Louisiana Birth Defects Monitoring Network

2025 Annual Legislative Report

Prepared by:

Bureau of Family Health

Office of Public Health

December 2025

LOUISIANA DEPARTMENT OF HEALTH

Submitted to:

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

By:

- Amy Zapata, MPH, Director, Bureau of Family Health
- Louisiana Department of Health, Office of Public Health, Bureau of Family Health Staff

For inquiries, contact LBDMN@la.gov.

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; Senior Epidemiologist Tri Tran, MD, MPH; and Maternal and Child Health Epidemiologist Coordinator Rimi Mandal, MS.

The Louisiana Birth Defects Monitoring Network surveillance system and public health actions, as described in this report, fulfill the legislative mandate of Louisiana R.S. 40:31.41-31.48[1] 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 Health Resources and Services Administration (HRSA), Department of Health and Human Services (HHS) provided financial support for the Louisiana Birth Defects Monitoring Network. The award provided 100% of total costs and totaled $13,152,546. The contents are those of the author. They may not reflect the policies of HRSA, HHS, or the U.S. Government. For more information, please visit HRSA.gov.

[1] (Legislature, Department of Administration Boards and Commissions Statutory Citations)

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Executive Summary

The Louisiana Department of Health (LDH), Office of Public Health (OPH), Bureau of Family Health (BFH) 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 birth defects surveillance system is authorized and mandated by Louisiana R.S. 40:31.43[2] and is guided by a statutorily defined advisory board.[3] Louisiana R.S. 40:31.45[4] requires the Louisiana Department of Health to produce a report on the results obtained through the surveillance system.

Of the 168,275 children born in Louisiana from 2020 to 2022, 4,739 children, or 2.81%, were diagnosed with at least one birth defect. According to the U.S. Centers for Disease Control and Prevention (CDC), the national average is about 3% of all babies born each year. Louisiana falls below that average. However, deaths due to congenital anomalies or birth defects were one of the top four leading causes of death in infants from birth to 1 year of age in Louisiana from 2020 to 2022.

Among Louisiana children with birth defects born from 2020 to 2022, cardiovascular system defects were the most common (66.4%). The six most common specific birth defects 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.[5] 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 Manning Family Children's in 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.

BFH provides referrals for early intervention services to families of children with birth defects that may affect development. Through the Family Resource Center, the Bureau conducts one-on-one needs assessments with families to offer resources and referrals to services. 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 June 2025, BFH has identified 4,652 families of children with qualifying birth defects who may benefit from a referral needs assessment. The bureau has successfully contacted 1,965 families and linked 75% (1,475 families) to services. The most

[2] (Legislature, Revised Statute 40:31.43, 2001)
[3] (Legislature, RS 40:31.46, 2001)
[4] (Legislature, RS 40:31.45, 2004)
[5] (NCBDDD, 2024)

commonly requested resources among families include family and youth support, health and insurance information, community resources, and disability services.

Introduction

The LDH, OPH, BFH is responsible for surveillance of birth defects in Louisiana's children and moving that data to action by informing recommendations 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. The bureau's broader mission is to promote the health of Louisiana families throughout their lifetime through programs and initiatives to support pregnant women, babies, children, teens, adults, and youth with special health care needs.

Our vision is for Louisiana to be a state where all people are valued and able 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 BFH 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."[6]

Now recognized as the Louisiana Birth Defects Monitoring Network, this system identifies and reports qualifying birth defects diagnosed before 3 years of age statewide in accordance with the definition of birth defect as provided in R.S. 40.31.42.[7] Individual identifying data is confidential and subject to discovery (R.S. 40:31.44).[8] BFH produces this annual report on behalf of the Department per R.S. 40:31.45.[9]

Clinicians, families, and individuals living with birth defects participating in the statutorily required advisory board developed the Louisiana Birth Defects Monitoring Network.[10] The advisory board members are subject matter experts who provide expertise and perspective to guide BFH 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. The Louisiana Birth Defects Monitoring Network posts notices of meetings, agendas, and minutes on the

[6] (Legislature, Act 194, 2001)
[7] (Legislature, RS40:31.42, 2001)
[8] (Legislature, RS 40:31.44, 2001)
[9] (Legislature, RS 40:31.45, 2004)
[10] (Health L. D., Boards-Commissions-Councils-Family-Health-Louisiana, 2025)

Louisiana Boards and Commissions webpage and the Bureau of Family Health's Boards and Commissions webpage.

BFH 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. In accordance with Act 393 of the 2023 Regular Session of the Louisiana Legislature (Legislature, Act 393), the bureau received a legal opinion confirming that the Louisiana Birth Defects Monitoring Network's Advisory Board meets the requirement of "strictly advisory" established in Louisiana R.S. 42:17 (F)(2).[11] Thus, the advisory board is allowed to conduct its meetings via electronic means. In 2025, the bureau worked to update the board roster by contacting current members to assess their intent to serve so that vacant positions can be filled with new members who possess diverse expertise and retain active members who have demonstrated engagement with the program and a willingness to attend meetings. In addition, the bureau developed a new member orientation and onboarding guide to provide clear expectations for advisory board members and increase active participation on the board.

While the advisory board's inability to meet quorum did not significantly disrupt operations to identify infants and young children with birth defects and connect them to resources, certain projects requiring their direct oversight were postponed. The 2023 the 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 the open meetings law.

Louisiana Birth Defects Monitoring Network

BFH 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 maternal and child health programs that identify and support children and youth with special health care needs and their families.

From January 2005 through June 2025, BFH investigated potential birth defects among 66,769 children. Just under 3% were identified with birth defects within the Louisiana Birth Defects Monitoring Network case definition. The bureau identifies approximately 1,500 to 2000 children with specified birth defects annually, averaging 260 to 280 per 10,000 live births.

BFH 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, support efforts in the state to improve maternal and child health outcomes, and

[11] (Legislature, Act 393)

prevent new occurrences of birth defects. The bureau can evaluate concerns about unexpected groups of birth defects (cluster investigations), as well as the effectiveness of preventive interventions.
As a part of BFH's system of monitoring birth outcomes in the state, the Louisiana Birth Defects Monitoring Network supports:

- **Policymakers** by identifying risk factors, such as maternal exposures and chronic conditions potentially linked to specific birth conditions, and recognizing preventive strategies to decrease birth defects;
- **Families of infants with birth defects from birth through 3 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 the Louisiana Birth Defects Monitoring Network webpage; and
- **Researchers from the CDC, universities, and other states** by investigating possible causes of specific birth defects.

BFH also provides the following:

- Active public health surveillance of hospital discharges of newborns until 3 years of age for major structural, functional, or genetic birth defects;
- Links to resources for families of children under 3 years of age with specified birth defects to health, social service, and developmental resources through the BFH Family Resource Center; and
- 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, 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; and
    - Spina Bifida of Louisiana.

Additional Reporting

In addition to providing annual reports to the Louisiana Legislature, the Secretary of LDH, and the advisory board, the Louisiana Birth Defects Monitoring Network includes its data in biennial reports produced by the National Birth Defects Prevention Network and special reports in the Birth Defects Research Journal. BFH reported birth defects data from the 2018-2022 birth years to the National Birth Defects Prevention Network in September 2025 for publication in January 2026.

In July 2025, BFH supplied birth defects prevalence and counts for data years spanning 2005 through 2020 birth years to the LDH, OPH's Environmental Public Health Tracking system, which provides data and information on health outcomes, the environment, population, and exposures. Louisiana's Environmental Public Health Tracking is one of the 33 entities (state and local health departments, cities, and jurisdictions) that are a 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 3 years of age. Staff review records from all birthing hospitals and pediatric hospitals in Louisiana. BFH maintains a longitudinal data system of all children born in Louisiana who are diagnosed with a structural, functional, or genetic birth defect. Maternal and child health epidemiologists statistically analyze de-identified medical record data for patterns and trends over time. Through the Family Resource Center, the Bureau links families to health, social service, and developmental resources for children identified with specified birth defects.

Case Inclusion

The following are requirements for BFH to review a case:

- The child must have a major structural, functional, or genetic birth defect. Major defects are those that can adversely affect the child's health and development. The bureau does not include children with 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.
- The 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, meaning the mother was pregnant for at least 20 weeks before the birth. In the absence of an age estimate, the infant must have a birth weight of at least 350 grams.

Methodology

BFH 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.

BFH 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, BFH includes children diagnosed before their third birthday, allowing adequate time to capture all birth defects within the case definition. Additionally, this timeframe allows hospitals adequate time for 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. Because birth defects are rare, especially when categorized by race and ethnicity, BFH reports data from three birth years to ensure enough case numbers for meaningful analysis. Refer to Appendix B for a complete list of International Classification of Diseases-Version 10 birth defects codes with corresponding description labels, CDC/British Pediatric Association Classification of Diseases (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 at any time during pregnancy, most occur within the first three months during rapid cell growth and organ development."[12]

Birth defects vary from cosmetic differences to the medically complex, which can 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 often 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.[13]

Infant Mortality: Birth to 1 Year

In Louisiana, from 2020 to 2022, the four leading causes of death in infants from birth to 1 year were deaths from conditions originating in the perinatal period, sudden unexpected infant deaths, deaths due to congenital anomalies or birth defects, and external causes of mortality such as injury.[14]

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.

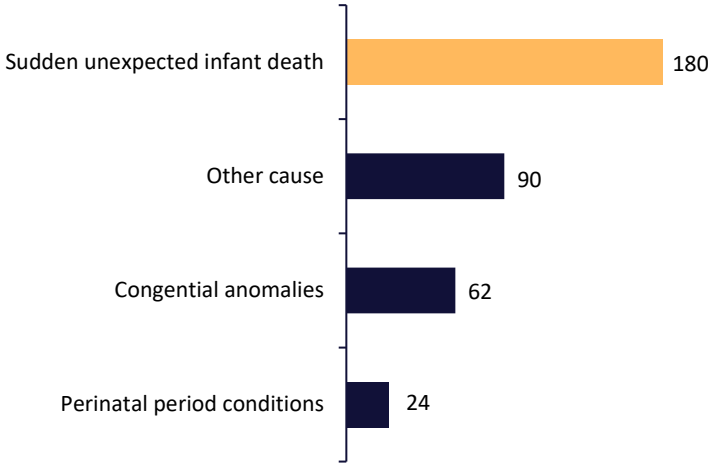
For more information on infant mortality, the latest Child Death Review report is available at partnersforfamilyhealth.org/childdeathreview.

[12] (CDC, CDC Birth Defects)
[13] (NCBDDD, 2024)
[14] (Child Death Review, 2020-2022)

From 2020-2022 in Louisiana, an average of 62 infants per year died from congenital anomalies.

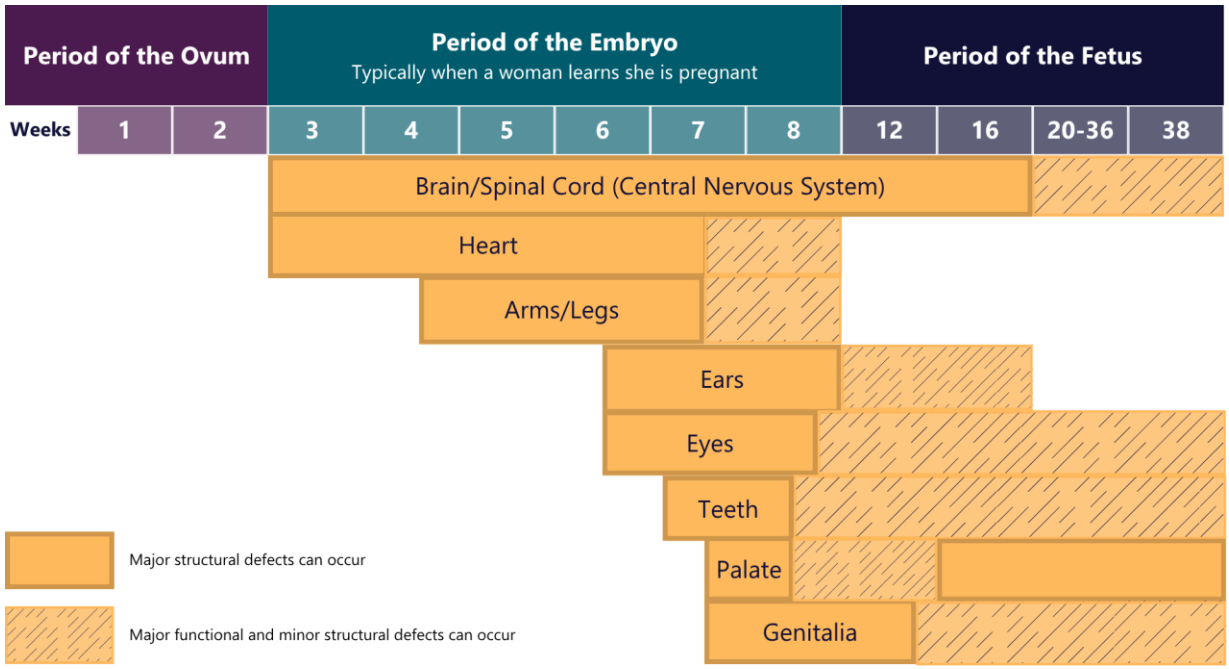
In Louisiana each year, an average of:

- One hundred eighty infants died from conditions originating in the perinatal period;
- Ninety infant deaths were classified as sudden unexpected infant deaths, which primarily occurred in the sleep environment;
- Sixty-two infants died from congenital anomalies; and
- Twenty-four infants died from other causes, including injury.



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,[15]

Not all birth defects are preventable. However, 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, and some prescribed medications;
- Obesity; and
- 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.[16] [17]

Preventing Birth Defects in Louisiana

BFH provides birth defects prevention education and materials for providers, community partners, and individuals. While not all birth defects are preventable, strengthening prevention messaging and improving systems of care for women throughout their childbearing years can reduce preventable birth defects.

The following 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 health care 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 can become pregnant or are 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**.

[15] (Bleyl SB, Schoenwolf GC, 2010, Carlson BM, 2008, Cochard LR, 2012, Moore KL 2013,, 2023)
[16] (Good Health Before Pregnancy: Prepregnancy Care)
[17] (Network M. , 2025)

This resource is adapted from March of Dimes.[18]

Additional Resources

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

Advanced Pediatric Care in Louisiana

Birth defects require specialized care at birth, during infancy and 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, families and children must travel to Manning Family Children's or The Gayle and Tom Benson Ochsner Children's Hospital, both in 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:

- Manning Family Children's: 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;
- The Gayle and Tom Benson Ochsner Children's Hospital: Neonatal intensive care unit, pediatric intensive care unit, cardiac intensive care unit, emergency department, acute care, and 30+ pediatric specialty clinics;
- Our Lady of the Lake Children's Hospital: Neonatal intensive care unit, pediatric intensive care unit, emergency department, acute care, and 30 pediatric specialty clinics;
- Ochsner LSU Shreveport Pediatrics: Neonatal intensive care unit, pediatric intensive care unit, emergency department, and 20+ pediatric specialty clinics; and
- Shriners Children's Shreveport: Outpatient orthopedic and craniofacial specialty clinics.

Children with birth defects often qualify for Medicaid to support their complex medical needs. Most children with birth defects and complex medical conditions use Medicaid to provide medical care, allied health services, and early interventions to improve quality of life and minimize the risk of developmental impacts.

[18] (Dolan, 2020)

Birth Defects Findings in Louisiana 2020-2022 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, BFH reports data in three-year birth year increments at the state level. This report presents birth defects data among children born from 2020 to 2022. For awareness, a single 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 (genes), 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 during pregnancy with known risk factors and specialists in Louisiana who can provide treatment to address particular birth defects.

Findings at a Glance

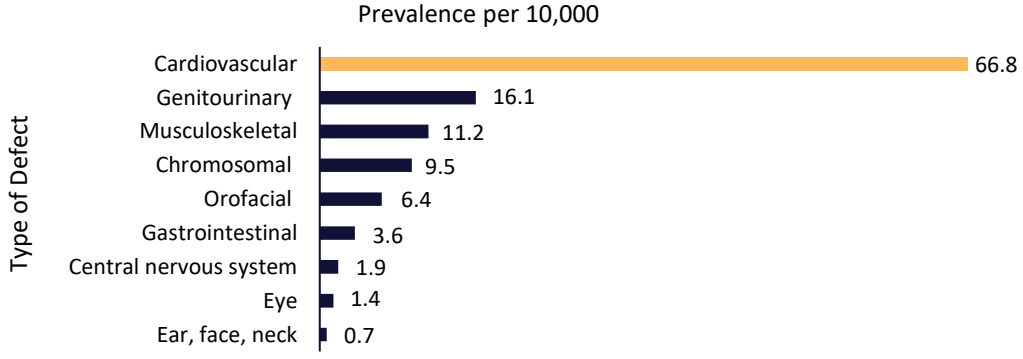
Of the 168,275 children born in Louisiana between 2020 and 2022, 4,739 children, or 2.81%, were diagnosed with at least one birth defect.

Among children born from 2020 to 2022, 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).

Number of Children per 10,000 Live Births Diagnosed with Birth Defects

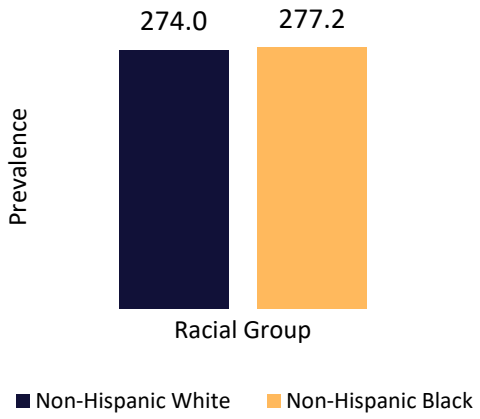
Atrial Septal Defect	Hypospadias	Ventricular Spetal Defects	Pulmonary Valve Artresia/Stenosis	Clubfoot	Down Syndrome
116.5	75.8	69.1	16	13.3	12.5

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

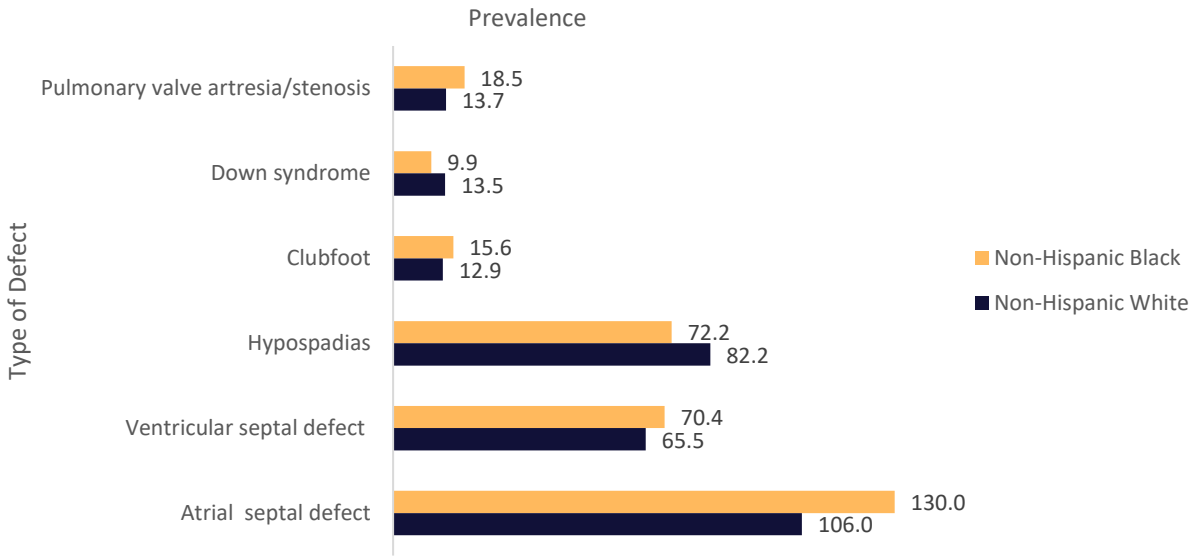


Racial Disparities in Birth Defects

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



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



Non-Hispanic white infants were almost twice as likely to have cleft palate without cleft lip (11.7 vs 6.0).*

Pulmonary valve atresia and stenosis were seen more often **among non-Hispanic Black infants (18.5)** than **non-Hispanic white infants (13.7).***

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

Cardiovascular System

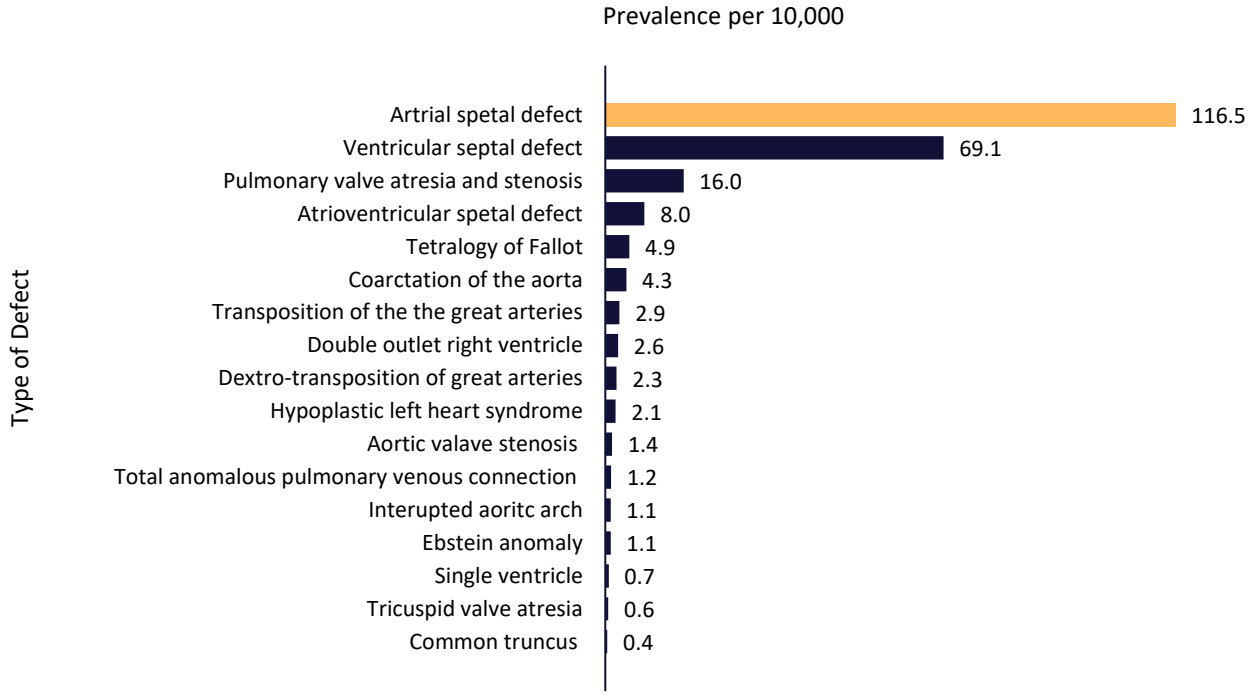
Congenital heart defects affect how blood flows through the heart and out to the rest of the body. Heart defects range in complexity from those that resolve with medical monitoring only to those that require surgical intervention. Newborn screening through pulse oximetry may detect critical heart defects 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: These include uncontrolled diabetes, maternal smoking, and certain medications during pregnancy.

Where to Receive Care: 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, or receive care out of state.

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



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

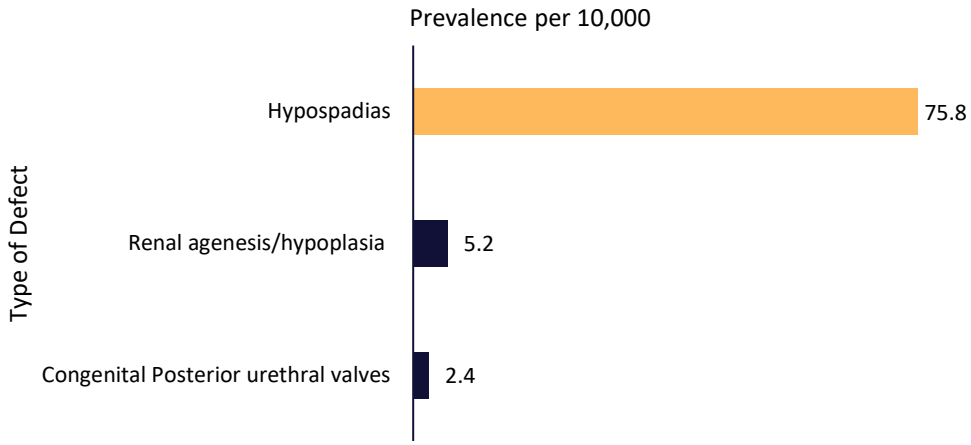
The urethra is the tube that carries urine from the bladder to the outside of the body. Hypospadias is an opening of the urethra not located at the tip of the penis. Most cases require surgical correction between 3 and 18 months. Posterior urethral valves is an obstruction of urine flow by abnormal membranes 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: These include maternal age of 30 or older, maternal obesity, assisted reproductive technology, and progestin use just before or during pregnancy.

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

Hypospadias is the leading genitourinary birth defect (85.7%).

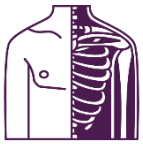


Hypospadias: *The opening of the urethra is not located at the tip of the penis.*

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.

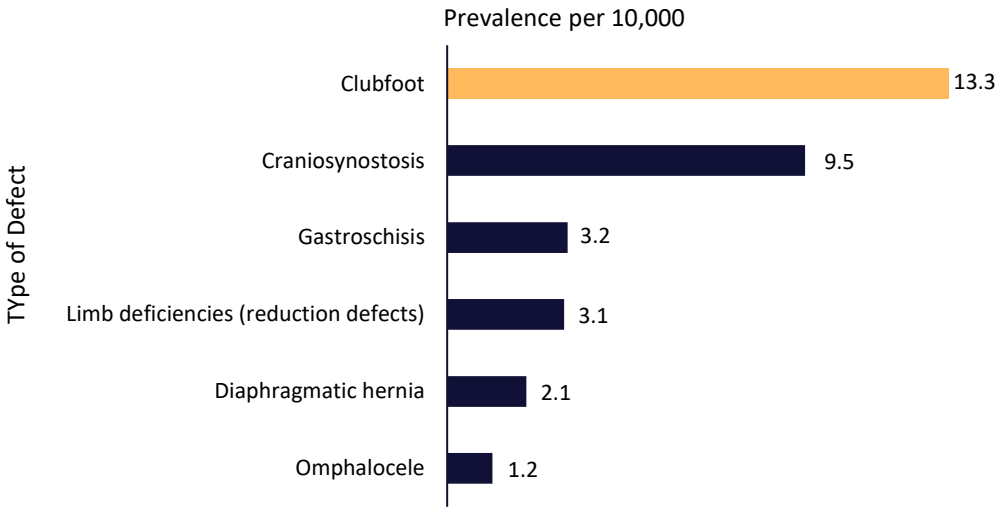


Critical Period of Development: The fetal skeletal system develops between weeks three through eight of pregnancy.

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

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

Clubfoot is the leading musculoskeletal birth defect (42%).



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

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

Chromosomal System

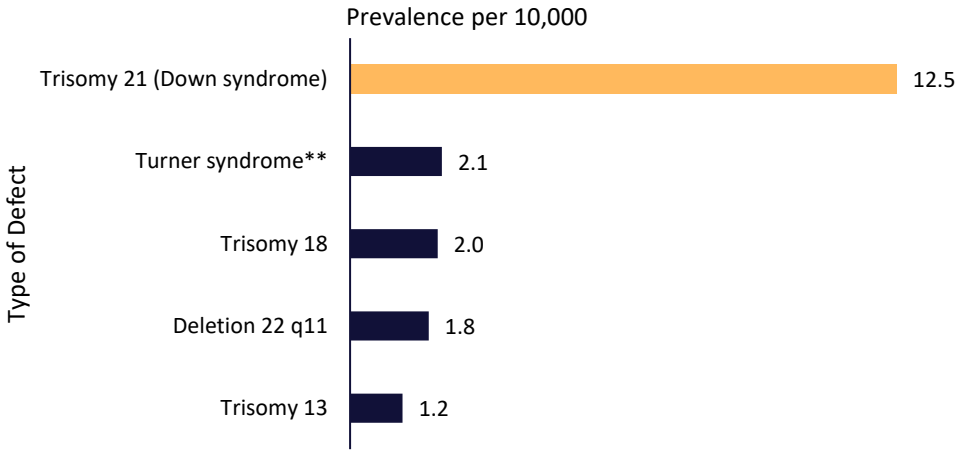
Chromosomal defects arise from abnormalities in the structure or number of chromosomes within a cell's genes, such as extra chromosomal material, missing chromosomal segments, or the absence of entire chromosomes. Chromosomes determine how the body forms and functions. Chromosomal rearrangements may 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: These include advanced maternal age (35+), family history of genetic diagnoses, radiation, and chemical exposure during pregnancy.

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

Down syndrome is the leading chromosomal birth defect (46.8%).



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 are included in Appendix C: Birth Defects Definitions.

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

Orofacial System

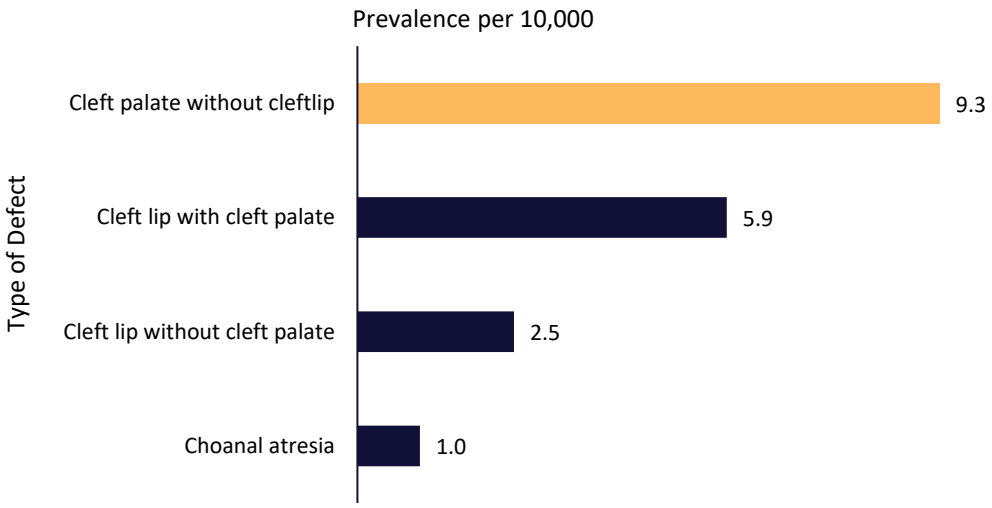
Cleft lips and cleft palates occur when the tissue of the lips and inside of the mouth do not completely join. Openings may happen anywhere along the top lip or roof of the mouth, and may or may not affect teeth, gums, nose, and uvula. Most clefts are diagnosed during pregnancy or at birth. Submucosal clefts (opening in the soft palate covered by a thin membrane) and bifid (split) 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: These include smoking during pregnancy, maternal diabetes, certain epilepsy medications during pregnancy, and genetic factors.

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

Cleft palate without cleft lip is the leading orofacial birth defect (52%).



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

Cleft lip: *Incomplete fusion of the lips.*

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

Gastrointestinal System

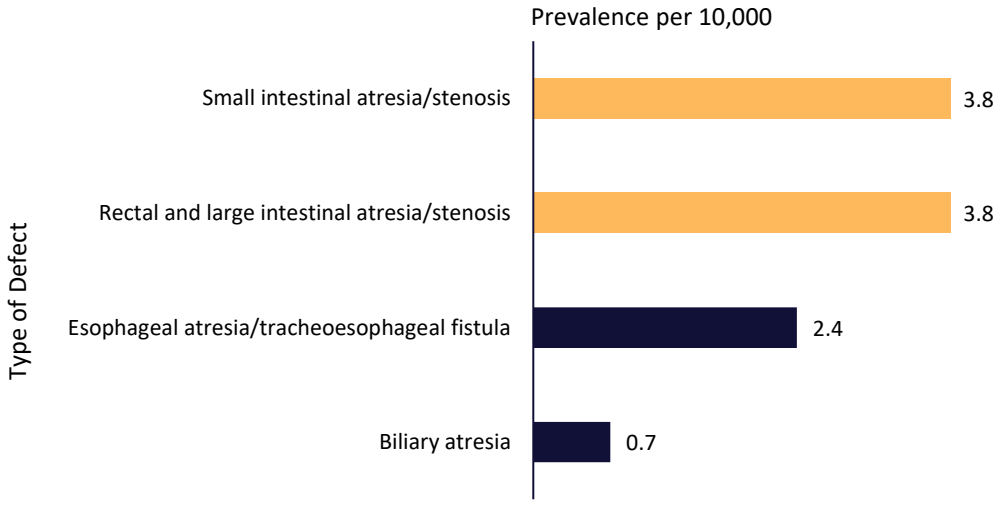
The gastrointestinal (GI) tract begins at the lips and ends at the anus. Gastrointestinal defects occur when connections are interrupted or underdeveloped. The liver and gallbladder are included in this category. The gastrointestinal tract is one of the first things to form and continues to develop throughout pregnancy. Therefore, premature babies have a higher rate of gastrointestinal challenges. Most gastrointestinal 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: These include genetic factors, use of assisted reproductive technology, or older paternal age (esophageal atresia).

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

Small intestinal atresia/stenosis and rectal and large intestinal atresia are the leading gastrointestinal birth defects (37.2%).



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

Rectal and large intestinal atresia/stenosis: *Complete or partial blockage due to narrowing of one or more segments of the large intestine and/or rectum.*

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

Central Nervous System

Central nervous system defects, also called neural tube defects, 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.

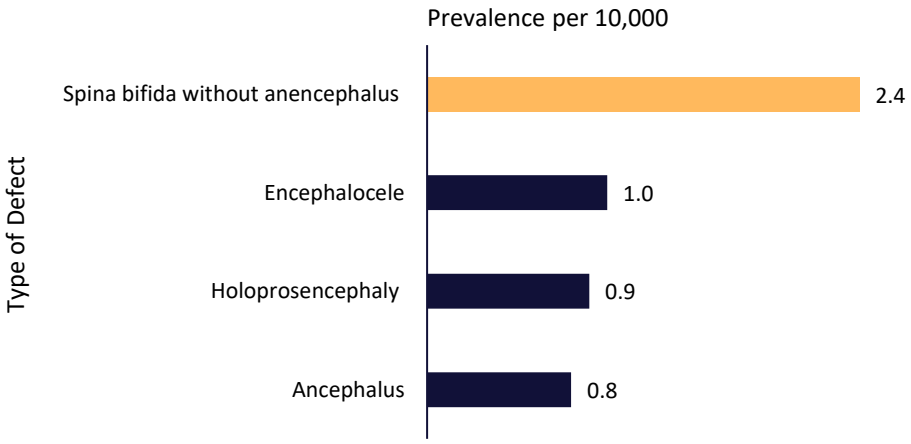
Getting 400 micrograms (mcg) of folic acid daily before and during early pregnancy can help prevent neural tube defects. These defects range in severity, from partial or complete absence of the brain, called anencephaly, to openings anywhere along the spinal cord, known as spina bifida. Neural tube defects may be diagnosed by ultrasound during pregnancy. Anencephaly is incompatible with life. Spina bifida may be eligible for repair before birth 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 in week three and continuing through the entire pregnancy.

Known Risk Factors: These include 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: 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 (43.5%).



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 form 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.

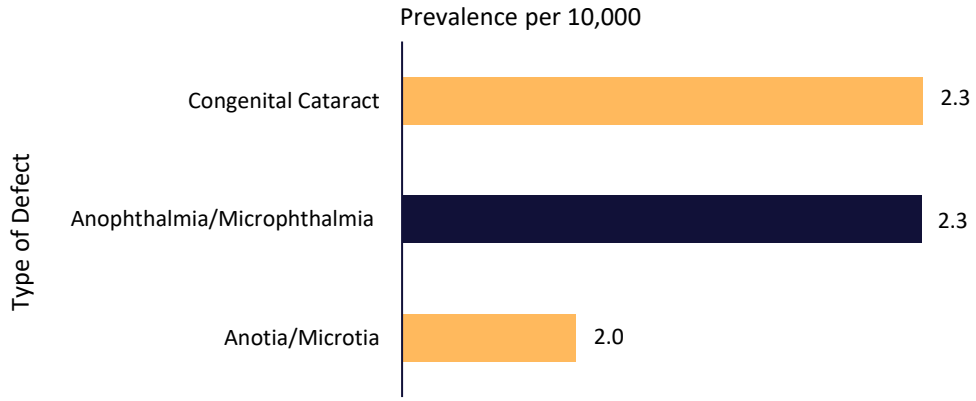
Anotia (absent ear) and microtia (small ear) defects range from cosmetic to obstructed hearing. Both require varying levels of audiology and plastic surgery management into early childhood. Eye and ear anomalies may be isolated or associated with certain genetic syndromes. Therefore, medical management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

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

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

Where to Receive Care: Pediatric ophthalmologists and pediatric ear, nose, and throat doctors are located in New Orleans, Baton Rouge, Lafayette, and Shreveport.

Congenital cataracts are the leading eye birth defects (58.2%).
Anotia/microtia are the leading ear defects (100%).



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

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: 2020-2022 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 understanding how to navigate those often-fragmented systems. The Bureau of Family Health 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, 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, from birth to 3 years of age (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 June 2025, BFH has identified 4,652 families of children with qualifying birth defects who may benefit from a referral needs assessment. The Bureau has successfully contacted more than 1,965 families and linked 75% (approximately 1,475) of those families to services. The most requested resources among families include insurance navigation, along with parent support and systems of care navigation services.

BFH uses multiple means to contact families, including mail, phone, email, and text messaging. The bureau also uses accommodation supports, such as the Language Line to communicate with families using non-English languages and Telecommunications Relay Services to communicate with people who are deaf or hard of hearing. 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.

Expanding Referrals

In 2024 and 2025, the Louisiana Birth Defects Monitoring Network increased the number of referrals to the Family Resource Center by developing protocols to include children who may benefit from a referral needs assessment, but who do not meet the Louisiana Birth Defects Monitoring Network case definition for inclusion in the registry. This includes children 3 three years old with developmental and medical involvement due to prematurity or a birth defect or medical condition outside of the National Birth Defects Prevention Network standards for reporting.

BFH offers resources 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 from 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, from the CDC Data and Statistics on Birth Defects, and on the Louisiana Environmental Public Health Tracking Network health data portal to enable analysis, visualization, and reporting. This data is available to environmental and public health practitioners, health care 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 BFH 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 birth defects data to action in 2025 by informing timely referrals to services for families.

Program goals for 2026 include revitalizing the advisory board, increasing the number of referrals of children to the BFH Resource Center, and achieving and maintaining the highest standards of quality indicators for birth defects surveillance.

Advisory Board Goal

In 2025, the bureau reviewed the membership roster and identified inactive members to remove from the board. In 2026, the Louisiana Birth Defects Monitoring Network will focus on filling board vacancies with actively engaged pediatric health providers, ensuring quorum is met at all open meetings to facilitate effective decision-making and the provision of valuable recommendations. Recruitment strategies include outreach to potential members recommended by the Louisiana Medical Society, Louisiana pediatric hospitals, local organizations serving families with children with special health care needs, and interested partners who have expressed interest in or used birth defects data for research.

The advisory board will work with BFH 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 Case Review Model 2.0 to critical congenital heart defects, neural tube defects, Trisomy 21, abdominal wall defects, and limb reduction defects. See Appendix E for model framework.

Referral Goal

Each year, Louisiana Birth Defects Monitoring Network works to increase the number of children referred to the Family Resource Center. In 2026, BFH will develop protocols to assess the needs of children of military families who may benefit from referrals. This includes children under 3 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.[19] Because the U.S. Department of Defense has a birth defects surveillance system, Louisiana does not include children of military service members in our registry to avoid duplications in prevalence. However, we recognize those service member families need local resources while serving in Louisiana. In May 2025, Louisiana Birth Defects Monitoring Network staff identified and had an introductory meeting with the Department of Defense Military Community Clinical Integration lead, who works as a clinical psychologist, the Complex Pediatric Clinical Community program manager, and the clinical lead for Autism Care Demonstration. This contact was extremely interested in connecting families with the Family Resource Center. As children of military service members are identified with needs in the course of birth defects surveillance, Louisiana Birth Defects Monitoring Network staff will refer them to the Family Resource Center for outreach and individual needs assessments.

Surveillance Quality Goal

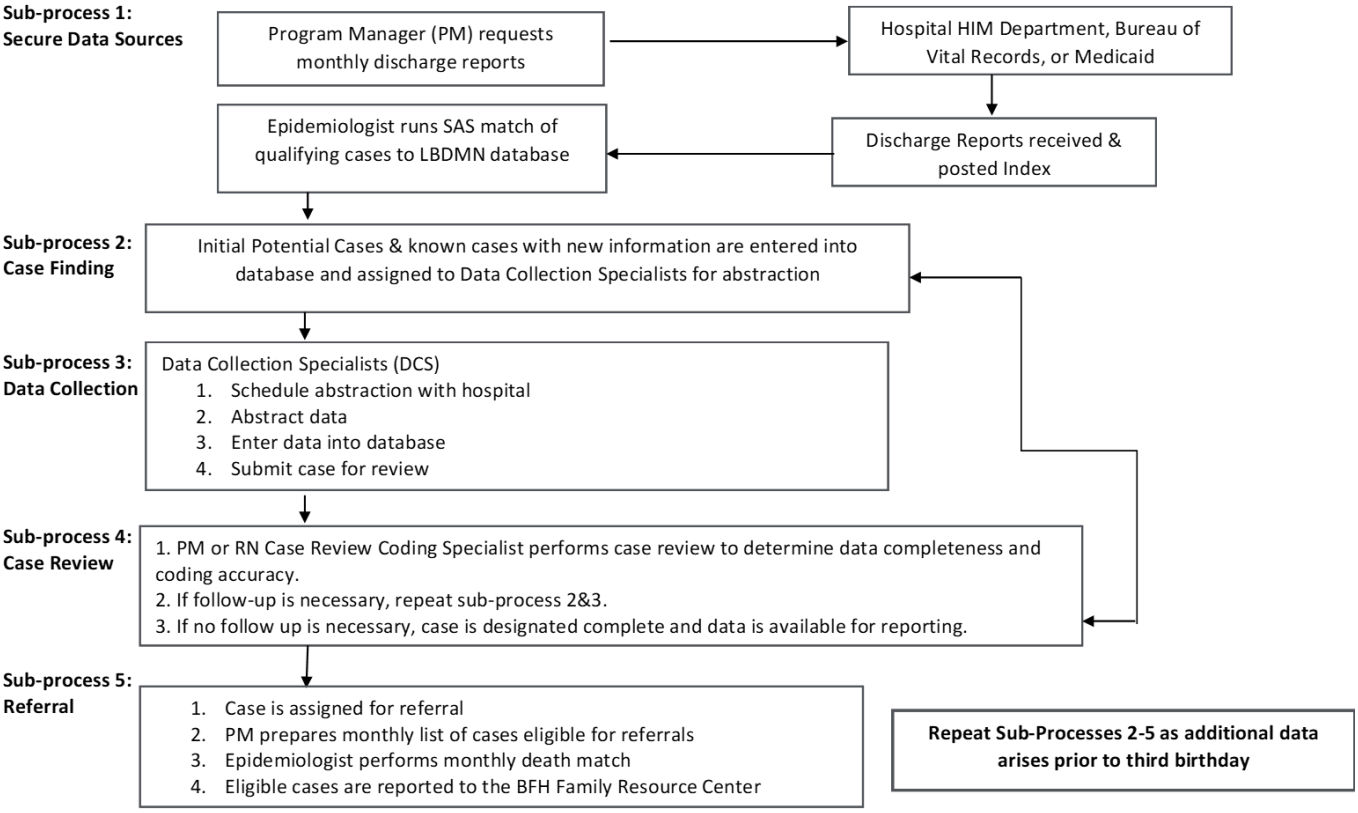
Our primary goal in 2026 is to achieve and maintain the highest data quality standards for accuracy, completion, and timeliness of state birth defects surveillance programs by identifying, processing, and completing 98% of initial potential cases within 45 days of identification. This will meet the National Birth Defects Prevention Network's highest standard of data quality by completing 99% of all core and recommended data within two years of birth.

[19] (NCBDDD, 2024)

Appendix

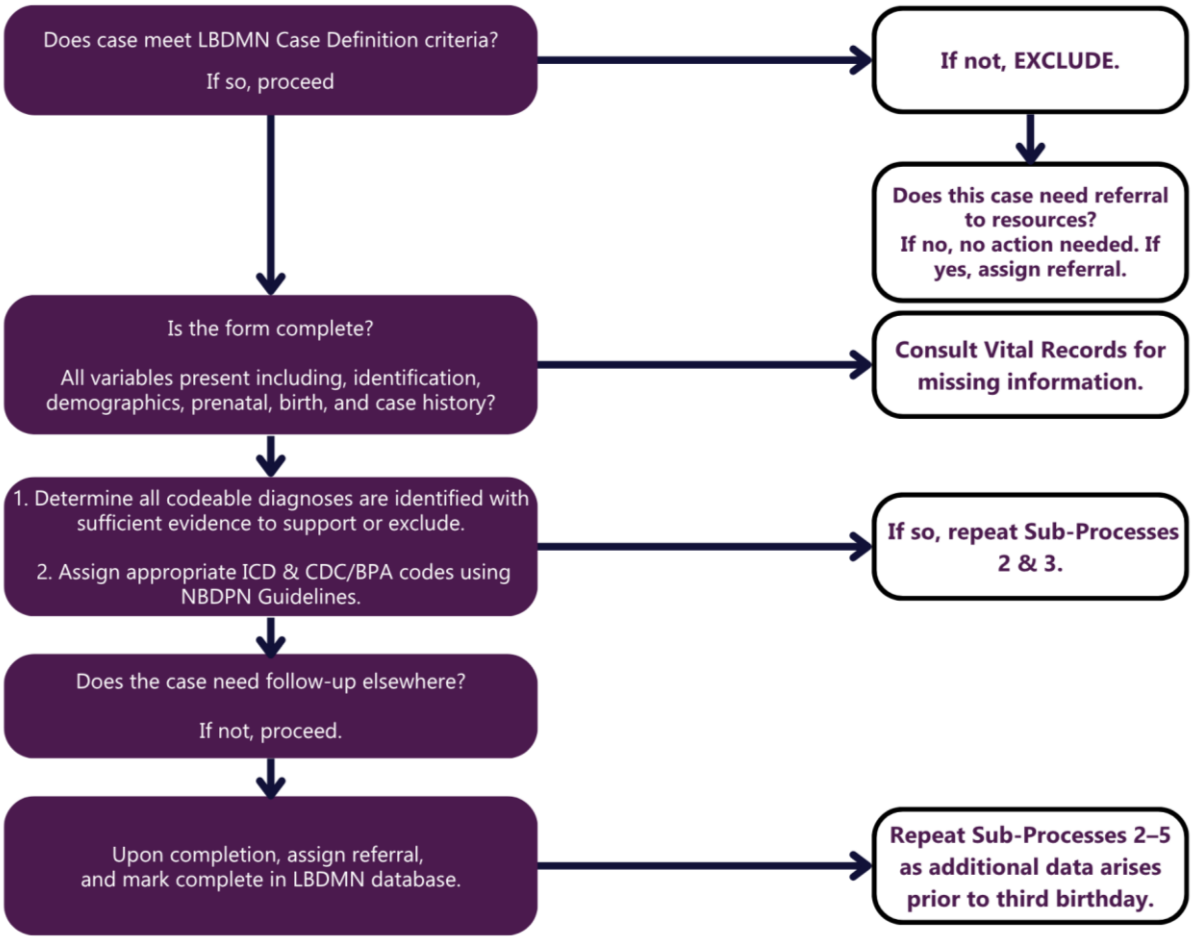
Appendix A: Case Ascertainment/Review/Quality Assurance Process Chart

This chart depicts how we identify cases, abstract, and review data to 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 description labels, Centers for Disease Control and Prevention/British Pediatric Association codes,[20] 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 cases with birth defects 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

[20] (Sever, 2021)

Appendix B: Birth Defects Codes, Descriptions, Referral Logic

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 Imp
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 sex chromosome abnormalities, female phenotype	CODING	758.810	YES

Appendix B: Birth Defects Codes, Descriptions, Referral Logic

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, males 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.[21]

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 valve that controls blood flow from the heart to the lung does not 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 (a hole in the wall [septum] that separates the two lower chambers [ventricles] of the heart.)

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**.**

[21] (Sever, 2021)

Appendix C: Birth Defects Definitions Continued

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.

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 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 chest 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 are usually covered by a nearly transparent membranous sac.

Appendix C: Birth Defects Definitions Continued

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 child with the observable characteristics of a female.

Deletion 22 q11: Chromosome abnormality resulting from 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.

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 upper region of the pharynx on either side.

Gastrointestinal System

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

Rectal and large intestinal atresia/stenosis: Complete or partial blockage due to narrowing 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 internal opening of the extrahepatic bile ducts of the liver. The extrahepatic bile ducts include the hepatic duct (formed by the two main ducts that carry bile out of the liver), the cystic duct (which carries bile out of the gallbladder where it is stored), and the common bile duct (formed by the junction of the hepatic duct and the cystic duct), which carries bile into the duodenum (small intestine) for excretion.

Appendix C: Birth Defects Definitions Continued

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, or push through the vertebrae**.**

Encephalocele: Herniation of brain tissue and/or meninges (membrane of the spinal cord) through a defect in the skull. The hernia sac is usually 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 (also called anencephalus): The 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: 2020-2022 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, 2020-2022 (n = 4,739)

Organ and chromosomal system	Number	Percent*
Cardiovascular	3165	66.8
Genitourinary	761	16.1
Musculoskeletal	533	11.2
Chromosomal	449	9.5
Orofacial including Choanal atresia	302	6.4
Gastrointestinal	172	3.6
Central nervous system	92	1.9
Eye	67	1.4
Ear, face and neck	34	0.7
Other**	-	-

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

***Numbers between one and four are suppressed for confidentiality protections*

Appendix D: 2020-2022 Data Tables Continued

Table 2: Prevalence of specific birth defects by organ and chromosomal system, 2020-2022 (N = 168,275 live births)

System	Birth defects	Number	%	Prev.	95%CI	
	Total	**4739**		**281.6**	**273.7**	**289.8**
Central nervous	Spina bifida without anencephalus	40	43.5	2.4	1.7	3.2
(n = 92)	Encephalocele	16	17.4	1.0	0.5	1.5
	Holoprosencephaly	15	16.3	0.9	0.5	1.5
	Anencephalus	14	15.2	0.8	0.5	1.4
Eyes	Congenital cataract	39	58.2	2.3	1.6	3.2
(n = 67)	Anophthalmia/microphthalmia	38	56.7	2.3	1.6	3.1
Ear, face, neck	Anotia/microtia	34	100.0	2.0	1.4	2.8
(n = 34)						
Cardiovascular	Atrial septal defect	1960	61.9	116.5	111.4	121.7
(n = 3165)	Ventricular septal defect	1162	36.7	69.1	65.1	73.1
	Pulmonary valve atresia and stenosis	269	8.5	16.0	14.1	18.0
	Atrioventricular septal defect (Endocardial cushion defect)	134	4.2	8.0	6.7	9.4
	Tetralogy of Fallot (TOF)	82	2.6	4.9	3.9	6.0
	Coarctation of the aorta	72	2.3	4.3	3.3	5.4
	Transposition of the great arteries (TGA)	49	1.5	2.9	2.2	3.9
	Double outlet right ventricle (DORV)	44	1.4	2.6	1.9	3.5
	Dextro-transposition of great arteries	38	1.2	2.3	1.6	3.1
	Hypoplastic left heart syndrome	35	1.1	2.1	1.4	2.9
	Pulmonary valve atresia	24	0.8	1.4	0.9	2.1
	Aortic valve stenosis	23	0.7	1.4	0.9	2.1
	Total anomalous pulmonary venous connection (TAPVC)	20	0.6	1.2	0.7	1.8
	Interrupted aortic arch (IAA)	19	0.6	1.1	0.7	1.8
	Ebstein anomaly	18	0.6	1.1	0.6	1.7

	Tricuspid valve atresia and stenosis	16	0.5	1.0	0.5	1.5
	Single Ventricle	12	0.4	0.7	0.4	1.2
	Tricuspid valve atresia	10	0.3	0.6	0.3	1.1
	Common truncus (truncus arteriosus or TA)	7	0.2	0.4	0.2	0.9
Oro-facial	Cleft palate without cleft lip	157	52.0	9.3	7.9	10.9
(n = 302)	Cleft lip with cleft palate	100	33.1	5.9	4.8	7.2
	Cleft lip without cleft palate	42	13.9	2.5	1.8	3.4
	Choanal atresia	16	5.3	1.0	0.5	1.5
Gastrointestinal	Small intestinal atresia/stenosis	64	37.2	3.8	2.9	4.9
(n = 172)	Rectal and large intestinal atresia/stenosis	64	37.2	3.8	2.9	4.9
	Esophageal atresia/tracheoesophageal fistula	40	23.3	2.4	1.7	3.2
	Biliary atresia	12	7.0	0.7	0.4	1.2
Genitourinary	Hypospadias*	652	85.7	75.8	70.1	81.8
(n = 761)	Renal agenesis/hypoplasia	88	11.6	5.2	4.2	6.4
	Congenital posterior urethral valves*	21	2.8	2.4	1.5	3.7
Musculoskeletal	Clubfoot	224	42.0	13.3	11.6	15.2
(n = 533)	Craniosynostosis	160	30.0	9.5	8.1	11.1
	Gastroschisis	53	9.9	3.2	2.4	4.1
	Limb deficiencies (reduction defects)	52	9.8	3.1	2.3	4.1
	Diaphragmatic hernia	36	6.8	2.1	1.5	3.0
	Omphalocele	21	3.9	1.2	0.8	1.9
Chromosomal	Trisomy 21 (Down syndrome)	210	46.8	12.5	10.8	14.3
(n = 449)	Trisomy 18	34	7.6	2.0	1.4	2.8
	Deletion 22 q11	31	6.9	1.8	1.3	2.6
	Trisomy 13	21	4.7	1.2	0.8	1.9
	Turner syndrome**	17	3.8	2.1	1.2	3.3

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

Appendix D: 2020-2022 Data Tables Continued

Table 3: Prevalence of specific birth defects by organ and chromosomal system and race and ethnicity, 2020-2022

	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	**2280**	**274.0**	**285.5**	**1786**	**290.5**	**277.2**	**304.3**	**465**	**293.4**	**267.3**	**321.3**	**203**	**268.6**	**232.9**	**308.1**
Central nervous system															
Spina bifida without anencephalus	18	2.2, 1.3-3.4		15	2.4, 1.4-4.0			6	3.8, 1.4-8.2			-			
Anencephalus	8	1.0, 0.4-1.9		-				-				-			
Encephalocele	6	0.7, 0.3-1.6		8	1.3, 0.6-2.6			-				0			
Holoprosencephaly	-			9	1.5, 0.7-2.8			-				0			
Eyes															
Congenital cataract	22	2.6, 1.7-4.0		10	1.6, 0.8-3.0			6	3.8, 1.4-8.2			-			
Anophthalmia/ microphthalmia	16	1.9, 1.1-3.1		15	2.4, 1.4-4.0			6	3.8, 1.4-8.2			-			
Ear, face, neck															
Anotia/microtia	16	1.9, 1.1-3.1		11	1.8, 0.9-3.2			7	4.4, 1.8-9.1			0			
Cardiovascular system															
Atrial septal defect	882	106.0, 99.1-113.2		799	130.0, 121.1-139.3			192	121.1, 104.6-139.5			84	111.1, 88.6-137.6		
Ventricular septal defect	545	65.5, 60.1-71.2		433	70.4, 63.9-77.4			145	91.5, 77.2-107.6			38	50.3, 35.6-69.0		
Pulmonary valve atresia and stenosis	114	13.7, 11.3-16.5		114	18.5, 15.3-22.3			29	18.3, 12.3-26.3			12	15.9, 8.2-27.7		
Atrioventricular septal defect	61	7.3, 5.6-9.4		55	8.9, 6.7-11.6			12	7.6, 3.9-13.2			6	7.9, 2.9-17.3		
Coarctation of the aorta	40	4.8, 3.4-6.5		24	3.9, 2.5-5.8			8	5.0, 2.2-9.9			0			
Tetralogy of Fallot (TOF)	39	4.7, 3.3-6.4		37	6.0, 4.2-8.3			-				-			

Transposition of the great arteries (TGA)	28	3.4, 2.2-4.9	11	1.8, 0.9-3.2	9	5.7, 2.6-10.8	-	
Double outlet right ventricle (DORV)	24	2.9, 1.8-4.3	17	2.8, 1.6-4.4	-		-	
Dextro-transposition of great arteries	23	2.8, 1.8-4.1	9	1.5, 0.7-2.8	5	3.2, 1.0-7.4	-	
Hypoplastic left heart syndrome	14	1.7, 0.9-2.8	13	2.1, 1.1-3.6	5	3.2, 1.0-7.4	-	
Pulmonary valve atresia	11	1.3, 0.7-2.4	12	2.0, 1.0-3.4	-		0	
Aortic valve stenosis	11	1.3, 0.7-2.4	5	0.8, 0.3-1.9	7	4.4, 1.8-9.1	0	
Interrupted aortic arch (IAA)	10	1.2, 0.6-2.2	7	1.1, 0.5-2.3	-		-	
Tricuspid valve atresia and stenosis	9	1.1, 0.5-2.1	-		-		0	
Ebstein anomaly	8	1.0, 0.4-1.9	5	0.8, 0.3-1.9	-		-	
Tricuspid valve atresia	6	0.7, 0.3-1.6	-		-		0	
Total anomalous pulmonary venous connection (TAPVC)	6	0.7, 0.3-1.6	-		7	4.4, 1.8-9.1	-	
Single Ventricle	-		-		5	3.2, 1.0-7.4	0	
Common truncus (truncus arteriosus or TA)	-		-		-		0	
Oro-facial system								
Cleft palate without cleft lip	97	11.7, 9.5-14.2	37	6.0, 4.2-8.3	17	10.7, 6.2-17.2	6	7.9, 2.9-17.3
Cleft lip with cleft palate	48	5.8, 4.3-7.6	33	5.4, 3.7-7.5	15	9.5, 5.3-15.6	-	
Cleft lip without cleft palate	24	2.9, 1.8-4.3	10	1.6, 0.8-3.0	6	3.8, 1.4-8.2	-	
Choanal atresia	9	1.1, 0.5-2.1	-		-		-	
Gastrointestinal system								
Small intestinal atresia/stenosis	30	3.6, 2.4-5.1	27	4.4, 2.9-6.4	6	3.8, 1.4-8.2	-	
Rectal and large intestinal atresia/stenosis	25	3.0, 1.9-4.4	25	4.1, 2.6-6.0	10	6.3, 3.0-11.6	-	
Esophageal atresia/ tracheoesophageal fistula	17	2.0, 1.2-3.3	17	2.8, 1.6-4.4	5	3.2, 1.0-7.4	-	
Biliary atresia	7	0.8, 0.3-1.7	-		0		-	

Gastrointestinal system								
Hypospadias*	351	82.2, 73.8-91.3	225	72.2, 63.1-82.3	41	50.2, 36.0-68.1	35	90.3, 62.9-125.5
Renal agenesis/hypoplasia	49	5.9, 4.4-7.8	29	4.7, 3.2-6.8	5	3.2, 1.0-7.4	5	6.6, 2.1-15.4
Congenital Posterior Urethral Valves*	6	1.4, 0.5-3.1	14	4.5, 2.5-7.5	-		0	
Musculoskeletal system								
Clubfoot	107	12.9, 10.5-15.5	96	15.6, 12.6-19.1	16	10.1, 5.8-16.4	5	6.6, 2.1-15.4
Craniosynostosis	91	10.9, 8.8-13.4	49	8.0, 5.9-10.5	11	6.9, 3.5-12.4	9	11.9, 5.4-22.6
Gastroschisis	23	2.8, 1.8-4.1	24	3.9, 2.5-5.8	-		-	
Limb deficiencies (reduction defects)	22	2.6, 1.7-4.0	21	3.4, 2.1-5.2	5	3.2, 1.0-7.4	-	
Diaphragmatic hernia	18	2.2, 1.3-3.4	15	2.4, 1.4-4.0	-		-	
Omphalocele	9	1.1, 0.5-2.1	10	1.6, 0.8-3.0	-		0	
Chromosomal system								
Trisomy 21 (Down syndrome)	112	13.5, 11.1-16.2	61	9.9, 7.6-12.7	31	19.6, 13.3-27.8	5	6.6, 2.1-15.4
Deletion 22 q11	17	2.0, 1.2-3.3	12	2.0, 1.0-3.4	-		-	
Trisomy 18	16	1.9, 1.1-3.1	14	2.3, 1.2-3.8	-		-	
Turner syndrome**	10	2.5, 1.2-4.5	5	1.6, 0.5-3.8	-		0	
Trisomy 13	7	0.8, 0.3-1.7	13	2.1, 1.1-3.6	-		0	

**Prevalence limited to males; **Prevalence limited to females*

- Numbers between one and four are suppressed for confidentiality protections

Appendix E: Birth Defects Case Review Model 2.0

LOUISIANA DEPARTMENT OF HEALTH
Office of Public Health

Birth Defects Case Review Model 2.0

Julie Johnston, BS; Tri Tran, MD, MPH, Rimi Mandal, MS

PARTNERS FOR FAMILY HEALTH LOUISIANA

Background

- [Specific birth defect], is one of the most prevalent congenital conditions.
- Based on the U.S. Centers for Disease Control and Prevention, in the United States, one in every [####] babies is born with [specific birth defect], and one in every [####] babies is born with [associated birth defect].
- Causes of [specific birth defect] among most infants are unknown. Some risk factors of [specific birth defect] are known, including [list of risk factors].
- Understanding risk factors and causes of birth defects plays an important role in primary prevention. Causes of birth defects may be identified through case review, a critical analysis of a patient's medical history.
- No national birth defects prevention case review model or best practice guidelines exist. Louisiana developed a birth defects case review model in 2022.

Study Question

The **study aimed to:**

- Use the case review model to evaluate timeliness of diagnosis, treatment, and referral to services for children born in [year] identified through the Louisiana Birth Defects Monitoring Network.

Methods

- **Data Sources:** Louisiana [dataset year] hospital discharge reports, vital records (birth, death, and fetal death), Medicaid data, and medical records, which included prenatal, delivery, and postnatal records, including genetic studies and autopsy reports.
- **Case review selection:** Children with [specific birth defect], born in [birth year], with or without [specific characteristics]. Birth defects cases only included children ages 0-36 months.
- **Review panelists:** One pediatrician, two epidemiologists, two maternal fetal medicine physicians, one geneticist, [specify pediatric specialists] and six key public health stakeholders and partners. [amend as necessary]
- **Analysis methods and timeline of completion:**
  - **Literature Review:** [date conducted], identified risk factors, diagnosis, treatment recommendations, and types of referral to services of [specific birth defect] from previous research
  - **Poisson regression:** [date conducted], analyzed [dataset years] linked birth defects, Medicaid, and birth data to identify timeliness of diagnosis, treatment, and referral to services associated with [specific birth defect].
  - **Model Framework and case review tool:** [date conducted], use the model framework and a case review tool to compile data, facilitate the review, and make a conclusion.
  - **Case Review:** [date conducted], completed [#] case reviews identified using case selection inclusion criteria.
  - **Pilot evaluation:** [date conducted], completed evaluation.

Results

Figure 1: Occurrence of [specify birth defect], [specify dataset years], n = [###]

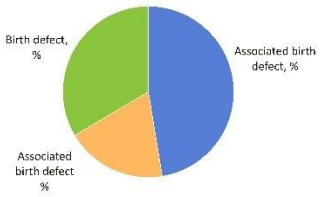


Figure 2: Prevalence of [specific birth defect] per 1,000 live births

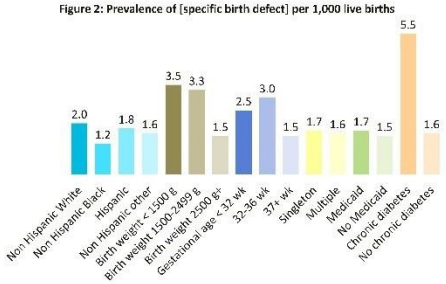


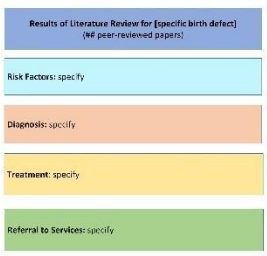
Table 1: Variables statistically significant in the final adjusted regression model

Variable	Value	Adjusted PR, CI95%	P value
Race/ethnicity	Non Hispanic White Non Hispanic Black Hispanic Non Hispanic other		
Birth weight	< 1500 g 1500-2499 g 2500+ g		
Gestational age	<32 weeks 32-36 weeks 37+ weeks		
Plurality	Singleton Multiple		
Medicaid	Yes No		
Other variable specific to birth defect	Yes No		

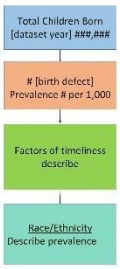
Table 2: Timeliness of birth defects diagnosis, treatment, and referral among [### of] cases selected for case review

Case #
1
2
3
4
5
6
7
8
9

Figure 3: Results of literature review and prevalence of [specific birth defect]



- **Results of literature review:** describe
- **Prevalence and Poisson regression:** describe (Table 1).
- **Case review:** # were selected for review. # live births and plurality; # race/ethnicity, # preterm births; and # deceased.
- **Findings:** describe



Conclusions

- Describe findings of timeliness of diagnosis of [specific birth defect]
- Describe findings timeliness of treatment of [specific birth defect]
- Describe findings timeliness of referral to services of [specific birth defect]

Limitations

- Lack of [describe findings]

Public Health Implications

The study provides:

- Insight into [describe findings]
- A method to standardize case review models for birth defects which can lead to prevention, implementation, and intervention efforts for the future.

Recommendations:

- Describe recommendations to increase early diagnosis, treatment, and referral to service for [specific birth defect]

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Louisiana Department of Health
628 N. 4th St., Baton Rouge, LA 70802
225-342-9500
ldh.la.gov

@ladepthealth