



## Western Australian Register of Developmental Anomalies

### Inclusion List/Definition Guide

Revised January 2014

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 9340 2735 to discuss.

#### **NERVOUS SYSTEM**

Anencephaly  
Spina bifida  
Encephalocele  
Congenital hydrocephalus  
Microcephaly  
Congenital Hearing Loss

#### **CARDIOVASUCLAR SYSTEM**

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

#### **RESPIRATORY SYSTEM**

Pulmonary hypoplasia  
Diaphragmatic hernia  
Choanal atresia/stenosis  
Congenital lung cyst

#### **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

#### **URINARY SYSTEM**

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

#### **MUSCULO-SKELETAL SYSTEM**

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

#### **EYE**

Anophthalmia  
Microphthalmia  
Congenital glaucoma  
Congenital cataract

#### **SKIN**

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

#### **METABOLIC DISORDERS**

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

#### **BLOOD**

Thalassaemia  
Sickle cell anaemia  
Haemophilia

#### **CONGENITAL INFECTIONS**

Rubella  
Toxoplasmosis  
Cytomegalovirus  
Syphilis

#### **CHROMOSOMAL ANOMALIES**

Trisomy 21  
Trisomy 18  
Trisomy 13  
Turner syndrome  
Klinefelter syndrome

#### **OTHER**

Fetal alcohol syndrome

#### **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