenital cataract

Partial or complete opacity on or in the lens or capsule of one or both eyes, impairing vision or causing blindness; typically diagnosed at birth

LA12.2 Congenital aphakia

Congenital primary aphakia is a developmental eye defect characterised by an absence of the lens, and can be associated with variable secondary ocular defects (including aplasia/dysplasia of the anterior segment of the eye, microphthalmia, and in some cases absence of the iris, retinal dysplasia, or sclerocornea).

LA12.3 Spherophakia

A disease of the eye, caused by homozygous mutations in the LTBP2 gene (Isolated spherophakia), or by other genetic mutations. This disease is characterised by small, spherical lenses. This disease can also present with lenticular myopia, glaucoma, or sublation of the lens into the vitreous cavity.

LA12.Y Other specified structural developmental anomalies of lens or zonula

LA12.Z Structural developmental anomalies of lens or zonula, unspecified

LA13 Structural developmental anomalies of the posterior segment of eye

Any condition caused by failure of the posterior segment of the eye to correctly develop during the antenatal period. These conditions are characterised by clinical, functional, or morphological changes to the posterior segment of the eye.

Coded Elsewhere: Juvenile retinoschisis (9B73.11)

Optic nerve hypoplasia or aplasia (LA13.7Y)

LA13.0 Congenital anomalies of the vitreous

Coded Elsewhere: Congenital vitreoretinal dysplasia (LA13.3)

Persistent hyperplastic primary vitreous (LA13.Y)

LA13.1 Coloboma of choroid or retina

A condition of the eye characterised by absence of the retina in the lower inside corner of the eye.

LA13.2 Coloboma of macula

A disease caused by malformation of the macula due to retinal inflammation during the antenatal period or by congenital genetic mutation. This disease is characterised by a clearly delineated defect in the macula.

LA13.3 Congenital vitreoretinal dysplasia

Any disease caused by the maldevelopment of the vitreous and retina.

Coded Elsewhere: Incontinentia pigmenti (LD27.00)

Walker Warburg syndrome (8C70.6)

Norrie disease (LD21.Y)

LA13.5 Congenital retinal aneurysm

LA13.6 Congenital malformations of choroid

These are single or multiple defects of the morphogenesis of the choroid, the vascular layer of the eye, identifiable at birth or during the intrauterine life.

LA13.7 Congenital malformation of optic disc

LA13.70 Isolated optic nerve hypoplasia

LA13.71 Optic nerve aplasia

LA13.72 Congenitally elevated optic disc

LA13.73 Optic disc dysplasia

deformed optic discs that fail to conform to any recognizable diagnostic category

LA13.74 Megalopapilla

LA13.76 Coloboma of optic disc

Congenital abnormal optic disc appearance due to incomplete coaptation of the proximal end of the embryonic fissure in ocular development

LA13.7Y Other specified congenital malformation of optic disc

LA13.7Z Congenital malformation of optic disc, unspecified

LA13.8 Certain congenital malformations of posterior segment of eye

Coded Elsewhere: Coloboma of choroid or retina (LA13.1)

LA13.80 Anastomosis of retinal or choroidal vessels

LA13.Y Other specified structural developmental anomalies of the posterior segment of eye

LA13.Z Structural developmental anomalies of the posterior segment of eye, unspecified

LA14 Structural developmental anomalies of eyelid, lacrimal apparatus or orbit

Any condition caused by failure of the eyelid lacrimal apparatus and orbit to correctly develop during the antenatal period.

Exclusions: cryptophthalmos NOS (LA10.0)

LA14.0 Structural developmental anomalies of eyelids

Coding Note: Code also any associated syndrome

LA14.00 Palpebral cleft or coloboma

LA14.01 Cryptophthalmia

Isolated cryptophtalmia is a congenital abnormality in which the eyelids are absent and skin covers the ocular bulb, which is often microphthalmic.

LA14.02 Congenital entropion

LA14.03 Congenital ectropion

LA14.04 Congenital ptosis

Congenital ptosis is characterised by superior eyelid drop present at birth.

LA14.05 Congenital eyelid retraction

LA14.06 Epibulbar choristoma

LA14.07 Ankyloblepharon filiforme adnatum

Isolated ankyloblepharon filiforme adnatum is characterised by the presence of single or multiple thin bands of connective tissue between the upper and lower eyelids, preventing full opening of the eye.

LA14.0Y Other specified structural developmental anomalies of eyelids

Coding Note: Code also any associated syndrome

LA14.1 Structural developmental anomalies of lacrimal apparatus

This refers to structural developmental anomalies of the physiologic system containing the orbital structures for tear production and drainage.

LA14.10 Aplasia of lacrimal or salivary glands

LA14.11 Agenesis of lacrimal ducts

Isolated congenital alacrima is characterised by deficient lacrimation (ranging from a complete absence of tears to hyposecretion of tears) that is present from birth.

Inclusions: Absence of punctum lacrimale

LA14.12 Congenital dacryocele

LA14.13 Congenital agenesis of lacrimal punctum

LA14.14 Congenital stenosis or stricture of lacrimal duct

LA14.1Y Other specified structural developmental anomalies of lacrimal apparatus

LA14.1Z Structural developmental anomalies of lacrimal apparatus, unspecified

LA14.2 Structural developmental anomalies of orbit

Any condition caused by failure of the orbit to correctly develop during the antenatal period.

LA14.Y Other specified structural developmental anomalies of eyelid, lacrimal apparatus or orbit

LA14.Z Structural developmental anomalies of eyelid, lacrimal apparatus or orbit, unspecified

LA1Y Other specified structural developmental anomalies of the eye, eyelid or lacrimal apparatus

LA1Z Structural developmental anomalies of the eye, eyelid or lacrimal apparatus, unspecified

Structural developmental anomalies of the ear (BlockL2‑LA2)

Any condition caused by failure of the ear to correctly develop during the antenatal period.

LA20 Structural anomaly of eustachian apparatus

LA21 Minor anomalies of pinnae

Any condition caused by failure of the pinna to correctly develop during the antenatal period. These conditions are characterised by asymptomatic abnormalities of the visible part of the ear.

LA21.0 Macrotia

1) Median longitudinal ear length greater than 2 SD above the mean and median ear width greater than 2 SD above the mean (objective). 2) Apparently increase in length and width of the pinna (subjective).

LA21.1 Protruding ear

LA21.2 Low-set ear

The upper third of the pinna is not above a plane defined by the lateral canthi and the maximum convexity of the occiput.

Exclusions: cervical auricle (LA23)

LA21.3 Misshapen ear

A condition caused by the malformation of the ear during the antenatal period.

Exclusions: Acquired deformity of pinna (AA41)

LA21.Y Other specified minor anomalies of pinnae

LA22 Structural developmental anomalies of ear causing hearing impairment

Any condition caused by the failure of the ear to correctly develop during the antenatal period. These conditions are characterised by hearing impairment.

LA22.0 Microtia

Microtia is a congenital malformation of variable severity of the external and middle ear. Both hereditary factors (evidence for familial craniofacial microsomia and patterns suggestive of multifactorial inheritance)

and vascular accidents are involved in the etiology of the disease. Specific causative factors also can include maternal rubella during the first trimester of pregnancy. Microtia commonly involves the external canal and middle ear; hence, hearing can be affected. Microtia may present within a spectrum of branchial arch defects (hemifacial microsomia, craniofacial microsomia) or may manifest as an independent malformation. The microtic auricle consists of a disorganised remnant of cartilage attached to a variable amount of soft tissue lobule.

LA22.1 Anotia

Complete absence of any auricular structures.

LA22.2 Aplasia or hypoplasia of external auditory canal

Exclusions: Microtia (LA22.0)

Anotia (LA22.1)

LA22.3 Structural developmental anomalies of ear ossicles

Any condition caused by failure of the tear ossicles to correctly develop during the antenatal period.

LA22.4 Structural developmental anomalies of inner ear

Any condition caused by failure of the inner ear to correctly develop during the antenatal period.

LA22.Y Other specified structural developmental anomalies of ear causing hearing impairment

LA22.Z Structural developmental anomalies of ear causing hearing impairment, unspecified

LA23 Otocephaly

Malplacement of the external ears with or without fusion microstomia, and persistence of the buccopharyngeal membrane likely being secondary effects of absence or hypoplasia of the mandibular arch.

LA24 Accessory auricle

A condition caused by development of an auricular appendage during the antenatal period.

LA2Y Other specified structural developmental anomalies of the ear

LA2Z Structural developmental anomalies of the ear, unspecified

Structural developmental anomalies of the face, mouth or teeth (BlockL2‑LA3)

Any condition caused by failure of the face, mouth and teeth to correctly develop during the antenatal period.

Coded Elsewhere: Dermoid cyst (LC40)

Congenital micrognathia (DA0E.00)

LA30 Structural developmental anomalies of teeth and periodontal tissues

Coded Elsewhere: Disturbances in tooth formation (DA07.3)

Root anomaly (DA07.4)

Disturbances in tooth eruption (DA07.6)

Anomalies of tooth position (DA0E.3)

LA30.0 Anodontia

Anodontia is a genetic disorder commonly defined as the absence of all teeth, affecting both, temporary and permanent dentitions, and is extremely rarely encountered in a pure form without any associated abnormalities. Rare but more common than complete anodontia are hypodontia.

LA30.1 Hypodontia

Hypodontia presents as a lack of one or a few (less than 6) permanent teeth, without any systemic disorders.

Inclusions: Congenital absence of one tooth

LA30.2 Oligodontia

A genetic condition characterised by the development of fewer than the normal number of teeth. The diagnosis of Oligodontia is usually made in cases in which more than six teeth are missing.

LA30.3 Hyperdontia

is the condition of having supernumerary teeth, or teeth which appear in addition to the regular number of teeth.

Inclusions: Supplementary teeth

Supernumerary teeth

distomolar

Fourth molar

Mesiodens

Paramolar

LA30.4 Abnormalities of size or form of teeth

A group of conditions characterised by abnormal size and form of teeth.

LA30.5 Anomalies in tooth resorption or loss

Coded Elsewhere: Pathological resorption of teeth (DA08.14)

LA30.50 Early exfoliation of teeth

LA30.51 Late exfoliation of teeth

LA30.5Y Other specified anomalies in tooth resorption or loss

LA30.5Z Anomalies in tooth resorption or loss, unspecified

LA30.6 Amelogenesis imperfecta

Amelogenesis imperfecta presents with a rare abnormal formation of the enamel or external layer of the crown of teeth. Amelogenesis imperfecta is due to the malfunction of the proteins in the enamel: ameloblastin, enamelin, tuftelin and amelogenin. People afflicted with amelogenesis imperfecta have teeth with abnormal colour: yellow, brown or grey; this disorder can afflict any number of teeth of both dentitions. The teeth have a higher risk for dental cavities and are hypersensitive to temperature changes as well as rapid attrition, excessive calculus deposition, and gingival hyperplasia.

LA30.7 Dentine dysplasia

LA30.8 Dentinogenesis imperfecta

LA30.9 Odontogenesis imperfecta

LA30.Y Other specified structural developmental anomalies of teeth and periodontal tissues

LA30.Z Structural developmental anomalies of teeth and periodontal tissues, unspecified

LA31 Structural developmental anomalies of mouth or tongue

Embryo fetal anomalies affecting structure of maxillo-labial or mandibular tissues or the tongue.

LA31.0 Congenital macroglossia

A condition caused by failure of the tongue to correctly develop during the antenatal period. This condition is characterised by a larger than normal tongue.

LA31.1 Hypoglossia or aglossia

Isolated aglossia and hypoglossia are terms covering the spectrum from partial to total absence of the tongue. These congenital malformations have been classified as part of the group of oromandibular-limb hypogenesis syndromes (OLHS).

LA31.2 Ankyloglossia

A condition of the tongue, caused by short, tight, lingual frenulum or fusion of the tongue to the floor of the mount. This condition is characterised by difficulty in speech articulation due to limitation or restriction in tongue movement.

Inclusions: Tongue tie

LA31.3 Macrostomia

Congenital macrostomia or transverse facial cleft is a rare congenital craniofacial anomaly. It is usually associated with deformities of other structures developed from the first and second branchial arches and is thought to be part of the manifestations of hemifacial microsomia, the second most common congenital craniofacial anomaly.

Inclusions: Transverse facial cleft

LA31.4 Microstomia

A condition of the mouth, caused by congenital genetic mutation, burns, or injury. This condition is characterised by reduction in the size of the oral aperture with or without involvement of the entire oral cavity.

LA31.Y Other specified structural developmental anomalies of mouth or tongue

Clefts of lip, alveolus or palate (BlockL3‑LA4)

A condition caused by failure of the structures of the mouth to correctly develop during the antenatal period. This condition is characterised by a fissure extending across the upper lip, nasal base, alveolar ridge or the palate. This condition may present with disruption of sucking or swallowing in neonates, recurrent otitis, transmission hypoacusia, or abnormalities of the maxillary lateral incisor.

LA40 Cleft lip

Isolated cleft lip is a fissure type embryopathy extending from the upper lip to the nasal base.

Exclusions: Cleft lip and alveolus (LA41)

LA40.0 Cleft lip, unilateral

Exclusions: Cleft lip and alveolus (LA41)

LA40.1 Cleft lip, bilateral

A condition caused by failure of the upper lip to correctly develop during the antenatal period. This condition is characterised by two fissures in the upper lip and a collapsed and stretched nose through one side of the upper lip. This condition may present with disruption of sucking or swallowing in neonates.

Exclusions: Cleft lip and alveolus (LA41)

LA40.2 Cleft lip, median

Exclusions: Cleft lip and alveolus (LA41)

LA41 Cleft lip and alveolus

Cleft lip and alveolus is a fissure type embryopathy that involves the upper lip, nasal base and alveolar ridge in variable degrees.

LA41.0 Cleft lip and alveolus, unilateral

LA41.1 Cleft lip and alveolus, bilateral

LA42 Cleft palate

Cleft palate is a fissure type embryopathy that affects the soft and hard palate to varying degrees.

LA42.0 Cleft hard palate

A condition caused by failure of the palate to correctly develop during the antenatal period. This condition is characterised by a fissure extending across the palate, including the bony portion of the palate. This condition may present with disruption of sucking or swallowing in neonates.

LA42.1 Cleft soft palate

Cleft velum is a fissure type embryopathy that affects in varying degrees the soft palate.

LA42.2 Cleft uvula

Bifid uvula is a fissure type embryopathy affecting the uvula at the back of the soft palate.

LA42.Y Other specified cleft palate

LA42.Z Cleft palate, unspecified

LA4Y Other specified clefts of lip, alveolus or palate

LA4Z Clefts of lip, alveolus or palate, unspecified

LA50 Congenital velopharyngeal incompetence

A condition caused by failure of the velum to correctly develop during the antenatal period. This condition is characterised by improper closing of the velopharyngeal sphincter, nasal speech, and difficulties in pronouncing certain letters or words.

LA51 Facial clefts

Any condition caused by failure of the structures of the face to correctly develop during the antenatal period. These conditions are characterised by a partition in bone, soft tissue, or skin of the face.

Exclusions: Frontofacionasal dysostosis (LD25.3)

Frontonasal dysplasia (LD25.3)

LA52 Facial asymmetry

A condition caused by failure of the face to develop symmetrically during the antenatal period.

LA53 Macrocheilia

A condition characterised by above normal lip volume. This condition may present with difficulties in speaking, drinking, salivary control, or mastication.

LA54 Microcheilia

A condition caused by failure of the lips to develop correctly during the antenatal period. This condition is characterised by below normal lip size.

LA55 Compression facies

A disease caused by neurovascular compression of the facial nerve. This disease is characterised by facial spasm, and abnormal facial expression.

LA56 Pierre Robin syndrome

Pierre-Robin syndrome (or Pierre-Robin sequence) is characterised by triad of orofacial morphological anomalies consisting of retrognathism, glossoptosis and a posterior median velopalatal cleft. This condition is referred to as a sequence because the posterior cleft palate is a secondary defect associated with abnormal mandibular development: mandibular hypoplasia occurring early in gestation causes the tongue to be maintained high-up in the oral cavity, preventing fusion of the palatal shelves.

LA5Y Other specified structural developmental anomalies of the face, mouth or teeth

LA5Z Structural developmental anomalies of the face, mouth or teeth, unspecified

Structural developmental anomalies of the neck (BlockL2‑LA6)

Any condition caused by failure of the neck to correctly develop during the antenatal period.

Coded Elsewhere: Thyroglossal cyst (DA05.Y)

LA60 Webbed neck

A condition caused by failure of the tissues of the neck to correctly develop during the antenatal period. This condition is characterised by a broad neck due to lateral folds of skin. This condition may present with limited range of motion of the neck.

Inclusions: Pterygium colli

LA61 Congenital sternomastoid tumour

LA62 Congenital torticollis

LA6Y Other specified structural developmental anomalies of the neck

LA6Z Structural developmental anomalies of the neck, unspecified

Structural developmental anomalies of the respiratory system (BlockL2‑LA7)

LA70 Structural developmental anomalies of the nose or cavum

Any condition caused by failure of the nose and cavum to correctly develop during the antenatal period.

LA70.0 Arrhinia

Also called nasal agenesis, it is a very rare anomaly in which external nose does not develop and the nasal cavity is totally or partially obliterated.

LA70.1 Bifid nose

Isolated bifid nose is defined as a median cleft of the nose due to fusion anomalies of the medial nasal processes, that may involve the nasal tip only, or the nose on its length. Bifid nose may also be seen in multiple malformation syndromes like frontonasal dysplasia.

LA70.2 Choanal atresia

Any condition in neonates, caused by failure of the nose to correctly develop during the antenatal period. This condition is characterised by narrowing or blockage of the nasal airway by tissue. This condition may also present with chest retraction unless child is breathing through mouth or crying, difficulty breathing, cyanosis, and inability to nurse and breathe at same time.

LA70.3 Congenital perforated nasal septum

A condition caused by trauma during birth or by failure of the nasal septum to correctly develop during the antenatal period. This condition is characterised by the presence of a hole in the nasal septum.

LA70.Y Other specified structural developmental anomalies of the nose or cavum

LA70.Z Structural developmental anomalies of the nose or cavum, unspecified

LA71 Structural developmental anomalies of larynx

Any condition caused by failure of the larynx to correctly develop during the antenatal period.

Coded Elsewhere: Airway obstruction in the neonate due to airway abnormality (KB2J)

Laryngeal lymphatic malformation (LA90.12)

LA71.0 Congenital laryngomalacia

A condition caused by failure of the larynx to correctly develop during the antenatal period. This disease is characterised by collapse of the supraglottic structures into the airway during the inspiratory phase of respiration, resulting in inspiratory stridor. This disease may also present with regurgitation, emesis, cough, choking, slow feedings, weight loss, failure to thrive, tachypnoea, suprasternal and substernal retractions, cyanosis, pectus excavatum, or obstructive sleep apnoea. Confirmation is through verification of supraglottic collapse by flexible laryngoscopy.

LA71.1 Laryngocele

A condition of the larynx, characterised by an abnormal saccular dilatation of the appendix of the laryngeal ventricle of Morgagni. This condition may also present with cough, hoarseness, stridor, sore throat, or swelling of the neck. Alternatively this condition may be asymptomatic.

LA71.2 Laryngeal hypoplasia

A condition caused by failure of the pharynx and larynx to correctly develop during the antenatal period. This condition is characterised by a narrowed airway and protrusion of intestines through the belly button. This condition may also present with high pitched voice, spinal curvature, or learning difficulties.

LA71.3 Congenital subglottic stenosis

A condition caused by failure of the tracheal rings to correctly develop during the antenatal period. This condition is characterised by inspiratory stridor. This condition may also present with shortness of breath, difficulty feeding, or failure to thrive. Confirmation is through observation of the stenosis by flexible laryngoscopy.

LA71.Y Other specified structural developmental anomalies of larynx

LA71.Z Structural developmental anomalies of larynx, unspecified

LA72 Laryngotracheooesophageal cleft

A laryngo-tracheo-oesophageal cleft (LC) is a congenital malformation characterised by an abnormal, posterior, sagittal communication between the larynx and the pharynx, possibly extending downward between the trachea and the oesophagus. Five types of laryngo-tracheo-oesophageal cleft have been described based on the downward extension of the cleft, which typically correlates with the severity of symptoms: Type 0 laryngo-tracheo-oesophageal cleft to Type 4 laryngo-tracheo-oesophageal cleft (see these terms).

LA73 Structural developmental anomalies of trachea

Coded Elsewhere: Congenital tracheobronchomegaly (CA27.1)

LA73.0 Congenital stenosis of trachea

Tracheal stenosis is a fixed intrinsic narrowing of the trachea. The narrowing can be localised to a short or long tracheal segment, often due to a complete tracheal rings. Alternatively, the tracheal lumen may become progressively narrow caudally

Inclusions: Atresia of trachea

LA73.1 Congenital tracheomalacia

Congenital tracheomalacia is a relatively uncommon anomaly that results from an intrinsic weakness of the cartilaginous support of the trachea such that it is prone to collapse especially during expiration.

LA73.Y Other specified structural developmental anomalies of trachea

LA73.Z Structural developmental anomalies of trachea, unspecified

LA74 Structural developmental anomalies of bronchi

This refers to the structural developmental anomalies of the passage of airway in the respiratory tract that conducts air into the lungs.

LA74.0 Congenital stenosis or atresia of bronchus

A condition caused by interruption of a lobar, segmental, or subsegmental bronchus with peripheral mucus impaction, during the antenatal period. This condition is characterised by hyperinflation of the blocked section of lung. This condition may present with respiratory distress, infiltrative pneumonia, or emphysema.

LA74.1 Congenital bronchomalacia

Bronchus characterised by excessive dynamic collapse

LA74.Y Other specified structural developmental anomalies of bronchi

LA74.Z Structural developmental anomalies of bronchi, unspecified

LA75 Structural developmental anomalies of lungs

Any condition caused by failure of the lungs to correctly develop during the antenatal period.

Coded Elsewhere: Primary ciliary dyskinesia, Kartagener type (LA75.Y)

Primary ciliary dyskinesia - retinitis pigmentosa (LA75.Y)

LA75.0 Accessory lobe of lung

An extra lobe of lung beyond the 3 on the right and the 2 on the left

LA75.1 Agenesis of lung

This refers to the absence or rudimentary residua of an undeveloped lung.

LA75.2 Congenital hypoplasia of lung

LA75.3 Congenital hyperplasia of lung

LA75.4 Congenital pulmonary airway malformations

A disease caused by failure of the bronchial structure to correctly develop during the antenatal period. This disease may present with severe respiratory distress in the newborn period, acute respiratory distress or infection later in life, or may be asymptomatic. This disease can be distinguished from other lesions and normal lung by polypoid projections of the mucosa, an increase in smooth muscle and elastic tissue within the cyst walls, an absence of cartilage in the cystic parenchyma, mucous secreting cells, and the absence of inflammation.

Inclusions: Congenital honeycomb lung

Congenital polycystic disease of lung

Exclusions: Cystic lung disease, acquired or unspecified (CB40)

LA75.5 Congenital lobar emphysema

Congenital lobar emphysema is a developmental lung anomaly characterised by over distension of the affected lobe and leading to compression and displacement of adjacent normal lung tissue and mediastinum. In the majority of cases, symptoms appear during the neonatal period or in early childhood. Clinically, children present with signs of respiratory distress, frequently occurring with a lower respiratory tract infection that aggravates air trapping and renders the patient symptomatic.

LA75.6 Congenital sequestration of lung

A medical condition wherein a piece of tissue that ultimately develops into lung tissue is not attached to the pulmonary arterial blood supply, as is the case in normally developing lung. As a result, this sequestered tissue is not connected to the normal bronchial airway. Intralobar sequestration: With intralobar sequestration, the lung tissue lies within the same visceral pleura as the lobe in which it occurs With extralobar sequestrations, an accessory lung is contained within its own pleura

LA75.Y Other specified structural developmental anomalies of lungs

LA75.Z Structural developmental anomalies of lungs, unspecified

LA76 Structural developmental anomalies of pleura

Anomalies of the lining of the lung (visceral pleura) and thoracic cavity (parietal pleura)

LA77 Congenital cyst of mediastinum

A condition caused by failure of the anterior intestine or coelomic cavity to correctly develop during the antenatal period. This condition may be asymptomatic or may present with adjacent organ compression. Confirmation is observation of the cysts by imaging.

LA7Y Other specified structural developmental anomalies of the respiratory system

LA7Z Structural developmental anomalies of the respiratory system, unspecified

Structural developmental anomalies of the circulatory system (BlockL2‑LA8)

Structural developmental anomaly of heart or great vessels (BlockL3‑LA8)

A congenital malformation of the heart and/or great vessels or an acquired abnormality unique to the congenitally malformed heart.

Coded Elsewhere: Congenital heart or great vessel related acquired abnormality (BE14.3)

LA80 Anomalous position-orientation of heart

A congenital cardiovascular finding or malformation in which there is an abnormality of the position or orientation of heart.

LA80.0 Laevocardia

A congenital cardiovascular finding in which the heart is predominantly to the left of the thoracic midline.

Additional information: this is independent of the orientation of the cardiac apex. This is a normal finding and should be coded only in the context of complex congenital heart disease.

Coding Note: This term should be coded only in the context of complex heart disease. This is independent of the orientation of the cardiac apex.

LA80.1 Dextrocardia

A congenital cardiovascular malformation in which the heart is predominantly to the right of the thoracic midline. This is independent of the orientation of the cardiac apex.

LA80.2 Mesocardia

A congenital cardiovascular malformation in which the heart is central or midline within the thorax.

LA80.3 Extrathoracic heart

A congenital cardiovascular malformation in which the heart is at least partially outside of the thorax.

LA80.Y Other specified anomalous position-orientation of heart

LA80.Z Anomalous position-orientation of heart, unspecified

LA81 Abnormal ventricular relationships

A congenital cardiovascular malformation in which the ventricular positions relative to each other or their laterality (sidedness) are abnormal.

LA82 Total mirror imagery

A congenital malformation in which there is complete mirror-imaged arrangement of the internal organs along the left-right axis of the body.

Inclusions: Situs inversus totalis

Exclusions: dextrocardia NOS (LA80.1)

laevocardia (LA80.0)

Primary ciliary dyskinesia, Kartagener type (LA75)

Kartagener triad (LA75)

LA83 Right isomerism

A congenital cardiovascular malformation that is a variant of heterotaxy syndrome in which some paired structures on opposite sides of the left-right axis of the body are symmetrical mirror images of each other, and have the morphology of the normal right-sided structures.

LA84 Left isomerism

A congenital cardiovascular malformation that is a variant of an heterotaxy syndrome in which some paired structures on opposite sides of the left-right axis of the body are symmetrical mirror images of each other, and have the morphology of the normal left-sided structures.

LA85 Congenital anomaly of an atrioventricular or ventriculo-arterial connection

A congenital cardiovascular malformation in which one or more of the following connections is abnormal 1) the morphologically right atrium to the morphologically right ventricle, 2) the morphologically left atrium to the morphologically left ventricle, 3) the morphologically right ventricle to the pulmonary trunk, 4) the morphologically left ventricle to the aorta.

This excludes codes for hearts with a univentricular atrioventricular connection (mitral atresia, tricuspid atresia and double inlet ventricle), as these are listed under Functionally Univentricular Heart.

Exclusions: Functionally univentricular heart (LA89)

LA85.0 Discordant atrioventricular connections

A congenital cardiovascular malformation in which the morphologically right atrium connects to the morphologically left ventricle and the morphologically left atrium connects to the morphologically right ventricle.

LA85.1 Transposition of the great arteries

A congenital cardiovascular malformation in which the morphologically right ventricle or its remnant connects to the aorta and the morphologically left ventricle or its remnant connects to the pulmonary trunk.

LA85.2 Double outlet right ventricle

A congenital cardiovascular malformation in which both great arteries arise entirely or predominantly from the morphologically right ventricle.

LA85.20 Double outlet right ventricle with subpulmonary ventricular septal defect, transposition type

A congenital cardiovascular malformation that is a variant of double outlet right ventricle with concordant atrioventricular connections that is associated with a subpulmonary ventricular septal defect (includes Taussig-Bing heart).

LA85.21 Double outlet right ventricle with non-committed ventricular septal defect

A congenital cardiovascular malformation that is a variant of double outlet right ventricle with concordant atrioventricular connections that is associated with ventricular septal defect that is remote from the ventricular outflow tracts and usually within the inlet or trabecular muscular septum.

LA85.22 Double outlet right ventricle with subaortic or doubly committed ventricular septal defect without pulmonary stenosis, ventricular septal defect type

A congenital cardiovascular malformation that is a variant of double outlet right ventricle with concordant atrioventricular connections, a subaortic or doubly-committed (with absence or deficiency of the conal septum) ventricular septal defect, and unobstructed pulmonary outflow tract.

LA85.23 Double outlet right ventricle with subaortic or doubly committed ventricular septal defect and pulmonary stenosis, Fallot type

A congenital cardiovascular malformation that is a variant of double outlet right ventricle with concordant atrioventricular connections, a subaortic or doubly-committed (with absence or deficiency of the conal septum) ventricular septal defect, and pulmonary outflow tract obstruction.

LA85.2Y Other specified double outlet right ventricle

LA85.2Z Double outlet right ventricle, unspecified

LA85.3 Double outlet left ventricle

A congenital cardiovascular malformation in which both great arteries arising entirely or predominantly from the morphologically left ventricle.

LA85.4 Common arterial trunk

A congenital cardiovascular malformation in which a single arterial trunk arises from the heart, giving origin sequentially to the coronary arteries, one or more pulmonary arteries, and the systemic arterial circulation.

Inclusions: persistent truncus arteriosus

truncus arteriosus

LA85.40 Common arterial trunk with aortic dominance

A congenital cardiovascular malformation in which a common arterial trunk is associated with an unobstructed aortic arch.

LA85.41 Common arterial trunk with pulmonary dominance and interrupted aortic arch

A congenital cardiovascular malformation in which a common arterial trunk is associated with an interrupted aortic arch.

LA85.4Y Other specified common arterial trunk

LA85.4Z Common arterial trunk, unspecified

LA85.Y Other specified congenital anomaly of an atrioventricular or ventriculo-arterial connection

LA85.Z Congenital anomaly of an atrioventricular or ventriculo-arterial connection, unspecified

LA86 Congenital anomaly of mediastinal vein

A congenital cardiovascular malformation in which there is an abnormality of a mediastinal vein including but not limited to: pulmonary veins, caval veins, coronary sinus, hepatic veins connecting to the heart, brachiocephalic veins, azygos veins, and/or levo-atrial cardinal veins.

LA86.0 Left superior caval vein

A congenital cardiovascular malformation in which there is a left superior caval vein (superior vena cava).

Additional information: unless the code for absent right superior caval vein is used, this term assumes that a right superior caval vein is present and, therefore, there are bilateral superior caval veins with or without a bridging vein.

Coding Note: Unless the code for absent right superior caval vein is used, this term assumes that a right superior caval vein is present and, therefore, there are bilateral superior caval veins with or without a bridging vein.

LA86.1 Unroofed coronary sinus

A congenital cardiovascular malformation in which there is direct communication between the left atrium and the coronary sinus.

Additional information: this term includes partial and complete unroofing of the coronary sinus in the presence or absence of an interatrial communication. If an interatrial communication is present through the coronary sinus orifice then also select the term 'Interatrial communication through coronary sinus orifice'. If a left superior caval vein (superior vena cava) is present then one should also select the term for 'Left superior caval vein (superior vena cava) to left-sided atrium'.

Coding Note: If an interatrial communication is present through the coronary sinus orifice code also interatrial communication through coronary sinus orifice. If a left superior caval vein (superior vena cava) is present code also left superior caval vein (superior vena cava) to left-sided atrium.

LA86.2 Anomalous pulmonary venous connection

A congenital cardiovascular malformation in which one or more pulmonary vein does not connect normally to the morphologically left atrium.

LA86.20 Total anomalous pulmonary venous connection

A congenital cardiovascular malformation in which none of the pulmonary veins connect to the morphologically left atrium.

LA86.21 Partial anomalous pulmonary venous connection

A congenital cardiovascular malformation in which one or more (but not all) of the pulmonary veins connect anomalously to the right atrium or to one or more of its venous tributaries and the remaining pulmonary veins connect to the left atrium.

LA86.22 Scimitar syndrome

A congenital cardiopulmonary malformation with “partial anomalous pulmonary venous connection of Scimitar type” and one or more of the following: hypoplasia of the right lung with bronchial anomalies, dextrocardia, hypoplasia of the right pulmonary artery, lobar lung sequestration, and anomalous systemic arterial supply to the lower lobe of the right lung directly from the aorta or its main branches.

LA86.2Y Other specified anomalous pulmonary venous connection

LA86.2Z Anomalous pulmonary venous connection, unspecified

LA86.3 Congenital pulmonary venous stenosis or hypoplasia

A congenital cardiovascular malformation with a pathologic narrowing of one or more pulmonary veins including diffuse hypoplasia, long segment focal/tubular stenosis and/or discrete stenosis.

LA86.Y Other specified congenital anomaly of mediastinal vein

LA86.Z Congenital anomaly of mediastinal vein, unspecified

LA87 Congenital anomaly of an atrioventricular valve or atrioventricular septum

A congenital cardiovascular malformation in which there is an abnormality of the atrioventricular valve or atrioventricular septum.

LA87.0 Congenital anomaly of tricuspid valve

A congenital cardiovascular malformation in which there is an abnormality of the tricuspid valve.

Inclusions: congenital anomaly of tricuspid subvalvular apparatus

Exclusions: Tricuspid atresia (LA89.1)

LA87.00 Congenital tricuspid regurgitation

A congenital cardiovascular finding in which there is backward flow through the tricuspid valve.

LA87.01 Congenital tricuspid valvar stenosis

A congenital cardiovascular malformation of the tricuspid valve in which there is narrowing or stricture (obstruction to flow).

LA87.02 Dysplasia of tricuspid valve

A congenital cardiovascular malformation of the tricuspid valve, commonly consisting of leaflet thickening and restricted mobility, with normally hinged leaflets.

Coding Note: This diagnosis is not used for patients with Ebstein malformation of tricuspid valve, which is characterised by abnormally hinged tricuspid valve.

Exclusions: Ebstein malformation of tricuspid valve (LA87.03)

LA87.03 Ebstein malformation of tricuspid valve

A congenital cardiovascular malformation of the tricuspid valve and right ventricle that is characterised by incomplete delamination of the septal and inferior (posterior) tricuspid valvar leaflets from the myocardium of the right ventricle, and varying degrees of downward (apical) rotational displacement of the functional annulus.

Additional information: associated cardiac anomalies include an interatrial communication, the presence of accessory conduction pathways and varying degrees of right ventricular outflow tract obstruction, including pulmonary atresia. In the setting of discordant atrioventricular and ventriculo-arterial connections ['Congenitally corrected transposition of great arteries'], 'Ebstein malformation of tricuspid valve' may be present.

LA87.0Y Other specified congenital anomaly of tricuspid valve

LA87.0Z Congenital anomaly of tricuspid valve, unspecified

LA87.1 Congenital anomaly of mitral valve

A congenital cardiac malformation in which there is an abnormality of the mitral valve.

Exclusions: Mitral atresia (LA89.2)

LA87.10 Congenital mitral regurgitation

A congenital cardiovascular finding in which there is backward flow through the mitral valve.

LA87.11 Congenital mitral valvar stenosis

A congenital cardiovascular malformation of the mitral valve in which there is narrowing or stricture of the valvar orifice (obstruction to flow).

LA87.12 Dysplasia of mitral valve

A congenital cardiac malformation that includes any structural abnormality of the mitral valvar leaflet(s), commonly consisting of leaflet thickening and restricted mobility.

LA87.13 Congenital anomaly of mitral subvalvar apparatus

A congenital cardiac malformation in which the mitral chords, chordal attachments, or papillary muscles are abnormal.

LA87.1Y Other specified congenital anomaly of mitral valve

LA87.1Z Congenital anomaly of mitral valve, unspecified

LA87.2 Common atrioventricular junction

A congenital cardiac malformation where both atria connect to a common atrioventricular valve which characteristically has 4 or 5 leaflets including superior and inferior bridging leaflets with a single annulus.

Additional information: the common valve may have one or two major orifices depending on the absence or presence of fusion of the bridging leaflets to each other or the septal crest. The left ventricular zone of apposition between the superior and inferior bridging leaflets is commonly referred to as a "cleft".

LA87.20 Atrioventricular septal defect

A congenital cardiac malformation with a common atrioventricular junction and an atrioventricular septal defect.

Inclusions: AVC - [atrioventricular canal]

LA87.2Y Other specified common atrioventricular junction

LA87.2Z Common atrioventricular junction, unspecified

LA87.Y Other specified congenital anomaly of an atrioventricular valve or atrioventricular septum

LA87.Z Congenital anomaly of an atrioventricular valve or atrioventricular septum, unspecified

LA88 Congenital anomaly of a ventricle or the ventricular septum

A congenital cardiac malformation in which there is an abnormality of a ventricle and/or the ventricular septum. The ventricles include the ventricular inlet, ventricular body and ventricular outflow tract.

LA88.0 Congenital right ventricular outflow tract obstruction

A congenital cardiovascular condition in which the flow through the right ventricular outflow tract (proximal to the valve[s] guarding the outflow from the right ventricle) is blocked or impeded.

Additional information: this code should not be used for obstruction immediately under the arterial valve(s) because specific codes exist for these entities, such as congenital subpulmonary and subaortic stenosis.

LA88.1 Double chambered right ventricle

A congenital cardiovascular malformation in which the right ventricle is divided into two chambers, one inferior including the inlet and trabecular portions of the right ventricle and one superior including the trabecular portion and infundibulum.

Additional information: Double chamber right ventricle is often associated with one or several closing ventricular septal defects. In some cases, the ventricular septal defect is already closed. Double chamber right ventricle is differentiated from the rare isolated infundibular stenosis that develops more superiorly.

LA88.2 Tetralogy of Fallot

A group of congenital cardiovascular malformations with biventricular atrioventricular alignments or connections characterised by anterosuperior deviation of the conal or outlet septum or its fibrous remnant, narrowing or atresia of the pulmonary outflow, a ventricular septal defect of the malalignment type, and biventricular origin of the aorta.

Additional information: tetralogy of Fallot will always have a ventricular septal defect, narrowing or atresia of the pulmonary outflow, aortic override, and most often right ventricular hypertrophy.

LA88.20 Tetralogy of Fallot with absent pulmonary valve syndrome

A congenital cardiovascular malformation that is a variant of tetralogy of Fallot in which the ventriculo-arterial junction of the right ventricle with the pulmonary trunk features an atypical valve with absent or rudimentary leaflets (cusps) that do not coapt.

Additional information: in its usual form there is dilatation of the pulmonary trunk and central right and left pulmonary arteries, which when extreme, is associated with abnormal arborization of lobar and segmental pulmonary artery branches and with compression of the trachea and mainstem bronchi, often with tracheobronchomalacia. The physiologic consequence is usually a combination of variable degrees of both stenosis and regurgitation of the pulmonary valve.

LA88.21 Tetralogy of Fallot with pulmonary atresia

A congenital cardiovascular malformation that is a variant of tetralogy of Fallot in which there is no direct communication between the right ventricle and the pulmonary arterial tree.

Exclusions: Tetralogy of Fallot with pulmonary atresia and systemic-to-pulmonary collateral artery (LA88.22)

LA88.22 Tetralogy of Fallot with pulmonary atresia and systemic-to-pulmonary collateral artery

A congenital cardiovascular malformation that is a variant of tetralogy of Fallot in which there is no direct communication between the right ventricle and the pulmonary arterial tree and there are collateral blood vessels between the systemic and pulmonary arteries.

Coding Note: This morphological abnormality usually is an integral part of other congenital cardiovascular anomalies and does not need to be coded separately. It should be coded as secondary to an accompanying congenital cardiovascular anomaly if the left ventricular hypoplasia is not considered an integral and understood part of the primary congenital cardiovascular diagnosis such as hypoplastic left heart syndrome.

LA88.2Y Other specified tetralogy of Fallot

LA88.2Z Tetralogy of Fallot, unspecified

LA88.3 Congenital left ventricular outflow tract obstruction

A congenital cardiac condition in which the flow through the left ventricular outflow tract (proximal to the valve[s] guarding the outflow from the left ventricle) is blocked or impeded.

This code should not be used for obstruction immediately under the arterial valve such as subaortic stenosis due to fibromuscular shelf or tunnel.

LA88.4 Ventricular septal defect

A congenital cardiac malformation in which there is a hole or pathway between the ventricular chambers.

LA88.40 Trabecular muscular ventricular septal defect

A congenital cardiac malformation in which there is a ventricular septal defect within the trabeculated component of the ventricular septum.

Additional information: the codes specifying defects within the trabecular part of the ventricular septum should not be used to code inlet or outlet muscular defects, as there are specific codes for these entities.

LA88.41 Perimembranous central ventricular septal defect

A congenital cardiovascular malformation in which there is a ventricular septal defect that 1) occupies the space that is usually closed by the interventricular part of the membranous septum, 2) is usually adjacent to the area of fibrous continuity between the leaflets of an atrioventricular valve and an arterial valve, 3) is adjacent to an area of mitral-tricuspid fibrous continuity, and 4) is located at the center of the base of the ventricular mass.

Additional information: This code is used by some as synonymous with the perimembranous, conoventricular, Type II, or the paramembranous defects. Although best used to describe the perimembranous defect that opens centrally at the base of the right ventricle, this term might be used to code perimembranous defects with inlet or outlet extension. It is recommended, however, that the more precise terms be used whenever possible for coding the latter lesions. It should also not be used to code an inlet ventricular septal defect, or the so-called atrioventricular canal ventricular septal defect. More specific terms exist for coding these entities. It is used by some to describe an isolated perimembranous ventricular septal defect without extension, although it is unlikely that perimembranous defects exist in the absence of deficiency of their muscular perimeter. The conoventricular ventricular septal defect with malalignment should be coded as an outlet defect, as should the perimembranous defect opening to the outlet of the right ventricle. Such defects can also extend to become doubly committed and juxta-arterial (conal septal hypoplasia) when there is also fibrous continuity between the leaflets of the arterial valves or when there is a common arterial valve. Specific codes exist for these variants, which ideally should not be coded using this term.

Coding Note: Although best used to describe the perimembranous defect that opens centrally at the base of the right ventricle, this term might be used to code perimembranous defects with inlet or outlet extension. It is recommended, however, that the more precise terms be used whenever possible for coding the latter lesions. This code is used by some as synonymous with the perimembranous, conoventricular, Type II, or the paramembranous defects. It should not be used to code an inlet VSD, or the so-called atrioventricular canal VSD. More specific terms exist for coding these entities. It is used by some to describe an isolated perimembranous VSD without extension, although it is unlikely that perimembranous defects exist in the absence of deficiency of their muscular perimeter. The conoventricular VSD with malalignment should be coded as an outlet defect, as should the perimembranous defect opening to the outlet of the right ventricle. All perimembranous defects, nonetheless, have part of their margins made up of fibrous continuity either between the leaflets of an atrioventricular and an arterial valve or, in the setting of double outlet right ventricle or overriding of the tricuspid valve, by fibrous continuity between the leaflets of the mitral and tricuspid valves. Such defects can also extend to become doubly committed and juxta-arterial (conal septal hypoplasia) when there is also fibrous continuity between the leaflets of the arterial valves or when there is a common arterial valve. Specific codes exist for these variants, which ideally should not be coded using this term.

LA88.42 Ventricular septal defect haemodynamically insignificant

A congenital cardiac malformation in which there is one or more small, clinically insignificant ventricular septal defect(s) in the absence of flow-related cardiac chamber dilation or abnormal elevation of pulmonary arterial pressure.

Additional information: though restrictive ventricular septal defect is listed as a synonym of hemodynamically insignificant VSD, it should be recognised that some pressure restrictive ventricular septal defects will lead to flow-related chamber dilation, and thus would be haemodynamically significant. In such instances, the term haemodynamically insignificant ventricular septal defect should not be coded.

LA88.4Y Other specified ventricular septal defect

LA88.4Z Ventricular septal defect, unspecified

LA88.Y Other specified congenital anomaly of a ventricle or the ventricular septum

LA88.Z Congenital anomaly of a ventricle or the ventricular septum, unspecified

LA89 Functionally univentricular heart

The term “functionally univentricular heart” describes a spectrum of congenital cardiovascular malformations in which the ventricular mass may not readily lend itself to partitioning that commits one ventricular pump to the systemic circulation, and another to the pulmonary circulation.

Additional information: a heart may be functionally univentricular because of its anatomy or because of the lack of feasibility or lack of advisability of surgically partitioning the ventricular mass. Common lesions in this category typically include double inlet right ventricle (DIRV), double inlet left ventricle (DILV), tricuspid atresia, mitral atresia, and hypoplastic left heart syndrome. Other lesions which sometimes may be considered to be a functionally univentricular heart include complex forms of atrioventricular septal defect, double outlet right ventricle, congenitally corrected transposition, pulmonary atresia with intact ventricular septum, and other cardiovascular malformations. Specific diagnostic codes should be used whenever possible, and not the term “functionally univentricular heart”.

LA89.0 Double inlet atrioventricular connection

A congenital cardiovascular malformation with a univentricular atrioventricular connection wherein both atria connect to one ventricle either via two separate atrioventricular valves or a common atrioventricular valve, such that all or nearly all of the total atrioventricular junctional (annular) area is committed to one ventricular chamber.

LA89.1 Tricuspid atresia

A congenital cardiovascular malformation with absence of the tricuspid valvar annulus (connection/junction) or an imperforate tricuspid valve.

LA89.2 Mitral atresia

A congenital cardiovascular malformation with absence of the mitral valvar annulus (connection/junction) or an imperforate mitral valve.

LA89.3 Hypoplastic left heart syndrome

A spectrum of congenital cardiovascular malformations with normally aligned great arteries without a common atrioventricular junction, characterised by underdevelopment of the left heart with significant hypoplasia of the left ventricle including atresia, stenosis, or hypoplasia of the aortic or mitral valve, or both valves, and hypoplasia of the ascending aorta and aortic arch.

LA89.Y Other specified functionally univentricular heart

LA89.Z Functionally univentricular heart, unspecified

LA8A Congenital anomaly of a ventriculo-arterial valve or adjacent regions

A congenital cardiovascular malformation of a ventriculo-arterial valve or its immediate subvalvar and supravalvar regions.

Exclusions: Common arterial trunk (LA85.4)

LA8A.0 Congenital anomaly of pulmonary valve

A congenital malformation of the heart where the pulmonary valve is abnormal.

LA8A.00 Congenital pulmonary valvar stenosis

A congenital cardiovascular malformation of the pulmonary valve in which there is narrowing or stricture causing obstruction to flow.

Additional information: congenital pulmonary valvar stenosis ranges from critical neonatal pulmonic valve stenosis with hypoplasia of the right ventricle to valvar pulmonary stenosis in the infant, child, or adult.

LA8A.01 Congenital pulmonary regurgitation

Congenital cardiovascular malformation of the pulmonary valve allowing backward flow into the ventricle. Congenital pulmonary valve regurgitation may be due to primary annular dilation, prolapse and leaflet underdevelopment.

LA8A.0Y Other specified congenital anomaly of pulmonary valve

LA8A.0Z Congenital anomaly of pulmonary valve, unspecified

LA8A.1 Congenital pulmonary atresia

A congenital cardiovascular malformation in which there is no opening between any ventricle and the pulmonary arterial tree.

Exclusions: Tetralogy of Fallot with pulmonary atresia (LA88.21)

LA8A.10 Pulmonary atresia with intact ventricular septum

A congenital cardiovascular malformation in which there are normally aligned great arteries, no opening between the morphologically right ventricle and the pulmonary trunk, and no ventricular level communication.

Additional information: pulmonary atresia with intact ventricular septum is a duct-dependent congenital malformation that forms a spectrum of lesions including atresia of the pulmonary valve, a varying degree of right ventricle and tricuspid valve hypoplasia, and anomalies of the coronary circulation. A right ventricular dependent coronary artery circulation is present when coronary artery fistulas are associated with a proximal coronary artery stenosis. Associated Ebstein anomaly of the tricuspid valve can be present.

LA8A.1Y Other specified congenital pulmonary atresia

LA8A.1Z Congenital pulmonary atresia, unspecified

LA8A.2 Congenital anomaly of aortic valve

A congenital cardiovascular malformation where the aortic valve is abnormal.

LA8A.20 Congenital aortic valvar stenosis

A congenital cardiovascular malformation of the aortic valve in which there is narrowing or stricture (obstruction to flow).

Additional information: 'Congenital aortic valvar stenosis' arises most commonly as a result of partial or complete fusion of one or more commissures, or is due to dysplasia of one or more aortic cusps. These congenital malformations of the aortic valve may not be initially obstructive but may become stenotic later in life due to leaflet thickening, poor relative growth and-or calcification. It is not until the congenitally malformed aortic valve is or becomes stenotic that this term should be used.

Exclusions: Congenital subaortic stenosis (LA8A.5)

that in hypoplastic left heart syndrome (LA89.3)

LA8A.21 Congenital aortic regurgitation

Congenital cardiovascular malformation of the aortic valve allowing backward flow into the ventricle.

Additional information: congenital aortic regurgitation is rare as an isolated entity. Aortic regurgitation is more commonly seen with other associated congenital cardiac anomalies.

LA8A.22 Bicuspid aortic valve

A congenital abnormality of the heart where the aortic valve has two commissures and two separate leaflets because of fusion or absence of one of the commissures

LA8A.23 Aortic valvar atresia

A congenital cardiovascular malformation in which there is no orifice of the aortic valve.

Additional information: aortic valvar atresia will most often not be coded independently, as it is frequently included within the 'Hypoplastic left heart syndrome' code as part of this spectrum of cardiovascular malformations. However, there is a small subset of patients with aortic valve atresia who have a well developed left ventricle and mitral valve and a large ventricular septal defect (nonrestrictive or restrictive).

Coding Note: Aortic valve atresia will most often be coded under the hypoplastic left heart syndrome/complex diagnostic codes since it most often occurs as part of a spectrum of cardiovascular malformations. However, there is a small subset of patients with aortic valve atresia who have a well developed left ventricle and mitral valve and a large ventricular septal defect (nonrestrictive or restrictive).

LA8A.24 Unicuspid aortic valve

A congenital cardiovascular malformation in which the aortic valve has a single commissure and a single or functionally single leaflet (cusp)

LA8A.2Y Other specified congenital anomaly of aortic valve

LA8A.2Z Congenital anomaly of aortic valve, unspecified

LA8A.3 Congenital supravalvar aortic stenosis

A congenital cardiovascular malformation with narrowing of the aorta at the level of the sinotubular junction which may extend into the ascending aorta.

Additional information: 'Congenital supravalvar aortic stenosis' is described as three forms: an hourglass deformity, a fibrous membrane, and a diffuse narrowing of the ascending aorta. Supravalvar aortic stenosis may involve the coronary artery ostia, and the aortic leaflets may be tethered. The coronary arteries can become tortuous and dilated due to elevated pressures and early atherosclerosis may ensue.

Exclusions: Congenital aortic valvar stenosis (LA8A.20)

LA8A.4 Aneurysm of aortic sinus of Valsalva

A congenital cardiovascular malformation in which there is dilation of one or more sinuses of Valsalva.

Additional information: the sinus of Valsalva is defined as that portion of the aortic root between the aortic root annulus and the sinotubular junction. Sinus of Valsalva aneurysm most commonly originates from the right sinus, less commonly from the non-coronary sinus and rarely from the left sinus (<5%). The aneurysm may rupture into an adjacent chamber or site (right atrium, right ventricle, left atrium, left ventricle, pulmonary artery, pericardium) and in this case should be coded specifically (‘Ruptured aortic sinus of Valsalva aneurysm’). This is to be distinguished from aortic root dilation associated with connective tissue disorders and aortopathies.

LA8A.5 Congenital subaortic stenosis

Exclusions: Subaortic stenosis due to fibromuscular tunnel (LA8A)

Subaortic stenosis due to fibromuscular shelf (LA8A)

LA8A.6 Congenital subpulmonary stenosis

A congenital cardiovascular malformation associated with narrowing within the outflow tract supporting the pulmonary valve.

Additional information: subvalvar (infundibular) pulmonary stenosis is a narrowing of the outflow tract of the ventricle immediately below the pulmonic valve. This term should preferably be used in the setting of abnormal ventriculo-arterial connections, such as double outlet ventricle. Although subvalvar pulmonary stenosis is a type of right ventricular outflow tract obstruction if the ventriculo-arterial connections are normal, in this setting 'Congenital right ventricular outflow tract obstruction' should be used. Subvalvar pulmonary stenosis is also a type of left ventricular outflow tract obstruction in the setting of discordant ventriculo-arterial connections; this term should be used when the obstruction is only apparent immediately below the pulmonary valve, otherwise the term 'Congenital left ventricular outflow tract obstruction' should be used

Exclusions: Double chambered right ventricle (LA88.1)

LA8A.Y Other specified congenital anomaly of a ventriculo-arterial valve or adjacent regions

LA8A.Z Congenital anomaly of a ventriculo-arterial valve or adjacent regions, unspecified

LA8B Congenital anomaly of great arteries including arterial duct

A congenital cardiovascular malformation of the great arteries (aorta, pulmonary trunk [main pulmonary artery], branch pulmonary arteries) or the arterial duct (ductus arteriosus).

Exclusions: Common arterial trunk (LA85.4)

LA8B.0 Congenital aortopulmonary window

A congenital cardiovascular malformation in which there is side-to-side continuity of the lumens of the ascending aorta and pulmonary trunk in association with separate aortic and pulmonary valves or their atretic remnants.

Additional information: side-to-side continuity of the lumens of the aorta and pulmonary arterial tree, which is distinguished from common arterial trunk (truncus arteriosus) by the presence of two arterial valves or their atretic remnants, and involvement of the pulmonary trunk (main pulmonary artery).

Inclusions: Aortic septal defect

Aortopulmonary window

LA8B.1 Congenital anomaly of pulmonary arterial tree

A congenital cardiovascular malformation of the pulmonary trunk (main pulmonary artery) and/or branch pulmonary arteries (right, left, and ramifications).

Inclusions: Aberrant pulmonary artery

Anomaly of pulmonary artery

LA8B.2 Congenital anomaly of aorta or its branches

A congenital cardiovascular malformation of the aorta and/or its branches.

LA8B.20 Aberrant origin of right subclavian artery

A congenital cardiovascular malformation in which the right subclavian artery arises distal to the left subclavian artery in the setting of a left aortic arch.

LA8B.21 Coarctation of aorta

A congenital cardiovascular malformation in which there is a discrete luminal narrowing of the junction between the aortic arch and the descending aorta.

Additional information: 'Coarctation of the aorta' generally indicates a narrowing of the descending thoracic aorta just distal to the left subclavian artery. However, the term may also be accurately used to refer to a region of narrowing anywhere in the thoracic or abdominal aorta.

LA8B.22 Interrupted aortic arch

A congenital cardiovascular malformation in which there is an absence of luminal continuity between the ascending and descending aorta.

Additional information: this includes luminal atresia with discontinuity between the aortic segments and also luminal atresia with fibrous continuity between the aortic segments. Interrupted aortic arch is defined as the loss of luminal continuity between the ascending and descending aorta. In most cases, blood flow to the descending thoracic aorta is through a patent arterial duct, and there is a large ventricular septal defect. Arch interruption is further defined by site of interruption.

In type A, interruption is distal to the left subclavian artery; in type B, interruption is between the left carotid and left subclavian arteries; and in type C, interruption occurs between the innominate and left carotid arteries.

LA8B.2Y Other specified congenital anomaly of aorta or its branches

LA8B.2Z Congenital anomaly of aorta or its branches, unspecified

LA8B.3 Tracheo-oesophageal compressive syndrome

A congenital cardiovascular malformation which causes compression of the trachea and/or the oesophagus.

LA8B.4 Patent arterial duct

A congenital cardiovascular finding in which the arterial duct (ductus arteriosus) is open beyond the normal age of spontaneous closure.

Additional information: a patent arterial duct is a vascular arterial connection between the thoracic aorta and the pulmonary artery. Most commonly, a patent arterial duct has its origin from the descending thoracic aorta, just distal and opposite the origin of the left subclavian artery. The insertion of the duct is most commonly into the very proximal left pulmonary artery at its junction with the main pulmonary artery. Origination and insertion sites can be variable, however.

LA8B.Y Other specified congenital anomaly of great arteries including arterial duct

LA8B.Z Congenital anomaly of great arteries including arterial duct, unspecified

LA8C Congenital anomaly of coronary artery

A congenital cardiovascular malformation of a coronary artery. This includes absence of a coronary, anomalous origin or course, dilation or stenosis, and fistulas.

Additional information: congenital anomalies of the coronary venous system should not be included here but rather under 'Congenital anomaly of mediastinal systemic vein'.

LA8C.0 Anomalous origin of coronary artery from pulmonary arterial tree

A congenital cardiovascular malformation in which a coronary artery origin is the pulmonary trunk or one of its branches. Although the most common of these malformations involves the left coronary artery arising from the pulmonary trunk (main pulmonary artery) rather than from the aorta, occasionally the right coronary artery, the circumflex, or both coronary arteries may arise from any of the central pulmonary arteries.

LA8C.1 Anomalous aortic origin or course of coronary artery

A congenital cardiovascular malformation in which the origin and/or course of a coronary artery is abnormal.

This is where coronary "anomalies" in the presence of discordant ventriculo-arterial connections should be coded.

LA8C.2 Congenital coronary arterial fistula

A congenital cardiovascular malformation in which a coronary artery communicates, through an anomalous channel, with a cardiac chamber or with any segment of the systemic or pulmonary circulation.

Additional information: this communication may be simple and direct or may be tortuous and dilated. In order of frequency the involved coronary artery is the right, the left and, rarely, both coronary arteries. Occasionally multiple fistulas are present.

Inclusions: congenital coronary fistula to pulmonary artery

Exclusions: anomalous origin of coronary artery from pulmonary arterial tree (LA8C.0)

LA8C.Y Other specified congenital anomaly of coronary artery

LA8C.Z Congenital anomaly of coronary artery, unspecified

LA8D Congenital pericardial anomaly

A congenital cardiovascular malformation in which there is a structural and/or functional abnormality of the pericardium.

Additional information: congenital pericardium anomalies include congenital absence of pericardium and pericardial cysts. This would include antenatal pericardial effusion and congenital tumours of the serous pericardium.

LA8E Congenital anomaly of atrial septum

A congenital cardiovascular malformation in which there is an abnormality of the atrial septum.

LA8E.0 Patent oval foramen

A congenital cardiovascular finding in which there is a small interatrial communication (or potential communication) confined to the region of the oval fossa (fossa ovalis) characterised by no deficiency of the primary atrial septum (septum primum) and a normal limbus with no deficiency of the septum secundum (superior interatrial fold).

LA8E.1 Atrial septal defect within oval fossa

A congenital cardiovascular malformation in which there is an interatrial communication confined to the region of the oval fossa (fossa ovalis), most commonly due to a deficiency of the primary atrial septum (septum primum) but deficiency of the septum secundum (superior interatrial fold) may also contribute. Source: ISNPCHD

LA8E.2 Sinus venosus defect

A congenital cardiovascular malformation in which there is a caval vein (vena cava) and/or pulmonary vein (or veins) that overrides the atrial septum or the septum secundum (superior interatrial fold) producing an interatrial or anomalous veno-atrial communication.

Additional information: although the term sinus venosus atrial septal defect is commonly used, the lesion is more properly termed a sinus venosus communication because, while it functions as an interatrial communication, this lesion is not a defect of the atrial septum.

LA8E.3 Interatrial communication through coronary sinus orifice

A congenital cardiovascular malformation in which there is a communication between the left atrium and the coronary sinus allowing interatrial communication through the coronary sinus ostium.

Additional information: 'Interatrial communication through coronary sinus orifice' may or may not be associated with a persistent left superior caval vein (superior vena cava). This occurs in the absence of the coronary sinus (total unroofing of the coronary sinus) or partial unroofing of the coronary sinus.

LA8E.Y Other specified congenital anomaly of atrial septum

LA8E.Z Congenital anomaly of atrial septum, unspecified

LA8F Congenital anomaly of right atrium

A congenital cardiovascular malformation in which there is an abnormality of the right atrium.

LA8G Congenital anomaly of left atrium

A congenital cardiovascular malformation in which there is an abnormality of the left atrium.

LA8G.0 Divided left atrium

A congenital cardiac malformation in which there is a partition that divides the left atrium into a posterosuperior chamber that receives some or all of the pulmonary veins and an antero-inferior chamber that communicates with the left atrial appendage and atrioventricular junction (usually the mitral valve).

Additional information: in differentiating 'Divided left atrium' from 'Congenital supravalvar or intravalvar mitral ring', in the latter, the antero-inferior compartment contains only the mitral valvar orifice.

LA8G.Y Other specified congenital anomaly of left atrium

LA8G.Z Congenital anomaly of left atrium, unspecified

LA8Y Other specified structural developmental anomaly of heart or great vessels

LA8Z Structural developmental anomaly of heart or great vessels, unspecified

LA90 Structural developmental anomalies of the peripheral vascular system

Exclusions: Congenital anomaly of coronary artery (LA8C)

Congenital anomaly of pulmonary arterial tree (LA8B.1)

haemangioma and lymphangioma (2E81)

Congenital retinal aneurysm (LA13.5)

LA90.0 Capillary malformations

This is a vascular anomaly consisting of superficial and deep dilated capillaries in the skin which produce a reddish to purplish discoloration of the skin.

Exclusions: Macrocephaly - Cutis Marmorata Telangiectatica Congenita (LD2F.1)

Coded Elsewhere: Developmental capillary vascular malformations of the skin (LC50)

LA90.00 Hereditary haemorrhagic telangiectasia

Rendu-Osler-Weber disease, also called hereditary haemorrhagic telangiectasia (HHT), is a genetic disorder of angiogenesis leading to arteriovenous dilatations: cutaneo-mucosal haemorrhagic telangiectasias and visceral shunting.

LA90.0Y Other specified capillary malformations

LA90.0Z Capillary malformations, unspecified

LA90.1 Lymphatic malformations

Lymphatic malformations (LM), formerly referred to by the term lymphangioma, are malformations of the lymphatic system which result in obstructed lymphatic drainage. There are two types of LM: macrocystic LM (including cystic hygroma/lymphangioma) and tissue-infiltrating microcystic LM (lymphangioma circumscriptum). The macro and microcystic forms of LM may occur in association.

Coded Elsewhere: Primary lymphoedema (BD93.0)

LA90.10 Macrocystic lymphatic malformation

A condition caused by failure of the lymphatic system to correctly develop during the antenatal period. This condition is characterised by large, soft, smooth clear masses under normal or bluish skin. This condition may be associated with cellulitis, bleeding within the malformation, pain, or leakage of lymphatic fluid internally.

LA90.11 Microcystic lymphatic malformation

Microcystic lymphatic malformations consist of clusters of dilated lymphatic vessels which have developed without connection to the systemic lymphatic circulation. They present with grouped clear or haemorrhagic vesicles anywhere on the skin or mucous membrane.

Inclusions: Lymphangioma circumscriptum

Exclusions: Circumscribed lymphatic malformation (LA90.10)

LA90.12 Lymphatic malformations of certain specified sites

LA90.13 Cystic hygroma in fetus

Development abnormalities of the lymphoid system that occur at sites of lymphatic-venous connection, most commonly in the posterior neck but may be anterior and may extend into chest. Frequently associated with karyotypic abnormalities, various malformation syndromes, and several teratogenic agents. When diagnosed prenatally, the overall prognosis is poor. Cystic hygroma diagnosed after birth is usually associated with a good prognosis.

LA90.1Y Other specified lymphatic malformations

LA90.1Z Lymphatic malformations, unspecified

LA90.2 Peripheral venous malformations

Coded Elsewhere: Developmental venous malformations involving the skin (LC51)

Blue rubber bleb naevus syndrome (LC51)

LA90.20 Vein of Galen aneurysm

Vein of Galen aneurysmal malformation (VGAM) is a congenital vascular malformation characterised by dilation of the embryonic precursor of the vein of Galen. It is a sporadic lesion that occurs during embryogenesis. Cardiac insufficiency of variable severity is the principle manifestation that leads to detection of the malformation in newborns.

LA90.21 Anomalous portal venous connection

LA90.2Y Other specified peripheral venous malformations

LA90.2Z Peripheral venous malformations, unspecified

LA90.3 Peripheral arteriovenous malformations

This is a peripheral, abnormal connection between arteries and veins, bypassing the capillary system. This vascular anomaly is widely known because of its occurrence in the central nervous system, but can appear in any location.

Inclusions: congenital arteriovenous varices NOS

Exclusions: acquired arteriovenous aneurysm (BD52.1)

Coded Elsewhere: Arteriovenous malformation of cerebral vessels (8B22.40)

LA90.30 Portal vein-hepatic artery fistula

LA90.31 Arteriovenous malformation of precerebral vessels

LA90.32 Uterine arteriovenous malformations

LA90.3Y Other specified peripheral arteriovenous malformations

LA90.3Z Peripheral arteriovenous malformations, unspecified

LA90.4 Peripheral arterial malformations

This is a peripheral lesion with a direct connection between an artery and a vein, without an intervening capillary bed, but with an interposed nidus of dysplastic vascular channels in between.

Coded Elsewhere: Hereditary cerebrovascular diseases (8B22.C)

LA90.40 Congenital renal artery stenosis

This is the congenital narrowing of the renal artery, most often caused by atherosclerosis or fibromuscular dysplasia. This narrowing of the renal artery can impede blood flow to the target kidney.

LA90.41 Congenital precerebral nonruptured aneurysm

LA90.42 Congenital cerebral nonruptured aneurysm

This is a cerebrovascular disorder in which weakness in the wall of a cerebral artery or vein causes a localised dilation or ballooning of the blood vessel (nonruptured).

Coded Elsewhere: Familial cerebral saccular aneurysm (8B22.6)

LA90.4Y Other specified peripheral arterial malformations

LA90.4Z Peripheral arterial malformations, unspecified

LA90.5 Pulmonary arteriovenous fistula

A congenital cardiovascular malformation in which there is an abnormal, direct connection between a pulmonary artery and pulmonary vein or left atrium without an intervening capillary bed.

LA90.Y Other specified structural developmental anomalies of the peripheral vascular system

LA90.Z Structural developmental anomalies of the peripheral vascular system, unspecified

LA9Y Other specified structural developmental anomalies of the circulatory system

LA9Z Structural developmental anomalies of the circulatory system, unspecified

Structural developmental anomalies of the diaphragm, abdominal wall or umbilical cord (BlockL2‑LB0)

Any condition caused by failure of the diaphragm, abdominal wall and umbilical cord to correctly develop during the antenatal period.

Exclusions: Prune belly syndrome (LD2F.10)

LB00 Structural developmental anomalies of diaphragm

LB00.0 Congenital diaphragmatic hernia

Congenital diaphragmatic hernia is a posterolateral defect of the diaphragm that allows passage of abdominal viscera into the thorax, leading to respiratory insufficiency and persistent pulmonary hypertension with high mortality.

Exclusions: Congenital hiatus hernia (LB13.1)

LB00.1 Absence of diaphragm

LB00.Y Other specified structural developmental anomalies of diaphragm

LB00.Z Structural developmental anomalies of diaphragm, unspecified

LB01 Omphalocele

Omphalocele is an embryopathy classified in the group of abdominal celosomias and is characterised by a large hernia of the abdominal wall, centred on the umbilical cord, in which the protruding viscera are protected by a sac.

Exclusions: Umbilical hernia (DD53)

LB02 Gastroschisis

Gastroschisis is a congenital abdominal wall defect characterised by viscera protruding, without a covering sac, from the fetal abdomen on the right lateral base of the umbilicus.

LB03 Structural developmental anomalies of umbilical cord

Any condition caused by failure of the umbilical cord to correctly develop during the antenatal period.

Coded Elsewhere: Umbilical cord haemangioma (2E81.00)

Fetus or newborn affected by short umbilical cord (KA03.20)

Fetus or newborn affected by long umbilical cord (KA03.21)

Developmental anomalies of the umbilicus (EC50)

LB03.0 Allantoic duct remnants or cysts

Any condition caused by failure of the umbilical cord to correctly develop during the antenatal period. These conditions are characterised by cysts or remnants of allantoic tissue within the umbilical cord, the umbilicus, or the urachus.

LB03.1 Single umbilical cord artery

A single umbilical artery arising from the either the allantoic arterial system (Type I), or vitelline artery (Type II). And has been associated with renal anomalies

LB03.Y Other specified structural developmental anomalies of umbilical cord

LB03.Z Structural developmental anomalies of umbilical cord, unspecified

LB0Y Other specified structural developmental anomalies of the diaphragm, abdominal wall or umbilical cord

LB0Z Structural developmental anomalies of the diaphragm, abdominal wall or umbilical cord, unspecified

Structural developmental anomalies of the digestive tract (BlockL2‑LB1)

Any condition caused by failure of the digestive tract to correctly develop during the antenatal period.

Coded Elsewhere: Genetic or developmental disorders involving lips or oral mucosa (DA02.0)

LB10 Structural developmental anomalies of salivary glands or ducts

Any condition caused by failure of the salivary glands and ducts to correctly develop during the antenatal period.

LB11 Congenital diverticulum of pharynx

A condition caused by failure of the pharynx to correctly develop during the antenatal period. This condition may present with difficulty swallowing, or may be asymptomatic. Confirmation is through observation of a diverted pharynx by imaging.

Exclusions: pharyngeal pouch syndrome (LD44.N0)

LB12 Structural developmental anomalies of oesophagus

Any congenital defect of oesophagus that results from interference with the normal growth and differentiation of the fetus. Such defects can arise at any stage of embryonic development, vary greatly in type and severity, and are caused by a wide variety of determining factors, including genetic mutations, chromosomal aberrations, teratogenic agents, and environmental factors. Most developmental defects are apparent at birth, especially any structural malformation, but some becomes evident later.

Coded Elsewhere: Bronchopulmonary foregut malformation (LA74.Y)

LB12.0 Congenital oesophageal web or ring

A rare form of incomplete oesophageal obstruction due to a developmental defect of the primitive foregut that presents as a mucosal lesion forming an incomplete diaphragm. Symptoms (apparent from birth) include dysphagia, regurgitation, and choking.

Exclusions: Oesophageal web (DA20.2)

LB12.1 Atresia of oesophagus

Oesophageal atresia encompasses a group of congenital anomalies with an interruption in the continuity of the oesophagus, with or without persistent communication with the trachea. In 86% of cases there is a distal tracheooesophageal fistula, in 7% of cases there is no fistulous connection, while in 4% of cases there is a tracheooesophageal fistula without atresia. The remaining cases are made up of patients with OA with proximal, or both proximal and distal, tracheooesophageal fistula.

LB12.10 Atresia of oesophagus with oesophagobronchial fistula

LB12.1Y Other specified atresia of oesophagus

LB12.1Z Atresia of oesophagus, unspecified

LB12.2 Oesophageal fistula without atresia

This is a birth defect (congenital anomaly) of oesophagus, and one type of EA/TEF, namely isolated "H"-shaped atresia. Tracheoesophageal fistula in which there is no oesophageal atresia because the oesophagus is continuous to the stomach. Fistula is present between the oesophagus and the trachea. Incidence of TE fistula without atresia varies between 1 -11% of oesophageal malformations.

LB12.3 Congenital stenosis or stricture of oesophagus

A form of incomplete oesophageal obstruction due to a developmental defect of the primitive foregut. Abnormal narrowing of the oesophagus occurs most often at the junction of the middle and lower thirds. Clinical manifestations, apparent 2 to 3 weeks after birth, include dysphagia and progressive vomiting.

LB12.4 Congenital diverticulum of oesophagus

A very rare congenital diverticulum which is typically located just above the cricopharyngeal junction. It is usually asymptomatic unless complicated by an inflammatory process. If the diverticulum compresses the trachea or is associated with oesophageal stenosis or fistula, the symptoms of stridor, progressive dysphagia, respiratory distress, severe choking, and regurgitation may be present from birth.

Inclusions: Congenital oesophageal pouch

oesophageal pouch

Exclusions: Diverticulum of oesophagus, acquired (DA20.1)

LB12.5 Congenital dilatation of oesophagus

This is a congenital abnormal enlargement of the lower portion of the oesophagus, as seen in patients with achalasia.

LB12.Y Other specified structural developmental anomalies of oesophagus

LB12.Z Structural developmental anomalies of oesophagus, unspecified

LB13 Structural developmental anomalies of stomach

Any congenital defect of stomach that results from interference with the normal growth and differentiation of the fetus. Such defects can arise at any stage of embryonic development, vary greatly in type and severity, and are caused by a wide variety of determining factors, including genetic mutations, chromosomal aberrations, teratogenic agents, and environmental factors. Most developmental defects are apparent at birth, especially any structural malformation, but some becomes evident later.

Coded Elsewhere: Gastric volvulus (DA40.2)

LB13.0 Congenital hypertrophic pyloric stenosis

A not uncommon congenital malformation of the stomach of unknown cause in which there is hypertrophy and hyperplasia of the circular muscle of the pylorus. Symptoms of gastric outlet obstruction usually appear between the third and sixth weeks of life. The anomaly is manifested by intermittent vomiting (which increases in frequency and becomes projectile), regurgitation, weight loss, dehydration, electrolyte imbalance, sometimes a small palpable pyloric mass, and visible peristaltic contractions across the epigastrium; there may also be jaundice. Some cases appear to be familial (possibly of autosomal dominant inheritance).

LB13.1 Congenital hiatus hernia

Congenital diaphragmatic hernia is an embryopathy which is defined by the absence of development of all or part of the diaphragmatic dome that results in the presence of abdominal viscera in the thorax, whit compression of the homolateral lung and impaired development of the contralateral lung.

Inclusions: Congenital displacement of cardia through oesophageal hiatus

Exclusions: Congenital diaphragmatic hernia (LB00.0)

Diaphragmatic hernia (DD50.0)

LB13.2 Congenital antral web

LB13.Y Other specified structural developmental anomalies of stomach

LB13.Z Structural developmental anomalies of stomach, unspecified

LB14 Structural developmental anomalies of duodenum

Any congenital defect of duodenum that results from interference with the normal growth and differentiation of the fetus. Such defects can arise at any stage of embryonic development, vary greatly in type and severity, and are caused by a wide variety of determining factors, including genetic mutations, chromosomal aberrations, teratogenic agents, and environmental factors. Most developmental defects are apparent at birth, especially any structural malformation, but some becomes evident later.

LB15 Structural developmental anomalies of small intestine

Congenital gross anatomical structural defect of small intestine that results from interference with the normal growth and differentiation of the fetus, which may be inherited genetically, acquired during gestation, or inflicted during parturition.

LB15.0 Meckel diverticulum

A congenital abnormality characterised by the outpouching or sac formation in the ILEUM. It is a remnant of the embryonic YOLK SAC in which the VITELLINE DUCT failed to close. During early gestation, the omphalomesenteric or vitelline duct connects the fetal yolk sac to the primitive gut. By 7-8 weeks of gestation, this duct is normally completely obliterated. A Meckel diverticulum results when this structure fails to resorb completely.

LB15.1 Atresia of small intestine

Jejunoileal atresias and stenoses are major causes of neonatal intestinal obstruction. Atresia refers to a congenital obstruction with complete occlusion of the intestinal lumen. It accounts for 95% of obstructions. Four types of jejunoileal atresias are described. They can range from having a small area of blockage or web to missing large sections of the intestines.

Intestinal atresia is one of the most frequent causes of bowel obstruction in the newborn. The ileal atresia is more common than jejunal atresia, and multiple foci are more common than isolated atresia. The most accepted theory regarding the etiology of jejunoileal atresia is that of an intrauterine vascular accident resulting in necrosis of the affected segment.

Stenosis, on the other hand, refers to a partial occlusion with incomplete obstruction and accounts for the remaining 5% of cases. A stenosis has an intact mesentery and is a localised narrowing of the bowel. No loss of continuity of the lumen exists.

Inclusions: Congenital absence of small intestine

Congenital stenosis of small intestine

LB15.2 Congenital short bowel

Short bowel syndrome is a condition in which nutrients are not properly absorbed due to a congenital defect where a large part of the small intestine is missing .

LB15.3 Congenital diverticulitis of small intestine

This refers to a clinical entity characterised by the presence of sac-like congenital herniations in the wall of the small intestine, in which the pouches of small intestine (diverticula) become infected or inflamed.

LB15.4 Congenital diverticulosis of small intestine

This refers to a condition characterised by the presence of congenital multiple sack-like mucosal herniations called diverticula through weak points in the wall or lining of the small intestine. Most people with diverticulosis do not have any discomfort or symptoms. However, some people may experience pain or discomfort in the abdomen, bloating, and bleeding.

LB15.5 Congenital diverticulum of small intestine

This refers to a morphological condition in which there is single small congenital pouch in the lining of the small intestine, bulging outward through a weak spot.

LB15.Y Other specified structural developmental anomalies of small intestine

LB15.Z Structural developmental anomalies of small intestine, unspecified

LB16 Structural developmental anomalies of large intestine

Any congenital defect of large intestine that results from interference with the normal growth and differentiation of the fetus. Such defects can arise at any stage of embryonic development, vary greatly in type and severity, and are caused by a wide variety of determining factors, including genetic mutations, chromosomal aberrations, teratogenic agents, and environmental factors. Most developmental defects are apparent at birth, especially any structural malformation, but some becomes evident later.

Exclusions: Congenital atresia of rectum (LB17)

Persistent cloaca (LB17.2)

Congenital atresia of anus (LB17)

Coded Elsewhere: Meconium ileus without perforation (KB87.2)

LB16.0 Congenital absence, atresia or stenosis of large intestine

Colonic atresia is a congenital intestinal malformation resulting in a non-latent segment of the colon and characterised by lower intestinal obstruction manifesting with abdominal distention and failure to pass meconium in newborns.

Exclusions: Congenital atresia of rectum (LB17)

Congenital absence of rectum (LB17)

LB16.1 Hirschsprung disease

This is a developmental anomaly affecting the intestinal tract characterised by congenital absence of myenteric ganglion cells (aganglionosis) in a segment of the large bowel. Due to the absence of intrinsic innervation of the muscle layers of the affected segment, there is a loss of motor function. This results in an abnormally large or dilated colon (congenital megacolon) with intestinal occlusion or constipation. This condition becomes evident shortly after birth.

LB16.2 Immature ganglionosis of large intestine

When the number of ganglion cells is normal but the ganglion cells are prominently immature, the disease is referred to as immature ganglionosis or immaturity of ganglia.

LB16.3 Congenital hypoganglionosis of large intestine

The number and size of ganglion cells are small at birth. The size of ganglion cells tends to increase over time, but because their numbers do not increase the symptoms of dysmotility do not improve.

Source: http://www.ncbi.nlm.nih.gov/pubmed/17161202

Exclusions: Acquired hypoganglionosis of large intestine (DB32.3)

LB16.Y Other specified structural developmental anomalies of large intestine

LB16.Z Structural developmental anomalies of large intestine, unspecified

LB17 Structural developmental anomalies of anal canal

LB17.0 Anorectal malformations

Anorectal malformations (ARMs) are birth defects (due to alterations in embryo development of hindgut or proctodeum) where the anus and rectum (the lower end of the digestive tract) do not develop properly. They occur in approximately 1 in 5000 live births. These comprise a wide spectrum of diseases, which can affect boys and girls, and involve the distal anus and rectum as well as the urinary and genital tracts. Several abnormalities can occur, including the following: A membrane may be present over the anal opening; The rectum may not connect to the anus (imperforate anus); The rectum may connect to a part of the urinary tract or the reproductive system through an abnormal passage called a fistula. The classification of ARMs is mainly based on the position of the rectal pouch relative to the puborectal sling, the presence or absence of fistulas, and the types and locations of the fistulas. The following classification is according to the level of the atretic rectal cul-de-sac with respect to the pubococcygeal line (the radiological landmark for the upper border or the levator ani muscle).

LB17.1 Ectopic anus

While children with imperforate or obviously mislocated anus are identified in the newborn period, some children with a very mild abnormality may escape identification until after the newborn period. This mild mislocation of the anus has been termed anterior ectopic anus. Anterior ectopic anus is different from imperforate anus with perineal fistula in that the anal opening is usually of normal size, and only mildly misplaced. Most of these children come to medical attention due to severe constipation.

LB17.2 Persistent cloaca

A congenital anomaly in which the intestinal, urinary, and reproductive ducts open into a common cavity, a result of the failure of the urorectal septum to form during prenatal development. They occur exclusively in girls and comprise the most complex defect in the spectrum of anorectal malformations.

LB17.3 Cloacal exstrophy

Rare and complex anorectal and genitourinary malformation in which rectum, vagina and urinary tract share a common everted orifice, accompanied by an omphalocele and an imperforate anus.

Exstrophy of the cloaca is a well-known malformation that includes the persistence and the exstrophy of a cloaca that receives ureters, ileum and a rudimentary hindgut. Cloacal exstrophy is a severe birth defect wherein much of the abdominal organs (the bladder and intestines) are exposed. It often causes the splitting of both male and female genitalia (specifically, the penis and clitoris respectively), and the anus is occasionally sealed.

LB17.4 Perineal groove

The perineal groove describes a normal vestibule but with a groove extending from the vestibule to the anus, which is both normal sized and positioned.

LB17.Y Other specified structural developmental anomalies of anal canal

LB17.Z Structural developmental anomalies of anal canal, unspecified

LB18 Congenital anomalies of intestinal fixation

A condition caused by failure of the intestines to correctly develop during the antenatal period. This condition may present with intermittent abdominal pain, vomiting, or diarrhoea. Confirmation is through observation of intestinal rotation by imaging.

LB1Y Other specified structural developmental anomalies of the digestive tract

LB1Z Structural developmental anomalies of the digestive tract, unspecified

Structural developmental anomalies of the liver, biliary tract, pancreas or spleen (BlockL2‑LB2)

Any condition caused by failure of the liver, biliary tract, pancreas and spleen to correctly develop during the antenatal period.

LB20 Structural developmental anomalies of gallbladder, bile ducts or liver

Any condition caused by failure of the gallbladder, bile ducts and liver to correctly develop during the antenatal period.

LB20.0 Structural developmental anomalies of liver

Exclusions: Non-alcoholic fatty liver disease (DB92)

Coded Elsewhere: Biliary atresia (LB20.21)

LB20.00 Fibropolycystic liver disease

Coded Elsewhere: Choledochal cyst (LB20.20)

LB20.0Y Other specified structural developmental anomalies of liver

LB20.0Z Structural developmental anomalies of liver, unspecified

LB20.1 Structural developmental anomalies of gallbladder

LB20.10 Agenesis, aplasia or hypoplasia of gallbladder

LB20.1Y Other specified structural developmental anomalies of gallbladder

LB20.1Z Structural developmental anomalies of gallbladder, unspecified

LB20.2 Structural developmental anomalies of bile ducts

Coded Elsewhere: Congenital bronchobiliary fistula (LA74.Y)

LB20.20 Choledochal cyst

Inclusions: Congenital bile duct dilatation

LB20.21 Biliary atresia

Biliary atresia is a rare disease characterised by an inflammatory biliary obstruction of unknown origin that presents in the neonatal period. It is the most frequent surgical cause of cholestatic jaundice in this age group. Untreated, this condition leads to cirrhosis and death within the first years of life.

LB20.22 Congenital stenosis or stricture of bile ducts

LB20.23 Structural developmental anomalies of cystic duct

LB20.24 Accessory bile duct

LB20.2Y Other specified structural developmental anomalies of bile ducts

LB20.2Z Structural developmental anomalies of bile ducts, unspecified

LB20.Y Other specified structural developmental anomalies of gallbladder, bile ducts or liver

LB20.Z Structural developmental anomalies of gallbladder, bile ducts or liver, unspecified

LB21 Structural developmental anomalies of pancreas

This is structural development of the ducts of the pancreas in which a single pancreatic duct is not formed, but rather remains as two distinct dorsal and ventral ducts.

LB21.0 Annular pancreas

Annular pancreas is a distinct form of duodenal atresia in which the head of the pancreas forms a ring around the second portion of the duodenum. During the neonatal period, the clinical picture is dominated by epigastric distension with vomiting, which is nonbilious as the obstruction is usually supra-vaterian. Chromosomal abnormalities are present in one-third of cases of annular pancreas, with trisomy 21 (followed by trisomy 18 and 13) being the most frequently detected anomaly. Annular pancreas is an embryopathy resulting from an anomaly occurring early (towards the fourth week) in development.

LB21.1 Pancreas divisum

This is a congenital anomaly in the anatomy of the ducts of the pancreas in which a single pancreatic duct is not formed, but rather remains as two distinct dorsal and ventral ducts.

LB21.2 Accessory pancreas

Accessory pancreas is an asymptomatic embryopathy characterised by the presence of pancreatic tissue in other sites of the body such as the splenic pedicle, gonadic pedicles, intestinal mesentery, duodenum wall, upper jejunum, or, more rarely, the gastric wall, ileum, gallbladder or spleen.

LB21.3 Agenesis-aplasia of pancreas

This refers to the failure of an organ to develop during embryonic growth and development due to the absence of primordial tissue of the pancreas.

LB21.4 Partial agenesis of pancreas

Partial agenesis of the pancreas is characterised by the congenital absence of a critical mass of pancreatic tissue. The severity of the disease depends on the amount of functional pancreatic tissue present. Pancreatic agenesis is commonly associated with other malformations, in particular pancreaticobiliary duct anomalies, leading to acute or chronic pancreatitis, hyperglycaemia (50% of cases), or, more rarely, polysplenia.

LB21.5 Hypoplasia of pancreas

LB21.Y Other specified structural developmental anomalies of pancreas

LB21.Z Structural developmental anomalies of pancreas, unspecified

LB22 Structural developmental anomalies of spleen

Any condition caused by failure of the spleen to correctly develop during the antenatal period.

Exclusions: isomerism of atrial appendages (with asplenia or polysplenia) (BlockL3‑LA8)

LB22.0 Congenital asplenia

A condition caused by failure of the spleen to develop during the antenatal period. This condition may present with pneumococcal sepsis. Confirmation is through verification of no spleen by imaging.

LB22.1 Polysplenia

A condition caused by the development of supernumerary spleens during the antenatal period. This condition may present with cardiac defects, anomalies in abdominal organs, or may be asymptomatic.

LB22.2 Ectopic spleen

LB22.Y Other specified structural developmental anomalies of spleen

LB22.Z Structural developmental anomalies of spleen, unspecified

LB2Y Other specified structural developmental anomalies of the liver, biliary tract, pancreas or spleen

LB2Z Structural developmental anomalies of the liver, biliary tract, pancreas or spleen, unspecified

Structural developmental anomalies of the urinary system (BlockL2‑LB3)

Any condition caused by failure of the urinary system to correctly develop during the antenatal period.

LB30 Structural developmental anomalies of kidneys

Any condition caused by failure of the kidneys to correctly develop during the antenatal period.

LB30.0 Renal agenesis or other reduction defects of kidney

A series of conditions resulting in reduced kidney function including a congenital absence of both kidneys

LB30.00 Renal agenesis

A condition where one or both kidneys does not form (or develop) in utero.

LB30.0Y Other specified renal agenesis or other reduction defects of kidney

LB30.0Z Renal agenesis or other reduction defects of kidney, unspecified

LB30.1 Renal dysplasia

A condition characterised by abnormal development of one or both kidneys.

Exclusions: Autosomal dominant polycystic kidney disease (GB81)

LB30.2 Congenital single renal cyst

A single cyst in a kidney, noted in utero or from birth. No other structural abnormality of the kidney or urinary tract noted.

LB30.3 Renal tubular dysgenesis

Abnormal renal development and kidney formation secondary to an underlying condition or exposure.

LB30.4 Oligomeganephronia

Oligomeganephronic renal hypoplasia is a severe developmental defect of both kidneys characterised by a reduced number of nephrons (the functional unit of the kidney), hypertrophic glomeruli with diameters twice the normal size, hypertrophic tubules and thickening of Bowman's capsule, occurring in the absence of a urinary tract malformation.

LB30.5 Accessory kidney

LB30.6 Fusion anomaly of kidneys

The embryological, incomplete fusion of renal lobules and/or kidneys

LB30.60 Lobulated kidney

Any condition caused by incomplete fusion of the developing renal lobules during the antenatal period. This condition may be asymptomatic.

LB30.61 Fused pelvic kidney

A condition caused by failure of the kidneys to correctly develop during the antenatal period. This condition is characterised by the presence of a single kidney, along the midline of the body. This condition may present with kidney stones, hydronephrosis, kidney infection, haematuria, or may be asymptomatic. Confirmation is through observation of a fused kidney by imaging.

LB30.62 Horseshoe kidney

Horseshoe kidney is the most frequent renal fusion anomaly and is characterised by the union of the inferior poles of the two kidneys through an isthmus. Horseshoe kidney is in fact an anatomical anomaly rather than a disease, but it does lead to predisposition to certain conditions such as hydronephrosis, nephrolithiasis or pyelonephritis. One third of individuals with horseshoe kidney are asymptomatic, with the anomaly being discovered fortuitously during a radiological examination. Urogenital or renal vessel anomalies may be associated with the condition. For cases requiring treatment, various therapeutic approaches are available and choice of treatment depends on the associated pathology.

LB30.6Y Other specified fusion anomaly of kidneys

LB30.6Z Fusion anomaly of kidneys, unspecified

LB30.7 Ectopic or pelvic kidney

A birth defect characterised by an abnormally positioned kidney; may be asymptomatic or result in urine blockage, infection or kidney stones

Inclusions: Congenital displaced kidney

Malrotation of kidney

LB30.8 Medullary sponge kidney

A condition characterised by cystic or saccular dilatations of the medullary collecting ducts seen with radiocontrast filling. A predisposition to stones and associated often with renal tubular acidosis. There is no clear genetic predisposition.

LB30.9 Multicystic renal dysplasia

A condition characterised by abnormal development of the kidney, specifically in which the abnormal kidney does not form a reniform structure but rather, a collection of non-communicating cysts, with no renal functional tissue.

LB30.Y Other specified structural developmental anomalies of kidneys

LB30.Z Structural developmental anomalies of kidneys, unspecified

LB31 Structural developmental anomalies of urinary tract

Any condition caused by failure of the urinary tract to correctly develop during the antenatal period.

Coded Elsewhere: Allantoic duct remnants or cysts (LB03.0)

Persistent urogenital sinus (LB42.Y)

LB31.0 Congenital hydronephrosis

Congenital hydronephrosis is a renal urinary disease characterised by distension and dilation of the renal pelvis and calyces secondary to various congenital obstructive malformations of the kidneys and urinary tract that can evolve to renal atrophy.

LB31.1 Congenital primary megaureter

Congenital primary megaureter is an idiopathic condition in which the bladder and bladder outlet are normal but the ureter is dilated to some extent. It may be obstructed, refluxing or unobstructed and not refluxing.

LB31.2 Fetal lower urinary tract obstruction

A disease caused by partial or complete obstruction of the urethra, during the antenatal period. This disease can present with enlarged bladder, oligohydramnios, or pulmonary hypoplasia. Confirmation is through observation of the obstruction by imaging.

LB31.3 Exstrophy of urinary bladder

Bladder exstrophy (or classic bladder exstrophy) is a congenital genitourinary malformation belonging to the spectrum of the exstrophy-epispadias complex and is characterised by an evaginated bladder plate, epispadias and an anterior defect of the pelvis, pelvic floor and abdominal wall.

Inclusions: Ectopia vesicae

Extroversion of bladder

LB31.4 Congenital diverticulum of urinary bladder

A condition caused by failure of the bladder to correctly develop. This condition is characterised by weakness in the bladder wall through which some of the lining of the bladder protrudes. This condition may present with urinary tract infections, difficulty voiding, or abdominal fullness. This condition may also be asymptomatic.

LB31.5 Duplication of urethra

A condition caused by failure of the urethra to correctly develop during the antenatal period. This condition is characterised by the presence of a second passage from the bladder. This condition may present with double urinary stream, urination from the anus, or may be asymptomatic. Confirmation is through observation of a second urethra by imaging.

LB31.6 Congenital megalourethra

A condition caused by failure of the penile corpora cavernosa and spongiosa to correctly develop during the antenatal period. This condition is characterised by dilatation of the penile urethra. This condition may present with poor stream, swelling of the penis, megacystis, oligohydramnios, renal failure, or pulmonary hypoplasia.

LB31.7 Megacystis-megaureter

Megacystic-megaureter syndrome describes the presence of a massive primary non-obstructive vesicoureteral reflux and a large capacity, smooth, thin walled bladder due to the continual recycling of refluxed urine. Recurrent urinary infections are commonly associated with this condition.

LB31.8 Atresia or stenosis of ureter

A condition caused by blockage or narrowing of the ureter due to failure to correctly develop during the antenatal period. This condition may present with bladder outlet obstruction, low amniotic fluid volume, pulmonary hypoplasia, megacystis, hydroureter, hydronephrosis, or renal dysplasia.

LB31.9 Agenesis of ureter

A condition caused by failure of the ureter to develop during the antenatal period. Confirmation verification that one or more ureters are missing by imaging.

Inclusions: Absent ureter

LB31.A Duplication of ureter

A condition caused by failure of the ureter to correctly develop during the antenatal period, resulting in incorrect connection of the ureter to the kidney. This condition may present with ureteroureteral reflux, or ureteropelvic junction obstruction of the lower pole of the kidney in the case of incomplete duplication. Complete duplication may present with vesicoureteral reflux, ectopic ureterocele, or ectopic ureteral insertion. Confirmation is through observation of two ureters on one side by imaging.

Inclusions: Double ureter

Accessory ureter

LB31.B Malposition of ureter

A condition caused by failure of the ureter to correctly develop during the antenatal period, resulting in partial or complete duplication of the ureter. This condition may present with hydronephrosis, urinary tract infection, or incontinence in females. Confirmation is through observation of an incorrectly positioned ureter by imaging.

LB31.C Congenital absence of bladder or urethra

Any condition caused by failure of both the bladder and the urethra to develop during the antenatal period. This condition may result in fetal death, or sepsis and sever complications in cases of live births.

LB31.D Congenital vesico-uretero-renal reflux

A condition caused by failure of the ureter to develop correctly during the antenatal period. This condition may present with urinary tract infection, or may be asymptomatic.

LB31.Y Other specified structural developmental anomalies of urinary tract

LB31.Z Structural developmental anomalies of urinary tract, unspecified

LB3Y Other specified structural developmental anomalies of the urinary system

LB3Z Structural developmental anomalies of the urinary system, unspecified

Structural developmental anomalies of the female genital system (BlockL2‑LB4)

Exclusions: Disorders of sex development leading to sexual ambiguity (LD2A)

LB40 Structural developmental anomalies of vulva

A deformation established before birth of an anatomical structure of the vulva.

LB40.0 Absence of vulva

This is a birth defect or congenital abnormality of the female genitourinary system that manifests itself in the absence of the vulva.

LB40.1 Embryonic cyst of vulva

Remnant tissue from embryological development of the development of the pelvic organs presenting as a closed fluid sac in or on the tissue of the vulva.

LB40.2 Fusion of labia

A condition of the labia commonly affecting females between 6 months and 6 years of age, caused by skin irritation during infancy. This condition is characterised by the sealing of the labia minor (usually completely) due to a thin membrane that seals the entrance to the vagina, leaving a very small gap for urination.

LB40.Y Other specified structural developmental anomalies of vulva

LB40.Z Structural developmental anomalies of vulva, unspecified

LB41 Structural developmental anomalies of clitoris

A deformation established before birth of an anatomical structure of the clitoris.

LB41.0 Agenesis of clitoris

LB41.1 Duplication of clitoris

An anatomical anomaly present at birth in which there are two clitoral structures present.

LB41.2 Clitoromegaly

LB41.Y Other specified structural developmental anomalies of clitoris

LB41.Z Structural developmental anomalies of clitoris, unspecified

LB42 Structural developmental anomalies of vagina

A deformation established before birth of an anatomical structure of the vagina.

LB42.0 Absence of vagina

A condition of the genitourinary system affecting females, caused by an abnormality arising during the antenatal period. This condition is characterised by vaginal agenesis.

LB42.1 Septate vagina

A condition of the genitourinary system affecting females, caused by the absence of Mullerian duct fusion during the antenatal period. This condition is characterised by a transverse of longitudinal septum, partitioning the vagina into two parts. This condition may also present with dyspareunia, abnormal vaginal bleeding, or is asymptomatic. Confirmation is by imaging.

Exclusions: doubling of vagina with doubling of uterus and cervix (LB44.3)

LB42.2 Congenital rectovaginal fistula

A condition of the genitourinary system affecting females, caused by an abnormality arising during the antenatal period. This condition is characterised by the formation of an abnormal passage between the rectum and the vagina.

Exclusions: Persistent cloaca (LB17.2)

LB42.3 Tight hymenal ring

A condition of the vagina, caused by determinants arising during the antenatal period. This condition is characterised by tightening of the hymen and stenosis of the external opening of the vagina, and dyspareunia.

Inclusions: Rigid hymen

Tight introitus

Exclusions: Imperforate hymen (LB42.4)

LB42.4 Imperforate hymen

A condition in which the hymen, the membrane that surrounds or partially covers the external vaginal opening, is harder than normal or is complete and sealed without any opening into the vaginal vault.

LB42.5 Stricture or atresia of vagina

A condition of the vagina, caused by an abnormality arising during the antenatal period. This condition is characterised by stenosis and occlusion of the vaginal opening.

Exclusions: Postoperative adhesions of vagina (GC70)

LB42.Y Other specified structural developmental anomalies of vagina

LB42.Z Structural developmental anomalies of vagina, unspecified

LB43 Structural developmental anomalies of cervix uteri

LB43.0 Embryonic cyst of cervix

A condition of the cervix, caused by a cluster of cells that have formed a closed sac or structures left behind from development during the antenatal period. This condition is characterised by air, fluid, or semi-solid material surrounded by a distinct membrane of cells with abnormal appearance and behaviour.

LB43.1 Agenesis or aplasia of cervix

A condition of the cervix, caused by the absence of primordial tissue development during the antenatal period. This condition is characterised by improper or lack of development of the cervix.

LB43.Y Other specified structural developmental anomalies of cervix uteri

LB43.Z Structural developmental anomalies of cervix uteri, unspecified

LB44 Structural developmental anomalies of uterus, except cervix

Absent vagina, absent or hypoplastic uterus (sometimes with areas of functional endometrium), and typical or hypoplastic fallopian tubes, mostly associated with typical ovaries and urologic abnormalities and skeletal abnormalities. Presenting symptoms and signs typically include primary amenorrhea, absent vagina, and typical breast and pubic hair development.

LB44.0 Agenesis or aplasia of uterine body

A condition of the uterus, caused by the absence of primordial tissue development during the antenatal period. This condition is characterised by improper or lack of development of the uterine body.

LB44.1 Hypoplasia of uterus

LB44.2 Unicornuate uterus

A uterine malformation where the uterus is formed from one only of the paired Müllerian ducts while the other Müllerian duct does not develop or develops only in a rudimentary fashion.

LB44.3 Bicornuate uterus

A condition of the uterus, caused by malformation in the development of the uterus during the antenatal period. This condition is characterised by a uterus with a bifurcated cephalo, and a unitary caudal part. Confirmation is by imaging.

LB44.4 Septate uterus

Longitudinal septum in uterus, subclassified as complete or partial

LB44.5 Congenital fistulae between uterus and digestive and urinary tracts

A condition caused by abnormal tissue development during the antenatal period. This condition is characterised by the formation of an abnormal passage between the uterus, digestive, and urinary tracts.

LB44.6 Uterovaginal malformation due to diethylstilbestrol syndrome

Fetal diethylstilbestrol syndrome is characterised by a group of symptoms likely to occur in children and grandchildren of a woman who was treated while pregnant with diethylstilbestrol (DES). The drug is a synthetic nonsteroidal oestrogen, used in the US until 1971 and in Europe until 1978 to try and prevent miscarriage, premature delivery, and other pregnancy complications. It has been estimated that 25% of female fetuses exposed to DES in utero during the first trimester have subsequently developed genital tract anomalies including vaginal adenosis, cervical malformations, vaginal septae, uterine cavity anomalies, or fallopian tube anomalies causing subsequent fertility problems.

LB44.Y Other specified structural developmental anomalies of uterus, except cervix

LB44.Z Structural developmental anomalies of uterus, except cervix, unspecified

LB45 Structural developmental anomalies of ovaries, fallopian tubes or broad ligaments

LB45.0 Congenital absence of ovary

A condition of the ovary, caused by determinants arising during the antenatal period. This condition is characterised by a female born with fewer than two ovaries.

Exclusions: Turner syndrome (LD50.0)

LB45.1 46,XX gonadal dysgenesis

Karyotype 46 XX; Gonads: gonadal dysgenesis (streak gonads); Phenotype female with symptoms like primary amenorrhea, hypergonadotrophic hypogonadism, normal stature and no other abnormalities.

LB45.2 Developmental ovarian cyst

A condition in which an individual is born with a benign, functional cyst, or cysts, on one or more ovaries which result from enlargement of otherwise normal follicles during third trimester or early neonatal period.

LB45.3 Congenital torsion of ovary

A condition of the ovary, caused by determinants arising during the antenatal period. This condition is characterised by a partial or complete rotation of the ovary, an occlusion to the venous or arterial blood supply of the ovary, severe lower abdominal pain that may radiate to the back, pelvis and thigh, and nausea, vomiting, diarrhoea or constipation.

LB45.4 Accessory ovary

A condition of the ovary, caused by determinants arising during the antenatal period. This condition is characterised by excess ovarian tissue situated near an anatomically correct ovary, which may or may not be connected to the original ovarian tissue.

LB45.5 Congenital absence of fallopian tube

A condition of the fallopian tube, caused by determinants arising during the antenatal period. This condition is characterised by a female born with fewer than two fallopian tubes.

LB45.6 Atresia of fallopian tube

A condition of the fallopian tube, caused by determinants arising during the antenatal period. This condition is characterised by unilateral or bilateral closure or absence of the fallopian tube(s), commonly within the proximal isthmic or proximal ampullary segments.

LB45.7 Accessory fallopian tube

A condition of the fallopian tube, caused by determinants arising during the antenatal period. This condition is characterised by the duplication of one or more fallopian tubes, commonly attached to the ampullary segment.

LB45.8 Embryonic cyst of fallopian tube

A condition of the Fallopian tube, caused by the overgrowth of pelvic tissue during the antenatal period. This condition is characterised by air, fluid, or semi-solid material surrounded by a distinct membrane of cells with abnormal appearance and behaviour.

Inclusions: Fimbrial cyst

LB45.9 Embryonic cyst of broad ligament

Remnant tissue from embryological development of the development of the pelvic organs presenting as a closed fluid sac on the broad ligament.

Inclusions: epoophoron cyst

parovarian cyst

LB45.Y Other specified structural developmental anomalies of ovaries, fallopian tubes or broad ligaments

LB45.Z Structural developmental anomalies of ovaries, fallopian tubes or broad ligaments, unspecified

LB4Y Other specified structural developmental anomalies of the female genital system

LB4Z Structural developmental anomalies of the female genital system, unspecified

Structural developmental anomalies of the male genital system (BlockL2‑LB5)

Any condition affecting the male genital system, caused by determinants arising during the antenatal period. These conditions are characterised by structural developmental anomalies.

Exclusions: Disorders of sex development leading to sexual ambiguity (LD2A)

LB50 Micropenis or penis agenesis

A condition caused by reduced androgen production during the antenatal period. This condition is characterised by an absent, or unusually small penis. Confirmation is by measuring the length of a dorsal erectile penis.

LB51 Anorchia or microorchidia

A disorder affecting males, caused by an abnormality occurring in sex development during the antenatal period. This disorder is characterised by individuals who are born with absence of the testes, or with testes that are deficient in size and function. Confirmation is by physical examination, identification of low testosterone levels but elevated follicle stimulating hormone and luteinizing hormone levels in a blood sample, or imaging.

Coded Elsewhere: Testicular agenesis (LD2A.2)

LB52 Cryptorchidism

A disorder affecting males, caused by an abnormality occurring in sex development during the antenatal period. This disorder is characterised by the absence of one or both testes from the scrotum. This disorder may also present with reduced fertility, psychological implications, or increased risk of testicular germ cell tumours. Confirmation is by imaging, karyotyping, or identification of male sex hormones in a blood sample.

Exclusions: Retractile testis migrans (MF42)

LB52.0 Ectopic testis

A condition of the testis, caused by determinants arising during the antenatal period. This condition is characterised by the abnormal location of the testis away from the normal line of decent such as in the superficial inguinal pouch, perineal, abdominal, pelvic, crural, penile or femoral positions, and with normal testis and spermatic cord anatomy. Confirmation is by diagnostic laparoscopy to rule out an intra-abdominal, inguinal or absent/vanishing testis.

LB52.1 Undescended testicle, unilateral

The situation in which one of the two testicles in a male has not transitioned from the abdomen, and therefore appears absent from the scrotum.

LB52.2 Undescended testicle, bilateral

The situation in which both testicles in a male have not transitioned from the abdomen, and therefore appear absent from the scrotum.

LB52.Y Other specified cryptorchidism

LB52.Z Cryptorchidism, unspecified

LB53 Hypospadias

Any condition of the urethra affecting males, caused by determinants arising during the antenatal period. These conditions are characterised by a malformation of the urethra and an abnormally placed urinary meatus.

Exclusions: Epispadias (LB55)

LB53.0 Hypospadias, balanic

A condition of the urethra affecting males, caused by determinants arising during the antenatal period. This condition is characterised by a malformation of the urethra and an abnormally placed urinary meatus that opens at the site of the frenulum. This condition may also present with an incomplete foreskin that forms a hood.

LB53.00 Hypospadias, coronal

A condition of the urethra affecting males, caused by determinants arising during the antenatal period. This condition is characterised by a malformation of the urethra and the ventral side of the penis, leading to an abnormally placed urinary meatus that opens in the ventral portion of the coronal sulcus. This condition may also present with an incomplete foreskin that forms a hood.

LB53.01 Hypospadias, glandular

A condition of the urethra affecting males, caused by determinants arising during the antenatal period. This condition is characterised by a malformation of the urethra and the ventral side of the penis, leading to an abnormally placed urinary meatus that opens at the site of the frenulum. This condition may also present with an incomplete foreskin that forms a hood.

LB53.0Y Other specified hypospadias, balanic

LB53.0Z Hypospadias, balanic, unspecified

LB53.1 Hypospadias, penile

A condition of the urethra affecting males, caused by determinants arising during the antenatal period. This condition is characterised by a malformation of the urethra and the ventral side of the penis and an abnormally placed urinary meatus that opens along the shaft of the penis. This condition may also present with an incomplete foreskin that forms a hood.

LB53.2 Hypospadias, penoscrotal

A condition of the urethra affecting males, caused by determinants arising during the antenatal period. This condition is characterised by a malformation of the urethra and the ventral side of the penis, leading to an abnormally placed urinary meatus that opens where the shaft of the penis meets the scrotum. This condition may also present with an incomplete foreskin that forms a hood.

LB53.3 Hypospadias, scrotal

A condition caused by determinants arising during the antenatal period. This condition is characterised by a malformation of the urethra and the ventral side of the penis, leading to an abnormally placed urinary meatus that opens on the scrotum. This condition may also present with an incomplete foreskin that forms a hood.

LB53.4 Hypospadias, perineal

A condition of the urethra affecting males, caused by determinants arising during the antenatal period. This condition is characterised by a malformation of the urethra and the ventral side of the penis and an abnormally placed urinary meatus that opens in the perineum. This condition may also present with an incomplete foreskin that forms a hood.

LB53.Y Other specified hypospadias

LB53.Z Hypospadias, unspecified

LB54 Congenital chordee

A condition caused by the development of fibrous bands of tissue along the corpus spongiosum and shortening of the ventral skin during the antenatal period. This condition is characterised by ventral or dorsal curvature of the head of the penis at the junction with the shaft, most apparent during erection. This condition may also present with hypospadias.

LB55 Epispadias

Epispadias is a congenital genitourinary malformation belonging to the spectrum of the exstrophy-epispadias complex and is characterised in males by an ectopic meatus or a mucosal strip in place of the urethra on the penile dorsum and in females by bifid clitoris and a variable cleft of the urethra.

Exclusions: Hypospadias (LB53)

LB56 Bifid scrotum

A condition caused by failure of the scrotum to correctly develop during the antenatal period resulting in incomplete fusion of the labioscrotal folds. This condition in characterised by a deep midline cleft in the scrotum. This condition may be asymptomatic.

LB57 Agenesis of vas deferens

A condition of the vas deferens affecting males, caused by determinants arising during the antenatal period. This condition is characterised by the unilateral or bilateral absence of the vas deferens, azoospermia, and infertility.

LB58 Polyorchidism

A condition of the testes, caused by determinants arising during the antenatal period. This condition is characterised by the presence of more than two testicles. Confirmation is by imaging.

LB59 Hypoplasia of testis or scrotum

A condition caused by incomplete development of the testis and scrotum during the antenatal period. This condition is characterised by a decrease or destruction in the number or size of cells testis and scrotal tissue.

LB5Y Other specified structural developmental anomalies of the male genital system

LB5Z Structural developmental anomalies of the male genital system, unspecified

Structural developmental anomalies of the breast (BlockL2‑LB6)

A deformation established before birth of an anatomical structure of the breast or breast tissue.

LB60 Breast aplasia

A condition caused by failure of the breast to develop during the antenatal period. This condition is characterised by a total lack of breast tissue. This condition may also present with absence of the nipple or pectoral muscle.

LB61 Absent nipple

A condition caused by failure of the nipple to develop during the antenatal period. This condition is characterised by a total lack of a nipple. This condition may also present with absence of the breast or pectoral muscle.

LB62 Supernumerary breasts

A condition caused by failure of the breasts to correctly develop during the antenatal period. This condition is characterised by supernumerary breasts, with or without nipples. This condition may be asymptomatic.

LB63 Accessory nipple

A condition caused by development of supernumerary nipples during the antenatal period. This condition is characterised by the presence of nipples or nipple-like structures additional to the normal two. This condition may be asymptomatic.

Inclusions: Supernumerary nipple

LB6Y Other specified structural developmental anomalies of the breast

LB6Z Structural developmental anomalies of the breast, unspecified

Structural developmental anomalies of the skeleton (BlockL2‑LB7)

A deformation established before birth of an anatomical structure of one or more bones.

LB70 Structural developmental anomalies of cranium

Any condition caused by failure of the cranium to correctly develop during the antenatal period.

Coded Elsewhere: Wide cranial sutures of newborn (KD31)

LB70.0 Craniosynostosis

Craniosynostosis consists of premature fusion of one or more cranial sutures, resulting in an abnormal head shape. It can be divided in several subgroups; the major different types are primary vs secondary craniosynostosis and isolated vs syndromic craniosynostosis.

Inclusions: Imperfect fusion of skull

LB70.00 Plagiocephaly

Isolated synostotic plagiocephaly is a form of nonsyndromic craniosynostosis characterised by premature fusion of one coronal or lambdoid suture leading to skull deformity and facial asymmetry.

LB70.0Y Other specified craniosynostosis

LB70.0Z Craniosynostosis, unspecified

LB70.1 Wormian bones

LB70.2 J-shaped sella turcica

LB70.3 Macrocephaly

A condition characterised by above normal head size.

Coding Note: Code aslo the casusing condition

LB70.Y Other specified structural developmental anomalies of cranium

LB70.Z Structural developmental anomalies of cranium, unspecified

LB71 Structural developmental anomalies of facial bones

Any condition caused by failure of the facial bones to correctly develop during the antenatal period.

Exclusions: Facial clefts (LA51)

Otomandibular dysplasia (LD2F.16)

Agnathia (BlockL2‑LA3)

Micrognathia (DA0E.00)

LB71.0 Hypotelorism

A condition caused by failure of the facial bones to correctly develop during the antenatal period. This condition is characterised by lower than normal distance between the eyes.

LB71.1 Hypertelorism

A condition caused by failure of the facial bones to correctly develop during the antenatal period. This condition is characterised by higher than normal distance between the eyes.

LB71.Y Other specified structural developmental anomalies of facial bones

LB71.Z Structural developmental anomalies of facial bones, unspecified

LB72 Structural developmental anomalies of shoulder girdle

Any condition caused by failure of the shoulder girdle to correctly develop during the antenatal period.

LB72.0 Cervical rib

LB72.1 Sprengel deformity

A condition caused by failure of the pectoral girdle to correctly develop during the antenatal period. This condition is characterised by abnormal descent, and altered position and anatomy of the scapula. This condition may present with muscle hypoplasia.

LB72.2 Deformation of scapula

LB72.Y Other specified structural developmental anomalies of shoulder girdle

LB72.Z Structural developmental anomalies of shoulder girdle, unspecified

LB73 Structural developmental anomalies of spine or bony thorax

Any condition caused by failure of the spine and bony thorax to correctly develop during the antenatal period.

LB73.0 Occult spinal dysraphism

Inclusions: Spina bifida occulta

Cryptomerorachischisis

Exclusions: meningocele (spinal) (LA02)

Spina bifida aperta (LA02.1)

Spina bifida cystica (LA02.0)

LB73.1 Structural developmental anomalies of chest wall

Any condition caused by failure of the chest wall to correctly develop during the antenatal period.

LB73.10 Poland syndrome

Poland syndrome is characterised by a unilateral absence or hypoplasia of the pectoralis major muscle (most frequently involving the sternocostal portion), and a variable degree of ipsilateral hand anomalies, including symbrachydactyly.

LB73.11 Bifid rib

LB73.12 Accessory rib

A condition caused by failure of the ribs to correctly develop during the antenatal period. This condition is characterised by a supernumerary rib arising for a cervical or lumbar vertebra. This condition may present with thoracic outlet syndrome, or may be asymptomatic.

LB73.13 Structural developmental anomalies of sternum

Any condition caused by failure of the sternum to correctly develop during the antenatal period.

LB73.1Y Other specified structural developmental anomalies of chest wall

LB73.1Z Structural developmental anomalies of chest wall, unspecified

LB73.2 Structural developmental anomalies of spine

Any condition caused by failure of the spine to correctly develop during the antenatal period.

LB73.20 Klippel-Feil anomaly

Klippel-Feil syndrome is characterised by improper segmentation of cervical segments resulting in congenitally fused cervical vertebrae.

Inclusions: Cervical fusion syndrome

LB73.21 Occipitalisation of atlas

A condition caused by failure of the atlas and occiput to correctly develop during the antenatal period. This condition is characterised by fusion of the atlas to the base of the occiput. This condition may present with headache, suboccipital stiffness, restricted motion, or dizziness. Confirmation is through observation of the fusion by imaging.

LB73.22 Atlanto-axial instability or subluxation

A condition caused by bony or ligamentous abnormality of the upper spinal column. This condition is characterised by excessive movement at the junction between C1 and C2 vertebrae. This condition may present with impaired rotation of the neck, neurological difficulties, or may be asymptomatic.

LB73.23 Aplasia or hypoplasia of the odontoid process of axis

LB73.24 Segmentation anomalies of vertebrae

Any condition caused by failure of the vertebrae to correctly develop during the antenatal period. These conditions are characterised by an abnormal number of fully developed vertebrae. Confirmation is through verification of absent or improperly formed vertebrae by imaging.

LB73.25 Congenital scoliosis due to congenital bony malformation

A condition caused by malformation of the ribs or spine. This condition is characterised by abnormal curving of the spine.

LB73.26 Sacralization of the last lumbar vertebra

LB73.27 Lumbarisation of the first sacral vertebra

LB73.28 Sacrum agenesis or hypoplasia

LB73.29 Caudal appendage

A condition caused by development of a malformation on the lower back during the antenatal period. This condition is characterised by a cutaneous protrusion superior to the buttocks. This condition may be associated with occult spinal dysraphism.

Exclusions: Caudal regression sequence (LD2F.1)

LB73.2A Congenital spondylolisthesis

A condition caused by vertebral malformation, which allows the vertebra to slip forward over the sacrum. This condition may present with lower back pain, or may be asymptomatic.

Exclusions: acquired spondylolisthesis (FA84)

acquired spondylolysis (FA81)

LB73.2Y Other specified structural developmental anomalies of spine

LB73.2Z Structural developmental anomalies of spine, unspecified

LB73.Y Other specified structural developmental anomalies of spine or bony thorax

LB73.Z Structural developmental anomalies of spine or bony thorax, unspecified

LB74 Structural developmental anomalies of pelvic girdle

Any condition caused by failure of the pelvic girdle to correctly develop during the antenatal period.

Exclusions: Clicking hip (ME80)

LB74.0 Developmental dysplasia of hip

A condition caused by failure of the hip to correctly develop during the antenatal period. This condition is characterised by slippage of the hip from the socket. This condition may present with outward turning of the leg, reduced movement on one side of the body, shortness of one leg, uneven skin folds on thigh or buttocks, walking difficulties, or inward rounding of the lower back.

LB74.1 Congenital subluxation of hip

LB74.2 Unstable hip

A condition caused by failure of the hip joint to correctly develop during the antenatal period. This condition is characterised by looseness of the hip joint. This condition may present with dislocation, multidirectional intra-operative instability, abductor insufficiency, or neuromuscular disability.

LB74.3 Congenital coxa vara

A condition caused by failure of the hip joint to correctly develop during the antenatal period. This condition is characterised by a decrease in the femoral neck-shaft angle. This condition may present with a shortened leg, or a limp.

LB74.4 Congenital coxa valga

A condition caused by failure of the hip joint to correctly develop during the antenatal period. This condition is characterised by an increase in the femoral neck-shaft angle.

LB74.5 Wide symphysis pubis

LB74.Y Other specified structural developmental anomalies of pelvic girdle

LB74.Z Structural developmental anomalies of pelvic girdle, unspecified

LB75 Brachydactyly

Brachydactyly ('short digits') is a general term that refers to disproportionately short fingers and toes, and forms part of the group of limb malformations characterised by bone dysostosis. The various types of isolated brachydactyly are rare, except for types A3 and D.

LB75.0 Brachydactyly of fingers

A condition caused by failure of the fingers to correctly develop during the antenatal period. This condition is characterised by below normal finger length.

LB75.1 Brachydactyly of toes

A condition caused by failure of the toes to correctly develop during the antenatal period. This condition is characterised by below normal toe length.

LB75.2 Symbrachydactyly of hands or feet

A condition caused by failure of the digits to correctly develop during the antenatal period. This condition is characterised by short digits, which may be webbed. This condition may also present with missing digits, shortened metacarpals, or short limb sections.

LB75.Y Other specified brachydactyly

LB75.Z Brachydactyly, unspecified

LB76 Triphalangeal thumb

A condition caused by failure of the thumb to correctly develop during the antenatal period. This condition is characterised by a long, finger-like thumb with three phalanges instead of two. Isolated triphalangeal thumbs may be associated with genetic abnormality in the 7q36 region.

LB77 Hyperphalangy

LB78 Polydactyly

Any condition caused by development of supernumerary fingers during the antenatal period.

LB78.0 Polydactyly of the thumb

A condition caused by development of supernumerary thumbs during the antenatal period.

LB78.1 Polysyndactyly

Polysyndactyly is a form of preaxial polydactyly of fingers characterised by the presence of a thumb showing the mildest degree of duplication, being broad, bifid or with radially deviated distal phalanx. Syndactyly of various degrees of third-and-fourth fingers is occasionally present. Two forms have been characterised: unilateral and bilateral.

LB78.2 Postaxial polydactyly of fingers

A condition caused by development of supernumerary fingers during the antenatal period. This condition is characterised by fifth digit duplications.

LB78.3 Polydactyly of toes

Any condition caused by development of supernumerary toes during the antenatal period.

LB78.Y Other specified polydactyly

LB78.Z Polydactyly, unspecified

LB79 Syndactyly

A condition caused by failure of the longitudinal interdigital necrosis that normally separates the digits during the antenatal period. This condition is characterised by the presence of two or more digits that are fused together.

Coded Elsewhere: Polysyndactyly (LB78.1)

LB79.0 Fused fingers

Inclusions: complex syndactyly of fingers with synostosis

LB79.1 Webbed fingers

A condition caused by failure of the longitudinal interdigital necrosis that normally separates the fingers to during the antenatal period. This condition is characterised by the presence of two or more fingers that are fused together.

Inclusions: Simple syndactyly of fingers without synostosis

LB79.2 Fused toes

Inclusions: Complex syndactyly of toes with synostosis

LB79.3 Webbed toes

A condition caused by failure of the longitudinal interdigital necrosis that normally separates the toes during the antenatal period. This condition is characterised by the presence of two or more toes that are fused together.

Inclusions: Simple syndactyly of toes without synostosis

LB79.Y Other specified syndactyly

LB79.Z Syndactyly, unspecified

Congenital deformities of fingers or toes (BlockL3‑LB8)

LB80 Congenital deformities of fingers

Any condition caused by failure of the fingers to develop correctly during the antenatal period.

Inclusions: Congenital deformities of hand

LB80.0 Clinodactyly of fingers

A condition caused by failure of the fifth finger to correctly develop during the antenatal period. This condition is characterised by bending of the fifth finger towards the fourth.

LB80.1 Congenital club finger

LB80.2 Radial deviation of fingers

LB80.Y Other specified congenital deformities of fingers

LB81 Congenital deformities of toes

LB81.0 Clinodactyly of toes

LB81.Y Other specified congenital deformities of toes

LB8Z Congenital deformities of fingers or toes, unspecified

LB90 Joint formation defects

Any condition of the skeletal system, caused by failure of joints to correctly develop during the antenatal period.

LB90.0 Humero-radio-ulnar synostosis

A condition caused by failure of the arm bones to correctly develop during the antenatal period. This condition is characterised by direct fusion of the humerus to the ulnar and radial bones of the arm, and consequent inability to straighten the elbow joint. This condition may be associated with thalidomide embryopathy. Confirmation is through observation of humero-radio-ulnar fusion by imaging.

LB90.1 Humero-radial synostosis

LB90.2 Humero-ulnar synostosis

A condition caused by failure of the arm bones to correctly develop during the antenatal period. This condition is characterised by direct fusion of the humerus and radial bones of the arm, and consequent inability to straighten the elbow joint. Confirmation is through observation of humero-ulnar fusion by imaging.

LB90.3 Radio-ulnar synostosis

A condition caused by failure of the arm bones to correctly develop during the antenatal period. This condition is characterised by direct fusion of the ulnar and radial bones of the arm, and consequent limitation of rotational movement of the forearm. Confirmation is through observation of radio ulnar fusion by imaging.

LB90.4 Madelung deformity

Madelung disease, or deformity is a predominantly bilateral wrist anomaly characterised by shortened and bowed radii and long ulnae leading to dorsal dislocation of the distal ulna and limited mobility of the wrist and elbow.

LB90.5 Congenital digital clubbing

Isolated congenital digital clubbing is a rare genodermatosis disorder characterised by enlargement of the terminal segments of fingers and toes with thickened nails without any other abnormality.

LB90.6 Tibio-fibular synostosis

LB90.7 Cubitus valgus

LB90.8 Cubitus varus

LB90.Y Other specified joint formation defects

LB90.Z Joint formation defects, unspecified

LB91 Congenital shoulder dislocation

LB92 Congenital elbow dislocation

LB93 Congenital knee dislocation

A condition characterised by hyperextension of the knee joint.

LB93.0 Congenital genu recurvatum

LB93.1 Congenital genu flexum

LB93.Y Other specified congenital knee dislocation

LB93.Z Congenital knee dislocation, unspecified

LB94 Congenital patella dislocation

LB95 Patella aplasia or hypoplasia

Isolated patella aplasia-hypoplasia is an extremely rare genetic condition characterised by congenital absence or marked reduction of the patellar bone. This condition may present with discomfort or abnormal gait. Confirmation is through verification of the reduced or absent patella by imaging

LB96 Congenital bowing of long bones

Congenital bowing of long bones is a congenital condition described by the presence of symmetric or asymmetric angular deformity and shortening of the long bones, particularly the femurs, tibiae and ulnae.

LB96.0 Congenital bowing of femur

A condition caused by failure of the femur to develop correctly during the antenatal period. This condition is characterised by abnormal angling of the femur. Confirmation is through observation of the bowed femur by imaging.

LB96.1 Congenital bowing of tibia

A condition caused by failure of the tibia to develop correctly during the antenatal period. This condition is characterised by abnormal angling of the tibia. Confirmation is through observation of the bowed tibia by imaging.

LB96.Y Other specified congenital bowing of long bones

LB96.Z Congenital bowing of long bones, unspecified

LB97 Limb overgrowth

Disproportionately long or asymmetric upper limbs

Exclusions: Hemihypertrophy (LD2C)

LB97.0 Macrodactyly of fingers

A condition caused by failure of the fingers to correctly develop during the antenatal period. This condition is characterised by overgrowth of bone and soft tissue, resulting in larger than normal fingers. This condition may be asymptomatic.

LB97.1 Macrodactyly of toes

LB97.2 Upper limb hypertrophy

LB97.3 Lower limb hypertrophy

LB97.Y Other specified limb overgrowth

LB97.Z Limb overgrowth, unspecified

LB98 Congenital deformities of feet

Any condition caused by malformation of the foot during the antenatal period.

LB98.0 Congenital varus deformities of feet

Any condition caused by failure of the bones of the foot to correctly develop during the antenatal period. These conditions are characterised by twisting of parts of the foot inward from the centre of the body.

LB98.00 Talipes equinovarus

A condition characterised by a foot that is fixated in adduction, in supination, and in varus. This condition may be associated with intrauterine position, genetic mutation, or can be idiopathic.

LB98.01 Talipes calcaneovarus

LB98.02 Metatarsus varus

A condition characterised by medial rotation of the cuneiform bones at the midtarsal joint, with associated medial deviation of the metatarsal, resulting in adduction and supination of the forefoot.

LB98.0Y Other specified congenital varus deformities of feet

LB98.0Z Congenital varus deformities of feet, unspecified

LB98.1 Congenital pes planus

Any condition caused by failure of the foot to correctly develop during the antenatal period. These conditions are characterised by severe rigid flat foot deformity.

Inclusions: congenital flat foot

LB98.2 Congenital valgus deformities of feet

Any condition caused by failure of the bones of the foot to correctly develop during the antenatal period. These conditions are characterised by twisting of parts of the foot outward from the centre of the body.

LB98.20 Congenital hallux valgus

A condition caused by failure of the hallux to correctly develop during the antenatal period. This condition is characterised by angling of the hallux medial to the metatarsophalangeal joint.

LB98.21 Metatarsus valgus

A condition caused by failure of the bones of the foot to correctly develop during the antenatal period. This condition is characterised by rotation of the forepart of the foot outward from the midline of the body.

LB98.22 Talipes calcaneovalgus

A condition caused by tightness of the muscles of the foot due to resting of the foot in a turned up position during the antenatal period. This condition is characterised by a foot that is turned upwards towards the shin.

LB98.2Y Other specified congenital valgus deformities of feet

LB98.2Z Congenital valgus deformities of feet, unspecified

LB98.3 Congenital pes cavus

A condition characterised by a high arch of the foot that does not flatten with weight bearing.

LB98.4 Congenital vertical talus

Isolated congenital vertical talus is a rare pedal deformity recognizable at birth by a dislocation of the talonavicular joint, resulting in a characteristic radiographic near-vertical orientation of the talus.

LB98.5 Congenital hammer toe

A condition characterised by angling downwards of the toe.

LB98.Y Other specified congenital deformities of feet

LB98.Z Congenital deformities of feet, unspecified

LB99 Reduction defects of upper limb

Any condition caused by the failure of an upper limb to correctly develop during the antenatal period. These conditions are characterised by reduction in size or absence of the limb.

LB99.0 Amelia of upper limb

A condition caused by the failure of an upper limb to develop during the antenatal period. This condition is characterised by absence of the upper limb.

LB99.1 Humeral agenesis or hypoplasia

LB99.2 Radial hemimelia

Radial hemimelia is a congenital longitudinal deficiency of the radius bone of the forearm characterised by partial or total absence of the radius.

Inclusions: Radial clubhand

LB99.3 Ulnar hemimelia

Ulnar hemimelia is a congenital ulnar deficiency of the forearm characterised by complete or partial absence of the ulna bone.

LB99.4 Congenital absence of upper arm or forearm with hand present

A condition caused by the failure of the upper arm and forearm to develop during the antenatal period, but with the hand present. This condition is characterised by direct connection of the hand to the shoulder.

LB99.5 Congenital absence of both forearm and hand

A condition caused by the failure of the forearm and hand to develop during the antenatal period.

LB99.6 Acheiria

A condition caused by failure of one or both hands to develop during the antenatal period.

LB99.7 Adactyly of hands

A condition caused by failure of the digits on the hand to correctly develop during the antenatal period. This condition is characterised by absence of digits on the hand.

LB99.8 Split hand

A condition caused by malformation of the hand during the antenatal period. This condition is characterised by a deep median cleft of the hand due to the absence of the central rays.

LB99.Y Other specified reduction defects of upper limb

LB99.Z Reduction defects of upper limb, unspecified

LB9A Reduction defects of lower limb

Any condition caused by the failure of a lower limb to correctly develop during the antenatal period. These conditions are characterised by reduction in size or absence of the limb.

LB9A.0 Amelia of lower limb

LB9A.1 Tibial hemimelia

Tibial hemimelia is a rare congenital anomaly characterised by deficiency of the tibia with a relatively intact fibula.

LB9A.2 Fibular hemimelia

Fibular hemimelia is a congenital longitudinal limb deficiency characterised by complete or partial absence of the fibula bone.

LB9A.3 Congenital absence of thigh or lower leg with foot present

Any condition caused by the failure of the thigh and lower leg to develop during the antenatal period. These conditions are characterised by direct connection of the foot to the hip.

LB9A.4 Apodia

A condition caused by failure of the foot to develop during the antenatal period.

LB9A.5 Adactyly of feet

A condition caused by failure of the digits on the foot to correctly develop during the antenatal period. This condition is characterised by absence of digits on the foot.

LB9A.6 Split foot

A condition caused by malformation of the foot during the antenatal period. This condition is characterised by a deep median cleft of the foot due to the absence of the central rays.

LB9A.7 Congenital absence of both lower leg and foot

Any condition caused by the failure of the lower leg and foot to develop during the antenatal period.

LB9A.8 Femoral agenesis or hypoplasia

Femoral agenesis/hypoplasia is a rare malformation of variable severity ranging from mild hypoplasia to complete absence of the femur.

LB9A.Y Other specified reduction defects of lower limb

LB9A.Z Reduction defects of lower limb, unspecified

LB9B Reduction defects of upper and lower limbs

LB9Y Other specified structural developmental anomalies of the skeleton

LB9Z Structural developmental anomalies of the skeleton, unspecified

Structural developmental anomalies of the skin (BlockL2‑LC0)

A deformation established before birth of an anatomical structure of the skin.

Exclusions: pilonidal cyst or sinus (EG63)

Congenital erythropoietic porphyria (5C58.12)

Acrodermatitis enteropathica (5C64.20)

Developmental hamartomata of the epidermis and epidermal appendages (BlockL3‑LC0)

Keratinocytic, pilosebaceous, eccrine, apocrine and other complex hamartomatous malformations of the skin.

LC00 Keratinocytic epidermal hamartoma

Keratinocytic epidermal hamartoma or epidermal naevus is a congenital hamartomatous epidermal malformation composed of keratinocytes. It is thought to arise as a result of somatic mutation: early embryonic mutations can give rise to extensive systematised naevi, though typically epidermal naevi are localised linear papillomatous or verrucous plaques. Histologically they exhibit acanthosis, papillomatosis and acanthosis.

Coded Elsewhere: Linear porokeratosis (ED52)

LC00.0 Epidermal naevus

LC00.Y Other specified keratinocytic epidermal hamartoma

LC01 Pilosebaceous hamartoma

Hamartomatous malformation involving elements originating from the developing pilosebaceous follicle.

LC02 Complex epidermal hamartoma

Hamartomatous malformation composed of elements deriving from several components of the developing epidermis and epidermal appendages.

LC0Y Other specified developmental hamartomata of the epidermis and epidermal appendages

Developmental anomalies of skin pigmentation (BlockL3‑LC1)

Hamartomatous cutaneous malformations involving melanocytes including congenital pigmented naevi.

Coded Elsewhere: Congenital melanocytic naevus (2F20.2)

Familial multiple café-au-lait macules (EC23.0)

LC10 Dermal melanocytosis

The presence at birth of functional melanocytes within the dermis. Most commonly this is as a result of incomplete migration of melanocytes to the epidermis as in lumbosacral dermal melanocytosis (Mongolian spot). Less commonly it is due to circumscribed hamartomatous proliferation of melanocytes in the dermis (e.g. Naevus of Ota).

Coded Elsewhere: Phakomatosis caesioflammea (LD2D.Y)

Phakomatosis caesiomarmorata (LD2D.Y)

LC1Y Other specified developmental anomalies of skin pigmentation

Hamartomata derived from dermal connective tissue (BlockL3‑LC2)

Hamartomatous malformations of dermal collagen and elastin.

LC20 Connective tissue hamartoma

Inclusions: Connective tissue naevus

LC2Y Other specified hamartomata derived from dermal connective tissue

Developmental defects of hair or nails (BlockL3‑LC3)

LC30 Developmental defects of hair or hair growth

LC31 Developmental defects of the nail apparatus

Congenital malformations of the nail apparatus.

Inclusions: congenital abnormalities of the nails

LC40 Dermoid cyst

Coded Elsewhere: Dermoid cyst of eyelid (2F36.4)

Developmental anomalies of cutaneous vasculature (BlockL3‑LC5)

Congenital vascular malformations affecting the skin

LC50 Developmental capillary vascular malformations of the skin

Coded Elsewhere: Phakomatosis pigmentovascularis (LD2D.Y)

LC50.0 Salmon patch

A common skin condition of neonates, characterised by flat, deep-pink localised areas of capillary dilation that occur predominantly on the back of the neck, lower occiput, upper eyelids, upper lip, and bridge of the nose. The areas disappear permanently by about 2 years of age.

LC50.1 Port-wine stain

A port-wine stain is defined as a macular telangiectatic area of skin which is present at birth and does not spontaneously involute. Port-wine stains may be localised or extensive and they are often associated with an underlying disorder.

Coded Elsewhere: Sturge-Weber syndrome (LD23)

LC50.Y Other specified cutaneous capillary vascular malformation

LC51 Developmental venous malformations involving the skin

Certain genetically-determined syndromes presenting with venous anomalies in the skin

LC52 Complex or combined developmental vascular malformations involving the skin

Coded Elsewhere: Angio-osteohypertrophic syndrome (LD26.60)

Cobb syndrome (LA90.3Y)

Maffucci syndrome (LD2F.1Y)

LC5Y Other specified developmental anomalies of cutaneous vasculature

LC5Z Developmental anomalies of cutaneous vasculature, unspecified

Congenital anomalies of skin development (BlockL3‑LC6)

Coded Elsewhere: Focal dermal hypoplasia (LD27.0Y)

Beckwith-Wiedemann syndrome (LD2C)

LC60 Aplasia cutis congenita

Congenital absence of skin. The commonest form presents as a defect limited to the scalp. It is also a component of a number of genetic syndromes.

LC7Y Other specified structural developmental anomalies of the skin

LC7Z Structural developmental anomalies of the skin, unspecified

Structural developmental anomalies of the adrenal glands (BlockL2‑LC8)

A deformation established before birth of an anatomical structure of the adrenal glands.

Exclusions: Congenital adrenal hyperplasia (5A71.01)

LC80 Congenital adrenal hypoplasia

Coded Elsewhere: Congenital adrenocortical insufficiency (5A74.Y)

LC8Y Other specified structural developmental anomalies of the adrenal glands

LC8Z Structural developmental anomalies of the adrenal glands, unspecified

LD0Y Other specified structural developmental anomalies primarily affecting one body system

LD0Z Structural developmental anomalies primarily affecting one body system, unspecified

Multiple developmental anomalies or syndromes (BlockL1‑LD2)

Complex developmental anomalies involving more than one body system

LD20 Syndromes with central nervous system anomalies as a major feature

Exclusions: Meckel syndrome (LD2F.13)

LD20.0 Syndromes with cerebellar anomalies as a major feature

Coded Elsewhere: Dysplastic cerebellar gangliocytoma (2A00.21)

LD20.00 Joubert syndrome

Joubert syndrome is a genetic midbrain-hindbrain malformation syndrome characterised by congenital malformation of the brainstem and agenesis or hypoplasia of the cerebellar vermis leading to an abnormal respiratory pattern, nystagmus, hypotonia, ataxia, and delay in achieving motor milestones.

Coded Elsewhere: Oral-facial-digital syndrome type 6 (LD25.00)

LD20.01 Pontocerebellar hypoplasia

Nonsyndromic pontocerebellar hypoplasias are a rare heterogeneous group of diseases characterised by hypoplasia and atrophy and/or early neurodegeneration of the cerebellum and pons. Eight subtypes named type 1-8 have been described, generally inherited in an autosomal recessive pattern.

LD20.0Y Other specified syndromes with cerebellar anomalies as a major feature

LD20.0Z Syndromes with cerebellar anomalies as a major feature, unspecified

LD20.1 Syndromes with lissencephaly as a major feature

The term lissencephaly covers a group of rare malformations sharing the common feature of anomalies in the appearance of brain convolutions (characterised by simplification or absence of folding) associated with abnormal organisation of the cortical layers as a result of neuronal migration defects during embryogenesis. Children with lissencephaly have feeding and swallowing problems, muscle tone anomalies (early hypotonia and subsequently limb hypertonia), seizures (in particular, infantile spasms) and severe psychomotor retardation. Two large groups can be distinguished: classical lissencephaly (and its variants) and cobblestone lissencephaly.

Inclusions: Agyria

Pachygyria

LD20.2 Syndromes with microcephaly as a major feature

Developmental syndromes in which an abnormally small head size is a significant feature.

LD20.3 Syndromes with holoprosencephaly as a major feature

Any syndrome caused by failure of the prosencephalon to divide in two during the antenatal period. These syndromes may present with closely spaced eyes, cyclopia, flat nasal bridge, single maxillary central incisor, small head size, and clefts of the lip and palate.

Coded Elsewhere: Arrhinencephaly (LA05.4)

LD20.4 Brain calcifications

LD20.Y Other specified syndromes with central nervous system anomalies as a major feature

LD20.Z Syndromes with central nervous system anomalies as a major feature, unspecified

LD21 Syndromes with eye anomalies as a major feature

Any syndrome caused by failure of one or both eyes to correctly develop during the antenatal period.

Exclusions: Septo-optic dysplasia (5A61.0)

Cat-eye syndrome (LD41.P)

Aicardi syndrome (LD20)

Papillorenal syndrome (LA13.7)

WAGR syndrome (LD2A)

LD21.0 Syndromes with microphthalmia as a major feature

Syndromes in which abnormally small eyes form an important component.

LD21.Y Other specified syndromes with eye anomalies as a major feature

LD21.Z Syndromes with eye anomalies as a major feature, unspecified

LD22 Syndromes with dental anomalies as a major feature

Coded Elsewhere: Amelogenesis imperfecta (LA30.6)

Dentine dysplasia (LA30.7)

Dentinogenesis imperfecta (LA30.8)

LD23 Syndromes with vascular anomalies as a major feature

Coded Elsewhere: Angio-osteohypertrophic syndrome (LD26.60)

Primary lymphoedema (BD93.0)

Cutis marmorata telangiectatica congenita (LC52)

LD24 Syndromes with skeletal anomalies as a major feature

Coded Elsewhere: Progressive osseous heteroplasia (FB31.0)

Fibrodysplasia ossificans progressiva (FB31.1)

Osteolysis syndromes (FB86.2)

Calcification or ossification of muscles of genetic origin (FB31.Y)

LD24.0 Syndromes with micromelia

Syndromes in which abnormally short limbs are a major feature

LD24.00 Achondroplasia

Achondroplasia is the most frequent form of chondrodysplasia and is a type of dwarfism is characterised by short limbs, hyperlordosis, short hands, and macrocephaly with high forehead and saddle nose, with normal intellectual development.

LD24.01 Hypochondroplasia

Hypochondroplasia is a skeletal dysplasia characterised by disproportionate short stature, mild lumbar lordosis and limited extension of the elbow joints.

LD24.02 Thanatophoric dysplasia

Thanatophoric Dysplasia is a severe skeletal disorder that is lethal in the neonatal period. Two clinically defined TD subtypes have been classified: type I (TDI), characterised by micromelia with bowed femurs and, occasionally, by the presence of cloverleaf skull deformity of varying severity and type II (TDII), characterised by micromelia with straight femurs and a moderate to severe cloverleaf skull deformity.

LD24.03 Diastrophic dysplasia

Diastrophic dwarfism is a rare autosomal recessive disorder marked by short stature with short extremities (final adult height is 120cm +/- 10cm), and joint malformations leading to multiple joint contractures (principally involving the shoulders, elbows, interphalangeal joints and hips)

LD24.04 Chondrodysplasia punctata

LD24.0Y Other specified syndromes with micromelia

LD24.0Z Syndromes with micromelia, unspecified

LD24.1 Bone diseases with increased bone density

Coded Elsewhere: Pycnodysostosis (5C56.Y)

Buschke-Ollendorff syndrome (EC4Y)

LD24.10 Osteopetrosis

Osteopetrosis ('marble bone disease') is a descriptive term that refers to a group of rare, heritable disorders of the skeleton characterised by increased bone density on radiographs. Osteopetrotic conditions vary greatly in their presentation and severity, ranging from neonatal onset with life-threatening complications such as bone marrow failure (as in classical or 'malignant' autosomal recessive osteopetrosis) to the incidental finding of osteopetrosis on radiographs (e.g. osteopoikilosis).

Coded Elsewhere: OL-EDA-ID syndrome (LD27.0Y)

Osteopetrosis - hypogammaglobulinaemia (4A01.0Y)

LD24.11 Osteopoikilosis

LD24.1Y Other specified bone diseases with increased bone density

LD24.1Z Bone diseases with increased bone density, unspecified

LD24.2 Bone diseases with disorganised development of skeletal components

Coded Elsewhere: Osteogenesis imperfecta (LD24.K0)

Enchondromatosis (2E83.Z)

X-linked cutis laxa (LD28.2)

Maffucci syndrome (LD2F.1Y)

Inherited bone dysplasia (FB80.Y)

LD24.20 Multiple osteochondromas

Inclusions: Diaphyseal aclasis

LD24.21 Exostoses with anetodermia and brachydactyly type E

LD24.22 Cherubism

Cherubism is a benign fibro-osseous hereditary disorder of childhood, limited to the lower half of the face, the maxilla and particularly the mandible, with bilateral painless swelling of jaws (giving the so-called cherubic look) associated with multicystic bone tumours and eyes-to-heaven appearance. Dentition is also abnormal at the sites concerned: tooth agenesis, noneruption, displacement, root resorption and malocclusions are common.

LD24.23 Yunis-Varon disease

A disease caused by failure of multiple body systems to correctly develop during the antenatal period, due to mutation of the FIG4 gene. This disease is characterised by cleidocranial dysplasia, digital anomalies, and severe neurological involvement.

LD24.2Y Other specified bone diseases with disorganised development of skeletal components

LD24.2Z Bone diseases with disorganised development of skeletal components, unspecified

LD24.3 Spondyloepiphyseal or spondyloepimetaphyseal dysplasias

Spondyloepiphyseal dysplasias (SED) are a heterogeneous group of congenital chondrodysplasias that specifically affect epiphyses and vertebrae. Their most frequent form is characterised by small neonatal size of ovid vertebrae and overall late growth of bones, more marked in the femoral heads, with a slightly irregular metaphyseal limit. Other clinical forms have been described, some of which were dominant and more or less severe with metaphyseal lesions, while others were recessive and included nephrotic syndrome, lymphopenia, and immune disorders (immune bone dysplasia).

LD24.4 Spondylometaphyseal dysplasias

Spondylometaphyseal dysplasias are a heterogeneous group of disorders associated with walking and growth disturbances that become evident during the second year of life. The disorders are characterised by platyspondyly (flattened vertebrae) and marked hip and knee metaphyseal lesions. The different forms of spondylometaphyseal dysplasia are distinguished by the localization and severity of involvement of the affected metaphyses.

LD24.5 Spondylodysplastic dysplasias

LD24.50 Achondrogenesis

LD24.51 Hypochondrogenesis

A condition caused by failure of the skeletal system to correctly develop during the antenatal period, due to mutation of the COL2A1 gene. This condition is characterised by a small body, short limbs, underdeveloped lungs, flat and oval-shaped face, hypertelorism, micrognathia, enlarged abdomen, and ossification in the spine and pelvis. This condition may also present with a cleft palate.

LD24.5Y Other specified spondylodysplastic dysplasias

LD24.5Z Spondylodysplastic dysplasias, unspecified

LD24.6 Multiple epiphyseal dysplasia or pseudoachondroplasia

LD24.60 Pseudoachondroplasia

Pseudoachondroplasia is a chondrodysplasia characterised by severe growth deficiency and deformations such as bow legs and hyperlordosis.

LD24.61 Multiple epiphyseal dysplasias

Multiple epiphyseal dysplasias (MED/EDMs) are characterised by epiphyseal anomalies causing joint pain early in life, recurrent osteochondritis and early arthrosis. The EDMs are a heterogeneous group of diseases with variable expression classed as MED/EDMs 1-6.

Coded Elsewhere: Wolcott-Rallison syndrome (5A13.6)

LD24.6Y Other specified multiple epiphyseal dysplasia or pseudoachondroplasia

LD24.6Z Multiple epiphyseal dysplasia or pseudoachondroplasia, unspecified

LD24.7 Multiple metaphyseal dysplasias

Exclusions: Pyle disease (LD24.1)

Coded Elsewhere: Cartilage-hair hypoplasia (LD27.0Y)

Metaphyseal dysostosis - intellectual deficit - conductive deafness (LD2H.Y)

LD24.8 Acromelic dysplasias

Coded Elsewhere: Microspherophakia or Weill Marchesani Syndrome (9C61.42)

Trichorhinophalangeal syndrome type 1 and 3 (LD27.0Y)

LD24.80 Langer-Giedion syndrome

Langer-Giedon syndrome or trichorhinophalangeal syndrome type 2 is a chromosomal anomaly syndrome characterised by the association of intellectual deficit and numerous other anomalies including redundant skin, multiple cartilaginous exostoses, characteristic facies and cone-shaped phalangeal epiphyses.

LD24.8Y Other specified acromelic dysplasias

LD24.8Z Acromelic dysplasias, unspecified

LD24.9 Acromesomelic dysplasias

A group of rare disorders characterised by shortening of the bones of the forearms, lower legs, hands and feet.

Exclusions: Sensenbrenner syndrome (LD27.0)

LD24.A Mesomelic or rhizomesomelic dysplasias

LD24.B Short rib syndromes

Exclusions: Oral-facial-digital syndrome type 4 (LD25.00)

Coded Elsewhere: Chondroectodermal dysplasia (LD27.0Y)

LD24.B0 Short rib-polydactyly syndrome

Short rib-polydactyly syndromes are a group of bone malformations characterised by a narrow thorax and polydactyly (usually preaxial). Prevalence as a group is unknown. The group is heterogeneous and includes Jeune syndrome and Ellis Van Creveld syndrome, neither of which are lethal, together with lethal chondrodysplasias: Saldino-Noonan (type 1), Majewski (type 2), Verma-Naumoff (type 3) and Beemer-Langer (Type 4).

LD24.B1 Asphyxiating thoracic dystrophy

Asphyxiating thoracic dystrophy, also called Jeune syndrome, is a short-rib dysplasia characterised by a narrow thorax, short limbs and radiological skeletal abnormalities including "trident" aspect of the acetabula and metaphyseal changes.

LD24.BY Other specified short rib syndromes

LD24.BZ Short rib syndromes, unspecified

LD24.C Bent bone dysplasias

Any syndromes are characterised by poor mineralization of the skull, craniosynostosis, hypoplastic pubis and clavicles, osteopenia, bent long bones, low-set ears, hypertelorism, midface hypoplasia, prematurely erupted fetal teeth, and micrognathia. These syndromes may be associated with mutation of the FGFR2 gene.

Coded Elsewhere: Campomelic dysplasia (LD2A.Y)

Juvenile osteochondrosis of tibia or fibula (FB82.1)

LD24.D Slender bone dysplasias

Any syndrome characterised by dwarfism, thin bones, multiple fractures, and prenatal or early postnatal death.

Coded Elsewhere: IMAGe syndrome (5A74.Y)

LD24.E Bone dysplasias with multiple joint dislocations

Any syndrome characterised by malformation of the musculoskeletal system during the antenatal period, which include the dislocations of multiple joints.

LD24.F Progressive ossification of skin, skeletal muscle, fascia, tendons or ligaments

Coded Elsewhere: Progressive osseous heteroplasia (FB31.0)

Fibrodysplasia ossificans progressiva (FB31.1)

LD24.G Syndromic craniosynostoses

Any syndrome caused by premature fusing of sections of the infant skull. These syndromes are characterised by disfiguring compensatory growth of the skull. These syndromes may also present with frequent worsening morning headache, recurrent vomiting, cephalocranial disproportion, raised intracranial pressure, optic atrophy, blindness, or developmental delay.

Exclusions: Sensenbrenner syndrome (LD27.0)

Shprintzen-Goldberg craniosynostosis syndrome (LD28.0)

Craniotelencephalic dysplasia (LD20.1)

Coded Elsewhere: Craniofrontonasal dysplasia (LD25.3)

LD24.G0 Pfeiffer syndrome

Pfeiffer syndrome (associated with mutations in the FGFR1 and 2 gene) is a syndromic form of craniosynostosis characterised by the association of craniosynostosis. Often pansynostosis. Severe midface hypoplasia. Broad and deviated thumbs and big toes, and partial syndactyly of the fingers and toes. Hydrocephaly may be found occasionally, along with severe ocular proptosis, ankylosed elbows.

Exclusions: Pfeiffer disease (1D81.0)

LD24.G1 Crouzon disease

Crouzon disease is a form of syndromic craniosynostosis characterised by craniosynostosis and facial hypoplasia.

LD24.G2 Apert syndrome

Apert syndrome is a syndromic craniosynostosis associated with mutations in the FGFR2 gene and characterised by premature closure of coronal suture and a later onset of pansynostosis. Pathognomonic is an osseous and membranous syndactyly of at least Digitus II-IV (fingers and toes). High incidence of midface hypoplasia with orbital- and facial stenosis, cleft palate, vertebral fusion. Mental deficits in 30%.

LD24.GY Other specified syndromic craniosynostoses

LD24.GZ Syndromic craniosynostoses, unspecified

LD24.H Dysostoses with predominant vertebral and costal involvement

Any syndrome characterised by malformation of the musculoskeletal system during the antenatal period, which include dysgenesis of the vertebrae and intercostal cartilage.

Exclusions: Spondylocostal dysostosis - anal and genitourinary malformations (LD2F.1)

LD24.J Patellar dysostoses

Any syndrome characterised by malformation of the patella during the antenatal period.

LD24.J0 Nail-patella syndrome

Nail patella syndrome is a hereditary osteo-onychodysplasia characterised by nail dysplasia with triangular lunula, hypoplastic or absent patellas, iliac exostoses (`iliac horns') and dysplastic elbows.

LD24.JY Other specified patellar dysostoses

LD24.JZ Patellar dysostoses, unspecified

LD24.K Genetic bone diseases with decreased bone density

Coded Elsewhere: Ehlers-Danlos-osteogenesis imperfecta syndrome (LD28.1Y)

LD24.K0 Osteogenesis imperfecta

Osteogenesis imperfecta (OI) comprises a heterogeneous group of genetic disorders characterised by increased bone fragility, low bone mass, and susceptibility to bone fractures with variable severity. The most clinically relevant characteristic of all types of OI is bone fragility, which manifests as multiple spontaneous fractures.

Inclusions: Fragilitas ossium

Osteopsathyrosis

LD24.KY Other specified genetic bone diseases with decreased bone density

LD24.KZ Genetic bone diseases with decreased bone density, unspecified

LD24.Y Other specified syndromes with skeletal anomalies as a major feature

LD24.Z Syndromes with skeletal anomalies as a major feature, unspecified

LD25 Syndromes with face or limb anomalies as a major feature

Exclusions: Freeman-Sheldon syndrome (LD26.4)

LD25.0 Oromandibular-limb anomaly syndrome

A syndrome caused by failure of the face and limbs to correctly develop during the antenatal period. This syndrome is characterised by malformations of the tongue, mandible, and limbs.

Exclusions: Ectrodactyly - cleft palate (LD2F.1)

Ectrodactyly - ectodermal dysplasia - cleft lip or palate (LD27.0)

LD25.00 Oral-facial-digital syndrome

A condition caused by failure of the head and digits to correctly develop during the antenatal period, due to mutation of the OFD1 gene. This condition may be associated with cleft or lobed tongue, noncancerous tumours or nodules of the tongue, abnormal shape or number of teeth, cleft palate, hyperplastic frenula of the lip or gums, cleft lip, hypertelorism, wide nose with broad, flat nasal bridge, syndactyly, brachydactyly, clinodactyly, polydactyly, polycystic kidney disease, neurological problems, bone abnormalities, vision loss, or heart defects.

LD25.0Y Other specified oromandibular-limb anomaly syndrome

LD25.0Z Oromandibular-limb anomaly syndrome, unspecified

LD25.1 Fronto-otopalatodigital syndromes

LD25.2 Acrofacial dysostoses

Any syndrome caused by failure of the face and limbs to correctly develop during the antenatal period.

LD25.3 Craniofacial dysostoses

Syndromes caused by abnormal development of skull and facial bones. They may present with acrocephaly, exophthalmos, hypertelorism, strabismus, parrot-beaked nose, or hypoplastic maxilla. Non-syndromic craniosynostosis, which is predominantly sporadic, is coded elsewhere.

Exclusions: Acrofacial dysostosis, Nager type (LD25.2)

Postaxial acrofacial dysostosis (LD25.2)

Acrofacial dysostosis, Weyers type (LD25.2)

Frontometaphyseal dysplasia (LD25.1)

Craniosynostosis (LB70.0)

LD25.Y Other specified syndromes with face or limb anomalies as a major feature

LD25.Z Syndromes with face or limb anomalies as a major feature, unspecified

LD26 Syndromes with limb anomalies as a major feature

LD26.0 Combined reduction defects of upper and lower limbs

LD26.1 Complex brachydactylies

A disease caused by failure of the digits to correctly develop during the antenatal period. This disease is characterised by multiple digits of below normal length. This condition may be associated with mutation in the GDF5 gene.

Exclusions: Catel-Manzke syndrome (LD2F.1)

LD26.2 Syndromes with limb duplication, polydactyly, syndactyly or triphalangism

Any syndrome caused by failure of the limbs to correctly develop during the antenatal period. These syndromes are characterised by supernumerary limbs or digits, fused digits, or supernumerary phalanges.

Exclusions: Townes-Brocks syndrome (LD2F.1)

LD26.3 Syndromes with synostoses of limbs

LD26.4 Arthrogryposis syndromes

Any syndrome caused by failure of elastic tissue to correctly develop during the antenatal period. These syndromes are characterised by the presence of multiple joint contractures, where elastic tissues are replaced by inelastic tissues, which results in fixation of the joint.

Exclusions: Arthrogryposis due to muscular dystrophy (8C70)

LD26.40 Multiple pterygium syndrome

LD26.41 Arthrogryposis multiplex congenita

Arthrogryposis multiplex congenita, comprises nonprogressive congenital conditions characterised by multiple joint contractures. The term is currently used in connection with a very heterogeneous group of disorders that all include multiple congenital joint contractures. The major cause of arthrogryposis is fetal akinesia due to fetal abnormalities (e.g. neurogenic, muscle, or connective tissue abnormalities; mechanical limitations to movement) or maternal disorders (e.g. infection, drugs, trauma, other maternal illnesses). generalised fetal akinesia can also lead to polyhydramnios, pulmonary hypoplasia, micrognathia, ocular hypertelorism, and short umbilical cord. Lack of fetal movement causes extra connective tissue to develop around the joint, limiting movement and further aggravating the joint contracture.

Exclusions: COFS syndrome (LD2B)

Arthrogryposis multiplex congenita - lissencephaly (LD2F.1)

Coded Elsewhere: Arthrogryposis - renal dysfunction - cholestasis (5C58.0Y)

LD26.4Y Other specified arthrogryposis syndromes

LD26.4Z Arthrogryposis syndromes, unspecified

LD26.5 Constriction rings

A condition caused by entangling of fibrous bands of the amniotic sac around a developing fetus. This condition may present with circular indentation around a digit or limb, swelling, restriction of the lymphatic or venous flow, limb development defects, or in utero amputation.

LD26.6 Congenital vascular bone syndromes

LD26.60 Angio-osteohypertrophic syndrome

Angio-osteohypertrophic (AOH) syndrome is a congenital vascular bone syndrome characterised by the presence of vascular malformations in a limb resulting in limb overgrowth. Depending on whether the malformations are slow flow venous or fast flow arteriovenous the syndrome may be divided into two subtypes, Klippel-Trénaunay and Parkes-Weber syndromes respectively. Some cases of the latter are associated with mutations in the RASA1 gene.

LD26.6Y Other specified congenital vascular bone syndromes

LD26.6Z Congenital vascular bone syndromes, unspecified

LD26.Y Other specified syndromes with limb anomalies as a major feature

LD26.Z Syndromes with limb anomalies as a major feature, unspecified

LD27 Syndromes with skin or mucosal anomalies as a major feature

Coded Elsewhere: Acrodermatitis enteropathica (5C64.20)

Non-syndromic ichthyosis (EC20.0)

Pseudoxanthoma elasticum (EC40)

Xeroderma pigmentosum-Cockayne syndrome complex (LD2B)

Hereditary ichthyosis (EC20.Y)

Palmoplantar keratoderma – oral leukokeratosis – oesophageal carcinoma (EC20.31)

LD27.0 Ectodermal dysplasia syndromes

Ectodermal dysplasias (EDs) are a heterogeneous group of disorders characterised by developmental dystrophies of ectodermal structures, such as hypohidrosis, hypotrichosis, onychodysplasia and hypodontia or anodontia. More than 160 clinically and genetically distinct hereditary ectodermal dysplasias have been catalogued.

Coded Elsewhere: Langer-Giedion syndrome (LD24.80)

Oral-facial-digital syndrome (LD25.00)

Solitary median maxillary central incisor syndrome (LA30.Y)

Rothmund-Thomson syndrome (LD2B)

Hallermann-Streiff-François syndrome (LD2B)

Keratitis – ichthyosis – deafness syndrome (LD27.2)

Papillon-Lefèvre syndrome (EC20.30)

Cataract – hypertrichosis – intellectual deficit (LD27.3)

Hypomelanosis of Ito (EC23.2Y)

Ectodermal dysplasia – skin fragility syndrome (EC30)

Dyskeratosis congenita (3A70.0)

LD27.00 Incontinentia pigmenti

Incontinentia pigmenti is an X-linked dominant gene disorder due to abnormalities of the NF-kappa-B (NEMO) gene on chromosome Xq28. It is lethal in male fetuses but the presence of a normal second X chromosome in females results in a mosaicism which is compatible with life. Affected females present in infancy with skin blisters in linear arrays (Blaschko lines) typically on the scalp and limbs. Within the first few months of life these are succeeded by warty changes and hyperpigmentation. These tend to resolve over time, often leaving atrophic streaks. Associated features include abnormal dentition, ocular defects and a variety of neurological complications.

LD27.01 Cronkhite-Canada syndrome

Cronkhite-Canada syndrome (CCS) is a sporadically occurring, noninherited disorder of generalised gastrointestinal polyps (hamartomas), cutaneous pigmentation, alopecia, and onychodystrophy. The possibility of progression to cancer is considered to be low. Chronic diarrhea and protein-losing enteropathy are often observed.

LD27.02 Hypohidrotic ectodermal dysplasia

Hypohidrotic ectodermal dysplasia is a genetic disorder of ectoderm development characterised by malformation of ectodermal structures such as skin, hair, teeth and sweat glands. It comprises three clinically almost indistinguishable subtypes with impaired sweating as the key symptom: Christ-Siemens-Touraine syndrome (X-linked), autosomal recessive and autosomal dominant hypohidrotic ectodermal dysplasia, as well as a fourth rare subtype with immunodeficiency as the key symptom.

LD27.03 Hidrotic ectodermal dysplasia, Clouston type

Clouston syndrome (or hidrotic ectodermal dysplasia) is an inherited disorder characterised by the clinical triad of nail dystrophy, alopecia, and palmoplantar hyperkeratosis.

LD27.0Y Other specified ectodermal dysplasia syndromes

LD27.1 Xeroderma pigmentosum

Xeroderma pigmentosum (XP) is a rare genodermatosis characterised by extreme sensitivity to ultraviolet (UV)-induced changes in the skin and eyes, and multiple skin cancers. It is subdivided into 8 complementation groups, according to the affected gene: XPA to XPG, and XP variant (XPV). The severity of the clinical manifestations and the age of onset are extremely variable and are in part dependent on exposure to sunlight and the complementation group.

Coded Elsewhere: Xeroderma pigmentosum variant (LD27.Y)

LD27.2 Syndromic ichthyosis

Hereditary disorders in which ichthyosis is associated with significant other abnormalities.

Coded Elsewhere: Sjögren-Larsson syndrome (5C52.03)

LD27.3 Genetic syndromes with hypertrichosis

Genetic syndromes in which excessive non-androgen-dependent hair growth is associated with other abnormalities.

Coded Elsewhere: Cone-rod type amaurosis congenita – congenital hypertrichosis (9B70)

Ramon syndrome (LD2F.1Y)

LD27.4 Genetic syndromes affecting nails

Coded Elsewhere: Wilson disease (5C64.00)

Nail-patella syndrome (LD24.J0)

Hidrotic ectodermal dysplasia, Clouston type (LD27.03)

Severe T-cell immunodeficiency - congenital alopecia - nail dystrophy (4A01.1Y)

Odonto-onycho-dermal dysplasia (LD27.0Y)

Onycho-tricho-dysplasia – neutropaenia syndrome (LD27.0Y)

Knuckle pads – leukonychia – sensorineural deafness (LD2H.Y)

Deafness – enamel hypoplasia – nail defects (LD27.0Y)

Anonychia with bizarre flexural pigmentation (LD27.0Y)

Anonychia or onychodystrophy – hypoplasia or absence of distal phalanges (LD27.0Y)

Autosomal dominant hypodontia with nail dysplasia (LD27.0Y)

Amelo-onycho-hypohidrotic syndrome (LD27.0Y)

Deafness – onychodystrophy (LD27.0Y)

Odonto-onycho-hypohidrotic dysplasia - midline scalp defects (LD27.0Y)

Tricho-odonto-onycho-dermal syndrome (LD27.0Y)

Tricho-odonto-onychodysplasia - dominant syndactyly (LD27.0Y)

Pili torti - onychodysplasia (LD27.0Y)

Dyskeratosis congenita (3A70.0)

Primary hypertrophic osteoarthropathy (FB86.10)

LD27.5 Genetic hamartoneoplastic syndromes affecting the skin

A heterogeneous group of inherited diseases characterised by the presence of multiple hamartomata and associated with an increased risk of malignancy.

Coded Elsewhere: Neurofibromatoses (LD2D.1)

Tuberous sclerosis (LD2D.2)

Gardner syndrome (LD2D.3)

Gorlin syndrome (LD2D.4)

Bannayan-Riley-Ruvalcaba syndrome (LD2D.Y)

Cowden syndrome (LD2D.Y)

Multiple familial trichoepithelioma (2F22)

LD27.6 Genetic lipodystrophy

Genetic lipodystrophies represent a heterogeneous group of rare diseases characterised by a generalised or localised loss of body fat (lipoatrophy).

Coded Elsewhere: Familial partial lipodystrophy (5A44)

Wiedemann-Rautenstrauch progeroid syndrome (LD2B)

LD27.60 Congenital generalised lipodystrophy

Coded Elsewhere: Berardinelli-Seip congenital lipodystrophy (5A44)

LD27.6Z Genetic lipodystrophy, unspecified

LD27.Y Other specified syndromes with skin or mucosal anomalies as a major feature

LD27.Z Syndromes with skin or mucosal anomalies as a major feature, unspecified

LD28 Syndromes with connective tissue involvement as a major feature

Exclusions: Cutis laxa (EE41.0)

Pseudoxanthoma elasticum (EC40)

LD28.0 Marfan syndrome or Marfan-related disorders

Coded Elsewhere: Aortic aneurysm syndrome, Loeys-Dietz type (BD50.Z)

Ectopia lentis syndrome (LA12.Y)

LD28.00 Congenital contractural arachnodactyly

Congenital contractural arachnodactyly (CCA, Beals syndrome) is a connective tissue disorder characterised by multiple flexion contractures, arachnodactyly, severe kyphoscoliosis, abnormal pinnae and muscular hypoplasia. Although the clinical features can be similar to Marfan syndrome (MFS), multiple joint contractures (especially of the elbow, knee, and finger joints), and crumpled ears in the absence of significant aortic root dilatation are characteristic of Beals syndrome and rarely found in MFS.

LD28.01 Marfan syndrome

Marfan syndrome is a systemic disease of connective tissue characterised by a variable combination of cardiovascular, musculo-skeletal, ophthalmic and pulmonary manifestations. Cardiovascular involvement is characterised by 1) progressive dilation of the aorta accompanied by an increased risk of aortic dissection, which affects prognosis and 2) mitral insufficiency. Skeletal involvement is often the first sign of the disease and can include dolichostenomelia, large size, arachnodactyly, joint hypermobility, scoliotic deformations, acetabulum protrusion, thoracic deformity, dolichocephaly of the anteroposterior axis, micrognathism or malar hypoplasia. Ophthalmic involvement results in axile myopia, which can lead to retinal detachment and lens displacement.

LD28.0Y Other specified Marfan syndrome or Marfan-related disorders

LD28.0Z Marfan syndrome or Marfan-related disorders, unspecified

LD28.1 Ehlers-Danlos syndrome

Ehlers – Danlos syndrome (EDS) is a heterogeneous group of inherited disorders of connective tissue, principally collagen, that range in severity from mild joint hypermobility to life-threatening fragility of soft tissue and vasculature.

LD28.10 Ehlers-Danlos syndrome, classical type

Ehlers-Danlos syndrome, classic type is a type of Ehlers-Danlos syndromes (EDS), a heterogeneous group of hereditary connective tissue diseases characterised by joint hyperlaxity, cutaneous hyperelasticity and tissue fragility, and is characterised by the following major clinical diagnostic criteria: hyperextensible skin, atrophic cutaneous scars due to tissue fragility and joint hyperlaxity.

LD28.1Y Other specified types of Ehlers-Danlos syndrome

LD28.2 Genetically-determined cutis laxa

LD28.Y Other specified syndromes with connective tissue involvement as a major feature

LD28.Z Syndromes with connective tissue involvement as a major feature, unspecified

LD29 Syndromes with obesity as a major feature

Exclusions: WAGR syndrome (LD2A)

Fragile X syndrome (LD55)

Coded Elsewhere: Prader-Willi syndrome (LD90.3)

Alström syndrome (LD2H.Y)

Cohen syndrome (LD90.Y)

Sotos syndrome (LD2C)

Weaver syndrome (LD2C)

Beckwith-Wiedemann syndrome (LD2C)

LD2A Malformative disorders of sex development

Any condition caused by failure of the genitals to correctly develop during the antenatal period.

Exclusions: pseudohermaphroditism: female, with adrenocortical disorder (5A71)

Coded Elsewhere: Chimaera 46, XX, 46, XY (LD56)

46,XX disorders of sex development induced by androgens of maternal origin (5A71.1)

Congenital adrenal hyperplasia (5A71.01)

LD2A.0 Ovotesticular disorder of sex development

Ovotesticular disorder of sex development, formerly called true hermaphroditism, is a rare cause of genital ambiguity characterised by the presence of ovarian and testicular tissue in an individual, leading to development of both male and female structures.

LD2A.1 46,XY gonadal dysgenesis

This is any congenital developmental disorder of the reproductive system characterised by a progressive loss of primordial germ cells on the developing gonads of an embryo.

LD2A.2 Testicular agenesis

LD2A.3 46,XY disorder of sex development due to a defect in testosterone metabolism

Exclusions: Congenital adrenal hyperplasia (5A71.01)

Coded Elsewhere: Smith-Lemli-Opitz syndrome (5C52.10)

LD2A.4 46,XY disorder of sex development due to androgen resistance

Androgen insensitivity syndrome (AIS) is a disorder of sex development (DSD) characterised by the presence of female external genitalia, ambiguous genitalia or variable defects in virilization in a 46,XY individual with absent or partial responsiveness to age-appropriate levels of androgens. It comprises two clinical subgroups: complete AIS (CAIS) and partial AIS (PAIS).

LD2A.Y Other specified malformative disorders of sex development

LD2A.Z Malformative disorders of sex development, unspecified

LD2B Syndromes with premature ageing appearance as a major feature

A heterogeneous group of hereditary syndromes in which affected individuals do or appear to age at an accelerated rate.

Inclusions: Progeroid syndromes

Exclusions: Xeroderma pigmentosum (LD27.1)

Cutis laxa (EE41.0)

Coded Elsewhere: Ehlers-Danlos syndrome, progeroid type (LD28.1Y)

Autosomal recessive cutis laxa, type 3 (LD28.2)

Bloom syndrome (4A01.31)

Ataxia-telangiectasia (4A01.31)

Mandibuloacral dysplasia (LD27.6Z)

LD2C Overgrowth syndromes

Exclusions: Sturge-Weber syndrome (LD23)

Diabetic embryopathy (KB60.1)

Enchondromatosis (2E83)

Maffucci syndrome (LD2F.1)

Coded Elsewhere: Perlman syndrome (2C90.Y)

LD2D Phakomatoses or hamartoneoplastic syndromes

Exclusions: Ataxia-telangiectasia (4A01.31)

familial dysautonomia [Riley-Day] (8C21.1)

Rendu-Osler-Weber disease (LA90.00)

Proteus syndrome (LD2C)

Sturge-Weber syndrome (LD23)

Enchondromatosis (2E83)

Maffucci syndrome (LD2F.1)

Angio-osteohypertrophic syndrome (LD26.60)

Coded Elsewhere: NAME syndrome (2F01)

Von Hippel-Lindau disease (5A75)

Focal dermal hypoplasia (LD27.0Y)

Epidermal naevus syndrome (LC02)

Lumbosacral dermal melanocytosis (LC10)

Naevus of Ota (LC10)

Naevus of Ito (LC10)

Dermal melanocyte hamartoma (LC10)

Hereditary leiomyomatosis and renal cell cancer (2C90.Y)

LD2D.0 Peutz-Jeghers syndrome

Peutz-Jeghers syndrome (PJS) is an autosomal dominant inherited disorder characterised by intestinal hamartomatous polyps in association with a distinct pattern of skin and mucosal macular melanin deposition. Patients have an increased risk of developing intestinal cancer.

LD2D.1 Neurofibromatoses

The neurofibromatoses (NF) are related genetic disorders which affect bone, soft tissue, skin and nervous system. In NF type 1 neurofibromas develop in the skin and elsewhere: these can cause problems as a result of their visibility in the skin, compression of vital internal structures or from malignant degeneration. Neuromas of the acoustic nerve are the predominant problem in NF type 2.

LD2D.10 Neurofibromatosis type 1

Neurofibromatosis type 1 (NF1) is an inherited, multi-system, neurocutaneous disorder that predisposes to the development of benign and malignant tumours. Two of the following criteria are required to diagnose NF1: six or more café au lait patches, neurofibromas, i.e. peripheral nerve sheath tumours manifesting as cutaneous, sub-cutaneous or plexiform lesions, skin-fold freckling, two or more iris Lisch nodules, an optic pathway glioma, a specific bony dysplasia (thinning of the long bone cortex, sphenoid wing dysplasia), an affected first-degree relative.

Inclusions: von Recklinghausen disease

LD2D.11 Neurofibromatosis type 2

Neurofibromatosis type 2 (NF2) is a tumour-prone disorder characterised by the development of multiple schwannomas and meningiomas.

LD2D.12 Neurofibromatosis type 3

LD2D.1Y Other specified neurofibromatoses

LD2D.1Z Neurofibromatosis, unspecified

LD2D.2 Tuberous sclerosis

A disease caused by a dominant mutation of 9q34 (TSC1) or 16p13 (TSC2). This disease may present with facial angiofibromas, Koenen tumours, fibrous plaques on the forehead and scalp, renal angiomyolipomas, subependymal nodules, multiple cortical tuber or retinal hamartoma, epilepsy, or mental retardation.

Inclusions: Bourneville disease

Coded Elsewhere: Autosomal dominant polycystic kidney disease type 1 with tuberous sclerosis (LD2F.1Y)

LD2D.3 Gardner syndrome

Gardner syndrome develops adenomatous polyps throughout the gastrointestinal tract, accompanied by extracolonic manifestations, including periampullary adenomas, papillary carcinoma of the thyroid, hepatoblastoma, osteomas of the mandible and skull, epidermal cysts, and desmoid tumours. Gardner syndrome is a term used to refer to patients in whom these extraintestinal features are unusually prominent.

LD2D.4 Gorlin syndrome

Gorlin syndrome, also known as naevoid basal cell carcinoma syndrome (NBCCS), is a hereditary condition characterised by a wide range of developmental abnormalities (odontogenic keratocysts of the jaws, hyperkeratosis of palms and soles, skeletal abnormalities, intracranial ectopic calcifications, and facial dysmorphism) and a predisposition to neoplasms (multiple basal cell carcinomas, medulloblastoma).

Inclusions: Naevoid basal cell carcinoma syndrome

LD2D.Y Other specified phakomatoses or hamartoneoplastic syndromes

LD2D.Z Phakomatoses or hamartoneoplastic syndromes, unspecified

LD2E Syndromes with structural anomalies due to inborn errors of metabolism

Coded Elsewhere: Disorders of cholesterol synthesis (5C52.10)

Pyruvate dehydrogenase complex deficiency (5C53.02)

Inborn errors of glycosylation or other specified protein modification (5C54)

Fabry disease (5C56.01)

Mucolipidosis (5C56.20)

Oligosaccharidosis (5C56.21)

Mucopolysaccharidosis (5C56.3)

Pseudo-Zellweger syndrome (5C57.Y)

Hypophosphatasia (5C64.3)

Classical homocystinuria (5C50.B)

Encephalopathy due to sulfite oxidase deficiency (5C50.B)

Mucosulfatidosis (5C56.0Y)

Zellweger syndrome (5C57.0)

Infantile Refsum disease (5C57.1)

Menkes disease (5C64.0Y)

LD2F Syndromes with multiple structural anomalies, without predominant body system involvement

Coded Elsewhere: Congenital rubella syndrome (KA62.8)

Congenital cytomegalovirus infection (KA62.3)

Perinatal Herpes simplex infection (KA62.A)

Congenital Epstein-Barr virus infection (KA62.1)

Congenital parvovirus syndrome (KA62.7)

Congenital enterovirus infection (KA62.5)

Congenital toxoplasmosis (KA64.0)

Embryofetopathy due to maternal phenylketonuria (5C50.02)

LD2F.0 Toxic or drug-related embryofetopathies

Coded Elsewhere: Uterovaginal malformation due to diethylstilbestrol syndrome (LB44.6)

LD2F.00 Fetal alcohol syndrome

Fetal alcohol syndrome is a malformation syndrome caused by maternal consumption of alcohol during pregnancy. It is characterised by prenatal and/or postnatal growth deficiency (weight and/or height <10th percentile); a unique cluster of minor facial anomalies (short palpebral fissures, flat and smooth philtrum, and thin upper lip) that presents across all ethnic groups, is identifiable at birth, and does not diminish with age. Affected children present severe central nervous system abnormalities including: microcephaly, cognitive and behavioural impairment (intellectual disability, deficit in general cognition, learning and language, executive function, visual-spatial processing, memory, and attention).

Coded Elsewhere: Neurodevelopmental syndrome due to prenatal alcohol exposure (6A0Y)

LD2F.01 Fetal hydantoin syndrome

Fetal hydantoin syndrome is a fetopathy likely to occur when a pregnant woman takes the anticonvulsant drug phenytoin (diphenylhydantoin) for epileptic seizures. In utero exposure to this drug may result in a characteristic dysmorphic syndrome in the newborn, including low-set hair, short neck with pterygium colli, small nose, deep nasal bridge, epicanthus, hypertelorism, large mouth, malformed ears, hypoplastic distal phalanges of the fingers and toes and finger-like thumbs. These dysmorphic features are often associated with growth retardation and delayed psychomotor development. The mechanism underlying these anomalies has been shown to depend on maternal genetic characteristics, i.e. on maternal capacity to detoxify intermediate metabolites of phenytoin.

LD2F.02 Embryofetopathy due to oral anticoagulant therapy

A condition caused by exposure of the embryo or fetus to anticoagulants during the antenatal period. This disease may present with optic nerve anomaly, optic atrophy, anomaly of the papilla, blindness, or choanal atresia.

LD2F.0Y Other specified toxic or drug-related embryofetopathies

LD2F.0Z Toxic or drug-related embryofetopathies, unspecified

LD2F.1 Syndromes with multiple structural anomalies, not of environmental origin

Coded Elsewhere: Fraser syndrome (LD2H.0)

Waardenburg-Shah syndrome (LD2H.3)

Oculocerebrorenal syndrome (5C60.0)

Albinism - black lock - cell migration disorder of the neurocytes of the gut - sensorineural deafness (LD2H.Y)

Bardet-Biedl syndrome (5A61.0)

Blepharocheilodontic syndrome (LD27.0Y)

Cat-eye syndrome (LD41.P)

Cataract - intellectual deficit - hypogonadism (5A61.0)

CHARGE syndrome (5A61.0)

Coffin-Siris syndrome (LD27.0Y)

Dubowitz syndrome (LD27.0Y)

Ectodermal dysplasia - ectrodactyly - macular dystrophy (LD27.0Y)

Ectrodactyly - ectodermal dysplasia - cleft lip or palate (LD27.0Y)

Ectrodactyly - ectodermal dysplasia without clefting (LD27.0Y)

Hirschsprung disease - deafness - polydactyly (LD2H.Y)

Limb-mammary syndrome (LD27.0Y)

Marshall syndrome (LD27.0Y)

MODY 5 syndrome (5A13.6)

Nijmegen breakage syndrome-like disorder (4A01.31)

Papillorenal syndrome (LA13.7Y)

Perrault syndrome (LD2H.Y)

Phocomelia - ectrodactyly - deafness - sinus arrhythmia (LD2H.Y)

Shwachman-Diamond syndrome (3A70.0)

Smith-Magenis syndrome (LD44.H1)

Split hand - split foot - deafness (LD2H.Y)

Triple A syndrome (5A74.Y)

Waardenburg syndrome (EC23.2Y)

WAGR syndrome (LD2A.Y)

Williams-Beuren syndrome (LD44.70)

Gorham-Stout disease (FB86.2)

Alagille syndrome (LB20.0Y)

Deafness – onychodystrophy (LD27.0Y)

Autosomal recessive cutis laxa, type 3 (LD28.2)

Macrocephaly – alopecia – cutis laxa – scoliosis syndrome (LD28.2)

SCARF syndrome (LD28.2)

Lethal restrictive dermopathy (EE6Y)

Encephalocraniocutaneous lipomatosis (EF02.1)

Dahlberg-Borer-Newcomer syndrome (LD27.0Y)

LD2F.10 Prune belly syndrome

A syndrome is characterised by cryptorchidism, urinary tract defects, and poor development of the abdominal muscles causing the skin on the abdomen to wrinkle.

LD2F.11 VATER association

VACTERL/VATER is an association of congenital malformations typically characterised by the presence of at least three of the following: vertebral defects, anal atresia, cardiac defects, tracheo-oesophageal fistula, renal anomalies, and limb abnormalities.

LD2F.12 Sirenomelia

Sirenomelia is a rare lethal malformation characterised by severe anomalies of the caudal part of the fetus that include a single lower limb, with various degrees of involvement ranging from single to separate femurs in the same skin shaft, presence of two feet (sympode mermaid) or one foot (monopode mermaid), to absence of both feet (ectromelic mermaid). Urogenital anomalies are also present and include bilateral renal agenesis, absence of outflow tract and absence of external genitalia. Imperforate anus and sacro-coccygeal agenesis have also been reported. Together these malformations comprise the extreme form of the caudal regression sequence.

LD2F.13 Meckel-Gruber syndrome

Meckel syndrome (MKS) is a monogenic disease characterised by a combination of renal cysts and variably associated features, including developmental anomalies of the central nervous system (usually occipital encephalocele), hepatic ductal dysplasia and cysts, and polydactyly., and a lethal course, with death occurring in the perinatal period.

LD2F.14 MURCS association

MURCS association, which stands for Müllerian duct aplasia (MU), congenital renal dysplasia (R), cervical somite anomalies (CS), is the atypical (or type II) form of Mayer-Rokitansky-Küster-Hauser syndrome, characterised by utero-vaginal atresia in otherwise normal females as well associated kidney and skeletal abnormalities and hearing problems.

LD2F.15 Noonan syndrome

Noonan Syndrome is characterised by short stature, facial dysmorphism and congenital heart defects. The main facial features of NS are hypertelorism with down-slanting palpebral fissures, ptosis and low-set posteriorly rotated ears with a thickened helix. The cardiovascular defects most commonly associated with this condition are pulmonary stenosis and hypertrophic cardiomyopathy. Other associated features are webbed neck, chest deformity, mild intellectual deficit, cryptorchidism, poor feeding in infancy, bleeding tendency and lymphatic dysplasia. The syndrome is transmitted as an autosomal dominant trait.

LD2F.16 Otomandibular dysplasia

Any condition characterised by malformation of facial bones and muscles. These conditions may present with eyes that slant downward, sparse eyelashes, eyelid coloboma, hearing loss, underdeveloped or absent vertebrae, or cleft palate.

LD2F.1Y Other specified syndromes with multiple structural anomalies, not of environmental origin

LD2F.1Z Syndromes with multiple structural anomalies, not of environmental origin, unspecified

LD2F.Y Other specified syndromes with multiple structural anomalies, without predominant body system involvement

LD2F.Z Syndromes with multiple structural anomalies, without predominant body system involvement, unspecified

LD2G Conjoined twins

A condition characterised as twins that are physically united at some part or parts of their bodies at the time of birth.

LD2H Syndromic genetic deafness

Coded Elsewhere: CATCH 22 phenotype (LD44.N0)

Pendred syndrome (5A00.02)

Generalised resistance to thyroid hormone (5A05)

CHARGE syndrome (5A61.0)

Deafness - opticoacoustic nerve atrophy - dementia (5C53.2Y)

Ectodermal dysplasia - sensorineural deafness (LD27.0Y)

Hypoparathyroidism - deafness - renal disease (LD27.0Y)

Renal tubular acidosis - deafness (GB90.44)

Stapes ankylosis with broad thumbs and toes (LD2F.1Y)

Stickler syndrome (LD2F.1Y)

Thiamine-responsive megaloblastic anaemia syndrome (5C63.Y)

Mitochondrial encephalopathy with lactic acidosis and stroke-like episodes (8C73.Y)

Norrie disease (LD21.Y)

Fechtner syndrome (3B64.01)

Spondyloepiphyseal dysplasia, MacDermot type (LD24.3)

Oral-facial-digital syndrome type 1 (LD25.00)

Oral-facial-digital syndrome type 2 (LD25.00)

Oral-facial-digital syndrome type 3 (LD25.00)

Oral-facial-digital syndrome type 4 (LD25.00)

Oral-facial-digital syndrome type 6 (LD25.00)

Oral-facial-digital syndrome type 8 (LD25.00)

Otopalatodigital syndrome (LD25.1)

Kearns-Sayre syndrome (9C82.0)

Multiple synostoses syndrome (LD26.3)

Arthrogryposis-like hand anomaly - sensorineural deafness (LD26.4Y)

Cockayne syndrome (LD2B)

Keratitis – ichthyosis – deafness syndrome (LD27.2)

Connexin palmoplantar keratoderma with sensorineural deafness (EC20.30)

Deafness – enamel hypoplasia – nail defects (LD27.0Y)

Tietz hypomelanosis – deafness syndrome (EC23.2Y)

LEOPARD syndrome (LD2F.1Y)

Cutis verticis gyrata - retinitis pigmentosa - sensorineural deafness (LD27.Y)

Deafness, lymphoedema and leukaemia syndrome (BD93.0)

Long QT syndrome with hearing impairment (BC65.0)

Infantile Bartter syndrome with deafness (GB90.43)

LD2H.0 Fraser syndrome

Fraser syndrome is a rare syndrome characterised by cryptophthalmos and syndactyly and associated with a wide variety of other anomalies including: middle and outer ear malformations; high-arched palate; cleavage along the midplane of nares and tongue; hypertelorism; laryngeal stenosis; wide separation of symphysis pubis; displacement of umbilicus and nipples; absent or multicystic kidneys; bicornuate uterus, malformed Fallopian tubes, fusion of labia and enlargement of clitoris in girls; and undescended testes and small penis with hypospadias in boys.

LD2H.1 Neuropathy with hearing impairment

Neuropathy with hearing impairment is characterised by the association of sensorineural hearing impairment and peripheral demyelinating and predominantly sensory neuropathy.

LD2H.2 Progressive deafness with stapes fixation

LD2H.3 Waardenburg-Shah syndrome

In this syndrome the phenotype includes not only the classical features of Waardenburg syndrome but also Hirschsprung disease. It may be caused by mutations in SOX10, EDN3 or EDNRB genes.

LD2H.4 Usher syndrome

Usher syndrome is the most common cause of hereditary combined deafness-blindness, and is characterised by the association of sensorineural deafness (usually congenital) with retinitis pigmentosa and progressive vision loss.

LD2H.Y Other specified syndromic genetic deafness

LD2H.Z Syndromic genetic deafness, unspecified

LD2Y Other specified multiple developmental anomalies or syndromes

LD2Z Multiple developmental anomalies or syndromes, unspecified

Chromosomal anomalies, excluding gene mutations (BlockL1‑LD4)

Any disease caused by alteration of the number or structure of chromosomes.

LD40 Complete trisomies of the autosomes

Any disease caused by the presence of one extra autosome, for a total of three. Confirmation is through observation of a supernumerary autosome by karyotyping.

LD40.0 Complete trisomy 21

Trisomy 21 is a chromosomal abnormality, characterised by the presence of a third (partial or total) copy of chromosome 21, which clinical manifestations include variable intellectual deficiency, muscular hypotonia and joint laxity, often associated with facial dysmorphism and variable malformations (essentially heart and digestive) and a risk of complications (epilepsy, leukemia, auto-immune and endocrine pathologies, earlier aging and Alzheimer disease.

Inclusions: Down syndrome

Coded Elsewhere: Keratoconus in Down syndrome (9A78.50)

LD40.1 Complete trisomy 13

Trisomy 13 is a chromosomal anomaly caused by the presence of an extra chromosome 13 and is characterised by brain malformations (holoprosencephaly), facial dysmorphism, ocular anomalies, postaxial polydactyly, visceral malformations (cardiopathy) and severe psychomotor retardation.

Inclusions: Patau syndrome

LD40.2 Complete trisomy 18

Trisomy 18 is a chromosomal abnormality associated with the presence of an extra chromosome 18 and characterised by growth delay, dolichocephaly, a characteristic facies, limb anomalies and visceral malformations.

LD40.Y Other specified complete trisomies of the autosomes

LD40.Z Complete trisomies of the autosomes, unspecified

LD41 Duplications of the autosomes

LD41.0 Duplications of chromosome 1

LD41.00 Duplications of the long arm of chromosome 1

LD41.01 Duplications of the short arm of chromosome 1

LD41.0Y Other specified duplications of chromosome 1

LD41.0Z Duplications of chromosome 1, unspecified

LD41.1 Duplications of chromosome 2

LD41.10 Duplications of the long arm of chromosome 2

LD41.11 Duplications of the short arm of chromosome 2

LD41.1Y Other specified duplications of chromosome 2

LD41.1Z Duplications of chromosome 2, unspecified

LD41.2 Duplications of chromosome 3

LD41.20 Duplications of the long arm of chromosome 3

LD41.21 Duplications of the short arm of chromosome 3

LD41.2Y Other specified duplications of chromosome 3

LD41.2Z Duplications of chromosome 3, unspecified

LD41.3 Duplications of chromosome 4

LD41.30 Duplications of the long arm of chromosome 4

LD41.31 Duplications of the short arm of chromosome 4

LD41.3Y Other specified duplications of chromosome 4

LD41.3Z Duplications of chromosome 4, unspecified

LD41.4 Duplications of chromosome 5

LD41.40 Duplications of the long arm of chromosome 5

LD41.41 Duplications of the short arm of chromosome 5

LD41.4Y Other specified duplications of chromosome 5

LD41.4Z Duplications of chromosome 5, unspecified

LD41.5 Duplications of chromosome 6

LD41.50 Duplications of the long arm of chromosome 6

LD41.51 Duplications of the short arm of chromosome 6

LD41.5Y Other specified duplications of chromosome 6

LD41.5Z Duplications of chromosome 6, unspecified

LD41.6 Duplications of chromosome 7

LD41.60 Duplications of the long arm of chromosome 7

LD41.61 Duplications of the short arm of chromosome 7

LD41.6Y Other specified duplications of chromosome 7

LD41.6Z Duplications of chromosome 7, unspecified

LD41.7 Duplications of chromosome 8

LD41.70 Duplications of the long arm of chromosome 8

LD41.71 Duplications of the short arm of chromosome 8

LD41.7Y Other specified duplications of chromosome 8

LD41.7Z Duplications of chromosome 8, unspecified

LD41.8 Duplications of chromosome 9

LD41.80 Duplications of the long arm of chromosome 9

LD41.81 Duplications of the short arm of chromosome 9

LD41.8Y Other specified duplications of chromosome 9

LD41.8Z Duplications of chromosome 9, unspecified

LD41.9 Duplications of chromosome 10

LD41.90 Duplications of the long arm of chromosome 10

LD41.91 Duplications of the short arm of chromosome 10

LD41.9Y Other specified duplications of chromosome 10

LD41.9Z Duplications of chromosome 10, unspecified

LD41.A Duplications of chromosome 11

LD41.B Duplications of chromosome 12

LD41.B0 Duplications of the long arm of chromosome 12

LD41.B1 Duplications of the short arm of chromosome 12

LD41.BY Other specified duplications of chromosome 12

LD41.BZ Duplications of chromosome 12, unspecified

LD41.C Duplications of chromosome 13

LD41.D Duplications of chromosome 14

LD41.E Duplications of chromosome 15

LD41.F Duplications of chromosome 16

LD41.F0 Duplications of the long arm of chromosome 16

LD41.F1 Duplications of the short arm of chromosome 16

LD41.FY Other specified duplications of chromosome 16

LD41.FZ Duplications of chromosome 16, unspecified

LD41.G Duplications of chromosome 17

LD41.G0 Duplications of the long arm of chromosome 17

LD41.G1 Duplications of the short arm of chromosome 17

LD41.GY Other specified duplications of chromosome 17

LD41.GZ Duplications of chromosome 17, unspecified

LD41.H Duplications of chromosome 18

LD41.H0 Duplications of the long arm of chromosome 18

LD41.H1 Duplications of the short arm of chromosome 18

LD41.HY Other specified duplications of chromosome 18

LD41.HZ Duplications of chromosome 18, unspecified

LD41.J Duplications of chromosome 19

LD41.J0 Duplications of the long arm of chromosome 19

LD41.J1 Duplications of the short arm of chromosome 19

LD41.JY Other specified duplications of chromosome 19

LD41.JZ Duplications of chromosome 19, unspecified

LD41.K Duplications of chromosome 20

LD41.K0 Duplications of the long arm of chromosome 20

LD41.K1 Duplications of the short arm of chromosome 20

LD41.KY Other specified duplications of chromosome 20

LD41.KZ Duplications of chromosome 20, unspecified

LD41.L Duplications of chromosome 21

LD41.M Duplications of chromosome 22

LD41.N Extra ring or dicentric chromosomes

LD41.P Duplications with other complex rearrangements

LD41.Q Extra marker chromosomes

LD41.Y Other specified duplications of the autosomes

LD41.Z Duplications of the autosomes, unspecified

LD42 Polyploidies

Any disease caused by one or more additional sets of chromosomes. Non mosaic version of these diseases are characterised by gross fetal malformation or death of the fetus. Confirmation is through observation of supernumerary sets of chromosomes by karyotyping.

LD42.0 Triploidy

A disease caused by one additional set of chromosomes, for a total of 69 chromosomes. Triploidy can present with albuminuria, oedema, or hypertension in the mother. The fetus may present with microcephaly and a placenta that is enlarged and filled with cysts in the case of extra maternally inherited chromosomes, while extra paternally inherited chromosomes cause severe growth problems, an enlarged head, and a small placenta that does not have cysts. Non-mosaic triploidy is highly lethal, and is rarely observed in live births. Confirmation is through observation of an additional set of chromosomes by karyotyping.

LD42.1 Tetraploidy

A disease caused by two additional sets of chromosomes, for a total of 92 chromosomes. This disease commonly results in spontaneous abortion during the first trimester. Live births of tetraploidy individuals are very rare. These cases are characterised by facial dysmorphism, severely delayed growth and developmental delay. Confirmation is through observation of two additional set of chromosomes by karyotyping.

LD42.Y Other specified polyploidies

LD42.Z Polyploidies, unspecified

LD43 Complete monosomies of the autosomes

LD43.0 Complete monosomy of autosome

LD43.1 Mosaic monosomy of autosome

Any disease caused by embryonic fusion or loss of an autosome early in embryonic development, resulting in a subset of cells in the body having only one of a pair of autosomes.

LD43.Y Other specified complete monosomies of the autosomes

LD43.Z Complete monosomies of the autosomes, unspecified

LD44 Deletions of the autosomes

LD44.0 Chromosome replaced with ring or dicentric with normal number of chromosomes

LD44.1 Deletions of chromosome 1

LD44.10 Deletions of the long arm of chromosome 1

LD44.11 Deletions of the short arm of chromosome 1

LD44.1Y Other specified deletions of chromosome 1

LD44.1Z Deletions of chromosome 1, unspecified

LD44.2 Deletions of chromosome 2

LD44.20 Deletions of the long arm of chromosome 2

LD44.21 Deletions of the short arm of chromosome 2

LD44.2Y Other specified deletions of chromosome 2

LD44.2Z Deletions of chromosome 2, unspecified

LD44.3 Deletions of chromosome 3

LD44.30 Deletions of the long arm of chromosome 3

LD44.31 Deletions of the short arm of chromosome 3

LD44.3Y Other specified deletions of chromosome 3

LD44.3Z Deletions of chromosome 3, unspecified

LD44.4 Deletions of chromosome 4

LD44.40 Deletions of the long arm of chromosome 4

LD44.41 Deletions of the short arm of chromosome 4

LD44.4Y Other specified deletions of chromosome 4

LD44.4Z Deletions of chromosome 4, unspecified

LD44.5 Deletions of chromosome 5

LD44.50 Deletions of the long arm of chromosome 5

LD44.51 Deletions of the short arm of chromosome 5

LD44.5Y Other specified deletions of chromosome 5

LD44.5Z Deletions of chromosome 5, unspecified

LD44.6 Deletions of chromosome 6

LD44.60 Deletions of the long arm of chromosome 6

LD44.61 Deletions of the short arm of chromosome 6

LD44.6Y Other specified deletions of chromosome 6

LD44.6Z Deletions of chromosome 6, unspecified

LD44.7 Deletions of chromosome 7

LD44.70 Deletions of the long arm of chromosome 7

LD44.71 Deletions of the short arm of chromosome 7

LD44.7Y Other specified deletions of chromosome 7

LD44.7Z Deletions of chromosome 7, unspecified

LD44.8 Deletions of chromosome 8

LD44.80 Deletions of the long arm of chromosome 8

Coded Elsewhere: Langer-Giedion syndrome (LD24.80)

LD44.81 Deletions of the short arm of chromosome 8

LD44.8Y Other specified deletions of chromosome 8

LD44.8Z Deletions of chromosome 8, unspecified

LD44.9 Deletions of chromosome 9

LD44.90 Deletions of the long arm of chromosome 9

LD44.91 Deletions of the short arm of chromosome 9

LD44.9Y Other specified deletions of chromosome 9

LD44.9Z Deletions of chromosome 9, unspecified

LD44.A Deletions of chromosome 10

LD44.A0 Deletions of the long arm of chromosome 10

LD44.A1 Deletions of the short arm of chromosome 10

LD44.AY Other specified deletions of chromosome 10

LD44.AZ Deletions of chromosome 10, unspecified

LD44.B Deletions of chromosome 11

LD44.B0 Deletions of the long arm of chromosome 11

LD44.B1 Deletions of the short arm of chromosome 11

These deletions may give rise to the Paris-Trousseau syndrome, a very rare disorder in which intellectual deficit, cardiac malformations and facial abnormalities are associated with thrombocytopenia and dysmegakaryopoiesis.

Coded Elsewhere: WAGR syndrome (LD2A.Y)

LD44.BY Other specified deletions of chromosome 11

LD44.BZ Deletions of chromosome 11, unspecified

LD44.C Deletions of chromosome 12

LD44.C0 Deletions of the long arm of chromosome 12

LD44.C1 Deletions of the short arm of chromosome 12

LD44.CY Other specified deletions of chromosome 12

LD44.CZ Deletions of chromosome 12, unspecified

LD44.D Deletions of chromosome 13

LD44.E Deletions of chromosome 14

LD44.F Deletions of chromosome 15

LD44.G Deletions of chromosome 16

LD44.G0 Deletions of the long arm of chromosome 16

LD44.G1 Deletions of the short arm of chromosome 16

Coded Elsewhere: Autosomal dominant polycystic kidney disease type 1 with tuberous sclerosis (LD2F.1Y)

Alpha thalassaemia - intellectual deficit syndrome (3A50.1)

LD44.GY Other specified deletions of chromosome 16

LD44.GZ Deletions of chromosome 16, unspecified

LD44.H Deletions of chromosome 17

LD44.H0 Deletions of the long arm of chromosome 17

LD44.H1 Deletions of the short arm of chromosome 17

Coded Elsewhere: Miller-Dieker syndrome (LD20.1)

LD44.HY Other specified deletions of chromosome 17

LD44.HZ Deletions of chromosome 17, unspecified

LD44.J Deletions of chromosome 18

LD44.J0 Deletions of the long arm of chromosome 18

LD44.J1 Deletions of the short arm of chromosome 18

LD44.JY Other specified deletions of chromosome 18

LD44.JZ Deletions of chromosome 18, unspecified

LD44.K Deletions of chromosome 19

LD44.K0 Deletions of the long arm of chromosome 19

LD44.K1 Deletions of the short arm of chromosome 19

LD44.KY Other specified deletions of chromosome 19

LD44.KZ Deletions of chromosome 19, unspecified

LD44.L Deletions of chromosome 20

LD44.L0 Deletions of the long arm of chromosome 20

LD44.L1 Deletions of the short arm of chromosome 20

LD44.LY Other specified deletions of chromosome 20

LD44.LZ Deletions of chromosome 20, unspecified

LD44.M Deletions of chromosome 21

LD44.N Deletions of chromosome 22

LD44.N0 CATCH 22 phenotype

Monosomy 22q11 (DiGeorge Velocardiofacial syndrome, DGS/VCF) syndrome is a chromosomal anomaly characterised by the association of several variable malformations: hypoplastic thymus and parathyroid glands, congenital conotruncal heart defects, a subtle but characteristic facial dysmorphism, cleft palate or velar insufficiency, and learning difficulties.

Inclusions: Pharyngeal pouch syndrome

DiGeorge syndrome

Velocardiofacial syndrome

LD44.NY Other specified deletions of chromosome 22

LD44.NZ Deletions of chromosome 22, unspecified

LD44.P Deletions with other complex rearrangements

LD44.Y Other specified deletions of the autosomes

LD44.Z Deletions of the autosomes, unspecified

LD45 Uniparental disomies

Any disease caused by the inheritance of two homologous copies of a chromosome from one parent, and none from the other parent. Confirmation is by observation of identical chromosomes pairs by genetic testing.

LD45.0 Uniparental disomies of maternal origin

Any disease characterised by the inheritance of two homologous copies of a chromosome from the mother, and none from the father. Confirmation is by observation of identical chromosome pairs, and matching to a maternal chromosome, by genetic testing.

LD45.1 Uniparental disomies of paternal origin

Any disease caused by the inheritance of two homologous copies of a chromosome from the father, and none from the mother. Confirmation is by observation of identical chromosome pairs, and matching to a paternal chromosome, by genetic testing.

LD45.Y Other specified uniparental disomies

LD45.Z Uniparental disomies, unspecified

LD46 Imprinting errors

LD46.0 Maternal imprinting error

LD46.1 Paternal imprinting error

LD46.Y Other specified imprinting errors

LD46.Z Imprinting errors, unspecified

LD47 Balanced rearrangements or structural markers

Any disease caused by alteration of chromosomal structure with no net gain or loss of genetic material, or by the presence of a marker chromosome. Confirmation is through observation of a balanced chromosomal rearrangement by genetic testing.

Inclusions: Robertsonian and balanced reciprocal translocations and insertions

LD47.0 Balanced translocation and insertion in normal individual

A condition caused by translocation of genetic material between chromosomes with no net gain or loss of genetic material, in an individual demonstrating no abnormalities. Confirmation is through observation of a balanced translocation and insertion by genetic testing.

LD47.1 Chromosome inversion in normal individual

Any disease caused by inversion of genetic material on a chromosome, in an individual demonstrating no abnormalities. Confirmation is through observation of a chromosomal inversion by genetic testing.

LD47.2 Balanced autosomal rearrangement in abnormal individual

Any disease caused by alteration of autosome structure with no net gain or loss of genetic material, in an individual demonstrating abnormalities. Confirmation is through observation of a balanced chromosomal rearrangement by genetic testing.

LD47.3 Balanced sex or autosomal rearrangement in abnormal individual

Any disease caused by alteration of chromosomal structure with no net gain or loss of genetic material, in an individual demonstrating abnormalities. Confirmation is through observation of a balanced chromosomal rearrangement by genetic testing.

LD47.4 Autosomal fragile site

Any disease caused by presence of a fragile site on an autosome. These diseases may present as asymptomatic. Confirmation is through observation of a fragile site by genetic testing.

LD47.Y Other specified balanced rearrangements or structural markers

LD47.Z Balanced rearrangements or structural markers, unspecified

Sex chromosome anomalies (BlockL2‑LD5)

Any disease caused by change in the number or structure of the X or Y chromosome. Confirmation is by observation of a chromosomal anomaly by genetic testing.

LD50 Number anomalies of chromosome X

LD50.0 Turner syndrome

Karyotype missing one X chromosome (45, X0 or 45,XO/46,XX mosaicism) ; gonads: ovaries (streak); phenotype female with short stature, amenorrhea (hypergonadotrophic hypogonadism), absence of sexual development, webbed neck, low set ears, posterior hairline, widely-spaced nipples, short fourth metacarpals, and increased carrying angle at the elbow (cubitus valgus). Often associated with renal, cardiac and ocular abnormalities.

Inclusions: Monosomy X

Exclusions: Noonan syndrome (LD2F.15)

LD50.00 Karyotype 45, X

A disease affecting females, caused by absence of one of the two X chromosomes. This disease may present with short stature, extra folds of skin on the neck, a low hairline at the back of the neck, puffiness or swelling of the hands and feet, skeletal abnormalities, ovarian hypofunction or premature ovarian failure, kidney problems, or heart defects. Confirmation is through observation of only one X chromosome by karyotyping.

LD50.01 Karyotype 46, X iso Xq

A disease affecting females, caused by one of the two X chromosomes consisting of two q arms, which are structurally identical and contain the same genes. This disease may present with short stature, extra folds of skin on the neck, a low hairline at the back of the neck, puffiness or swelling of the hands and feet, skeletal abnormalities, ovarian hypofunction or premature ovarian failure, kidney problems, or heart defects. This disease may be differentiated from classical Turner Syndrome by a near complete lack of gonadal development, resulting in a lack of menstruation or breast development. Confirmation is through observation of an iso Xq chromosome by karyotyping.

LD50.02 Karyotype 46, X with abnormal sex chromosome, except iso Xq

LD50.03 Mosaicism, 45, X, 46, XX or XY

A disease caused by embryonic fusion, or by the loss of one of the sex chromosomes from a cell early in embryonic development; Gonadal status: normal or variable abnormalities of sexual anatomy, maturation or function. Phenotype: normal, or abnormal sexual development.

LD50.04 Mosaicism, 45, X or other cell line with abnormal sex chromosome

A disease caused by embryonic fusion or the structural mutation of a sex chromosome early in embryonic development, resulting in a subset of cells in the body having one normal copy of the X chromosome and one abnormal sex chromosome. This disease may present with short stature, sexual organ dysfunction, or may be asymptomatic.

LD50.1 Karyotype 47,XXX

Trisomy X is a sex chromosome anomaly with a variable phenotype caused by the presence of an extra X chromosome in females (47,XXX instead of 46,XX). Most individuals are only mildly affected or asymptomatic, the most common physical features including tall stature, epicanthal folds, hypotonia and clinodactyly, with seizures, renal and genitourinary abnormalities, and premature ovarian failure being also associated findings.

LD50.2 Mosaicism, lines with various numbers of X chromosomes

A disease caused by embryonic fusion or gain or loss of X chromosomes early in embryonic development, resulting in a subset of cells in the body having an abnormal number of X chromosomes. This disease may present with abnormal height, genitourinary abnormalities, or may be asymptomatic.

LD50.3 Klinefelter syndrome

Klinefelter syndrome defines a group of chromosomal disorders in which there is at least one extra X chromosome compared with the normal 46,XY male karyotype. The effects on physical features and on physical and cognitive development increase with the number of extra X's, and each extra X is associated with an intelligence quotient (IQ) decrease of approximately 15-16 points, with language most affected, particularly expressive language skills.

LD50.30 Klinefelter syndrome with karyotype 47,XXY, regular

Karyotype 47 XXY; gonads: testes (hypogonadism) small and firm with decreased spermatogenesis ; phenotype male with associated congenital abnormalities (decreased virilization due to decreased testosterone production, long arms and legs, short trunk, psychosocial problems).

LD50.31 Klinefelter syndrome, male with more than two X chromosomes

A disease affecting males, caused by the presence of more than two X chromosomes in each cell. This disease is characterised by impaired sexual development, intellectual disability, distinctive facial features, skeletal abnormalities, poor coordination, and severe problems with speech. This disease may be differentiated from classic Klinefelter syndrome by increased severity of symptoms. Confirmation is through observation of more than two X chromosomes by karyotyping.

LD50.3Y Other specified Klinefelter syndrome

LD50.Y Other specified number anomalies of chromosome X

LD50.Z Number anomalies of chromosome X, unspecified

LD51 Structural anomalies of chromosome X, excluding Turner syndrome

LD52 Number anomalies of chromosome Y

LD52.0 Male with 46,XX karyotype

A disease affecting males, characterised by hypergonadotrophic hypogonadism, testosterone deficiency, and infertility. This condition may also present with hypospadias. This disease may be associated with abnormal crossing over of the sex chromosomes during meiosis in the father, resulting in the SRY gene being present on one or both copies of the X chromosome.

LD52.1 Male with double or multiple Y

A condition affecting males, caused by the presence of supernumerary Y chromosomes. This condition is asymptomatic. Confirmation is through observation of supernumerary Y chromosomes by karyotyping.

LD52.Y Other specified number anomalies of chromosome Y

LD52.Z Number anomalies of chromosome Y, unspecified

LD53 Structural anomalies of chromosome Y

Coded Elsewhere: Chromosome Y deletion (5A81.1)

LD54 Male with sex chromosome mosaicism

Any disease affecting males, caused by embryonic fusion or gain or loss of a sex chromosome early in embryonic development, resulting in a subset of cells in the body having an abnormal number of sex chromosomes. These diseases may present with deficiencies in testosterone, abnormalities of sexual development, or infertility.

LD55 Fragile X chromosome

Fragile X syndrome is a rare genetic disease associated with mild to severe intellectual deficit that may be associated with behavioural disorders and characteristic physical features.

Inclusions: Fragile X syndrome

LD56 Chimaera 46, XX, 46, XY

A disease caused by XX and XY embryonic fusion or two distinct loss event of a sex chromosome from a XXY embryo early in development. This results in a subset of cells in the body having a XX karyotype, while other cells demonstrate a XY karyotype. This disease may present with abnormal genital development.

LD56.0 Androgenetic chimaera

LD56.1 Gynogenetic chimaera

LD56.Y Other specified chimaera 46, XX, 46, XY

LD56.Z Chimaera 46, XX, 46, XY, unspecified

LD5Y Other specified sex chromosome anomalies

LD5Z Sex chromosome anomalies, unspecified

LD7Y Other specified chromosomal anomalies, excluding gene mutations

LD7Z Chromosomal anomalies, excluding gene mutations, unspecified

LD90 Conditions with disorders of intellectual development as a relevant clinical feature

Coded Elsewhere: Lesch-Nyhan syndrome (5C55.01)

Hydrocephalus with stenosis of the aqueduct of Sylvius (LA04.0)

Pelizaeus-Merzbacher disease (8A44.0)

Hereditary sensory and autonomic neuropathy type IV (8C21.2)

Joubert syndrome (LD20.00)

Phenylketonuria (5C50.0)

Tyrosinaemia type 2 (5C50.12)

Carbamoylphosphate synthetase deficiency (5C50.A1)

Carnosinaemia (5C50.F1)

Homocarnosinosis (5C50.F2)

Syndromes with lissencephaly as a major feature (LD20.1)

Sjögren-Larsson syndrome (5C52.03)

Polymicrogyria (LA05.50)

Porencephaly (LA05.60)

Pyruvate dehydrogenase complex deficiency (5C53.02)

Brain-lung-thyroid syndrome (CB04.5)

Metachromatic leukodystrophy (5C56.02)

Neuronal ceroid lipofuscinosis (5C56.1)

Mucopolysaccharidosis type 2 (5C56.31)

Mucopolysaccharidosis type 6 (5C56.33)

Oculocerebrorenal syndrome (5C60.0)

CATCH 22 phenotype (LD44.N0)

Langer-Giedion syndrome (LD24.80)

Crigler-Najjar syndrome (5C58.00)

Fragile X chromosome (LD55)

Incontinentia pigmenti (LD27.00)

Tuberous sclerosis (LD2D.2)

Noonan syndrome (LD2F.15)

Congenital rubella syndrome (KA62.8)

Congenital cytomegalovirus infection (KA62.3)

Complete trisomy 21 (LD40.0)

Klinefelter syndrome, male with more than two X chromosomes (LD50.31)

Intellectual disability – enteropathy – deafness – neuropathy – ichthyosis – keratoderma syndrome (LD2H.Y)

Microcephaly - deafness - intellectual disability (LD2H.Y)

Schizophrenia - intellectual disability - deafness - retinitis (LD2H.Y)

Corneal anaesthesia - deafness - intellectual disability (LD2H.Y)

Ataxia - deafness - intellectual disability syndrome (LD2H.Y)

Retinitis pigmentosa - intellectual disability - deafness - hypogenitalism (LD2H.Y)

LD90.0 Angelman syndrome

Angelman syndrome is a neurogenetic disorder characterised by severe intellectual deficit and distinct facial dysmorphic (microcephaly, macrostomia, maxillary hypoplasia, prognathia), behavioural (outbursts of laughter with hand flapping, a happy demeanour, hyperactivity without aggression, short attention span, excitability and sleeping problems with decreased need to sleep, increased sensitivity to heat, attraction to and fascination with water), and neurological features (a puppet-like gait, ataxia and epileptic seizures).

LD90.1 Early-onset parkinsonism - intellectual deficit

Early-onset parkinsonism with intellectual deficit is a basal ganglia disorder characterised by parkinsonian-type symptoms (postural changes, tremor, rigidity), megalencephaly and variable intellectual deficit. Other signs are frontal bossing, persistent frontal lobe reflexes, strabismus and seizures.

LD90.2 Pelizaeus-Merzbacher-like disease

Pelizaeus-Merzbacher like disease (PMLD) is an autosomal recessive leukodystrophy sharing identical clinical and radiological features as X-linked Pelizaeus-Merzbacher disease (PMD; ).

LD90.3 Prader-Willi syndrome

Prader-Willi syndrome is a rare genetic disorder characterised by hypothalamic-pituitary abnormalities with severe hypotonia during the neonatal period and first two years of life and the onset of hyperphagia with a risk of morbid obesity during infancy and adulthood, learning difficulties and behavioural problems or severe psychiatric problems.

LD90.4 Rett syndrome

A condition in which apparently normal early development is followed by partial or complete loss of speech and of skills in locomotion and use of hands, together with deceleration in head growth, usually with an onset between seven and 24 months of age. Loss of purposive hand movements, hand-wringing stereotypies, and hyperventilation are characteristic. Social and play development are arrested but social interest tends to be maintained. Trunk ataxia and apraxia start to develop by age four years and choreoathetoid movements frequently follow. Severe mental retardation almost invariably results.

LD90.Y Other specified conditions with disorders of intellectual development as a relevant clinical feature

LD90.Z Conditions with disorders of intellectual development as a relevant clinical feature, unspecified

LD9Y Other specified developmental anomalies

LD9Z Developmental anomalies, unspecified