BIRTH DEFECTS

CASE DEFINITION FOR SURVEILLANCE

Case definition

Case definition for Washington State Birth Defects Surveillance System is based on ICD-9-CM diagnostic and procedure codes as they appear in the hospital medical records. Any child up to age one year, diagnosed or treated, with a reportable birth defect who was a Washington State resident at the time of birth, or treated in a Washington facility is reportable. Information for all stillbirths over 20 weeks gestation diagnosed with a reportable birth defect should also be reported. Currently, we receive a completed hard copy form including the following data elements: Child's name, medical record number, date of birth, sex, admission date, zip code, discharge date, ICD-9-CM code for diagnosis, diagnosis, ICD code for procedure, and procedure.

Table. List of Currently Reportable Birth Defects and the Corresponding ICD-9-CM and ICD-10-CM Codes

Birth Defect	ICD-9-CM	ICD-10-CM	Procedure Code (used with ICD9)
1. Anencephaly and similar anomalies	***740.0-740.2***	***Q00***	
Acrania	740.0	Q00.0	
Amyelencephalus	740.0	Q00.0	
Hemianencephaly	740.0	Q00.0	
Hemicephaly	740.0	Q00.0	
Craniorachischisis	740.1	Q00.1	
Iniencephaly	740.2	Q00.2	
2. Spina Bifida	***741.0, 741.9***	***Q05, Q07***	
With Hydrocephalus	741.0	Q05.4	
Arnold-Chiari syndrome, type II	741.0	Q07.0	
Without mention of Hydrocephalus	741.9	*Must select region of defect*	
Hydromeningocele	741.9	Q05.9	
Hydromyelocele	741.9	Q05.9	
Meningocele (spinal)	741.9	Q05.9	
3. Cleft Palate	***749.0***	***Q35***	
Cleft Palate, Unspecified	749.00	*No longer valid; must select bilateral, unilateral or medial*	
Cleft Palate, Unilateral, Complete	749.01	Q35.5	
Cleft Palate, Unilateral, Incomplete	749.02	*Hard* = Q35.1 *Soft* = Q35.3	
Cleft Uvula	749.02	Q35.7	

Birth Defect	ICD-9-CM	ICD-10-CM	Procedure Code (used with ICD9)
Cleft Palate, Bilateral, Complete	749.03	Q35.4	
Cleft Palate, Bilateral, Incomplete	749.04	*Hard* = Q35.0 *Soft* = Q35.2	
4. Cleft Lip	***749.1***	***Q36***	
Cheiloschisis *Congenital fissure of lip* *Harelip* *Labium Leporinum*	749.1 749.1 749.1 749.1	*Q36.9*	
Cleft Lip, Unspecified	749.10	Q36.9	
Cleft Lip, Unilateral, Complete	749.11	Q36.9	
Cleft Lip, Unilateral, Incomplete	749.12	Q36.9	
Cleft Lip, Bilateral, Complete	749.13	Q36.0	
Cleft Lip, Bilateral, Incomplete	749.14	Q36.0	
5. Cleft palate with cleft lip	***749.2***	***Q37***	
Cheilopalatoschisis	749.2	*Term no longer used*	
Cleft palate with cleft lip, unspecified	749.20	*No longer valid, must select bilateral or unilateral*	
Unilateral, complete	749.21	Q37.5	
Unilateral, incomplete	749.22	*Hard* = Q37.1 *Soft* = Q37.3	
Bilateral, complete	749.23	Q37.4	
Bilateral, incomplete	749.24	*Hard* = Q37.0 *Soft* = Q37.2	
6. Abnormalities of Abdominal Wall	***756.70***	***Q79.2, Q79.3***	
Omphalocele	756.79	Q79.2	53.41
Gastroschisis	756.79	Q79.3	54.71
7. Limb reduction defects	***755.20-755.4***	***Q71-Q73***	
Unspecified reduction deformity of upper limb	755.20	Q71.9	
Transverse deficiency of upper limb	755.21	Q71.0 - Q71.3	
Longitudinal deficiency of upper limb	755.22	Q71.4 – Q71.5	
Longitudinal deficiency combined, Involving Humerus, Radius, and Ulna (Complete or incomplete)	755.23	*Must select specific bone(s)* Q71.4, Q71.5, Q71.6	

Birth Defect	ICD-9-CM	ICD-10-CM	Procedure Code (used with ICD9)
Longitudinal deficiency. Humeral, complete or Partial(with or without distal deficiencies, Incomplete)	755.24	Q71.8	
Longitudinal deficiency, Radioulnar, complete or partial (with or without distal deficiencies, incomplete)	755.25	*Must select specific bone(s)* Q71.4, Q71.5, Q71.6	
Longitudinal deficiency, Radial, Complete or partial (with or without distal deficiencies, Incomplete)	755.26	Q71.4	
Longitudinal deficiency ulnar, complete or partial (with or without distal deficiencies, incomplete)	755.27	Q71.5	
Longitudinal deficiency Carpals or Metacarpals, complete or partial (with or without incomplete phalangeal deficiencies)	755.28	Q71.8	
Longitudinal deficiency, Phalanges Complete or Partial	755.29	Q71.8	
Unspecified reduction deformity of Lower Limbs	755.30	Q72	
Transverse deficiency of Lower Limbs	755.31	Q72.0 – Q72.3	
Longitudinal deficiency of Lower Limb	755.32	Q72.4 – Q72.6	
Longitudinal deficiency, combined, involving Femur, Tibia, and Fibula (complete or incomplete)	755.33	Q72.8	
Longitudinal deficiency Femoral, complete or partial (with or without distal deficiencies, incomplete)	755.34	Q72.4	
Longitudinal deficiency, Tibiofubular complete or partial (with or without distal deficiencies, incomplete)	755.35	*Must select specific bone(s)* Q72.5, Q72.6	

Birth Defect	ICD-9-CM	ICD-10-CM	Procedure Code (used with ICD9)
Longitudinal deficiency Tibia, complete or partial(with or without distal deficiencies, incomplete)	755.36	Q72.5	
Longitudinal deficiency, Fibular complete or partial, (with or without distal deficiencies, incomplete)	755.37	Q72.6	
Longitudinal deficiency, Tarsals or Metatarsals,complete or Partial (with or without incomplete phalangeal deficiency)	755.38	Q72.8	
Longitudinal deficiency, Phalanges, complete or partial	755.39	Q72.8	
Reduction deformities unspecified limb *Amelia (absence)* *Ectromelia* *Hemimelia* *Phocomelia* *Other*	755.4 755.4 755.4 755.4 755.4	**Q73.0** Q73.0 Q73.8 Q73.8 Q73.1 Q73.8	
8. Hypospadias and Epispadias	***752.6***	***Q54, Q64***	
Hypospadias	752.61	*Report specific degree/location when available* Q54.0 -Q54.3 *or* *Other* Q54.8 *or* *Unspecified* Q54.9	58.45
Epispadias	752.62	Q64.0	58.45
9. Down Syndrome	***758.0***	**Q90.9** *for NOS* *Use specific condition third digits when possible(Q90.0 – Q90.2)*	
10. Cerebral Palsy	***343***	**G80.9** *for NOS* *Use specific condition third digits when possible(G80.0 – G80.8)*	
11. Fetal Alcohol Syndrome/Alcohol Related Birth Defects*	***760.71***	**Q86.0, P04.3**	
Fetal Alcohol Syndrome		Q86.0	
Fetus/ Newborn affected by maternal alcohol		P04.3	

Birth Defect	ICD-9-CM	ICD-10-CM	Procedure Code (used with ICD9)
12. Autism Spectrum Disorder**	***299.0***	**F84**	
Autism	299.0	F84.0 *for Childhood* <u>*or*</u> F84.1 *for Atypical*	
Asperger's		F84.5	
Pervasive Developmental Disorder		*Other* F84.8 <u>*or*</u> *Unspecified* F84.9	

A. Description

1. General

Birth defects are inborn syndromes, diseases, disorders and malformations that occur before birth. They can affect the organs, senses, limbs, physical and mental development. They also cause pregnancy loss through miscarriage and stillbirth. Some conditions are recognized at birth, others become apparent later in life.

2. Cause(s)

The cause(s) of about 70% of all birth defects are unknown. Environmental pollutants may cause birth defects, developmental disabilities, or other adverse reproductive outcomes. Similarly, occupational hazards, dietary factors, medications, infections, and personal behaviors may cause or contribute to birth defects. The causes of birth defects may be a defect in any part of the genome, an interaction between genes or between genes and the environment.

3. World Wide Occurrence

Congenital abnormalities may occur any where in the world. It is possible to see random or systematic geographical variation in the occurrence of some forms of birth defects. This could be due to certain genetic factor(s) or due to a specific exposure(s).

4. High risk factors

Factors that can cause an adverse outcome of pregnancy exist in the environment, where some are known and most of them are not. In addition to what genetic factors may contribute, exposure before or during pregnancy to factors such as smoking, alcohol, medications, illicit drugs, or dietary factors may predispose a pregnant woman to have a child with one or more abnormalities.

B. Methods of Control

1. Prevention Activities

Education of health professionals and the public regarding factors that may be related to birth defects is crucial for prevention. The use of folic acid, a B-vitamin (B9) found in fortified foods and vitamin pills has been shown to be effective in reducing the incidence of neural tube defects. The U.S. Public Health Service recommends that all women who could possibly become pregnant get 400 micrograms (or 0.4mg) of folic acid every day. This could prevent up to 70% of some types of serious birth defects. But to do this, women need folic acid before they get pregnant. Since over 50% of births in Washington are unintended, it is important to promote daily consumption of folic acid to all women of childbearing age. In addition, early identification of birth defects promotes care coordination and secondary prevention activities to improve the quality of life.

2. Occurrence of Clusters

The occurrence of a new or an already known birth defect in a cluster is an indicator of exposure to a known or an unknown factor(s) or may be by chance. Suspected clusters (3 or more cases of the same or developmentally similar conditions) may be reported to Jeanette Robbins, at (360) 236-3591 or to Juliet VanEenwyk, the state epidemiologist for non-communicable conditions at (360) 236-4250.

3. International Measures

Once a teratogen or a risk factor proves to be causal, that knowledge should be communicated internationally and appropriate measure should be taken to avoid exposures. Efforts to prevent exposures of pregnant women to Thalidomide, Aminopterin, Isotretinoin all over the world are examples of this kind of international effort.

4. Control Measures

Environmental protection, surveillance and research

5. Public Health Importance

Birth defects are the leading cause of infant mortality in the United States, accounting for more than 20% of all infant deaths. Of about 120,000 U.S. babies born each year with a birth defect, 8,000 die during their first year of life. Birth defects are the 5th leading cause of years of potential life lost and contribute substantially to childhood morbidity and long-term disability. The health care cost associated with birth defects is also immense.