

Appendix 1 Types of fetus defects and birth defects

Types of fetus defects and birth defects

Diagnosis

Anencephalus

Spina bifida

Encephalocele

Congenital Hydrocephalus

Cleft Palate

Cleft Lip

Cleft Lip with Cleft Palate

Microtia (including Anotia)

Deformity of external ear(s) (except Microtia and Anotia)

Esophageal atresia or stenosis

Anorectal atresia (including Congenital Anorectal Malformations)

Hypospadias

Ectopocystis

Pes Equinovarus

Polydactylism

Syndactylia

Limb shortening

Congenital Diaphragmatic Hernia

Pcmphalus

Celoschisis

Conjoined Twins

Trisomy 21 syndrome

Congenital heart disease

Others

Note: Down's syndrome, neural tube defects, congenital heart defects, hydrocephalus, digestive tract

malformations and urinary malformations are most common defects in China. Defects were detected by prenatal Down's syndrome screening, NT examination and Ultrasound image examination during the second trimester; and the number and type of birth defects after childbirth are diagnosed by professional clinical team. Down syndrome (DS), caused by the trisomy, translocation, or partial trisomy of chromosome 21, is the most common genetic cause of intellectual disability. DS is diagnosed by neonatologist. Neural tube defects including spina bifida and hypospadias, were diagnosed by ultrasound examination and pediatric neurosurgeon. Congenital heart disease was diagnosis by Doppler ultrasound and CHD screening (pulse oximetry plus cardiac murmurs). Hydrocephalus, digestive tract malformations, urinary malformations and other defects also were also diagnosed either by ultrasound or some other specific diagnosis methods.