



Baby's Last Name: _____ DOB: _____ Accession #: _____ Sex: _____ Birth Sequence: _____

Mother's Last Name: _____ Mother's First Name: _____

Birth Hospital: _____ Medical Record #: _____

Is a birth defect present in this child? Yes No Referral to CDHI? Yes No Refused Expired

Report Submitted by: _____ Title: _____ Date: _____ Reporting Facility: _____

How was PCP notified of birth defect(s): we are the PCP Discharge Summary Electronic Health Record (EHR-networked) Electronic Medical Record (EMR-single record) Email/Fax Phone Call Other – specify _____

CENTRAL NERVOUS SYSTEM

Anencephalus (Q00.0-Q00.1)

- Q00.0-Anencephaly
- Q00.1-Craniorachischisis

Microcephalus

- Q02-Microcephaly

Spina Bifida

(Q05.0-Q05.9, Q07.01, Q07.03 w/o Q00.0-Q00.1)

- Q05.0-Cervical spina bifida with hydrocephalus
- Q05.1-Thoracic spina bifida with hydrocephalus
- Q05.2-Lumbar spina bifida with hydrocephalus
- Q05.3-Sacral spina bifida with hydrocephalus
- Q05.4 (D)-Unspecified spina bifida with hydrocephalus
- Q05.5-Cervical spina bifida without hydrocephalus
- Q05.6-Thoracic spina bifida without hydrocephalus
- Q05.7-Lumbar spina bifida without hydrocephalus
- Q05.8-Sacral spina bifida without hydrocephalus
- Q05.9 (D)-Spina bifida, unspecified
- Q07.01-Arnold-Chiari syndrome with spina bifida
- Q07.03-Arnold-Chiari syndrome with spina bifida and hydrocephalus

Encephalocele (Q01.0-Q01.9)

- Q01.0-Frontal encephalocele
- Q01.1-Nasofrontal encephalocele
- Q01.2-Occipital encephalocele
- Q01.8-encephalocele of other sites
- Q01.9 (D)-encephalocele, unspecified

Holoprosencephaly

- Q04.2 Holoprosencephaly

If condition not listed, please specify:

- Q0 _____
- Q0 _____

EYE

Anophthalmia/microphthalmia (Q11.0-Q11.2)

- Q11.0- Cystic eyeball
- Q11.1- Other anophthalmos
- Q11.2 (D)- Microphthalmos
- Q12.0-Congenital cataract

If condition not listed, please specify:

- Q1 _____
- Q1 _____

EAR

Anotia/microtia (Q16.0, Q17.2)

- Q16.0-Congenital absence of (ear) auricle
- Q17.2-Microtia

If condition not listed, please specify:

- Q1 _____
- Q1 _____

CARDIOVASCULAR

Aortic Valve Stenosis

- Q23.0 (D)-Congenital stenosis of aortic valve

Atrial Septal Defect

- Q21.1 (D)-Atrial septal defect

Atrioventricular septal defect

- Q21.2-Atrioventricular septal defect

Coarctation of aorta

- Q25.1-Coarctation of aorta

Common truncus

- Q20.0 (D)-Common arterial trunk

Double outlet right ventricle (DORV)

- Q20.1 (D)-Double outlet right ventricle

Ebstein's anomaly

- Q22.5-Ebstein's anomaly

Hyperplastic left heart syndrome

- Q23.4-Hypoplastic left heart syndrome

Interrupted Aortic Arch (IAA) (Q25.2 and Q25.4)

- Q25.2 (D)-Atresia of aorta
- Q25.4-Other congenital malformations of aorta

Pulmonary Valve Atresia and Stenosis (Q22.0 and Q22.1)

- Q22.0-Pulmonary valve atresia
- Q22.1-Congenital pulmonary valve stenosis

Single Ventricle

- Q20.4 (D)-Double inlet ventricle

Tetralogy of Fallot (TOF)

- Q21.3 (D)-Tetralogy of Fallot

Total anomalous pulmonary venus connection (TAPVC)

- Q26.2 (D)-Total anomalous pulmonary venous connection

Transposition of great arteries (TGA) (Q20.3 and Q20.5)

- Q20.3-Discordant ventriculoarterial connection
- Q20.5-Discordant atrioventricular connection

Tricuspid valve atresia and stenosis

- Q22.4 (D)-Congenital tricuspid stenosis

Ventricular septal defect

- Q21.0 (D)-Ventricular septal defect

If condition not listed, please specify:

- Q2 _____
- Q2 _____



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<p>OROFACIAL</p> <p><input type="checkbox"/> Q30.0-Choanal atresia</p> <p>Cleft lip WITH cleft palate (Q37.0-Q37.9)</p> <p><input type="checkbox"/> Q37.0-Cleft hard palate with bilateral cleft lip</p> <p><input type="checkbox"/> Q37.1-Cleft hard palate with unilateral cleft lip</p> <p><input type="checkbox"/> Q37.2-Cleft soft palate with bilateral cleft lip</p> <p><input type="checkbox"/> Q37.3-Cleft soft palate with unilateral cleft lip</p> <p><input type="checkbox"/> Q37.4-Cleft hard and soft palate with bilateral cleft lip</p> <p><input type="checkbox"/> Q37.5-Cleft hard and soft palate with unilateral cleft lip</p> <p><input type="checkbox"/> Q37.8 (D)-Unspecified cleft palate with bilateral cleft lip</p> <p><input type="checkbox"/> Q37.9 (D)-Unspecified cleft palate with unilateral cleft lip</p> <p>Cleft lip alone withOUT cleft palate (Q36.0-Q36.9)</p> <p><input type="checkbox"/> Q36.0- Cleft lip, bilateral</p> <p><input type="checkbox"/> Q36.1- Cleft lip, median</p> <p><input type="checkbox"/> Q36.9 (D)- Cleft lip, unilateral</p> <p>Cleft palate alone (without cleft lip) (Q35.1-Q35.9)</p> <p><input type="checkbox"/> Q35.1-Cleft hard palate</p> <p><input type="checkbox"/> Q35.3-Cleft soft palate</p> <p><input type="checkbox"/> Q35.5-Cleft hard palate with cleft soft palate</p> <p><input type="checkbox"/> Q35.7-Cleft uvula</p> <p><input type="checkbox"/> Q35.9 (D)-Cleft palate, unspecified</p> <p>GASTROINTESTINAL</p> <p>Biliary atresia (Q44.2-Q44.3)</p> <p><input type="checkbox"/> Atresia of bile ducts-Q44.2</p> <p><input type="checkbox"/> Congenital stenosis and stricture of bile ducts-Q44.3</p> <p>Esophageal atresia/tracheoesophageal fistula (Q39.0-Q39.4)</p> <p><input type="checkbox"/> Atresia of esophagus without fistula-Q39.0</p> <p><input type="checkbox"/> Atresia of esophagus w/tracheo-esophageal fistula –Q39.1 (D)</p> <p><input type="checkbox"/> Congenital tracheo-esophageal fistula without atresia-Q39.2</p> <p><input type="checkbox"/> Congenital stenosis and stricture of esophagus-Q39.3</p> <p><input type="checkbox"/> Esophageal web-Q39.4</p> <p>Rectal and large intestinal atresia/stenosis (Q42.0-Q42.9)</p> <p><input type="checkbox"/> Congenital absence, atresia, & stenosis of rectum with fistula-Q42.0</p> <p><input type="checkbox"/> Congenital absence, atresia, & stenosis of rectum without fistula-Q42.1</p> <p><input type="checkbox"/> Congenital absence, atresia, and stenosis of anus with fistula-Q42.2</p> <p><input type="checkbox"/> Congenital absence, atresia, and stenosis of anus without fistula-Q42.3</p> <p><input type="checkbox"/> Congenital absence, atresia, and stenosis of other parts of large intestine-Q42.8</p> <p><input type="checkbox"/> Congenital absence, atresia, and stenosis of large intestine, part unspecified-Q42.9 (D)</p> <p>Small intestine atresia/stenosis (Q41.0-Q41.9)</p> <p><input type="checkbox"/> Congenital absence, atresia, and stenosis of duodenum-Q41.0</p> <p><input type="checkbox"/> Congenital absence, atresia, and stenosis of jejunum-Q41.1</p> <p><input type="checkbox"/> Congenital absence, atresia, and stenosis of ileum-Q41.2</p> <p><input type="checkbox"/> Congenital absence, atresia, and stenosis of other parts of small intestine-Q41.8</p> <p><input type="checkbox"/> Congenital absence, atresia, and stenosis of small intestine, part unspecified-Q41.9</p> <p>If condition not listed, please specify:</p> <p><input type="checkbox"/> Q4 _____</p> <p><input type="checkbox"/> Q4 _____</p>	<p>GENITOURINARY</p> <p>Bladder exstrophy (Q64.10, Q64.19)</p> <p><input type="checkbox"/> Q64.10-Exstrophy of urinary bladder, unspecified</p> <p><input type="checkbox"/> Q64.19 (D)-Other exstrophy of urinary bladder</p> <p>Cloacal exstrophy</p> <p><input type="checkbox"/> Q64.12-Cloacal exstrophy of urinary bladder</p> <p>Congenital posterior</p> <p><input type="checkbox"/> Q64.2-Congenital posterior urethral valves</p> <p>Hypospadias (Q54.0-Q54.9 excluding Q54.4)</p> <p><input type="checkbox"/> Q54.0-Hypospadias, balanic</p> <p><input type="checkbox"/> Q54.1-Hypospadias, penile</p> <p><input type="checkbox"/> Q54.2-Hypospadias, penoscrotal</p> <p><input type="checkbox"/> Q54.3-Hypospadias, perineal</p> <p><input type="checkbox"/> Q54.8-Other hypospadias</p> <p><input type="checkbox"/> Q54.9 (D)-Hypospadias, unspecified</p> <p>Renal agenesis/hypoplasia (Q60.0-Q60.6)</p> <p><input type="checkbox"/> Q60.0-Renal agenesis, unilateral</p> <p><input type="checkbox"/> Q60.1-Renal agenesis, bilateral</p> <p><input type="checkbox"/> Q60.2 (D)-Renal agenesis, unspecified</p> <p><input type="checkbox"/> Q60.3-Renal hypoplasia, unilateral</p> <p><input type="checkbox"/> Q60.4-Renal hypoplasia, bilateral</p> <p><input type="checkbox"/> Q60.5-Renal hypoplasia, unspecified</p> <p><input type="checkbox"/> Q60.6-Potter's syndrome</p> <p>If not listed, please specify:</p> <p><input type="checkbox"/> Q _____</p> <p>CHROMOSOMAL ABNORMALITIES</p> <p>Deletion 22q11</p> <p><input type="checkbox"/> Q93.81-DiGeorge syndrome-deletion 22q11 Velo-cardio-facial syndrome</p> <p>Trisomy 13-Patau's syndrome (Q91.4-Q91.7)</p> <p><input type="checkbox"/> Q91.4-Trisomy 13, non-mosaicism (meiotic nondisjunction)</p> <p><input type="checkbox"/> Q91.5-Trisomy 13, mosaicism (mitotic nondisjunction)</p> <p><input type="checkbox"/> Q91.6-Trisomy 13, translocation</p> <p><input type="checkbox"/> Q91.7 (D)- Trisomy 13, unspecified</p> <p>Trisomy 18-Edward syndrome (Q91.0-Q91.3)</p> <p><input type="checkbox"/> Q91.0-Trisomy 18, non-mosaicism (meiotic nondisjunction)</p> <p><input type="checkbox"/> Q91.1-Trisomy 18, mosaicism (mitotic nondisjunction)</p> <p><input type="checkbox"/> Q91.2-Trisomy 18, translocation</p> <p><input type="checkbox"/> Q91.3 (D)-Trisomy 18, unspecified</p> <p>Trisomy 21-Down syndrome (Q90.0-Q90.9)</p> <p><input type="checkbox"/> Q90.0-Trisomy 21, non-mosaicism (meiotic nondisjunction)</p> <p><input type="checkbox"/> Q90.1-Trisomy 21, mosaicism (mitotic nondisjunction)</p> <p><input type="checkbox"/> Q90.2-Trisomy 21, translocation</p> <p><input type="checkbox"/> Q90.9 (D)-Down syndrome, unspecified</p> <p>Turner Syndrome (Q96.0-96.9)</p> <p><input type="checkbox"/> Q96.0-Karyotype 45,X</p> <p><input type="checkbox"/> Q96.1-Karyotype 45,X iso (Xq)</p> <p><input type="checkbox"/> Q96.2-Karyotype 45,X w/abnormal sex chromosome, except iso (Xq)</p> <p><input type="checkbox"/> Q96.3-Mosaicism, 45,X/46,XX or XY</p> <p><input type="checkbox"/> Q96.4-Mosaicism, 45,X/other cell line(s) w/abnormal sex chromosome</p> <p><input type="checkbox"/> Q96.8-Other variants of Turner's syndrome</p> <p><input type="checkbox"/> Q96.9 (D)-Turner's syndrome, unspecified</p> <p>If condition not listed, please specify: <input type="checkbox"/> Q9 _____</p>
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**Enter this information into the Maven Newborn Screening System, or
If needed fax to: Department of Public Health, Attn: Alessandra Bogacki, at 860-509-7720**



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MUSCULOSKELETAL

Clubfoot (Q66.0, Q66.89)

- Q66.0-congenital talipes equinovarus
- Q66.89-Other specified congenital deformities of feet

Craniosynostosis

- Q75.0-Craniosynostosis

Diaphragmatic hernia (Q79.0, Q79.1)

- Q79.0 (D)-Congenital diaphragmatic hernia
- Q79.1-Other congenital malformations of diaphragm

Gastroschisis

- Q79.3-Gastroschisis

Limb deficiencies (reduction defects) (Q71.0-Q71.9)

- Q71.0-Congenital complete absence of unspecified upper limb
- Q71.01-Congenital complete absence of right upper limb
- Q71.02-Congenital complete absence of left upper limb
- Q71.03-Congenital complete absence of upper limb, bilateral
- Q71.10 (D)-Congenital absence of unspecified upper arm and forearm with hand present
- Q71.11-Congenital absence of right upper arm and forearm with hand present
- Q71.12-Congenital absence of left upper arm and forearm with hand present
- Q71.13-Congenital absence of upper arm and forearm with hand present, bilateral
- Q71.20-Congenital absence of both forearm and hand, unspecified upper limb
- Q71.21-Congenital absence of both forearm and hand, right upper limb
- Q71.22-Congenital absence of both forearm and hand, left upper limb
- Q71.23-Congenital absence of both forearm & hand, bilateral
- Q71.30-Congenital absence of unspecified hand and finger
- Q71.31-Congenital absence of right hand and finger
- Q71.32-Congenital absence of left hand and finger
- Q71.33-Congenital absence of hand and finger, bilateral
- Q71.40 (D)-Longitudinal reduction defect of unspecified radius
- Q71.41-Longitudinal reduction defect of right radius
- Q71.42-Longitudinal reduction defect of left radius
- Q71.43-Longitudinal reduction defect of radius, bilateral
- Q71.50 (D)-Longitudinal reduction defect of unspecified ulna
- Q71.51-Longitudinal reduction defect of right ulna
- Q71.52-Longitudinal reduction defect of left ulna
- Q71.53-Longitudinal reduction defect of ulna, bilateral
- Q71.60-Lobster-claw hand, unspecified hand
- Q71.61-Lobster-claw right hand
- Q71.62-Lobster-claw left hand
- Q71.63-Lobster-claw hand, bilateral
- Q71.90 (D)-Unspecified reduction defect of unspecified upper limb
- Q71.91-Unspecified reduction defect of right upper limb
- Q71.92-Unspecified reduction defect of left upper limb
- Q71.93-Unspecified reduction defect of upper limb, bilateral
- Q71.891-Other reduction defects of right upper limb
- Q71.892-Other reduction defects of left upper limb
- Q71.893-Other reduction defects of upper limb, bilateral
- Q71.899 (D)-Other reduction defects of unspec upper limb

(Q72.0-Q72.9)

- Q72.00-Congenital complete absence of unspecified lower limb
- Q72.01-Congenital complete absence of right lower limb
- Q72.02-Congenital complete absence of left lower limb
- Q72.03-Congenital complete absence of lower limb, bilateral
- Q72.10 (D)- Congenital absence of unspecified thigh and lower leg with foot present
- Q72.11-Congenital absence of right thigh and lower leg with foot present
- Q72.12-Congenital absence of left thigh and lower leg with foot present
- Q72.13-Congenital absence of thigh and lower leg with foot present, bilateral
- Q72.20-Congenital absence of both lower leg and foot, unspecified lower limb
- Q72.21-Congenital absence of both lower leg and foot, right lower limb
- Q72.22-Congenital absence of both lower leg and foot, left lower limb
- Q72.23-Congenital absence of both lower leg and foot, bilateral
- Q72.30-Congenital absence of unspecified foot and toe(s)
- Q72.31-Congenital absence of right foot and toe(s)
- Q72.32-Congenital absence of left foot and toe(s)
- Q72.33-Congenital absence of foot and toe(s), bilateral
- Q72.40 (D)-Longitudinal reduction defect of unspecified femur
- Q72.41-Longitudinal reduction defect of right femur
- Q72.42-Longitudinal reduction defect of left femur
- Q72.43-Longitudinal reduction defect of femur, bilateral
- Q72.50 (D)-Longitudinal reduction defect of unspecified tibia
- Q72.51-Longitudinal reduction defect of unspecified tibia
- Q72.52-Longitudinal reduction defect of unspecified tibia
- Q72.53-Longitudinal reduction defect of unspecified tibia
- Q72.60 (D)-Longitudinal reduction defect of unspecified fibula
- Q72.61-Longitudinal reduction defect of right fibula
- Q72.62-Longitudinal reduction defect of left fibula
- Q72.63-Longitudinal reduction defect of fibula, bilateral
- Q72.70-Split foot, unspecified lower limb
- Q72.71-Split foot, right lower limb
- Q72.72-Split foot, left lower limb
- Q72.73-Split foot, bilateral
- Q72.891-Other reduction defects of right lower limb
- Q72.892-Other reduction defects of left lower limb
- Q72.893-Other reduction defects of lower limb, bilateral
- Q72.899 (D)-Other reduction defects of unspecified lower limb
- Q72.90 (D)-Unspecified reduction defect of unspecified lower limb
- Q72.91-Unspecified reduction defect of right lower limb
- Q72.92-Unspecified reduction defect of left lower limb
- Q72.93-Unspecified reduction defect of lower limb, bilateral

(Q73.0-Q73.8)

- Q73.0-Congenital absence of unspecified limb(s)
- Q73.1-Phocomelia, unspecified limb(s)
- Q73.8 (D)-Other reduction defects of unspecified limb(s)

Omphalocele

- Q79.2-Exomphalos

If condition not listed, please specify:

- Q7 _____
- Q7 _____