Maine CDC
Birth Defects Program

January 1, 2011 - December 31, 2011

Submitted to the Joint Standing Committee on Health and Human Services

2011 Annual Report
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EXECUTIVE SUMMARY
January 2012

Background

Effective May 1999, State of Maine statutory language (Title 22: Health and Welfare: Subtitle: Facilities for Children and Adults; Chapter 1687; Birth effects Program; §8941-§13375) authorized the Department of Health and Human Services to collect information on birth defects in Maine. The Maine CDC Birth Defects Program was established within the Department of Health and Human Services, Maine Center for Disease Control and Prevention, Division of Family Health, Children with Special Health Needs Program. The program started collecting data in 2003.

Purpose

The overall purpose of the Maine CDC Birth Defects Program (BDP), using a public health approach, is to assess the full impact of birth defects on Maine children and their families, to improve access to specialty services for families and locate resources for emotional and economic support which includes referring the infant with a confirmed birth defect to the Department of Education Part C – Children’s Developmental Services (CDS) Program, monitor trends related to prevalence of selected birth defects in Maine and to educate the provider community and the general public on prevention strategies to decrease the incidence of birth defects in Maine.

Highlights

This 2011 annual report summarizes the current activities of the Maine CDC BDP as well as the ongoing and upcoming activities for calendar year 2012. It also shows selected birth defects counts and birth prevalence for the years 2003 – 2009. Some of the activities Maine CDC BDP undertook in 2011:

- Contact families with a baby with a confirmed birth defect by letter to offer support and information regarding birth defects.
- Follow-up with families via telephone call to ensure families had an opportunity to have their questions or concerns addressed.
- Refer babies with a confirmed birth defect to the Part C Agency (CDS).
- Work with Maine CDC WIC staff in the distribution of multivitamins with folic acid to pregnant women as well as survey participants on their use of multivitamins in conjunction with the other New England States.
- Meet regularly with the Maine CDC Environmental Public Health Tracking Unit to discuss specific issues relating to the Environmental Tracking Portal and birth defects.
- Meet regularly with the 2 abstractors that work with the Maine CDC BDP to develop skills and share information pertinent to birth defects and the abstraction process.
• Provide educational materials regarding the prevention of birth defects to parents, health providers and other interested parties.
• Provide educational presentations to Hospital staff as well as other interested parties, including parents, regarding the Maine CDC BDP and prevention activities.
• Meet with the advisory board once to discuss issues relating to the Maine CDC BDP.
• Work in collaboration with the New Hampshire Birth Conditions Program on a research project looking at the relationship between arsenic and birth defects in the two states.
• Amend MBDP rules May 2011 to include the 45 birth defects recommended by the National Birth Defects Prevention Network and U.S. CDC and start to collect data on the expanded list.
• Convene monthly discussions with the epidemiology staff related to birth defects data; currently working on updating the abstraction process and discussing ways to present data which is collected.
• Update the Maine CDC BDP manual as well as program forms.
• Attend the annual National Birth Defects Prevention Network.
• Meet regularly with ChildLINK staff to discuss the database system and work to expand and increase its use.

For more information on the Maine CDC Birth Defects Program:
Contact Diane Haberman, Program Coordinator, diane.haberman@maine.gov or 207-287-8424
http://www.mainehealth.gov/BirthDefects
Maine CDC Birth Defects Program Overview

Background

A birth defect is defined as an abnormal condition that occurs before or at the time of birth. Birth defects include a wide range of abnormalities with varying levels of impact. Some birth defects are serious and can result in death, while others are less severe and can be treated with appropriate medical services. Birth defects may be caused by genetic factors, environmental, drug or medication exposures while others remain unexplained. Birth defects can cause both mental and physical disabilities that affect children and their families for life.\(^1\)

Birth defects affect about one in every 33 babies born in the United States each year. They are the leading cause of infant deaths, accounting for more than 20% of all infant deaths. Babies born with birth defects have a greater chance of illness and long-term disability than babies born without birth defects.\(^2\) In order to prevent birth defects, it is essential to know what types of birth defects are occurring. A population-based birth defects surveillance program that uses multiple sources of data allows a surveillance program to accurately quantify morbidity and mortality, detect temporal trends, and assess the financial burden that birth defects may cause. Many children who survive have a lifetime of major expenses. In addition, specialty medical care, special education, rehabilitation and developmental services are often essential.

The Maine CDC Birth Defects Program (BDP) is committed to fulfilling its mission that all infants with birth defects are identified early and referred to an established network of services, in order for all to achieve optimal health and develop to their fullest potential. The Maine CDC BDP is a comprehensive surveillance program that benefits the residents of Maine through the early identification of infants who have birth defects. Early identification ensures timely and appropriate access to systems of care that are family-centered, culturally competent, and community-based. It is the intention of the Maine CDC BDP to participate fully in epidemiological investigations as a means of informing public policy, to develop prevention strategies in order to reduce birth defects and to assess for timely referrals and follow-up care to reduce mortality and morbidity among children identified with birth defects.

Legislation and Rules

Legislation supporting the Maine CDC Birth Defects Program has and continues to define the purpose of the program. The statutes defining these roles and responsibilities are listed below.

May 1999, Public Law (P.L.) 1322, 22 M.R.S.A. c. 1687, establishes the Maine CDC Birth Defects Program (BDP) within the Department of Health and Human Services. Program rules

\(^1\) Trust for America’s Health “Birth defects and Developmental Disabilities: A Major Public Health Challenge”
\(^2\) http://www.cdc.gov/node.do/id0900f38000dffe
were formally adopted April 2003 outlining reporting responsibilities and access to medical
records. Mandated reporting began May 2003.

**May 2008** – Rules were updated to include 3 more reportable birth defects.

**April 2011** – Rules were amended to include the 45 birth defects recommended by the U.S. CDC
and the National Birth Defects Prevention Network. Referral to the Department of Education
Part C Agency (CDS) was also included.

**Stakeholders**

The following is a brief listing of organizations that have a strong association with the Maine
CDC Birth Defects Program. In addition at the national level the March of Dimes and the
National Birth Defects Prevention Network are key partners.

<table>
<thead>
<tr>
<th>• Consumers</th>
<th>• Early Intervention Agencies</th>
</tr>
</thead>
<tbody>
<tr>
<td>o Parents and families</td>
<td>o Department of Education – Child Development Services</td>
</tr>
</tbody>
</table>

| • External groups involved with screening,      | • Other State Programs                               |
| follow-up and diagnosis                         | o Maine CDC Newborn Hearing Screening Program        |
| o Hospitals and their staff                     | o Maine CDC Newborn Bloodspot Screening Program      |
| o Nurses                                        | o Maine CDC Environmental Health Tracking Unit       |
| o Primary care providers                        | o Maine CDC Data, Research and Vital Statistics      |
| o Specialty physicians                          | o Office of MaineCare                                |
| o Genetic Counselors                            |                                                     |

**State Advisory Committee**

The Committee meets at least annually and at times more frequently. The committee consists of
interested parties including parents, health professionals, outside agencies including the Maine
Chapter of March of Dimes and other state, community and private sector agencies and the
Maine CDC Children with Special Health Needs staff. The group provides consultation to the
Maine CDC BDP on development, implementation and evaluation of program policies,
procedures and activities.

**Summary of Activities**

The Committee met once this past year. Items discussed included the Maine CDC BDP’s
participation in the New England Consortium multivitamin project and the Maine CDC BDP’s
participation with the New Hampshire Birth Conditions Program in investigating the possible
link of arsenic and the development of birth defects. Referral of children confirmed with a birth
defect to Department of Education’s Child Development Services (CDS) and contacting families
by the Maine CDC BDP was also discussed.
Maine CDC Birth Defects Program Summary

Program Description

The Maine CDC Birth Defects Program began developing a birth defects surveillance system in 1999 with funding from the U.S. Centers for Disease Control and Prevention (CDC). The Maine CDC Birth Defects Program is located at the Maine Center for Disease Control and Prevention. The Maine CDC BDP was established to identify newborns with birth effects, ensure that they receive appropriate specialty services and monitor trends.

The Maine CDC Birth Defects Program began passive case ascertainment with confirmation by active case ascertainment on May 1, 2003. Passive case ascertainment with active case ascertainment is an approach whereby the surveillance program receives case reports of birth defects from a variety of data sources. As required by statute those entities licensed under Title 22: Hospitals and Title 32: Licensed Professionals are required to provide or make available health records and information relating to the occurrence of birth defects. Passive data sources include hospital case reports, birth and death certificates, and medical discharge records using ICD-9 codes. Once a case is identified as a possible reportable birth defect, the case is assigned to an abstractor. The abstractor goes to the hospital to review records to confirm the presence of a birth defect; active case ascertainment.

The Maine CDC Birth Defects Program recently updated the listing of reportable birth defects to reflect the birth defects surveillance guidelines developed by the U.S. CDC National Birth Defects Prevention Network. The current listing of reportable birth defects may be found in Appendix A.

The Maine CDC BDP receives medical discharge data electronically from 28 of the 30 birth hospitals. Maine CDC BDP is currently working with 1 of the remaining hospitals to submit data electronically. The hospital is having difficulty with their computer system. The Maine CDC BDP will continue to work on helping them to submit the data electronically. The remaining hospital has a policy that they do not send protected health information electronically and had chosen to submit data in hardcopy. Maine CDC BDP sends each hospital a monthly reminder electronically to submit data.

In order for a case to be considered by the Maine CDC BDP it must meet the following criteria:

- Infant was live born, stillborn or prenatally diagnosed, with a gestational age of greater than 20 weeks,
- Fetuses less than 20 weeks gestation but with a prenatal diagnosis,
- The birth must occur in Maine and the mother must be a Maine resident,
- The diagnosis was made before the infant reached 1 year of age, and
The birth defect is included in the Maine CDC BDP list of reportable birth defects.

Potential cases are identified through weekly downloads of both the electronic birth and infant death certificates and medical record discharge data. Once a potential case is identified, abstraction is performed using a comprehensive electronic abstraction method. Information collected includes the nature and details of the birth defect, demographics, mother’s health history, prenatal information, cytogenic and laboratory data, family history, and when available father’s history, mother’s exposure to illegal drugs, medications, smoking and alcohol exposure.

The data collected by the abstractors is reviewed and entered in the birth defects surveillance and tracking system, called ChildLINK. ChildLINK was built to link existing state information systems with data obtained from hospitals, health care providers and others mandated to report. Once a child is confirmed to have a birth defect the family is notified by mail of services available to them.

Currently, the Maine CDC Birth Defects Program reports on and gathers information on 45 birth defects, see Appendix A for a complete listing of reportable birth defects. These cases are confirmed usually within the first three months after birth. However, this time frame can be longer depending on when the birth defect was reported and the ability of the abstractors to gather the necessary information from the birth hospital. There is also a lag time in verifying a birth defect because, by law, a birth defect can be diagnosed within the first year of life.

**Funding Sources**

The Maine CDC Birth Defects Coordinator position is funded through the Maternal and Child Health Block Grant. Abstractor positions are funded in part by a U.S. Center for Disease Control and Prevention Environmental Health Tracking Grant and state general fund.

**Personnel**

The Maine CDC Birth Defects Program consists of one full-time Coordinator and two part-time contractors. The Maine CDC BDP contracts with Maine Medical Center and a private individual for part-time abstraction services.

**Goals, Activities and Achievements**

The Maine CDC Birth Defects Program (BDP) gathers data about infants born each year with certain birth defects diagnosed within the first year of life. The statute requires that the “Program”:

- Collect, analyze and distribute information to identify birth defects with regards to the following: causes, risk factors and strategies for prevention and the provision of services,
- Establish a system for data collection that identifies prevalence and incidence rates by region and population group and identifies the morbidity and mortality rates resulting from birth defects, and
◊ Contact families to provide information about available resources and services,
◊ Conduct investigations to determine the nature, and extent of the disease or known or suspected causes of birth defects. Examples of investigation include:
  
  - Maine collects data on 45 birth defects and collects further information through the abstraction process on 22 of those birth defects. Maine CDC BDP is in the process of expanding the abstraction process to include all 45 conditions. The Maine CDC BDP submits data annually to the National Birth Defects Prevention Network and U.S. CDC.
  - The Maine CDC BDP works in collaboration with the Maine CDC Environmental Health Tracking Unit to show 12 of the 22 birth defects on the Environmental Health Tracking Portal.
  - The Maine CDC BDP connects with a family once a baby is identified with a confirmed birth defect with an initial letter. The family is then contacted by phone to follow-up to identify any unmet needs and to connect them to the resources that they may need. The baby identified with a confirmed birth defect is also referred to the Department of Education Part C agency (CDS) for follow-up for early intervention services.
  - The Maine CDC BDP is currently working with the New Hampshire Birth Conditions Program in investigating the effects of arsenic on the development of birth defects.

**Maine CDC Birth Defects Tracking System**

*Tracking System*

Maine CDC DBP began collaborating with the University of Maine at Orono in 2001 to develop and implement a comprehensive surveillance and tracking system. Today, ChildLINK tracks approximately 13,000 infants born in Maine each year. ChildLINK links birth defect data with multiple data sources that include birth and death certificates, hospital discharge date, metabolic screen data, and newborn hearing screening data. By linking information from these existing data sources ChildLINK can: 1) help ensure that children with birth defects and their families receive information about resources and services that may be of assistance; and, 2) provide valuable public health data to state and national policy makers. ChildLINK maintains security/confidentiality of all records by assigning permission to access the system on an individual basis. Access is monitored by Symantec on a 24/7 basis.

*Process Overview*

Once a month hospitals send in a discharge report reflecting the previous month’s birth defects data identified at the hospital. An automatic reminder is sent from ChildLINK to the hospital contact person if the data is not sent in by the 15th of the month. This information is put into an excel format and then submitted electronically to the ChildLINK database system. Once the system receives the report, the report is reviewed for proper formatting and then uploaded to the ChildLINK system. The data is then linked to the birth certificate of the corresponding child.
Statistical Reports

Birth Defects data is submitted annually to the National Birth Defects Prevention Network and U.S. CDC. The most recent data years are below. At the end of the calendar year 2011, data will be available covering 2004 – 2008. The 15-month lag time in data being available is based on the definition of a birth defect. By definition, a birth defect meets criteria and is included in the Maine CDC Birth Defects count if the birth defect is identified within the first year of life. This means that the Maine CDC BDP may still be gathering data as late as December of the year after the requested year. Consequently time is needed to abstract the data necessary to confirm the birth defect. The 2010 birth defects data will not be available until March 2012.

Secondly, the U.S. CDC requests data for a specific span of years and once requested, the data is not published until the end of the request year. The 2009 birth defects data has been sent to the U.S. CDC.
(Prevalence per 10,000 Live Births)

<table>
<thead>
<tr>
<th>Defect</th>
<th>Non-Hispanic White</th>
<th>Non-Hispanic Black or African</th>
<th>Hispanic</th>
<th>Asian or Pacific Islander</th>
<th>American Indian or Alaskan Native</th>
<th>Total**</th>
<th>Notes (+)</th>
</tr>
</thead>
<tbody>
<tr>
<td>Anencephalus</td>
<td>6</td>
<td>0.93</td>
<td>0</td>
<td>0.00</td>
<td>0.00</td>
<td>1.17</td>
<td></td>
</tr>
<tr>
<td>Cleft lip with and without cleft palate</td>
<td>56</td>
<td>8.70</td>
<td>0.00</td>
<td>0.00</td>
<td>27.99</td>
<td>9.07</td>
<td></td>
</tr>
<tr>
<td>Cleft palate without cleft lip</td>
<td>56</td>
<td>8.70</td>
<td>0.00</td>
<td>0.00</td>
<td>18.90</td>
<td>8.34</td>
<td></td>
</tr>
<tr>
<td>Coarctation of aorta</td>
<td>28</td>
<td>4.35</td>
<td>0.00</td>
<td>0.00</td>
<td>0.00</td>
<td>4.10</td>
<td></td>
</tr>
<tr>
<td>Common truncais</td>
<td>3</td>
<td>0.47</td>
<td>0.00</td>
<td>0.00</td>
<td>0.00</td>
<td>0.44</td>
<td>+</td>
</tr>
<tr>
<td>Down syndrome (Trisomy 21)</td>
<td>70</td>
<td>10.87</td>
<td>0.00</td>
<td>0.00</td>
<td>18.90</td>
<td>11.56</td>
<td></td>
</tr>
<tr>
<td>Encephalocele</td>
<td>2</td>
<td>0.21</td>
<td>0.00</td>
<td>10.81</td>
<td>0.00</td>
<td>0.59</td>
<td></td>
</tr>
<tr>
<td>Gastroschisis</td>
<td>33</td>
<td>5.13</td>
<td>0.00</td>
<td>0.00</td>
<td>0.00</td>
<td>5.12</td>
<td>+</td>
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<td>Hypoplastic left heart syndrome</td>
<td>18</td>
<td>2.80</td>
<td>0.00</td>
<td>0.00</td>
<td>0.00</td>
<td>2.97</td>
<td></td>
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<tr>
<td>Omphalocele</td>
<td>14</td>
<td>2.17</td>
<td>0.00</td>
<td>0.00</td>
<td>0.00</td>
<td>2.49</td>
<td>+</td>
</tr>
<tr>
<td>Pulmonary valve atresia and stenosis</td>
<td>10</td>
<td>0.15</td>
<td>0.00</td>
<td>0.00</td>
<td>0.00</td>
<td>0.29</td>
<td></td>
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<tr>
<td>Spina bifida without anencephalus</td>
<td>19</td>
<td>2.95</td>
<td>0.00</td>
<td>0.00</td>
<td>0.00</td>
<td>3.07</td>
<td></td>
</tr>
<tr>
<td>Tetralogy of Fallot</td>
<td>21</td>
<td>3.26</td>
<td>0.00</td>
<td>10.81</td>
<td>0.00</td>
<td>3.51</td>
<td></td>
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<tr>
<td>Transposition of great arteries</td>
<td>25</td>
<td>3.88</td>
<td>0.00</td>
<td>0.00</td>
<td>0.00</td>
<td>3.88</td>
<td></td>
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<tr>
<td>Tricuspid valve atresia and stenosis</td>
<td>1</td>
<td>0.15</td>
<td>0.00</td>
<td>0.00</td>
<td>0.00</td>
<td>0.15</td>
<td></td>
</tr>
<tr>
<td>Total Live Births</td>
<td>64379</td>
<td>1244</td>
<td>925</td>
<td>1072</td>
<td>529</td>
<td>68350</td>
<td></td>
</tr>
<tr>
<td>Total Male Live Births</td>
<td>33187</td>
<td>650</td>
<td>485</td>
<td>557</td>
<td>282</td>
<td>35264</td>
<td></td>
</tr>
</tbody>
</table>

*Hypospadias: prevalence per 10,000 male live births
**Total includes other and unknown race

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# Maine Trisomy Counts and Prevalence by Maternal Age 2003-2007
(Prevalence per 10,000 Live Births)

<table>
<thead>
<tr>
<th>Defect</th>
<th>&lt;35</th>
<th>35 and &gt;</th>
<th>Total**</th>
</tr>
</thead>
<tbody>
<tr>
<td>Down syndrome (Trisomy 21)</td>
<td>40</td>
<td>39</td>
<td>79</td>
</tr>
<tr>
<td>Total Live Births</td>
<td>58744</td>
<td>9603</td>
<td>68380</td>
</tr>
</tbody>
</table>

**Total includes unknown age

### Notes (+)
1. Common truncus: Truncus arteriosus
2. Gastroschisis and Omphalocele: Record abstraction confirms the diagnosis
3. Unless otherwise noted, birth defect counts include live births plus stillbirths
4. Case finding is limited to babies born in Maine to Maine residents
5. Race/ethnicity is based on maternal race and ancestry fields on the birth certificate
6. Total live birth counts and male live birth counts are of live births born in Maine to Maine residents
7. Terminations are not available
Public Awareness

Education and Informational Materials

The Maine CDC Birth Defects Program has a range of materials available, including brochures that are sent to interested parties and stakeholders. Topics include prevention information regarding folic acid use, information on the various birth defects and other topics of interest regarding birth defects.

Website


Maine CDC Birth Defects Program

Accomplishments and Future Direction

Accomplishments

- Initiated follow-up with families by phone after an initial letter was sent to them to offer support.
- Worked collaboratively with the other New England states and the WIC program to determine the use of multivitamins with folic acid use in women participating in the WIC program. This project will be completed at the end of the year. (2011)
- Expanded the list of reportable birth defects to the 45 birth defects recommended by the National Birth Defects Prevention Network and U.S. CDC.
- Continued arsenic and birth defects project with the New Hampshire Birth Condition Program.
- Referred 104 babies with confirmed birth defects to the Part C agency (CDS)
- Continued to meet regularly with abstractors to discuss ways to make the abstraction process more complete and to discuss other issues related to birth defects.
- Continued to educate the provider community by going to birth hospitals and sharing information regarding birth defects and the Maine CDC Birth Defects Program in collaboration with the Maine CDC Cleft Lip and Palate Program Director.

Future Direction

- Expand the ability to follow-up with families to offer support by phone on a timely basis.
- Continue to work in collaboration with the other New England States on projects of mutual interest.
- Expand the number of birth defect cases being abstracted to include all 45 birth defects.
- Continue to explore ways to share information with stakeholders and interested parties in regards to the information gathered through the abstraction process including risk factors.
- Continue to explore ways to share prevention information with stakeholders and interested parties.
• Continue to work with the Maine CDC Environmental Health Tracking Unit to expand the number of birth defects being reported on the portal.
• Expand the number of times the Birth Defects Advisory Committee meets from 1 time a year to 3 to 4 times a year.
Appendix A – Maine CDC Birth Defects Program
Reportable Birth Defects

Central Nervous System

- **Anencephalus** - Partial or complete absence of the brain and skull.
- **Spina Bifida without Anencephalus** - Incomplete closure of the vertebral spine (usually posteriorly) through which spinal cord tissue and/or the membranes covering the spine (meninges) herniate.
- **Hydrocephalus without Spina Bifida** – An increase in the amount of cerebrospinal fluid (CSF) within the brain resulting in enlargement of the cerebral ventricles and increased intracranial pressure.
- **Encephalocele** - Herniation of brain tissue and/or meninges through a defect in the skull. The hernia sac is usually covered by skin.
- **Microcephalus** – A cranial vault that is smaller than normal for age.

Eye

- **Anophthalmia** – Total absence of eye tissue or apparent absence of the globe in an otherwise normal orbit.
- **Microphthalmia** – Reduced volume of the eye.
- **Congenital cataract** – An opacity of the lens of the eye that has its origin prenatally.
- **Aniridia** – Hypoplasia of the iris of both eyes.

Ear

- **Anotia** – Total absence of the external ear and canal
- **Microtia** – Malformation or hypoplasia of the external ear (auricle, pinna)

Cardiovascular

- **Common Truncus (Truncus Arteriosus or TA)** - Failure of separation of the aorta and the pulmonary artery, resulting in a single common arterial trunk carrying blood from the heart to both the body and lungs.
- **Transposition of the Great Arteries (TGA)** - 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).
- **Tetralogy of Fallot** - The simultaneous presence of a ventricular septal defect (VSD), pulmonic stenosis, a malpositioned aorta that overrides the ventricular septum, and right ventricular hypertrophy.
- **Ventricular septal defect** – An opening in the septum that separates the left and right ventricles of the heart.
- **Atrial septal defect** – An opening in the septum that separates the left and right atria of the heart.
Endocardial cushion defect – A defect in both the lower portion of the atrial septum and the upper portion of the ventricular septum, producing a large opening (canal) in the central part of the heart.

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.

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.

Ebstein's anomaly – Downward displacement of the tricuspid valve into the right ventricle.

Aortic valve stenosis – Obstruction or narrowing of the aortic valve, which may impair blood flow from the left ventricle to the aorta.

Hypoplastic Left Heart Syndrome (HLHS) - A condition in which the structures on the left side of the heart and the aorta are extremely small. Classically, this condition includes hypoplasia of the left ventricle, atresia or severe hypoplasia of the mitral and aortic valves, and hypoplasia and coarctation of the aorta.

Patent ductus arteriosus – Abnormally persistent blood flow through the ductus arteriosus beyond the first few days of life.

Coarctation of the Aorta - Narrowing of the descending aorta, which may obstruct blood flow from the heart to the rest of the body.

Orofacial

Cleft Palate without Cleft Lip - 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.

Cleft Lip with and without Cleft Palate - A defect in the upper lip resulting from incomplete fusion of the parts of the lip.

Choanal atresia – Congenital obstruction of the opening of the nasal cavity into the nasopharynx on either side.

Gastrointestinal

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.

Rectal and large intestinal atresia/stenosis – Complete or partial occlusion of the lumen of one or more segments of the large intestine and/or rectum.

Pyloric stenosis – hypertrophy (thickening) of the muscles of the pylorus connecting the stomach to the duodenum, resulting in complete or partial obstruction of the passage of food and gastric contents.
Hirshsprung’s disease (congenital megacolon) – Absence of the parasympathetic ganglion nerve cells (aganglionosis) of the wall of the colon or rectum, which may result in congenital megacolon. Megacolon – enlargement of the diameter of part or all of the colon.

Biliary atresia – Congenital absence of the lumen of the extrahepatic bile ducts.

Genitourinary

Renal agenesis – Complete absence of the kidney.

Renal hypoplasia - Incomplete development of the kidney.

Bladder exstrophy – 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.

Obstructive genitourinary defect – Partial or complete obstruction of the flow of urine at any level of the genitourinary tract from the kidney to the urethra.

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.

Epispadias – Displacement of the opening of the urethra (urethral meatus) dorsally and proximally (on the top and closer to the body) in relation to the tip of the glans of the penis.

Musculoskeletal

Reduction Deformity, Upper Limbs - Complete or partial absence of the upper arm (humerus), lower arm (radius and/or ulna), wrist (carpals), hand (metacarpals), or fingers (phalanges).

Reduction Deformity, Lower Limbs - Complete or partial absence of the upper leg (femur), lower leg (tibia and/or fibula), ankle (tarsals), foot (metatarsals), or toes (phalanges).

Gastroschisis - 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.

Omphalocele - 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.

Congenital hip dislocation – Location of the head of the femur (bone of the upper leg) outside its normal location in the cup-shaped cavity formed by the hip bones (acetabulum).

Diaphragmatic hernia – Incomplete formation of the diaphragm through which a portion of the abdominal contents herniated into the thoracic cavity.

Chromosomal

Trisomy 13 – The presence of three copies of all or a large part of chromosome 13.

Down Syndrome (Trisomy 21) - The presence of three copies of all or a large part of chromosome 21.

Trisomy 18 – The presence of three copies of all or a large part of chromosome 18

Other

Fetal alcohol syndrome – A spectrum of abnormalities resulting from exposure to alcohol in utero. While the specific abnormalities vary among individuals, the hallmarks include growth deficiency, microcephaly, facial dysmorphisms, and neurodevelopmental abnormalities.
◊ **Amniotic bands** – Strands of tissue that float in the amniotic fluid as a consequence of tears or ruptures in the amniotic membrane which surrounds the fetus during development.