Louisiana

Birth Defects

Monitoring Network

2022 Annual Legislative Report

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LOUISIANA DEPARTMENT OF HEALTH

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Acknowledgements:

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." Funding for LBDMN surveillance is provided through the federal Title V Maternal and Child Health (MCH) Block Grant.

We recognize the LBDMN Data Collection Specialists and Case Review Coding Specialist who abstracted the medical records to collect these data. Their dedication and hard work made this report possible.

We are thankful for the volunteer members of the LBDMN 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 LBDMN will improve the systems of care serving Louisiana's families.

Contents

Introduction

The Louisiana Birth Defects Monitoring Network (LBDMN) within 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 children. Mandated in 2004, it was the intent of the legislature to "establish 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" (Louisiana Revised Statute (R.S.) 40:31.43 and Louisiana Administrative Code LAC Title 48, Part V, Subpart 55, Chapters 161 & 163 et al).

The following report summarizes the key findings related to the prevalence of birth defects and the performance of this essential surveillance system.

Our Mission

The mission of LBDMN is to collect, analyze, and disseminate high quality, timely, actionable data to inform policy and to improve Louisiana's maternal and child health system to eliminate preventable birth defects, mitigate disability, and connect families with resources to improve their quality of life.

What We Do

LBDMN 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. LBDMN can evaluate concerns about unexpected groups of birth defects (cluster investigation) as well as the effectiveness of preventive interventions.

Who We Serve

As a part of BFH's system of monitoring birth outcomes in the state, LBDMN 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. These data can inform policy development to improve the maternal and child health system in Louisiana.
- 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 via our website.
- Researchers from the Centers for Disease Control and Prevention (CDC), universities, and other states investigating possible causes of specific birth defects, by providing relevant data for collaborative research studies.

Approximately 1,500 children with specified birth defects are identified annually, averaging 295 per 10,000 live births. Since 2005, LBDMN has investigated potential birth defects among 44,083 children [10/2022]. LBDMN case definition criteria include all of the following:

- 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. Children who have minor defects posing no significant health or social burdens are excluded.
- 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 made before the child's 3rd 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.

Services

LBDMN is a core activity of the state's Federal Title V Maternal Child Health Block Grant programs, which identify and support children and youth with special health care needs (CYSHCN) and their families and are administered by BFH.

LBDMN services include:

- Active public health surveillance of hospital discharges of newborns until three years of age for major structural, functional, or genetic birth defects.
- Coordination with the BFH Family Resource Center to link families of children under three years of age with specified birth defects to health, social, and developmental resources.
- Partnership with national, state, and local stakeholders such as CDC, National Birth Defects Prevention Network, Louisiana Chapters of the American Academy of Pediatrics and the American College of Obstetricians and Gynecologists, March of Dimes, regional Families Helping Families, and Spina Bifida of Louisiana on public awareness campaigns focused on the prevention of future birth defects.
    - Campaigns include education to inform men and women of reproductive age of healthy prenatal lifestyle choices such as: daily consumption of 400 micrograms of folic acid daily; reducing exposures to infections and toxins; and controlling chronic conditions such diabetes and hypertension to prevent risks of associated birth defects.

Operations

Role of the LDH OPH Bureau of Family Health

This public health activity is administered by senior epidemiologists and is carried out by a statewide network of regionally-assigned Data Collection Specialists (DCS) who evaluate patient discharge information of newborns until three years of age. Records are reviewed from all birthing hospitals in Louisiana, as well as at Children's Hospital, Ochsner Medical Center, and Tulane University Medical Center in New Orleans. LBDMN maintains a longitudinal data system of all children born in Louisiana diagnosed with a structural, functional, and/or genetic birth defect. De-identified medical record data are analyzed statistically for patterns and trends over time. Families of children with certain birth defects are linked to appropriate health, social service, and developmental resources through the BFH Family Resource Center. This new referral to resources operations activity began in 2022.

Data to Action Spotlight: Referral to Resources

Timely data enables LBDMN to move data to action in the form of meaningful referrals to resources for families. When babies are born with birth defects, families need help learning about complex systems of care and how to navigate those often fragmented systems. In 2022, LBDMN partnered with BFH's Family Resource Center (FRC) to conduct a pilot of one-on-one needs assessments with families of children born in 2020 with birth defects most likely to impact developmental outcomes.

FRC staff contacted families to assess their need for health and social services including specialty medical care, early intervention services, insurance, advocacy, disability agencies, peer support, transportation, food, and housing security. In the pilot, FRC staff made contact with 54% of identified families (142/261) via phone calls, text messages, and postcards. Of these families, **93% identified at least one needed resource.** FRC staff facilitated connections to the appropriate resources for these families. This speaks to the high need for assistance with resource navigation that exists into and beyond the first two years of life.

Since evaluation of the initial referral pilot in July 2022, LBDMN and FRC staff have completed a second cohort of 2020 referrals, testing and tracking different methods to improve contact success rates. The third cohort 2021 is scheduled to be evaluated in the first quarter of 2023. Children born in 2021 and 2022 will be contacted in 2023. This timing is critical to connect children to early intervention services within the first two years of life to improve health and developmental outcomes as well as the quality of life for those impacted by birth defects.

Role of the Advisory Board

As mandated in the authorizing statute, LA R.S. 40:31.43, LBDMN is guided by an advisory board of volunteer stakeholders appointed by the secretary of LDH.

The role of LBDMN advisory board as prescribed in the law is *"to make recommendations on the implementation and continuing operation of the surveillance system."* The advisory board meets on the third Friday of January, June, and October. Notices of meetings, agendas, and minutes are posted on the Louisiana Boards & Commissions website.

Data to Action Spotlight: Birth Defects Prevention Case Review Model

The focus of 2021-2022 board activities was to move birth defects data to action through data analysis. LBDMN program staff worked through the advisory board to develop a birth defects prevention ***case review model***. The purpose of structured on-going case review is to move LBDMN data to action by identifying preventable birth defects to make evidence-based recommendations for systems level changes to improve primary prevention and intervention efforts in Louisiana's maternal health system.

The ***case review model*** consists of three frameworks to support case review including the:

- *Evidence Base* as presented in the literature review of risk factors and best practices.
- *Data Framework* which included a State of the State of maternal health for the identified birth year using a variety of state datasets (Vital Records, Louisiana Pregnancy Risk Assessment Monitoring System (Louisiana PRAMS), LA Bias or Racism & Social Determinants of Health (LABoRS) tool, Environmental Public Health Tracking (EPHT) for data mapping & visualization, and LBDMN birth defects data analysis. Additionally Maternal Fetal Medicine subject matter experts provided insight into actual maternal fetal healthcare practice and risk factor management.

- *Case Review Approach* which included case selection criteria and tools such as the *Case Summary Form and Considerations for Birth Defects Prevention Case Review* – a one pager of risk factors, general prevention strategies, and *Considerations for Recommendations & Strategies* – a one pager of recommendations criteria (evidence-based and partner networks to facilitate implementation), examples of types of strategies for primary prevention & systems improvements, and definitions of public health prevention levels.

In October 2022, the ***case review model*** was tested using LBDMN 2017-2019 orofacial defects data. After suggesting minor adjustments to the case summary form, the advisory board adopted the following schedule to apply the prevention case review model to fourteen birth defects designated as *Core* by the National Birth Defects Prevention Network:

- 2023: Critical congenital heart defects
- 2024: Neural tube defects and Trisomy 21
- 2025: Abdominal wall and limb reduction defects

Please refer to Appendix B: *NBDPN Birth Defects Codes and Descriptions* for respective *Core* and *Recommended* designations. Upon completion of this cycle of case review, the advisory board will bring recommendations of the identified strategies to aid in advancing prevention efforts and interventions to increase health and developmental outcomes for children identified with birth defects in Louisiana.

Methodology

LBDMN contacts health providers to identify cases and collect data. Potential cases of interest are identified from hospital discharge indices, Medicaid, Louisiana Hospital Inpatient Discharge Data (LAHIDD), as well as birth, death, and fetal death record data from the Louisiana Vital Records Electronic Event Registration System (LEERS). Medical and vital statistic records are reviewed to collect and validate data among children diagnosed from birth up to their 3rd birthday.

Data are reviewed for completeness and coding accuracy by a Registered Nurse Case Review Clinical Coding Specialist and/or the LBDMN Program Manager before data are accepted into the Registry and are available for reporting. Data are stored and managed in a database integrated with LEERS birth and death certificates as well as Early Hearing Detection and Intervention (LA-EHDI) data.

Not all defects are evident at birth, therefore, LBDMN includes children diagnosed before their 3rd birthday allowing adequate time to capture all birth defects within our case definition. Additional time is allowed for records to be processed by hospitals, reported to LBDMN, and abstracted to more accurately 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 are converted into the CDC clinical coding system, based on the British Pediatric Association and Classification of Diseases and the ICD-10CM. Prevalence rate is calculated as the number of children with birth defects per 10,000 total live births. There is an exception for hypospadias and Turner Syndrome, which is limited to per 10,000 males and 10,000 females respectively. Data are reported in three year increments to have an adequate number of cases to be representative at the state level when stratified by race/ethnicity and type of major birth defects.

2022 Performance Assessment and Improvements

LBDMN follows national standards and guidelines for birth defects surveillance. CDC monitors national birth defects surveillance through a branch called the National Center for Birth Defects and Developmental Disabilities (NCBDDD). NCBDDD coordinates standards for state birth defects programs through the National Birth Defects Prevention Network (NBDPN). The NBDPN Standards Workgroup produces Data Quality Standards and Assessment Tools for population-based birth defects surveillance systems.

Performance standards are used to improve and standardize operations, outcomes, and surveillance functions across state programs, thereby making data comparable at the state, multi-state, and national levels. Eleven data quality measures around completeness, timeliness, and accuracy are associated with three performance levels: (1) Rudimentary, (2) Essential, and (3) Optimal.

Using NBDPN data quality assessments, Louisiana ranks among the nation's top active surveillance programs in completeness, accuracy, and overall quality. **From 2019 to 2022 assessments, LBDMN increased from Level 1 to Levels 2 and 3 in both data quality measures of timeliness. This means 95-99% of data from NBDPN's lists of 19 *Core* and 33 *Recommended* birth defects are identified, collected, reviewed, and available for reporting within two years of birth.** See Appendix B for NBDPN Birth Defects Codes and Descriptions. Per LA R.S. 40:31.42, LBDMN continues to identify and report qualifying birth defects diagnosed before three years of age.

This increase in timeliness is a direct result of efficiencies realized from performance improvement strategies adopted in 2020-2021. Strategies included code list revisions, automated case-finding, database updates, and expanded remote access to medical records for data collection thereby eliminating travel and enabling staffing reassignments for equitable caseloads.

Findings

Of 176,643 children born between 2017 and 2019, 4,657 children were diagnosed with at least one birth defect, yielding an overall prevalence of 263.6 per 10,000 live births or 2.63%. Among children with birth defects, cardiovascular system defects (63.3%) were the most common followed (in order of occurrence) by defects of the genitourinary, musculoskeletal, chromosomal, orofacial, central nervous, gastrointestinal, eye, and ear/face/neck systems. Other birth defects contributed just under 2% (Table 1).

Table1: Type of birth defects by organ and chromosome system among children with birth defects, 2017-2019 (n = 4,657)

Organ and Chromosome System	Number	Percent*
Cardiovascular	2,947	63.3
Genitourinary	841	18.1
Musculoskeletal	585	12.6
Chromosomal	492	10.6
Oro-facial	322	6.9
Central nervous	220	4.7
Gastrointestinal	184	4.0
Eye	60	1.3

Ear, face, and neck	40	0.9
Other	80	1.7

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

The six most common specific birth defects overall, regardless of the organ or chromosomal system to which it belongs, with a prevalence greater than 10 per 10,000 live births among children born in 2017-2019 included:

- Atrial septal defect (114.9)
- Ventricular septal defect (51.9)
- Hypospadias (72.1)
- Clubfoot (14.0)
- Down syndrome (13.1)
- Craniosynostosis (9.9).

Stratified by organ and chromosomal system, the most common birth defects were:

- For cardiovascular: atrial septal defects and ventricular septal defects
- For genitourinary: hypospadias
- For central nervous: spina bifida
- For eyes: congenital cataract and anophthalmia/microphthalmia
- For ear, face, and neck: anotia/microtia
- For orofacial: cleft palate without cleft lip
- For gastrointestinal: rectal, large, and small intestinal atresia or stenosis
- For musculoskeletal: clubfoot and craniosynostosis
- For chromosomal: Down syndrome

See Table 2 below for more information.

Table 2: Occurrence of specific birth defects by organ and chromosomal system, 2017-2019 (N = 176,643)

System	Birth defects	Number	%	Prev.	95%CI
	Total	**4,657**		**263.6**	**256.1, 271.3**
Central nervous	Spina bifida without anencephalus	50	22.7	2.8	2.1, 3.7
(n = 220)	Anencephalus	29	13.2	1.6	1.1, 2.4
	Holoprosencephaly	27	12.3	1.5	1.0, 2.2
	Encephalocele	21	9.5	1.2	0.7, 1.8
Eyes	Congenital cataract	36	60.0	2.0	1.4, 2.8
(n = 60)	Anophthalmia/microphthalmia	21	35.0	1.2	0.7, 1.8
Ear, face, neck	Anotia/microtia	34	85.0	1.9	1.3, 2.7
(n = 40)					
Cardiovascular	Atrial septal defect	2,030	68.9	114.9	110.0, 120.0
(n = 2,947)	Ventricular septal defect	917	31.1	51.9	48.6, 55.4
	Pulmonary valve atresia and stenosis	173	5.9	9.8	8.4, 11.4
	Atrioventricular septal defect	139	4.7	7.9	6.6, 9.3
	Coarctation of the aorta	91	3.1	5.2	4.1, 6.3
	Tetralogy of Fallot	82	2.8	4.6	3.7, 5.8
	Transposition of the great arteries	49	1.7	2.8	2.1, 3.7
	Dextro-transposition of great arteries	43	1.5	2.4	1.8, 3.3
	Double outlet right ventricle	43	1.5	2.4	1.8, 3.3
	Hypoplastic left heart syndrome	40	1.4	2.3	1.6, 3.1
	Aortic valve stenosis	20	0.7	1.1	0.7, 1.7
	Total anomalous pulmonary venous connection	19	0.6	1.1	0.6, 1.7
	Tricuspid valve atresia and stenosis	14	0.5	0.8	0.4, 1.3
	Common truncus	12	0.4	0.7	0.4, 1.2
	Tricuspid valve atresia	12	0.4	0.7	0.4, 1.2
	Pulmonary valve atresia	10	0.3	0.6	0.3, 1.0
	Interrupted aortic arch	10	0.3	0.6	0.3, 1.0
	Ebstein anomaly	9	0.3	0.5	0.2, 1.0
	Single Ventricle	9	0.3	0.5	0.2, 1.0
Oro-facial	Cleft palate without cleft lip	146	45.3	8.3	7.0, 9.7
(n = 322)	Cleft lip with cleft palate	115	35.7	6.5	5.4, 7.8
	Cleft lip without cleft palate	52	16.1	2.9	2.2, 3.9
	Choanal atresia	17	5.3	1.0	0.6, 1.5
Gastrointestinal	Small intestinal atresia/stenosis	81	44.0	4.6	3.6, 5.7
(n = 184)	Rectal and large intestinal atresia/stenosis	69	37.5	3.9	3.0, 4.9
	Esophageal atresia/tracheoesophageal fistula	28	15.2	1.6	1.1, 2.3
	Biliary atresia	13	7.1	0.7	0.4, 1.3
Genitourinary	Hypospadias	651	77.4	72.1	66.7, 77.9
(n = 841)	Renal agenesis/hypoplasia	94	11.2	5.3	4.3, 6.5
	Congenital posterior urethral valves	28	3.3	3.1	2.1, 4.5
Musculoskeletal	Clubfoot	247	42.2	14.0	12.3, 15.8
(n = 585)	Craniosynostosis	174	29.7	9.9	8.4, 11.4
	Gastroschisis	54	9.2	3.1	2.3, 4.0
	Diaphragmatic hernia	47	8.0	2.7	2.0, 3.5
	Limb deficiencies	40	6.8	2.3	1.6, 3.1
	Omphalocele	37	6.3	2.1	1.5, 2.9
Chromosomal	Trisomy 21 (Down syndrome)	231	47.0	13.1	11.4, 14.9
(n = 492)	Trisomy 18	33	6.7	1.9	1.3, 2.6
	Deletion 22 q11	28	5.7	1.6	1.1, 2.3
	Turner syndrome	14	2.8	1.6	0.9, 2.7
	Trisomy 13	11	2.2	0.6	0.3, 1.1

**Prevalence limited to male (90,254); **Prevalence limited to female (86,386)*

Stratified by race and ethnicity, the total prevalence of birth defects was a bit higher in non-Hispanic white (NHW) (270.2) than in non-Hispanic Black (NHB) (261.4). The five most common birth defects with a prevalence equal or greater than 10 per 10,000 live births in both groups included atrial septal defect (NHW: 108.3 vs. NHB: 130.6), hypospadias (NHW: 84.0 vs. NHB: 65.3), ventricular septal defect (NHW: 53.3 vs. NHB: 44.3), clubfoot (NHW: 14.2 vs. NHB: 14.8), and Down syndrome (NHW: 12.0 vs. NHB: 11.1). In addition, cleft palate without cleft lip (9.9) and craniosynostosis (12.0) were seen with a prevalence greater than 10 per 10,000 live births in NHW (Table 3).

Table 3: Occurrence of specific birth defects by organ and chromosomal system and race and ethnicity, 2017-2019

Birth defects	Non-Hispanic White		Non-Hispanic Black		Hispanic		Non-Hispanic Other	
	n	Prevalence, 95%CI	n	Prevalence, 95%CI	n	Prevalence, 95%CI	n	Prevalence, 95%CI
Total	**2409**	**270.2, 259.6-281.3**	**1713**	**261.4, 249.1-274.1**	**366**	**257.5, 231.8-285.2**	**164**	**217.3, 185.3-253.2**
Central nervous system								
Spina bifida without anencephalus	27	3.0, 2.0-4.4	13	2.0, 1.1-3.4	9	6.3, 2.9-12.0	-	
Anencephalus	15	1.7, 0.9-2.8	9	1.4, 0.6-2.6	-		-	
Holoprosencephaly	9	1.0, 0.5-1.9	12	1.8, 0.9-3.2	-		-	
Encephalocele	7	0.8, 0.3-1.6	12	1.8, 0.9-3.2	-		-	
Eyes								
Congenital cataract	17	1.9, 1.1-3.1	14	2.1, 1.2-3.6	-		-	
Anophthalmia/microphthalmia	8	0.9, 0.4-1.8	8	1.2, 0.5-2.4	-		-	
Ear, face, neck								
Anotia/microtia	12	1.3, 0.7-2.4	10	1.5, 0.7-2.8	11	7.7, 3.9-13.8	-	
Cardiovascular system								
Atrial septal defect	965	108.3, 101.5-115.3	856	130.6, 122.0-139.7	137	96.4, 80.9-113.9	71	94.1, 73.5-118.7
Ventricular septal defect	475	53.3, 48.6-58.3	290	44.3, 39.3-49.6	117	82.3, 68.1-98.6	35	46.4, 32.3-64.5
Pulmonary valve atresia and stenosis	79	8.9, 7.0-11.0	76	11.6, 9.1-14.5	15	10.6, 5.9-17.4	-	
Atrioventricular septal defect	68	7.6, 5.9-9.7	54	8.2, 6.2-10.8	12	8.4, 4.4-14.7	5	6.6, 2.2-15.5
Coarctation of the aorta	52	5.8, 4.4-7.7	32	4.9, 3.3-6.9	5	3.5, 1.1-8.2	-	
Tetralogy of Fallot	49	5.5, 4.1-7.3	24	3.7, 2.3-5.4	7	4.9, 2.0-10.1	-	
Hypoplastic left heart syndrome	28	3.1, 2.1-4.5	10	1.5, 0.7-2.8	-		-	
Transposition of the great arteries	26	2.9, 1.9-4.3	14	2.1, 1.2-3.6	6	4.2, 1.5-9.2	-	
Dextro-transposition of great arteries	24	2.7, 1.7-4.0	11	1.7, 0.8-3.0	5	3.5, 1.1-8.2	-	
Double outlet right ventricle	20	2.2, 1.4-3.5	18	2.7, 1.6-4.3	5	3.5, 1.1-8.2	0	
Aortic valve stenosis	13	1.5, 0.8-2.5	-		-		-	
Total anomalous pulmonary venous connection	10	1.1, 0.5-2.1	7	1.1, 0.4-2.2	-		0	
Common truncus	9	1.0, 0.5-1.9	-		-		0	
Single Ventricle	6	0.7, 0.2-1.5	-		0		0	
Tricuspid valve atresia and stenosis	5	0.6, 0.2-1.3	5	0.8, 0.2-1.8	-		0	
Tricuspid valve atresia	5	0.6, 0.2-1.3	-		-		0	
Ebstein anomaly	5	0.6, 0.2-1.3	-		-		-	
Pulmonary valve atresia	-		5	0.8, 0.2-1.8	-		0	
Interrupted aortic arch	-		6	0.9, 0.3-2.0	0		0	

Table 3: Occurrence of specific birth defects by organ and chromosomal system and race and ethnicity, 2017-2019 (continued)

Birth defects	Non-Hispanic White		Non-Hispanic Black		Hispanic		Non-Hispanic Other	
	n	Prevalence, 95%CI	n	Prevalence, 95%CI	n	Prevalence, 95%CI	n	Prevalence, 95%CI
Oro-facial system								
Cleft palate without cleft lip	88	9.9, 7.9-12.2	40	6.1, 4.4-8.3	10	7.0, 3.4-12.9	8	10.6, 4.6-20.9
Cleft lip with cleft palate	68	7.6, 5.9-9.7	31	4.7, 3.2-6.7	12	8.4, 4.4-14.7	-	
Cleft lip without cleft palate	30	3.4, 2.3-4.8	11	1.7, 0.8-3.0	7	4.9, 2.0-10.1	-	
Choanal atresia	13	1.5, 0.8-2.5	-		-		0	
Gastrointestinal system								
Small intestinal atresia/stenosis	42	4.7, 3.4-6.4	27	4.1, 2.7-6.0	7	4.9, 2.0-10.1	5	6.6, 2.2-15.5
Rectal and large intestinal atresia/stenosis	32	3.6, 2.5-5.1	29	4.4, 3.0-6.4	7	4.9, 2.0-10.1	-	
Esophageal atresia/tracheoesophageal fistula	14	1.6, 0.9-2.6	11	1.7, 0.8-3.0	-		-	
Biliary atresia	5	0.6, 0.2-1.3	8	1.2, 0.5-2.4	0		0	
Genitourinary system								
Hypospadias	385	84.0, 75.9-92.9	217	65.3, 56.9-74.6	26	36.0, 23.5-52.7	20	51.7, 31.6-79.8
Renal agenesis/hypoplasia	54	6.1, 4.6-7.9	25	3.8, 2.5-5.6	11	7.7, 3.9-13.8	-	
Congenital Posterior Urethral Valves	13	2.8, 1.5-4.9	11	3.3, 1.7-5.9	-		-	
Musculoskeletal system								
Clubfoot	127	14.2, 11.9-17.0	97	14.8, 12.0-18.1	16	11.3, 6.4-18.3	7	9.3, 3.7-19.1
Craniosynostosis	107	12.0, 9.8-14.5	43	6.6, 4.7-8.8	17	12.0, 7.0-19.1	6	7.9, 2.9-17.3
Gastroschisis	31	3.5, 2.4-4.9	18	2.7, 1.6-4.3	-		-	
Diaphragmatic hernia	27	3.0, 2.0-4.4	15	2.3, 1.3-3.8	-		-	
Omphalocele	23	2.6, 1.6-3.9	11	1.7, 0.8-3.0	-		0	
Limb deficiencies (reduction defects)	20	2.2, 1.4-3.5	17	2.6, 1.5-4.2	-		0	
Chromosomal system								
Trisomy 21 (Down syndrome)	107	12.0, 9.8-14.5	73	11.1, 8.7-14.0	33	23.2, 16.0-32.6	18	23.8, 14.1-37.7
Deletion 22 q11	18	2.0, 1.2-3.2	9	1.4, 0.6-2.6	-		0	
Trisomy 18	15	1.7, 0.9-2.8	12	1.8, 0.9-3.2	6	4.2, 1.5-9.2	0	
Trisomy 13	5	0.6, 0.2-1.3	-		-		0	
Turner syndrome	5	1.2, 0.4-2.7	9	2.8, 1.3-5.3	0		0	

- Case number not shown between one and four

In addition to providing annual reports to the Louisiana Legislature, LBDMN data are included in biennial reports produced by the National Birth Defects Prevention Network and special reports such as the manuscript *Short Interpregnancy Interval and Prevalence of Birth Defects: A multi-state study* published in the January 2022 special issue of *Birth Defects Research.*

Summary

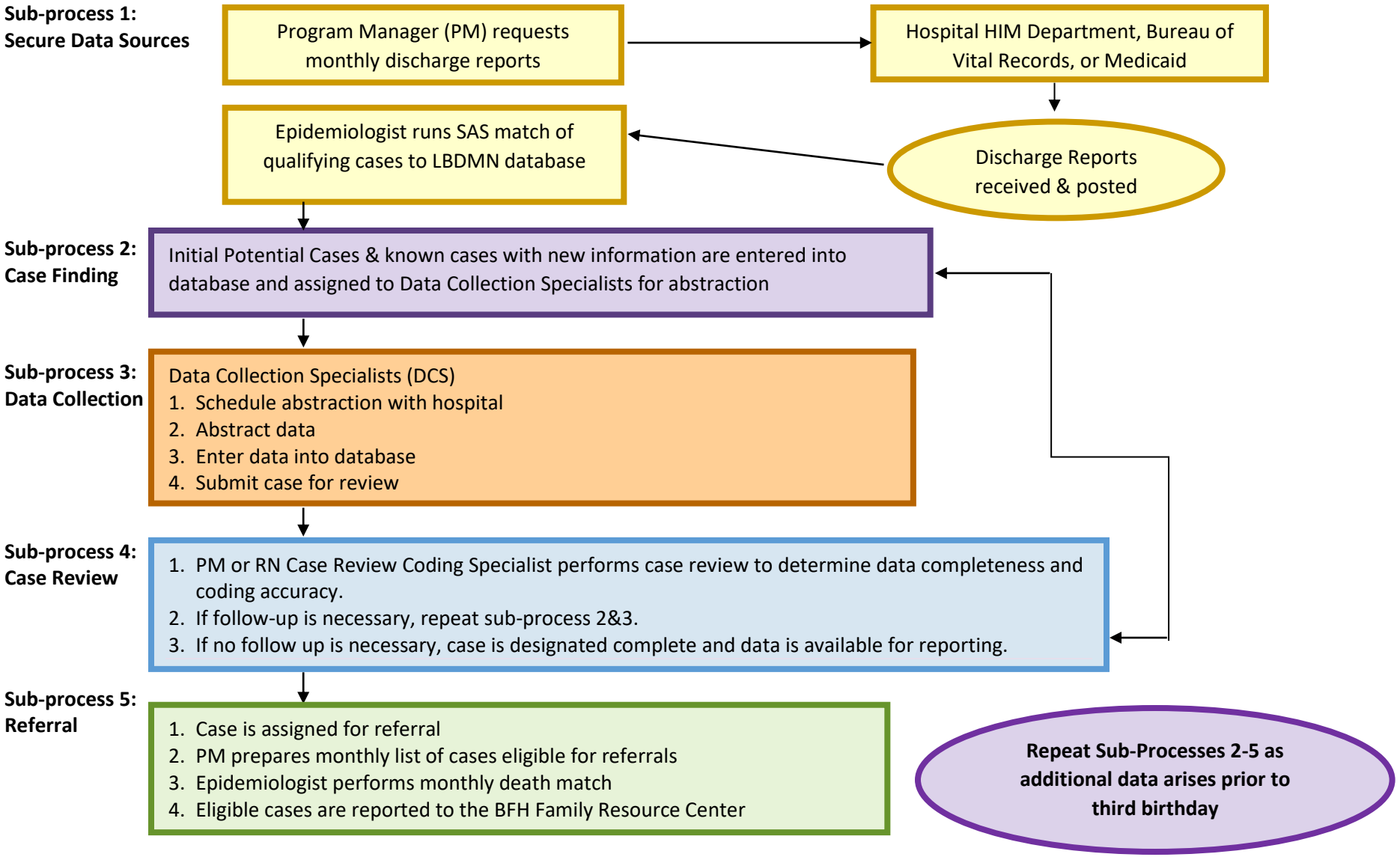
Of 176,643 children born between 2017 and 2019, 4,657 children were diagnosed with at least one birth defect, yielding an overall prevalence of 263.6 per 10,000 live births or 2.63%. According to the CDC the US average is about 3% of all babies born each year. Among Louisiana children with birth defects born in 2017-2019, cardiovascular system defects (about 63%) were the most common.

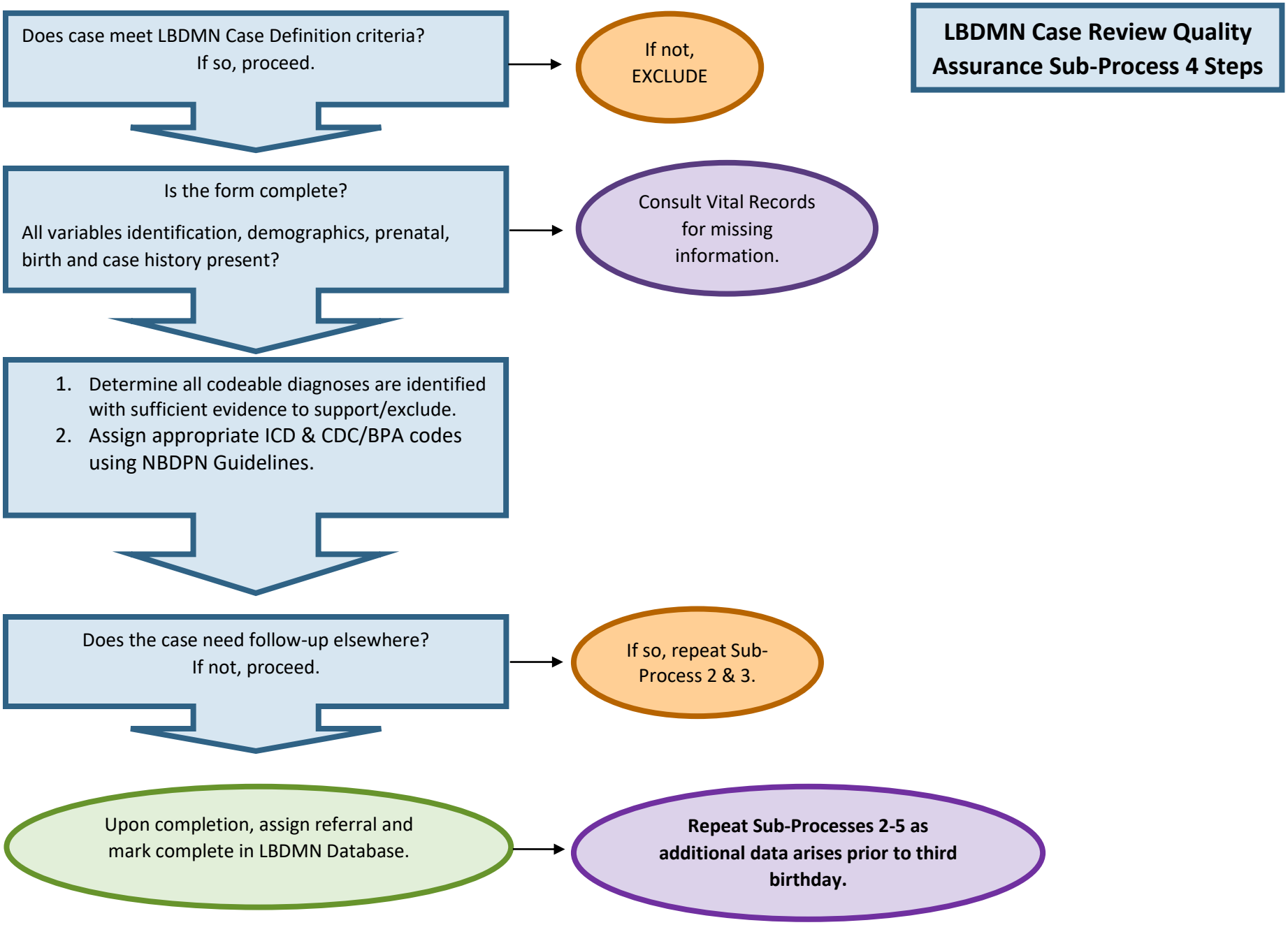
As the established statewide mechanism for tracking and monitoring birth defects in Louisiana, LBDMN incorporates evidence-based public health surveillance best practices and continues to seek opportunities for quality improvement. Increased efficiencies in data collection and reporting approaches moved our data to action in 2022 by informing timely referrals to services for families and development of the birth defects prevention case review model. Ultimately, this surveillance system will generate actionable recommendations for systems level changes and positively impact the occurrence of preventable birth defects in the future.

LBDMN data are available at PartnersForFamilyHealth.org, the National Birth Defects Prevention Network, from CDC Data & Statistics on Birth Defects and on the Louisiana's Environmental Public Health Tracking Network health data portal to enable analysis, visualization, and reporting. These data can be accessed by environmental and public health practitioners, healthcare providers, community members, policy makers, and others to make data-driven decisions that affect the health of Louisiana citizens.

Appendix

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




LBDMN Case Review Quality Assurance Sub-Process 4 Steps
Does case meet LBDMN Case Definition criteria? If so, proceed.
If not, EXCLUDE
Is the form complete?
All variables identification, demographics, prenatal, birth and case history present?
Consult Vital Records for missing information.
1. Determine all codeable diagnoses are identified with sufficient evidence to support/exclude.
2. Assign appropriate ICD & CDC/BPA codes using NBDPN Guidelines.
Does the case need follow-up elsewhere? If not, proceed.
If so, repeat Sub-Process 2 & 3.
Upon completion, assign referral and mark complete in LBDMN Database.
Repeat Sub-Processes 2-5 as additional data arises prior to third birthday.

Appendix B: Birth Defects Codes and Descriptions

ICD-10 CM CODES BY STANDARD LEVEL	DESCRIPTION	2018-2020 CASE DEFINITION	CDC 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 745.510 only
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 (exclude Imperforate anus)
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% mortality rate
Q91.4 - Q91.7	Trisomy 13	CASE FINDING & CODING	758.100-758.190	91% mortality rate
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

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