Chapter 3

Case Definition
Table of Contents

3.1 Introduction.................................................................................................................3-1
3.2 What Is Meant by a ‘Birth Defect’ ..........................................................3-2
3.3 Terminology ..................................................................................................................3-3
3.4 Case Definition Criteria..........................................................................................3-6
   3.4.1 Diagnoses to Be Included ...........................................................................3-6
   3.4.2 Residence .............................................................................................................3-6
   3.4.3 Pregnancy Outcome .........................................................................................3-7
   3.4.4 Gestational Age ..................................................................................................3-8
   3.4.5 Age at Which Defects Are Diagnosed ..........................................................3-9
   3.4.6 Pregnancies Resulting from Assisted Reproductive Technology ............3-10
3.5 Case Definition and Sensitivity and Specificity ..................................................3-11
3.6 References .....................................................................................................................3-12

Appendices

Appendix 3.1 and 3.2  As of March 2015, these two appendices have been updated and combined into one file (Appendix 3.1 - Birth Defects Descriptions for NBDPN Core, Recommended, and Extended Conditions)..............................................................................................................A3.1-1
Appendix 3.3  Examples of Conditions Considered to be Minor Anomalies...............A3.3-1
Appendix 3.4  Conditions Related to Prematurity in Infants Born at Less than 36 Weeks Gestation...A3.4-1
Appendix 3.5 Case Inclusion Guidance for Potentially Zika-related Birth Defects.................A3.5-1
3.1 Introduction

A case definition is a set of criteria that define the parameters of what is included for quantitative description and analysis. In birth defects surveillance a case refers to an individual with characteristics that fit into the defined parameters. Important characteristics in birth defects surveillance include the diagnosis, pregnancy outcome information, and demographics.

In the absence of a single national birth defects surveillance program in the United States, pooled data from state-based programs across the country serve to estimate national rates, indicate regional variations, and describe the epidemiology of defects that occur rarely. Because, at any given time, these programs may be in different stages of development, employ different methods of ascertainment, and have different goals and objectives, the elements of the case definition used by each must be clearly identified in order to make valid comparisons and to minimize birth defects rate variations across surveillance programs and among individual defects ascertained by the same program.

Therefore, it is necessary for a surveillance program to develop a clear, concise case definition. Consistent application of a standard definition facilitates the accurate monitoring of clinically relevant conditions, identification of true changes over time, and comparison among populations in order to meet surveillance goals.

In the remainder of this chapter we discuss what is meant by the term ‘birth defect’ (Section 3.2), some important terminology for case definition (Section 3.3), and case definition criteria (Section 3.4). The relationship between case definition and the two terms ‘sensitivity’ and ‘specificity’ is discussed in Section 3.5. References cited in this chapter may be found in Section 3.6. Appendices to this chapter include birth defects included in the NBDPN’s case definition (Appendix 3.1 and 3.2 – As of 3/2015, these two appendices have been merged into one), examples of minor anomalies (Appendix 3.3), and conditions related to prematurity (Appendix 3.4).

As of 11/16/16, the network has adopted the list of potentially Zika-related birth defects\(^1\), including changes to this list as determined by CDC as more information becomes available, as a special category of NBDPN birth defects.

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\(^1\) Honein, et al. 2016
3.2 What Is Meant by a ‘Birth Defect’

The general term ‘birth defect’ may take on a variety of meanings depending on the context in which it is used and the perspective of the person using it. ‘Congenital abnormality’, ‘congenital anomaly’, and ‘congenital malformation’ are terms often used as synonyms for ‘birth defect’. However, the word ‘congenital’ may describe any condition present at birth, regardless of its etiology or timing of occurrence. In the broadest sense, the term birth defect encompasses a diversity of conditions including physical malformations, sensory deficits, chromosomal abnormalities, metabolic defects, neurodevelopmental disorders, and complications related to prematurity and low birth weight, among others.

While such a broad definition may be very helpful when seeking legislation and funding for screening, intervention, or prevention programs, a more specific definition is needed for surveillance purposes. Traditionally, birth defects surveillance programs have monitored major structural and genetic defects that adversely affect health and development (Correa-Villaseñor et al., 2003). The specific conditions monitored by an individual program will vary depending on the goals and objectives of that program, the case ascertainment methods used, and the resources available.
3.3 Terminology

**General Terminology**

<table>
<thead>
<tr>
<th>Term</th>
<th>Definition</th>
</tr>
</thead>
<tbody>
<tr>
<td>Major anomaly</td>
<td>A congenital abnormality that requires medical or surgical treatment, has a serious adverse effect on health and development, or has significant cosmetic impact. Individual major anomalies occur in less than 1 percent of the population. Together, they are seen in approximately 3 percent of births. Examples include cleft lip and tracheo-esophageal fistula.</td>
</tr>
<tr>
<td>Minor anomaly</td>
<td>A congenital abnormality that does not require medical or surgical treatment, does not seriously affect health and development, and does not have significant cosmetic impact. Individual minor anomalies generally occur in less than 4 percent of the population. The presence of multiple minor anomalies in the same child may provide clues to the timing of a prenatal insult and may indicate the presence of an undiagnosed major anomaly, syndrome, or functional deficit. Examples of minor anomalies are listed in Appendix 3.3.</td>
</tr>
<tr>
<td>Normal variant</td>
<td>A minor anomaly that occurs in approximately 4 percent or more of the population. Examples of normal variants include webbing of the second and third toes and a single umbilical artery in an otherwise normal infant.</td>
</tr>
</tbody>
</table>

**Terminology Related to the Formation of Major Anomalies**

<table>
<thead>
<tr>
<th>Term</th>
<th>Definition</th>
</tr>
</thead>
<tbody>
<tr>
<td>Malformation</td>
<td>A major anomaly that arises during the initial formation of a structure, i.e., during organogenesis. For most organs, this occurs during the first eight weeks after fertilization. The resulting structure may be abnormally formed, incompletely formed, or may fail to form altogether. Examples of malformations include spina bifida and hypoplastic left heart. The term ‘congenital malformation’ is also used more broadly to indicate any major anomaly.</td>
</tr>
<tr>
<td>Disruption</td>
<td>A major anomaly that results from alteration of a structure after its initial formation. The resulting structure may have an altered shape and configuration, abnormal division or fusion of its component parts, or loss of parts that were previously present. Examples of disruption defects include intestinal atresia and possibly gastroschisis.</td>
</tr>
<tr>
<td>Deformation</td>
<td>A major anomaly that results from molding of part of a structure, usually over a prolonged time, by mechanical forces after its initial formation. Examples of forces that may lead to a deformation include oligohydramnios (diminished amniotic fluid) and intrauterine crowding in twin, triplet, or higher order pregnancies. Examples of deformations include the compression (Potter’s) facies seen with bilateral renal agenesis and some instances of clubfoot.</td>
</tr>
</tbody>
</table>

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### Terminology Related to Patterns of Multiple Anomalies Occurring in a Single Child

<table>
<thead>
<tr>
<th>Syndrome</th>
<th>A pattern of anomalies that form a specific diagnosis for which the natural history and recurrence risk are usually known. Use of the term ‘syndrome’ implies that the anomalies have a common specific etiology. Examples include Beckwith-Weidemann syndrome and Rubinstein-Taybi syndrome.</th>
</tr>
</thead>
<tbody>
<tr>
<td>Sequence</td>
<td>A pattern of anomalies that results from a single primary anomaly or mechanical factor. The presence of the initial anomaly or factor leads to one or more secondary anomalies, which may then lead to one or more tertiary anomalies, etc., in cascade fashion. Examples include Robin sequence (micrognathia, posterior displacement of the tongue, cleft soft palate) and the oligohydramnios, or Potter, sequence (pulmonary hypoplasia, flattened facies, abnormal positioning of the limbs).</td>
</tr>
<tr>
<td>Association</td>
<td>A nonrandom pattern of anomalies that occur together more frequently than expected by chance alone, but for which no etiology has been demonstrated. Examples include VACTERL association (Vertebral, Anal, Cardiac, Tracheo-Esophageal, Renal, and Limb anomalies) and CHARGE association (Colobomas, Heart defects, choanal Atresia, Retarded growth and development and/or central nervous system anomalies, Genital anomalies and/or hypogonadism, Ear anomalies and/or deafness). Use of the term ‘association’ does not indicate that a specific diagnosis has been made.</td>
</tr>
</tbody>
</table>

### Terminology Related to Tissue and Organ Formation

| Agenesis | Failure of an organ to form. |
| Dysgenesis | Anomalous or disorganized formation of an organ. |
| Aplasia | Absence of a tissue or organ due to lack of cell proliferation. |
| Dysplasia | Disorganized cell structure or arrangement within a tissue or organ. |
| Hypoplasia | Undergrowth of a tissue or organ due to insufficient proliferation of otherwise normal cells. |
| Hyperplasia | Overgrowth of a tissue or organ due to excess proliferation of otherwise normal cells. |

### Terminology Related to the Timing of Gestation and Delivery

| Embryonic period | The first eight weeks after fertilization, during which most, but not all, organs are formed. |
| Fetal period | The period from the ninth week after fertilization through delivery. |
| Neonatal (newborn) period | The first 28 days following delivery of a live-born infant. |
**Terminology Related to the Timing of Gestation and Delivery (continued)**

<table>
<thead>
<tr>
<th>Term</th>
<th>Definition</th>
</tr>
</thead>
<tbody>
<tr>
<td>Prenatal</td>
<td>Before delivery.</td>
</tr>
<tr>
<td>Perinatal</td>
<td>Before, during, or after delivery. The exact time period may vary from 20 to 28 completed weeks of gestation through 7 to 28 days after delivery, depending on the context in which the term is used.</td>
</tr>
<tr>
<td>Postnatal</td>
<td>After delivery.</td>
</tr>
</tbody>
</table>

**Terminology Related to Pregnancy Outcome**

<table>
<thead>
<tr>
<th>Term</th>
<th>Definition</th>
</tr>
</thead>
<tbody>
<tr>
<td>Live birth</td>
<td>Spontaneous delivery of an infant that exhibits signs of life, including a heartbeat, spontaneous breathing, or movement of voluntary muscles. Transient cardiac contractions and fleeting respiratory efforts or gasps are not necessarily considered signs of life by all programs.</td>
</tr>
<tr>
<td>Fetal death (stillbirth)</td>
<td>Spontaneous delivery of an infant or fetus at 20 weeks or greater gestation that does not exhibit signs of life. Transient cardiac contractions and fleeting respiratory efforts or gasps are not necessarily considered signs of life by all programs. A late fetal death is a fetal death that occurs at 28 weeks or greater gestation.</td>
</tr>
<tr>
<td>Spontaneous abortion (miscarriage)</td>
<td>Spontaneous delivery of a fetus at less than 20 weeks gestation.</td>
</tr>
<tr>
<td>Induced abortion (elective termination)</td>
<td>The purposeful interruption of pregnancy with the intention other than to produce a live birth and which does not result in a live birth.</td>
</tr>
<tr>
<td>Term infant</td>
<td>An infant born after 37 completed weeks and before 42 completed weeks of gestation.</td>
</tr>
<tr>
<td>Preterm infant</td>
<td>An infant born before 37 completed weeks of gestation.</td>
</tr>
<tr>
<td>Postterm infant</td>
<td>An infant born after 42 completed weeks of gestation.</td>
</tr>
<tr>
<td>Low birth weight</td>
<td>Birth weight less than 2,500 grams, regardless of gestational age.</td>
</tr>
<tr>
<td>Very low birth weight</td>
<td>Birth weight less than 1,500 grams, regardless of gestational age.</td>
</tr>
<tr>
<td>Extremely low birth weight</td>
<td>Birth weight less than 1,000 grams, regardless of gestational age.</td>
</tr>
<tr>
<td>Neonatal death</td>
<td>Death of a live-born infant within the first 28 days after birth. <strong>Early neonatal death</strong> refers to death during the first 7 days. <strong>Late neonatal death</strong> refers to death after 7 days but before 29 days.</td>
</tr>
<tr>
<td>Infant death</td>
<td>Death of a live-born infant before 12 months of age.</td>
</tr>
</tbody>
</table>
3.4 Case Definition Criteria

In this section we discuss the various components of the case definition, that is, the criteria a birth defects surveillance program uses to define a case. These include diagnoses to be included (Section 3.4.1), residence (Section 3.4.2), pregnancy outcome (Section 3.4.3), gestational age (Section 3.4.4), age at which defects are diagnosed (Section 3.4.5), as well as the issue of pregnancies resulting from assisted reproductive technology (Section 3.4.6). Each of these criteria is discussed further below.

3.4.1 Diagnoses to Be Included

For the purposes of generating and reporting birth defects surveillance data across multiple states, the National Birth Defects Prevention Network (NBDPN) recommends the 47 major anomalies listed in Appendix 3.1. These were chosen on the basis of their frequency, their impact on public health, the state of knowledge about their etiologies and risk factors, and other considerations. Individual surveillance programs may want to expand this list to include additional defects of interest. Programs with limited resources may need to ascertain a subset of this list. Descriptions of each of the 47 diagnoses, its ICD-9-CM and CDC/BPA codes, inclusion and exclusion criteria, and defect-specific information that may be helpful when abstracting medical records are provided in the NBDPN birth defects descriptions (see Appendix 3.1). Examples of conditions considered to be minor anomalies are provided in Appendix 3.3. Conditions related to prematurity that are not considered to be major anomalies are listed in Appendix 3.4.

As of 11/16/16, the network has adopted the list of potentially Zika-related birth defects, including any changes to this list as determined by CDC as more information becomes available, as a special category of NBDPN birth defects. The addition of this list of potentially Zika-related birth defects does not affect the list of core, recommended and extended birth defects that a program must ascertain in order to meet specific NBDPN Data Quality standard levels.

3.4.2 Residence

When monitoring the frequency of any condition, it is critical to define the population in which the cases occur. This allows one to calculate rates within the population, evaluate changes in these rates over time, plan for prevention and intervention services, and assess program goals and effectiveness. Population-based birth defects surveillance programs should strive to ascertain defects that occur among the offspring of all women who reside within a defined geographic area at the time of pregnancy outcome.

While this charge for surveillance programs appears straightforward, there are some special considerations. One such consideration is the fact that women who reside in one state or community may travel outside that area – such as to an adjacent state, specialty care center, or military facility – for obstetric care. In these instances, the mother’s place of residence at the time of delivery (rather than the actual location of the delivery) should be used to determine whether to include her pregnancy in the surveillance. Including in-area residents who deliver outside the surveillance area, and excluding out-of-area residents who deliver within the surveillance area, is essential in order to conduct comprehensive surveillance. Whether an individual program attains this level of comprehensiveness will depend on how frequently women travel outside the surveillance area for delivery, the magnitude of the potential impact this may have on defect rates, the staff and resources available, and, most importantly, the existence of data-sharing relationships with facilities and programs outside the surveillance area. Recent changes in regulations concerning the privacy of medical records under the Health Information Portability and Accountability Act (HIPAA) have added to the complexity of these data-sharing relationships. The HIPAA regulations are discussed further in Chapter 2 on Legislation.
Another consideration is the fact that a surveillance program may identify more than one residential address for an individual woman. For example, the address of the health insurance policyholder listed in a hospital delivery record may differ from the mother’s address listed on a birth certificate. If a patient changes residence during pregnancy, programs that employ multisource ascertainment may identify one address from prenatal or laboratory records and another from the hospital delivery record. For these reasons, surveillance programs should develop standard procedures for deciding which of multiple addresses to accept as the mother’s residence at delivery. Usually, this is the address at the time of delivery as listed on the vital record. If a vital record is not available or is not generated, as when an elective termination is performed outside the hospital setting, considering the mother’s address from the termination record or from the prenatal visit closest in time to the delivery may be appropriate alternatives.

A third consideration requires detailing the method of determining whether a particular address lies within the surveillance area, particularly if the population under surveillance is not that of an entire state. For example, zip codes often cross city or county boundaries, streets may be renamed, new zip codes may be added, and city or county boundaries may change over time. Addresses that contain only a post office box number do not provide information about a person’s actual place of residence. For these reasons, surveillance programs should develop standard procedures for distinguishing addresses that lie within or outside their surveillance area. Potential reference sources include street maps, United States Postal Service listings, tax assessor records, census tracts, and the latitude and longitude of the surveillance area (geocoding). While the latter can be extremely precise, the accuracy of geocoding will depend upon the accuracy of the addresses to which the latitude and longitude are assigned.

### 3.4.3 Pregnancy Outcome

Ideally, for births defects surveillance to be comprehensive with high sensitivity, all defects occurring in a population should be ascertained regardless of whether a pregnancy ends in live birth, fetal death, or spontaneous abortion, or whether an elective termination is performed. It is estimated that approximately 10 to 15 percent of all recognized pregnancies end in spontaneous abortion, and approximately 6 to 7 percent of those that reach 20 weeks gestation end in fetal death (Gabbe et al., 1996; National Center for Health Statistics, 2001). Surveillance systems that ascertain defects only among live-born infants may report incomplete data for defects that occur frequently among these outcomes. However, it is important to recognize that even late fetal deaths may not be scrutinized for defects as closely or as systematically as are live births. Unless an autopsy (including internal examination) and chromosome analysis are performed routinely, defects present in fetal deaths, yet not immediately evident in the delivery room, may remain unidentified. Even if an autopsy and chromosome analysis are performed, the presence of minor defects may not be recognized and syndromes may not be diagnosed. Whether it is beneficial for an individual program to ascertain defects reported in outcomes other than live births will depend upon the program’s goals and objectives, the staff and resources available, the accessibility of information about these outcomes, and the magnitude of the potential impact on individual defect rates of excluding them.

The development and widespread use of prenatal diagnostic technology has posed additional issues for birth defects surveillance. These procedures have provided women with the option of electively terminating affected pregnancies, particularly those with defects that are life-threatening or that are likely to result in significant mental or functional impairment, usually before 20 weeks gestation. In the absence of prenatal diagnosis, many of these pregnancies would end in live birth or fetal death and would be included in birth defects surveillance data from many programs. Failure to ascertain prenatal diagnoses among electively terminated pregnancies may, therefore, limit the comprehensiveness and sensitivity of surveillance programs for some defects, such as neural tube defects and chromosomal abnormalities, even when defects among fetal deaths are ascertained. And – because the availability and utilization of prenatal diagnosis and elective termination may vary among populations, across geographic regions, and over time
Chapter 3

Calculating Gestational Age

The ability to make valid comparisons of some defect rates may be compromised unless pregnancies electively terminated after prenatal diagnosis are regularly ascertained.

Unfortunately, including these prenatal diagnoses will likely require expansion of a program’s case ascertainment sources to include settings such as prenatal diagnostic clinics and termination centers. Furthermore, as is the case with fetal deaths, pregnancies that are electively terminated may not be fully scrutinized for confirmation of the prenatal diagnosis or the presence of additional defects or syndromes upon completion of the procedure. Again, whether it is beneficial for an individual program to ascertain defects reported in these pregnancies will depend upon the program’s goals and objectives, the staff and resources available, the accessibility of information about these outcomes, and the magnitude of the potential impact on individual defect rates of excluding them. Regardless, it is important for birth defects surveillance programs to clearly state which outcomes are included when reporting surveillance data and to include pregnancies electively terminated after prenatal diagnosis whenever possible.

3.4.4 Gestational Age

Another important component of the case definition is the gestational age at delivery of the cases included in the surveillance data. The frequency of some defects may vary by gestational age, leading to variations in their rates depending on the length of gestation. For example, some defects are identified more frequently among preterm infants (Rasmussen et al., 2001; Shaw et al., 2001). Others, such as patent ductus arteriosus and undescended testes, may be abnormal in term infants but physiologically normal in preterm infants. Some ventricular septal defects that are present at birth in preterm infants might have closed during the last weeks of gestation if the pregnancy had continued to term. If surveillance systems differ in the gestational age at delivery of cases they include, or in their use of exclusion criteria based on gestational age, their rates of some defects may not be comparable.

Again, the inclusion of pregnancies electively terminated after prenatal diagnosis poses additional issues. Many of these pregnancies would have delivered spontaneously at a considerably later gestational age had they not been terminated. In order not to underestimate the frequency of defects for which elective termination may be performed, pregnancies terminated after prenatal diagnosis should be included in surveillance data regardless of the gestational age at which they were terminated. However, this may slightly overestimate the frequency of some defects relative to their frequency in the absence of prenatal diagnosis. For example, the majority of pregnancies electively terminated before 20 weeks gestation would have otherwise continued beyond 20 weeks to be included in birth defects surveillance programs that monitor pregnancies of 20 weeks or greater. However, a small proportion might have ended in spontaneous abortion before 20 weeks and would not appropriately be included in data from these programs. While the frequency of spontaneous abortion for pregnancies with Down syndrome has been estimated for each week of gestation, the natural history of pregnancies with other defects has not been well described (Hook et al., 1995). The frequency of spontaneous abortion by gestational week probably varies depending on the defect. Unfortunately, this effect is likely to be greater the earlier in gestation that affected pregnancies are terminated.

For the purposes of generating and reporting birth defects surveillance data across multiple states, the NBDPN recommends monitoring defects among live births and fetal deaths of 20 weeks or greater and among pregnancies electively terminated after prenatal diagnosis at any gestational age. Gestational age may be derived in various ways based on the date of the last menstrual period, measurement of the fetus by prenatal ultrasound, or the newborn clinical exam. Because these methods may not be equally accurate and may yield conflicting results, an important consideration is which method to use to determine whether a case fulfills the gestational age criterion for inclusion in surveillance data (Alexander et al., 1990; Hall, 1990). The methods below are listed in descending order of their generally accepted accuracy for calculating gestational age:
- Prenatal ultrasound with a reported gestational age of less than 14 weeks
- Date of the last menstrual period
- Prenatal ultrasound with a reported gestational age of 14 weeks or greater
- Clinical examination after delivery

When multiple estimates of gestational age are ascertained for an individual case, the NBDPN recommends that the value derived using the method highest on this list be used to determine case status. Regardless of which method is used, it is important for birth defects surveillance programs to clearly state the gestational ages of the cases included when reporting surveillance data.

### 3.4.5 Age at Which Defects Are Diagnosed

The age at which a defect is diagnosed is also an important component of the case definition. The frequency of some defects may vary depending on the age of the child at diagnosis. While defects that are visible in the delivery room or symptomatic shortly after birth may be ascertained by most surveillance systems with high sensitivity, some internal defects may not be apparent for weeks or months after birth. Examples include cardiac defects that do not produce overt cyanosis, such as many atrial or ventricular septal defects, many obstructive renal defects, and some instances of intestinal malrotation. In addition, some chromosomal abnormalities may not be diagnosed until a year or more after birth when developmental delay or behavioral symptoms prompt a more in-depth evaluation. The rates of such conditions reported by surveillance systems that ascertain defects only among infants in the newborn nursery may not be comparable with those from systems that ascertain defects among older infants and children.

For the purposes of generating and reporting birth defects surveillance data across multiple states, the NBDPN recommends monitoring defects among live-born infants up to one year of age. Whether an individual program is able to ascertain defects beyond the newborn period will depend on the accessibility of information from sources other than the newborn nursery and the availability of staff and resources to add these additional sources. Programs should regularly state the range of ages at diagnosis included when reporting surveillance data.

As with other case definition criteria, the inclusion of defects that are diagnosed prenatally poses additional issues. The sensitivity and specificity of fetal ultrasound may vary for different defects depending on the gestational age, the skill and experience of the technician, the presence of maternal obesity, and other factors. The sensitivity and specificity of fetal ultrasound also may differ from that of newborn ultrasound and other postnatal diagnostic procedures. In addition, some conditions identified at mid-gestation by prenatal ultrasound may resolve spontaneously before delivery. Examples include renal obstructions, such as pyelectasis and uretero-pelvic junction obstructions, choroid plexus cysts of the brain, and some ventricular septal defects. Even chorionic villus sampling (CVS) may yield placental cells that contain chromosomal mosaicism not actually present in the fetus. For these reasons, many abnormalities diagnosed or suspected prenatally must be evaluated postnatally to determine their true nature. When such postnatal assessment is not possible or the medical records are not available, decisions about whether to include these defects in the surveillance must be made individually based on the certainty and specificity of the prenatal diagnosis for each case. General abstractor’s instructions for the inclusion and exclusion of prenatal diagnoses for the 47 defects reported by the NBDPN are provided in Appendix 3.1. When reporting surveillance data, it is important for birth defects surveillance programs to state clearly the ages at which the defects were diagnosed and whether prenatal diagnoses without postnatal confirmation are included.
3.4.6 Pregnancies Resulting from Assisted Reproductive Technology

The use of assisted reproductive technology raises unique issues for birth defects surveillance, particularly in pregnancies where the egg from one woman (the biological mother) is used to conceive, but the pregnancy is carried by another woman (the birth mother). In this instance, genetic characteristics of the biological mother will be transmitted to the infant, but the birth mother’s environment and lifestyle during pregnancy may also affect the infant. This situation may become quite complex when examining etiologic factors for birth defects. However, for surveillance purposes, the NBDPN recommends that the person listed on the child’s birth certificate should be mother of record.
3.5 Case Definition and Sensitivity and Specificity

Use of a consistent case definition is critical when evaluating the sensitivity and specificity of surveillance data and the efficiency and utility of surveillance programs.

The sensitivity of a surveillance program is defined as the proportion of cases occurring within a population that the program ascertains. Factors that may affect the sensitivity of a birth defects surveillance program include which pregnancy outcomes are ascertained (live births, fetal deaths, elective terminations), the gestational age at which they are ascertained (term infants only, pregnancies > 20 weeks, all pregnancies), the child’s age at the time the defect is diagnosed (prenatally, in the newborn period, before one year, at any age), and the diagnostic setting and methods used for ascertainment. For example, defects that are symptomatic in a live born infant may not be recognized in pregnancies that end in fetal death unless an autopsy is performed. Defects that are not immediately life-threatening, such as many cardiac septal defects, may not be diagnosed until several weeks or months after birth. If managed solely in the outpatient setting, these defects may be missed entirely by hospital-based programs unless surgical correction is required.

The specificity of a surveillance program is defined as the proportion of cases within a population that are ascertained by the program and that truly have defects. Factors that affect the sensitivity of a birth defects surveillance program may also affect its specificity. For example, patent ductus arteriosus (PDA) may be entirely normal in a preterm or one-day-old term infant but distinctly abnormal in a three-month-old child. Inclusion of all occurrences of PDA, regardless of gestational or postnatal age, may lead to ascertainment of false positive cases. Variations in the quality of prenatal ultrasound and in the natural course of some prenatal conditions necessitate postnatal confirmation of many diagnoses to avoid including false positive or clinically nonsignificant cases. Such confirmation may not be possible for pregnancies that end in fetal death or elective termination unless fetal autopsies are performed. Similarly, the exact nature of a congenital heart defect may not be finalized until the time of corrective surgery.
3.6 References


Appendix 3.1

Birth Defects Descriptions
for NBDPN Core, Recommended, and Extended Conditions

Updated March 2015

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Table of Contents

List of Birth Defects ......................................................................................................................... i
Detailed Descriptions of Birth Defects ............................................................................................. 1
  Format for Birth Defect Descriptions ............................................................................................. 1
Central Nervous System .................................................................................................................... 2
  Anencephaly .................................................................................................................................... 2
  Encephalocele ................................................................................................................................ 3
  Holoprosencephaly .......................................................................................................................... 4
  Spina Bifida without Anencephaly .................................................................................................. 5
Eye .................................................................................................................................................... 7
  Anophthalmia/Microphthalmia ......................................................................................................... 7
  Congenital Cataract ......................................................................................................................... 8
Ear ..................................................................................................................................................... 9
  Anotia/Microtia ................................................................................................................................. 9
Cardiovascular .................................................................................................................................. 11
  Aortic Valve Stenosis ....................................................................................................................... 11
  Atrial Septal Defect (ASD) ............................................................................................................. 12
  Atrioventricular Septal Defect ....................................................................................................... 13
  (Atrioventricular Canal Defect; Endocardial Cushion Defect) ....................................................... 13
  Coarctation of the Aorta .................................................................................................................. 15
  Common Truncus (Truncus Arteriosus or TA) ............................................................................... 16
  Double Outlet Right Ventricle (DORV) ........................................................................................... 17
  Ebstein Anomaly ............................................................................................................................. 18
  Hypoplastic Left Heart Syndrome (HLHS) .................................................................................... 19
  Interrupted Aortic Arch (IAA) .......................................................................................................... 20
  Pulmonary Valve Atresia and Stenosis ............................................................................................ 21
  Single Ventricle .............................................................................................................................. 23
  Tetralogy of Fallot (TOF) ................................................................................................................ 24
  Total Anomalous Pulmonary Venous Connection (TAPVC) ........................................................... 26
  Transposition of the Great Arteries (TGA) ..................................................................................... 27
  Tricuspid Valve Atresia and Stenosis ............................................................................................... 29
  Ventricular Septal Defect (VSD) .................................................................................................... 30
Orofacial ............................................................................................................................................. 31
  Choanal Atresia ............................................................................................................................... 31
  Cleft Lip Alone (without Cleft Palate) ............................................................................................ 32
  Cleft Lip with Cleft Palate ................................................................................................................ 33
  Cleft Palate Alone (without Cleft Lip) ............................................................................................ 34
Gastrointestinal ................................................................................................................................. 35
  Biliary Atresia ................................................................................................................................. 35
  Esophageal Atresia/Tracheoesophageal Fistula .......................................................................... 36
  Rectal and Large Intestinal Atresia/Stenosis .................................................................................. 37
  Small Intestinal Atresia/Stenosis .................................................................................................... 38
Genitourinary ..................................................................................................................................... 39
  Bladder Exstrophy .......................................................................................................................... 39
  Cloacal Exstrophy ........................................................................................................................... 41
  Congenital Posterior Urethral Valves .............................................................................................. 42
  Hypospadias ................................................................................................................................... 43
Appendix 3.1

Case Definition

Renal Agenesis/Hypoplasia ................................................................................................................................................. 45

**Musculoskeletal** ................................................................................................................................................................. 47
  Clubfoot ................................................................................................................................................................................. 47
  Craniosynostosis .................................................................................................................................................................... 48
  Diaphragmatic Hernia ............................................................................................................................................................... 49
  Gastrochisis ............................................................................................................................................................................. 50
  Limb Deficiencies (Reduction Defects) ................................................................................................................................. 52
  Omphalocele ........................................................................................................................................................................... 55

**Chromosomal** ........................................................................................................................................................................ 57
  Deletion 22q11.2 ....................................................................................................................................................................... 57
  Trisomy 13 ................................................................................................................................................................................ 59
  Trisomy 18 ................................................................................................................................................................................ 61
  Trisomy 21 (Down Syndrome) ................................................................................................................................................. 63
  Turner Syndrome ....................................................................................................................................................................... 65
Note: As of January 2014, the following conditions were dropped from the NBDPN list:

- Amniotic bands
- Aniridia
- Congenital hip dislocation
- Epispadias
- Fetus or newborn affected by maternal alcohol use
- Hirschsprung disease (congenital megacolon)
- Hydrocephalus without spina bifida
- Microcephalus
- Patent ductus arteriosus
- Pyloric stenosis

The following conditions were added:

- Clubfoot
- cloacal extrophy
- Congenital posterior urethral valves
- Craniosynostosis
- Deletion 22q11.2
- Double outlet right ventricle (DORV)
- Holoprosencephaly
- Interrupted aortic arch (IAA)
- Single ventricle
- Small intestine atresia/stenosis
- Turner syndrome

The following conditions were merged:

- Reduction deformity, lower limbs; reduction deformity, upper limbs. Merged to limb deficiencies (reduction defects).

The following conditions were separated:

- Cleft lip with and without cleft palate separated to cleft lip with cleft palate; cleft lip alone (without cleft palate).
## List of Birth Defects

<table>
<thead>
<tr>
<th>Birth Defects</th>
<th>ICD-9-CM Codes</th>
<th>CDC/BPA Codes</th>
<th>ICD-10-CM Codes</th>
<th>Standard Level*</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>CNS</strong></td>
<td></td>
<td></td>
<td></td>
<td>Level 1, Level 2, Level 3</td>
</tr>
<tr>
<td>Anencephalus</td>
<td>740.0 – 740.1</td>
<td>740.00 – 740.10</td>
<td>Q00.0 - Q00.1</td>
<td>Level 1</td>
</tr>
<tr>
<td>Spina bifida without anencephalus</td>
<td>741.0, 741.9 w/o 740.0 - 740.10</td>
<td>741.00 – 741.99 w/o 740.0 – 740.10</td>
<td>Q05.0 - Q05.9, Q07.01, Q07.03 w/o Q00.0 - Q00.1</td>
<td>Level 1</td>
</tr>
<tr>
<td>Encephalocele</td>
<td>742.0</td>
<td>742.00 – 742.09</td>
<td>Q01.0 – Q01.9</td>
<td>Level 2</td>
</tr>
<tr>
<td>Holoprosencephaly</td>
<td>742.2</td>
<td>742.26</td>
<td>Q04.2</td>
<td>Level 3</td>
</tr>
<tr>
<td><strong>Eye</strong></td>
<td></td>
<td></td>
<td></td>
<td>Level 2, Level 3</td>
</tr>
<tr>
<td>Anophthalmia / microphthalmia</td>
<td>743.0, 743.1</td>
<td>743.00 – 743.10</td>
<td>Q11.0 – Q11.2</td>
<td>Level 2</td>
</tr>
<tr>
<td>Congenital cataract</td>
<td>743.30 – 743.34</td>
<td>743.32</td>
<td>Q12.0</td>
<td>Level 3</td>
</tr>
<tr>
<td><strong>Ear</strong></td>
<td></td>
<td></td>
<td></td>
<td>Level 2</td>
</tr>
<tr>
<td>Anotia/microtia</td>
<td>744.01, 744.23</td>
<td>744.01, 744.21</td>
<td>Q16.0, Q17.2</td>
<td>Level 2</td>
</tr>
<tr>
<td><strong>Cardiovascular</strong></td>
<td></td>
<td></td>
<td></td>
<td>Level 2, Level 2</td>
</tr>
<tr>
<td>Aortic valve stenosis</td>
<td>746.3</td>
<td>746.3</td>
<td>Q23.0</td>
<td>Level 2</td>
</tr>
<tr>
<td>Atrial septal defect</td>
<td>745.5</td>
<td>745.51 – 745.59</td>
<td>Q21.1</td>
<td>Level 2</td>
</tr>
<tr>
<td>Atrioventricular septal defect (Endocardial cushion defect)</td>
<td>745.60, .61, .69</td>
<td>745.60 – 745.69</td>
<td>Q21.2</td>
<td>Level 1</td>
</tr>
<tr>
<td>Coarctation of the aorta</td>
<td>747.1</td>
<td>747.10 – 747.19</td>
<td>Q25.1</td>
<td>Level 2 (CCHD 2° target)</td>
</tr>
<tr>
<td>Birth Defects</td>
<td>ICD-9-CM Codes</td>
<td>CDC/BPA Codes</td>
<td>ICD-10-CM Codes</td>
<td>Standard Level*</td>
</tr>
<tr>
<td>---------------------------------------------</td>
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<td>--------------------------</td>
</tr>
<tr>
<td>Common truncus (truncus arteriosus or TA)</td>
<td>745.0</td>
<td>745</td>
<td>Q20.0</td>
<td>Level 1 (CCHD 1° target)</td>
</tr>
<tr>
<td>Double outlet right ventricle (DORV)</td>
<td>745.11</td>
<td>745.13 – 745.15</td>
<td>Q20.1</td>
<td>Level 2 (CCHD 2° target)</td>
</tr>
<tr>
<td>Ebstein anomaly</td>
<td>746.2</td>
<td>746.20</td>
<td>Q22.5</td>
<td>Level 2 (CCHD 2° target)</td>
</tr>
<tr>
<td>Hypoplastic left heart syndrome</td>
<td>746.7</td>
<td>746.7</td>
<td>Q23.4</td>
<td>Level 1 (CCHD 1° target)</td>
</tr>
<tr>
<td>Pulmonary valve atresia and stenosis</td>
<td>746.01 (pulmonary valve atresia), 746.02 (pulmonary valve stenosis) Note: for CCHD, 746.01 only (pulmonary atresia, intact ventricular septum)</td>
<td>746.00 (pulmonary valve atresia), 746.01 (pulmonary valve stenosis) Note: for CCHD, 746.00 only (pulmonary atresia, intact ventricular septum)</td>
<td>Q22.0, Q22.1 (Note: for CCHD, Q22.0 only (pulmonary atresia, intact ventricular septum))</td>
<td>Level 2 (CCHD 1° target)</td>
</tr>
<tr>
<td>Single Ventricle</td>
<td>745.3</td>
<td>745.3</td>
<td>Q20.4</td>
<td>Level 2 (CCHD 2° target)</td>
</tr>
<tr>
<td>Tetralogy of Fallot (TOF)</td>
<td>745.2</td>
<td>745.20 – 745.21, 747.31 Note: code 746.84 has been removed</td>
<td>Q21.3</td>
<td>Level 1 (CCHD 1° target)</td>
</tr>
<tr>
<td>Birth Defects</td>
<td>ICD-9-CM Codes</td>
<td>CDC/BPA Codes</td>
<td>ICD-10-CM Codes</td>
<td>Standard Level*</td>
</tr>
<tr>
<td>------------------------------------------------------------------------------</td>
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<tr>
<td><strong>Total anomalous pulmonary venous connection (TAPVC)</strong></td>
<td>747.41</td>
<td>747.42</td>
<td>Q26.2</td>
<td>Level 1 (CCHD 1&lt;sup&gt;o&lt;/sup&gt; target)</td>
</tr>
<tr>
<td><strong>Transposition of the great arteries (TGA)</strong></td>
<td>745.10, 745.12, 745.19 (Note: for CCHD, 745.10 only (d-TGA only))</td>
<td>745.10 – 745.12, 745.18 – 745.19 (Note: for CCHD, 745.10 (TGA complete, no VSD), 745.11 (TGA incomplete, with VSD), 749/18 (other specified TGA), 745.19 (unspecified TGA))</td>
<td>Q20.3, Q20.5 (Note: for CCHD, Q20.3 only)</td>
<td>Level 1 (CCHD 1&lt;sup&gt;o&lt;/sup&gt; target)</td>
</tr>
<tr>
<td><strong>Tricuspid valve atresia and stenosis</strong></td>
<td>746.1</td>
<td>746.100 (tricuspid atresia), 746.106 (tricuspid stenosis) (excl. 746.105 – tricuspid insufficiency) Note: for CCHD, 746.100 only. Only tricuspid atresia is a CCHD. Many cases of tricuspid stenosis are not critical.</td>
<td>Q22.4</td>
<td>Level 2 (CCHD 1&lt;sup&gt;o&lt;/sup&gt; target)</td>
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<tr>
<td><strong>Ventricular septal defect</strong></td>
<td>745.4</td>
<td>745.40 – 745.49 (excl. 745.487, 745.498)</td>
<td>Q21.0</td>
<td>Level 2</td>
</tr>
<tr>
<td><strong>Orofacial</strong></td>
<td></td>
<td></td>
<td></td>
<td></td>
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<tr>
<td>Birth Defects</td>
<td>ICD-9-CM Codes</td>
<td>CDC/BPA Codes</td>
<td>ICD-10-CM Codes</td>
<td>Standard Level*</td>
</tr>
<tr>
<td>--------------------------------------------------</td>
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<td>-----------------</td>
</tr>
<tr>
<td>Choanal atresia</td>
<td>748.0</td>
<td>748.0</td>
<td>Q30.0</td>
<td>Level 2</td>
</tr>
<tr>
<td>Cleft lip with cleft palate</td>
<td>749.2</td>
<td>749.20 – 749.29</td>
<td>Q37.0 – Q37.9</td>
<td>Level 1</td>
</tr>
<tr>
<td>Cleft lip alone (without cleft palate)</td>
<td>749.1</td>
<td>749.10-749.19</td>
<td>Q36.0 – Q36.9</td>
<td>Level 1</td>
</tr>
<tr>
<td>Cleft palate alone (without cleft lip)</td>
<td>749.0</td>
<td>749.00 – 749.09</td>
<td>Q35.1 – Q35.9</td>
<td>Level 1</td>
</tr>
<tr>
<td><strong>Gastrointestinal</strong></td>
<td></td>
<td></td>
<td></td>
<td></td>
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<tr>
<td>Biliary atresia</td>
<td>751.61</td>
<td>751.65</td>
<td>Q44.2 - Q44.3</td>
<td>Level 3</td>
</tr>
<tr>
<td>Esophageal atresia/tracheoesophageal fistula</td>
<td>750.3</td>
<td>750.30 – 750.35</td>
<td>Q39.0 – Q39.4</td>
<td>Level 2</td>
</tr>
<tr>
<td>Rectal and large intestinal atresia/stenosis</td>
<td>751.2</td>
<td>751.20 – 751.24</td>
<td>Q42.0 – Q42.9</td>
<td>Level 2</td>
</tr>
<tr>
<td>Small intestinal atresia/stenosis</td>
<td>751.1</td>
<td>751.10-751.19</td>
<td>Q41.0 – Q41.9</td>
<td>Level 2</td>
</tr>
<tr>
<td><strong>Genitourinary</strong></td>
<td></td>
<td></td>
<td></td>
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</tr>
<tr>
<td>Bladder extrophy</td>
<td>753.5</td>
<td>753.5</td>
<td>Q64.10, Q64.19</td>
<td>Level 2</td>
</tr>
<tr>
<td>Cloacal extrophy</td>
<td>751.5</td>
<td>751.555</td>
<td>Q64.12</td>
<td>Level 2</td>
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<tr>
<td>Congenital Posterior Urethral Valves</td>
<td>753.6</td>
<td>753.60</td>
<td>Q64.2</td>
<td>Level 2</td>
</tr>
<tr>
<td>Hypospadias</td>
<td>752.61</td>
<td>752.60 – 752.62 (excluding 752.61 and 752.621)</td>
<td>Q54.0 – Q54.9 (excluding Q54.4)</td>
<td>Level 2</td>
</tr>
<tr>
<td>Renal agenesis/hypoplasia</td>
<td>753.0</td>
<td>753.00 – 753.01</td>
<td>Q60.0 – Q60.6</td>
<td>Level 2</td>
</tr>
<tr>
<td><strong>Musculoskeletal</strong></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Clubfoot</td>
<td>754.51, 754.70</td>
<td>754.50, 754.73</td>
<td>Q66.0, Q66.89</td>
<td>Level 2</td>
</tr>
<tr>
<td>Craniosynostosis</td>
<td>No specific code</td>
<td>756.00-756.03</td>
<td>Q75.0</td>
<td>Level 3</td>
</tr>
<tr>
<td>Diaphragmatic hernia</td>
<td>756.6</td>
<td>756.61</td>
<td>Q79.0, Q79.1</td>
<td>Level 2</td>
</tr>
<tr>
<td>Birth Defects</td>
<td>ICD-9-CM Codes</td>
<td>CDC/BPA Codes</td>
<td>ICD-10-CM Codes</td>
<td>Standard Level*</td>
</tr>
<tr>
<td>--------------------------------------</td>
<td>-------------------------------------------------------------------------------</td>
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</tr>
<tr>
<td><strong>Gastroschisis</strong></td>
<td>756.73 (as of 10/1/09; previously a shared code 756.79 with omphalocele)</td>
<td>756.71</td>
<td>Q79.3</td>
<td>Level 1</td>
</tr>
<tr>
<td><strong>Limb deficiencies (reduction defects)</strong></td>
<td>755.2 – 755.4</td>
<td>755.20 – 755.49</td>
<td>Q71.0 – Q71.9, Q72.0 – Q72.9, Q73.0 – Q73.8</td>
<td>Level 1</td>
</tr>
<tr>
<td><strong>Omphalocele</strong></td>
<td>756.72 (as of 10/1/09; previously a shared code 756.79 with gastroschisis)</td>
<td>756.7</td>
<td>Q79.2</td>
<td>Level 2</td>
</tr>
<tr>
<td><strong>Chromosomal</strong></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td><strong>Deletion 22 q11</strong></td>
<td>758.32</td>
<td>758.37</td>
<td>Q93.81</td>
<td>Level 3</td>
</tr>
<tr>
<td><strong>Trisomy 13</strong></td>
<td>758.1</td>
<td>758.10 – 758.19</td>
<td>Q91.4 – Q91.7</td>
<td>Level 2</td>
</tr>
<tr>
<td><strong>Trisomy 18</strong></td>
<td>758.2</td>
<td>758.20 – 758.29</td>
<td>Q91.0 – Q91.3</td>
<td>Level 2</td>
</tr>
<tr>
<td><strong>Trisomy 21 (Down syndrome)</strong></td>
<td>758.0</td>
<td>758.00 – 758.09</td>
<td>Q90.0 – Q90.9</td>
<td>Level 1</td>
</tr>
<tr>
<td><strong>Turner syndrome</strong></td>
<td>758.6</td>
<td>758.60-758.69</td>
<td>Q96.0-.9</td>
<td>Level 3</td>
</tr>
</tbody>
</table>
**Detailed Descriptions of Birth Defects**

**Format for Birth Defect Descriptions**

<table>
<thead>
<tr>
<th>Defect Name</th>
<th>Description</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>Description</strong></td>
<td>Description of the defect.</td>
</tr>
<tr>
<td></td>
<td>Standard level (SL): Each condition is listed as core (SL 1), recommended (SL 2) or extended (SL 3). In order to meet the standard level specified, a program needs to ascertain that condition.</td>
</tr>
<tr>
<td><strong>Inclusions</strong></td>
<td>Other names or conditions that should be included in the code for the defect.</td>
</tr>
<tr>
<td><strong>Exclusions</strong></td>
<td>Other names or conditions that should not be included in the code for the defect.</td>
</tr>
<tr>
<td><strong>ICD-9-CM Codes</strong></td>
<td>Applicable ICD-9-CM codes for the defect.</td>
</tr>
<tr>
<td><strong>ICD-10-CM Codes</strong></td>
<td>Applicable ICD-10-CM codes for the defect.</td>
</tr>
<tr>
<td><strong>CDC/BPA Codes</strong></td>
<td>Applicable CDC/BPA codes for the defect.</td>
</tr>
<tr>
<td><strong>Diagnostic Methods</strong></td>
<td>Postnatal procedures by which the defect may be accurately and reliably diagnosed.</td>
</tr>
<tr>
<td><strong>Prenatal Diagnoses Not Confirmed Postnatally</strong></td>
<td>Guidance on whether cases with only a prenatal diagnosis should be included in the defect code.</td>
</tr>
<tr>
<td><strong>Additional Information</strong></td>
<td>Tips and useful information about the defect.</td>
</tr>
</tbody>
</table>
Central Nervous System

Anencephaly
(Core Condition)

Description
Partial or complete absence of the brain and skull.

Inclusions
- Acrania – Absence of skull bones with some brain tissue present.
- Absent brain, with or without skull bones present.
- Anencephaly
- Craniorachischisis – Anencephaly continuous with an open posterior spinal defect with no meninges covering the neural tissue.

Exclusions
- Encephalocele
- Iniencephaly
- Rachischisis – When used alone, this term refers only to the spinal defect and should be coded as spina bifida without anencephaly.

ICD-9-CM Codes
740.0 – 740.1

ICD-10-CM Codes
Q00.0 - Q00.1

CDC/BPA Codes
740.00 – 740.10

Diagnostic Methods
Anencephaly is easily recognized on physical examination at delivery.

Prenatal Diagnoses Not Confirmed Postnatally
Anencephaly may be included when only diagnosed prenatally. However, if it is possible to ascertain the degree of certainty of the prenatal diagnosis, this should factor into the decision as to whether or not to include an individual case in the surveillance data.

Additional Information:
Anencephaly is one of a group of defects that result from failure of the neural tube to close.

Maternal serum alphafetoprotein (MSAFP) and/or amniotic fluid alphafetoprotein (AFAFP) and amniotic fluid acetylcholinesterase (ACHE) may be elevated with anencephaly. However, these screening tests alone are not sufficient to diagnose the condition.

In cases where both anencephaly and spina bifida are present but are not continuous (i.e., not craniorachischisis), both anencephaly and spina bifida should be coded.
### Encephalocele
(Recommended Condition)

<table>
<thead>
<tr>
<th>Description</th>
<th>Herniation of brain tissue and/or meninges through a defect in the skull. The hernia sac is usually covered by skin.</th>
</tr>
</thead>
</table>

| Inclusions  | Cephalocele  
Cranial meningocele – Herniation of meninges only.  
Encephalocele  
Encephalomyelocele - Herniation through a defect in a portion of both the skull and the upper spine.  
Encephalocystomeningocele  
Hydranencephalocele  
Meningoencephalocele  
Ventriculocele |
|-------------|---------------------------------------------------------------------------------------------------------------|

<table>
<thead>
<tr>
<th>Exclusions</th>
<th>NA</th>
</tr>
</thead>
</table>

<table>
<thead>
<tr>
<th>ICD-9-CM Codes</th>
<th>742.0</th>
</tr>
</thead>
<tbody>
<tr>
<td>ICD-10-CM Codes</td>
<td>Q01.0 – Q01.9</td>
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<tr>
<td>CDC/BPA Codes</td>
<td>742.00 – 742.09</td>
</tr>
</tbody>
</table>

<table>
<thead>
<tr>
<th>Diagnostic Methods</th>
<th>Most cases of encephalocele are recognizable on physical examination after delivery. However, they may be conclusively diagnosed only through direct visualization of the brain by cranial ultrasound, CT or MRI scan, surgery, or autopsy. This is particularly true for internal herniations through the sphenoid, maxillary, or ethmoid bones, the orbit, or pharynx.</th>
</tr>
</thead>
</table>

<table>
<thead>
<tr>
<th>Prenatal Diagnoses Not Confirmed Postnatally</th>
<th>Encephalocele may be included when only diagnosed prenatally. However, if it is possible to ascertain the degree of certainty of the prenatal diagnosis, this should factor into the decision as to whether or not to include an individual case in the surveillance data. In addition, the absence of a small encephalocele on prenatal ultrasound does not necessarily mean that it will not be diagnosed after delivery.</th>
</tr>
</thead>
</table>

**Additional Information:**
Encephaloceles are often included as one of a group of defects that result from failure of the neural tube to close. Maternal serum alphafetoprotein (MSAFP) and/or amniotic fluid alphafetoprotein (AFAFP) and amniotic fluid acetylcholinesterase (ACHE) may be elevated with encephaloceles. However, these screening tests alone are not sufficient to diagnose the condition. Occipital encephalocele is a component of Meckel-Gruber syndrome.
## Holoprosencephaly
*(Extended Condition)*

### Description
Structural brain anomaly that results from variable degrees of incomplete cleavage of the prosencephalon (embryonic forebrain), which fails to cleave sagittally into the right and left cerebral hemispheres and transversely into telencephalon and diencephalon.

### Inclusions
- Alobar holoprosencephaly
- Semilobar holoprosencephaly
- Lobar holoprosencephaly
- Middle interhemispheric variant (MIHV)
- Holotelencephaly
- Cyclopia
- Cebocephaly
- Ethmocephaly

### Exclusions
- Atelencephaly
- Hydranencephaly
- Porencephaly
- Arhinencephaly without holoprosencephaly

### ICD-9-CM Codes
- 742.2

### ICD-10-CM Codes
- Q04.2

### CDC/BPA Codes
- 742.26

### Diagnostic Methods
Confirmation of a diagnosis of holoprosencephaly is by CT, MRI, or autopsy.

### Prenatal Diagnoses Not Confirmed Postnatally
Holoprosencephaly may be included when only diagnosed prenatally. However, if it is possible to ascertain the degree of certainty of the prenatal diagnosis, this should factor into the decision as to whether or not to include an individual case in the surveillance data. For example, clear diagnoses of cyclopia, ethmocephaly, or cebocephaly are virtually always associated with holoprosencephaly, but prenatal diagnoses of lobar holoprosencephaly and middle interhemispheric variants are more problematic without postnatal imaging or autopsy confirmation.

### Additional Information:
Holoprosencephaly, especially the alobar type, is commonly associated with facial anomalies that range from hypotelorism and median cleft lip (premaxillary agenesis) to cyclopia, a rare abnormality characterized by a single central eye in the low frontal area and a missing nose or a proboscis (a tubular-shaped nose) located above the eye. Other similarly uncommon facial anomalies include ethmocephaly, in which a proboscis is found close to the root of the nose, and cebocephaly, characterized by a small nose with a single nostril situated below underdeveloped eyes.
Spina Bifida without Anencephaly
(Core Condition)

Description
Incomplete closure of the vertebral spine (usually posteriorly) through which spinal cord tissue and/or the membranes covering the spine (meninges) herniate.

Inclusions
- Lipomeningocele
- Lipomyelomeningocele
- Meningocele – Herniation of meninges only.
- Meningomyelocele, Myelomeningocele – Herniation of meninges and spinal cord tissue
- Myelocystocele
- Myelodysplasia
- Myeloschisis
- Open spina bifida
- Rachischisis – Open spina bifida without meninges covering the spinal cord tissue
- Spina bifida aperta
- Spina bifida cystica

Exclusions
- Diastematomyelia
- Diplomyelia
- Hydromyelia
- Spina bifida with coexisting anencephaly – Code only as anencephaly
- Spina bifida occulta
- Syringomyelia
- Tethered spinal cord

ICD-9-CM Codes
741.0 or 741.9 without 740.0 – 740.1

ICD-10-CM Codes
Q05.0 - Q05.9 or Q07.01 or Q07.03 without Q00.0 - Q00.1

CDC/BPA Codes
741.00 – 741.99 without 740.00 – 740.10

Diagnostic Methods
The majority of defects result in a direct opening on the infant’s back that is easily recognized on physical examination at delivery. However, the exact nature of the defect (meningocele vs. myelomeningocele) may only be distinguished by CT or MRI scan, at surgery, or at autopsy.

Prenatal Diagnoses Not Confirmed Postnataally
Spina bifida may be included when only diagnosed prenatally. However, if it is possible to ascertain the degree of certainty of the prenatal diagnosis, this should factor into the decision as to whether or not to include an individual case in the surveillance data. In addition, the absence of spina bifida on prenatal ultrasound does not necessarily mean that it will not be diagnosed after delivery.
**Additional Information:**
Spina bifida is one of a group of defects that result from failure of the neural tube to close.

Open lesions (spina bifida cystica, spina bifida aperta) are those with no covering or with only meninges covering the neural tissue. They usually leak cerebrospinal fluid. Closed lesions are covered by normal skin.

Hydrocephalus and Arnold-Chiari malformation of the brain frequently, though not always, result from spina bifida. When present, there is no need to code them separately from the spina bifida.

Maternal serum alphafetoprotein (MSAFP) and/or amniotic fluid alphafetoprotein (AFAFP) and amniotic fluid acetylcholinesterase (ACHE) may be elevated in spina bifida. However, these screening tests alone are not sufficient to diagnose the condition.

In cases where both anencephaly and spina bifida are present but are not continuous (i.e., not craniorachischisis), both anencephaly and spina bifida should be coded.

If the defect coding system includes unique codes for different levels of spina bifida (cervical; thoracic; lumbar; sacral) and a defect involves more than one level (cervicothoracic; thoracolumbar; lumbosacral), the highest level at which it occurs should be coded (i.e., cervical; thoracic; lumbar). The highest level of involvement determines the degree of associated neurologic impairment.
### Eye

#### Anophthalmia/Microphthalmia

(Recommended Condition)

| Description | Anophthalmia – Total absence of eye tissue or apparent absence of the globe in an otherwise normal orbit. Microphthalmia – Reduced volume of the eye. The corneal diameter is usually less than 10 millimeters, or the anteroposterior globe diameter is less than 20 millimeters. |
| Inclusions | Anophthalmia Microphthalmia Nanophthalmia – Microphthalmia with normal internal eye (intraocular) structures. This is a distinct genetic condition. |
| Exclusions | Small eyes or small palpebral fissures for which the diagnosis of microphthalmia or anophthalmia has not been made. Microcornea with otherwise normal eye size. |
| ICD-9-CM Codes | 743.0, 743.1 |
| ICD-10-CM Codes | Q11.0 – Q11.2 |
| CDC/BPA Codes | 743.00 – 743.10 |
| Diagnostic Methods | These conditions are usually recognized on physical examination after delivery, especially by an ophthalmologist. However, the anteroposterior diameter of the globe may be measured only by ultrasound, CT or MRI scan, or at autopsy. |
| Prenatal Diagnoses Not Confirmed Postnatally | While these conditions may be identified by prenatal ultrasound, they should not be included in surveillance data without postnatal confirmation. In addition, the absence of anophthalmia or microphthalmia on prenatal ultrasound does not necessarily mean that it will not be diagnosed after delivery. |

#### Additional Information:

- Microphthalmia may occur in association with colobomas (gaps) in the uvea, iris, choroid and/or optic nerve (colobomatous microphthalmia).

- Anophthalmia and microphthalmia often are accompanied by malformations of the brain and face, and frequently are components of genetic syndromes.
### Congenital Cataract
(Extended Condition)

<table>
<thead>
<tr>
<th>Description</th>
<th>An opacity of the lens of the eye that has its origin prenatally.</th>
</tr>
</thead>
</table>
| Inclusions  | Anterior polar cataract  
Cataract, type not specified  
Infantile cataract  
Lamellar cataract  
Nuclear cataract  
Posterior lentiglobus/lenticonus cataract  
Posterior cortical cataract  
Sectoral cataract  
Zonular cataract |
| Exclusions  | Any of the above types of cataract that has its origin after birth  
Corneal opacities |
| ICD-9-CM Codes | 743.30 – 743.34 |
| ICD-10-CM Codes | Q12.0 |
| CDC/BPA Codes | 743.32 |
| Diagnostic Methods | Some cataracts are readily apparent on physical examination. Others are visible with an ophthalmoscope. However, they may be conclusively diagnosed only through examination by an ophthalmologist. |
| Prenatal Diagnoses Not Confirmed Postnatally | While this condition may be identified by prenatal ultrasound, it should not be included in surveillance data without postnatal confirmation. In addition, the absence of a cataract on prenatal ultrasound does not necessarily mean that it will not be diagnosed after delivery. |

**Additional Information:**
Cataracts may be congenital, acquired, or inherited. They may involve all or only part of the lens of either or both eyes. They may be an isolated finding in an otherwise normal eye, or may be part of a more general eye malformation. They may be seen with metabolic disorders, such as galactosemia; genetic syndromes, such as chondrodysplasia punctata; chromosomal abnormalities, such as Trisomy 21; intrauterine infection, such as congenital rubella; or trauma.

In some instances, the severity of the cataract progresses over time. The need for surgical treatment depends on the degree of visual impairment.

When congenital cataract occurs with microphthalmia in the same infant, both conditions should be coded.
## Ear

### Anotia/Microtia

(Recommended Condition)

<table>
<thead>
<tr>
<th>Description</th>
<th>Anotia – Total absence of the external ear and canal. Microtia – Malformation or hypoplasia of the external ear (auricle, pinna).</th>
</tr>
</thead>
<tbody>
<tr>
<td>Inclusions</td>
<td>Anotia, Microtia</td>
</tr>
<tr>
<td>Exclusions</td>
<td>Small ears that retain most of the overall structure of the normal auricle, including lop or cup ear defects. In these, the auditory meatus is usually patent and defects of the ossicular chain of the middle ear are infrequent. However, these defects are sometimes designated as Type I Microtia. Isolated absence, atresia, stenosis or malformation of the ear canal with a normal external ear. Congenital absence of the ear not diagnosed as anotia or microtia.</td>
</tr>
<tr>
<td>ICD-9-CM Codes</td>
<td>744.01, 744.23</td>
</tr>
<tr>
<td>ICD-10-CM Codes</td>
<td>Q16.0, Q17.2</td>
</tr>
<tr>
<td>CDC/BPA Codes</td>
<td>744.01, 744.21</td>
</tr>
<tr>
<td>Diagnostic Methods</td>
<td>Anotia and microtia are usually easily recognized on physical examination after delivery. However, abnormalities of the middle and inner ear may be conclusively diagnosed only by CT or MRI scan, surgery, or autopsy.</td>
</tr>
<tr>
<td>Prenatal Diagnoses Not Confirmed Postnatally</td>
<td>While these conditions may be identified by prenatal ultrasound, they should not be included in surveillance data without postnatal confirmation. In addition, the absence of anotia or microtia on prenatal ultrasound does not necessarily mean that they will not be diagnosed after delivery.</td>
</tr>
</tbody>
</table>

### Additional Information:
The spectrum of severity of microtia may range from a measurably small external ear with minimal structural abnormality to major structural alteration of the external ear with an absent or blind-ending canal. Following is the classification system of Meurman (modified from Marks):

- **Type I** – Generally small ears that retain most of the overall structure of the normal auricle. These
should not be coded as microtia.

Type II B – A moderately severe anomaly with a longitudinal mass of cartilage with some resemblance to a pinna. The rudimentary auricle may be hook-shaped, have an S-shape, or the appearance of a question mark.

Type III B – The ear is a rudiment of soft tissue and the auricle has no resemblance to a normal pinna.

Type IV B – Complete absence of all external ear structures (anotia).

Abnormalities that may be associated with anotia/microtia include anomalies of the middle and/or inner ear, the mandible and face, and hearing loss.

Anotia/microtia may be a component of Goldenhar and other syndromes.
## Cardiovascular

### Aortic Valve Stenosis

(Recommended Condition)

<table>
<thead>
<tr>
<th>Description</th>
<th>Obstruction or narrowing of the aortic valve, which may impair blood flow from the left ventricle to the aorta.</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>Inclusions</strong></td>
<td>Stenosis of the aortic valve</td>
</tr>
<tr>
<td><strong>Exclusions</strong></td>
<td>Stenosis of the aorta without mention of the aortic valve. Supra-valvular or sub-valvular aortic stenosis.</td>
</tr>
<tr>
<td><strong>ICD-9-CM Codes</strong></td>
<td>746.3</td>
</tr>
<tr>
<td><strong>ICD-10-CM Codes</strong></td>
<td>Q23.0</td>
</tr>
<tr>
<td><strong>CDC/BPA Codes</strong></td>
<td>746.30</td>
</tr>
<tr>
<td><strong>Diagnostic Methods</strong></td>
<td>While aortic valve stenosis may be suspected by clinical presentation, it may be conclusively diagnosed only through direct visualization of the heart by cardiac echo (echocardiography), catheterization, surgery, or autopsy.</td>
</tr>
<tr>
<td><strong>Prenatal Diagnoses Not Confirmed Postnatally</strong></td>
<td>While this condition may be identified by prenatal ultrasound, it should not be included in surveillance data without postnatal confirmation. In addition, the absence of aortic valve stenosis on prenatal ultrasound does not necessarily mean that it will not be diagnosed after delivery.</td>
</tr>
<tr>
<td><strong>Additional Information</strong></td>
<td>NA</td>
</tr>
</tbody>
</table>
### Atrial Septal Defect (ASD)

**Description**
An opening in the wall (septum) that separates the left and right top chambers (atria) of the heart.

| Inclusions | Atrial septal defect (ASD), type not specified (NOS)  
ASD other specified (OS) – which includes sinus venosus type  
ASD secundum type (ASD 2 or ASD II)  
ASD vs. PFO – In the first days of life, it may not be possible to distinguish whether the opening in the atrial septum is a true ASD or a patent foramen ovale that has not yet closed (see below). ASD vs. PFO should be included only if the exact nature of the condition was never resolved.  
Exclusions | Atroventricular septal defects (AVSD)  
ASD primum type (1° ASD) – This is included under atroventricular septal defects (see below).  
Patent foramen ovale (PFO) – A PFO is normal *in utero* to allow blood to flow properly during fetal circulation. This usually closes shortly after birth, but frequently does not close until 24 to 48 hours after birth.  
ICD-9-CM Codes | 745.5  
ICD-10-CM Codes | Q21.1  
CDC/BPA Codes | 745.51 – 745.59  
Diagnostic Methods | Some isolated ASDs may be diagnosed based on physical examination and/or EKG without direct imaging of the heart. However, many ASDs may be conclusively diagnosed only through direct visualization of the heart by cardiac echo (echocardiography), catheterization, surgery, or autopsy.  
Prenatal Diagnoses Not Confirmed Postnatally | While ASDs may be identified by prenatal ultrasound, they may close spontaneously before delivery. For this reason, ASDs that are diagnosed prenatally should not be included unless they have been confirmed postnatally. In addition, the absence of an ASD on prenatal ultrasound does not necessarily mean that an ASD will not be diagnosed after delivery, as it is not always possible to accurately visualize the entire atrial septum by prenatal ultrasound.  
Additional Information:  
Types of ASDs are denoted by location on the septum and when they formed in utero. Secundum ASDs are usually located toward the middle of the atrial septum. Some close spontaneously without treatment. Primum ASDs are located in the lower portion of the atrial septum near the atrioventricular valves, are etiologically related to atroventricular septal defects, and never close spontaneously.  

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*Appendix 3.1*

*Case Definition*
## Atrioventricular Septal Defect

(Atrioventricular Canal Defect; Endocardial Cushion Defect)

(Core Condition)

### Description

A defect in both the lower portion of the atrial septum and the upper portion of the ventricular septum. In extreme cases, virtually the entire atrial and ventricular septae may be missing. The valves controlling blood flow from the atria to the ventricles, the tricuspid and mitral valves may also be abnormal. They may not form from the endocardial cushions during cardiac development into two separate valves, and thus be a single common atrioventricular valve. Together, these defects producing a large opening (canal) in the central part of the heart.

### Inclusions

- Atrioventricular septal defect (AVSD)
- Common or complete atrioventricular (AV) canal
- Endocardial cushion defect

Primum type atrial septal defect (1° ASD) – A defect only in the lower portion of the atrial septum. While this does not involve a defect in the upper portion of the ventricular septum, it is etiologically related to the more complete form of AVSD. A cleft mitral valve is often present with a primum type ASD (see partial AVC).

Common atrium – Near absence of the atrial septum.

Partial AV canal (partial endocardial cushion defect) – Refers to a primum ASD with cleft mitral valve.

Inflow-type, subtricuspid, or canal-type ventricular septal defect (VSDAVC) – A defect in the upper (inflow) portion of the ventricular septum. While this does not also involve a defect in the lower portion of the atrial septum, it is etiologically related to the more complete form.

### Exclusions

Secundum ASDs that coexist with a VSD. In this instance, both the ASD and the VSD should be coded.

### ICD-9-CM Codes

- 745.60
- 745.61
- 745.69

### ICD-10-CM Codes

- Q21.2

### CDC/BPA Codes

- 745.60 – 745.69, 745.487

### Diagnostic Methods

While atrioventricular septal defects may be suspected by clinical presentation, examination, and EKG changes, it may be conclusively diagnosed only through direct visualization of the heart by cardiac echo (echocardiography), catheterization, surgery, or autopsy.

### Prenatal Diagnoses Not

These conditions may be included as cases when only diagnosed prenatally.
**Confirmed Postnatally**

However, if it is possible to ascertain the degree of certainty of the prenatal diagnosis, this should factor into the decision as to whether or not to include an individual case in the surveillance data, as it may be difficult to distinguish this condition from other abnormalities of the cardiac septae prenatally. Live-born children who survive should always have confirmation of the defect postnatally.

**Additional Information:**

Atrioventricular septal defects are known to be associated with Down syndrome. Approximately 40% of children with Down syndrome have some type of CHD, 20% have an atrioventricular septal defect. Conversely, approximately 70% of children with an atrioventricular septal defect have Down syndrome.
Coarctation of the Aorta  
(Recommended Condition)

**Description**
Narrowing of the descending aorta, which may obstruct blood flow from the heart to the rest of the body. The most common site of coarctation occurs distal to the origin of the left subclavian artery in the region of the ductus arteriosus. If there is complete loss of communication in this location, it is a form of interruption of the aorta (Type A).

**Inclusions**
Coarctation of the aorta, type not specified  
Preductal, juxtaductal, and postductal coarctations – These terms refer to the exact placement of the segment of coarctation relative to the insertion of the ductus arteriosus.

**Exclusions**
NA

**ICD-9-CM Codes**
747.10

**ICD-10-CM Codes**
Q25.1

**CDC/BPA Codes**
747.10 – 747.19

**Diagnostic Methods**
While coarctation of the aorta may be suspected by clinical presentation and examination, it may be conclusively diagnosed only through direct visualization of the heart by cardiac echo (echocardiography), catheterization, surgery, or autopsy.

**Prenatal Diagnoses Not Confirmed Postnatally**
While this condition may be identified by prenatal ultrasound, it should not be included in surveillance data without postnatal confirmation. In addition, the absence of coarctation of the aorta on prenatal ultrasound does not necessarily mean that it will not be diagnosed after delivery.

**Additional Information:**
Left-sided obstructive lesions of the heart, such as coarctation, have been associated with Turner syndrome (karyotype 45,X and other variants).
## Common Truncus (Truncus Arteriosus or TA)  
(Core Condition)

<table>
<thead>
<tr>
<th><strong>Description</strong></th>
<th>Failure of separation of the aorta and the pulmonary artery during development, resulting in a single common arterial trunk carrying blood from the heart to both the body and lungs.</th>
</tr>
</thead>
</table>
| **Inclusions**  | Common truncus  
Truncus arteriosus (TA)  
Persistent truncus arteriosus |
| **Exclusions**  | Aorto-pulmonary window. In ICD-9-CM, this related defect is not distinguished from truncus. An AP window is a hole (aka “window”) between a separate aorta and pulmonary artery. This is distinct from truncus, when neither vessel forms separately. |
| **ICD-9-CM Codes** | 745.0 |
| **ICD-10-CM Codes** | Q20.0 |
| **CDC/BPA Codes** | 745.00 only (excluding 745.01, aortic septal defect which including aorto-pulmonary window) |
| **Diagnostic Methods** | Truncus arteriosus is conclusively diagnosed only through direct visualization of the heart by cardiac imaging (typically echocardiography but also MRI), catheterization, surgery, or autopsy. A clinical diagnosis is considered insufficient to make the diagnosis. |
| **Prenatal Diagnoses Not Confirmed Postnatally** | These conditions may be included as cases when only diagnosed prenatally by a pediatric cardiologist through fetal echocardiography. However, if it is possible to ascertain the degree of certainty of the prenatal diagnosis, this should factor into the decision as to whether or not to include an individual case in the surveillance data. Live-born children who survive should always have confirmation of the defect postnatally. |

**Additional Information:**

A ventricular septal defect is often present in association with truncus defects and should be coded separately. Truncus arteriosus is one of several abnormalities of the outflow tract of the heart known as conotruncal defects. Some infants (1 in 5 to 1 in 3) with these defects have a deletion on the short arm of chromosome 22 (deletion 22q11.2). This deletion may not necessarily be detected on a routine karyotype analysis and is more reliably diagnosed by fluorescent in situ hybridization (FISH) or microarray technology.
## Double Outlet Right Ventricle (DORV)

### (Recommended Condition)

| **Description** | Both the pulmonary artery and the aorta arise from the right ventricle, usually accompanied by a ventricular septal defect (VSD). DORV subtypes are usually distinguished by the great artery anatomic relationship: DORV with normally related great arteries and DORV with “transposed” or malposed or side-by-side great arteries. Actually, the arteries are not truly “transposed”, which refers to the aorta arising from the right ventricle and pulmonary artery from the left ventricle, since in DORV both great arteries arise from the right ventricle. |
| **Inclusions** | Double outlet right ventricle (DORV) with normally related great vessels DORV with transposed great vessels DORV with unknown relationship of great vessels Taussig-Bing syndrome – |
| **Exclusions** | NA |
| **ICD-9-CM Codes** | 745.11 |
| **ICD-10-CM Codes** | Q20.1 |
| **CDC/BPA Codes** | 745.13 - 745.15 |
| **Diagnostic Methods** | DORV is conclusively diagnosed through direct visualization of the heart by cardiac echo (echocardiography), catheterization, surgery, or autopsy. |
| **Prenatal Diagnoses Not Confirmed Postnatally** | These conditions may be included as cases when only diagnosed prenatally. However, if it is possible to ascertain the degree of certainty of the prenatal diagnosis, this should factor into the decision as to whether or not to include an individual case in the surveillance data. Live-born children who survive should always have confirmation of the defect postnatally. |

### Additional Information:

Distinction of types of DORV is often done on the basis of the anatomic relationship of the great arteries/vessels, which can be malposed, side-by-side, normal or undetermined. However, the coding systems are somewhat confusing in representing these anatomic distinctions. In ICD-9-CM, the single code for DORV is contained under the broader category of Transposition of Great Arteries (TGA). Actually, the arteries are not truly “transposed”, which refers to the aorta arising from the right ventricle and pulmonary artery from the left ventricle, since in DORV both great arteries arise from the right ventricle, regardless of how they are related positionally. In ICD-10-CM, there also is no distinction for great artery relationship, but the single code for DORV is no longer a subtype under TGA. In the latest version of modified CDC/BPA codes there are separate DORV codes depended on knowledge of the great artery relationship.

Previously, for surveillance guidelines, all DORV was included in the TGA category, following the coding system structure. However, now there is a new separate category for all types of DORV.
### Ebstein Anomaly
(Recommended Condition)

**Description**
Abnormal formation and downward displacement of the tricuspid valve into the right ventricle. The tricuspid valve is usually hypoplastic and regurgitant. As a result, the right atrium is enlarged and the right ventricle is small. There may also be associated pulmonary stenosis as the abnormal tricuspid valve tissue obstructs blood flow out of the pulmonary valve.

**Inclusions**
- Ebstein’s anomaly
- Ebstein malformation

**Exclusions**
NA

**ICD-9-CM Codes**
746.2

**ICD-10-CM Codes**
Q22.5

**CDC/BPA Codes**
746.20

**Diagnostic Methods**
While Ebstein’s anomaly may be suspected by clinical presentation, it may be conclusively diagnosed only through direct visualization of the heart by cardiac echo (echocardiography), catheterization, surgery, or autopsy.

**Prenatal Diagnoses Not Confirmed Postnatally**
While this condition may be identified by prenatal ultrasound, it should not be included in surveillance data without postnatal confirmation. In addition, the absence of Ebstein’s anomaly on prenatal ultrasound does not necessarily mean that it will not be diagnosed after delivery.

**Additional Information:**
Ebstein’s anomaly has been associated with lithium exposure during gestation. However, the magnitude of this association is probably very small.
# Hypoplastic Left Heart Syndrome (HLHS)

(Phenotypic Condition)

| **Description** | A condition in which the structures on the left side of the heart and the aorta are extremely small, insufficient to support systemic circulation and with normally related great arteries. Classically, this condition includes hypoplasia of the left ventricle, atresia or severe hypoplasia of both the mitral and aortic valves, hypoplasia of the aortic arch, and coarctation of the aorta. |
| **Inclusions** | Any diagnosis of hypoplastic left heart syndrome, regardless of whether all conditions in the classical definition are present. |
| **Exclusions** | Hypoplasia or diminished size of the left ventricle alone without involvement of other structures on the left side of the heart or the aorta. Hypoplastic left heart or small left ventricle that occurs as part of another complex heart defect, such as atrioventricular septal defect. |
| **ICD-9-CM Codes** | 746.7 |
| **ICD-10-CM Codes** | Q23.4 |
| **CDC/BPA Codes** | 746.70 |
| **Diagnostic Methods** | While hypoplastic left heart may be suspected by clinical presentation, examination, and EKG changes, it may be conclusively diagnosed only through direct visualization of the heart by cardiac echo (echocardiography), catheterization, surgery, or autopsy. |
| **Prenatal Diagnoses Not Confirmed Postnatally** | These conditions may be included as cases when only diagnosed prenatally. However, if it is possible to ascertain the degree of certainty of the prenatal diagnosis, this should factor into the decision as to whether or not to include an individual case in the surveillance data, as it may be difficult to distinguish this condition from other abnormalities of the left ventricle prenatally. Live-born children who survive should always have confirmation of the defect postnatally before being included. |
| **Additional Information** | NA |
# Interrupted Aortic Arch (IAA)

(Recommended Condition)

<table>
<thead>
<tr>
<th>Description</th>
<th>Complete loss of communication (interruption) between the ascending and descending aorta, usually associated with a malalignment-type ventricular septal defect (VSD). Types of IAA are defined by where the interruption occurs along the arch from the conotruncus to the descending aorta. Type A involves the distal descending aorta distal to the left subclavian artery in the same region as coarctation of the aorta, and is considered an extreme version of that obstructive defect. Type B interruption occurs between the left carotid and subclavian, and is considered a conotruncal heart defect; it is the more common form of interrupted aortic arch.</th>
</tr>
</thead>
<tbody>
<tr>
<td>Inclusions</td>
<td>IAA types A, B or C, or all IAA if type unknown or not otherwise specified (NOS).</td>
</tr>
<tr>
<td>Exclusions</td>
<td>NA</td>
</tr>
<tr>
<td>ICD-9-CM Codes</td>
<td>747.11</td>
</tr>
<tr>
<td>ICD-10-CM Codes</td>
<td>Q25.2, Q25.4</td>
</tr>
<tr>
<td>CDC/BPA Codes</td>
<td>747.215 - 747.217</td>
</tr>
<tr>
<td>Diagnostic Methods</td>
<td>IAA is conclusively diagnosed through direct visualization of the heart by cardiac echo (echocardiography), catheterization, surgery, or autopsy.</td>
</tr>
<tr>
<td>Prenatal Diagnoses Not Confirmed Postnatally</td>
<td>These conditions may be included as cases when only diagnosed prenatally. However, if it is possible to ascertain the degree of certainty of the prenatal diagnosis, this should factor into the decision as to whether or not to include an individual case in the surveillance data. Live-born children who survive should always have confirmation of the defect postnatally.</td>
</tr>
<tr>
<td>Additional Information</td>
<td>NA</td>
</tr>
</tbody>
</table>
Pulmonary Valve Atresia and Stenosis
(Recommended Condition)

Description
Pulmonary valve atresia – Lack of patency, or failure of formation altogether, of the pulmonary valve, resulting in obstruction of blood flow from the right ventricle to the pulmonary artery.

Pulmonary valve stenosis – Obstruction or narrowing of the pulmonary valve, which may impair blood flow from the right ventricle to the pulmonary artery.

Inclusions
Pulmonary valve atresia with intact ventricular septum
Pulmonary valve stenosis (PS) (most cases of PS)
Pulmonic stenosis (PS)

Exclusions
Atresia or stenosis of the main or branch (right or left) pulmonary arteries, not involving the pulmonary valve.
Pulmonary stenosis that occurs as part of Tetralogy or Pentalogy of Fallot. Supra-valvular or sub-valvular pulmonic stenosis.

ICD-9-CM Codes
For CCHD Screening
746.01 (pulmonary valve atresia), 746.02 (pulmonary valve stenosis)
746.01 only (pulmonary atresia, intact ventricular septum)

ICD-10-CM Codes
For CCHD Screening
Q22.0, Q22.1
Q22.0 only (pulmonary atresia, intact ventricular septum)

CDC/BPA Codes
For CCHD Screening
746.00 (pulmonary valve atresia), 746.01 (pulmonary valve stenosis)
746.00 only (pulmonary atresia, intact ventricular septum)

Diagnostic Methods
While pulmonary valve atresia or stenosis may be suspected by clinical presentation, it may be conclusively diagnosed only through direct visualization of the heart by cardiac echo (echocardiography), catheterization, surgery, or autopsy.

Prenatal Diagnoses Not Confirmed Postnatally
While these conditions may be identified by prenatal ultrasound, they should not be included in surveillance data without postnatal confirmation. In addition, the absence of pulmonary valve atresia or stenosis on prenatal ultrasound does not necessarily mean that it will not be diagnosed after delivery.

Additional Information:
These defects have important physiological and coding differences among systems as seen here in the Table, which is also discussed in the Tetralogy of Fallot section.

<table>
<thead>
<tr>
<th></th>
<th>CCHD</th>
<th>ICD-9</th>
<th>CDC/BPA</th>
</tr>
</thead>
<tbody>
<tr>
<td>PVS</td>
<td>746.02</td>
<td>746.01</td>
<td></td>
</tr>
<tr>
<td>PA, IVS</td>
<td>746.01</td>
<td>746.00</td>
<td></td>
</tr>
<tr>
<td>PA, VSD (TOF)</td>
<td>--</td>
<td>747.31</td>
<td></td>
</tr>
<tr>
<td>TOF</td>
<td>745.2</td>
<td>745.20 - 21</td>
<td></td>
</tr>
</tbody>
</table>

Additional Information:
Pulmonary valve atresia or stenosis may occur with or without a coexisting ventricular septal defect. For pulmonary valve atresia without a VSD (intact ventricular septum), the CDC/BPA code 746.00 (‘atresia,
hypoplasia of pulmonary valve”) is used, corresponding to the ICD-9-CM code 746.01. In CDC/BPA, 746.01 refers to pulmonary valve stenosis.

Pulmonary atresia with a VSD is similar to severe forms of Tetralogy of Fallot, and is included in Tetralogy of Fallot for surveillance (see below). There is no good code depicting valvular pulmonary atresia with VSD; hence in CDC/BPA the code 747.31 (“pulmonary artery atresia with septal defect”) is used.
### Single Ventricle
(Recommended Condition)

#### Description
Instead of two separate ventricles, there is only one morphological ventricle, most commonly a double-inlet left ventricle. This is always a complex heart with several associated heart defects.

#### Inclusions
Single ventricle or common ventricle WITHOUT more specific diagnosis related to hypoplastic ventricle or atroventricular valve (e.g. Hypoplastic left heart syndrome or tricuspid atresia). Forms include double-inlet left ventricle (most common), double inlet right ventricle, single ventricle indeterminate morphology, and other specified type of single ventricle.

#### Exclusions
“Functional” single ventricles, which have 2 ventricles, one of which is very small, so the heart functions as a single ventricle; these are usually due to atresia of one of the atroioventricular valves. Single/common ventricle WITH more specific diagnosis related to hypoplastic ventricle or atroioventricular valves (e.g. hypoplastic left heart syndrome or tricuspid atresia) are excluded from this category but included elsewhere:
- Hypoplastic Left Heart Syndrome (single right ventricle)
- Tricuspid Atresia (single left ventricle)
- Complete atroioventricular canal with malalignment of the AV valves to either the right or left side (creating a single ventricle)
- Some severe forms of DORV (single right ventricle)

#### ICD-9-CM Codes
745.3

#### ICD-10-CM Codes
Q20.4

#### CDC/BPA Codes
745.3

#### Diagnostic Methods
Single ventricle is conclusively diagnosed through direct visualization of the heart by cardiac echo (echocardiography), catheterization, surgery, or autopsy.

#### Prenatal Diagnoses Not Confirmed Postnatally
These conditions may be included as cases when only diagnosed prenatally. However, if it is possible to ascertain the degree of certainty of the prenatal diagnosis, this should factor into the decision as to whether or not to include an individual case in the surveillance data or which category to include the case in. Live-born children who survive should always have confirmation of the defect postnatally.

#### Additional Information
These are very difficult hearts to code and categorize, as they often have many different descriptions. Forms include double-inlet left ventricle (most common), double inlet right ventricle, single ventricle indeterminate morphology, and other specified type of single ventricle. Other associated heart defects may include transposed/malposed great vessels, pulmonary stenosis, coarctation of aorta, and rudimentary outlet chambers (the tiny second ventricle).
# Tetralogy of Fallot (TOF)

## (Core Condition)

### Description

The simultaneous presence of a ventricular septal defect (VSD), pulmonic and subpulmonic stenosis, a malpositioned aorta that overrides the ventricular septum, and right ventricular hypertrophy.

### Inclusions

- Pentalogy of Fallot – Tetralogy of Fallot with an associated inter-atrial communication, either a patent foramen ovale (PFO) or an atrial septal defect (ASD).
- Tetralogy of Fallot (TOF)
- Pulmonary atresia with VSD (see ‘Additional information’)

### Exclusions

Simultaneous occurrence of a VSD and pulmonary stenosis that has TOF physiology but has not been diagnosed as Tetralogy of Fallot. Also, some coding systems may also include Trilogy of Fallot, or Fallot’s Triad – the simultaneous presence of an atrial septal defect, pulmonic stenosis, and right ventricular hypertrophy. This is not to be included as TOF.

### ICD-9-CM Codes

- 745.2

### ICD-10-CM Codes

- Q21.3

### CDC/BPA Codes

- 745.20 – 745.21, 747.31
- (Note: code 746.84 (trilogy of Fallot) has been removed)

### Diagnostic Methods

While Tetralogy of Fallot may be suspected by clinical presentation, it may be conclusively diagnosed only through direct visualization of the heart by cardiac echo (echocardiography), catheterization, surgery, or autopsy.

### Prenatal Diagnoses Not Confirmed Postnataally

These conditions may be included as cases when only diagnosed prenatally. However, if it is possible to ascertain the degree of certainty of the prenatal diagnosis, this should factor into the decision as to whether or not to include an individual case in the surveillance data. Live-born children who survive should always have confirmation of the defect postnatally.

### Additional Information:

Children with Tetralogy of Fallot may experience episodes of cyanosis or hypoxia that result from shunting of unoxygenated blood across the VSD from the right to the left ventricle. Children who have a coexisting VSD and pulmonary stenosis, but do not have Tetralogy of Fallot, may experience similar episodes. Thus, the occurrence of cyanosis or hypoxia does not necessarily mean a child has been diagnosed with Tetralogy of Fallot.

Tetralogy of Fallot is one of several abnormalities of the outflow tract of the heart known as conotruncal...
defects. Some infants (approximately 1 in 7) with these defects have a deletion on the short arm of chromosome 22 (deletion 22q11.2). This deletion is diagnosed using fluorescent in situ hybridization (FISH) and will not necessarily be detected on a routine karyotype analysis.

Tetralogy of Fallot is on a spectrum with other defects having important physiological and coding differences among systems as seen here in the table.

<table>
<thead>
<tr>
<th>CCHD</th>
<th>ICD-9</th>
<th>CDC/BPA</th>
</tr>
</thead>
<tbody>
<tr>
<td>PVS</td>
<td>746.02</td>
<td>746.01</td>
</tr>
<tr>
<td>PA, IVS</td>
<td>746.01</td>
<td>746.00</td>
</tr>
<tr>
<td>PA, VSD (TOF)</td>
<td>--</td>
<td>747.31</td>
</tr>
<tr>
<td>TOF</td>
<td>745.2</td>
<td>745.20 - 21</td>
</tr>
</tbody>
</table>

Pulmonary atresia with a VSD is similar to severe forms of Tetralogy of Fallot and is included here for surveillance. There is no good code depicting valvular pulmonary atresia with VSD; hence in CDC/BPA the code 747.31 (“pulmonary artery atresia with septal defect”) is used. For pulmonary valvular atresia without a VSD (intact ventricular septum), the code 746.00 (“atresia, hypoplasia of pulmonary valve”) is used – see separate section on Pulmonary valve atresia/stenosis.

When pulmonary valve atresia occurs with a VSD, the child may experience episodes of cyanosis or hypoxia similar to those seen in children with Tetralogy of Fallot. This results from shunting of unoxygenated blood across the VSD from the right to the left ventricle. Thus, the occurrence of cyanosis or hypoxia does not necessarily mean that the child has Tetralogy of Fallot.
## Total Anomalous Pulmonary Venous Connection (TAPVC)

(Core Condition)

### Description

A condition in which all 4 pulmonary veins connect anomalously into the systemic venous circulation to the right atrium or the body (systemic veins) instead of the left atrium; often occurs with other cardiac defects.

### Inclusions

- TAPVC (total anomalous pulmonary venous connection)
- TAPVR (total anomalous pulmonary venous return)
- TAPVD (total anomalous pulmonary venous drainage)

### Exclusions

If not all 4 veins are visibly connecting/draining anomalously (e.g. Partial Anomalous Venous Return, ICD-9-CM code 747.42 or CDC/BPA code 747.41 or Q26.3)

### ICD-9-CM Codes

- 747.41

### ICD-10-CM Codes

- Q26.2

### CDC/BPA Codes

- 747.42

### Diagnostic Methods

While TAPVR may be suspected by clinical presentation, it may be conclusively diagnosed only through direct visualization of the heart by cardiac echo (echocardiography), catheterization, surgery, or autopsy. The difficulty in viewing all 4 veins may mean that several echocardiograms may be needed to confirm the diagnosis.

### Prenatal Diagnoses Not Confirmed Postnatally

TAPVR is difficult to identify prenatally. If identified by prenatal ultrasound, it should not be included in surveillance data without postnatal confirmation. In addition, the absence of TAPVR on prenatal ultrasound does not necessarily mean that it will not be diagnosed after delivery.

### Additional Information:

Total anomalous pulmonary venous return and partial anomalous pulmonary venous return have not been shown to be developmentally related, although they share a similar description. Also, there are subtle differences in the meaning of anomalous venous connection, return, and drainage, but the terms are often used interchangeably.
Transposition of the Great Arteries (TGA)  
(Core Condition)

**Description**
Transposition of the aorta and the pulmonary artery such that the aorta arises from the right ventricle (instead of the left) and the pulmonary artery arises from the left ventricle (instead of the right).

**Inclusions**
- Complete or “dextro” transposition (d-TGA without a VSD)
- Corrected, or “levo” transposition (l-TGA) (but exclude for CCHD screening)
- Incomplete transposition (d-TGA with a VSD)
- Transposition of the Great Arteries (TGA), not otherwise specified
- Transposition of the Great Vessels (TGV)

**Exclusions**
Cases with codes for both DORV and TGA are counted in the DORV category. DORV subtype with malposed/”transposed” great arteries (CDC/BPA 745.14 are also counted in the DORV category, along with 745.13, and 745.15.

**ICD-9-CM Codes**
For CCHD Screening: 745.10, 745.12, 745.19

**ICD-10-CM Codes**
For CCHD Screening: Q20.3, Q20.5

**CDC/BPA Codes**
For CCHD Screening: 745.10 – 745.12, 745.18 – 745.19

**Diagnostic Methods**
d-TGA is conclusively diagnosed through direct visualization of the heart by cardiac echo (echocardiography), catheterization, surgery, or autopsy.

**Prenatal Diagnoses Not Confirmed Postnatally**
These conditions may be included as cases when only diagnosed prenatally. However, if it is possible to ascertain the degree of certainty of the prenatal diagnosis, this should factor into the decision as to whether or not to include an individual case in the surveillance data. Live-born children who survive should always have confirmation of the defect postnatally.

**Additional Information:**
In order for a child with d-TGA to survive, a communication must be present between the pulmonary and systemic circulations to allow oxygenated blood from the lungs to reach the right ventricle for distribution to the rest of the body through the abnormally placed aorta. In most instances, this communication is through a ventricular septal defect (incomplete TGA). If a VSD is not present, oxygenated blood from the
lungs is returned directly to the lungs without being distributed to the rest of the body (complete TGA).

If the defect coding system does not include unique codes to differentiate TGA with and without a VSD (complete vs. incomplete), the VSD should be coded separately when present.

L-TGA (corrected transposition or “levo” transposition) is a defect in which the ventricle on the right side of the heart has the anatomic appearance of the left ventricle, and the ventricle on the left side of the heart has the anatomic appearance of the right ventricle (ventricular inversion). The pulmonary artery arises from the anatomic left ventricle and the aorta arises from the anatomic right ventricle (hence the designation of transposition). Because blood from the ventricle on the right flows through the pulmonary artery, and that from the ventricle on the left flows through the aorta, circulation is normal as long as there are no other defects.

Transposition of the great arteries is one of several abnormalities of the outflow tract of the heart known as conotruncal defects. Very few infants with these defects have a deletion on the short arm of chromosome 22 (deletion 22q11.2). This deletion is diagnosed using fluorescent *in situ* hybridization (FISH) and will not necessarily be detected on a routine karyotype analysis.
| **Tricuspid Valve Atresia and Stenosis**  
<table>
<thead>
<tr>
<th>(Recommended Condition)</th>
</tr>
</thead>
</table>
| **Description** | Tricuspid valve atresia – Lack of patency, or failure of formation altogether, of the tricuspid valve, resulting in obstruction of blood flow from the right atrium to the right ventricle.  
Tricuspid valve stenosis – Obstruction or narrowing of the tricuspid valve, which may impair blood flow from the right atrium to the right ventricle. |
| **Inclusions** | Tricuspid atresia  
Tricuspid stenosis |
| **Exclusions** | Tricuspid regurgitation without specific mention of tricuspid atresia or stenosis. |
| **ICD-9-CM Codes** | 746.1 |
| **ICD-10-CM Codes** | Q22.4 |
| **CDC/BPA Codes** | 746.100 (tricuspid atresia), 746.106 (tricuspid stenosis) (excluding 746.105 – tricuspid insufficiency), |
| **For CCHD Screening** | 746.100 only  
Note: Only the tricuspid atresia is a CCHD. Many cases of tricuspid stenosis are not critical. |
| **Diagnostic Methods** | While tricuspid valve atresia or stenosis may be suspected by clinical presentation, it may be conclusively diagnosed only through direct visualization of the heart by cardiac echo (echocardiography), catheterization, surgery, or autopsy. |
| **Prenatal Diagnoses Not Confirmed Postnatally** | While these conditions may be identified by prenatal ultrasound, they should not be included in surveillance data without postnatal confirmation. In addition, the absence of tricuspid valve atresia or stenosis on prenatal ultrasound does not necessarily mean that it will not be diagnosed after delivery. |
| **Additional Information** | NA |
### Ventricular Septal Defect (VSD) (Recommended Condition)

<table>
<thead>
<tr>
<th>Description</th>
<th>An opening in the wall (septum) that separates the left and right ventricles of the heart.</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>Inclusions</strong></td>
<td>Ventricular septal defect VSD</td>
</tr>
<tr>
<td><strong>Exclusions</strong></td>
<td>Ventricular septal defects that occur as part of Tetralogy of Fallot or an atrioventricular septal defect. Inflow-type, subtricuspid, and canal-type VSDs are assumed to be part of an atrioventricular septal defect and should not be coded separately</td>
</tr>
<tr>
<td><strong>ICD-9-CM Codes</strong></td>
<td>745.4</td>
</tr>
<tr>
<td><strong>ICD-10-CM Codes</strong></td>
<td>Q21.0</td>
</tr>
<tr>
<td><strong>CDC/BPA Codes</strong></td>
<td>745.40 – 745.49 (excluding 745.487 (inlet VSD in AVSD category), 745.498 (possible VSD))</td>
</tr>
<tr>
<td><strong>Diagnostic Methods</strong></td>
<td>Some isolated VSDs may be diagnosed on physical examination and/or EKG without direct imaging of the heart. However, many VSDs may be conclusively diagnosed only through direct visualization of the heart by cardiac echo (echocardiography), catheterization, surgery, or autopsy.</td>
</tr>
<tr>
<td><strong>Prenatal Diagnoses Not Confirmed Postnatally</strong></td>
<td>While VSDs may be identified by prenatal ultrasound, many close spontaneously before delivery. For this reason, VSDs that are diagnosed prenatally should not be included unless they have been confirmed postnatally. In addition, the absence of a VSD on prenatal ultrasound does not necessarily mean that a VSD will not be diagnosed after delivery, as it is not always possible to accurately visualize the entire ventricular septum by prenatal ultrasound.</td>
</tr>
</tbody>
</table>

**Additional Information:**
VSDs may be of several types, depending on the location of the opening along the ventricular septum. The most common are: Muscular, Membranous, Perimembranous.

However, in many instances the type of VSD may not be specified in the medical record. Many muscular, membranous and perimembranous VSDs may close spontaneously in the first weeks or months of life without treatment. An aneurysm of the ventricular septum indicates a membranous or perimembranous VSD that is in the process of closing.
### Orofacial

#### Choanal Atresia

**(Recommended Condition)**

<table>
<thead>
<tr>
<th>Description</th>
<th>Congenital obstruction of the opening of the nasal cavity into the nasopharynx on either side. This prevents communication of the nasal cavity with the pharynx.</th>
</tr>
</thead>
</table>
| Inclusions | Choanal atresia, type not specified  
Choanal stenosis  
Membranous choanal atresia, with or without a bony rim  
Completely bony choanal atresia |
| Exclusions | NA |
| ICD-9-CM Codes | 748.0 |
| ICD-10-CM Codes | Q30.0 |
| CDC/BPA Codes | 748.00 |

**Diagnostic Methods:**

- Bilateral choanal atresia is usually easily recognized at birth from the clinical presentation of obligate mouth-breathing. Unilateral choanal atresia may be suspected by clinical examination. Both conditions may be diagnosed by the inability to pass a feeding tube from the nasal passage(s) into the posterior pharynx. Both conditions may also be seen on CT or MRI scan, at surgery or autopsy.

**Prenatal Diagnoses Not Confirmed Postnatally:**

While these conditions may be identified by prenatal ultrasound, they should not be included in birth defects surveillance data without postnatal confirmation. In addition, the absence of choanal atresia on prenatal ultrasound does not necessarily mean that it will not be diagnosed after delivery.

**Additional Information:**

Choanal atresia or stenosis may be unilateral or bilateral. If the defect coding system includes unique codes for these different types, the location should be coded.

Choanal atresia is one of the defects reported as part of the CHARGE association, which may also include colobomas, heart defects, retarded growth and development, genital hypoplasia, and ear anomalies and/or deafness.
Cleft Lip Alone (without Cleft Palate)  
(Core Condition)

**Description**
A defect in the upper lip resulting from incomplete fusion of the parts of the lip.

**Inclusions**
- Complete cleft lip – The defect extends through the entire lip into the floor of the nose.
- Incomplete cleft lip – The defect extends through part of the lip but not into the floor of the nose.
- Cheiloschisis

**Exclusions**
- Pseudocleft lip – An abnormal linear thickening, depressed groove, or scar-like pigmented change on the skin of the lip without an actual cleft.
- Oblique facial clefts
- Cleft palate without an associated cleft lip

**ICD-9-CM Codes**
- 749.1

**ICD-10-CM Codes**
- Q36.0 – Q36.9

**CDC/BPA Codes**
- 749.10-749.19

**Diagnostic Methods**
Cleft lip is usually easily recognized on physical examination after delivery. It may also be seen on CT or MRI scan, at surgery or autopsy; plastic surgery consultation reports are often useful.

**Prenatal Diagnoses Not Confirmed Postnatally**
While this condition may be identified by prenatal ultrasound, it should not be included in birth defects surveillance data without postnatal confirmation. In addition, the absence of cleft lip on prenatal ultrasound does not necessarily mean that it will not be diagnosed after delivery.

**Additional Information:**
Cleft lip may be unilateral, bilateral, or central in location, or not otherwise specified, as well as incomplete and complete. If the defect coding system includes unique codes for these different types, the location of the cleft should be coded.
# Cleft Lip with Cleft Palate

## Description
A defect in the upper lip resulting from incomplete fusion of the parts of the lip, with an opening in the roof of the mouth.

## Inclusions
- Cleft lip with cleft of the hard and soft palate
- Cleft lip with cleft of the hard palate
- Cleft lip with cleft of the soft palate
- Cleft lip with cleft palate, not otherwise specified
- Cheilopalatoschisis

## Exclusions
- Pseudocleft lip with cleft palate – An abnormal linear thickening, depressed groove, or scar-like pigmented change on the skin of the lip without an actual cleft.
- Oblique facial clefts with cleft palate
- Cleft palate without an associated cleft lip
- Cleft lip without an associated cleft palate

## ICD-9-CM Codes
749.20 - 749.25 (only these combined cleft palate with cleft lip codes should be used, not cleft lip or cleft palate codes individually)

## ICD-10-CM Codes
Q37.0 – Q37.9 (only these combined cleft palate with cleft lip codes should be used, not cleft lip or cleft palate codes individually)

## CDC/BPA Codes
749.20 – 749.29 (only these combined cleft lip with cleft palate codes should be used, not cleft lip or cleft palate codes individually)

## Diagnostic Methods
Cleft lip is usually easily recognized on physical examination after delivery. It may also be seen on CT or MRI scan, at surgery or autopsy; plastic surgery consultation reports are often useful.

## Prenatal Diagnoses Not Confirmed Postnatally
While this condition may be identified by prenatal ultrasound, it should not be included in birth defects surveillance data without postnatal confirmation. In addition, the absence of cleft lip on prenatal ultrasound does not necessarily mean that it will not be diagnosed after delivery.

## Additional Information:
Cleft lip with cleft palate may be unilateral, bilateral, or central in location, or not otherwise specified. If the defect coding system includes unique codes for these different types, the location of the cleft should be coded.
Cleft Palate Alone (without Cleft Lip)
(Core Condition)

**Description**
An opening in the roof of the mouth resulting from incomplete fusion of the shelves of the palate. The opening may involve the hard palate only, the soft palate only, or both.

**Inclusions**
- Bifid or cleft uvula
- Cleft palate, type not specified
- Cleft hard palate
- Cleft soft palate
- Submucous cleft palate – A cleft in the soft palate that is covered by the mucosa or a thin muscle layer.

**Exclusions**
Cleft palate that coexists with a cleft lip. These should be coded as cleft lip with cleft palate (see above).

**ICD-9-CM Codes**
749.0

**ICD-10-CM Codes**
Q35.1 – Q35.9

**CDC/BPA Codes**
749.00 – 749.09

**Diagnostic Methods**
Cleft palate is usually recognized on physical examination by direct visualization of the pharynx after delivery. It may also be seen on CT or MRI scan, at surgery or autopsy; plastic surgery consultation reports are often useful. However, submucous cleft palate and bifid uvula may be difficult to diagnose by physical examination during the first year of life.

**Prenatal Diagnoses Not Confirmed Postnatally**
This condition should not be included in birth defects surveillance data without postnatal confirmation.

**Additional Information:**
Cleft palate may be unilateral, bilateral, or central in location. If the defect coding system includes unique codes for these different types, the location of the cleft should be coded. Cleft palate sometimes may be described as U-shaped or V-shaped. This distinction is not clinically meaningful and these conditions should not be coded differently.

Bifid uvula is often seen in association with a submucous cleft palate. However, bifid uvula also may occur alone. The presence of submucous cleft palate does not necessarily mean that a bifid uvula is present. Cleft palate is one component of the Pierre Robin sequence, which also includes micrognathia and glossoptosis (when the tongue falls backward into the posterior pharynx). When diagnosed, Pierre Robin sequence should be coded separately.
### Gastrointestinal

#### Biliary Atresia
(Extended Condition)

<table>
<thead>
<tr>
<th>Description</th>
<th>Congenital absence of the lumen of the extrahepatic bile ducts.</th>
</tr>
</thead>
<tbody>
<tr>
<td>Inclusions</td>
<td>Agenesis, absence, hypoplasia, obstruction or stricture of the bile duct(s)</td>
</tr>
<tr>
<td>Exclusions</td>
<td>Congenital or neonatal hepatitis&lt;br&gt;Intrahepatic biliary atresia (absence or paucity of bile ducts within the liver) not associated with extrahepatic biliary atresia</td>
</tr>
<tr>
<td>ICD-9-CM Codes</td>
<td>751.61</td>
</tr>
<tr>
<td>ICD-10-CM Codes</td>
<td>Q44.2 - Q44.3</td>
</tr>
<tr>
<td>CDC/BPA Codes</td>
<td>751.65</td>
</tr>
<tr>
<td>Diagnostic Methods</td>
<td>Biliary atresia may be suspected by the clinical presentation and the presence of elevated direct bilirubin and liver function tests. However, it may be conclusively diagnosed only through direct assessment of the bile ducts by abdominal ultrasound, CT or MRI scan, biliary excretion study (HIDA scan), surgery, or autopsy.</td>
</tr>
<tr>
<td>Prenatal Diagnoses Not Confirmed Postnatally</td>
<td>While biliary atresia may be suspected by prenatal ultrasound, it should not be included in surveillance data without postnatal confirmation. In addition, the absence of biliary atresia on prenatal ultrasound does not necessarily mean that it will not be diagnosed after delivery.</td>
</tr>
</tbody>
</table>

**Additional Information:**
The liver contains within its substance intrahepatic bile ducts and passages that join and coalesce to form two main ducts that carry bile out of the liver.

The extrahepatic bile ducts include the hepatic duct (formed by the two main ducts that carry bile out of the liver), the cystic duct (which carries bile out of the gallbladder where it is stored), and the common bile duct (formed by the junction of the hepatic duct and the cystic duct), which carries bile into the duodenum for excretion.

When extrahepatic biliary atresia is present, the intrahepatic bile ducts may also be abnormal or atretic.

Patients with biliary atresia may have jaundice due to direct hyperbilirubinemia, which is not treated with phototherapy. The more common type of neonatal jaundice due to indirect hyperbilirubinemia may be treated with phototherapy and does not indicate the presence of biliary atresia.
Esophageal Atresia/Tracheoesophageal Fistula
(Recommended Condition)

**Description**
Esophageal atresia – A condition in which the esophagus ends in a blind pouch and fails to connect with the stomach.

Tracheoesophageal fistula – An abnormal communication between the esophagus and the trachea. This is almost always associated with some form of esophageal atresia.

**Inclusions**
- Esophageal atresia alone
- Esophageal atresia with tracheoesophageal (TE) fistula
- Esophageal stenosis, stricture, ring, or web
- TE fistula
- Tracheoesophageal fistula, all types

**Exclusions**
- Tracheal atresia
- Tracheoesophageal cleft

**ICD-9-CM Codes**
750.3

**ICD-10-CM Codes**
Q39.0 – Q39.4

**CDC/BPA Codes**
750.30 – 750.35

**Diagnostic Methods**
The diagnosis may be suspected by the clinical presentation of polyhydramnios, vomiting, or respiratory distress. Esophageal atresia may be diagnosed by x-ray documentation of failure of a feeding tube to pass from the pharynx into the stomach. Tracheoesophageal atresia may be conclusively diagnosed only by CT or MRI scan, surgery, or autopsy.

**Prenatal Diagnoses Not Confirmed Postnatally**
These conditions may be included when only diagnosed prenatally. However, if it is possible to ascertain the degree of certainty of the prenatal diagnosis, this should factor into the decision as to whether or not to include an individual case in the surveillance data. Live-born children who survive should always have confirmation of the defect postnatally before being included.

**Additional Information:**
In some instances, TE fistula without esophageal atresia may not be diagnosed until weeks, months, or even a year or more after birth if the communication between the esophagus and stomach remains patent.

TE fistula is one of the defects reported as part of the VATER, or VACTERL, association, which may also include vertebral and cardiac defects, anal atresia, renal defects, and limb anomalies.
## Rectal and Large Intestinal Atresia/Stenosis

(Recommended Condition)

<table>
<thead>
<tr>
<th>Description</th>
<th>Complete or partial occlusion of the lumen of one or more segments of the large intestine and/or rectum.</th>
</tr>
</thead>
</table>
| Inclusions  | Anal atresia or stenosis  
Colonic atresia or stenosis  
Imperforate anus  
Large intestinal atresia or stenosis  
Rectal atresia or stenosis |
| Exclusions  | Apple peel intestinal atresia  
Duodenal atresia or stenosis  
Ileal atresia or stenosis  
Jejunal atresia or stenosis  
Small intestinal atresia or stenosis |
| ICD-9-CM Codes | 751.2 |
| ICD-10-CM Codes | Q42.0 – Q42.9 |
| CDC/BPA Codes | 751.20 – 751.24 |
| Diagnostic Methods | Anal atresia (imperforate anus) is usually easily recognized at birth by physical examination. While large intestinal and rectal atresia or stenosis may be suspected by the clinical presentation of failure to pass meconium or stool, they may be conclusively diagnosed only through direct imaging of the bowel by x-ray, barium enema, surgery, or autopsy. |
| Prenatal Diagnoses Not Confirmed Postnataally | While these conditions may be identified by prenatal ultrasound, they should not be included in surveillance data without postnatal confirmation. In addition, the absence of intestinal, rectal or anal atresia or stenosis on prenatal ultrasound does not necessarily mean that it will not be diagnosed after delivery. |

**Additional Information:**

These conditions may occur with or without a fistula.

Anal atresia is one of the defects reported as part of the VATER, or VACTERL, association, which may also include vertebral and cardiac defects, TE fistula, renal defects, and limb anomalies.
### Small Intestinal Atresia/Stenosis

(Recommended Condition)

| **Description** | Complete or partial occlusion of the lumen of one or more segments of the small intestine. Small intestinal atresias are often assigned a type descriptor in the surgical or autopsy report, depending upon the severity of the atresia (types include I, II, IIIA, IIIB, and VI). |
| **Inclusions** | Duodenal atresia or stenosis (also include duodenal web, membrane, diaphragm, or windsock); include all types: I, II, IIIA, IIIB, VI, and not stated. 
Jejunal atresia or stenosis (also include jejunal web or membrane); include all types: I, II, IIIA, IIIB, VI, and not stated. 
Ileal atresia or stenosis also (include ileal web or membrane); include all types: I, II, IIIA, IIIB, VI, and not stated. 
Small intestinal atresia or stenosis, not otherwise specified; include all types: I, II, IIIA, IIIB, VI, and not stated. |
| **Exclusions** | Intestinal atresia/stenosis in an infant with cystic fibrosis. 
Sirenomelia. 
Anal atresia or stenosis. 
Anal stenosis, anteriorly displaced anus. 
Colonic atresia or stenosis. 
Imperforate anus. 
Large intestinal atresia or stenosis. 
Rectal atresia or stenosis. |
| **ICD-9-CM Codes** | 751.1 |
| **ICD-10-CM Codes** | Q41.0 – Q41.9 |
| **CDC/BPA Codes** | 751.10-751.19 |
| **Diagnostic Methods** | While the diagnosis may be suspected by clinical presentation of abdominal distension, vomiting, lack of passage of meconium, “double bubble” sign on abdominal ultrasound, dilated loops of bowel on abdominal x-ray, or failure of contrast to advance on upper GI or barium enema studies, small intestinal atresia or stenosis requires conclusive diagnosis through surgery or autopsy. |
| **Prenatal Diagnoses Not Confirmed Postnatally** | While these conditions may be suspected by prenatal ultrasound, they should not be included in surveillance data without postnatal confirmation; postnatal diagnosis of the small intestinal atresia or stenosis requires a surgical or autopsy report (i.e., ultrasound or abdominal x-ray studies, such as an upper GI or barium enema, are not sufficient). In addition, the absence of small intestinal atresia or stenosis on prenatal ultrasound does not necessarily mean that it will not be diagnosed after delivery. |

**Additional Information:**
If contiguous regions of the small intestine are involved, a compound descriptor may be used, e.g., jejunooileal atresia; codes for both affected areas should be included, but the descriptor should indicate whether these are contiguous or non-contiguous regions. One-third of all infants with duodenal atresia or stenosis have Down syndrome.
### Genitourinary

#### Bladder Exstrophy

(Recommended Condition)

<table>
<thead>
<tr>
<th>Description</th>
<th>A defect in the lower abdominal wall and anterior wall of the bladder through which the lining of the bladder is exposed to the outside.</th>
</tr>
</thead>
</table>
| Inclusions  | Classic bladder exstrophy  
Ectopia vesicae  
Epispadias-exstrophy complex  
Extroversion of the bladder  
Variants of bladder exstrophy  
Vesical exstrophy |
| Exclusions  | Ambiguous genitalia without mention of bladder exstrophy  
Cloacal exstrophy  
Isolated epispadias |
| ICD-9-CM Codes | 753.5 |
| ICD-10-CM Codes | Q64.10, Q64.19 |
| CDC/BPA Codes | 753.50 |
| Diagnostic Methods | Bladder exstrophy is easily recognized on physical examination at delivery. However, the exact nature of the defect and associated anomalies may only be distinguished by abdominal ultrasound, contrast x-ray studies, CT or MRI scan, surgery, or autopsy. |
| Prenatal Diagnoses Not Confirmed Postnatally | These conditions may be included when only diagnosed prenatally. However, if it is possible to ascertain the degree of certainty of the prenatal diagnosis, this should factor into the decision as to whether or not to include an individual case in the surveillance data, as it may be difficult to distinguish bladder exstrophy from cloacal exstrophy. Live-born children who survive should always have confirmation of the defect postnatally before being included. |

**Additional Information:**

In the classic form of bladder exstrophy, the entire urinary tract is open anteriorly from the urethral meatus to the umbilicus. The pubic bones are widely separated, as are the abdominal muscles and fascia. There is eversion/exposure of the posterior bladder wall. The genitalia of either gender may be involved and may be bifid or duplicated. The classic form of bladder exstrophy occurs more frequently in males.

Variants of bladder exstrophy occur more rarely and affect females more often than males. Included among these variants are superior vesical fistula, closed exstrophy, duplicate exstrophy, pseudoexstrophy, inferior vesicle. Epispadias is almost uniformly present, but should not be coded separately.

Ambiguous genitalia may be noted in patients with bladder exstrophy if an obvious scrotum and testes are not present. However, ambiguous genitalia should not be coded as a separate defect in these instances.
Bladder exstrophy should be distinguished from cloacal exstrophy, in which the urinary, intestinal, and genital structures open into a common cavity (the cloaca). The distinction may only be possible with detailed diagnostic studies, surgery, or at autopsy. In cloacal exstrophy, bladder exstrophy and imperforate anus are also present. In bladder exstrophy without cloacal exstrophy, the anus is patent. When both bladder and cloacal exstrophy are present, only cloacal exstrophy should be coded.
**Cloacal Exstrophy**
(Recommended Condition)

**Description**
Congenital persistence of a common cloacal cavity into which gut, urethra, and reproductive tracts open with exstrophy of the cavity: usually accompanied by a low omphalocele, imperforate anus, and a (closed) neural tube defect.

**Inclusions**
cloacal exstrophy
OEIS complex (Omphalocele, bladder Exstrophy, Imperforate anus, Spinal defects)

**Exclusions**
persistent cloaca (urorectal septum malformation sequence)
bladder exstrophy without omphalocele/imperforate anus

**ICD-9-CM Codes**
751.5

**ICD-10-CM Codes**
Q64.12

**CDC/BPA Codes**
751.555

**Diagnostic Methods**
Cloacal exstrophy is easily recognized on physical examination at delivery. However, the exact nature of the defect and associated anomalies may only be distinguished by abdominal ultrasound, contrast x-ray studies, CT or MRI scan, surgery, or autopsy.

**Prenatal Diagnoses Not Confirmed Postnatally**
This condition may be included when only diagnosed prenatally. However, if it is possible to ascertain the degree of certainty of the prenatal diagnosis, this should factor into the decision as to whether or not to include an individual case in the surveillance data, as it may be difficult to distinguish cloacal exstrophy from bladder exstrophy. Live-born children who survive should always have confirmation of the defect postnatally before being included. At a minimum, omphalocele with bladder exstrophy (which cannot be distinguished from cloacal exstrophy prenatally) must be evident.

**Additional Information:**
In the classic form of cloacal exstrophy, the entire bladder is open anteriorly from the urethral meatus to the low placed omphalocele. The pubic bones are widely separated, as are the abdominal muscles and fascia. The genitalia of either gender may be involved and may be bifid or duplicated.

Ambiguous genitalia may be noted in patients with cloacal exstrophy if an obvious scrotum and testes are not present. However, ambiguous genitalia should not be coded as a separate defect in these instances.

Cloacal exstrophy should be distinguished from bladder exstrophy. The distinction may only be possible with detailed diagnostic studies, surgery, or at autopsy. In cloacal exstrophy, bladder exstrophy and imperforate anus are also present. When both bladder and cloacal exstrophy are present, only cloacal exstrophy should be coded.
### Congenital Posterior Urethral Valves
*(Recommended Condition)*

| **Description** | Posterior urethral valves (PUV) are tissue folds of the posterior urethra and function as valves obstructing urine outflow. Congenital PUV is an abnormal congenital obstructing membrane that is located within the posterior male urethra; this valve is the most common cause of bladder outlet obstruction in male children. Congenital PUV can also be found in virilized females and rarely in normal females. Obstruction could vary from mild to severe. |
| **Inclusions** | Posterior urethral valves |
| **Exclusions** | Inhibition of urinary flow at any of the above sites resulting solely from neurologic impairment. |
| **ICD-9-CM Codes** | 753.6 |
| **ICD-10-CM Codes** | Q64.2 |
| **CDC/BPA Codes** | 753.60 |
| **Diagnostic Methods** | Congenital PUV may be suspected by the clinical presentation. Newborns can present at birth with abdominal masses, distended bladder, hydronephrosis, or with respiratory distress, oligohydramnions, and Potter facies. However, the exact nature of the defect and PUV may only be distinguished by direct visualization such as cystoscopy or urethral endoscopy, or with contrast studies such as voiding cystourethrogram (VCUG). With routine obstetric ultrasonography the prenatal diagnosis of PUV is becoming increasingly common. PUV also may be diagnosed at surgery or autopsy. |
| **Prenatal Diagnoses Not Confirmed Postnatally** | While obstructive genitourinary defects including congenital PUV may be identified by prenatal ultrasound, many lesions diminish or resolve spontaneously prior to birth. For this reason, PUV should not be included in surveillance data without postnatal confirmation. In addition, the absence of genitourinary obstruction on prenatal ultrasound does not necessarily mean that an obstructive defect such as PUV will not be diagnosed after delivery. |

**Additional Information:**
When urine flow is obstructed, the portion of the genitourinary tract proximal to the affected area may become enlarged and dilated with urine. Mild lesions may produce only partial or intermittent urinary obstruction without permanent damage. More severe lesions may substantially or completely obstruct urine flow, resulting in permanent damage to proximal structures, and sometimes impaired kidney function, if not relieved by surgery.
### Hypospadias
(Recommended Condition)

<table>
<thead>
<tr>
<th>Description</th>
<th>Hypospadias – Displacement of the opening of the urethra (urethral meatus) ventrally and proximally (underneath and closer to the body) in relation to the tip of the glans of the penis.</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>Inclusions</strong></td>
<td>First-degree hypospadias – The urethral meatus is located on the glans of the penis. Also called primary, 1°, glandular, or coronal hypospadias.</td>
</tr>
<tr>
<td></td>
<td>Second-degree hypospadias – The urethral meatus is located on the shaft of the penis. Also called secondary, 2°, or penile hypospadias.</td>
</tr>
<tr>
<td></td>
<td>Third-degree hypospadias – The urethral meatus is located at the base of the penis on the scrotum or perineum. Also called tertiary, 3°, scrotal, penoscrotal, or perineal hypospadias.</td>
</tr>
<tr>
<td></td>
<td>Hypospadias, degree not specified</td>
</tr>
<tr>
<td></td>
<td>Hypospadias of any type with chordee</td>
</tr>
<tr>
<td><strong>Exclusions</strong></td>
<td>Chordee alone without associated hypospadias</td>
</tr>
<tr>
<td></td>
<td>Ambiguous genitalia</td>
</tr>
<tr>
<td></td>
<td>Epispadias</td>
</tr>
<tr>
<td><strong>ICD-9-CM Codes</strong></td>
<td>Hypospadias 752.61</td>
</tr>
<tr>
<td><strong>ICD-10-CM Codes</strong></td>
<td>Q54.0 – Q54.9 (excluding Q54.4)</td>
</tr>
<tr>
<td><strong>CDC/BPA Codes</strong></td>
<td>Hypospadias 752.60 – 752.62 (excluding 752.61 and 752.621)</td>
</tr>
<tr>
<td><strong>Diagnostic Methods</strong></td>
<td>Hypospadias is usually easily recognized on physical examination at delivery. They may also be seen on contrast x-rays of the urinary tract, at surgery or autopsy.</td>
</tr>
<tr>
<td><strong>Prenatal Diagnoses Not Confirmed Postnatally</strong></td>
<td>While this condition may be diagnosed by prenatal ultrasound, it should not be included in surveillance data without postnatal confirmation. In addition, the absence of hypospadias on prenatal ultrasound does not necessarily mean that they will not be diagnosed after delivery.</td>
</tr>
<tr>
<td><strong>Additional Information:</strong></td>
<td>Chordee indicates a ventral (downward) curve of the penis, which may result from cutaneous or fibrous restriction. It is present in approximately 35% to 50% of cases of hypospadias.</td>
</tr>
</tbody>
</table>
In mild forms of first-degree hypospadias, the foreskin may appear hooded but there may be no overt clinical symptoms.

In contrast, third-degree hypospadias may be described as ambiguous genitalia. In this instance, it is important to search the medical record for detailed information (including chromosome, molecular, and hormone analyses; genetics and endocrinology consultations; surgery or autopsy reports) that may clarify the anatomy and/or indicate whether an underlying genetic condition or endocrinopathy associated with ambiguous genitalia is present. Ambiguous genitalia should not be coded if hypospadias is the only diagnosis. Hypospadias generally should not be coded if a normal female karyotype (46,XX) is reported.
**Renal Agenesis/Hypoplasia**  
*(Recommended Condition)*

| **Description** | Renal agenesis – Complete absence of the kidney
| | Renal hypoplasia – Incomplete development of the kidney |
| **Inclusions** | Renal agenesis, dysgenesis, aplasia, or hypoplasia
| | Potter syndrome secondary to renal agenesis/hypoplasia |
| **Exclusions** | Cystic renal dysplasia
| | Cystic kidney disease
| | Multicystic kidney
| | Multicystic dysplastic kidney
| | Polycystic kidney
| | Renal cysts
| | Renal dysplasia
| | Small kidney |

**ICD-9-CM Codes**  
753.0

**ICD-10-CM Codes**  
Q60.0 – Q60.6

**CDC/BPA Codes**  
753.00 – 753.01

**Diagnostic Methods**  
Bilateral renal agenesis is often suspected on physical examination after delivery because of the Potter phenotype: low-set cartilage-deficient ears, prominent epicanthal folds, flattened “parrot-beaked” nose, recessed chin, limb contractures, malformed hands, and clubbed feet. Bilateral renal hypoplasia may or may not be recognized after delivery, depending on the severity and degree of residual kidney function.

Unilateral renal agenesis or hypoplasia may not be symptomatic at delivery if the contralateral kidney is not impaired.

Each of these diagnoses may be conclusively diagnosed only through direct assessment by abdominal ultrasound, CT or MRI scan, surgery, or autopsy.

**Prenatal Diagnoses Not Confirmed Postnatally**  
Bilateral renal agenesis may be included when only diagnosed prenatally. However, if it is possible to ascertain the degree of certainty of the prenatal diagnosis, this should factor into the decision as to whether or not to include an individual case in the surveillance data. Live-born children who survive should always have confirmation of the defect postnatally before being included.

While bilateral renal hypoplasia and unilateral renal agenesis/hypoplasia may be suspected by prenatal ultrasound, they should not be included in surveillance data without postnatal confirmation. Lack of visualization of a kidney on prenatal ultrasound does not always indicate that the kidney is truly absent.
Additional Information:
Renal agenesis and hypoplasia may be unilateral or bilateral. If the defect coding system includes unique codes for these different types, the location should be coded.

Bilateral renal agenesis, or any condition that significantly impairs the function of both kidneys in utero, may lead to the oligohydramnios sequence (Potter syndrome) due to lack of fetal urine production and the resulting decreased amniotic fluid volume. The sequence includes minor facial dysmorphism (flat face, small chin, large ears), pulmonary hypoplasia, and joint contractures.

Bilateral renal agenesis is incompatible with long-term survival unless a kidney transplant is performed. In contrast, unilateral renal agenesis/hypoplasia may not be diagnosed until weeks, months, or even years after birth if the contralateral kidney function is normal. Some unilateral cases may be diagnosed only as incidental findings during evaluation for other conditions, and some may never be recognized.
Musculoskeletal

Clubfoot
(Recommended Condition)

Description
An abnormality consisting of plantar flexion (downward pointing of the foot and toes), inversion (varus, or internal rotation), and metatarsus adductus (deviation of the forefoot toward the body) of the foot. An abnormally high arch (pes cavus) and midfoot flexion crease usually are also present.

Inclusions
Talipes equinovarus (including congenital, idiopathic, and neurogenic), talipes not otherwise specified, clubfoot not otherwise specified.

Exclusions
Talipes equinovalgus, talipes calcaneovalgus, talipes calcaneovalgus, talipes varus, talipes valgus, vertical talus, metatarsus adductus alone, metatarsus varus alone, pes varus, pes valgus, pes planus, rocker-bottom foot, positional or postural clubfoot.

ICD-9-CM Codes
754.51, 754.70

ICD-10-CM Codes
Q66.0, Q66.89

CDC/BPA Codes
754.50, 754.73 excluding 754.735

Diagnostic Methods
Clubfoot is diagnosed by physical exam. X-rays and imaging studies may provide supplemental information but are not necessary for diagnosis.

Prenatal Diagnoses Not Confirmed Postnatally
Clubfoot can be identified or suspected on prenatal ultrasound; however, it should not be included in birth defects surveillance data without postnatal confirmation. The primary utility of prenatal diagnosis of clubfoot is in its indication for additional genetic counseling and testing through amniocentesis or other means.

Additional Information:
Clubfoot can occur on either side alone or in both feet. The calf muscles on the affected side are permanently small. While in some instances the affected foot can be moved passively to a normal or near-normal position (so-called positional clubfoot), more commonly there is a component of rigidity which can be severe.

Clubfoot often occurs alone, but can be associated with other musculoskeletal abnormalities such as torticollis or developmental dysplasia of the hip, and with genetic syndromes such as triploidy, Larsen syndrome, or Moebius sequence. Neurogenic clubfoot results from impaired innervation of the foot during development. Examples of conditions that can result in such impairment include spina bifida, arthrogryposis, sacral agenesis, spinal muscular atrophy, and other paralytic states.
**Craniosynostosis**  
(Extended Condition)

<table>
<thead>
<tr>
<th>Description</th>
<th>Premature closure (fusion) of one or several cranial sutures (connective tissue membranes that separate the bones of the developing skull)</th>
</tr>
</thead>
</table>
| Inclusions | Craniosynostosis subtypes are typically named by the cranial sutures involved: sagittal, coronal, lambdoidal, or metopic craniosynostoses are the most common conditions. Mixed or multiple sutures can be involved, and rarely basilar or squamosal sutures fuse prematurely. Cranial shapes that may or may not result from craniosynostosis:  
• DOLICHOCEPHALY/SCAPHOCEPHALY--long, wedge-shaped skull with a prominent forehead and occiput resulting from premature closure of sagittal suture  
• BRACHYCEPHALY--high, wide, short skull resulting from premature fusion of coronal sutures  
• OXYCEPHALY/TURRICEPHALY/ACROCEPHALY--tall, tower-like skull (sometimes pointed) resulting from premature fusion of coronal and usually sagittal sutures  
• PLAGIOCEPHALY--asymmetric skull shape which can result from unilateral closure of coronal and/or lambdoidal suture  
• TRIGONOCEPHALY--triangular-shaped skull resulting from premature closure of metopic suture |
| Exclusions | Deformational plagiocephaly without synostosis  
Other abnormal head shapes described above without craniosynostosis |
| ICD-9-CM Codes | No specific code; 756.0 includes craniosynostosis and “other anomalies of skull and face bones” |
| ICD-10-CM Codes | Q75.0 |
| CDC/BPA Codes | 756.00-756.03 |
| Diagnostic Methods | Confirmation of a diagnosis of craniosynostosis is by postnatal skull X-ray and/or tomography (CT or CAT scan, the “gold standard”), operative/pathology reports, or autopsy; plastic surgery or neurosurgery consultation reports are often useful |
| Prenatal Diagnoses Not Confirmed Postnatally | Craniosynostosis can be identified or suspected on prenatal ultrasound; however, it should not be included in birth defects surveillance data without postnatal confirmation. |

**Additional Information:**  
Craniosynostosis is seen in many syndromes such as the acrocephalosyndactylies, in which there are limb abnormalities such as syndactyly. A particularly severe form of craniosynostosis of multiple sutures is called cloverleaf skull or Kleeblattschädel; this condition is usually associated with a syndrome diagnosis.
### Diaphragmatic Hernia
(Recommended Condition)

<table>
<thead>
<tr>
<th>Description</th>
<th>Incomplete formation of the diaphragm through which a portion of the abdominal contents herniate into the thoracic cavity.</th>
</tr>
</thead>
</table>
| Inclusions  | Absence of the diaphragm  
Bochdalek hernia – Herniation through a defect in the posterolateral portion of the diaphragm.  
Diaphragmatic hernia, type not specified  
Hemidiaphragm  
Morgagni hernia – Herniation through a defect in the anterior portion of the diaphragm.  
Paraesophageal hernia – Herniation through a defect in the central portion of the diaphragm surrounding the esophagus. |
| Exclusions  | Eventration of the diaphragm – Weakness in, or absence of, the muscles of the diaphragm which allows upward displacement of a portion of the abdominal contents. However, there is no true herniation of contents through the diaphragm into the thoracic cavity. |
| ICD-9-CM Codes | 756.6 |
| ICD-10-CM Codes | Q79.0, Q79.1 |
| CDC/BPA Codes | 756.610 – 756.617 |
| Diagnostic Methods | While diaphragmatic hernia may be suspected by the clinical presentation of respiratory distress, feeding intolerance, and/or cardiac compromise, it may be conclusively diagnosed only through x-ray, contrast study of the bowel, CT or MRI scan, surgery, or autopsy. |
| Prenatal Diagnoses Not Confirmed Postnatally | Diaphragmatic hernia may be included in surveillance data when only diagnosed prenatally. However, if it is possible to ascertain the degree of certainty of the prenatal diagnosis, this should factor into the decision as to whether or not to include an individual case in the surveillance data. Live-born children who survive should always have confirmation of the defect postnatally before being included. |
| Additional Information: | Children with diaphragmatic hernia often have accompanying abnormalities of the heart, intestine, and lungs, including hypoplastic lungs, which result from the abnormal location of abdominal organs within the thoracic cavity during development. |
## Gastroschisis
(Core Condition)

| Description | A congenital opening or fissure in the anterior abdominal wall lateral to the umbilicus through which the small intestine, part of the large intestine, and occasionally the liver and spleen, may herniate. The opening is separated from the umbilicus by a small bridge of skin, and the herniating organs are not covered by a protective membrane. Gastroschisis usually occurs on the right side of the umbilicus, although it may occur on the left. |
| Inclusions | Gastroschisis |
| Exclusions | Omphalocele |
| ICD-9-CM Codes | Prior to October 1, 2009 - 756.79 (shared code with omphalocele) October 1, 2009 and later – 756.73 |
| ICD-10-CM Codes | Q79.3 |
| CDC/BPA Codes | 756.71 |
| Diagnostic Methods | Gastroschisis is usually easily recognized on physical examination after delivery. However, in some instances, it may be conclusively distinguished from omphalocele only at surgery or autopsy. |
| Prenatal Diagnoses Not Confirmed Postnatally | Gastroschisis may be included when only diagnosed prenatally. However, it may be difficult to distinguish gastroschisis from omphalocele on prenatal ultrasound, and the terms sometimes are used interchangeably. If it is possible to ascertain the degree of certainty of the prenatal diagnosis and the location of the umbilical cord insertion relative to the abdominal defect, this should factor into the decision as to whether or not to include an individual case in the surveillance data. Live-born children who survive should always have confirmation of the defect postnatally before being included. In addition, the absence of gastroschisis on prenatal ultrasound does not necessarily mean that it will not be diagnosed after delivery. |

**Additional Information:**
The distinction between gastroschisis and omphalocele is important because they have different etiologies and different implications for treatment and long-term survival.

In gastroschisis, the umbilicus and cord are normal and separated from the abdominal wall defect by a small bridge of skin. The herniating organs are not covered by a protective membrane. However, they
may appear matted and covered by a thick fibrous material as a result of prolonged exposure to amniotic fluid \textit{in utero}.

In omphalocele, abdominal organs herniate through the umbilicus into the umbilical cord. There is no bridge of skin between the abdominal wall defect and the umbilicus and cord. While the herniating organs are covered by a protective membrane, this may rupture before, during, or after delivery.

Gastroschisis may be one of the defects reported as part of the Limb-Body Wall complex. This is a disruption complex of the lateral body wall, which may also include limb reductions, neural tube defects, heart defects, and other anomalies.

Maternal serum alphafetoprotein (MSAFP) and/or amniotic fluid alphafetoprotein (AFAFP) may be elevated with gastroschisis. However, these screening tests alone are not sufficient to diagnose the condition.
Limb Deficiencies (Reduction Defects)
(Core Condition)

Description
Complete or partial absence of the upper arm (humerus), lower arm (radius and/or ulna), wrist (carpals), hand (metacarpals), fingers (phalanges), thigh (femur), lower leg (tibia and/or fibula), ankle (tarsals), foot (metatarsals), or toes (phalanges).

Inclusions
Transverse limb deficiency (reduction) – Complete or partial absence of the distal (furthest from the body) structures of the arm or leg in a transverse (cross-wise) plane at the point where the deficiency begins. Structures proximal to the point where the deficiency begins remain essentially intact. Selected terms used for types of transverse limb deficiencies include:
- Acheiria – Absence of a hand
- Adactyly – Absence of digits (fingers or toes), excluding isolated missing thumb (see below)
- Aphalangia – Absence of phalanges. Fingers contain 3 phalanges each. The thumb (pollex) and big toe (hallux) contain 2 phalanges. The other toes contain 3 phalanges each.
- Amelia – Complete absence of the upper limb (humerus, radius, ulna, wrist, hand and fingers) or complete absence of the lower limb (femur, tibia, fibula, ankle, foot, and toes).
- Hemimelia, Meromelia – Partial absence of a limb. This may refer to either transverse or longitudinal deficiency (reduction).
- Oligodactyly – Deficiency of fewer than 5 digits.

Transverse terminal deficiency (reduction) – Complete absence of the distal structures of the arm with the proximal structures intact. This term usually refers to deficiency below the elbow, or complete absence of the distal structures of the leg with the proximal structures intact.

Congenital amputation, type not specified.

Longitudinal limb deficiency (reduction) – Partial absence of the upper limb in parallel with the long axis of the arm or partial absence of the lower limb in parallel with the long axis of the leg. These may involve preaxial (on the thumb side/on the big toe side), postaxial (on the fifth finger side/on the fifth toe side), or central parts of the arm or leg. Selected terms used for types of longitudinal limb reductions include:
- Ectrodactyly
- Ectromelia
- Isolated missing thumb
- Lobster claw hand
- Radial, ulnar, tibial, or fibular aplasia or hypoplasia
- Radial, ulnar, tibial, or fibular ray deficiency

Split-hand malformation (split hand/split foot malformation, SHSF) – A central longitudinal limb deficiency (reduction) in which there is complete or partial absence of one or more of the central rays (second through fourth
fingers and their associated metacarpal bones) of the hand.

Split-foot malformation (split hand/split foot malformation, SHSF) – A central longitudinal limb deficiency (reduction) in which there is complete or partial absence of one or more of the central rays (second through fourth toes and their associated metatarsal bones) of the foot.

Intercalary limb reduction – Complete or partial absence of the proximal (closest to the body) or middle segments of the upper limb or lower limb with all or part of the distal segment present.

Phocomelia is a general term used for any type of intercalary limb reduction.

Deficiency (reduction defect) of the upper limb or lower limb not elsewhere coded or of unspecified type – Complete or partial absence of the upper limb or lower limb that does not fall within the above categories or for which there is no specific description.

**Exclusions**

Shortened arms, forearms, hands, upper and/or lower legs, feet, toes or fingers that have all of their component parts, including those that are part of a generalized chondodystrophy, osteodystrophy, or dwarfism.

Hypoplastic nails

**ICD-9-CM Codes**

755.2 – 755.4

**ICD-10-CM Codes**

Q71.0 – Q71.9, Q72.0 – Q72.9, Q73.0 – Q73.8

**CDC/BPA Codes**

755.20 – 755.49

**Diagnostic Methods**

Limb deficiencies (reductions) are usually easily recognized on physical examination at delivery. However, the exact nature of the defect may only be distinguished by x-ray, surgery, or autopsy.

**Prenatal Diagnoses Not Confirmed Postnatally**

While these conditions may be identified by prenatal ultrasound, they generally should not be included in surveillance data without postnatal confirmation. However, if it is possible to ascertain the degree of certainty of the prenatal diagnosis, this should factor into the decision as to whether or not to include an individual case in the surveillance data. Lack of visualization of a bone or limb on prenatal ultrasound does not necessarily mean that the bone or limb truly is not present. Live-born children who survive should always have confirmation of the defect postnatally before being included.

**Additional Information:**

The terminology for limb deficiency (reduction) is often confusing. Some terms (such as “phocomelia”) have been misused and others (such as “ectrodactyly”) have been used for both longitudinal and transverse defects. If medical record review is available, it is important to look for a complete description of all structures that are present and absent in order to verify the diagnosis.
Transverse limb deficiency (reduction) may be seen in association with amniotic bands. When both are present, both conditions should be coded.

Rudimentary or nubbin toes may be present at the distal end of a transverse limb deficiency (reduction). Their presence alone does not change the classification of the defect as transverse.

Joint contractures or clubfoot/clubhand are commonly seen in association with longitudinal limb deficiencies.

Intercalary deficiency (phocomelia) has been associated with the use of thalidomide during early pregnancy. However, thalidomide use may result in a number of other defects, including longitudinal deficiency. Intercalary defects also may occur without exposure to thalidomide.

Limb deficiency is one of the defects that may be reported as part of:

The VATER or VACTERL association, which also may include vertebral, cardiac and renal defects, TE fistula, and anal atresia.

Oromandibular-Limb Hypogenesis spectrum, which also may include a small mouth, small chin (micrognathia), small tongue (hypoglossia), and sixth and seventh cranial nerve palsies (Moebius sequence).
## Omphalocele
(Recommended Condition)

| Description | A defect in the anterior abdominal wall in which the umbilical ring is widened, allowing herniation of abdominal organs, including the small intestine, part of the large intestine, and occasionally the liver and spleen, into the umbilical cord. The herniating organs are covered by a nearly transparent membranous sac. |
| Inclusions | Omphalocele |
| Exclusions | Gastrochisis  
Umbilical hernia |
| ICD-9-CM Codes | Prior to October 1, 2009 - 756.79 (shared code with gastrochisis)  
After October 1, 2009 – 756.72 |
| ICD-10-CM Codes | Q79.2 |
| CDC/BPA Codes | 756.70 |
| Diagnostic Methods | Omphalocele is usually easily recognized on physical examination after delivery. However, in some instances, it may be conclusively distinguished from gastrochisis only at surgery or autopsy. |
| Prenatal Diagnoses Not Confirmed Postnatally | Omphalocele may be included when only diagnosed prenatally. However, it may be difficult to distinguish omphalocele from gastrochisis on prenatal ultrasound, and the terms sometimes are used interchangeably. If it is possible to ascertain the degree of certainty of the prenatal diagnosis and the location of the umbilical cord insertion relative to the abdominal defect, this should factor into the decision as to whether or not to include an individual case in the surveillance data. Live-born children who survive should always have confirmation of the defect postnatally before being included. In addition, the absence of omphalocele on prenatal ultrasound does not necessarily mean that it will not be diagnosed after delivery. |

### Additional Information:
The distinction between omphalocele and gastrochisis is important because they have different etiologies and different implications for treatment and long-term survival.

In omphalocele, abdominal organs herniate through the umbilicus into the umbilical cord. There is no bridge of skin between the abdominal wall defect and the umbilicus and cord. While the herniating organs are covered by a protective membrane, this may rupture before, during, or after delivery.

In gastrochisis, the umbilicus and cord are normal and separated from the abdominal wall defect by a small bridge of skin. The herniating organs are not covered by a protective membrane. However, they may
appear matted and covered by a thick fibrous material as a result of prolonged exposure to amniotic fluid in utero.

Omphalocele is one of the defects reported as part of the Omphalocele-Exstrophy-Imperforate Anus-Spina Bifida (OEIS) complex.

Maternal serum alphafetoprotein (MSAFP) and/or amniotic fluid alphafetoprotein (AFAFP) may be elevated with omphalocele. However, these screening tests alone are not sufficient to diagnose the condition.

In contrast to omphalocele, umbilical hernias are completely covered by normal skin.
**Chromosomal**

### Deletion 22q11.2
(Extended Condition)

**Description**
Chromosome abnormality resulting from genomic microdeletions within a critical region on the long arm of chromosome 22 (22q11.2)

**Inclusions**
- Deletion 22q11.2 syndrome
- Chromosome deletion 22q11.2
- DiGeorge syndrome with chromosome 22q11.2 deletion
- Thymic aplasia syndrome with chromosome 22q11.2 deletion
- Velo-cardio-facial (VCF) syndrome with chromosome 22q11.2 deletion
- Conotruncal anomaly face (CTAF) syndrome with chromosome 22q11.2 deletion
- Cayler cardiofacial (asymmetric crying facies) syndrome with chromosome 22q11.2 deletion
- Shprintzen syndrome with chromosome 22q11.2 deletion
- Sedlackova (velofacial hypoplasia) syndrome with chromosome 22q11.2 deletion
- Takao syndrome with chromosome 22q11.2 deletion

**Exclusions**
Named phenotypes without cytogenetic abnormalities
- TBX1 mutations without cytogenetic abnormalities
- Deletion 22q13.3
- Duplication 22q11.2
- Shprintzen-Goldberg syndrome

**ICD-9-CM Codes**
758.32

**ICD-10-CM Codes**
Q93.81

**CDC/BPA Codes**
758.37

**Diagnostic Methods**
Deletion 22q11.2 syndrome might be suspected on physical examination. However, it is diagnosed conclusively only through molecular cytogenetic analysis (typically chromosomal microarray or fluorescence in situ hybridization). Rarely, direct analysis of the child’s chromosomes (karyotype) can suggest a 22q11.2 deletion associated with an unbalanced translocation involving another chromosome, but molecular cytogenetic analysis would be used to confirm the 22q11.2 deletion. All of these laboratory techniques may be done with blood or tissue cells.

**Prenatal Diagnoses Not Confirmed Postnatally**
Deletion 22q11.2 can be included only when diagnosed through molecular cytogenetic analysis obtained from amniocentesis, chorionic villus sampling (CVS), or percutaneous umbilical blood sampling (PUBS).

**Additional Information:**
The deletion 22q11.2 syndrome phenotype can include cardiac abnormalities, abnormal or dysmorphic facial features, thymic aplasia, cleft palate or velopharyngeal insufficiency, or hypocalcemia due to
hypoparathyroidism; the “CATCH” acronym appeared in the literature previously to describe these cardinal features, but this term is no longer used. Chromosome 22q11.2 deletions can be found with any of these features in isolation, and is sometimes not diagnosed until adulthood, e.g., in subtly affected parents of children with deletion 22q11.2 syndrome phenotypes or defects.

The term “DiGeorge syndrome” was used originally (before 22q11.2 deletions were described) for children with the combination of thymic and parathyroid defects; the ICD-9-CM code 279.11 or ICD-10-CM code D82.1 is sometimes still found in medical records with this diagnosis, but should be used in combination with the chromosomal codes listed above for individuals with documented 22q11.2 deletions.

The most common 22q11.2 deletions can be detected by commercially-available fluorescence in situ hybridization (FISH) probes, but normal FISH results with smaller 22q11.2 deletions seen on chromosomal microarrays are occasionally reported. These findings are sometimes called “atypical” deletions and labelled with specific letters (e.g., “C-D” deletion) or numbers describing the chromosomal loci; such cases should be included for surveillance purposes if the microarray interpretation is consistent with a pathogenic or clinically-significant 22q11.2 deletion.
# Trisomy 13

(Recommended Condition)

<table>
<thead>
<tr>
<th>Description</th>
<th>The presence of three copies of all or a large part of chromosome 13.</th>
</tr>
</thead>
</table>
| **Inclusions** | Patau syndrome  
Mosaic Patau syndrome  
Mosaic trisomy 13  
Translocation Patau syndrome  
Translocation trisomy 13  
Trisomy 13, not otherwise specified  
Trisomy D₁, not otherwise specified |
| **Exclusions** | Balanced translocations involving chromosome 13 |
| **ICD-9-CM Codes** | 758.1 |
| **ICD-10-CM Codes** | Q91.4 – Q91.7 |
| **CDC/BPA Codes** | 758.10 – 758.19 |

**Diagnostic Methods**

Trisomy 13 may be suspected on physical examination. However, it may be diagnosed conclusively only through direct analysis of the infant’s chromosomes (karyotype). The chromosomes may be obtained from blood or tissue cells.

**Prenatal Diagnoses Not Confirmed Postnatally**

Trisomy 13 may be included when only diagnosed through direct analysis of fetal chromosomes or molecular cytogenetic analysis (typically chromosomal microarray or fluorescence in situ hybridization) of cells obtained from amniocentesis, chorionic villus sampling (CVS), or percutaneous umbilical blood sampling (PUBS). However, when mosaic trisomy 13 is noted, the defect should be confirmed postnatally on a specimen obtained directly from the infant or fetus after birth (see below).

**Additional Information:**

When the two copies of chromosome 13 from one parent do not separate during egg or sperm formation, three copies of the entire chromosome 13 will be present in the fetus. In this instance, the karyotype is written as 47,XX,+13 or 47,XY,+13. This is the most common type of trisomy 13 and is associated with advanced maternal age, particularly of 35 years or greater.

Translocation trisomy 13 occurs when two separate copies of chromosome 13 are present, but a third copy of part of chromosome 13 is attached to another chromosome. In this instance, there are 46 total chromosomes present, but 3 copies of part of chromosome 13.

Mosaic trisomy 13 occurs when some, but not all, of the cells in the body contain three copies of all or a large part of chromosome 13. In this instance, the karyotype is written as 46,XY/47,XY,+13, for example. Because the placenta may contain mosaic cell lines not present in the fetus, mosaic trisomy 13 diagnosed through chorionic villus sampling should always be confirmed by direct examination of fetal chromosomes from amniocentesis, PUBS, or preferably postnatal blood or tissue samples.
Approximately 80% of infants with trisomy 13 do not survive beyond the first month of life. Major malformations associated with trisomy 13 may include holoprosencephaly, microcephaly, meningomyelocele, cleft lip and/or palate, microphthalmia, retinal dysplasia, polydactyly, heart defects (most commonly a VSD), omphalocele, and genitourinary defects, among others. Among children who survive the newborn period, severe developmental delay is virtually always present as may be deafness, visual impairment, minor motor seizures, and apneic spells.

Infants with mosaic trisomy 13 may be less severely affected with variable degrees of developmental delay and longer survival. Infants with partial trisomy for the proximal segment of chromosome 13 (13pter→q14) exhibit a nonspecific pattern of abnormalities with near-normal survival. Approximately 25% of infants with partial trisomy for the distal segment of chromosome 13 (13q14→qter) die during early postnatal life.

Children who survive exhibit severe developmental delay and specific abnormalities.

Major malformations that occur with trisomy 13 in the same infant should be coded separately, as their presence may varies among affected individuals.
## Trisomy 18
*(Recommended Condition)*

<table>
<thead>
<tr>
<th><strong>Description</strong></th>
<th>The presence of three copies of all or a large part of chromosome 18.</th>
</tr>
</thead>
</table>
| **Inclusions**  | Edwards syndrome  
Mosaic Edwards syndrome  
Mosaic trisomy 18  
Translocation Edwards syndrome  
Translocation trisomy 18  
Trisomy 18, not otherwise specified |
| **Exclusions**  | Balanced translocations involving chromosome 18 |
| **ICD-9-CM Codes** | 758.2 |
| **ICD-10-CM Codes** | Q91.0 – Q91.3 |
| **CDC/BPA Codes** | 758.20 – 758.29 |
| **Diagnostic Methods** | Trisomy 18 may be suspected on physical examination. However, it may be diagnosed conclusively only through direct analysis of the infant’s chromosomes (karyotype). The chromosomes may be obtained from blood or tissue cells. |
| **Prenatal Diagnoses Not Confirmed Postnatally** | Trisomy 18 may be included when only diagnosed through direct analysis of fetal chromosomes or molecular cytogenetic analysis (typically chromosomal microarray or fluorescence in situ hybridization) of cells obtained from amniocentesis, chorionic villus sampling (CVS), or percutaneous umbilical blood sampling (PUBS). However, when mosaic trisomy 13 is noted, the defect should be confirmed postnatally on a specimen obtained directly from the infant or fetus after birth (see below). |

**Additional Information:**
When the two copies of chromosome 18 from one parent do not separate during egg or sperm formation, three copies of the entire chromosome 18 will be present in the fetus. In this instance, the karyotype is written as 47,XX,+18 or 47,XY,+18. This is the most common type of trisomy 18 and is associated with advanced maternal age, particularly of 35 years or greater.

Translocation trisomy 18 occurs when two separate copies of chromosome 18 are present, but a third copy of part of chromosome 18 is attached to another chromosome. In this instance, there are 46 total chromosomes present, but 3 copies of part of chromosome 18.

Mosaic trisomy 18 occurs when some, but not all, of the cells in the body contain three copies of all or a large part of chromosome 18. In this instance, the karyotype is written as 46,XY/47,XY,+18, for example. Because the placenta may contain mosaic cell lines not present in the fetus, mosaic trisomy 18 diagnosed through chorionic villus sampling should always be confirmed by direct examination of fetal chromosomes from amniocentesis, PUBS, or preferably postnatal blood or tissue samples.

Most pregnancies affected with trisomy 18 result in spontaneous abortion. Approximately 50% of live-
born infants with trisomy 18 do not survive beyond the first week of life. Only 5% to 10% survive beyond the first year of life. Major malformations associated with trisomy 18 may include microcephaly, micrognathia, cleft lip and/or palate, heart defects, omphalocele, and renal defects, among others. Minor anomalies associated with trisomy 18 may include low-set malformed auricles (external ears), overlapping of the index and fifth fingers over the third and fourth fingers, absent distal crease on the fifth finger, hirsutism (excess hair) of the forehead and back, lateral deviation of the hands, a hypoplastic thumb, a single transverse palmar crease, and rocker-bottom feet, among others. Developmental delay is virtually always present, as may be hypertonicity, a weak cry, growth retardation, hypoplasia of skeletal muscle and subcutaneous fat, and clenched hands.

Infants with mosaic trisomy 18 may be less severely affected, with variable degrees of developmental delay and longer survival. Infants with trisomy of only the short arm of chromosome 18 (partial trisomy 18) exhibit a nonspecific pattern of abnormalities with mild to no developmental delay. Infants with trisomy of the short arm, centromere, and proximal third of the long arm of chromosome 18 exhibit features of trisomy 18 but not the entire spectrum of abnormalities. Infants with trisomy of only one-third to one-half of the long arm of chromosome 18 exhibit features of trisomy 18 but have longer survival and less severe developmental delays.

Major malformations that occur with trisomy 18 in the same infant should be coded separately, as their presence varies among affected individuals.
### Trisomy 21 (Down Syndrome) (Core Condition)

<table>
<thead>
<tr>
<th>Description</th>
<th>The presence of three copies of all or a large part of chromosome 21.</th>
</tr>
</thead>
</table>

#### Inclusions
- Down syndrome
- Mosaic Down syndrome
- Mosaic trisomy 21
- Translocation Down syndrome
- Translocation trisomy 21
- Trisomy 21, not otherwise specified

#### Exclusions
- Balanced translocations involving chromosome 21

#### ICD-9-CM Codes
- 758.0

#### ICD-10-CM Codes
- Q90.0 – Q90.9

#### CDC/BPA Codes
- 758.00 – 758.09

#### Diagnostic Methods
Down syndrome may be suspected on physical examination. However, it may be diagnosed conclusively only through direct analysis of the infant’s chromosomes (karyotype). The chromosomes may be obtained from blood or tissue cells.

#### Prenatal Diagnoses Not Confirmed Postnatally
Down syndrome may be included when only diagnosed through direct analysis of fetal chromosomes or molecular cytogenetic analysis (typically chromosomal microarray or fluorescence in situ hybridization) of cells obtained from amniocentesis, chorionic villus sampling (CVS), or percutaneous umbilical blood sampling (PUBS). However, when mosaic trisomy 21 is noted, the defect should be confirmed postnatally on a specimen obtained directly from the infant or fetus after birth (see below).

#### Additional Information:
When the two copies of chromosome 21 from one parent do not separate during egg or sperm formation, three copies of the entire chromosome 21 will be present in the fetus. In this instance, the karyotype is written as 47,XX,+21 or 47,XY,+21. This is the most common type of trisomy 21 and is associated with advanced maternal age, particularly of 35 years or greater.

Translocation trisomy 21 occurs when two separate copies of chromosome 21 are present, but a third copy...
of part of chromosome 21 is attached to another chromosome. In this instance, there are 46 total chromosomes present, but 3 copies of part of chromosome 21.

Mosaic trisomy 21 occurs when some, but not all, of the cells in the body contain three copies of all or a large part of chromosome 21. In this instance, the karyotype is written as 46,XY/47,XY,+21, for example. Because the placenta may contain mosaic cell lines not present in the fetus, mosaic trisomy 21 diagnosed through chorionic villus sampling should always be confirmed by direct examination of fetal chromosomes from amniocentesis, PUBS, or preferably postnatal blood or tissue samples.

Infants with Down syndrome have a typical appearance and other characteristics, including decreased muscle tone (hypotonia), a weak startle (Moro) reflex, hyperflexible joints, a flattened facial profile, upslanting eyes, abnormally shaped external ears (auricles), loose skin on the back of the neck, dysplasia of the pelvic bones, incurving of the fifth finger (clinodactyly), and a single transverse crease in the palm of the hand (Simian crease). Developmental delay is virtually always present. Major malformations associated with Down syndrome include heart defects (most notably atrioventricular septal defects), gastrointestinal defects, and vertebral abnormalities, among others.

Major malformations that occur with Down syndrome in the same infant should be coded separately, as their presence may varies among affected individuals.

Mongolism is an outdated term for Down syndrome.
### Turner Syndrome
(Extended Condition)

**Description**
Presence of an absent or structurally abnormal second X chromosome in a phenotypic female.

**Inclusions**
- Turner syndrome
- Turner syndrome mosaicism (45,X with 46,XX, 46,XY, 47,XXX, autosomal translocation, or combinations of the above)
- Turner syndrome with a ring X chromosome
- Turner syndrome with an isochromosome X
- Turner syndrome with Xp deletion
- Gonadal dysgenesis, many forms

**Exclusions**
- Chromosome Xq24 deletions without Turner syndrome phenotype
- Chromosomal deletions distal to Xp22.3 without Turner syndrome phenotype
- Males, e.g., with 46,XY/45,X mosaicism

**ICD-9-CM Codes**
758.6

**ICD-10-CM Codes**
Q96.0- Q96.9

**CDC/BPA Codes**
758.60-758.69

**Diagnostic Methods**
Physical examination often provides a strong clinical suspicion of Turner syndrome. However, it is diagnosed conclusively only through direct analysis of the infant’s chromosomes (karyotype). The chromosomes may be obtained from blood (lymphocytes), or tissue cells (skin fibroblasts, chorionic villi). Cheek (buccal) swab analysis is inadequate for diagnosis, although it is useful to evaluate mosaicism. Molecular cytogenetic analysis (typically chromosomal microarray or fluorescence in situ hybridization) is not the standard type of laboratory investigation for Turner syndrome, but recent studies show that chromosomal microarray can detect the missing X chromosome for both complete and mosaic forms. Fluorescence in situ hybridization (FISH) performed prenatally can detect 45,X but not other forms; if Turner syndrome is strongly suspected in the fetus and FISH is negative, postnatal blood karyotyping must be performed.

**Prenatal Diagnoses Not Confirmed Postnatally**
Turner syndrome can be included only when diagnosed through direct analysis of fetal chromosomes (karyotype) or molecular cytogenetic analysis of cells obtained from amniocentesis, chorionic villus sampling (CVS), or percutaneous umbilical blood sampling (PUBS). However, when mosaic Turner syndrome is noted, the abnormality should be confirmed postnatally on a specimen obtained directly from the infant or fetus after birth (see below).

**Additional Information:**
The appearance of a fetus or infant with Turner syndrome varies greatly from a severely hydropic nonviable fetus to a normal appearing infant. The classic phenotype includes physical features that represent the residua of fetal lymphatic distention (body edema, neck edema, low hairline, low-set ears, downslanted eyes, loose neck skin, puffy hands and feet), and congenital heart defects (coarctation, other forms of left-heart...
The facial appearance might include wide-spaced eyes and small chin. Renal anomalies are seen in 30% (horseshoe kidney, absent kidney). Although short stature occurs in most children with Turner syndrome, infants usually have normal size.

There are different causes of the different chromosome types of Turner syndrome. When 45,X is present, the chromosomal abnormality occurred as a random event during the formation of reproductive cells (eggs and sperm) in the affected person's parent. An error in cell division called nondisjunction resulted in reproductive cells with an abnormal number of chromosomes.

Mosaic Turner syndrome occurs as a random event during cell division in early fetal development. As a result, some of an affected person’s cells have the usual two sex chromosomes, and other cells have only one copy of the X chromosome. Other sex chromosome abnormalities are also possible in females with X chromosome mosaicism. Rarely, Turner syndrome caused by a partial deletion of the X chromosome can be passed from one generation to the next.

Birth defects, especially heart and kidney defects, that occur with Turner syndrome should be coded separately, as their presence may varies among affected individuals.

Bonnevie-Ullrich and Ullrich-Turner are outdated terms for Turner syndrome.
Appendix 3.3

Examples of Conditions Considered to Be Minor Anomalies
Appendix 3.3
Examples of Conditions Considered to Be Minor Anomalies

Eye
- Epicanthal folds
- Iris freckles, Brushfield spots
- Upward or downward palpebral slant

Ear
- Darwinian point or tubercle
- Thickened or excessively folded helix
- Lack of helical folding
- Creased, notched, or bifid ear lobe
- Lop, cup-shaped, or retroverted ear
- Preauricular sinus, cyst, pit, or skin tag

Head, Face and Neck
- Flat occiput
- Frontal bossing
- Flat brow
- Flat or prominent bridge of nose
- Anteverted nostrils
- Long nasal septum
- Webbed or redundant neck skin

Hands and Feet
- Single or horizontal palmar crease
- Clinodactyly
- Tapered fingers
- Overlapping digits
- Webbed or widely spaced 2nd and 3rd toes
- Prominent heel

Other
- Sacral dimples
- Nevi
- Cafe-au-lait spots
- Mongolian spots
- Accessory nipples
- Umbilical hernia
- Vaginal tag
- Single umbilical artery

3 This is not a comprehensive list. The exact abnormalities considered to be minor defects may vary among experts.
Appendix 3.4

Conditions Related to Prematurity in Infants Born at Less Than 36 Weeks Gestation
Appendix 3.4
Conditions Related to Prematurity in Infants Born at Less Than 36 Weeks Gestation

- Dolichocephaly
- Scaphocephaly
- Blue sclera
- Fused eyelids
- Absent or decreased ear cartilage
- Patent foramen ovale
- Patent ductus arteriosus
- Hypoplastic lungs
- Small or hypoplastic nipples
- Hypoplastic labia majora
- Undescended testicles
- Inguinal hernia
Appendix 3.5

Case Inclusion Guidance for Potentially Zika-related Birth Defects
Appendix 3.5

Case Inclusion Guidance for Potentially Zika-related Birth Defects

Contents

Background ........................................................................................................................................ 1

Brain Abnormalities with and without Microcephaly ................................................................. 2
  Microcephaly .................................................................................................................................. 2
  Intracranial Calcifications .............................................................................................................. 5
  Cerebral / Cortical Atrophy ........................................................................................................... 7
  Abnormal Cortical Gyral Patterns .................................................................................................. 9
  Corpus Callosum Abnormalities .................................................................................................... 11
  Cerebellar abnormalities ................................................................................................................ 12
  Porencephaly .................................................................................................................................. 14
  Hydranencephaly ............................................................................................................................ 16
  Ventriculomegaly/Hydrocephaly .................................................................................................. 18
  Fetal Brain Disruption Sequence .................................................................................................. 21
  Intraventricular Hemorrhage that occurs in utero ..................................................................... 24
  Other Major Brain Abnormalities .................................................................................................. 26

Neural Tube Defects and Other Early Brain Malformations ....................................................... 28
  Anencephaly/Acrania ....................................................................................................................... 28
  Encephalocele ............................................................................................................................... 30
  Spina Bifida without Anencephaly ................................................................................................. 32
  Holoprosencephaly/Ahinencephaly ............................................................................................... 35

Eye Abnormalities .......................................................................................................................... 37
  Microphthalmia/Anophthalmia ........................................................................................................ 37
  Coloboma ....................................................................................................................................... 39
  Congenital Cataract ......................................................................................................................... 41
  Intraocular Calcification ................................................................................................................ 43
  Chorioretinal Atrophy, Scarring, Pigmentary Changes, Retinitis ................................................ 45
  Optic Nerve Atrophy, Pallor, other Optic Nerve Abnormalities .................................................. 47

Consequences of Central Nervous System (CNS) Dysfunction .................................................... 49
  Arthrogryposis ............................................................................................................................... 49
  Clubfoot with associated brain abnormalities .............................................................................. 51
  Congenital Hip Dislocation / Developmental Dysplasia of the Hip with associated brain abnormalities .......................................................... 54
  Congenital Hearing Loss (unilateral or bilateral) ........................................................................... 57
  Figure 1. Brain – Exterior View ..................................................................................................... 60
  Figure 2. Brain - Cross-section View ........................................................................................... 61
  Figure 3. Skull – Exterior View ..................................................................................................... 62
  Figure 4. Eye – Cross-section View ............................................................................................. 63

Glossary of Terms ............................................................................................................................. 64
Background

This document has been developed to provide guidance for reviewing and abstracting medical records of infants with defects potentially related to Zika virus. While it does not provide comprehensive information about each condition to be ascertained, it covers basic clinical descriptions, definitions of terms, and tips on how to look for and where to find information in the medical records.

Some of the conditions listed for ascertainment are not malformations themselves, but conditions that may result from the effects of Zika virus infection in utero. The intent is two-fold: 1) assist in identification of all infants potentially infected with Zika virus in utero, and 2) provide background information on the prevalence of these conditions regardless of the cause.

For programs that have never conducted population-based birth defects surveillance, the list of conditions for ascertainment will form the initial case definition for their activities. For programs that already conduct birth defect surveillance, the list of conditions may require a change in the case inclusion and/or case finding approach.

We hope this guide is helpful. Please contact Jan Cragan (jcragan@cdc.gov) or Cara Mai (cmai@cdc.gov) for questions or comments.
Brain Abnormalities with and without Microcephaly

### Microcephaly

**Description**

Microcephaly, or microcephalus, is the clinical finding of a small head when compared with infants of the same sex and age. The head circumference (HC), also known as the occipitofrontal circumference (OFC), is considered a reliable assessment of the volume of the underlying brain. Microcephaly itself is not a malformation but a sign that the brain is abnormally small.

**Inclusions**

Congenital microcephaly – microcephaly that is present prenatally or at the time of birth/delivery.

For the purposes of surveillance for birth defects potentially linked to Zika, confirmed or possible congenital microcephaly is defined as:

1) Diagnosis of microcephaly or mention of microcephaly or small head in the medical record

AND EITHER 2a OR 2b:

2a) For Live Births: measured HC adjusted for gestational age and sex <3rd percentile at birth\(^\text{†}\); or if not measured at birth, within first 2 weeks of life

2b) For Pregnancy Losses: prenatal HC\(^*\) more than 3 standard deviations (SDs) below the mean on prenatal ultrasound; or postnatal HC\(^\dagger\) <3rd percentile

\(^\text{†}\) HC percentiles for birth measurements should be based on the InterGrowth-21\(^\text{st}\) standards. A tool for calculating percentiles for birth HC, weight, and length is available at: [http://intergrowth21.ndog.ox.ac.uk/](http://intergrowth21.ndog.ox.ac.uk/). These standards are based on measurements within 24 hours of birth, and therefore measurements within 24 hours of birth are appropriate for this assessment.

\(^*\) HC percentiles for prenatal ultrasound measurement should be based on the Society for Maternal Fetal Medicine standards. A table of fetal HC means and SDs by gestational age is available at: [http://www.ajog.org/pb/assets/raw/Health%20Advance/journals/ymob/SMFM%20Statement_Fetal\%20microcephaly.pdf](http://www.ajog.org/pb/assets/raw/Health%20Advance/journals/ymob/SMFM%20Statement_Fetal%20microcephaly.pdf)
‡ Prenatal findings should be confirmed by postnatal evaluation when possible. A suspected brain abnormality noted on prenatal evaluation that is clearly not present on postnatal evaluation should not be included.

Exclusions

For the purpose of surveillance for birth defects potentially linked to Zika, the following should not be included:

- Children with a diagnosis or mention of microcephaly or small head in the medical record for whom the HC measurement is outside of the range mentioned above (see Inclusions)
- Children with a diagnosis or mention of microcephaly or small head in the medical record for whom no HC measurement is available. However, attempt should be made to ascertain the HC measurement at birth or within the first 2 weeks of life.
- Acquired microcephaly - Microcephaly that develops after birth due to a delivery complication or postnatal insult such as trauma or infection in infancy or childhood. In this instance, the head circumference (HC) is normal for sex and age at birth. However, the head becomes disproportionately smaller as the baby grows in length.

The diagnosis of microcephaly should not be assigned by surveillance staff based only on the HC value in the medical record. For the purpose of surveillance for birth defects potentially linked to Zika, there must be diagnosis or mention of microcephaly or small head in the medical record.

ICD-9-CM Codes

742.1 – Microcephalus

ICD-10-CM Codes

Q02 – Microcephaly

CDC/BPA Codes

742.10 – Microcephalus
742.486 – Small brain

Diagnostic Methods

Gold Standard – Head circumference measurement soon after delivery. Prenatal ultrasound or fetal MRI scan can estimate the HC during development. Microcephaly may be mentioned on head/brain ultrasound, CT or MRI scan, but not always. These procedures are not diagnostic.

Medical Records – what and where to look for information

Mention of microcephaly on newborn physical exam (with HC measurement); results of prenatal ultrasound or fetal MRI scan; clinicians’ or nurses’ notes; results of postnatal head/brain ultrasound, CT or MRI scan

Associated Defects / Conditions

Depending on the underlying cause of microcephaly, a variety of brain abnormalities may also be present. Brain abnormalities that have been described in children with potential Zika-associated microcephaly include intracranial calcifications (see page 5); hydranencephaly (see page 12);
polymicrogyria and other neuronal migration disorders (see page 7); agenesis of the corpus callosum (see page 8); cortical loss (see page 6); hydrocephalus *ex-vacuo* (see page 13); and fetal brain disruption sequence (see page 15).

Microcephaly also can result from the presence of other major congenital malformations such as spina bifida (see page 22) and holoprosencephaly (see page 24).

### Prenatal Diagnoses

**Not Confirmed Postnatally**

Microcephaly can be detected on a mid-pregnancy anomaly scan (ultrasound) at 18-20 weeks. However, it may not be evident until the late 2nd or into the 3rd trimester. It is usually present by 36 weeks gestation. Serial prenatal ultrasounds may be needed to detect the development of microcephaly *in utero*. Prenatal findings should be confirmed by postnatal evaluation when possible.

### Additional Information:

Some clinicians use other cut-points, such as less than the 5th or 10th percentile, to make a diagnosis of microcephaly. Microcephaly may also be mentioned in the medical record when the HC measurement is in the normal range for age and sex but small relative to the baby’s weight and length. In other instances, microcephaly or a small head may not be mentioned in the medical record at all even though the measured HC is less than the 3rd percentile (or less than 3 SDs on prenatal ultrasound for a pregnancy loss). Surveillance programs may want to include infants with these conditions in their data. However, for the purposes of surveillance for birth defects potentially linked to Zika, only those infants or fetuses with mention of microcephaly or a small head in the medical record and a HC measurement that fits the stated criteria should be reported (see Inclusions).

The shape of the head after delivery can affect the accuracy of the HC measurement due to molding of the head from the birth canal.

Congenital microcephaly can result from: 1) an abnormality in the very early formation of the brain, often with a genetic etiology, or 2) arrest or destruction of normally-forming brain tissue, e.g., from infection or interruption of the blood supply during gestation. Although not all cases of microcephaly have an identifiable cause, known causes include:

- *In utero* infections such as cytomegalovirus (CMV), rubella, or *toxoplasmosis gondii*
- Chromosomal abnormalities, single gene disorders (syndromes), and mitochondrial mutations
- Teratogens including maternal alcohol use, certain medications, and toxins
- Maternal conditions such as poorly controlled diabetes, hyperphenylalaninemia, and severe malnutrition
- *In utero* ischemia or hypoxia (e.g., placental insufficiency or abruption)
Intracranial Calcifications

**Description**
Accumulations or deposits of calcium within the brain tissue. The calcifications themselves are not malformations but a sign of brain injury such as from infection, hemorrhage, or hypoxia (lack of oxygen).

**Inclusions**
Calcifications noted anywhere within the substance of the brain. Brightly echogenic foci on ultrasound, CT or MRI scan.

**Exclusions**
Calcifications associated with a brain tumor or thrombosis (blood clot) in a large blood vessel within the brain, such as might be seen with tuberous sclerosis or a transverse/straight sinus thrombosis.

**ICD-9-CM Codes**
No specific code; may be included under 742.4 – Other specified anomalies of brain.

**ICD-10-CM Codes**
No specific code; may be included under:
Q04.8 – Other specified congenital malformations of brain
Q04.9 – Congenital malformations of brain, unspecified.

**CDC/BPA Codes**
742.48 – Other specified anomalies of brain.

**Diagnostic Methods**
Gold standard – Prenatal or postnatal head/brain ultrasound, CT or MRI scan; autopsy or pathology. Intracranial calcifications cannot be detected by physical exam.

**Medical Records – what and where to look for information**
Results of prenatal ultrasound or fetal MRI scan; consultation reports by neurologist, geneticist, infectious disease specialist, or other subspecialist; clinicians’ or nurses’ notes; results of postnatal head/brain CT or MRI scan; autopsy or pathology report.

**Associated Defects / Conditions**
Depending on the underlying injury or cause of the calcification, a variety of brain abnormalities may also be present.

**Prenatal Diagnoses Not Confirmed Postnatally**
Intracranial calcifications may be included when only diagnosed prenatally on serial ultrasounds or a single fetal MRI scan. The certainty of the finding on a single prenatal ultrasound that does not persist on subsequent prenatal ultrasounds may be questionable. Prenatal findings should be confirmed by postnatal evaluation when possible.

**Additional Information:**
Some calcifications can be normal variants but usually in the context of an older person.
Causes of intracranial calcifications in a fetus or newborn include in utero infections such as cytomegalovirus (CMV), rubella, or toxoplasmosis gondii. In toxoplasmosis, the intracranial calcifications tend to be randomly distributed within the brain. In CMV, they tend to be distributed periventricularly (around the cerebral ventricles). The intracranial calcifications that have been described in children with birth defects potentially linked to Zika virus infection tend to be distributed in the region below the cerebral cortex (subcortical) and in other areas of the brain including the basal ganglia and brainstem. Other non-infectious causes include damage from anoxia (lack of oxygen) or intracranial hemorrhage (bleeding within the substance of the brain); vascular malformations within the brain, such as Sturge-Weber syndrome; storage diseases, such as Krabbe disease; and mitochondrial diseases.
### Cerebral / Cortical Atrophy

#### Description
Atrophy is a general term which means the loss of cells, and hence the loss of size of the organ or tissue, usually after initial normal development. Cerebral, or cortical, atrophy refers to loss of cells within the two cerebral hemispheres, the main portion of the brain. It can affect all or part of one or both hemispheres. Cerebral atrophy itself is not a malformation but a sign of an underlying problem.

#### Inclusions
- Atrophy of any part of the cerebral hemispheres
- Cerebral atrophy
- Cortical atrophy
- Cortical loss

#### Exclusions
- Cerebral or cortical cysts
- Cerebral atrophy that is secondary to prematurity

#### ICD-9-CM Codes
No specific code; may be included under 742.2 – Reduction deformities of brain

#### ICD-10-CM Codes
No specific code; may be included under Q04.3 – Other reduction deformities of brain

#### CDC/BPA Codes
- 742.48 – Other specified anomalies of brain

#### Diagnostic Methods
Gold standard – Postnatal CT or MRI scan; autopsy or pathology

Cerebral atrophy can also be described on prenatal or postnatal ultrasound.

Cerebral atrophy cannot be diagnosed by physical exam, although associated neurologic symptoms may be noted.

#### Medical Records – what and where to look for information
- Results of prenatal ultrasound or fetal MRI scan; consultation reports by neurologist, geneticist, or other subspecialist; clinicians’ or nurses’ notes; results of postnatal head/brain CT or MRI scan; autopsy or pathology report

#### Associated Defects / Conditions
Depending on the degree, cerebral atrophy can lead to reduced brain volume. As a result, the lateral ventricles are larger than normal (ventriculomegaly, see page 13). Likewise, there is often an increase in the cerebrospinal fluid between the brain and skull (extra-axial fluid). This is sometimes called “benign hydrocephalus”.

Depending on the underlying condition that leads to cerebral atrophy, a variety of other brain abnormalities may also be present.

#### Prenatal Diagnosis
Cerebral atrophy may be included when only diagnosed prenatally on
Not Confirmed Postnatally

serial ultrasounds.

The certainty of the finding on a single prenatal ultrasound that does not persist on subsequent prenatal ultrasounds may be questionable. Prenatal findings should be confirmed by postnatal evaluation when possible.

Additional Information:

When the cerebral ventricles are enlarged for any reason, the surrounding cerebral tissue (cortex) can be compressed. This may give the erroneous appearance of cerebral atrophy on diagnostic ultrasounds or scans. It is important to carefully review all of the medical record to be certain of the diagnosis.

There are numerous events and disorders which can lead to cerebral atrophy, including fetal stroke, leukodystrophy and other inherited conditions, and congenital infections other than Zika.

Cerebral atrophy can also develop postnatally as a result of brain injury from postnatal intraventricular hemorrhage and other complications of prematurity. Cerebral atrophy that is related to prematurity should not be included in surveillance for birth defects potentially linked to Zika.
Abnormal Cortical Gyral Patterns

Description
The surface of the normal brain has convolutions (gyri) and grooves (sulci), which look like folding of the brain. Changes in the pattern of the gyri and sulci reflect gross abnormalities in the structure of the cerebral (main portion of the brain) cortex. They may involve all or part of one or both cerebral hemispheres. There are several distinct and recognizable patterns of gyral abnormalities, and more than one abnormal pattern may be present in the same brain.

Inclusions
- Lissencephaly/Agyria – The terms mean “smooth brain.” The surface of the brain is smooth with no apparent gyri or only partially formed gyri.
- Pachygyria/Macrogyria/Incomplete lissencephaly – An area of the brain shows a reduced number of gyri which are wider than normal.
- Polymicrogyria – An area of the brain has an excessive number of small gyri.
- Gray matter heterotopia – The term heterotopia means “out of place.” It refers to neurons (brain cells) that have arrested (stopped) in their normal path of migration during brain development.
- Ectopia/Marginal glioneuronal heterotopias/Leptomenigeal heterotopias – Collections of neurons that have migrated beyond their normal limits during brain development.
- Neuronal migration disorder/Neuronal maturation disorder – Abnormal migration of neurons during brain development, which can lead to the various types of gyral malformations and heterotopia.
- Schizencephaly – Abnormal slits or clefts in the brain.
- Minor cortical dysplasias – Subtle disturbances in brain architecture that are more difficult to detect.

Exclusions
- Megalencephaly/Macrencephaly – The brain is abnormally large and heavy. It is thought to result from a disturbance in the regulation of the number of brain cells.

ICD-9-CM Codes
- No specific code; may be included under:
  - 742.2 – Reduction deformities of brain
  - 742.4 – Other specified anomalies of brain

ICD-10-CM Codes
- Q04.3 – Other reduction deformities of brain
- Q04.6 – Congenital cerebral cysts
- Q04.8 – Other specified congenital malformations of brain
CDC/BPA Codes

742.24 – Agyria and lissencephaly
742.25 – Microgyria
742.28 – Other specified reduction defect of brain

Diagnostic Methods

Gold standard: postnatal CT or MRI scan; autopsy or pathology. Abnormal gyral patterns may be suspected on prenatal ultrasound, fetal MRI scan, or postnatal head/brain ultrasound. They cannot be detected by physical exam.

Medical Records – what and where to look for information

Results of prenatal ultrasound or fetal MRI scan; consultation reports by neurologist, geneticist, or other subspecialist; clinicians’ or nurses’ notes; results of postnatal head/brain CT or MRI scan; autopsy or pathology report

Associated Defects / Conditions

A variety of other brain abnormalities may also be present.

Prenatal Diagnoses Not Confirmed Postnatally

Abnormal gyral patterns diagnosed by fetal MRI can be included. Gyral abnormalities suspected by prenatal ultrasound should be confirmed by postnatal evaluation for inclusion.

Additional Information:

During fetal development there are three steps to neuron (brain cell) development: first, the neurons develop and multiply; then they migrate to specific areas of the brain; and finally, they organize to form specific layers of the brain. Interference with any of these steps can result in abnormal migration and abnormal formation of the cerebral cortex. The clinical symptoms observed with these conditions depend on the extent of brain involvement and can range from profound developmental delay to mild dyslexia to none.

Abnormal gyral patterns have been described with fetal alcohol exposure and in a variety of genetic syndromes.
### Corpus Callosum Abnormalities

<table>
<thead>
<tr>
<th>Description</th>
<th>The corpus callosum is a broad band of nerve fibers in the central area of the brain that joins the two cerebral hemispheres. Most abnormalities reflect some degree of failure of development of the corpus callosum.</th>
</tr>
</thead>
<tbody>
<tr>
<td>Inclusions</td>
<td>Agenesis (absence) of the corpus callosum (ACC) – This can be either complete absence or partial absence. Hypoplasia (underdevelopment) of the corpus callosum. Dysgenesis (defective development) of the corpus callosum. Thinning of the corpus callosum.</td>
</tr>
<tr>
<td>Exclusions</td>
<td>--</td>
</tr>
<tr>
<td>ICD-9-CM</td>
<td>No specific code; may be included under: 742.2 – Reduction deformities of brain 742.4 – Other specified anomalies of brain</td>
</tr>
<tr>
<td>ICD-10-CM</td>
<td>Q04.0 – Congenital malformations of corpus callosum</td>
</tr>
<tr>
<td>CDC/BPA Codes</td>
<td>742.21 – Anomalies of corpus callosum</td>
</tr>
<tr>
<td>Diagnostic Methods</td>
<td>Gold standard – Postnatal head/brain ultrasound, CT or MRI scan; autopsy or pathology. Abnormalities of the corpus callosum may be suspected on prenatal ultrasound, fetal MRI scan, or postnatal head/brain ultrasound. Abnormalities of the corpus callosum cannot be detected by physical exam.</td>
</tr>
<tr>
<td>Medical records – What and where to look for information</td>
<td>Results of prenatal ultrasound or fetal MRI scan; consultation reports by neurologist, geneticist, or other subspecialist; clinicians’ or nurses’ notes; results of postnatal head/brain ultrasound, CT or MRI scans; autopsy or pathology report</td>
</tr>
<tr>
<td>Associated defects/Conditions</td>
<td>Corpus callosum abnormalities can be associated with a variety of other brain abnormalities, including microcephaly, macrocephaly, microgyria, pachygyria, or lissencephaly. Brain cysts in the area can block development of the corpus callosum. Abnormalities of the corpus callosum may also be seen with eye anomalies.</td>
</tr>
<tr>
<td>Prenatal Diagnoses Not Confirmed Postnatally</td>
<td>Abnormalities of the corpus callosum suspected prenatally should be confirmed by postnatal evaluation for inclusion.</td>
</tr>
<tr>
<td>Additional Information:</td>
<td>Abnormalities of the corpus callosum can result from congenital infections, chromosomal anomalies, fetal exposures such as alcohol, or blocked growth of the nerve fibers by brain cysts. They can occur in isolation, with other brain anomalies, or as part of a syndrome. Many people with isolated corpus callosum abnormalities appear to function normally and are diagnosed incidentally on procedures undertaken for other reasons.</td>
</tr>
</tbody>
</table>
### Cerebellar abnormalities

<table>
<thead>
<tr>
<th>Description</th>
<th>Inclusions</th>
</tr>
</thead>
<tbody>
<tr>
<td>The cerebellum (&quot;little brain&quot;) is located at the back of the cerebral cortex (the main portion of the brain). It is divided into two hemispheres with a midline structure called the vermis. A variety of congenital abnormalities in its structure have been described.</td>
<td>Cerebellar agenesis – Partial or complete absence of the cerebellum or any of its structures, the vermis, or hemispheres</td>
</tr>
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<td></td>
<td>Cerebellar hypoplasia – Underdevelopment (decreased size) of the cerebellum or any of its structures, the vermis, or hemispheres</td>
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<td></td>
<td>Cerebellar dysplasia – Disorganized development of the cerebellar tissues. This can involve one area or the entire cerebellum</td>
</tr>
<tr>
<td></td>
<td>Cerebellar atrophy – Decrease in size (due to loss of cells) after initial normal development of the cerebellum or any of its structures, the vermis, or hemispheres. This may be difficult to distinguish from hypoplasia if the process occurs early in development.</td>
</tr>
<tr>
<td></td>
<td>Dandy Walker malformation – A constellation of abnormalities that includes hypoplasia of the cerebellar vermis, cystic enlargement of the 4th ventricle (the channel through which cerebrospinal fluid [CSF] flows from the brain to the spinal cord), and enlargement of the posterior fossa (base of the skull that contains the cerebellum). It results from narrowing, absence (atresia), or obstruction of the foramina of Magendie and Luschka (openings in the roof of the fourth ventricle) through which CSF normally flows out of the brain. The obstruction leads to hydrocephalus.</td>
</tr>
<tr>
<td></td>
<td>Dandy Walker Blake continuum/Dandy Walker variant – These terms are sometimes used to denote the presence of a posterior fossa cyst and some degree of cerebellar dysgenesis. When encountering them, carefully review the medical record and abstract all of the specific cerebellar abnormalities described.</td>
</tr>
<tr>
<td></td>
<td>Mega cisterna magna; large or prominent cisterna magna – Excessive prominence of the CSF space posterior to the cerebellum.</td>
</tr>
<tr>
<td></td>
<td>Tectocerebellar dysraphia – Hypoplasia or aplasia of the cerebellar vermis with displacement of the cerebellar hemispheres</td>
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<tr>
<td></td>
<td>Rhombencephalsynapsis – Fusion of the two cerebellar hemispheres and absence of the vermis.</td>
</tr>
<tr>
<td></td>
<td>Cerebellar cyst – A cyst described in any area of the cerebellum, the vermis, or hemispheres which is not part of any of the conditions described above.</td>
</tr>
</tbody>
</table>

### Exclusions

<table>
<thead>
<tr>
<th>Exclusions</th>
</tr>
</thead>
<tbody>
<tr>
<td>Chiari/Arnold-Chiari malformation – Herniation of part of the cerebellum through the foramen magnum into the spinal canal. There</td>
</tr>
</tbody>
</table>
are several types, one of which is often a complication of spina bifida. When present with associated spina bifida, code only as spina bifida. When present without associated spina bifida, code under Other major brain abnormalities (see page 18)

**ICD-9-CM Codes**
No specific code; may be included under:
- 742.2 – Reduction deformities of brain
- 742.4 – Other specified anomalies of brain

**ICD-10-CM Codes**
No specific code; may be included under Q04.3 – Other reduction deformities of brain

**CDC/BPA Codes**
- 742.23 – Anomalies of cerebellum
- 742.31 – Dandy-Walker syndrome

**Diagnostic Methods**
- **Gold standard** – Postnatal CT or MRI scan; autopsy or pathology
- **Prenatal** – Fetal MRI scan
- Postnatal head/brain ultrasound (performed through the anterior fontanelle of the skull) cannot reliably evaluate the posterior fossa containing the cerebellum.
- Cerebellar abnormalities cannot be diagnosed by physical exam, although associated neurologic symptoms may be noted.

**Medical Records** – what and where to look for information
- Results of prenatal ultrasound or fetal MRI scan; consultation reports by neurologist, geneticist, or other subspecialist; clinicians’ or nurses’ notes; results of postnatal CT or MRI scans; autopsy or pathology report

**Associated Defects / Conditions**
- Hydrocephaly (see page 13).
- A variety of other brain abnormalities may also be present, such as agenesis of the corpus callosum (see page 8).

**Prenatal Diagnoses Not Confirmed Postnatally**
Cerebellar abnormalities diagnosed by fetal MRI can be included. Cerebellar abnormalities suspected by prenatal ultrasound should be confirmed by postnatal evaluation for inclusion.

**Additional Information:**
The cerebellum is one of the earliest structures of the brain to develop and its development one of the longest. Hence, the cerebellum is very vulnerable to developmental events.

Cerebellar anomalies are part of a number of genetic syndromes, including Joubert syndrome.
### Porencephaly

**Description**

Porencephaly refers to cysts or cavities within the substance of the brain that become filled with cerebrospinal fluid (the fluid which surrounds the brain and spinal cord). The cysts are not malformations themselves but often a sign of brain injury. Examples of potential causes of such brain injury include infection, trauma, interruption of blood flow to the brain, or hypoxia (lack of oxygen).

**Inclusions**

- Porencephaly
- Porencephalic cyst or cavity
- Encephaloclastic porencephaly
- Developmental porencephaly

**Exclusions**

- Arachnoid cyst
- Cerebral cysts not described as porencephalic (see page 18)
- Choroid plexus cyst

**ICD-9-CM Codes**

- 742.4 – Other specified anomalies of brain

**ICD-10-CM Codes**

- Q04.6 – Congenital cerebral cysts

**CDC/BPA Codes**

- 742.41 – Porencephaly
- 742.42 – Cerebral cysts

**Diagnostic Methods**

Gold standard – Postnatal CT or MRI scan; autopsy or pathology

Prenatal ultrasound; fetal MRI scan; postnatal head/brain ultrasound

Porencephaly cannot be detected by physical exam.

**Medical Records – what and where to look for information**

Results of prenatal ultrasound or fetal MRI scan; consultation reports by neurologist, geneticist, or other subspecialist; clinicians’ or nurses’ notes; postnatal head/brain ultrasound, CT or MRI scan; autopsy or pathology report

**Associated Defects / Conditions**

Porencephaly can be associated with a variety of other brain abnormalities, including microcephaly (see page 3) or macrocephaly (large head), microgyria (see page 7), absence of corpus callosum (see page 8), or absence of the septum pellucidum (a membrane separating the two cerebral hemispheres that is connected to the corpus callosum).

**Prenatal Diagnoses Not Confirmed**

Porencephaly diagnosed by fetal MRI can be included. Porencephaly suspected by prenatal ultrasound should be confirmed by postnatal
Postnatally evaluation for inclusion.

Additional Information:
Porencephalic cysts can occur sporadically or can be familial or genetic. The severity of clinical symptoms varies greatly depending on the size and location of the porencephaly.
### Hydranencephaly

| Description | Hydranencephaly is a condition in which the brain’s cerebral hemispheres (the main portion of the brain) are replaced by cerebrospinal fluid (the fluid that surrounds the brain and spinal cord). The brain stem and cerebellum may be normal. Hydranencephaly is thought to result from a destructive process rather than a primary malformation, and may be an extreme form of porencephaly (see page 11). |
| Inclusions | Hydrancephaly – This can be either bilateral or unilateral |
| Exclusions | -- |

#### ICD-9-CM Codes
No specific code; may be included under:
- 742.3 - Congenital hydrocephalus
- 742.4 – Other specified anomalies of brain

#### ICD-10-CM Codes
No specific code; should be included under Q04.3 – Other reduction deformities of brain

#### CDC/BPA Codes
- 742.32 - Hydranencephaly

#### Diagnostic Methods
Gold standard – Postnatal head/brain ultrasound, CT or MRI scan; autopsy or pathology.
Can be noted on prenatal ultrasound or fetal MRI scan.
Hydranencephaly cannot be diagnosed by physical exam, although associated neurologic symptoms may be noted.

#### Medical Records – what and where to look for information
Results of prenatal ultrasound or fetal MRI scan; consultation reports by neurologist, geneticist, or other subspecialist; clinicians’ or nurses’ notes; results of postnatal head/brain ultrasound, CT or MRI scans; autopsy or pathology report

#### Associated Defects / Conditions
--

#### Prenatal Diagnoses Not Confirmed Postnatally
Hydranencephaly may be included when only diagnosed prenatally. Prenatal findings should be confirmed by postnatal evaluation when possible.
Additional Information:
Hydranencephaly may result from congenital infection or interruption of the blood supply to the brain that disrupts normal development.

Infants may appear normal at birth as the brainstem is intact. Head size is usually normal or maybe enlarged. After a few months, there are indications of developmental delay, increased muscle tone, and seizures. Few children with bilateral hydranencephaly survive past one year. Unilateral hydranencephaly has a much better prognosis with some children having only mild delays.

It is critical to distinguish true hydranencephaly from severe hydrocephalus (see page 13) through careful review of the medical record. In hydrocephalus when the cerebral ventricles are severely enlarged, the cerebral hemispheres may be so compressed as to appear nonexistent. This can be mistaken for hydranencephaly. Severe hydrocephalus can be treated with shunting of the CSF to allow expansion of the cerebral hemispheres. There is no treatment for hydranencephaly.
Ventriculomegaly/Hydrocephaly

**Description**

Ventriculomegaly refers to enlargement of the cerebral ventricles (the cavities within the brain that contain cerebrospinal fluid or CSF) as measured on diagnostic imaging (prenatal or postnatal ultrasound, CT or MRI scan).

Hydrocephaly, or hydrocephalus, refers to an increase in the amount of CSF within the cerebral ventricles, which enlarges their size and increases the pressure within the brain (intracranial pressure). It most commonly results from obstruction to the normal flow of CSF within the brain and spinal cord, but can also result from impaired absorption of CSF by brain tissue.

The distinction between ventriculomegaly and hydrocephalus has not been clearly defined, and these terms can be used interchangeably in medical records.

**Inclusions**

Aqueductal stenosis – Narrowing or obstruction of the aqueduct of Sylvius between the third and fourth ventricles. This is the most common type of obstructive hydrocephalus.

Occlusion of the foramina of Monro – Narrowing or obstruction of the channels that connect the lateral ventricles (the ventricles in the cerebral hemispheres) to the third ventricle in the midline.

Communicating hydrocephalus – Impaired absorption of CSF due to either 1) occlusion of the subarachnoid cisterns around the brainstem or 2) obliteration of the subarachnoid spaces around the exterior of the brain, leading to an increased amount of CSF within the brain.

Hydrocephaly due to other anatomic lesions such as agenesis of the corpus callosum, arachnoid and interhemispheric cysts, or Dandy-Walker malformation.

Hydrocephalus of unspecified type.

Ventriculomegaly that is described as moderate or severe.

Note: For an explanation of hydrocephalus *ex vacuo*, see Other Major Brain Abnormalities on page 18.
Exclusions

For the purpose of surveillance for birth defects potentially linked to Zika, the following should not be included:

- Hydrocephalus diagnosed postnatally that results from a prior intracranial hemorrhage that occurred after delivery. In particular, this may be seen in preterm infants.
- Hydrocephalus that occurs in association with spina bifida or encephalocele. Only the appropriate spina bifida or encephalocele code should be used.
- Hydrocephaly that is associated with bone dysplasias such as achondroplasia (a form of dwarfism).

Colpocephaly – Enlargement of the posterior portion of the lateral ventricles resulting from abnormal development of the posterior part of the cerebral hemispheres.

Ventriculomegaly that is described as mild.

ICD-9-CM Codes

742.3 – Congenital hydrocephalus

ICD-10-CM Codes

Q03.0 – Malformations of aqueduct of Sylvius
Q03.1 – Atresia of foramina of Magendie and Luschka
Q03.8 – Other congenital hydrocephalus
Q03.9 – Congenital hydrocephalus, unspecified

CDC/BPA Codes

742.30 – Anomalies of aqueduct of Sylvius
742.38 – Other specified hydrocephaly
742.39 – Unspecified hydrocephaly

Diagnostic Methods

Gold standard – Postnatal head/brain ultrasound, CT or MRI scan; surgery; autopsy or pathology

Hydrocephalus also can be seen on prenatal ultrasound or fetal MRI scan.

Severe cases may be suspected by physical exam at delivery, but the diagnosis should be confirmed by postnatal imaging.

Medical Records – what and where to look for information

Results of prenatal ultrasound or fetal MRI scan; consultation reports by neurologist, geneticist, or other subspecialist; clinicians’ or nurses’ notes (signs can include sunsetting eyes, tense fontanelle); results of postnatal head/brain ultrasound, CT or MRI scan; surgical notes; autopsy or pathology report

Associated Defects / Conditions

Hydrocephaly itself is not a malformation but a sign of an underlying condition causing increased CSF in the brain. A variety of other brain abnormalities may also be present, such as Chiari II malformation and neural tube defects (spina bifida and encephalocele).
Prenatal Diagnoses Not Confirmed Postnatally

Severe cases may be included when only diagnosed prenatally. However, milder enlargement of the ventricles, when compared with prenatal reference values, may not be of clinical significance. Prenatal findings should be confirmed by postnatal evaluation when possible, and excluded if postnatal imaging studies are normal.

Additional Information

The ventricular system is made up of four ventricles connected by narrow passages – two lateral ventricles within the cerebral hemispheres, the third ventricle in the midline between the two lateral ventricles, and the fourth ventricle located within the brainstem and connected to the third ventricle. CSF normally flows through the ventricles and exits into cisterns that serve as reservoirs at the base of the brain. It bathes the surface of the brain and the spinal cord and is reabsorbed into the bloodstream.

Ventriculomegaly may be described as mild, moderate, or severe. How these designations correlate with the presence of true hydrocephalus, particularly when seen on prenatal ultrasound, has not been clearly defined.

While a child’s head circumference may be increased for age in the presence of hydrocephaly, this measurement alone is not sufficient to make the diagnosis.

Hydrocephalus has a variety of etiologies, including infection, hemorrhage, and tumors as well as anatomic lesions of the brain such as agenesis of the corpus callosum, encephaloceles, cysts, and some bone dysplasias. In many cases, the etiology is not known.

It is critical to distinguish severe hydrocephalus from true hydranencephaly (see page 12) through careful review of the medical record. In hydrocephalus, when the cerebral ventricles are severely enlarged, the cerebral hemispheres may be so compressed as to appear nonexistent. This can be mistaken for hydranencephaly. Severe hydrocephalus can be treated with shunting of the CSF to allow expansion of the cerebral hemispheres. There is no treatment for hydranencephaly.
Fetal Brain Disruption Sequence

Description
Fetal brain disruption sequence is a pattern of congenital abnormalities that include severe microcephaly, overlapping cranial sutures, prominence of the occipital bone, and scalp rugae (excessive folding of the skin). These abnormalities are thought to result from partial disruption of the previously normal fetal brain during the 2nd or 3rd trimester of gestation which leads to significant decrease in intracranial pressure and collapse of the skull.

Inclusions
For inclusion, all components of the fetal brain disruption sequence (microcephaly, overlapping sutures, prominent occipital bone, scalp rugae) must be present.

Exclusions
Abnormally shaped head without associated microcephaly, overlapping sutures, or scalp rugae (e.g., asymmetric head/skull, brachycephaly, plagiocephaly, dolichocephaly, etc.).
Overlapping cranial sutures without associated brain abnormalities or scalp rugae; do not code overlapping sutures if an isolated abnormality.
Prominence or unusual shape of the occipital bone without associated brain abnormalities or scalp rugae; do not code prominence of the occipital bone if an isolated abnormality.

ICD-9-CM Codes
Case finding: There is no specific code for fetal brain disruption sequence. It might be coded as microcephaly or another single brain malformation, or all of the components might be coded individually.
742.1 – Microcephalus
742.4 – Other specified anomalies of brain
742.8 – Other specified anomalies of nervous system
742.9 – Other and unspecified malformations of brain
Defect coding: If fetal brain disruption sequence is present, code every component individually along with any additional brain or other abnormalities described.

Note: For the purpose of surveillance for birth defects potentially linked to Zika, do not include the following abnormalities if there are no coexisting abnormalities of the brain:
756.0 – Anomalies of skull and face bones
757.39 – Other specified anomalies of skin
ICD-10-CM Codes

Case finding: There is no specific code for fetal brain disruption sequence. It might be coded as microcephaly or another single brain malformation, or all of the components might be coded individually.

Q02 – Microcephaly
Q04.8 – Other specified congenital malformations of brain
Q04.9 – Congenital malformation of brain, unspecified

Defect coding: If fetal brain disruption sequence is present, code every component individually along with any additional brain or other abnormalities described.

Note: For the purpose of surveillance for birth defects potentially linked to Zika, do not include the following abnormalities if there are no coexisting abnormalities of the brain:

Q67.4 – Other congenital deformities of skull, face and jaw
Q75.8 – Other specified congenital malformations of skull and face bones
Q75.9 – Congenital malformation of skull and face bones, unspecified
Q82.8 – Other specified congenital malformations of skin

CDC/BPA Codes

Case finding: There is no specific code for fetal brain disruption sequence. It might be coded as microcephaly or another single brain malformation, or all of the components might be coded individually.

742.10 – Microcephalus
742.48 – Other specified anomalies of brain

Defect coding: If fetal brain disruption sequence is present, code every component individually along with any additional brain or other abnormalities described.

Note: For the purpose of surveillance for birth defects potentially linked to Zika, do not include the following abnormalities if there are no coexisting abnormalities of the brain:

fetal brain disruption sequence has not been diagnosed or described:

754.08 – Other specified deformity of skull
754.09 – Unspecified deformity of skull
756.08 – Other specified skull and face bone anomalies
756.09 – Unspecified skull and face bone anomalies
757.39 – Other specified anomalies of skin
757.80 – Other specified anomalies of skin

Diagnostic Methods

Gold standard – Definitive description of all components of the sequence (microcephaly, overlapping sutures, prominent occipital bone, scalp rugae) postnatally by physical exam, with or without confirmation by x-ray, CT or MRI scan. Look for mention of severe microcephaly, overlapping or overriding sutures/cranial bones, collapse of the skull, increased or redundant skin folds or rugae of the
scalp, and excessive scalp skin. Collapse of the skull and associated brain abnormalities may be observed on prenatal ultrasound.

<table>
<thead>
<tr>
<th>Medical records – what and where to look for information</th>
</tr>
</thead>
<tbody>
<tr>
<td>Results of prenatal ultrasound or fetal MRI scan describing the skull and brain abnormalities; consultation reports by neurologist, geneticist, or other subspecialist; clinicians’ or nurses’ notes; head x-ray, CT or MRI scan; autopsy or pathology report</td>
</tr>
</tbody>
</table>

<table>
<thead>
<tr>
<th>Associated Defects / Conditions</th>
</tr>
</thead>
<tbody>
<tr>
<td>Loss or destruction (partial or total) of cortical tissue in the brain</td>
</tr>
<tr>
<td>Paucity or absence of gyri</td>
</tr>
<tr>
<td>Hydranencephaly</td>
</tr>
<tr>
<td>Ventriculomegaly/Hydrocephalus</td>
</tr>
<tr>
<td>Alteration/disruption of the normal pattern of the cerebral ventricles</td>
</tr>
<tr>
<td>Absence of the thalamus and/or basal ganglia</td>
</tr>
</tbody>
</table>

<table>
<thead>
<tr>
<th>Prenatal Diagnoses Not Confirmed Postnatally</th>
</tr>
</thead>
<tbody>
<tr>
<td>These cases can be included when only diagnosed prenatally if there is specific description of the skull abnormalities indicating collapse with associated evidence of severe microcephaly or partial brain destruction. Excess folding of the scalp is sometimes seen on fetal MRI.</td>
</tr>
</tbody>
</table>

<table>
<thead>
<tr>
<th>Additional Information:</th>
</tr>
</thead>
<tbody>
<tr>
<td>The occurrence of fetal brain disruption sequence has rarely been described with other congenital infections and is primarily seen with congenital Zika infection.</td>
</tr>
</tbody>
</table>
### Intraventricular Hemorrhage that occurs in utero

**Description**
Intraventricular hemorrhage (IVH) is bleeding inside or around the cerebral ventricles, the spaces within the brain that contain the cerebral spinal fluid. The bleeding can occur inside the ventricles only or can extend to the surrounding brain tissues. It can occur in small amounts or be extensive enough to enlarge the ventricles or compress the brain tissue. Bleeding in the brain can put pressure on the nerve cells and damage them. Severe damage to the nerve cells can lead to permanent brain injury.

Bleeding from an IVH occurs most commonly in preterm infants during the first days after birth. This is postnatal IVH and is considered a complication of prematurity, not a congenital defect. However, bleeding from an IVH can occur in utero and can lead to enlargement of the ventricles and/or damage to the brain during gestation. Because this occurs prior to delivery, the resulting abnormalities are considered congenital for the purposes of reporting birth defects potentially linked to Zika.

**Inclusions**
Any brain abnormalities that are described as related to in utero IVH. The specific abnormalities can vary depending on the timing during gestation and extent of the bleeding.

**Exclusions**
Postnatal IVH (when the bleeding occurs at some time after birth) is excluded. This is most common in preterm infants. If a postnatal IVH occurs in a full term infant, review the medical record closely to identify any qualifying brain abnormality that might have led to the IVH, but do not code the postnatal IVH itself.

**ICD-9-CM Codes**
- 742.4 – Other specified anomalies of brain
- 742.9 – Unspecified anomaly of brain, spinal cord, and nervous system

*Note: These are the most likely codes for in utero IVH, but any of the individual brain abnormalities might be coded.*

**ICD-10-CM Codes**
- Q04.8 – Other specified congenital malformations of brain
- Q04.9 – Congenital malformation of brain, unspecified

*Note: These are the most likely codes for in utero IVH, but any of the individual brain abnormalities might be coded.*

**CDC/BPA Codes**
- 742.48 – Other specified anomalies of brain
- 742.90 – Unspecified anomalies of brain

*Note: These are the most likely codes for in utero IVH, but any of the individual brain abnormalities might be coded.*
| **Diagnostic Methods** | Gold standard – Postnatal head/brain ultrasound, CT, or MRI scan; autopsy or pathology  
|                       | Prenatal ultrasound or fetal MRI scan  
|                       | IVH cannot be diagnosed by physical exam. |
| **Medical Records – what and where to look for information** | Results of prenatal ultrasound or fetal MRI scan; consultation reports by neurologist, geneticist, or other subspecialist; clinicians’ or nurses’ notes; results of postnatal head ultrasound, CT, or MRI scan; autopsy or pathology report. Look for specific mention of an IVH that occurred or likely occurred *in utero*, during gestation, or before birth. |
| **Associated Defects / Conditions** | Ventriculomegaly  
|                       | Hydrocephalus  
|                       | Cerebral atrophy |
| **Prenatal Diagnoses Not Confirmed Postnataally** | *In utero* IVH may be included only when diagnosed prenatally. Prenatal findings should be confirmed by postnatal evaluation when possible. |
| **Additional Information:** | -- |
### Other Major Brain Abnormalities

<table>
<thead>
<tr>
<th>Description</th>
<th>Inclusions</th>
</tr>
</thead>
<tbody>
<tr>
<td>Other congenital abnormalities of any part of the brain that are not</td>
<td>Absence of the septum pellucidum</td>
</tr>
<tr>
<td>included in other sections of this guide including, but not limited to,</td>
<td>Arnold-Chiari or Chiari malformation – Note: If associated with spina bifida, code only the</td>
</tr>
<tr>
<td>abnormalities of the thalamus, hypothalamus, pituitary, basal ganglia,</td>
<td>spina bifida</td>
</tr>
<tr>
<td>and brainstem.</td>
<td>Septo-optic dysplasia</td>
</tr>
<tr>
<td></td>
<td>Colpocephaly</td>
</tr>
<tr>
<td></td>
<td>Cranial nerve defects</td>
</tr>
<tr>
<td></td>
<td>Periventricular leukomalacia not due to prematurity</td>
</tr>
<tr>
<td></td>
<td>Enlarged or truncated frontal horns</td>
</tr>
<tr>
<td></td>
<td>Bilateral or multiple unilateral (all on the same side) subependymal cysts or pseudocysts</td>
</tr>
<tr>
<td></td>
<td>Hydrocephalus ex vacuo – This is when the damaged brain shrinks and there is a resulting excess</td>
</tr>
<tr>
<td></td>
<td>of CSF. However, the pressure within the brain is normal.</td>
</tr>
<tr>
<td></td>
<td>Atrophy, aplasia, hypoplasia, or dysplasia of any part of the brain not included elsewhere</td>
</tr>
<tr>
<td></td>
<td>Any congenital abnormality of any component of the brain not included elsewhere</td>
</tr>
</tbody>
</table>

| Exclusions                                                                  |
|                                                                             | Choroid plexus cyst                                                                           |
|                                                                             | Arachnoid cyst                                                                                 |
|                                                                             | Isolated (single) subependymal cyst or pseudocyst                                               |
|                                                                             | Brain abnormalities included in other sections of this guide                                   |

| ICD-9-CM Codes                 | 742.2 – Reduction deformities of brain                                                         |
|                               | 742.4 – Other specified anomalies of brain                                                     |
|                               | 742.9 – Unspecified anomaly of brain, spinal cord, or nervous system                          |

| ICD-10-CM Codes                | Q04.0, Q04.3–Q04.9 – Other congenital malformations of brain                                  |
|                               | Q07.00, Q07.02 – Arnold-Chiari syndrome                                                         |

### CDC/BPA Codes

Note: This list includes codes for brain anomalies that have not been specified in other defect categories. There may be conditions with codes specified in other categories that should be included under Other major brain abnormalities. All qualifying brain abnormalities not included in other defect categories should be included here regardless of the coding.
<table>
<thead>
<tr>
<th>Code</th>
<th>Description</th>
</tr>
</thead>
<tbody>
<tr>
<td>742.20</td>
<td>Anomalies of cerebrum</td>
</tr>
<tr>
<td>742.22</td>
<td>Anomalies of hypothalamus</td>
</tr>
<tr>
<td>742.29</td>
<td>Unspecified reduction defect of brain</td>
</tr>
<tr>
<td>742.48</td>
<td>Other specified anomalies of brain</td>
</tr>
<tr>
<td>742.90</td>
<td>Unspecified anomalies of brain</td>
</tr>
</tbody>
</table>

**Diagnostic Methods**

Gold standard – Postnatal head ultrasound, CT, or MRI scan; autopsy or pathology.
Prenatal ultrasound or fetal MRI scan

**Medical Records – what and where to look for information**

Results of prenatal ultrasound or fetal MRI scan; consultation reports by neurologist, geneticist, or other subspecialist; clinicians’ or nurses’ notes; results of postnatal head ultrasound, CT, or MRI scan; autopsy or pathology report. Look for mention of any abnormality of the cerebrum, cerebral hemispheres, cerebellum, thalamus, hypothalamus, corpus callosum, pituitary, basal ganglia, or brainstem.

**Associated Defects / Conditions**

A variety of other brain abnormalities may also be present, including those in other sections of this guide.

**Prenatal Diagnoses Not Confirmed Postnatally**

Many of these abnormalities may be described prenatally. Prenatal findings should be confirmed by postnatal evaluation when possible.

**Additional Information:**

This category is included in order to ascertain congenital brain abnormalities not specifically mentioned in the other defect categories.
## Neural Tube Defects and Other Early Brain Malformations

### Anencephaly/Acrania

| **Description** | Anencephaly – Partial or complete absence of the brain and skull.  
Acrania – Absence of skull bones with some brain tissue present. These conditions may occur with or without co-occurring spina bifida. |

| **Inclusions** | Anencephaly  
Acrania  
Absent brain, with or without skull bones present.  
Craniorachischisis – Anencephaly continuous with an open posterior spinal defect with no meninges covering the nerve tissue (open spina bifida). Can be as limited as the cervical region or as extensive as the entire spine.  
Craniorachischisis with spinal retroflexion – Defect associated with severe flexion of the anterior portion of the spine.  
Exencephaly – Absence of the skull with some protruding brain tissue.  
Iniencephaly – A rare form of anencephaly where the head is bent severely backward, the neck is virtually absent, and the scalp is directly connected to the skin of the back.  
Holoanencephaly – Anencephaly that extends thorough the foramen magnum (involves the entire skull).  
Meroanencephaly – Defect limited to the anterior part of the brain and skull  
NOTE: The distinction between holoanencephaly and meroanencephaly is rarely made in the medical record. |

| **Exclusions** | -- |

| **ICD-9-CM Codes** | 740.0 – Anencephalus  
740.1 – Craniorachischisis  
740.2 – Iniencephaly |

| **ICD-10-CM Codes** | Q00.0 – Anencephaly  
Q00.1 – Craniorachischisis |
Q00.2 – Iniencephaly

**CDC/BPA Codes**

- 740.00 – Absence of brain
- 740.01 – Acrania
- 740.02 – Anencephaly
- 740.03 – Hemianencephaly
- 740.08 – Other anomalies similar to anencephaly
- 740.10 – Craniorachischisis
- 740.20 - 740.29 – Iniencephaly

**Diagnostic Methods**

- Gold standard – Anencephaly is easily recognized on physical examination at delivery and autopsy or pathology

**Medical Records**

- Results of prenatal ultrasound or fetal MRI scan; clinicians’ or nurses’ notes; physical exam; autopsy or pathology report. Look for a description of the infant/fetus after delivery.

**Associated Defects / Conditions**

- Spina bifida that is not continuous with the anencephaly may also be present. Strings of tissue within the amniotic fluid surrounding the fetus may be noted on prenatal ultrasound or after delivery (amniotic band sequence).

**Prenatal Diagnoses Not Confirmed Postnatally**

- Anencephaly may be included when only diagnosed prenatally. However, the prenatal findings should be confirmed by postnatal examination when possible.

**Additional Information:**

Anencephaly is one of a group of defects that result from failure of the neural tube to close (neural tube defects). In most instances, anencephaly is fatal within the first days or weeks after birth. Many cases can be prevented through consumption of folic acid before and during pregnancy. When present during gestation, strings of tissue within the amniotic fluid surrounding the fetus may interfere with growth and formation of the brain leading to anencephaly. This is called amniotic band sequence.

Maternal serum alphafetoprotein (MSAFP) and/or amniotic fluid alphafetoprotein (AFAFP) and amniotic fluid acetylcholinesterase (ACHE) may be elevated with anencephaly during pregnancy since the brain tissue is in contact with the amniotic fluid. However, these screening tests alone are not sufficient to diagnose the condition.
### Encephalocele

#### Description
Herniation of brain tissue and/or meninges (membranes covering the brain) through a defect in the skull. The hernia sac is usually covered by skin.

#### Inclusions
- Cephalocele
- Cranial meningocele – Herniation of meninges only.
- Encephalocele
- Encephalomyelocele - Herniation through a defect in a portion of both the skull and the upper spine.
- Encephalocystomeningocele
- Hydranencephalocele
- Meningoencephalocele
- Ventriculoocele

#### Exclusions
---

#### ICD-9-CM Codes
- 742.0 – Encephalocele

#### ICD-10-CM Codes
- Q01.0 - Q01.9 – Encephalocele

#### CDC/BPA Codes
- 742.00 - 742.09 – Encephalocele

#### Diagnostic Methods
- Gold standard – Postnatal head/brain ultrasound, CT or MRI scan; surgery; autopsy or pathology.
- Prenatal ultrasound or fetal MRI scan.
- Most cases of encephalocele are recognizable on physical examination after delivery but conclusively diagnosed only through imaging or direct visualization at surgery.

#### Medical Records – what and where to look for information
- Results of prenatal ultrasound or fetal MIR; physical exam after delivery; consultation reports by neurologist, geneticist, or other subspecialist; clinicians’ or nurses’ notes; results of postnatal head/brain ultrasound, CT or MRI scan; surgery notes; autopsy or pathology report

#### Associated Defects / Conditions
Strings of tissue within the amniotic fluid surrounding the fetus may be noted on prenatal ultrasound or after delivery (amniotic band sequence).
**Prenatal Diagnoses Not Confirmed Postnatally**

Encephalocele may be included when only diagnosed prenatally. However, the prenatal findings should be confirmed by postnatal evaluation when possible. In addition, the absence of a small encephalocele on prenatal ultrasound does not necessarily mean that it will not be diagnosed after delivery.

**Additional Information:**

Encephalocele is one of a group of defects that result from failure of the neural tube to close (neural tube defects). Some cases may be prevented through consumption of folic acid before and during pregnancy. When present during gestation, strings of tissue within the amniotic fluid surrounding the fetus may interfere with growth and formation of the brain leading to encephalocele. This is called amniotic band sequence. Occipital encephalocele is a component of Meckel-Gruber syndrome.

While encephaloceles that herniate through the visible exterior surface of the skull are most common, internal herniations through the sphenoid, maxillary, or ethmoid bones or the orbit or pharynx are also possible.

Maternal serum alphafetoprotein (MSAFP) and/or amniotic fluid alphafetoprotein (AFAFP) and amniotic fluid acetylcholinesterase (ACHE) usually are not elevated with encephalocele during pregnancy since the brain tissue is covered by skin and not in contact with the amniotic fluid. However, elevation of these screening tests does not necessarily rule out encephalocele.
**Spina Bifida without Anencephaly**

**Description**
Incomplete closure of the vertebral spine (usually posteriorly) through which spinal cord tissue and/or meninges (membranes that cover the spine) herniate.

Spina bifida may co-occur with anencephaly or acrania, either as a continuous or discontinuous defect. Include these cases only under anencephaly/acrania (see page 19).

**Inclusions**
Any of the following defects in which anencephaly/acrania does not coexist:
- Lipomeningocele
- Lipomyelomeningocele
- Meningocele – Herniation of meninges only.
- Meningomyelocele, myelomeningocele – Herniation of both meninges and nerve/spinal cord tissue
- Myelocystocele
- Myelodysplasia
- Myeloschisis
- Open spina bifida – Spina bifida not covered by skin.
- Rachischisis – Open spina bifida without meninges covering the spinal cord tissue
- Spina bifida aperta
- Spina bifida cystica

**Exclusions**
Closed spina bifida – Spina bifida that is covered by skin
- Diastematomyelia
- Diplomyelia
- Hydromyelia
- Spina bifida occulta – Incomplete closure of the spine without external herniation of meninges or spinal cord tissue. This usually is not visible exteriorly and may be asymptomatic.
- Syringomyelia (hydromyelia)
- Tethered spinal cord – Spinal cord tissue that is attached to one of the spinal vertebrae.

**ICD-9-CM Codes**
Any of the following codes without an associated code in the range 740.0 – 740.2 (anencephaly/acrania, see page 19)
- 741.0 – Spina bifida with hydrocephalus
- 741.9 – Spina bifida without mention of hydrocephalus
ICD-10-CM Codes

Any of the following codes without an associated code in the range Q00.0 – Q00.2 (anencephaly/acrania, see page 19)
Q05.0 - Q05.9 – Spina bifida with or without hydrocephalus
Q07.01 – Arnold-Chiari syndrome with spina bifida
Q07.03 – Arnold-Chiari syndrome with spina bifida and hydrocephalus

CDC/BPA Codes

Any of the following codes without an associated code in the range
740.00 – 740.29 (anencephaly/acrania, see page 19)
741.00 - 741.99 – Spina bifida with and without hydrocephalus

Diagnostic Methods

Gold standard – Physical exam; postnatal head/brain ultrasound, CT or MRI scan; surgery; autopsy or pathology
Prenatal ultrasound or fetal MRI scan.
Most instances of spina bifida result in a direct opening on the infant’s back that is easily recognized on physical examination after delivery. However, the exact nature of the defect (meningocele vs. myelomeningocele) may only be distinguished by CT or MRI scan, at surgery, or at autopsy.

Medical records – what and where to look for information

Results of prenatal ultrasound or fetal MRI; physical exam after delivery; consultation reports by neurologist, geneticist, or other subspecialist; clinicians’ or nurses’ notes; results of postnatal x-ray, head/brain ultrasound, CT or MRI scan; surgery notes, autopsy or pathology report

Associated Defects / Conditions

Ventriculomegaly (see page 13)
Hydrocephalus (see page 13)
Arnold-Chiari malformation (see page 18)
Clubfoot (see page 34)
Congenital hip dislocation, developmental dysplasia of the hip (see page 34)
Strings of tissue within the amniotic fluid surrounding the fetus may be noted on prenatal ultrasound or after delivery (amniotic band sequence).

Prenatal Diagnoses Not Confirmed Postnatally

Spina bifida may be included when only diagnosed prenatally.
However, the prenatal findings should be confirmed by postnatal evaluation when possible. In addition, the absence of spina bifida on prenatal ultrasound does not necessarily mean that it will not be diagnosed after delivery.

Additional Information:
Spina bifida is one of a group of defects that result from failure of the neural tube to close (neural tube defects). Many cases may be prevented through consumption of folic acid before and during pregnancy. When present during gestation, strings of tissue within the amniotic fluid surrounding the fetus may interfere with growth and formation of the spine leading to spina bifida. This is called amniotic band sequence.

Spina bifida can occur at any level along the spinal column, from cervical (highest, at the neck) to thoracic, lumbar, and sacral (the lowest). When coding spina bifida, select the code for the highest level at which the spina bifida occurs. If the defect involves more than one level (e.g., cervicothoracic, thoracolumbar, lumbosacral), select the code for the highest level at which the spina bifida occurs. The highest level of involvement determines the degree of associated neurologic impairment.

Open spina bifida (spina bifida cystica, spina bifida aperta) are lesions that have no covering or are covered only by meninges (the membranes that cover the spinal cord). They usually leak cerebrospinal fluid (CSF). Closed lesions are covered by normal skin and do not leak CSF.

Hydrocephalus and Arnold-Chiari malformation of the brain frequently, though not always, result from spina bifida. When present, code only the spina bifida.

Maternal serum alphafetoprotein (MSAFP) and/or amniotic fluid alphafetoprotein (AFAFP) and amniotic fluid acetylcholinesterase (ACHE) may be elevated with anencephaly during pregnancy since the brain tissue is in contact with the amniotic fluid. However, these screening tests alone are not sufficient to diagnose the condition.

Maternal serum alphafetoprotein (MSAFP) and/or amniotic fluid alphafetoprotein (AFAFP) and amniotic fluid acetylcholinesterase (ACHE) may be elevated with spina bifida during pregnancy since the spinal cord tissue is in contact with the amniotic fluid. However, these screening tests alone are not sufficient to diagnose the condition.
Holoprosencephaly/Arhinencephaly

Description
Holoprosencephaly results from variable degrees of incomplete division of the brain into right and left cerebral hemispheres. There are four types which vary in severity: alobar, semi-lobar, lobar, and middle interhemispheric (MIHV). The condition can also affect development of the face and eyes. The most severely affected have one central eye (cyclopia) and a single tubular-shaped nose located above the eye (proboscis).

Inclusions
Alobar holoprosencephaly – Complete lack of division of the cerebral hemispheres, resulting in one single ventricle instead of right and left lateral cerebral ventricles.
Semi-lobar holoprosencephaly – Partial division of the cerebral hemispheres, with absence of the olfactory bulbs, absence of the corpus callosum, and underdeveloped (rudimentary) lobes of the cerebral hemispheres.
Lobar holoprosencephaly – The cerebral hemispheres are mostly divided but remain fused in the front.
Middle interhemispheric variant of holoprosencephaly (MIHV) – Lack of division of the posterior frontal and parietal lobes of the brain.
Arhinencephy – An older term for holoprosencephaly which refers more specifically to structural defects of the olfactory system or nose.
Holotelencephaly – Holoprosencephaly with associated arhinencephaly
Cyclopia – A form of holoprosencephaly where a single, central eye is present.
Cebocephaly – A form of holoprosencephaly where the nose is underdeveloped (e.g., single nostril; proboscis) and closely set eyes (hypotelorism) are present.
Ethmocephaly – A form of holoprosencephaly where the eyes are closely set (hypotelorism), the usual nose is absent, and a proboscis is present.

Exclusions
Arhinencephaly without associated holoprosencephaly

ICD-9-CM Codes
742.2 – Reduction deformities of brain

ICD-10-CM Codes
Q04.1 – Arhinencephaly
Q04.2 – Holoprosencephaly

CDC/BPA Codes
742.26 – Holoprosencephaly
742.27 – Arhinencephaly

**Diagnostic Methods**
Gold standard – Postnatal CT or MRI scan; autopsy or pathology
Prenatal ultrasound or fetal MRI scan; postnatal head/brain ultrasound.
Severe cases may be recognized on physical examination after delivery. However, the exact nature of the defect may only be distinguished by CT or MRI scan, or at autopsy.

**Medical Records – what and where to look for information**
Results of prenatal ultrasound or fetal MRI scan; physical exam after delivery; consultation reports by neurologist, geneticist, or other subspecialist; clinicians’ or nurses’ notes; results of postnatal head/brain ultrasound, CT or MRI scan; surgery notes, autopsy, or pathology report.

**Associated Defects / Conditions**
Associated facial features include cyclopia, proboscis, cebocephaly, ethmocephaly, cleft lip (usually midline), closely set eyes (hypotelorism), and/or absent or very small eyes (anophthalmia, microphthalmia, see page 26).
Associated brain malformations include microcephaly (see page 3), hydrocephalus (see page 13), a single cerebral ventricle, and abnormal gyral patterns (agyria, microgyria, heterotopias, see page 7).

**Prenatal Diagnoses Not Confirmed Postnatally**
Holoprosencephaly may be included when only diagnosed prenatally. However, the certainty of the prenatal diagnosis may vary depending on the type and severity of holoprosencephaly. Prenatal findings should be confirmed by postnatal evaluation when possible.

**Additional Information:**
The different types of holoprosencephaly represent a continuum of anatomic severity. When possible, the specific type should be recorded. Alobar holoprosencephaly is commonly associated with facial anomalies that range from closely set eyes (hypotelorism) and median cleft lip (premaxillary agenesis) to cyclopia (a single central eye in the low frontal area) with absence of the usual nose and a proboscis (tubular-shaped nose located above the eye). Cebocephaly and ethmocephaly represent varying combinations of these facial anomalies.
# Eye Abnormalities

## Microphthalmia/Anophthalmia

**Description**

- **Anophthalmia** – Total absence of eye tissue or apparent absence of the globe of the eye in an otherwise normal orbit.
- **Microphthalmia** – Reduced volume of the eye. The corneal diameter is usually less than 10 millimeters, or the anteroposterior globe diameter is less than 20 millimeters.

Anophthalmia or microphthalmia may affect one or both eyes, or there may be anophthalmia of one eye and microphthalmia of the other.

**Inclusions**

- Anophthalmia
- Microphthalmia
- Nanophthalmia – Microphthalmia with normal internal eye (intraocular) structures. This is a distinct genetic condition.

**Exclusions**

- “Small eyes” or “small palpebral fissures” for which the diagnosis of microphthalmia or anophthalmia has not been made.
- Microcornea with otherwise normal eye size.
- Cryptophthalmos – Failure of the eyelids to form. The eye is totally or partially covered with skin. However, if microphthalmia/anophthalmia or other qualifying eye abnormalities also are present, they should be included.

**ICD-9-CM Codes**

- 743.0 – Anophthalmos
- 743.1 – Microphthalmos

**ICD-10-CM Codes**

- Q11.0 – Cystic eyeball
- Q11.1 – Other anophthalmos
- Q11.2 – Microphthalmos

**CDC/BPA Codes**

- 743.00 – Anophthalmos
- 743.10 – Microphthalmos

**Diagnostic Methods**

- Gold standard – Physical examination after birth by an ophthalmologist; autopsy or pathology report
- These conditions also may be recognized after birth by a neonatologist, geneticist, or other clinician. However, the anteroposterior diameter of the globe can only be measured by postnatal ultrasound, CT or MRI scan, or autopsy.
### Medical Records – what and where to look for information
Clinicians’ exam with close inspection of the eyes; consultation reports by ophthalmologist or geneticist; postnatal ultrasound of the head/brain/eye, CT or MRI scan with measurement of the anteroposterior diameter of the globe; autopsy or pathology report

### Associated Defects / Conditions
- Coloboma of the uvea, iris, choroid, and/or optic nerve (see page 27)
- Anophthalmia and microphthalmia can be associated with a variety of brain abnormalities.

### Prenatal Diagnoses Not Confirmed Postnatally
Anophthalmia and microphthalmia may be suspected on prenatal ultrasound. However, they should not be included in surveillance data without postnatal confirmation. In addition, the absence of anophthalmia or microphthalmia on prenatal ultrasound does not necessarily mean that it will not be diagnosed after delivery.

### Additional Information:
Anophthalmia and microphthalmia are often accompanied by malformations of the brain and face, and frequently are components of genetic syndromes. Ophthalmologic examination of other family members, including the parents, for microphthalmia or blindness may be helpful in determining the cause.
Coloboma

**Description**
A coloboma is an abnormality of the eye where pieces of the eye structure are missing. A coloboma can be present in the iris (the colored part of the eye around the pupil), the lens (the clear structure behind the iris which focuses the light onto the retina), the retina (the light-sensitive tissue in the back of the eye), the choroid (the tissue layer behind the retina which contains the blood vessels), or the optic nerve which carries information from the eye to the brain. Colobomas can be found in one or both eyes.

**Inclusions**
Coloboma of any part of the eye, including the iris, lens, retina, choroid, optic nerve, or disc
Ocular coloboma
Uveoretinal coloboma

**Exclusions**
Coloboma of the eyelids

**ICD-9-CM Codes**
743.36 – Anomalies of lens shape
743.46 – Other specified anomalies of iris and ciliary body
743.47 – Specified anomalies of sclera
743.49 – Other coloboma and anomalies of anterior segment
743.52 – Fundus coloboma
743.56 – Other retinal changes, congenital
743.57 – Specified anomalies of optic disc
743.59 – Other congenital anomalies of posterior segment

**ICD-10-CM Codes**
Q12.2 – Coloboma of lens
Q13.0 – Coloboma of iris
Q14.1 - Q14.8 – Congenital malformations of posterior segment of eye

**CDC/BPA Codes**
743.34 – Coloboma of lens
743.43 – Coloboma of iris
743.48 – Other specified colobomas and anomalies of anterior segment
743.49 – Unspecified colobomas and anomalies of anterior segment
743.51 – Specified anomalies of retina
743.52 – Specified anomalies of optic disc
743.535 – Coloboma of choroid
743.58 – Other specified anomalies of posterior segment
743.59 – Unspecified anomalies of posterior segment

**Diagnostic Methods**
Gold standard - Physical examination, including a retinal exam, after
birth by an ophthalmologist; autopsy or pathology
Colobomas of the iris can be apparent on physical exam after birth.
The pupil appears keyhole-shaped rather than round. Colobomas of
the lens and most posterior structures require examination with an
ophthalmoscope.

<table>
<thead>
<tr>
<th>Medical Records – what and where to look for information</th>
</tr>
</thead>
<tbody>
<tr>
<td>Clinicians’ exam with close inspection of the eyes; consultation reports by ophthalmologist or geneticist; surgery reports, as examination of the eyes may be performed under anesthesia in young infants; autopsy or pathology report</td>
</tr>
</tbody>
</table>

<table>
<thead>
<tr>
<th>Associated Defects / Conditions</th>
</tr>
</thead>
<tbody>
<tr>
<td>Other eye anomalies also may be present, including cataracts (clouding of the lens of the eye), glaucoma (increased pressure inside the eye, also known as buphthalmos), and microphthalmos (see page 26).</td>
</tr>
</tbody>
</table>

<table>
<thead>
<tr>
<th>Prenatal Diagnoses Not Confirmed Postnatally</th>
</tr>
</thead>
<tbody>
<tr>
<td>Colobomas are unlikely to be diagnosed prenatally and should not be included if mentioned only on prenatal ultrasound without postnatal confirmation.</td>
</tr>
</tbody>
</table>

**Additional Information:**
During development, the eye begins as a bud and then folds in on itself leaving a small gap called the fetal cleft. This fetal cleft helps maintain the blood supply during eye development. At the final stage of development, the cleft closes from the back of the eye forward. A coloboma results when the cleft does not close properly.

Colobomas can be part of a genetic syndrome such as CHARGE.
## Congenital Cataract

### Description
A cataract is an opacity of the lens of the eye (the clear structure behind the iris which focuses light onto the retina in the back of the eye). Cataracts can affect any part of the lens, including the anterior, posterior, and zonular segments. Only cataracts that originate before birth should be included.

### Inclusions
- Infantile cataract
- Anterior polar cataract
- Lamellar cataract
- Nuclear cataract
- Posterior lentiglobus/lenticonus cataract
- Posterior cortical cataract
- Sectoral cataract
- Zonular cataract
- Cataract, type not specified

### Exclusions
- Any of the above types of cataract that has its origin after birth.
- Opacities of the cornea (the clear transparent membrane covering the front of the eye over the iris)

### ICD-9-CM Codes
743.30 - 743.34 – Congenital cataract

### ICD-10-CM Codes
Q12.0 – Congenital cataract

### CDC/BPA Codes
743.32 – Cataract

### Diagnostic Methods
Gold standard - Physical examination after birth by an ophthalmologist; autopsy or pathology
Some cataracts are readily apparent on physical examination. Others are only visible with an ophthalmoscope.

### Medical Records – what and where to look for information
Clinicians’ exam with close inspection of the eyes; consultation reports by ophthalmologist or geneticist; surgery reports, as examination of the eyes may be performed under anesthesia in young infants; autopsy or pathology report

### Associated Defects / Conditions
Other eye anomalies also may be present, especially anomalies of the pupils including polycoria (more than one pupil in each eye) and ectopic (off-center) pupils, and anomalies of the lens. Anomalies of the head and central nervous system (brain and spinal cord) also may be present.
Prenatal Diagnoses Not Confirmed Postnatally

Cataracts may be suspected by prenatal ultrasound, but should not be included in surveillance data without postnatal confirmation. In addition, the absence of a cataract on prenatal ultrasound does not necessarily mean that it will not be diagnosed after delivery.

Additional Information:
Factors potentially contributing to congenital cataracts include congenital viral infections, chromosome anomalies, mutations in certain genes associated with cataracts, and a family history of eye defects.
**Intraocular Calcification**

<table>
<thead>
<tr>
<th>Description</th>
<th>Abnormal deposits of calcium in the eye. These are not specific birth defects per se, but signs of injury.</th>
</tr>
</thead>
<tbody>
<tr>
<td>Inclusions</td>
<td>Calcifications in any part of the eye, usually in the anterior segment. Brightly echogenic foci in the eye on ultrasound, CT or MRI scan.</td>
</tr>
<tr>
<td>Exclusions</td>
<td>--</td>
</tr>
</tbody>
</table>

**ICD-9-CM Codes**

<table>
<thead>
<tr>
<th>Code</th>
<th>Description</th>
</tr>
</thead>
<tbody>
<tr>
<td>743.44</td>
<td>Specified anomalies of anterior chamber, chamber angle, and related structures</td>
</tr>
<tr>
<td>743.48</td>
<td>Multiple and combined anomalies of anterior segment</td>
</tr>
<tr>
<td>743.49</td>
<td>Other coloboma and anomalies of anterior segment</td>
</tr>
<tr>
<td>743.54</td>
<td>Congenital folds and cysts of posterior segment</td>
</tr>
<tr>
<td>743.55</td>
<td>Congenital macular changes</td>
</tr>
<tr>
<td>743.56</td>
<td>Other retinal changes, congenital</td>
</tr>
<tr>
<td>743.57</td>
<td>Specified anomalies of optic disc</td>
</tr>
<tr>
<td>743.59</td>
<td>Other congenital anomalies of posterior segment</td>
</tr>
</tbody>
</table>

**ICD-10-CM Codes**

<table>
<thead>
<tr>
<th>Code</th>
<th>Description</th>
</tr>
</thead>
<tbody>
<tr>
<td>Q13.8</td>
<td>Other congenital malformations of anterior segment of eye</td>
</tr>
<tr>
<td>Q13.9</td>
<td>Congenital malformations of anterior segment of eye, unspecified</td>
</tr>
<tr>
<td>Q14.1 - Q14.9</td>
<td>Congenital malformations of posterior segment of eye</td>
</tr>
</tbody>
</table>

**CDC/BPA Codes**

<table>
<thead>
<tr>
<th>Code</th>
<th>Description</th>
</tr>
</thead>
<tbody>
<tr>
<td>743.48</td>
<td>Other specified colobomas and anomalies of anterior segment</td>
</tr>
<tr>
<td>743.49</td>
<td>Unspecified colobomas and anomalies of anterior segment</td>
</tr>
<tr>
<td>743.51</td>
<td>Specified anomalies of retina</td>
</tr>
<tr>
<td>743.52</td>
<td>Specified anomalies of optic disc</td>
</tr>
<tr>
<td>743.58</td>
<td>Other specified anomalies of posterior segment</td>
</tr>
<tr>
<td>743.59</td>
<td>Unspecified anomalies of posterior segment</td>
</tr>
</tbody>
</table>

**Diagnostic Methods**

Gold standard - Physical examination, including retinal exam, after birth by an ophthalmologist; autopsy or pathology
Intraoculat calcifications also might be seen on postnatal brain CT or MRI scan.

**Medical Records – what and where to look for information**

Consultation reports by ophthalmologist or geneticist; postnatal brain CT or MRI scan; surgery reports, as examination of the eyes may be performed under anesthesia in young infants; autopsy or pathology report

Look for mention of calcium deposits or calcification in any part of the eye.

**Associated Defects / Conditions**

There may be associated abnormalities of the optic nerve, choroid or retina.

Intracranial calcifications within the brain can also be described on brain CT or MRI scan.

**Prenatal Diagnoses Not Confirmed Postnatally**

It is unlikely that these abnormalities would be detected by prenatal ultrasound, although they might be seen on a fetal MRI. However, they should not be included without postnatal confirmation.

**Additional Information:**

Intraocular calcifications have been reported very rarely in infants with congenital Zika infection, but have not been well described.
### Chorioretinal Atrophy, Scarring, Pigmentary Changes, Retinitis

**Description**

Changes in the retina (the light-sensitive tissue in the back of the eye) and/or the choroid (the tissue layer behind the retina which contains the blood vessels). The changes are not malformations themselves but a sign of injury from infection, bleeding, hypoxia (lack of oxygen), or other insults to structures in the back two-thirds of the eye.

**Inclusions**

Any abnormality of any part of the choroid, retina, or macula (area of the retina directly across from the pupil where vision is most perfect; its center is the known as the fovea), including but not limited to:

- atrophy
- hypoplasia
- scarring
- calcification
- pigmentary mottling or clumping
- hyperpigmentation
- abnormal blood vessels
- inflammation or infection

**Exclusions**

Retinopathy of prematurity

**ICD-9-CM Codes**

Note: No specific code. This might be coded under the affected part of the eye:

- 743.53 – Chorioretinal degeneration, congenital
- 743.54 – Congenital folds and cysts of posterior segment
- 743.55 – Congenital macular changes
- 743.56 – Other retinal changes, congenital
- 743.57 – Specified anomalies of optic disc
- 743.58 – Vascular anomalies of posterior segment
- 743.59 – Other congenital anomalies of posterior segment

**ICD-10-CM Codes**

Note: No specific code. This might be coded under the affected part of the eye:

- Q14.1 – Congenital malformation of retina
- Q14.2 – Congenital malformation of optic disc
- Q14.3 – Congenital malformation of choroid
- Q14.8 – Other congenital malformations of posterior segment of eye
- Q14.9 – Congenital malformation of posterior segment of eye, unspecified

**CDC/BPA Codes**

Note: No specific code. This might be coded under the affected part of the eye:

- 743.51 – Specified anomalies of retina
- 743.52 – Specified anomalies of optic disk
- 743.53 – Specified anomalies of choroid
743.58 – Other specific anomalies of posterior segment
743.59 – Unspecified anomalies of posterior segment

**Diagnostic Methods**
Gold standard - Physical examination, including retinal exam, after birth by an ophthalmologist; autopsy or pathology

**Medical Records – what and where to look for information**
Consultation reports by ophthalmologist or geneticist; surgery reports, as examination of the eyes may be performed under anesthesia in young infants; autopsy or pathology report. Look for mention of abnormalities of the choroid and/or retina (chorioretinal), or macula (fovea).

**Associated Defects / Conditions**
There may be associated abnormalities, such as atrophy or hypoplasia, etc., of the optic nerve.

**Prenatal Diagnoses Not Confirmed Postnatally**
It is unlikely that these abnormalities would be detected by prenatal ultrasound. They should not be included without postnatal confirmation.

**Additional Information:**
The lining of the back two-thirds of the eye is composed of several layers (see illustration on page 41). The outer layer is the sclera, which is continuous with the cornea at the front of the eye. It is made up of a tough membrane that maintains the shape of the eye. The middle layer is the choroid, which is continuous with the ciliary body and iris at the front of the eye. The choroid is made up mostly of blood vessels with a layer of dark pigmentation. The inner layer is the retina, which is primarily made up by the nerves of the eye. The retina also contains a dark pigmented layer. It is the retina that receives the images of external objects. In the center of the retina posteriorly and directly across from the pupil (the opening in the iris), is an oval yellowish area called the macula. In its center is a depression called the fovea. It is here that the vision of external objects is most perfect.

Chorioretinal changes have been observed in congenital infections other than Zika, most notably toxoplasmosis and cytomegalovirus (CMV).
### Optic Nerve Atrophy, Pallor, other Optic Nerve Abnormalities

**Description**

Abnormalities of the optic nerve that can be seen on eye examination where the optic nerve exits the retina at the back of the eye.

**Inclusions**

Any abnormality of the optic nerve, optic disc, or optic cup including but not limited to:
- atrophy
- hypoplasia
- pallor (pale color)
- increased optic cup to disc ratio
- increased optic disc cupping

**Exclusions**

--

**ICD-9-CM Codes**

743.57 – Specified anomalies of optic disc

**ICD-10-CM Codes**

Q14.2 – Congenital malformation of optic disc  
H47.03 – Optic nerve hypoplasia

**CDC/BPA Codes**

743.52 – Specified anomalies of optic disc

**Diagnostic Methods**

Gold standard - Physical examination, including retinal exam, after birth by an ophthalmologist; autopsy or pathology

**Medical Records – what and where to look for information**

Consultation reports by ophthalmologist or geneticist; surgery reports, as examination of the eyes may be performed under anesthesia in young infants; autopsy or pathology report  
Look for mention of abnormalities of the optic nerve, optic disc, or optic cup.

**Associated Defects / Conditions**

There may be associated abnormalities, such as atrophy, hypoplasia, scarring, etc., of the choroid, retina, or macula (fovea).

**Prenatal Diagnoses Not Confirmed Postnatally**

It is unlikely that these abnormalities would be detected on prenatal ultrasound. They should not be included without postnatal confirmation.

**Additional Information:**


The lining of the back two-thirds of the eye is composed of several layers. The innermost layer is the retina, which is made up mostly of the nerves of the eye. The optic disc is the area of the retina where the optic nerve exits the eye to the brain. It is at the back of the eye slightly to the nasal side of the macula (area of the retina directly across from the pupil where vision is most perfect). In the center of the optic disc is a white depression known as the optic cup. It usually measures about one-third or less of the diameter of the total optic disc.

Optic nerve abnormalities have been observed in congenital infections other than Zika, most notably toxoplasmosis and cytomegalovirus (CMV).
Consequences of Central Nervous System (CNS) Dysfunction

### Arthrogryposis

<table>
<thead>
<tr>
<th>Description</th>
<th>Contracture (abnormal shortening and stiffness of the muscles, tendons, and/or ligaments) of the limbs that is present at birth. Arthrogryposis is not a single disease or diagnosis, but a characteristic appearance of the joints, which can vary from mild to severe. Most of the joints are flexed, but some can be extended. The contractures can be fixed or more flexible, and can involve all or most joints or a single joint. The surrounding muscles can be thin, absent (amyoplasia), or contain excess fibrous tissue (fibrotic).</th>
</tr>
</thead>
<tbody>
<tr>
<td>Inclusions</td>
<td>Distal arthrogryposis – Involves just the hands and feet Arthrogryposis multiplex congenita (AMC) – Involves all joints Multiple pterygia – The contractures are accompanied by webbing of the skin across the affected joint(s) For the purpose of surveillance for birth defects potentially linked to Zika, include contracture of a single joint.</td>
</tr>
<tr>
<td>Exclusions</td>
<td>Posturing of the limbs in the flexed position due to increased muscle or nerve tone (hypertonia). Non-fixed, reducible positioning of the limbs or joints that can easily be moved to their typical neutral position.</td>
</tr>
<tr>
<td>ICD-9-CM Codes</td>
<td>754.89 – Other specified nonteratogenic anomalies</td>
</tr>
<tr>
<td>ICD-10-CM Codes</td>
<td>Q68.8 – Other specified congenital musculoskeletal deformities Q74.3 – Arthrogryposis multiplex congenita</td>
</tr>
<tr>
<td>CDC/BPA Codes</td>
<td>755.80 – Arthrogryposis multiplex congenita</td>
</tr>
<tr>
<td>Diagnostic Methods</td>
<td>Gold standard – Physical examination by a pediatric neurologist, geneticist, or orthopedic specialist. There is no single diagnostic test for arthrogryposis. Prenatal ultrasound of fetal limbs may suggest the diagnosis but is not considered diagnostic. Postnatal procedures that may assist in making the diagnosis include x-rays of the limbs (skeletal survey), muscle or skin biopsy, nerve testing (electromyogram or EMG, nerve conduction velocity or NCV), and CT or MRI scan of the brain; autopsy or pathology.</td>
</tr>
</tbody>
</table>
Medical Records – What and where to look for information

Results of prenatal ultrasound of the limbs or fetal MRI scan; consultation reports by neurologist, geneticist, orthopedist, or other specialists; clinicians’ or nurses’ notes; results of muscle or skin biopsy, nerve testing (electromyogram or EMG, nerve conduction velocity or NCV); results of postnatal CT or MRI scan of brain.; autopsy or pathology report

Associated Defects / Conditions

There may be associated brain or neurologic abnormalities.

Prenatal Diagnoses

Arthrogryposis may be suggested on prenatal ultrasound of the limbs. Prenatal findings should be confirmed by postnatal evaluation when possible.

Additional Information:

Because arthrogryposis is not a single disease or diagnosis, its identification in a newborn can lead to an extensive search for the underlying cause involving multiple subspecialists. Known factors that can contribute to arthrogryposis include lack of fetal movement in utero (fetal akinesia), which can have a variety of causes, chromosome abnormalities such as trisomy 18, and single gene disorders for which distal arthrogryposis is a component. The majority of people with the most common type of arthrogryposis have normal intelligence.
Clubfoot with associated brain abnormalities

Description
An abnormality of the foot consisting of plantar flexion (downward pointing of the foot and toes), inversion (internal rotation, or varus), and metatarsus adductus (deviation of the forefoot toward the body). An abnormally high arch (pes cavus) and midfoot flexion crease are also usually present, and the middle of the foot twists inward. A clubfoot usually cannot be returned to normal position and will interfere with normal walking if not corrected.

Clubfoot can occur alone or with other abnormalities as a consequence of neurologic impairment of the foot during development. For the purpose of surveillance for birth defects potentially linked to Zika, clubfoot should only be included if there are coexisting abnormalities of the brain.

Inclusions
Note: For the purpose of surveillance for birth defects potentially linked to Zika, include the following abnormalities only if there are coexisting abnormalities of the brain:
- Talipes equinovarus – Types include congenital, idiopathic, and neurogenic
- Talipes, not otherwise specified,
- Clubfoot, not otherwise specified.

Exclusions
- Talipes equinovarus
- Talipes calcaneovalgus
- Talipes calcaneovarus
- Talipes calcaneovalgus
- Talipes varus
- Talipes valgus
- Vertical talus
- Metatarsus adductus without the associated components of clubfoot
- Metatarsus varus without the associated components of clubfoot
- Pes varus
- Pes valgus
- Pes planus
- Rocker-bottom foot
- Positional or postural clubfoot

Note: For the purpose of surveillance for birth defects potentially linked to Zika, do not include the following abnormalities if there are no coexisting abnormalities of the brain:
- Talipes equinovarus – Types include congenital, idiopathic, and
neurogenic
Talipes, not otherwise specified,
Clubfoot, not otherwise specified.

**ICD-9-CM Codes**
- 754.51 – Talipes equinovarus
- 754.70 – Talipes, unspecified

**ICD-10-CM Codes**
- Q66.0 - Q66.9 – Congenital deformities of feet

**CDC/BPA Codes**
- 754.50 – Talipes equinovarus
- 754.73 – Clubfoot, not otherwise specified (NOS), but exclude
- 754.735 – Congenital deformities of foot, NOS

**Diagnostic Methods**
Gold standard - Physical examination by an orthopedic specialist or geneticist; autopsy or pathology
Clubfoot can be diagnosed by other clinicians. Prenatal ultrasound and postnatal X-rays of the foot may provide supplemental information but are not necessary for the diagnosis.

**Medical Records – What and where to look for information**
Results of prenatal ultrasound; consultation reports by orthopedics or genetics; clinicians’ and nurses’ notes; postnatal x-ray of the foot; results of surgical procedures; autopsy or pathology report

**Associated Defects / Conditions**
Clubfoot can also be associated with other musculoskeletal abnormalities such as torticollis (shortening of the neck muscle that tilts the head to one side) or developmental dysplasia of the hip (see page 36). It can also be a consequence of neurologic impairment of the foot during development.

**Prenatal Diagnoses Not Confirmed Postnatally**
Clubfoot can be identified or suspected on prenatal ultrasound.
However, it should not be included without postnatal confirmation.

**Additional Information:**
Clubfoot can occur on one foot or on both feet. The calf muscles on the affected side are usually permanently small. While in some instances the affected foot can be moved passively to a normal or near-normal position (so-called positional clubfoot), more commonly there is a component of rigidity, which can be severe.

Clubfoot often occurs alone, but can be associated with other musculoskeletal abnormalities such as torticollis (shortening of the neck muscle that tilts the head to one side) or developmental dysplasia of the hip (see page 36) and with genetic syndromes such as triploidy, Larsen syndrome, or Moebius sequence. Neurogenic clubfoot results from neurologic impairment of the foot during development due to conditions such as spina bifida, arthrogryposis, sacral agenesis, spinal muscular atrophy, and others that cause paralysis.
The terminology describing foot deformities can be confusing. The term “clubfoot” is often used in the medical record to mean talipes equinovarus, but it can also be used to refer to other conditions such as metatarsus adductus or talipes calcaneovarus. Terms used in describing foot deformities include:

- talus – ankle
- pes – foot
- talipes – ankle/foot
- equino – heel elevated (like a horse)
- varus – turned inward
- valgus – turned outward
- dorsi flex – flexed upward
- plantar flex – flexed downward
- adductis – toward midline
- abductis – away from midline
### Congenital Hip Dislocation / Developmental Dysplasia of the Hip with associated brain abnormalities

**Description**

Congenital hip dislocation (also known as developmental dysplasia of the hip or DDH) occurs when the head of the femur (bone of the upper leg) is located outside its normal position in the cup-shaped cavity formed by the hip bone (acetabulum). In some instances, the femur can be passively placed back into position; in others, physical treatment with surgery is required. The depth and shape of the acetabulum can also be abnormal.

Congenital hip dislocation can occur alone or with other abnormalities as a consequence of neurologic impairment during development. For the purpose of surveillance for birth defects potentially linked to Zika, congenital hip dislocation should only be included if there are coexisting abnormalities of the brain.

**Inclusions**

Note: For the purpose of surveillance for birth defects potentially linked to Zika, include the following abnormalities only if there are coexisting abnormalities of the brain:

- Congenital hip dislocation
- Developmental dysplasia of the hip (DDH)
- Teratologic hip dislocation

**Exclusions**

- Flexion deformity or contracture of the hip
- Hip click
- Predislocation of the hip
- Preluxation of the hip
- Subluxation of the hip
- Unstable hip

Note: For the purpose of surveillance for birth defects potentially linked to Zika, do not include the following abnormalities if there are no coexisting abnormalities of the brain:

- Congenital hip dislocation
- Developmental dysplasia of the hip (DDH)
- Teratologic hip dislocation

**ICD-9-CM Codes**

- 754.30 – Congenital dislocation of hip, unilateral
- 754.31 – Congenital dislocation of hip, bilateral
- 754.35 – Congenital dislocation of one hip with subluxation of other hip

**ICD-10-CM Codes**

- Q65.0 - Q65.9 – Congenital deformities of hip
**CDC/BPA Codes**

- 754.30 – Congenital dislocation of hip
- 754.31 – Unstable hip

**Diagnostic Methods**

- Gold standard – Postnatal ultrasound of the hip
- Hip dislocation may be suspected on prenatal ultrasound and is sometimes diagnosed by physical examination or postnatal x-ray of the hip.

**Medical Record – What and where to look**

- Results of prenatal ultrasound; physical examination of the hip after delivery; consultation reports by orthopedics or genetics; clinicians’ and nurses’ notes; postnatal ultrasound or x-ray of the foot; results of surgical procedures; autopsy or pathology report

**Associated Defects/Conditions**

- Congenital hip dislocation can be associated with other musculoskeletal abnormalities such as torticollis (shortening of the neck muscle that tilts the head to one side) or clubfoot (see page 34). It also can be a consequence of neurologic impairment during development.

**Prenatal Diagnoses Not Confirmed Postnatally**

- While this condition may be suspected by prenatal ultrasound, it should not be included in surveillance data without postnatal confirmation. In addition, the absence of hip dislocation on prenatal ultrasound does not necessarily mean that it will not be diagnosed after delivery.

**Additional Information:**

The hip on either side alone can be dislocated, or both hips can be dislocated. The terminology describing congenital hip dislocation has changed over time, and congenital hip dislocation is now more often referred to as hip dysplasia or developmental dysplasia of the hip (DDH). An unstable hip, in which the femoral head may be moved in and out of the acetabulum on physical examination of the newborn, often resolves spontaneously over time in young infants. However, a truly dislocated hip in which the femoral head remains out of the acetabulum for a prolonged period may result in acetabular deformity unless treated. In some instances, the hip joint is already abnormal (dysplastic) at the time of birth, which can result in hip dislocation. Hence, the designation developmental dysplasia of the hip.

The stability of the hip joint may be evaluated on physical examination using the Barlow test or Ortolani maneuver. Pressure is applied to the hip with the knees flexed to attempt to move the head of the femur out of the hip joint or to move it back into normal position in the acetabulum. The presence of either sign indicates a hip dislocation is present. However, the absence of these signs does not necessarily mean that a dislocation is not present. In some instances, the femoral head may be fixed in a dislocated position and cannot be moved in or out of the joint.

Congenital hip dislocation occurs more frequently after footling or breech deliveries and is more common in females than males. It is most often an isolated condition, although it may occur with
generalized skeletal abnormalities and in some genetic syndromes. It can be part of the caudal regression sequence. Some instances of congenital hip dislocation are probably familial.
### Congenital Hearing Loss (unilateral or bilateral)

**Description**
Loss of hearing in one or both ears present at birth or loss that may develop later but is due to infection, genetic causes, or other influences that affected the fetus while *in utero*. Hearing loss can be of two basic types: 1) Sensorineural - Hearing loss that occurs when there is a problem in the way the inner ear or nerve works; 2) Conductive hearing loss - Hearing loss caused by something that stops sounds from getting through the outer or middle ear. Hearing loss can also be of mixed type with both sensorineural and conductive components. It can also result from damage to the inner ear or nerve that results in failure of sound to be organized in a way that the brain can understand (auditory neuropathy).

**Inclusions**
- Sensorineural hearing loss
- Sensory hearing loss
- Neural hearing loss
- Permanent conductive hearing loss
- Mixed hearing loss (mixed conductive and sensory hearing loss)
- Auditory neuropathy
- Auditory neuropathy spectrum disorder
- Auditory dyssynchrony
- Central hearing loss
- External auditory canal atresia
- Aural atresia

**Exclusions**
-transient conductive hearing loss

**ICD-9-CM Codes**
- 389.0 - 389.9 – Hearing loss
- 744.00 - 744.09 – Anomalies of ear causing impairment of hearing
- 794.15 Abnormal Auditory Function Studies

**ICD-10-CM Codes**
- H90.0 - H90.8 and H90.A – Conductive and sensorineural hearing loss
- H91.0 - H91.9 – Other and unspecified hearing loss
- Q16.0 - Q16.9 – Congenital malformations of ear causing impairment of hearing

**CDC/BPA Codes**
- 744.09 – Unspecified anomalies of ear with hearing impairment

Note: The CDC/BPA code does not include hearing loss not associated
with an ear anomaly

**Diagnostic Methods**

Gold standard: Auditory Evoked Potentials (also known as Auditory Brainstem Response or ABR) using frequency-specific stimuli and including air and bone conduction thresholds to determine peripheral hearing levels in infants less than 4-6 months.

Visual Reinforcement Audimetry (VRA) is recommended for behavioral evaluation in children from 4-6 months (corrected for gestational age if preterm) until approximately 24 months. This includes audiologic assessment for ear specific tones and speech stimuli along with Otoacoustic Emissions (OAE) testing and tympanometry or potentially acoustic reflex thresholds. ABR should be performed if the behavioral audiologic evaluation yields conflicting or inconsistent results.

**Medical Records – what and where to look for information**

Often diagnosis is completed in an outpatient setting. Look for consultation reports by audiology, otolaryngology (ENT), genetics, or craniofacial specialist/team; results of postnatal CT or MRI scan of the ear and brain.

**Associated Defects / Conditions**

Craniofacial anomalies

Microtia (small abnormally-shaped ear).

Absence/atresia of the external auditory (ear) canal

Absence/atresia of the ear

**Prenatal Diagnoses Not Confirmed Postnatally**

Hearing loss cannot be diagnosed prenatally.

**Additional Information:**

For the purposes of surveillance for birth defects potentially linked to Zika, it is suggested that ascertainment be limited to congenital hearing loss in infants one year of age or younger.

All infants receive hearing screening soon after birth. This is usually done at the birth hospital before the newborn is discharged, but sometimes may be done later. A failed hearing screen does not diagnose hearing loss, but requires follow up evaluation, which is usually done on an out-patient basis. Verifying a diagnosis of hearing loss may require review of out-patient physician’s and/or audiologist’s records.

Because ABR is not a test of hearing itself but rather a measure of electrophysiologic response to
auditory stimulation, confirmation of hearing perception requires behavioral evaluation as soon as the child is developmentally capable of providing reliable and valid behavioral responses to sound.

Most hearing loss associated with congenital Zika infection is assumed to be sensorineural. Diagnostic ABR is more indicative of possible hearing loss in these children. Results of automated ABR screening and OAE screening or non-ear-specific soundfield studies are not sufficient for a diagnosis of hearing loss. While hearing loss related to congenital Zika infection may be evident on testing at birth, the onset of hearing loss might be delayed or progressive over time in some infants.

A CT or MRI scan of the ear and brain can identify an abnormally formed cochlea, absent or reduced auditory nerve volume (cranial nerve VIII), or malformed or absent auditory cortex or temporal lobe. These would indicate the presence of permanent end organ hearing loss or a disorder of auditory processing. Hearing loss may be part of many genetic syndromes with DNA mutations in genes known to cause hearing loss (e.g., Connexin 26). Prenatal genetic testing potentially could reveal a syndrome that is known to include hearing loss as one of the sequelae.
Figure 1. Brain – Exterior View
Figure 2. Brain - Cross-section View
Figure 3. Skull – Exterior View
Figure 4. Eye – Cross-section View
Glossary of Terms¹⁻⁵

General Terminology

Major anomaly - A congenital abnormality that requires medical or surgical treatment, has a serious adverse effect on health and development, or has significant cosmetic impact. Individual major anomalies occur in less than 1% of the population. Together, they are seen in approximately 3% of births. Examples include cleft lip and tracheo-esophageal fistula.

Minor anomaly - A congenital abnormality that does not require medical or surgical treatment, does not seriously affect health and development, and does not have significant cosmetic impact. Individual minor anomalies generally occur in less than 4% of the population. The presence of multiple minor anomalies in the same child may provide clues to the timing of a prenatal insult and may indicate the presence of an undiagnosed major anomaly, syndrome, or functional deficit. Examples of minor anomalies are listed in Appendix B.

Normal variant - A minor anomaly that occurs in approximately 4% or more of the population. Examples of normal variants include webbing of the second and third toes and a single umbilical artery in an otherwise normal infant.

Terminology related to the formation of major anomalies

Malformation - A major anomaly that arises during the initial formation of a structure, i.e. during organogenesis. For most organs, this occurs during the first 8 weeks after fertilization. The resulting structure may be abnormally formed, incompletely formed, or may fail to form altogether. Examples of malformations include spina bifida and hypoplastic left heart. The term "congenital malformation" is also used more broadly to indicate any major anomaly.

Disruption - A major anomaly that results from alteration of a structure after its initial formation. The resulting structure may have an altered shape and configuration, abnormal division or fusion of its component parts, or loss of parts that were previously present. Examples of disruption defects include intestinal atresia and possibly gastroschisis.

Deformation - A major anomaly that results from molding of part of a structure, usually over a prolonged time, by mechanical forces after its initial formation. Examples of forces that may lead to a deformation include oligohydramnios (diminished amniotic fluid) and intrauterine crowding in twin, triplet, or higher order pregnancies. Examples of deformations include the compression (Potter's) facies seen with bilateral renal agenesis and some instances of clubfoot.

Terminology related to patterns of multiple anomalies occurring in a single child

Syndrome - A pattern of anomalies that form a specific diagnosis for which the natural history and recurrence risk are usually known. Use of the term “syndrome” implies that the anomalies have a common specific etiology. Examples include Beckwith-Weidemann syndrome and Rubinstein-Taybi syndrome.

Sequence - A pattern of anomalies that results from a single primary anomaly or mechanical factor. The presence of the initial anomaly or factor leads to one or more secondary anomalies, which may then lead to one or more tertiary anomalies, etc., in cascade fashion. Examples include Robin sequence (micrognathia; posterior displacement of the tongue; cleft soft palate) and oligohydramnios (Potter’s) sequence (pulmonary hypoplasia; flattened facies; abnormal positioning of the limbs).
Association – A nonrandom pattern of anomalies that occur together more frequently than expected by chance alone, but for which no etiology has been demonstrated. Examples include VACTERL association (vertebral, anal, cardiac, tracheo-esophageal, renal, and limb anomalies) and CHARGE association (colobomas; heart defects; choanal atresia; retarded growth and development and/or central nervous system anomalies; genital anomalies and/or hypogonadism; ear anomalies and/or deafness). Use of the term “association” does not indicate that a specific diagnosis has been made.

Terminology related to tissue and organ formation
Agenesis - Failure of an organ to form.
Dysgenesis - Anomalous or disorganized formation of an organ.
Aplasia - Absence of a tissue or organ due to lack of cell proliferation.
Dysplasia – Disorganized cell structure or arrangement within a tissue or organ.
Hypoplasia - Undergrowth of a tissue or organ due to insufficient proliferation of otherwise normal cells.
Hyperplasia - Overgrowth of a tissue or organ due to excess proliferation of otherwise normal cells.

Terminology related to the timing of gestation and delivery
Embryonic period - The first eight weeks after fertilization during which most, but not all, organs are formed.
Fetal period - The period from the ninth week after fertilization through delivery.
Neonatal (Newborn) period - The first 28 days following delivery of a live born infant.
Prenatal - Before delivery.
Perinatal – Before, during, or after delivery. The exact time period may vary from 20 to 28 completed weeks of gestation through 7 to 28 days after delivery, depending on the context in which the term is used.
Postnatal - After delivery.

Terminology related to pregnancy outcome
Live birth – Spontaneous delivery of an infant that exhibits signs of life, including a heartbeat, spontaneous breathing, or movement of voluntary muscles. Transient cardiac contractions and fleeting respiratory efforts or gasps are not necessarily considered signs of life by all programs.
Fetal death (Stillbirth) – Spontaneous delivery of an infant or fetus at 20 weeks or greater gestation that does not exhibit signs of life. Transient cardiac contractions and fleeting respiratory efforts or gasps are not necessarily considered signs of life by all programs. A late fetal death is a fetal death that occurs at 28 weeks or greater gestation.
Spontaneous abortion (Miscarriage) - Spontaneous delivery of a fetus at less than 20 weeks gestation.
Induced abortion (Elective termination) - The purposeful interruption of pregnancy with the intention other than to produce a live birth and which does not result in a live birth.
Term infant - An infant born after 37 completed weeks and before 42 completed weeks of gestation.
Preterm infant - An infant born before 37 completed weeks of gestation.
Post term infant - An infant born after 42 completed weeks of gestation.
Low birth weight - Birth weight less than 2500 grams, regardless of gestational age.
Very low birth weight - Birth weight less than 1500 grams, regardless of gestational age.
Extremely low birth weight - Birth weight less than 1000 grams, regardless of gestational age.
Neonatal death - Death of a live-born infant within the first 28 days after birth. Early neonatal death refers to death during the first 7 days. Late neonatal death refers to death after 7 days but before 29 days.
Infant death - Death of a live-born infant before 12 months of age.

References