Louisiana Birth Defects Monitoring Network

2024 Annual Legislative Report

Prepared by:

Bureau of Family Health

Office of Public Health

December 2024

LOUISIANA DEPARTMENT OF HEALTH

Submitted to:

- Louisiana Legislature
- Louisiana Department of Health, Office of the Secretary
- Louisiana Birth Defects Monitoring Network Advisory Board

By:

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Acknowledgements:

This report was compiled and written by the Louisiana Department of Health, Office of Public Health, Bureau of Family Health staff responsible for the Louisiana Birth Defects Monitoring Network. Amy Zapata, MPH, is the director of the Bureau of Family Health. Key Bureau of Family Health contributors include Louisiana Birth Defects Monitoring Network Program Manager Julie Johnston and Senior Epidemiologist Tri Tran, MD, MPH.

The Louisiana Birth Defects Monitoring Network's surveillance system and public health actions, as described in this report, fulfill the legislative mandate of Louisiana Revised Statutes Title 40, Part VII, Sections 31.41–31.48 to maintain "a system to collect, analyze and disseminate data regarding birth defects in the state and to provide information to families of children born with birth defects regarding services available in their community and the development of appropriate prevention programs."

We recognize the team at the Bureau of Family Health whose dedication and hard work made this report possible, including the data collection specialists and case review coding specialist who abstracted the medical records to collect these data, and the communications staff who edited and designed the report.

We are thankful for the volunteer members of the Louisiana Birth Defects Monitoring Network Advisory Board who provide clinical expertise for the operations and management of the birth defects surveillance system.

Lastly, we honor the families of children impacted by birth defects represented in this report. It is our sincere hope that the activities of the Louisiana Birth Defects Monitoring Network will improve the systems of care serving Louisiana's families.

The federal Title V Maternal and Child Health (MCH) Block Grant provides funding for Louisiana Birth Defects Monitoring Network surveillance activities. The Health Resources and Services Administration (HRSA) of the U.S. Department of Health and Human Services (HHS), as part of an award totaling $31,634,077, with 58 percent financed with non-governmental sources, supported this publication. The contents are those of the author(s) and do not necessarily represent the official views of, nor an endorsement, by HRSA, HHS, or the U.S. Government. For more information, please visit HRSA.gov.

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Executive Summary

The Louisiana Department of Health, Office of Public Health, Bureau of Family Health is responsible for surveillance of birth defects in Louisiana's children and moving that data to action by informing recommendations of strategies to improve Louisiana's maternal and child health systems from preconception through referrals to resources post-diagnosis. This public health monitoring system is authorized and mandated through the Louisiana Legislature in LA R.S. 40:31.45 and is guided by a statutorily defined advisory board.

Of the 170,799 children born in Louisiana between 2019 and 2021, 4,694 children or 2.75% were diagnosed with at least one birth defect. According to the Centers for Disease Control and Prevention (CDC), the national average is about 3% of all babies born each year. Fortunately, Louisiana falls below the national average for children born with birth defects. However, deaths due to congenital anomalies or birth defects were one of the top four leading causes of death in infants from birth to one year of age in Louisiana from 2019-2021.

Among Louisiana children with birth defects born in 2019-2021, cardiovascular system defects were the most common (about 65.4%). The six most common specific birth defects overall were atrial septal defect (a heart defect), hypospadias (a penile defect), ventricular septal defects (heart defects), pulmonary valve atresia/stenosis (a heart defect), clubfoot (a musculoskeletal defect) and Down syndrome (a chromosomal defect).

There is not a single cause of birth defects and not all birth defects are preventable. Scientists think most birth defects are attributable to a combination of factors involving genes, maternal behaviors and environmental exposures. Birth defects require specialized care at birth, in infancy, during childhood and sometimes beyond. Pediatric specialists are located at five pediatric hospitals in Louisiana. Each hospital varies in the range of specialists and services available. For example, although pediatric cardiologists are available in every region of the state, patients must travel to Children's Hospital New Orleans or Ochsner Hospital for Children in Jefferson for cardiac surgery. Pediatric specialty provider shortages exist in Monroe, Lake Charles, Houma/Thibodaux and Central Louisiana.

The Bureau of Family Health provides families of children with birth defects that may impact development with access to early intervention services. The Bureau conducts one-on-one needs assessments to offer resource and referral services to families. The resources include health and Medicaid support, disability agencies, food security support, childcare, family and youth support, and advocacy and legal support.

From the start of the resource referral work in March 2022 through July 2024, the Bureau of Family Health has identified 2,605 families of children with qualifying birth defects who may benefit from a referral needs assessment. The Bureau has successfully contacted more than 1,500 families and linked 78% (approximately 1,170) to services. The most requested resources among families include insurance navigation, parent support and systems of care navigation services.

Introduction

The Louisiana Department of Health, Office of Public Health, Bureau of Family Health is responsible for surveillance of birth defects in Louisiana's children and moving that data to action by informing recommendations of strategies to improve Louisiana's maternal and child health systems of care, from preconception through referrals to resources post-diagnosis. The Louisiana Birth Defects Monitoring Network is one of the state's core public health monitoring systems. Overall, the Bureau works to promote the health of Louisiana families throughout their lifetime through programs and initiatives to improve the health of pregnant women, babies, children, teens, adults and youth with special healthcare needs.

Our vision is for Louisiana to be a state where all people are valued to reach their full potential, from birth through the next generation. Our mission is to elevate the strengths and voices of individuals, families and communities to catalyze transformational change to improve population health and achieve equity. The Louisiana Birth Defects Monitoring Network, the advisory board and this report are some of the ways the Bureau of Family Health works to advance maternal and child health outcomes in the state.

Legislative Mandates

In the 2001 Regular Session of the Louisiana Legislature, Act 194 established the authorities for a "… system to collect, analyze and disseminate data regarding birth defects in the state and to provide information to families of children born with birth defects regarding services available in their community and the development of appropriate prevention programs."

Now recognized as the Louisiana Birth Defects Monitoring Network, this system identifies and reports qualifying birth defects diagnosed before three years of age statewide in accordance with the definition of birth defect outlined in R.S. 40.31.42. Individual identifying data are confidential and are subject to discovery (R.S. 40:31.44). The Bureau of Family Health also produces an annual report per R.S. 40:31.45.

The Louisiana Birth Defects Monitoring Network was developed in partnership with clinicians, families and individuals living with birth defects participating in the statutorily required advisory board. The advisory board members are subject matter experts who provide expertise and perspective to guide the Bureau of Family Health in surveillance operations, referral to resources initiatives and birth defects prevention strategies. The current law specifies board membership by medical institutions, some of which have changed affiliation over time. Notices of meetings, agendas and minutes are posted on the Louisiana Boards and Commissions webpage.

The Bureau of Family Health is working to revitalize the advisory board and increase in-person attendance to meet quorum requirements. From January 2021 through October 2022, advisory board meetings did not meet quorum, having no more than three appointed board members in attendance at the scheduled meetings. The Bureau is working to update the board roster with additional members with diverse expertise and active members who have demonstrated engagement with the program and a willingness to travel to attend meetings.

While the advisory board's inability to meet quorum did not significantly disrupt most of the work, certain projects requiring their direct oversight have been postponed. The 2023 Louisiana Birth Defects Monitoring Network legislative report outlined advisory board plans to apply the Prevention Case Review Model to critical congenital heart defects, neural tube defects, Trisomy 21, abdominal wall defects and limb reduction defects. This work is paused until the advisory board can convene a quorum of appointed members to make official decisions for public advisory board meetings, as required by open meetings law.

Louisiana Birth Defects Monitoring Network

The Bureau of Family Health collects, analyzes and disseminates high quality, timely, actionable data through the Louisiana Birth Defects Monitoring Network. The data informs policy to improve Louisiana's maternal health system to eliminate preventable birth defects, mitigate disability and connect families with resources to improve their quality of life. The Bureau's goal is to help every child in Louisiana have a healthy start in life through birth defects monitoring. This work is a core activity of the Bureau's Title V Maternal and Child Health programs that identify and support children and youth with special healthcare needs and their families.

From January 2005 through July 2024, the Bureau of Family Health has investigated potential birth defects among 62,810 children. The Bureau identified approximately 1,500 children with specified birth defects annually, averaging 260 per 10,000 live births.

The Bureau of Family Health incorporates evidence-based public health surveillance best practices, including current technology and advanced methodologies, to conduct active surveillance of birth defects in children born in Louisiana. Monitoring the health status of newborns provides population-based data to inform policies, educate the public and support efforts in the state to improve maternal and child health outcomes, and prevent new occurrences of birth defects. The Bureau can evaluate concerns about unexpected groups of birth defects (cluster investigation), as well as the effectiveness of preventive interventions.

As a part of the Bureau of Family Health's system of monitoring birth outcomes in the state, the Louisiana Birth Defects Monitoring Network supports:

- **Policy makers** by identifying risk factors, such as maternal exposures and chronic conditions potentially linked to specific birth conditions, and identifying preventive strategies to decrease birth defects.
- **Families of infants with birth defects from birth through three years of age** by informing them of appropriate medical, educational, public health and peer support resources available in their region.
- **Men and women of reproductive age** by providing birth defects prevention education materials through our website.
- **Researchers from CDC, universities and other states** by investigating possible causes of specific birth defects.

The Bureau of Family Health provides:

- Active public health surveillance of hospital discharges of newborns until three years of age for major structural, functional or genetic birth defects.
- Links to resources for families of children under three years of age with specified birth defects to health, social service and developmental resources through our Family Resource Center.
- Prevention of future birth defects through public awareness efforts that include education to inform men and women of reproductive age of healthy prenatal lifestyle choices. Education topics include recommendations for daily consumption of 400 micrograms of folic acid daily, reducing exposures to infections and toxins, and controlling chronic conditions such as diabetes and hypertension to prevent risks of associated birth defects. This education is done in partnership with national, state and local partners such as:
    - CDC
    - National Birth Defects Prevention Network
    - Louisiana Chapter of the American Academy of Pediatrics
    - Louisiana Chapter of the American College of Obstetricians and Gynecologists
    - March of Dimes
    - Regional Families Helping Families
    - Spina Bifida of Louisiana

Additional Reporting

In addition to providing annual reports to the Louisiana Legislature, Secretary of the Louisiana Department of Health and advisory board, Louisiana Birth Defects Monitoring Network data is included in biennial reports produced by the National Birth Defects Prevention Network and special reports in the Birth Defects Research Journal. The National Birth Defects Prevention Network collects data in even years and publishes reports in odd years. The Bureau of Family Health reported birth defects data from the 2019-2021 birth years to the National Birth Defects Prevention Network in September 2024 for publication in January 2025.

In October 2023, the Bureau of Family Health supplied birth defects data from the 2018-2020 birth years to the Louisiana Department of Health, Office of Public Health's Environmental Public Health Tracking system, which provides data and information on health outcomes, the environment, population and exposures. Louisiana is one of 33 entities that include state and local health departments, cities and jurisdictions to be part of the CDC's National Environmental Public Health Tracking Network.

Birth Defects Surveillance Process

Birth defects surveillance is a public health activity, supported by senior epidemiologists and health policy leaders, and carried out by a statewide network of regionally assigned data collection specialists who evaluate patient discharge information of newborns until three years of age. Staff review records from all birthing hospitals and pediatric hospitals in Louisiana. The Bureau of Family Health maintains a longitudinal data system of all children born in Louisiana diagnosed with a structural, functional and/or genetic birth defect. Maternal and child health epidemiologists statistically analyze de-identified medical

record data for patterns and trends over time. The Bureau links families to health, social service and developmental resources for children identified with specified birth defects through the Family Resource Center.

Case Inclusion

The following are requirements for the Bureau of Family Health to review a case:

- The child must have a major structural, functional or genetic birth defect. Major defects are generally those that can adversely affect the child's health and development. We do not include children who have minor defects posing no significant health or developmental risk.
- The mother's residence at the time of the birth must be the state of Louisiana as determined by the mother's hospital records, or if still in question, by vital records birth registration data.
- Diagnosis of the qualifying condition must be confirmed before the child's third birthday.
- Pregnancy outcomes include only live births with a gestational age at birth of at least 20 weeks. In the absence of an age estimate, the infant must have a birth weight of at least 350 grams.

Methodology

The Bureau of Family Health contacts health care providers to find cases and collect data. The Bureau identifies potential cases of interest from hospital discharge indices, Medicaid and Louisiana hospital inpatient discharge data, as well as birth, death and fetal death record data from the Louisiana Vital Records Electronic Event Registration System. Staff review medical and vital statistic records to collect and validate data among children diagnosed from birth to their third birthday.

The Bureau of Family Health reviews birth defects data for completeness and coding accuracy prior to including the data in the Louisiana birth defects database. Data is stored and managed in the database and is integrated with the Louisiana Vital Records Electronic Event Registration System birth and death certificates as well as Early Hearing Detection and Intervention data.

Not all birth defects are evident at birth; therefore, the Bureau of Family Health includes children diagnosed before their third birthday allowing adequate time to capture all birth defects within our case definition. Additionally, this timeframe allows hospitals adequate records processing and reporting, as well as time for staff to abstract medical records to capture all diagnoses identified among those born in each calendar year. Please refer to Appendix A for the Case Ascertainment/Review/Quality Assurance flow chart.

Diagnoses by International Classification of Diseases-Version 10 (ICD-10) billing codes are converted to the appropriate corresponding codes from the CDC clinical coding system based on the British Pediatric Association and Classification of Diseases. Prevalence is calculated as the number of children with birth defects per 10,000 total live births. There is an exception for hypospadias and congenital posterior urethral valves, which only occur in males, and Turner syndrome, which only occurs in females. Therefore, prevalence for these defects is calculated exclusively among each gender respectively. To ensure sufficient case numbers for meaningful analysis, particularly when stratified by race/ethnicity and major birth defect type, the Bureau of Family Health reports data in three-year birth year

increments at the state level. Please refer to Appendix B for a complete list of ICD-10 birth defects codes with corresponding description labels, CDC/BPA codes, and respective referral logic.

Understanding Birth Defects

Information on Birth Defects

According to the CDC, "Birth defects are structural changes present at birth that can occur to any part of a baby's body during fetal development. While birth defects may occur any time during pregnancy, most occur within the first three months during rapid cell growth and organ development."

Birth defects vary from cosmetic differences to the medically complex, which interrupt the way the body functions. Individual health outcomes and life expectancies depend upon the body part affected and the degree to which it is affected.

Critical birth defects not only affect the baby's health, but may also result in developmental disabilities. Therefore, babies born with birth defects need special care and intervention to survive and thrive.

There is not a single cause of birth defects and not all birth defects are preventable. Scientists think most birth defects are attributable to a combination of factors involving genes, maternal behaviors and environmental exposures. (NCBDDD, 2024)

Infant Mortality: Birth to 1 Year

In Louisiana from 2019 to 2021, the four leading causes of death in infants from birth to one year were deaths from conditions originating in the perinatal period, deaths related to injury and other medical causes, Sudden Unexpected Infant Deaths and deaths due to congenital anomalies or birth defects.

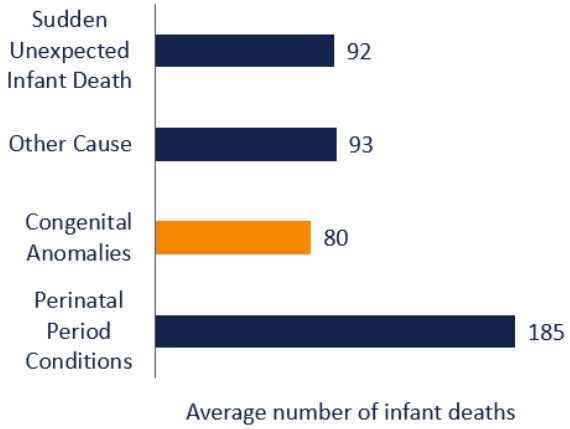
As a known leading cause of death, we know that reducing preventable birth defects reduces the number of infant deaths in Louisiana each year. However, while we know the number of infant deaths due to congenital anomalies, it is difficult to identify the exact cause of death attributable to specific birth defects because the birth defect itself is not listed as the primary cause of death on the death certificate. Therefore, death certificate data is not an adequate source to determine birth defects occurrence and prevalence.

For more information, the latest Child Death Review report is available at partnersforfamilyhealth.org/childdeathreview.

From 2019 to 2021 in Louisiana, an average of 80 infants per year died from congenital anomalies.

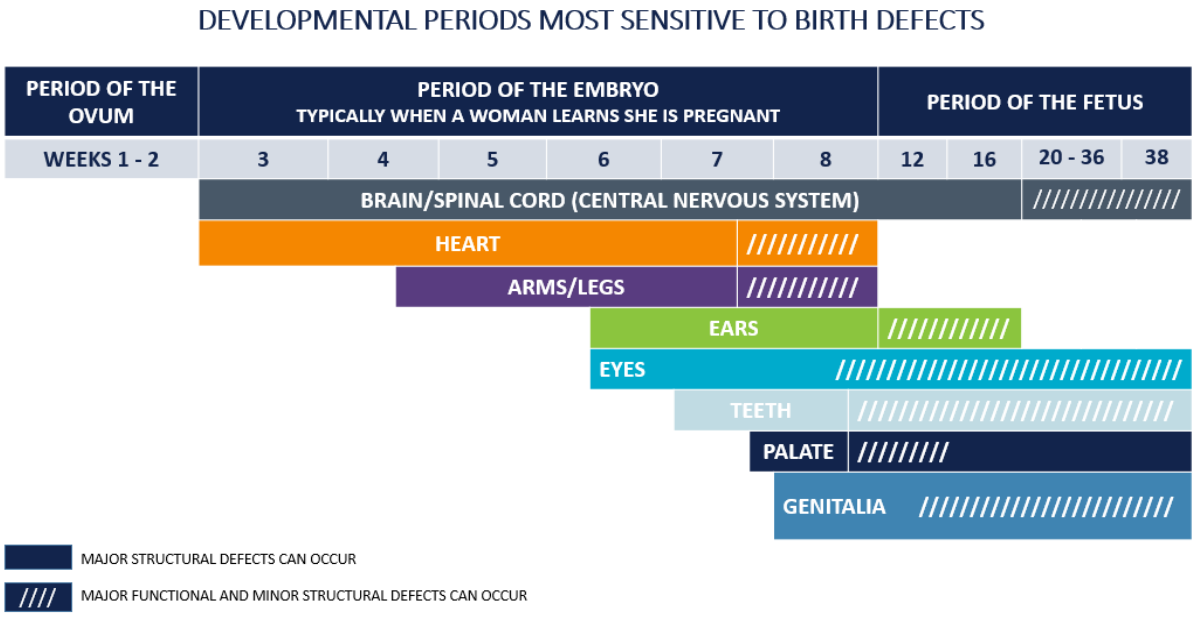
In Louisiana each year, an average of:

- 185 infants died from conditions originating in the perinatal period
- 93 infants died from other causes, including injury and other medical causes
- 92 infant deaths were classified as Sudden Unexpected Infant Deaths
- **80 infants died from congenital anomalies**



Critical Periods of Development and Risk Factors for Birth Defects

In pregnancy, each body part of the fetus forms during a specific time. During the critical periods of development for each body part, exposures may interrupt normal development resulting in a birth defect.



Adapted from Moore 1993 and the National Organization of Fetal Alcohol Syndrome (Bleyl SB, Schoenwolf GC, 2010, Carlson BM, 2008, Cochard LR, 2012, Moore KL 2013,,

Not all birth defects are preventable. Importantly, some birth defects arise from preventable exposures and risk factors. By effectively managing these factors, we can strive for positive pregnancy outcomes. These include:

- Unmanaged chronic conditions including diabetes, depression, eating disorders, high blood pressure, seizure disorder and sexually transmitted infections
- Maternal infections with fever
- Inadequate nutrition—lack of folic acid and iron
- Substance use including alcohol, tobacco or vaping, recreational/illegal drugs, some prescribed medications
- Obesity
- Environmental toxins and workplace exposures

For more information on limiting risk factors of birth defects, visit the American College of Obstetricians and Gynecologists and MotherToBaby Network.

Preventing Birth Defects in Louisiana

The Bureau of Family Health provides birth defects prevention education and materials for providers, community partners and individuals. Not all birth defects are preventable. Strengthening prevention messaging and improving systems of care for women throughout their childbearing years will reduce preventable birth defects.

These steps can help a mother and baby be as healthy as possible before, during and after pregnancy:

1. **Plan**: Decide if you want to have children. If you do, make a plan for when you would like to become pregnant.
2. **Visit a healthcare professional regularly**: Discuss medications, managing chronic conditions, recommended vaccines, testing for sexually transmitted infections and genetic counseling if you have a family history of birth defects or if you are over 35 years of age and pregnant.
3. **Take a multivitamin with 400 micrograms (mcg) of folic acid daily if you are trying to get pregnant or pregnant.**
4. **Prevent infections with good handwashing and safe sex practices.**
5. **Avoid alcohol, tobacco and other recreational drugs.**
6. **Manage obesity.**
7. **Limit exposure to environmental toxins.**

This resource is adapted from March of Dimes. For more information, visit: March of Dimes Healthy Moms. Strong Babies (Dolan, 2020).

Additional Resources

The Louisiana Reproductive Health program provides support to men, women and adolescents in the preconception period so they can establish healthy habits before they start a family, which may prevent certain birth defects. There are over 60 Louisiana Reproductive Health Program clinics statewide. Learn more at healthychoicesla.org.

Advanced Pediatric Care in Louisiana

Birth defects require specialized care at birth, in infancy, during childhood and sometimes beyond. Pediatric specialists are located at five pediatric hospitals in Louisiana. Each hospital varies in the range of specialists and services available. For example, although pediatric cardiologists are available in every region of the state, patients must travel to Children's Hospital New Orleans for cardiac surgery. Pediatric provider shortages exist in Monroe, Lake Charles, Houma, Thibodaux and Central Louisiana.

Advanced care is available at the following pediatric hospitals in Louisiana:

- Children's Hospital New Orleans: Neonatal intensive care unit (NICU), pediatric intensive care unit (PICU), cardiac intensive care unit (CICU), emergency department (ED), acute care, behavioral health and 40+ pediatric specialty clinics
- Ochsner Children's Hospital New Orleans: NICU, PICU, CICU, ED, acute careand 30+ pediatric specialty clinics
- Our Lady of the Lake Children's Hospital: NICU, PICU, ED, acute careand 30 pediatric specialty clinics
- Ochsner LSU Shreveport Pediatrics: NICU, PICU, ED and 20+ pediatric specialty clinics
- Shriners Children's Shreveport: Limited to outpatient orthopedic and craniofacial specialty clinics

Birth Defects Findings in Louisiana 2019-2021 Births

Reporting Data on Birth Defects

To ensure sufficient case numbers for meaningful analysis, particularly when stratified by race/ethnicity and major birth defect type, the Bureau of Family Health reports data in three-year birth year increments at the state level. This report presents birth defects data among children born from 2019 to 2021. For awareness, one child may have more than one qualifying birth defect.

Eight different body systems present with the most prevalent birth defects—cardiovascular (heart), genitourinary (urinary tract and kidneys), musculoskeletal (bones and muscles), chromosomal, orofacial (mouth, lips and face), gastrointestinal (digestion organs), central nervous (brain and spine) and the eyes, ears, face and neck. Each system has a critical period of development in utero, known risk factors and specialists in Louisiana who can provide treatment to address particular birth defects.

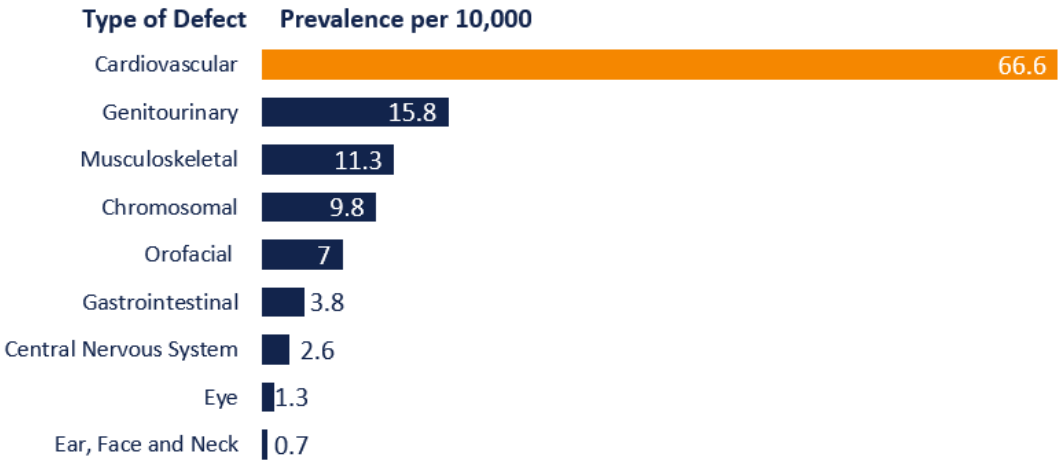
Findings At-a-Glance

Of the 170,799 children born in Louisiana between 2019 and 2021, 4,694 children or 2.75% of all children were diagnosed with at least one birth defect.

Among children born from 2019 to 2021, six birth defects with a prevalence greater than 10 per 10,000 live births were identified: atrial septal defect (a heart defect), hypospadias (a penile defect), ventricular septal defects (heart defects), pulmonary valve atresia/stenosis (a heart defect), clubfoot (a musculoskeletal defect) and Down syndrome (a chromosomal defect).

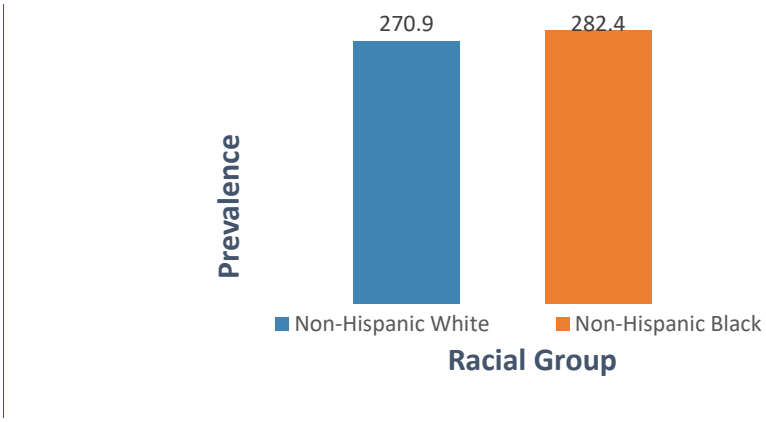
Atrial Septal Defect	**Hypospadias**	**Ventricular Septal Defects**	**Pulmonary Valve Atresia/Stenosis**	**Clubfoot**	**Down Syndrome**
114 Children Diagnosed	71 Children Diagnosed	62 Children Diagnosed	13 Children Diagnosed	12 Children Diagnosed	12 Children Diagnosed

Among children with birth defects, cardiovascular system defects were the most common.

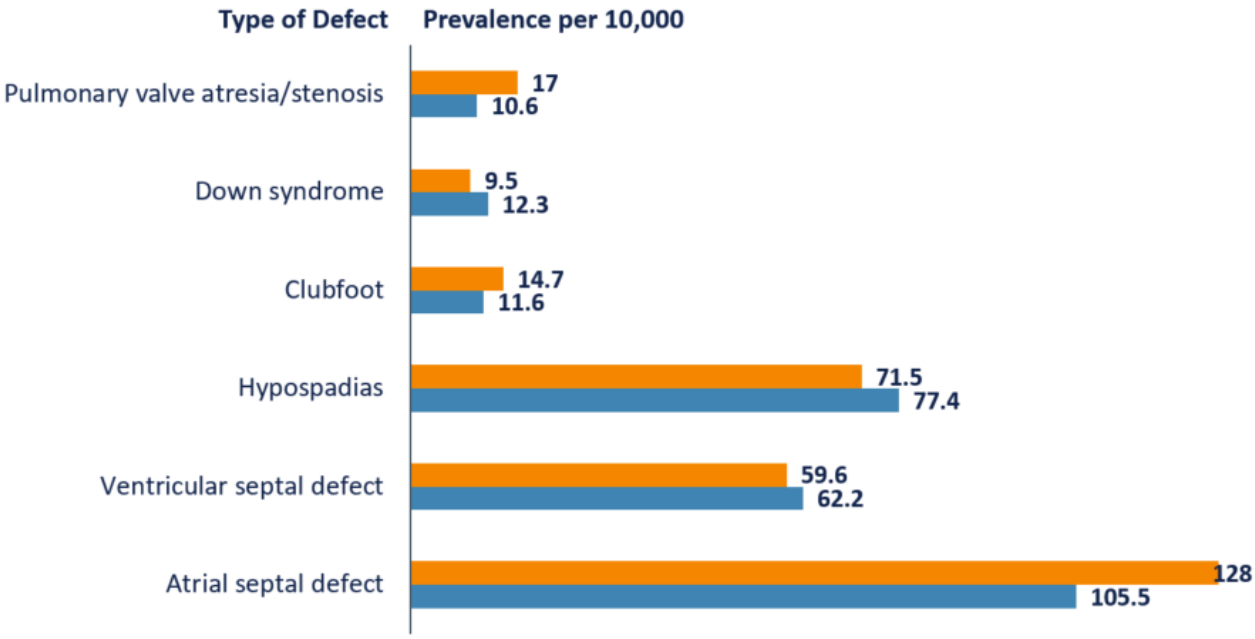


Racial Disparities in Birth Defects

The proportion of birth defects in
non-Hispanic white populations is lower than in non-Hispanic Black populations.



Among the six most common birth defects among both groups, **Black infants** have higher prevalence of pulmonary valve atresia/stenosis, clubfoot and atrial septal defects, while **white infants** have higher prevalence of Down syndrome, hypospadias and ventricle septal defect.



Non-Hispanic white infants were almost twice as likely to have cleft palate without cleft lip (11.9 vs 6.3) and craniosynostosis (11.1 vs 6.9).*

Pulmonary valve atresia and stenosis were seen more often among **non-Hispanic Black infants (17.0)** than **non-Hispanic white infants (10.6)**.*

**Among birth defects seen with a prevalence greater than 10 per 10,000 births*

Cardiovascular System

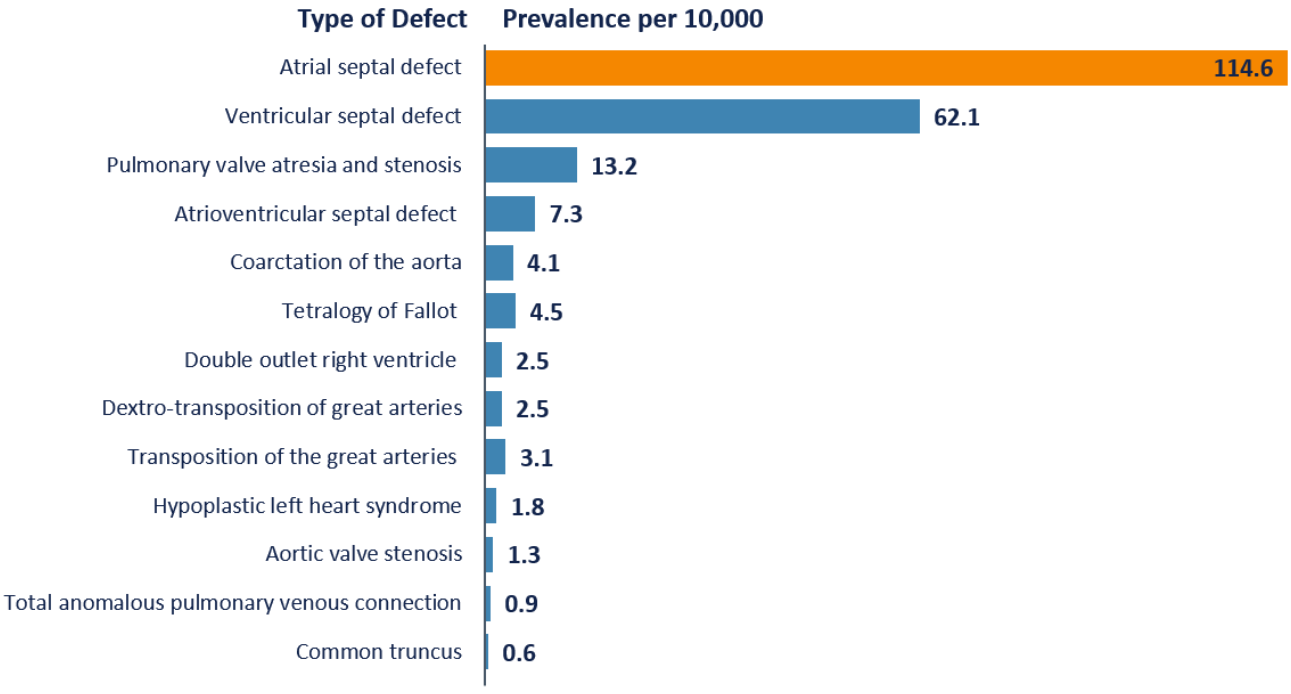
Congenital heart defects affect how blood flows through the heart and out to the rest of the body. Heart defects range in complexity from those that resolve with medical monitoring only to those that require surgical intervention. Newborn screening through pulse oximetry may detect critical heart defects for early treatment, which may prevent early death or developmental disabilities.

Critical Period of Development: The fetal heart develops between weeks three through eight of pregnancy.

Known Risk Factors: Uncontrolled diabetes, maternal smoking and certain medications during pregnancy.

Where to Receive Care in Louisiana: Pediatric cardiologists are located in New Orleans, Baton Rouge, Lafayette, Alexandria, Shreveport and Monroe. Infants in Louisiana requiring heart surgery must travel to New Orleans, as it is the only city in the state with pediatric cardiovascular surgeons.

Atrial septal defect is the leading cardiovascular birth defect among babies born in Louisiana (62 percent).



Atrial septal defect: *A hole in the wall (septum) that divides the upper chambers (atria) of the heart.*

Additional defect definitions are included in **Appendix C: Birth Defects Definitions**.

Genitourinary System

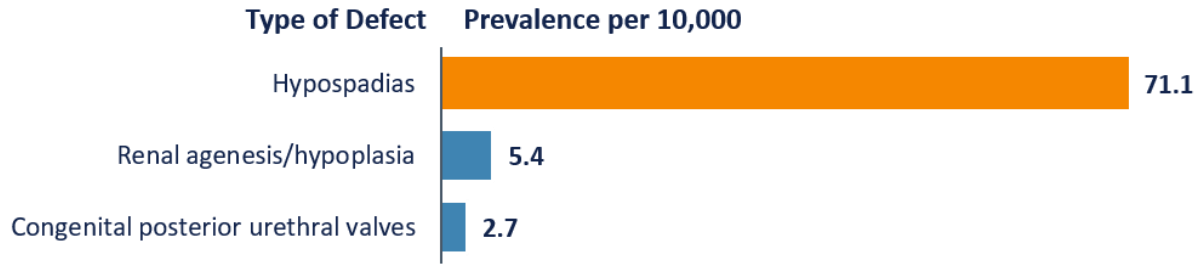
The urethra is the tube that carries urine from the bladder to the outside of the body. Hypospadias is an opening of the urethra not located at the tip of the penis. Most cases require surgical correction between 3 and 18 months. Posterior urethral valves is an obstruction of urine flow by abnormal membranes within the urethra that requires surgical correction. Renal agenesis/hypoplasia is the complete absence or incomplete development of one or both kidneys. Absence of one kidney may require medical monitoring only. Absence of both kidneys is not survivable.

Critical Period of Development: The fetal renal system and genitalia develop between weeks seven through 37 of pregnancy.

Known Risk Factors: Maternal age of 30 or older, maternal obesity, assisted reproductive technology and progestin use just before or during pregnancy.

Where to Receive Care in Louisiana: Pediatric urologists are located in New Orleans, Baton Rouge, Lafayette and Alexandria.

Hypospadias is the leading genitourinary birth defect (83 percent).



Hypospadias: *The opening of the urethra is not located at the tip of the penis.*

Additional defect definitions are included in **Appendix C: Birth Defects Definitions**.

Musculoskeletal System

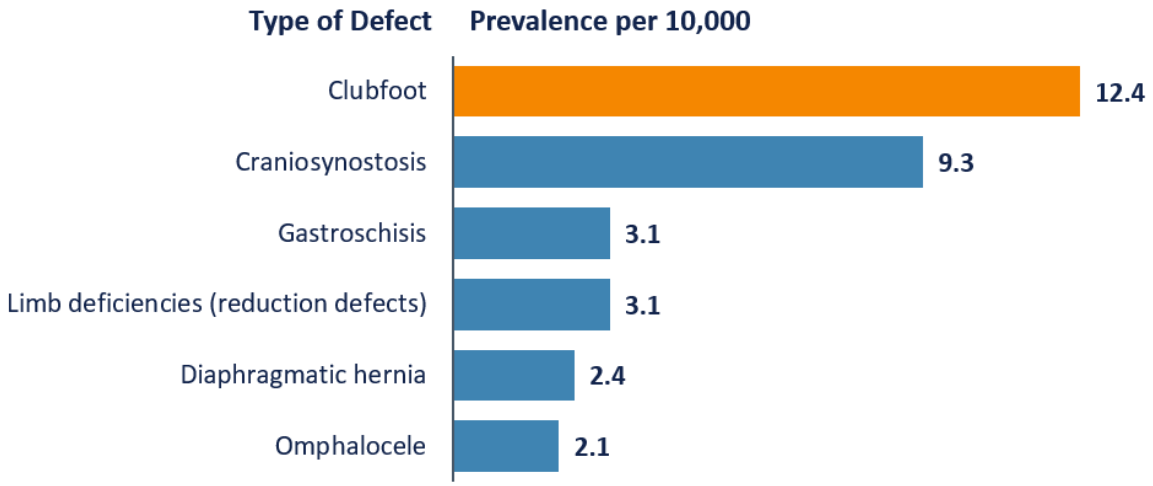
Musculoskeletal defects involve malformations of the bones and muscles. Skeletal defects include missing bones in arms and legs, rotated bones of the feet and premature closure of the developing bones in the skull. Skeletal treatments range from serial casting, helmet therapy and surgical correction. Muscle defects include herniation of the diaphragm into the thoracic cavity or herniation of the intestines through the abdominal wall. Muscle defects require surgical correction.

Critical Period of Development: The fetal skeletal system develops between weeks three through eight of pregnancy.

Known Risk Factors: Genetics factors, substance use or medications during pregnancy, and young maternal age (gastroschisis).

Where to Receive Care in Louisiana: Pediatric orthopedists and pediatric plastic surgeons are located in New Orleans, Baton Rouge, Lafayette and Shreveport.

Clubfoot is the leading musculoskeletal birth defect (40 percent).



Clubfoot: *When the foot is in a twisted position.*

Additional defect definitions are included in **Appendix C: Birth Defects Definitions**.

Chromosomal System

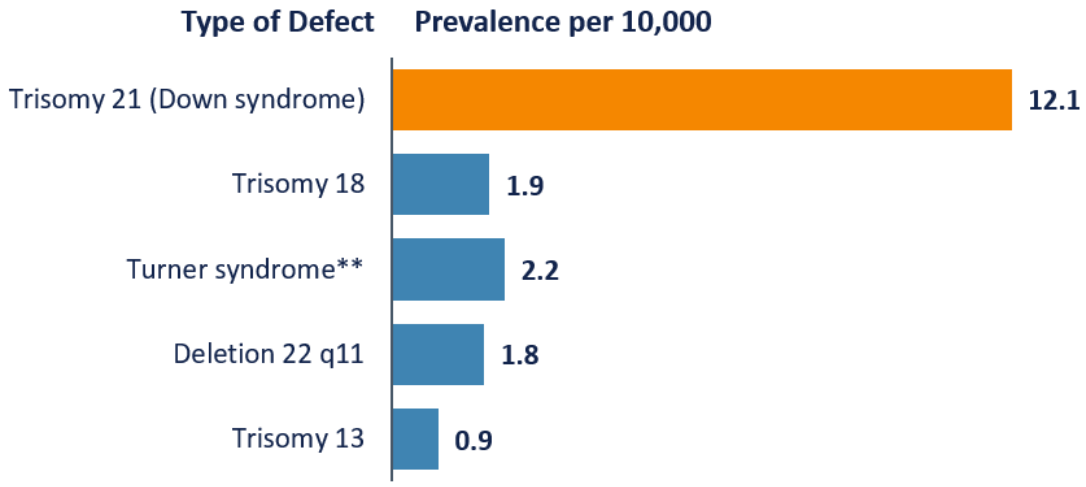
Chromosomal defects arise from abnormalities in the structure or number of chromosomes within a cell's genes, such as extra chromosomal material, missing chromosomal segments or the absence of entire chromosomes. Chromosomes determine how the body forms and functions. Chromosomal rearrangements may result in distinctive physical characteristics and various developmental challenges. Just as chromosomal defects vary, so does the degree of medical complexity. Many people with a chromosomal diagnosis lead healthy and productive lives into adulthood. Some chromosomal diagnoses are incompatible with life. Screening during pregnancy may suggest a higher chance of a chromosomal diagnosis, but screening alone is not definitive. Specific diagnoses must be confirmed through lab testing with a certified geneticist.

Critical Period of Development: Fetal chromosomes develop between weeks one and two of pregnancy.

Known Risk Factors: Advanced maternal age (35+), family history of genetic diagnoses, radiation and chemical exposure during pregnancy.

Where to Receive Care in Louisiana: Pediatric geneticists are located in New Orleans, Baton Rouge and Lafayette.

Down syndrome is the leading chromosomal birth defect (45 percent).



Trisomy 21 (Down syndrome): *When cell division results in extra genetic material from chromosome 21 resulting in the presence of three copies of all or part of chromosome 21.*

Additional defect definitions included in **Appendix C: Birth Defects Definitions**.

** Turner syndrome is exclusively in females when the second X chromosome is absent or structurally malformed.

Orofacial System

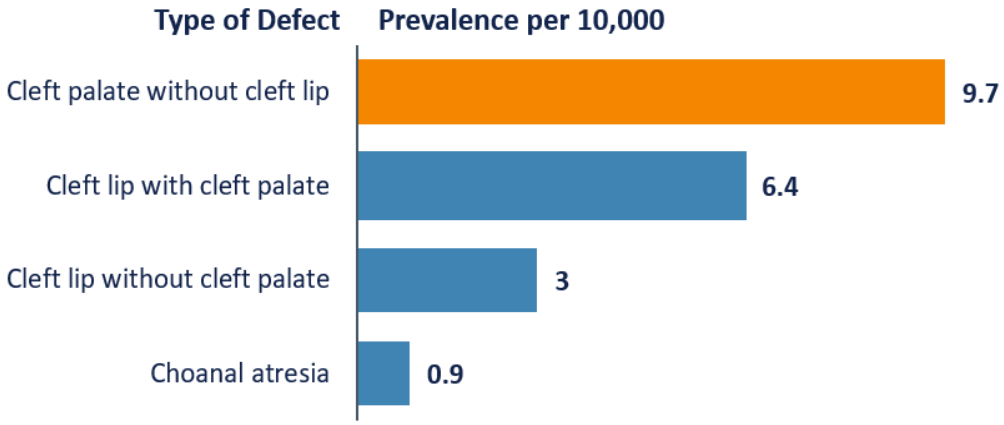
Cleft lips and cleft palates occur when the tissue of the lips and inside of the mouth do not completely join. Openings may happen anywhere along the top lip or roof of the mouth, and may or may not affect teeth, gums, nose and uvula. Most clefts are diagnosed during pregnancy or at birth. Submucosal clefts and bifid uvulas may not be diagnosed until later in childhood. Clefts affect feeding and speech. Surgical repair is required, usually in a staged approach, with multiple surgeries in the first two years.

Critical Period of Development: The fetal mouth, lips and face develop between weeks six through 38 of pregnancy.

Known Risk Factors: Smoking during pregnancy, maternal diabetes, certain epilepsy medications during pregnancy and genetic factors.

Where to Receive Care in Louisiana: Pediatric plastic surgeons are located in New Orleans, Baton Rouge, Lafayette and Shreveport.

Cleft palate without cleft lip is the leading orofacial birth defect (50 percent).



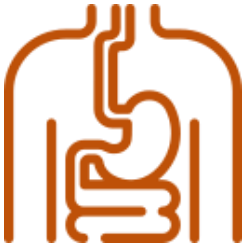
Cleft palate: *Incomplete fusion of the roof (palate) of the mouth.*

Cleft lip: *Incomplete fusion of the lips.*

Additional defect definitions included in **Appendix C: Birth Defects Definitions**.

Gastrointestinal System

The gastrointestinal (GI) tract begins at the lips and ends at the anus. GI defects occur when connections are interrupted or underdeveloped. The liver and gallbladder are included in this category. The GI tract is one of the first things to form and continues to develop throughout pregnancy. Therefore, premature babies have a higher rate of gastrointestinal challenges. Most GI defects require surgical intervention and repair within the first weeks of life.

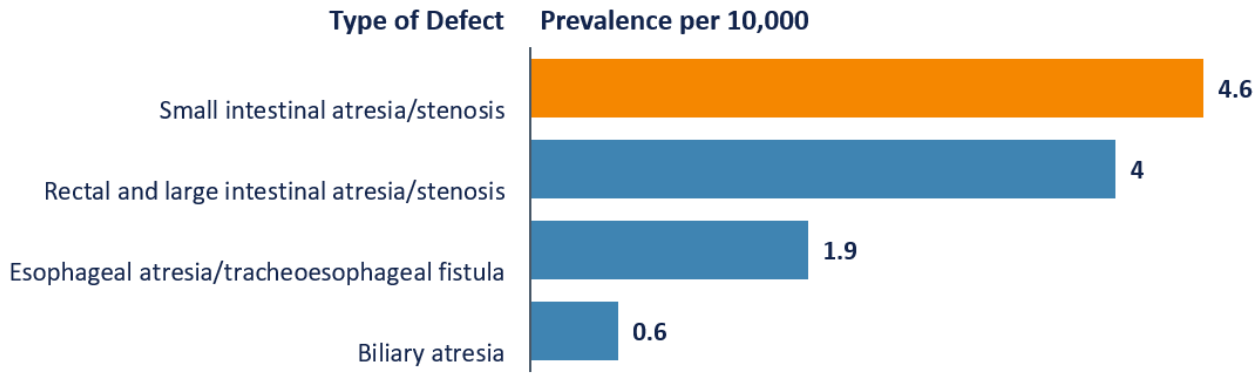


Critical Period of Development: The fetal gastrointestinal tract develops between weeks eight through 22 weeks of pregnancy.

Known Risk Factors: Genetic factors, use of assisted reproductive technology, or older paternal age (esophageal atresia).

Where to Receive Care in Louisiana: Pediatric gastroenterologists are located in New Orleans, Baton Rouge, Lafayette and Shreveport.

Small intestinal atresia/stenosis is the leading gastrointestinal birth defect (44 percent).



Small intestinal atresia/stenosis: *Absence of or narrowing of all or a section of the small intestine.*

Additional defect definitions are included in **Appendix C: Birth Defects Definitions**.

Central Nervous System

Central nervous system defects, also called neural tube defects (NTD), are severe birth defects caused when the neural tube does not close properly, interrupting development of the brain and spine. The brain and spine develop by the third week of pregnancy before most women know they are pregnant.

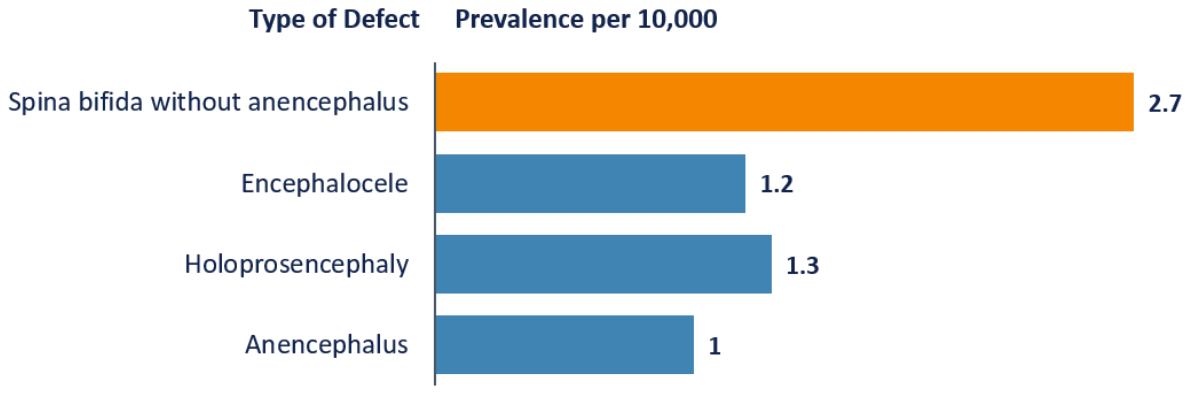
Getting 400 micrograms (mcg) of folic acid daily before and during early pregnancy can help prevent NTDs. NTDs range in severity from partial or complete absence of the brain, called anencephaly, to openings anywhere along the spinal cord, known as spina bifida. NTDs may be diagnosed by ultrasound during pregnancy. Anencephaly is incompatible with life. Spina bifida may be eligible for in utero repair to lessen further spinal cord damage. If not, surgical repair is required immediately upon birth.

Critical Period of Development: The fetal spine and brain develop starting week three and continuing through the entire pregnancy.

Known Risk Factors: Low folate in early pregnancy, MTHFR gene, maternal diabetes, some seizure medications, maternal fever or exposure to high temperatures such as hot tub use.

Where to Receive Care in Louisiana: Pediatric neurosurgeons are located in New Orleans, Baton Rouge and Shreveport. Pediatric neurologists are located in each region of Louisiana.

Spina bifida without anencephalus is the leading central nervous system birth defect (37 percent).



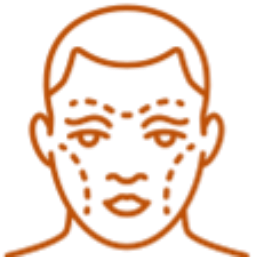
Spina bifida without anencephalus: *Incomplete closure of the vertebra of the spine from which spinal cord tissue and/or membranes covering the spine may protrude.*

Additional defect definitions are included in **Appendix C: Birth Defects Definitions**.

Eyes, Ears, Face and Neck

Eye and ear defects occur during the first 12 weeks of pregnancy when one or both eyes and ears fail to form entirely or incompletely. Anophthalmia (absent eye) and microphthalmia (small eye) result in blindness or limited vision. Both require ophthalmology and plastic surgery management as the baby grows.

Anotia (absent ear) and microtia (small ear) defects range from cosmetic to obstructed hearing. Both require varying levels of audiology and plastic surgery management into early childhood. Eye and ear anomalies may be isolated or associated with certain genetic syndromes. Therefore, medical management should include genetic testing.

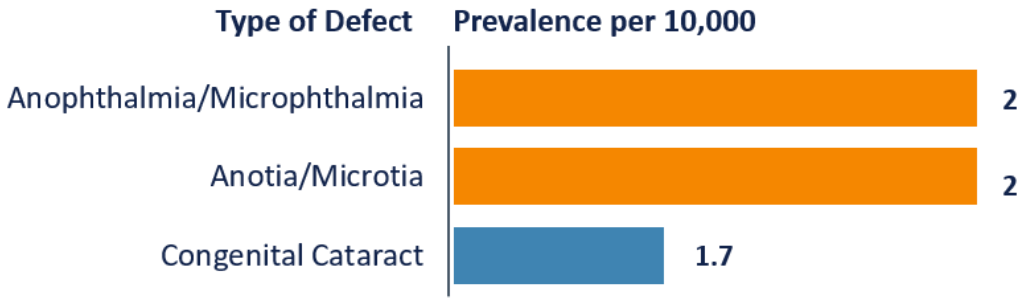


Critical Period of Development: Fetal eyes and ears develop between weeks four through 12 of pregnancy.

Known Risk Factors: Genetic/chromosomal changes, maternal diabetes, low folate levels, isotretinoin (Accutane) or thalidomide use during pregnancy, maternal infections such as rubella and syphilis.

Where to Receive Care in Louisiana: Pediatric ophthalmologists and pediatric ENTs are located in New Orleans, Baton Rouge, Lafayette and Shreveport.

Anophthalmia/microphthalmia are the leading eye birth defects (58 percent).
Anotia/microtia are the leading ear defects (100 percent).



Anophthalmia/microphthalmia: *Total absence of or reduced volume of eye tissue.*

Anotia/microtia: *Total absence of or malformation of the external ear and canal.*

Additional defect definitions are included in **Appendix C: Birth Defects Definitions**.

Please refer to **Appendix D: 2019-2021 Data Tables** for of types of birth defects, prevalence of specific birth defects by organ and chromosomal system, and by race and ethnicity.

Moving Data to Action

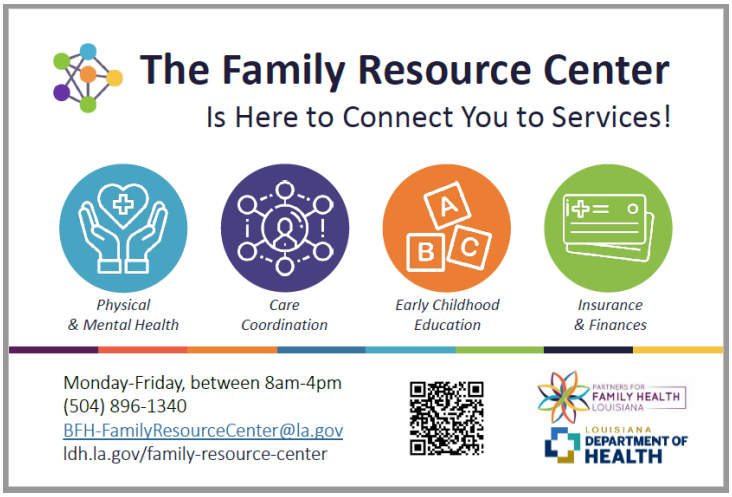
Referral to Resources

When babies are born with birth defects, families need help not only learning about complex systems of care but also how to navigate those often-fragmented systems. The Bureau of Family Health's Family Resource Center uses monthly data from the Louisiana Birth Defects Monitoring Network to contact families with children who may benefit from early intervention services. The Bureau conducts individualized needs assessments to offer resource and referral services to families of children with birth defects most likely to affect developmental outcomes. Resources include health and insurance, disability agencies, food security, childcare, family and youth support, and advocacy and legal support.

Timely birth defects data is critical to facilitate referrals because early intervention can have a significant impact on children's ability to learn new skills and increase their success in school and life. For example, Louisiana EarlySteps provides services to families with infants and toddlers, ages birth to three years (36 months) who have developmental delays or a medical condition likely to result in a developmental delay.

From the start of the referral project in March 2022 through July 2024, the Bureau of Family Health has identified 2,605 families of children with qualifying birth defects who may benefit from a referral needs assessment. The Bureau has successfully contacted more than 1,500 families and linked 78% (approximately 1,170) of those families to services. The most requested resources among families include insurance navigation along with parent support and systems of care navigation services.

The Bureau of Family Health uses multiple means to contact families including mail, phone, email and text messaging along with accommodation supports such as the Language Line and Telecommunications Relay Services. This ensures families receive information in the language and communication method that works best for them. Printed materials are also available in Arabic, English, Portuguese, Spanish and Vietnamese.



The Bureau of Family Health offers resource and referral services to families and providers statewide through the Family Resource Center. Service requests can be submitted via phone 504-896-1340 (Monday through Friday 8 a.m. to 4 p.m.) or via email at BFH-FamilyResourceCenter@la.gov. Visit ldh.la.gov/family-resource-center to learn more.

Conclusion

Louisiana Birth Defects Monitoring Network data is available at partnersforfamilyhealth.org, at the National Birth Defects Prevention Network (nbdpn.org/birth_defects_data_tables_and.php), from the CDC Data & Statistics on Birth Defects (cdc.gov/birth-defects/data-research/facts-stats/) and on the Louisiana Environmental Public Health Tracking Network health data portal (healthdata.ldh.la.gov) to enable analysis, visualization and reporting. This data is available to environmental and public health practitioners, healthcare providers, community members, policymakers and others to make data-driven decisions that affect the health of Louisiana residents.

Looking Ahead

As the established statewide mechanism for tracking and monitoring birth defects in Louisiana, the Bureau of Family Health's Louisiana Birth Defects Monitoring Network incorporates evidence-based public health surveillance best practices and continues to seek opportunities for quality improvement. Increased efficiencies in data collection and reporting approaches moved our data to action in 2024 by informing timely referrals to services for families.

Program goals include revitalizing the advisory board and ensuring quorum is met at all open meetings to facilitate effective decision-making and the provision of valuable recommendations. The first step will be to review and update the membership roster, removing inactive members and filling vacancies with actively engaged pediatric health providers. Second, the Bureau of Family Health will submit a legislative proposal and seek approval from the Secretary of the Louisiana Department of Health and the Governor to amend R.S. 31:46 to update the membership of the advisory board to include additional subject matter expertise not currently represented. The advisory board will work with the Bureau of Family Health to advance birth defects surveillance, identify data-informed and evidence-based prevention strategies, and make recommendations to improve systems of care for children with birth defects. This work includes applying the Birth Defects Prevention Case Review Model to critical congenital heart defects, neural tube defects, Trisomy 21, abdominal wall defects and limb reduction defects.

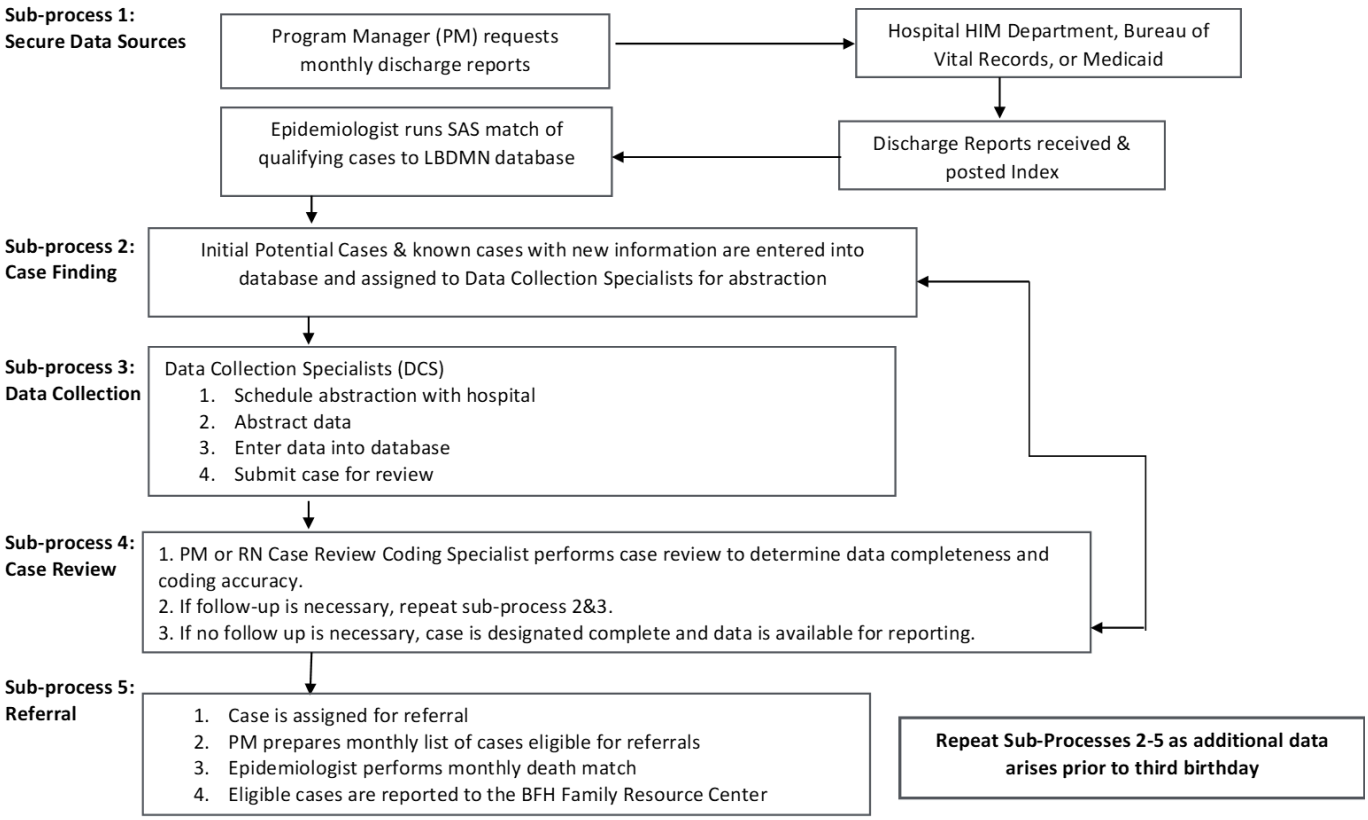
Additional goals include increasing the number of children referred to the Family Resource Center. The Bureau of Family Health will develop protocols to assess the needs of children who may benefit from referrals, even if they do not meet the strict criteria for inclusion in the birth defects database. This includes children under three years old with developmental and medical involvement due to prematurity or to a birth defect or medical condition outside of the National Birth Defects Prevention Network standards for reporting.

Our final goal is to achieve and maintain the highest data quality standards for accuracy, completion, and timeliness of state birth defects surveillance programs by identifying, processing and completing 97 percent of initial potential cases within 45 days of identification. This will meet the National Birth Defects Prevention Network's highest standard of data quality by completing 99% of all core and recommended data within two years of birth.

Appendix

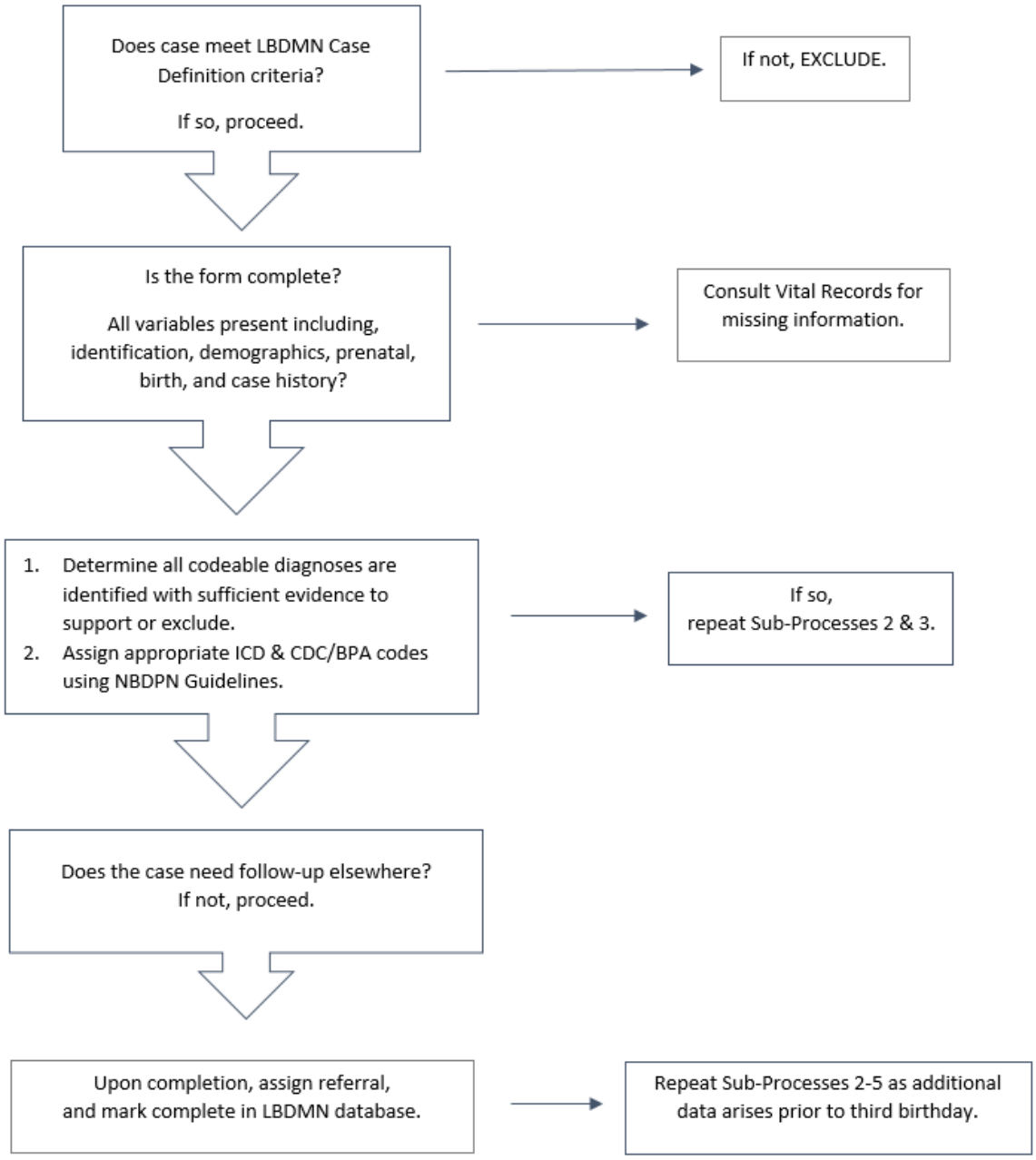
Appendix A: Case Ascertainment/Review/Quality Assurance Process Chart

This chart depicts how we identify cases, abstract and review data, and ensure completeness, accuracy and timeliness of data collection for reporting and referral to services.



Appendix A: Case Ascertainment/Review/Quality Assurance Process Chart Continued

Louisiana Birth Defects Monitoring Network Case Review Quality Assurance Sub-Process Four Steps



Appendix B: Birth Defects Codes, Descriptions, Referral Logic

These tables include a complete list of birth defects ICD-10 codes with corresponding descriptions labels, CDC/BPA codes and respective referral logic. “Standard level” refers to National Birth Defects Prevention Network designations for data quality completeness. “Case definition” refers to how the program uses codes for case finding and coding. Referral indicates if the birth defect qualifies for referral. The Bureau of Family Health refers only to birth defects that are most likely to affect developmental outcomes. Some birth defects are not survivable, or once repaired, do not impact health and developmental outcomes.

ICD-10 CM CODES BY STANDARD LEVEL	DESCRIPTION	CASE DEFINTION	CDC/BPA CODES	REFERRAL
CORE				
Q00.0-Q00.1	Anencephalus	CASE FINDING & CODING	740.000 –740.100	NO
Q05.0-Q05.9	Spina bifida without anencephalus	CASE FINDING & CODING	741.000; 741.020 -741.090; 741.900-	YES
Q07.01		CASE FINDING & CODING	741.010	YES
Q07.03 w/o Q00.0 - Q00.1		CASE FINDING & CODING	741.010	YES
Q20.0	Common Truncus	CASE FINDING & CODING	745.000	YES
Q20.3, Q20.5	TGA	CASE FINDING & CODING	745.100-745.120, 745.180-745.190	YES
Q21.2	AVSD	CASE FINDING & CODING	745.600-745.690	YES
Q21.3	TOF	CASE FINDING & CODING	74.20-745.21	YES
Q23.4	HLHS	CASE FINDING & CODING	746.700	YES
Q26.2	TAPVR	CASE FINDING & CODING	747.420	YES
Q35.1 - Q35.9	Cleft palate without Cleft lip	CASE FINDING & CODING	749.000-749.090	YES
Q36.0 - Q36.9	Cleft lip without Cleft palate	CASE FINDING & CODING	749.100-749.190	YES
Q37.0 - Q37.9	Cleft lip with Cleft palate	CASE FINDING & CODING	749.200-749.290	YES
Q71.0 - Q71.9	Limb reduction defects	CASE FINDING & CODING	755.200-755.290	YES
Q72.0 - Q72.9		CASE FINDING & CODING	755.300-755.390	YES
Q73.0 - Q73.8		CASE FINDING & CODING	755.400-755.490	YES
Q79.2	Omphalocele	CASE FINDING & CODING	756.700	YES
Q79.3	Gastroschisis	CASE FINDING & CODING	756.710	YES
Q90.0 - Q90.9	Trisomy 21	CASE FINDING & CODING	758.000-758.090	YES
RECOMMENDED				
Q01.0 - Q01.9	Encephalocele	CASE FINDING & CODING	742.000-742.090	YES
Q04.2	Holoprosencephaly*	CASE FINDING & CODING	742.260	YES
Q11.0 - Q11.2	Anophthalmia/Microphthalmia	CASE FINDING & CODING	743.000-743.100	YES
Q12.0	Congenital cataract*	CASE FINDING & CODING	743.320-743.326	YES
Q13.1	Aniridia	CASE FINDING & CODING	743.420	YES
Q16.0, Q17.2	Anotia/microtia	CASE FINDING & CODING	744.010, 744.210	YES
Q20.1	DORV	CASE FINDING & CODING	745.130-745.150	YES
Q20.4	Single Ventricle	CASE FINDING & CODING	745.300	YES
Q21.0	VSD	CASE FINDING & CODING	745.400-745.490	YES
Q21.1	ASD	CASE FINDING & CODING	745.500-745.590	YES
Q22.0, Q22.1	Pulmonary valve atresia and stenosis	CASE FINDING & CODING	746.000, 746.010	YES
Q22.4	Tricuspid valve atresia and stenosis	CASE FINDING & CODING	746.100, 746.106	YES
Q22.5	Ebstein anomaly	CASE FINDING & CODING	746.200	YES
Q23.0	Aortic valve stenosis	CASE FINDING & CODING	746.300	YES
Q25.1	COA	CASE FINDING & CODING	747.100-747.190	YES

Q25.2, Q25.4	IAA	CASE FINDING & CODING	747.215-747.217	YES
Q30.0	Choanal atresia	CASE FINDING & CODING	748.000	YES
Q39.0 - Q39.4	TEF/ EA	CASE FINDING & CODING	750.300-750.350	YES
Q41.0 - Q41.9	Small intestine atresia/stenosis	CASE FINDING & CODING	751.100-751.190	YES
Q42.0 - Q42.9	Rectal and large intestine atresia/ stenosis	CASE FINDING & CODING	751.200-751.240	YES Impe
Q44.2 - Q44.3	Biliary atresia*	CASE FINDING & CODING	751.650	YES
Q54.0 - Q54.9 (not Q54.4)	Hypospadias	CASE FINDING & CODING	752.600-752.607;752.620;752.625-752.627	NO
Q60.0 - Q60.6	Renal agenesis/ hypoplasia	CASE FINDING & CODING	753.000-753.010	YES
Q64.10, Q64.19	Bladder exstrophy	CASE FINDING & CODING	753.500	YES
Q64.12	Cloacal exstrophy	CASE FINDING & CODING	751.550	YES
Q64.2	PUV	CASE FINDING & CODING	753.600	YES
Q66.0, Q66.89	Clubfoot	CASE FINDING & CODING	754.500-754.004; 754.730-754.734	YES
Q75.0	Craniosynostosis*	CASE FINDING & CODING	756.000-756.030	YES
Q79.0, Q79.1	Diaphragmatic hernia	CASE FINDING & CODING	756.610-756.616	YES
Q91.0 - Q91.3	Trisomy 18	CASE FINDING & CODING	758.200-758.295	91%
Q91.4 - Q91.7	Trisomy 13	CASE FINDING & CODING	758.100-758.190	91%
Q93.81	22q11 deletion*	CASE FINDING & CODING	758.370	YES
Q96.0 - Q96.9	Turner syndrome*	CASE FINDING & CODING	758.600-758.690	YES
LBDMN				
Q87.40	Marfan syndrome, Stickler	CODING	759.860	YES
Q87.81	Alport syndrome	CODING	759.870	YES
Q93.3	Other autosomal deletions	CODING	758.380	YES
Q93.4	Cri du chat syndrome (5p deletion)	CODING	758.310	YES
Q93.59	Other deletions of part of a chromosome	CODING	758.390	YES
Q93.7	Deletions with other complex rearrangements	CODING	758.390	YES
Q93.88	Other microdeletions	CODING	758.380	YES
Q93.89	Other deletions from the autosomes	CODING	758.380	YES
Q97.0	Other conditions due to sex chromosome anomalies	CODING	758.8**	YES
Q97.1	Female with more than three X chromosomes	CODING	758.850	YES
Q97.2	Mosaicism, lines with various numbers of X chromosomes	CODING	758.800, 758.820, 758.830	YES
Q97.8	Other specified sexchromosome abnormalities, female phenotype	CODING	758.810	YES
Q98.4	Klinefelter's syndrome	CODING	758.700, 758.710, 758.790	YES
Q98.5	Karyotype 47,XYY	CODING	758.840	YES

Q98.7	Male with sex chromosome mosaicism	CODING	758.840	YES
Q98.8	Other specified sex chromosome abnormalities, male phenotype	CODING	758.820	YES
Q99.2	Fragile X syndrome	CODING	758.880	YES
Q99.8	Other conditions due to autosomal material	CODING	758.580	YES
Q99.8	Other conditions due to chromosome anomalies	CODING	758.880	YES
Q99.9	Conditions due to anomaly of unspecified chromosome	CODING	758.890	YES

Appendix C: Birth Defects Definitions

The following appendix includes explanations of birth defects mentioned in this report. Definitions are adapted from National Birth Defects Prevention Network Guidelines for Conducting Birth Defects Surveillance Appendix 3.1 Birth Defects Descriptions for National Birth Defects Prevention Network Core, Recommended and Extended Conditions, Updated March 2021 (Sever, 2021).

Cardiovascular System

Atrial septal defect: A hole in the wall (septum) that divides the upper chambers (atria) of the heart.

Ventricular septal defect: A hole in the wall (septum) that separates the two lower chambers (ventricles) of the heart.

Pulmonary valve atresia and stenosis: The pulmonary valve that controls blood flow from the heart to the lung doesn't form or is narrowed.

Atrioventricular septal defect: A defect in both the lower portion of the atrial septum and the upper portion of the ventricular septum. The valves controlling blood flow from the atria to the ventricles, the tricuspid and mitral valves may be malformed.

Coarctation of the aorta: The aorta is narrower than usual, impacting blood flow to the body.

Tetralogy of Fallot: Combines four defects of the heart and its blood vessels: ventricular septal defect, pulmonary stenosis, enlarged aortic valve and ventricular hypertrophy.

Double outlet right ventricle: The pulmonary artery and the aorta**,** both of the heart's major arteries, connect to the right ventricle with an accompanying ventricular septal defect.

Dextro-transposition of great arteries: The aorta arises from the right ventricle instead of the left and the pulmonary artery arises from the left ventricle instead of the right. Oxygen-rich blood remains in the heart instead of being pumped out to the body.

Transposition of the great arteries: The aorta arises from the right ventricle instead of the left and the pulmonary artery arises from the left ventricle instead of the right, with an accompanying ventricular septal defect. Therefore, some oxygen rich blood is pumped out to the body**.**

Hypoplastic left heart syndrome: The structures on the left side of the heart and the aorta are extremely small and insufficient to support circulation. This includes the left ventricle, the mitral and aortic valves, the aortic arch and the aorta.

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

Total anomalous pulmonary venous connection: The four pulmonary veins do not connect to the left atrium (left upper chamber). Instead, the four pulmonary veins drain through an abnormal (anomalous) connection to the right atrium (right upper chamber).

Common truncus: 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.

Appendix C: Birth Defects Definitions Continued

Genitourinary System

Hypospadias: Displacement of the opening of the urethra, other than at the tip of the penis.

Renal agenesis/hypoplasia: Complete absence or incomplete development of one or both kidneys.

Congenital posterior urethral valves: Obstruction of urine flow by abnormal membranes within the posterior male urethra.

Musculoskeletal System

Clubfoot: Abnormalities twisting of one or both feet out of shape to either side, upward or downward.

Craniosynostosis: Premature closure (fusion) of one or several cranial sutures (connective tissue membranes that separate the bones of the developing skull).

Gastroschisis: A fissure (opening) in the anterior abdominal wall lateral to (beside) the umbilicus through which the small intestine, part of the large intestine, and occasionally the liver and spleen, may herniate.

Limb deficiencies (reduction defects): 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).

Diaphragmatic hernia: Absence of or incomplete formation of the diaphragm through which a portion of the abdominal contents push (herniate) into the thoracic cavity.

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. Although outside of the body, the herniating organs usually are covered by a nearly transparent membranous sac.

Chromosomal System

Trisomy 21 (Down syndrome): 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.

Turner syndrome: The presence of an absent or structurally abnormal second X chromosome in a phenotypic female.

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

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

Appendix C: Birth Defects Definitions Continued

Orofacial System

Cleft palate: An opening in the roof of the mouth resulting from incomplete fusion of the shelves of the palate.

Cleft lip: Incomplete fusion of the parts of the lip.

Choanal atresia: Obstruction of the opening of the nasal cavity into the nasopharynx on either side.

Gastrointestinal System

Small intestinal atresia/stenosis: Complete or partial occlusion of the lumen of one or more segments of the small intestine.

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.

Esophageal atresia/tracheoesophageal fistula: Esophageal atresia is a condition in which the esophagus ends in a blind pouch and fails to connect with the stomach. A tracheoesophageal fistula is an abnormal communication between the esophagus and the trachea.

Biliary atresia: Absence of the lumen of the extrahepatic bile ducts.

Central Nervous System

Spina bifida without anencephalus: Incomplete closure of the vertebral spine through which spinal cord tissue and/or the membranes covering the spine (meninges) herniate**.**

Encephalocele: Herniation of brain tissue and/or meninges (membrane of the spinal cord) through a defect in the skull. The hernia sac usually is covered by skin**.**

Holoprosencephaly: A structural brain anomaly from failure of the developing brain to separate in half completely into the left and right cerebral hemispheres.

Anencephaly: Partial or complete absence of the brain and skull.

Eyes, Ears, Face and Neck Defects

Anophthalmia/microphthalmia: Total absence of or reduced volume of eye tissue.

Anotia: Total absence of the external ear and canal.

Microtia: Malformation or hypoplasia of the external ear.

Congenital cataract: Opacity of the lens of the eye that forms in utero and is present at birth.

Appendix D: 2019-2021 Data Tables

The following data tables include numbers and percentages of types of birth defects, prevalence of specific birth defects by organ and chromosomal system and by race and ethnicity.

Table 1: Type of birth defects by organ and chromosome system among children with birth defects, 2019-2021 (n = 4,694)

Organ and chromosomal system	Number	Percent*
Cardiovascular	3,127	66.6
Genitourinary	743	15.8
Musculoskeletal	530	11.3
Chromosomal	459	9.8
Orofacial including choanal atresia	328	7.0
Gastrointestinal	179	3.8
Central nervous system	124	2.6
Eye	60	1.3
Ear, face and neck	34	0.7
Other	12	0.3

**Because one child may have more than one birth defect, the total percentages are greater than 100% when totaled.*

Appendix D: 2019-2021 Data Tables Continued

Table 2: Prevalence of specific birth defects by organ and chromosomal system, 2019-2021 (N = 170,799 live births)

System	Birth defects	Number	%	Prev.	95%CI	
	Total	**4694**		**274.8**	**267.0**	**282.8**
Central nervous	Spina bifida without anencephalus	46	37.1	2.7	2.0	3.6
(n = 124)	Holoprosencephaly	22	17.7	1.3	0.8	2.0
	Encephalocele	20	16.1	1.2	0.7	1.8
	Anencephalus	17	13.7	1.0	0.6	1.6
Eyes	Anophthalmia/microphthalmia	35	58.3	2.0	1.4	2.9
(n = 60)	Congenital cataract	29	48.3	1.7	1.1	2.4
Ear, face, neck	Anotia/microtia	34	100.0	2.0	1.4	2.8
(n = 34)						
Cardiovascular	Atrial septal defect	1958	62.6	114.6	109.6	119.8
(n = 3127)	Ventricular septal defect	1060	33.9	62.1	58.4	65.9
	Pulmonary valve atresia and stenosis	226	7.2	13.2	11.6	15.1
	Atrioventricular septal defect	125	4.0	7.3	6.1	8.7
	Tetralogy of Fallot	76	2.4	4.5	3.5	5.6
	Coarctation of the aorta	70	2.2	4.1	3.2	5.2
	Transposition of the great arteries	53	1.7	3.1	2.3	4.1
	Double outlet right ventricle	43	1.4	2.5	1.8	3.4
	Dextro-transposition of great arteries	43	1.4	2.5	1.8	3.4
	Hypoplastic left heart syndrome	30	1.0	1.8	1.2	2.5
	Aortic valve stenosis	22	0.7	1.3	0.8	2.0
	Interrupted aortic arch	15	0.5	0.9	0.5	1.4
	Ebstein anomaly	15	0.5	0.9	0.5	1.4
	Total anomalous pulmonary venous connection	15	0.5	0.9	0.5	1.4
	Tricuspid valve atresia and stenosis	13	0.4	0.8	0.4	1.3

	Common truncus	11	0.4	0.6	0.3	1.2
	Pulmonary valve atresia	11	0.4	0.6	0.3	1.2
	Tricuspid valve atresia	10	0.3	0.6	0.3	1.1
	Single ventricle	7	0.2	0.4	0.2	0.8
Oro-facial	Cleft palate without cleft lip	165	50.3	9.7	8.2	11.3
(n = 328)	Cleft lip with cleft palate	110	33.5	6.4	5.3	7.8
	Cleft lip without cleft palate	51	15.5	3.0	2.2	3.9
	Choanal atresia	15	4.6	0.9	0.5	1.4
Gastrointestinal	Small intestinal atresia/stenosis	78	43.6	4.6	3.6	5.7
(n = 179)	Rectal and large intestinal atresia/stenosis	68	38.0	4.0	3.1	5.0
	Esophageal atresia/tracheoesophageal fistula	33	18.4	1.9	1.3	2.7
	Biliary atresia	10	5.6	0.6	0.3	1.1
Genitourinary	Hypospadias*	621	83.6	71.1	65.6	76.9
(n = 743)	Renal agenesis/hypoplasia	92	12.4	5.4	4.3	6.6
	Congenital posterior urethral valves*	24	3.2	2.7	1.8	4.1
Musculoskeletal	Clubfoot	212	40.0	12.4	10.8	14.2
(n = 530)	Craniosynostosis	158	29.8	9.3	7.9	10.8
	Gastroschisis	53	10.0	3.1	2.3	4.1
	Limb deficiencies (reduction defects)	53	10.0	3.1	2.3	4.1
	Diaphragmatic hernia	41	7.7	2.4	1.7	3.3
	Omphalocele	30	5.7	1.8	1.2	2.5
Chromosomal	Trisomy 21 (Down syndrome)	206	44.9	12.1	10.5	13.8
(n = 459)	Trisomy 18	33	7.2	1.9	1.3	2.7
	Deletion 22 q11	31	6.8	1.8	1.2	2.6
	Trisomy 13	21	4.6	1.2	0.8	1.9
	Turner syndrome**	18	3.9	2.2	1.3	3.4

**Prevalence limited to male; **Prevalence limited to female*

Appendix D: 2019-2021 Data Tables Continued

Table 3: Prevalence of specific birth defects by organ and chromosomal system and race and ethnicity, 2019-2021

	Non-Hispanic White		Non-Hispanic Black		Hispanic		Non-Hispanic Other	
Birth defects	**n**	**Prevalence, 95%CI**	**n**	**Prevalence, 95%CI**	**n**	**Prevalence, 95%CI**	**n**	**Prevalence, 95%CI**
Total	**2296**	**270.9, 259.9-282.2**	**1791**	**282.4, 269.5-295.8**	**408**	**273.0, 247.1-300.8**	**193**	**258.7, 223.5-297.9**
Central nervous system								
Spina bifida without anencephalus	21	2.5, 1.5-3.8	17	2.7, 1.6-4.3	7	4.7, 1.9-9.7	-	
Anencephalus	11	1.3, 0.6-2.3	-		-		-	
Encephalocele	6	0.7, 0.3-1.5	12	1.9, 1.0-3.3	-		-	
Holoprosencephaly	6	0.7, 0.3-1.5	11	1.7, 0.9-3.1	-	2.7, 0.7-6.9	-	
Eyes								
Anophthalmia/microphthalmia	14	1.7, 0.9-2.8	15	2.4, 1.3-3.9	-		-	
Congenital cataract	13	1.5, 0.8-2.6	12	1.9, 1.0-3.3	-		0	
Ear, face, neck								
Anotia/microtia	14	1.7, 0.9-2.8	11	1.7, 0.9-3.1	9	6.0, 2.8-11.4	0	
Cardiovascular system								
Atrial septal defect	894	105.5, 98.7-112.6	812	128.0, 119.4-137.1	168	112.4, 96.0-130.7	81	108.6, 86.2-135.0
Ventricular septal defect	527	62.2, 57.0-67.7	378	59.6, 53.7-65.9	116	77.6, 64.1-93.1	39	52.3, 37.2-71.5
Pulmonary valve atresia and stenosis	90	10.6, 8.5-13.1	108	17.0, 14.0-20.6	20	13.4, 8.2-20.7	8	10.7, 4.6-21.1
Atrioventricular septal defect	61	7.2, 5.5-9.2	48	7.6, 5.6-10.0	12	8.0, 4.1-14.0	-	
Tetralogy of Fallot	41	4.8, 3.5-6.6	28	4.4, 2.9-6.4	5	3.3, 1.1-7.8	-	
Coarctation of the aorta	40	4.7, 3.4-6.4	25	3.9, 2.6-5.8	-		-	

Transposition of the great arteries	27	3.2, 2.1-4.6	17	2.7, 1.6-4.3	7	4.7, 1.9-9.7	-	
Dextro-transposition of great arteries	23	2.7, 1.7-4.1	13	2.1, 1.1-3.5	5	3.3, 1.1-7.8	-	
Double outlet right ventricle	23	2.7, 1.7-4.1	17	2.7, 1.6-4.3	-		-	
Hypoplastic left heart syndrome	16	1.9, 1.1-3.1	9	1.4, 0.6-2.7	-		-	
Aortic valve stenosis	12	1.4, 0.7-2.5	-		6	4.0, 1.5-8.7	0	
Tricuspid valve atresia and stenosis	10	1.2, 0.6-2.2	0		-		0	
Ebstein anomaly	9	1.1, 0.5-2.0	-		-		-	
Common truncus	8	0.9, 0.4-1.9	-		-		0	
Interrupted aortic arch	7	0.8, 0.3-1.7	7	1.1, 0.4-2.3	-		0	
Tricuspid valve atresia	7	0.8, 0.3-1.7	0		-		0	
Pulmonary valve atresia	6	0.7, 0.3-1.5	5	0.8, 0.3-1.8	0		0	
Total anomalous pulmonary venous connection	6	0.7, 0.3-1.5	-		-		-	
Single ventricle	-		-		-		0	
Orofacial system								
Cleft palate without cleft lip	101	11.9, 9.7-14.5	40	6.3, 4.5-8.6	16	10.7, 6.1-17.4	8	10.7, 4.6-21.1
Cleft lip with cleft palate	58	6.8, 5.2-8.8	30	4.7, 3.2-6.8	15	10.0, 5.6-16.6	7	9.4, 3.8-19.3
Cleft lip without cleft palate	30	3.5, 2.4-5.1	9	1.4, 0.6-2.7	7	4.7, 1.9-9.7	-	
Choanal atresia	12	1.4, 0.7-2.5	-		-		0	
Gastrointestinal system								
Small intestinal atresia/stenosis	38	4.5, 3.2-6.2	31	4.9, 3.3-6.9	7	4.7, 1.9-9.7	-	
Rectal and large intestinal atresia/stenosis	21	2.5, 1.5-3.8	32	5.0, 3.5-7.1	13	8.7, 4.6-14.9	-	

Esophageal atresia/tracheoesophageal fistula	13	1.5, 0.8-2.6	17	2.7, 1.6-4.3	-		0	
Biliary atresia	5	0.6, 0.2-1.4	-		0		-	
Genitourinary system								
Hypospadias*	337	77.4, 69.3-86.1	230	71.5, 62.6-81.4	23	29.8, 18.9-44.8	29	76.2, 51.1-109.5
Renal agenesis/hypoplasia	49	5.8, 4.3-7.6	29	4.6, 3.1-6.6	9	6.0, 2.8-11.4	5	6.7, 2.2-15.6
Congenital posterior urethral valves*	6	1.4, 0.5-3.0	16	5.0, 2.8-8.1	-		-	
Musculoskeletal system								
Clubfoot	98	11.6, 9.4-14.1	93	14.7, 11.8-18.0	18	12.0, 7.1-19.0	-	
Craniosynostosis	94	11.1, 9.0-13.6	44	6.9, 5.0-9.3	13	8.7, 4.6-14.9	7	9.4, 3.8-19.3
Limb deficiencies (reduction defects)	24	2.8, 1.8-4.2	23	3.6, 2.3-5.4	-		-	
Diaphragmatic hernia	21	2.5, 1.5-3.8	16	2.5, 1.4-4.1	-		-	
Gastroschisis	20	2.4, 1.4-3.6	28	4.4, 2.9-6.4	-		-	
Omphalocele	15	1.8, 1.0-2.9	12	1.9, 1.0-3.3	-		0	
Chromosomal system								
Trisomy 21 (Down syndrome)	104	12.3, 10.0-14.9	60	9.5, 7.2-12.2	35	23.4, 16.3-32.6	6	8.0, 3.0-17.5
Deletion 22 q11	20	2.4, 1.4-3.6	9	1.4, 0.6-2.7	-		-	
Trisomy 18	14	1.7, 0.9-2.8	15	2.4, 1.3-3.9	-		0	
Turner syndrome**	10	2.4, 1.2-4.5	6	1.9, 0.7-4.2	-		0	
Trisomy 13	7	0.8, 0.3-1.7	11	1.7, 0.9-3.1	-		0	

*-Case numbers between one and four are not shown; *Prevalence limited to male; **Prevalence limited to female*

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