



WA Register of Developmental Anomalies

Inclusion List / Definition Guide

Below is a list of the most common developmental anomalies. It is not exhaustive. If there are anomalies you think should be notified but are not listed here, please contact the Register on 6458 2735 to discuss.

Blood

Thalassaemia
Sickle cell anaemia
Haemophilia

Cardiovascular System

Coarctation of aorta
Fallot's Tetralogy
VSD, ASD, PDA

Cerebral Palsy

Spastic cerebral palsy

- Quadriplegia
- Triplegia
- Diplegia
- Hemiplegia

Ataxic cerebral palsy

Dyskinetic cerebral palsy

- Athetoid
- Dystonic

Hypotonic cerebral palsy

Mixed types

Postnatally acquired up to 5 years of age

Chromosomal Anomalies

Trisomy 21

Trisomy 18

Trisomy 13

Turner syndrome

Klinefelter syndrome

Congenital Infections

Rubella
Toxoplasmosis
Cytomegalovirus
Syphilis

Eye

Anophthalmia
Microphthalmia
Congenital glaucoma
Congenital cataract

Gastro-Intestinal System

Cleft lip and/or palate
Tracheo-oesophageal
atresia/fistula
Pyloric stenosis
Intestinal atresia
Hirschsprung's disease
Ectopic/imperforate anus

Genital Organs

Undescended testis (if treated)
Hypospadias
Indeterminate sex

Metabolic Disorders

Phenylketonuria
Cystic Fibrosis
Congenital hypothyroidism
Adeno-genital disorder
Glycogen storage disorder
Lipid storage disorder
Albinism

Musculo-Skeletal System

Talipes (requiring treatment)
Polydactyly / syndactyly
Absent limbs/digits
Craniosynostosis
Osteogenesis imperfecta
Developmental dysplasia of hip (if treated)



Nervous System

Anencephaly
Spina bifida
Encephalocele
Congenital hydrocephalus
Microcephaly
Congenital Hearing Loss

Other

Fetal alcohol syndrome

Respiratory System

Pulmonary hypoplasia
Diaphragmatic hernia
Choanal atresia/stenosis
Congenital lung cyst

Skin

Naevi / haemangioma / birth mark if large, multiple or requiring treatment

Urinary System

Cystic kidney
Absent kidney
Ectopic kidney
Double ureter/collecting system
Vesico-ureteric reflux

This document can be made available in alternative formats on request.

© North Metropolitan Health Service 2020

