TN Department of Health

TENNESSEE BIRTH DEFECTS DATA REPORT 2016-2020

Tennessee
Birth Defects
Surveillance System

Division of
Family Health and
Wellness

December 2023

BIRTH DEFECTS SURVEILLANCE IN TENNESSEE

Birth defects are CRITICAL, COMMON, and COSTLY.

Birth defects (also known as congenital anomalies) are changes that can affect almost any part of the body and alter how the body looks and/or functions. Birth defects are identified ***before*** birth, ***at*** birth, or ***after*** birth. Not all birth defects are the same; some are very mild while others are severe. One's life expectancy may vary depending on the severity of the birth defect and affected body part(s).

Early recognition of and response to birth defects often promotes early intervention and treatment, which may decrease further complications and disability. ***A birth defects surveillance and information system is essential for the development of programs and policies that can reduce birth defects and infant mortality. These programs also serve a critical role in connecting families with support services in each community.***

The Tennessee Department of Health's Tennessee Birth Defects Surveillance System (TNBDSS), as outlined in Tennessee Code Annotated (TCA) §68-5-506, is a statewide surveillance program that:

- Identifies children with birth defects
- Provides information on the incidence, prevalence, and trends of birth defects
- Informs partners and the public on birth defects and risk factors
- Provides information to determine whether environmental hazards are associated with birth defects
- Provides guidance on prevention efforts
- Provides families of children with birth defects information on available supportive services in Tennessee and when appropriate provides service referrals

Enhanced Surveillance

TNBDSS is currently in **year 3** of a **5-year CDC cooperative agreement** to **enhance birth defects surveillance** in Tennessee. The activities outlined in this cooperative agreement help the program to not only meet those TCA activities, but also surpass those activities. In addition to improving surveillance capacity and data quality, TNBDSS is working to develop and implement prevention strategies centered around preconception health and improve connections for qualifying families to supportive services.

About this Report

This annual report provides details on the prevalence of 46 major birth defects for Tennessee infants born in the years 2016-2020. The primary data sources for this report are the Hospital Discharge Data System (HDDS) and the Birth, Death, and Fetal Death Statistical Data Systems. The methodology of data collection used for this report results in a time lag for analysis, since finalization of the HDDS files occurs one year after the birth year.

Unless otherwise noted, all data represented in this report is from 2016-2020.

Although clinically very different, atrial septal defect (a known congenital heart defect) and patent foramen ovale (considered a normal variant) shared a single diagnostic code (ICD-10 Q21.1). This shared coding leads to an overestimation of totals for congenital heart defects (CHD). Therefore, the general analyses in this report ***do not*** include the diagnosis of atrial septal defect (ASD) (except in Appendix I) and should not be compared with reports from previous years.

NOTE: All clinical diagnoses in this report are defined in Appendix II.

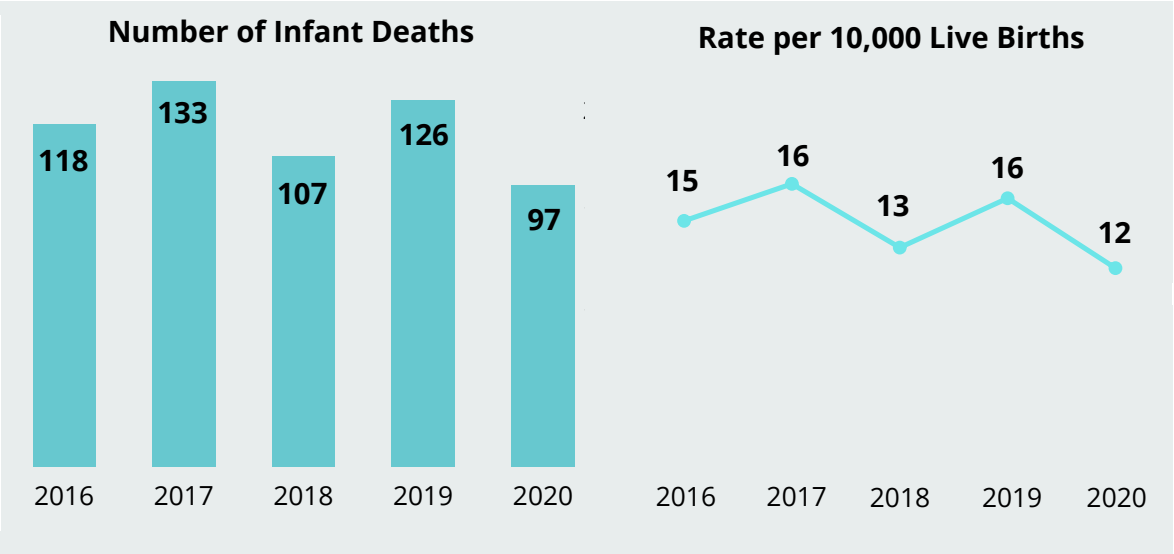
Birth Defects Are CRITICAL

Birth defects accounted for approximately **21% of infant deaths** in the state of Tennessee.

This made birth defects the ***leading cause of infant mortality*** in our state between 2016-2020.

From 2016-2020, an average of **116 Tennessee infants** died due to birth defects **each year**. There was some fluctuation in the annual number and rate of death per 10,000 live births, but no sign of a strong increasing or decreasing trend over this period.

Infant Deaths Due to Birth Defects



Infant mortality due to birth defects does not impact all race/ethnic groups equally in Tennessee. **Hispanic** infants had the highest rate of death due to birth defects over the 2016-2020 time period.

Non-English speaking and **immigrant families** encounter additional barriers when accessing healthcare, which may influence overall mortality rates.

Infant Deaths per 10,000 Live Births

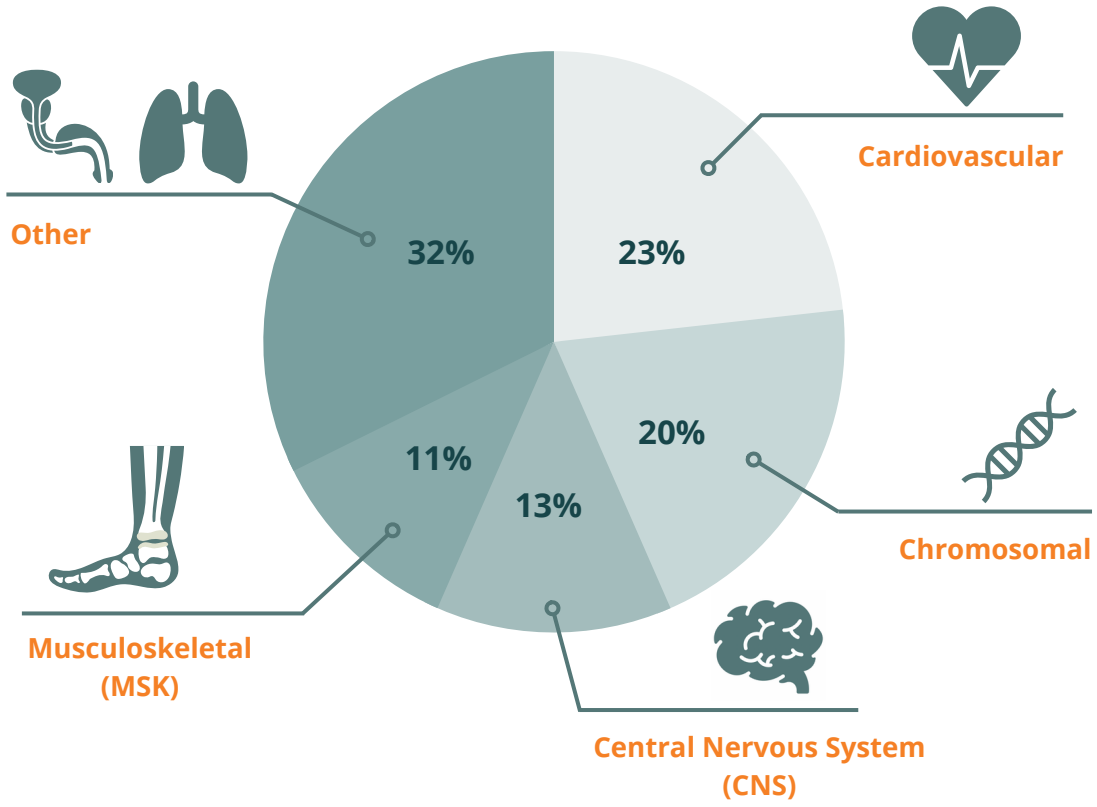
Birth defects cause about 1 in 5 infant deaths in Tennessee, and can lead to lifelong disability.

Birth Defects Are CRITICAL (continued)

About one in four (23%) infant deaths due to birth defects had **cardiovascular** conditions, and **one in five (20%)** had **chromosomal** conditions. Among infant deaths due to chromosomal birth defects, Trisomy 18 was by far the most common condition.

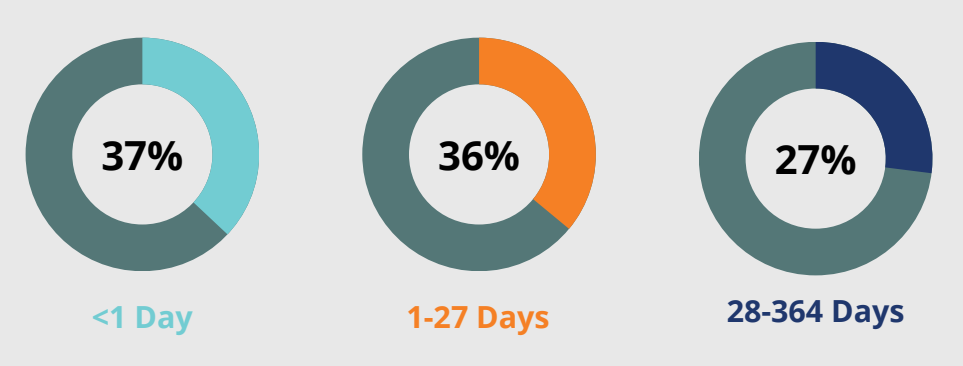
The 'Other' category shown below includes infant deaths due to genitourinary (GU), respiratory, and other types of birth defects.

Birth Defect Deaths by Type of Defect (Organ System)



Birth Defects Are CRITICAL (continued)

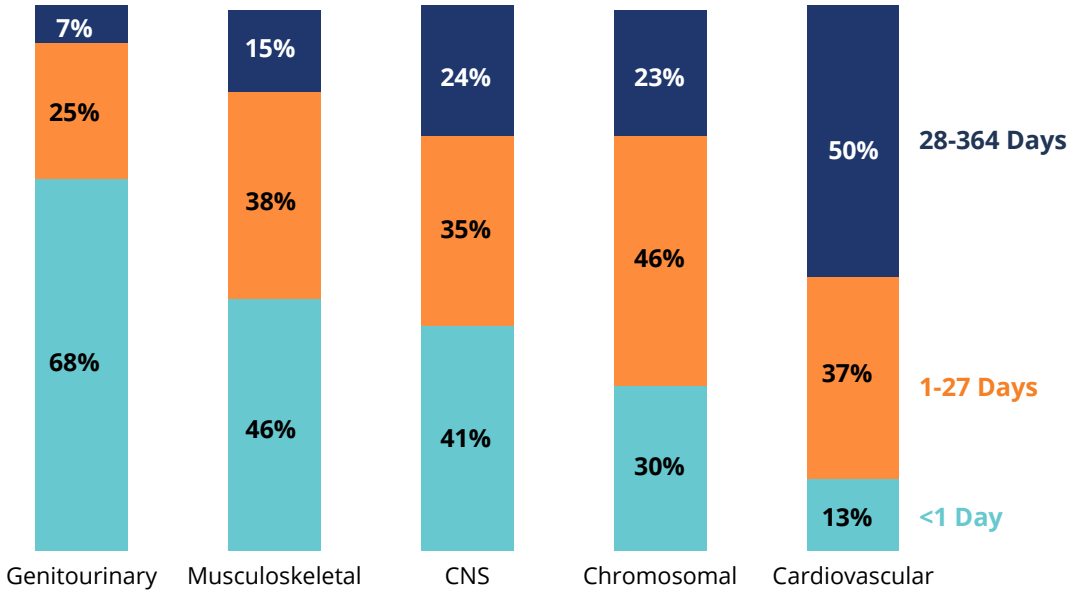
Infant Age at Death Due to All Birth Defects



Age at death differed sharply by type of defect, classified by organ system. Over two-thirds of infant deaths due to **genitourinary** defects occurred the same day as birth (when documented as the underlying cause of death on the death certificate). Infants with a genitourinary defect comprised a small number of cases, but often had additional comorbidities or defects that may have contributed to the early mortality numbers.

In contrast, most infants who died due to **cardiovascular** defects died later in the post-neonatal period. Advances in mandatory screening and medical care have led to infants born with critical congenital heart defects (CCHDs) living beyond the first few weeks of life.

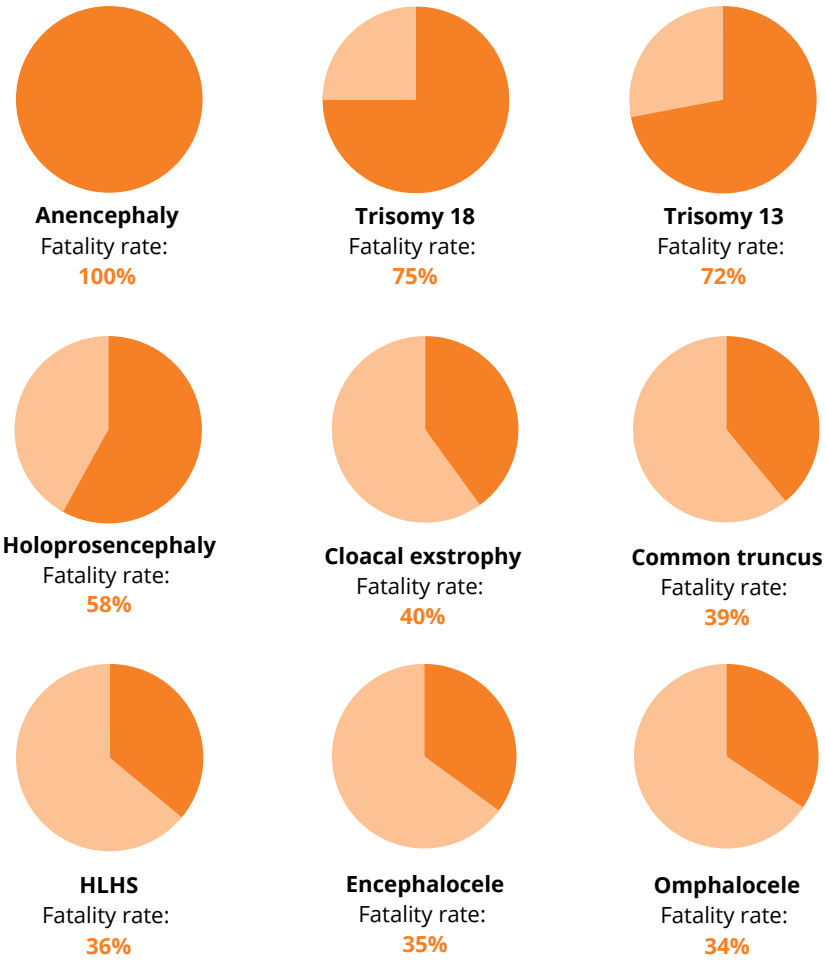
Percent of Birth Defect Deaths by Age at Death and Type (Organ System)



Birth Defects Are CRITICAL (continued)

The figures below demonstrate the tracked birth defects with the highest rates of mortality, either **during pregnancy** or **up to one year of age**.

Fetal/Infant Mortality Rate by Birth Defect



Common truncus and **hypoplastic left heart syndrome (HLHS)** were two of the cardiovascular defects with the highest mortality rates.

The children who survived infancy often passed away later in childhood. Many that survive have lifelong disability and comorbidities. Furthermore, birth defects can have a significant **emotional, medical,** and **financial impact** on affected children and their families.

Birth Defects Are CRITICAL (continued)

The conditions included in this report vary widely in terms of severity. One example of this is the cardiovascular defects monitored.

Cardiovascular conditions can vary greatly in severity - from **congenital heart defects (CHDs)** that can often be managed with medications and may self-resolve, to **critical congenital heart defects (CCHDs)** that can cause serious, life-threatening symptoms. Often, these symptoms require intervention within the first days or first year of life.

As noted on the first page of this report, a major change to the data analysis introduced this year was the exclusion of cases with atrial septal defect (ASD) as their only diagnosis. Historically, ASD has been an extremely common diagnosis, accounting for approximately 1700 reported cases each year. Prior to October 2022, ASD and patent foramen ovale (PFO) were grouped within a *single* diagnosis code. Clinically speaking, these are two *different* diagnoses. However, because ASD did not have a unique diagnosis code during the time frame of this report, it was impossible to distinguish true ASD cases from infants with PFOs.

Also important to note, most cases, identified by the shared ICD-10 code, as having ASD (68%) had no other birth defect. These cases, now excluded from our analysis, noted extremely different **demographic and geographic findings,** compared to the other birth defect cases. They were much more likely to be infants born to non-Hispanic Black women (30% versus 18% for the other cases) and to be born in Shelby County (25% versus 14%). *Therefore, their exclusion has had a significant impact on the trends reflected throughout this report.* Direct comparisons should not be made to previous reports. This change was necessary to improve the accuracy and usefulness of the information by removing cases likely to be "false positives," or infants with no actual birth defect.

NOTE: unique data for these diagnoses will be available after October 2022, when they will be represented by their own ICD-10 code.

Birth Defects Are CRITICAL (continued)

As previously stated, there are clinical differences between congenital heart defects (CHDs) and critical congenital heart defects (CCHDs).

The table below summarizes high-level statistics for one congenital heart defect, ventricular septal defect (**VSD**), and three ***critical*** congenital heart defects (CCHDs): hypoplastic left heart syndrome (**HLHS**), interrupted aortic arch (**IAA**), and total anomalous pulmonary venous connection (**TAPVC**).

While VSD was much more common than the CCHDs shown, the associated infant mortality rate and median hospital charge were far lower for VSD. Some conditions like HLHS had a similar prevalence rate across racial/ethnic groups, but others like TAPVC varied significantly.

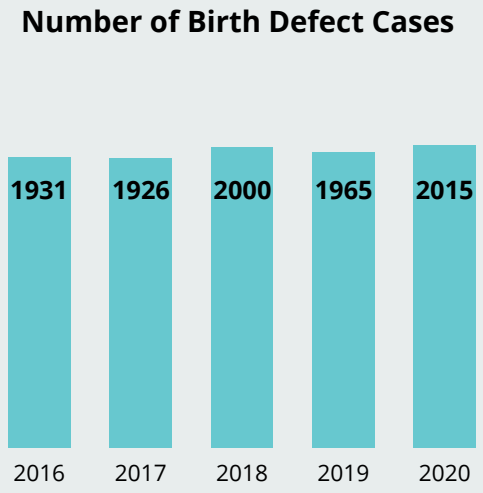
	VSD	HLHS	IAA	TAPVC
Number of cases per year in TN	418	30	12	9
Mortality rate Percent of cases that died during the first year of life	7%	36%	24%	20%
Hospitalization Charge Median total charge for an inpatient hospitalization of an infant with defect as primary diagnosis	$181,365	$395,326	$675,273	$493,046
Prevalence by Race/Ethnicity per 10,000 live births				
NH White	50.6	3.9	1.3	1.0
NH Black	55.1	3.4	2.2	0.5
Hispanic	60.9	3.7	2.0	3.5

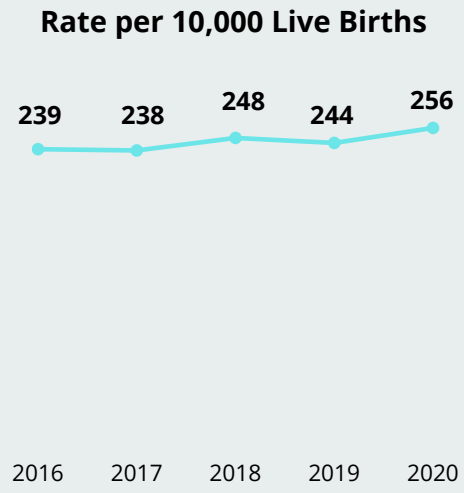
Birth Defects Are COMMON

Every **4.5 minutes** a baby is born with a birth defect in the United States. This translates to approximately **120,000 babies** each year.[1]

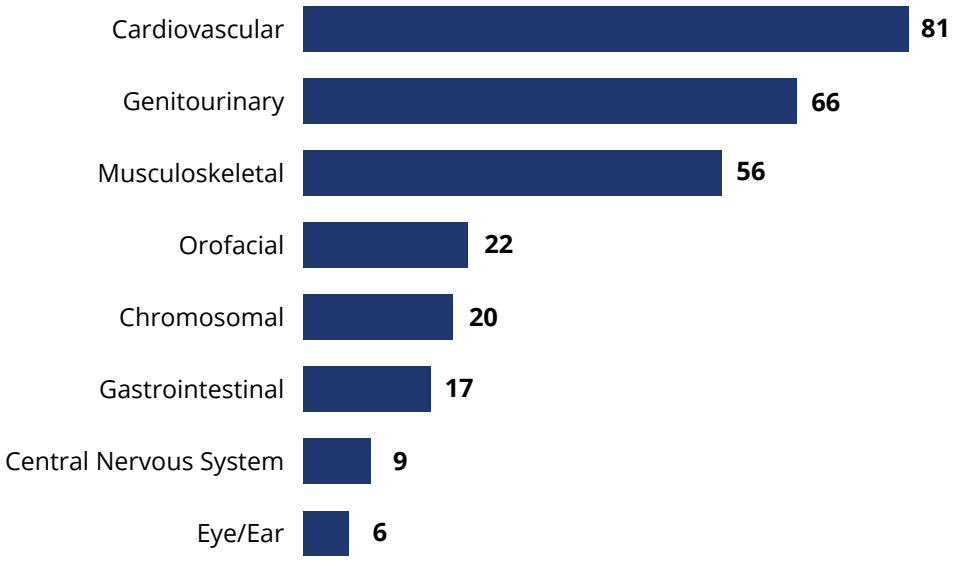
From 2016 to 2020, approximately 2,000 Tennessee infants per year were diagnosed with a birth defect. Cardiovascular defects were most common, affecting 81 infants per 10,000 live births. No significant trend was detected during this time period.

Total Cases of Infants with Birth Defects





Rate (per 10,000 Live Births) of Defects by Organ System



In Tennessee, 1 in every 41 babies is born with a birth defect.

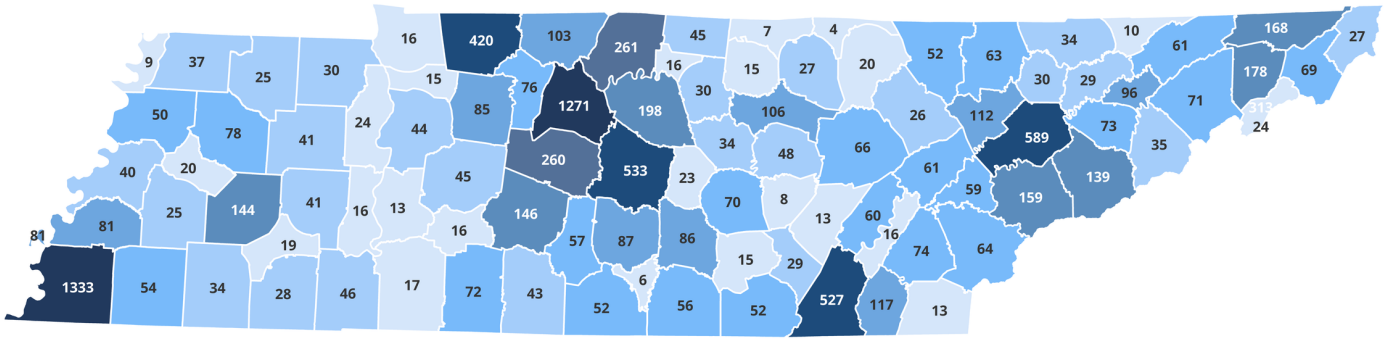
1. Birth Defects are Common, Costly, and Critical. Centers for Disease Control and Prevention. Updated June 28, 2023. Accessed October 16, 2023. https://www.cdc.gov/ncbddd/birthdefects/infographic.html

Birth Defects Are COMMON (continued)

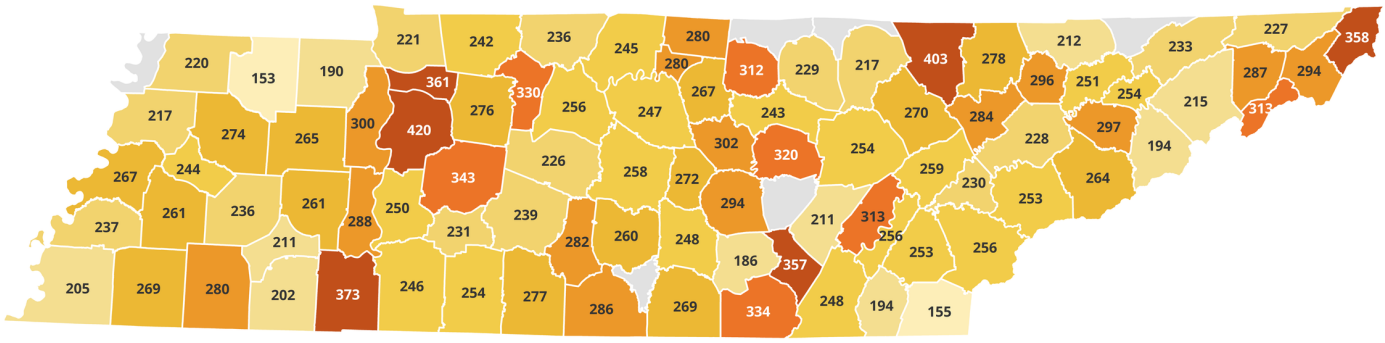
The maps below demonstrate the number, and rate, of birth defect cases by county. The highest case numbers from the 2016-2020 time period were for the metro counties of Shelby (1,333 cases) and Davidson (1,217 cases).

The second map shows several areas with **multiple high-rate counties** clustered together, including Houston, Humphreys, and Hickman in Middle Tennessee.

Number of Infants with Birth Defects



Rate (per 10,000 Live Births) of Infants with Birth Defects

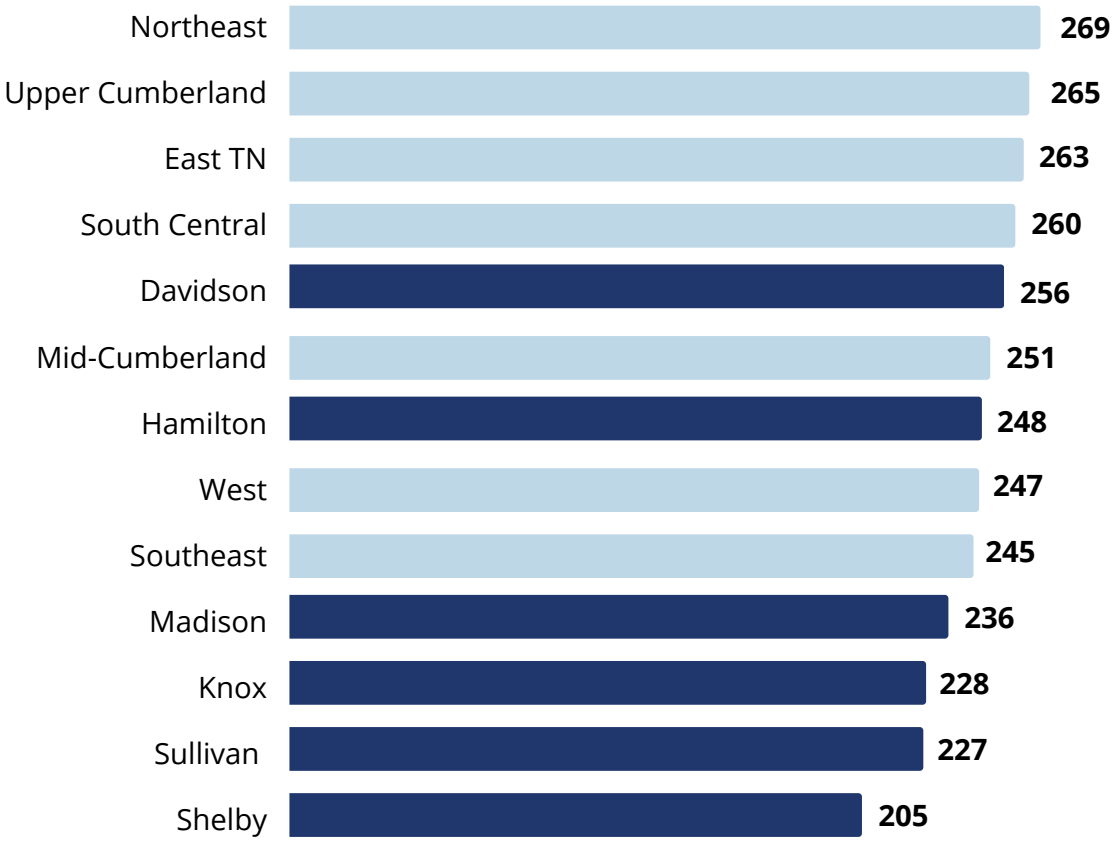


Note: the counties shown in grey are suppressed (e.g. data is not shown) due to small numbers, as rates based on counts less than 11 are statistically unreliable.

In Humphreys County (the highest statewide rate of 420 per 10,000 live births), infants were **nearly 3x as likely** to be diagnosed with a birth defect compared to Weakley County (the lowest statewide rate of 153 per 10,000 live births).

Birth Defects Are COMMON (continued)

Rate (per 10,000 Live Births) of Cases for Rural and Metro Regions



When grouping the counties into the 13 health department regions, a clear pattern emerges of the regions of **Northeast Tennessee, Upper Cumberland** and **East Tennessee** having the highest prevalence of birth defects overall.

Birth Defects are COSTLY

Of the over $2 billion in hospital charges for infants with birth defects over the past 5 years, 66% was billed to TennCare

In addition to the significant morbidity and mortality caused by birth defects, these conditions impose a significant financial burden on Tennessee families and the state as a whole. The below figure summarizes **total hospital charges** for Tennessee-resident infants at Tennessee hospitals ***between 2016-2020***:

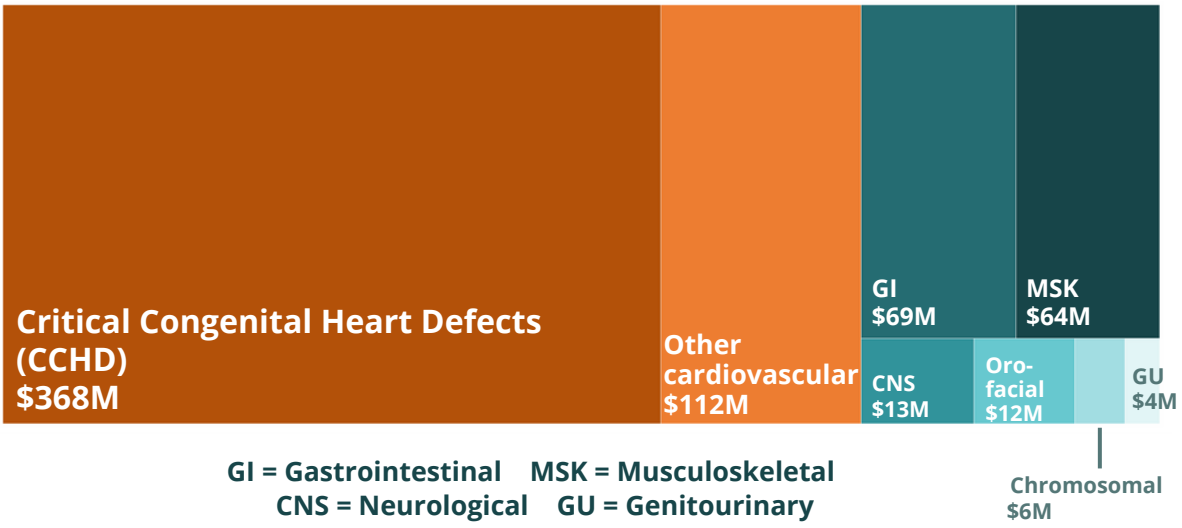
$648 M
where the birth defect was the primary reason for the hospitalization

$2.2 B
where the birth defect was the primary *or* contributing reason for the hospitalization

Most hospitalizations included in the $2.2 billion total shown above were for iinfant deliveries. The presence of a birth defect can increase charges, on average, to **10 times greater** than for infants without a birth defect.

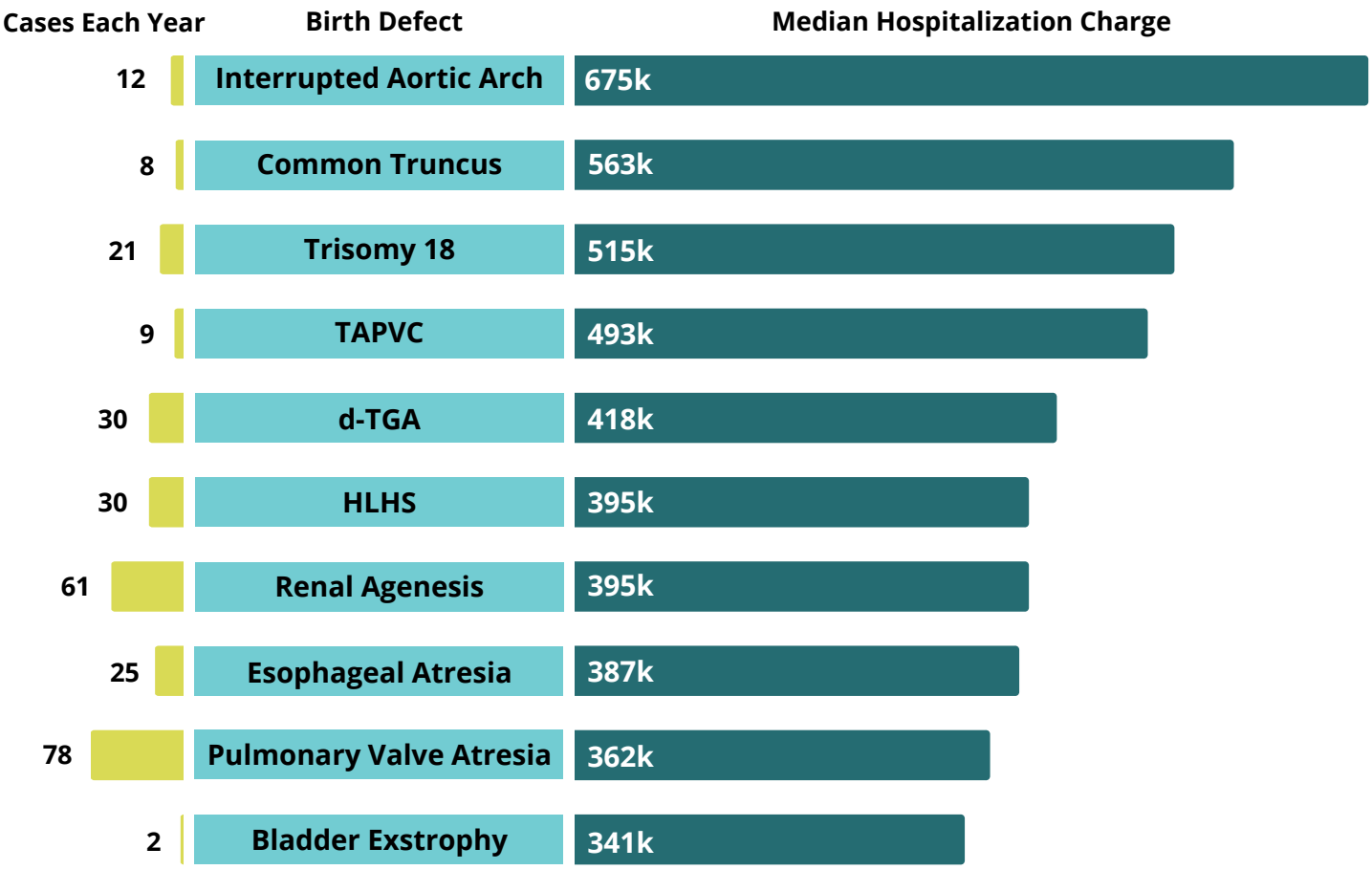
For inpatient hospitalizations with a major birth defect as the primary diagnosis (~$670M), CCHDs were *by far* the costliest conditions, accounting for **nearly $370 million** in hospital charges alone.

Total Hospitalization Charges for Birth Defects from Major Organ Systems, 2016-2020



Birth Defects are COSTLY (continued)

Top Ten Defects by Median Hospitalization Charge



The figure above demonstrates the average annual number of cases and the median hospitalization charge for infants with the listed birth defects as a primary diagnosis.

These ten conditions had the **highest median hospitalization charges** of all diagnoses included in this report. Six of the ten (interrupted aortic arch, common truncus, TAPVC, d-TGA, HLHS, and pulmonary valve atresia) are CCHDs which typically require surgery soon after birth.

Risk Factors

6% of infants born to women with pre-pregnancy diabetes had a birth defect, compared to 2% of infants overall

Although not all birth defects can be prevented, some health behaviors (such as smoking and drinking alcohol) and chronic health conditions (such as diabetes and hypertension) are associated with an increased risk of specific birth defects.

MORE THAN HALF of all pregnancies in Tennessee are unplanned.

Unplanned pregnancies can be associated with unhealthy behaviors and delayed access to prenatal care.

The majority of fetal development occurs in the first 4-12 weeks, often before a woman knows she is pregnant.

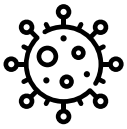
Closely-spaced pregnancies (less than 12 months apart) can increase the risk for pregnancy complications.

There was a **higher prevalence of all birth defects** for women with a BMI categorized as **underweight** or **obese**; women with a normal BMI had the lowest prevalence of infants with birth defects.

Women with **pre-pregnancy hypertension** were nearly **2 times as likely** to have a baby with a birth defect compared to women without hypertension. Women with gestational hypertension were 1.2 times as likely.

Women with **pre-pregnancy diabetes** were nearly **3 times as likely** to have a baby with a birth defect compared to women without diabetes. Women with gestational diabetes were 1.3 times as likely.

Infants born to women who **smoked** at all during pregnancy had **1.2 times the risk** of birth defects compared to women who did not smoke during pregnancy.



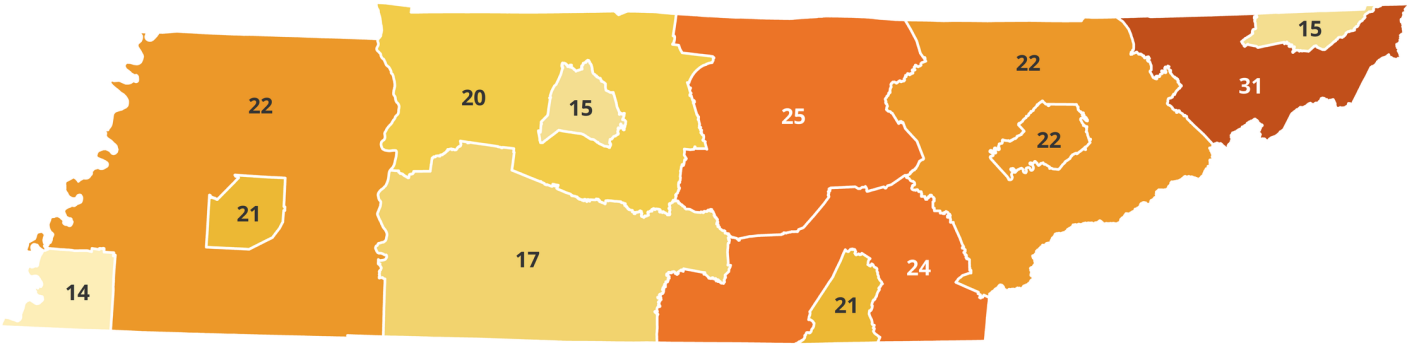
Some **infections** that a woman can get during pregnancy, such as **Zika, Rubella, Toxoplasmosis, Syphilis, Hepatitis C, and CMV**, can be harmful to the developing baby and can even cause birth defects. [2-4]

2. 10 Tips for Preventing Infections Before and During Pregnancy. Centers for Disease Control and Prevention. Updated September 29, 2022. Accessed October 17, 2023. https://www.cdc.gov/pregnancy/infections.html
3. Pregnancy and Rubella. Centers for Disease Control and Prevention. Updated December 31, 2020. Accessed October 19, 2023. https://www.cdc.gov/rubella/pregnancy.html
4. Congenital Syphilis. Centers for Disease Control and Prevention. Updated April 11, 2023. Accessed October 19, 2023. https://www.cdc.gov/std/syphilis/stdfact-congenital-syphilis.htm

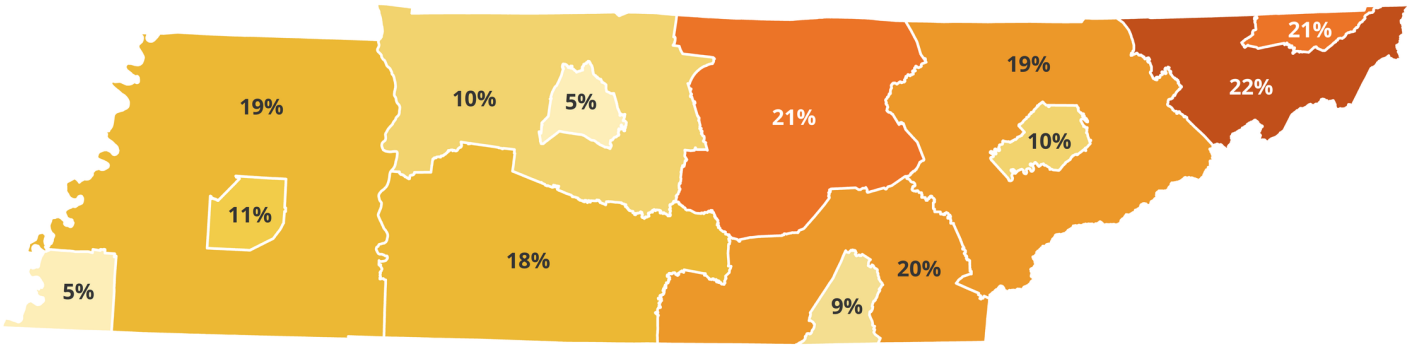
Risk Factors (continued)

Smoking (for both men and women) during pregnancy is another known risk factor for birth complications and defects, including orofacial clefts. The maps below do not establish causality, but do show similar patterns, specifically in the Northeast region, which had by far the highest statewide rate of infants with clefts and the highest percentage of smoking during pregnancy.

Rate (per 10,000 Live Births) of Orofacial Cleft



Percentage of Women who Smoked During Pregnancy

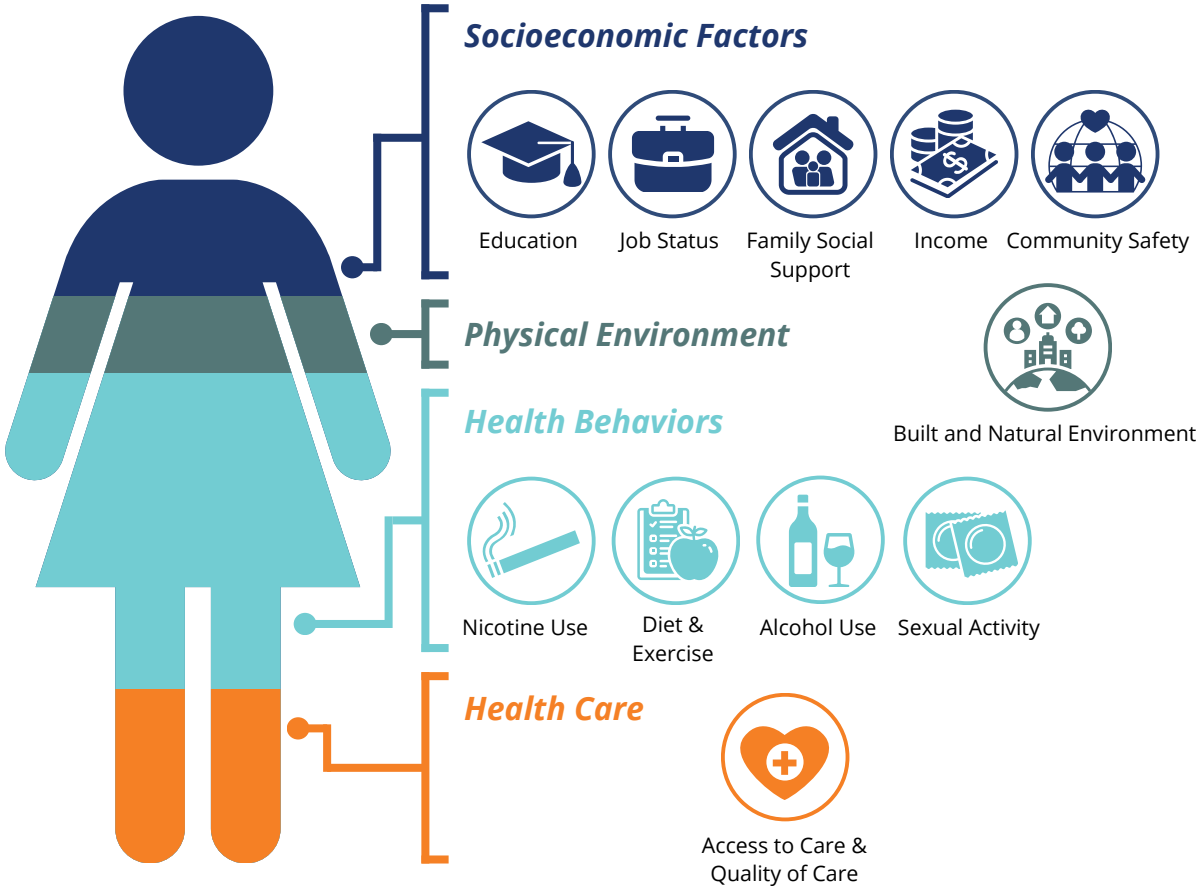


Occupational and environmental exposures (such as radiation, certain chemicals, and strenuous physical labor) may harm the health of mother and baby.[5] Hazardous work environments should be avoided during pregnancy. Pregnant workers, and those planning to become pregnant, should understand these risks and work with their employers to assure safety measures are in place.

5. Learn about Specific Exposures during Pregnancy & Breastfeeding. The National Institute for Occupational Safety and Health. Updated May 1, 2023. Accessed October 17, 2023. https://www.cdc.gov/niosh/topics/repro/specificexposures.html

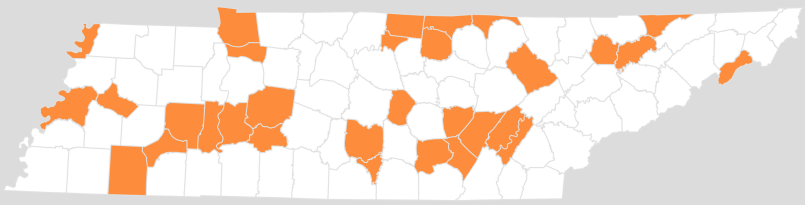
Social Determinants of Health

There are many factors that contribute to an individual's health, or conversely, illness. "Social determinants of health (SDOH) are conditions in the **places where people live, learn, work, and play** that affect a wide range of health and quality-of life-risks and outcomes." [6]



Access to Healthcare

March of Dimes defines **Maternity Care Deserts** as regions with "no hospitals providing obstetric care, no birth centers, no OB/GYN and no certified nurse midwives." [7]



33%

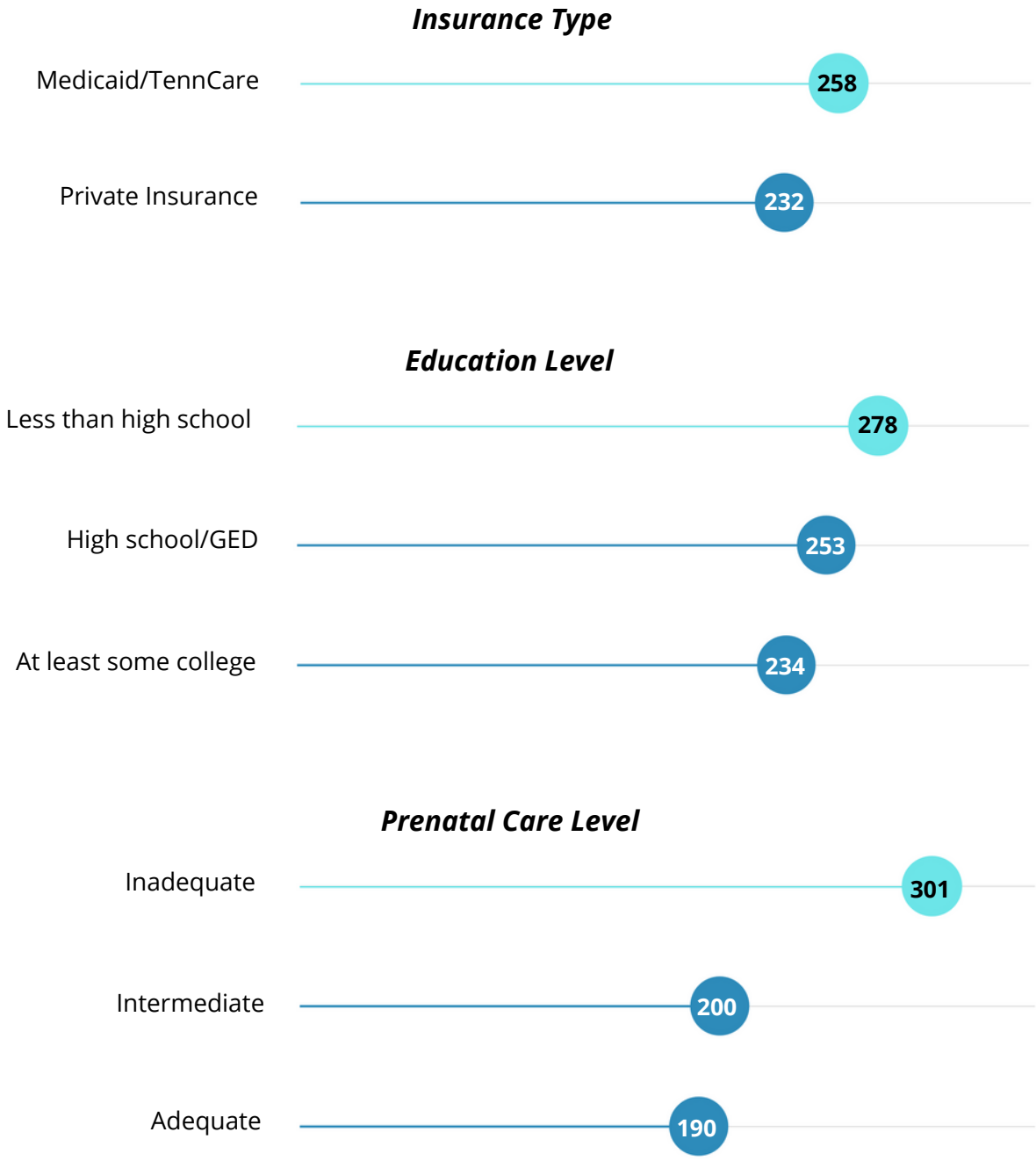
of TN counties are defined as Maternity Care Deserts [7]

6. Social Determinants of Health. Centers for Disease Control and Prevention. https://www.cdc.gov/socialdeterminants/index.htm. Updated December 8, 2022. Accessed September 15, 2023.
7. Where you live matters: Maternity care access in Tennessee. March of Dimes. https://www.marchofdimes.org/peristats/reports/tennessee/maternity-care-deserts. Published August 2, 2023. Accessed September 14, 2023.

Social Determinants of Health (continued)

The rate of birth defects (per 10,000 live births) differed by the SDOH factors listed below, with infants born to women with *Medicaid coverage*, *less than a high school education*, and *inadequate prenatal care* significantly more likely to be born with these conditions.

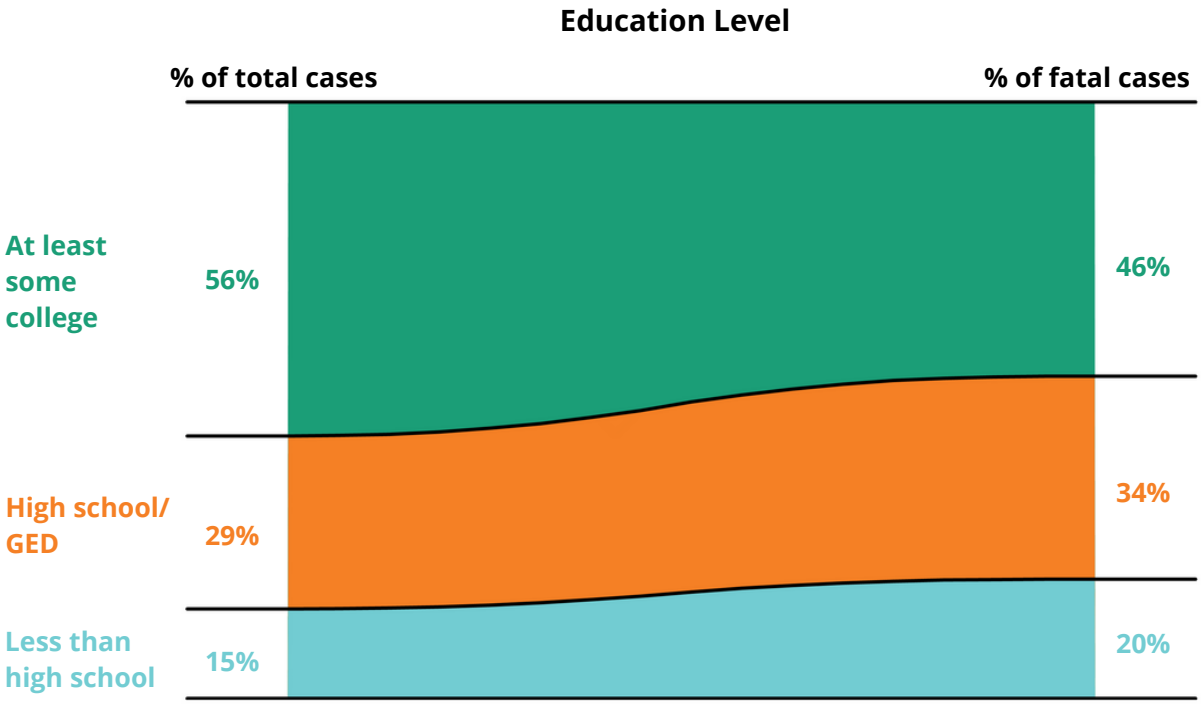
Rate (per 10,000 live births) by SDOH Factors

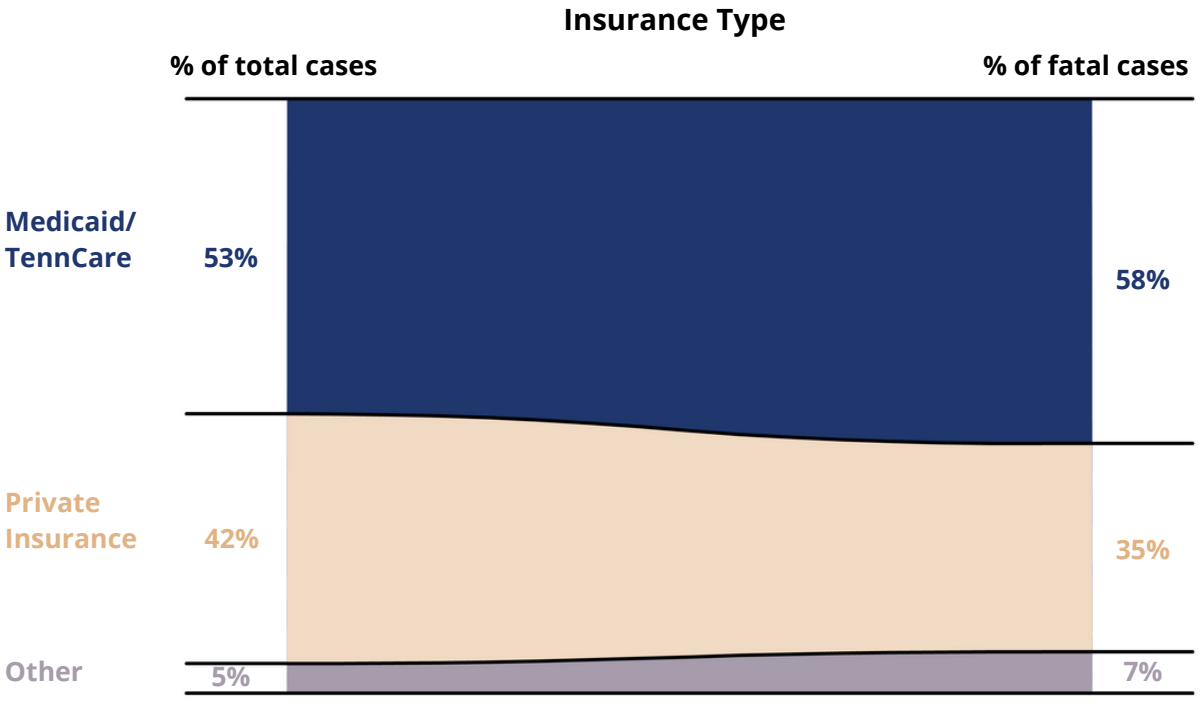


19% of women receiving Medicaid reported transportation issues as a barrier to getting early prenatal care, compared to just 5% who had private insurance

Social Determinants of Health (continued)

The impact of SDOHs is also reflected in the outcomes experienced by children with birth defects. In the charts below, the percentages on the left are for **children with birth defects overall** while the percentages on the right are for **children with birth defects who died during the first year of life**. Infants with birth defects born to women with *low socioeconomic status* (indicated by education level and Medicaid coverage), were significantly more likely to *die before their first birthday*.





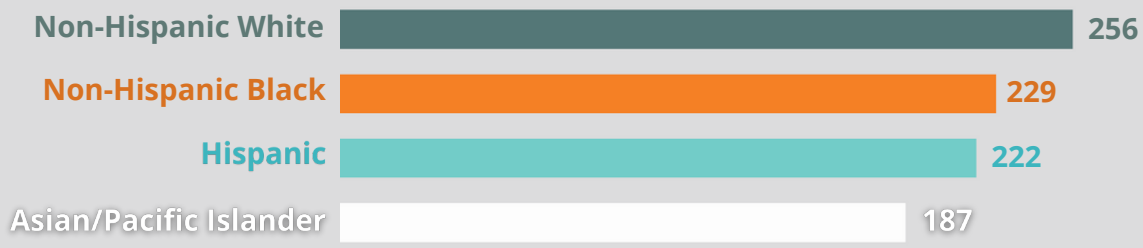
Maternal Characteristics

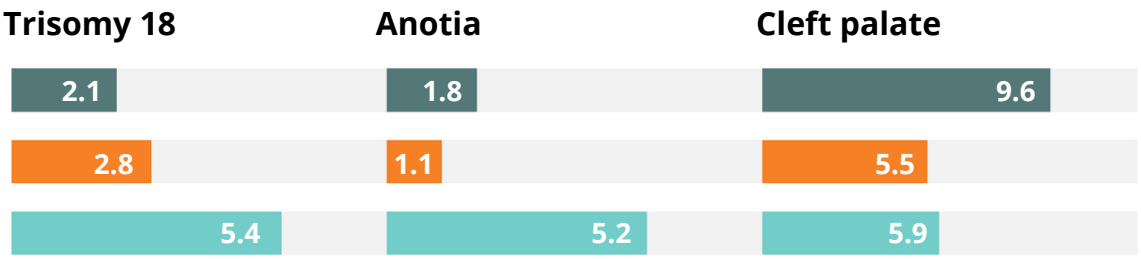
Maternal Race/Ethnicity

Overall, infants born to non-Hispanic White women had the highest rate (per 10,000 live births) of birth defects. However, the breakdown by race/ethnicity varied significantly across the individual conditions. For example, Trisomy 18 and anotia were much more common in infants born to Hispanic women, while cleft palate was most common amongst infants born to non-Hispanic White women. Infants of Asian/Pacific Islander descent are not included in the diagnosis-specific charts below due to small numbers.

Hispanic infants were nearly 3x as likely to be diagnosed with Trisomy 18 and anotia, compared to non-Hispanic White infants

Birth Defects Overall





Maternal Characteristics (continued)

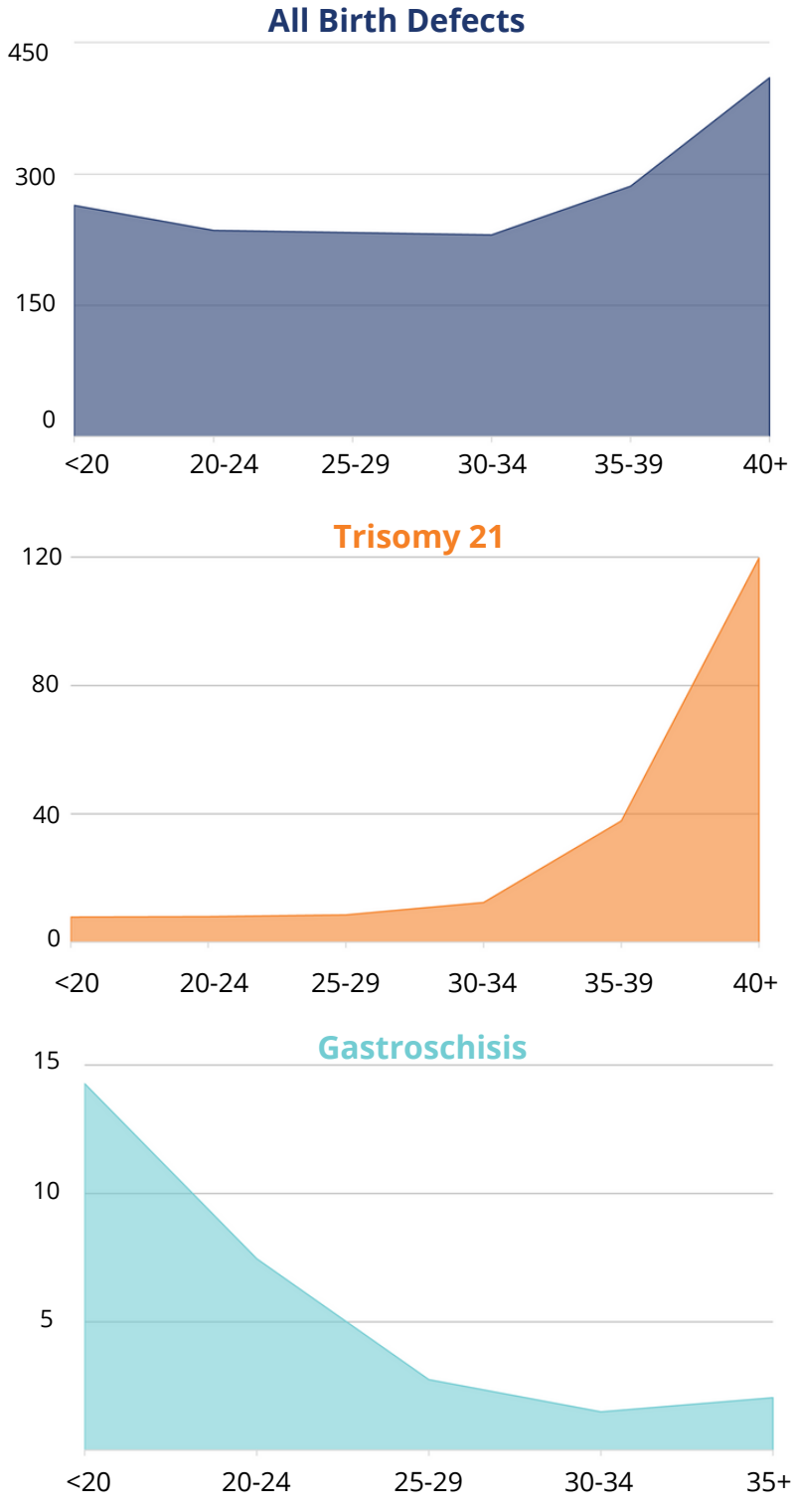
Maternal Age

Overall, women 40 and older were mostly likely to give birth to an infant with a birth defect. This trend was especially pronounced for chromosomal birth defects, including Trisomy 21, the most common chromosomal condition. Other conditions like gastroschisis, an abdominal wall defect, exhibited the opposite trend, with infants born to young women most likely to be diagnosed.

Trisomy 21 (Down syndrome) was 15x as common among infants born to women 40 and older, compared to those born to women less than 20

Gastroschisis was 7x as common among infants born to women less than 20, compared to those born to women 35 and older

Rate (per 10,000 live births) by Maternal Age



COVID-19

Here's what is known about **COVID-19 and pregnancy in Tennessee**:

- Between March 1, 2020, and March 31, 2023, 8% of women who gave birth were diagnosed with COVID-19 during pregnancy.[8]
- Among pregnant women with COVID-19, Intensive Care Unit admissions at delivery were 4 times as high as pregnant women without COVID-19.[9]
- Data shows that women with COVID-19 infection during pregnancy might be at a higher risk for preterm birth or stillbirth.[10-13]

Here's what is known about **COVID-19 in pregnancy and birth defects**:

- To date, national studies have not indicated an increased risk of having a baby born with a birth defect for mothers who are diagnosed with COVID-19 during pregnancy.[14]
- Current studies have not found an increased risk of birth defects for those who are pregnant and receive a COVID-19 vaccine.[15,16]
- COVID-19 vaccinations during pregnancy are considered safe, effective, and are recommended to prevent serious illness and potential complications.[17-19]

The 2020 birth defects data included in this report does not distinguish between those who were COVID-19 positive while pregnant and those who were not.

In 2020, the rate of infant death due to birth defects and overall case rate remained stable from previous years, with no signs of a significant increase or decrease from previous years. TNBDSS will continue to monitor these trends as further years of data are finalized.

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** "Organization of Teratology Information Specialists (OTIS) is a professional organization comprised of experts engaged in assessing and evaluating risks to pregnancy and breastfeeding outcomes from medications and other exposures."*

Service Referrals

TNBDSS will continue to coordinate with a child's healthcare provider and parent/guardian to ensure awareness of Children's Special Services (CSS), Tennessee Early Intervention Services (TEIS), Family Voices of Tennessee, and when appropriate, assist with service referrals. *(Please scan each QR code below to learn more).*

For the Fiscal Year 2020,

549

Tennessee children (with a qualifying birth defect diagnosis) were enrolled in CSS.

Over

$350k

was paid out as last resort payments for these children and their families.

TEIS (TN EARLY INTERVENTION SYSTEM)

- Referrals may be made for eligible children from birth - 3 years of age
- Provides services and referrals for children who have disabilities or other developmental delays (refer to qualifying diagnosis list)
- Supports families in helping their child's development
- Helps the child participate in family and community activities
- Encourages the active participation of the whole family in the child's development
- Referral form can also be found in the MyTN app (available in any mobile app store)

CHANT (COMMUNITY HEALTH ACCESS AND NAVIGATION IN TENNESSEE)

- Provides care coordination/assistance in navigating the health and social service needs of qualifying families and individuals for various programs
- Referrals are accepted from all medical providers and social service agencies
- Self-referrals to CHANT are also accepted

CSS (CHILDREN'S SPECIAL SERVICES)

- Provides reimbursement for medical care for eligible children with physical disabilities from birth to 21 years of age
- Eligibility is determined by: residency, income, age and diagnosis
- The program consists of two components:
  - Care coordination
  - Reimbursement of medical services as a payor of last resort
- CSS provides: case management, referrals, community resources, collaboration with schools, managed care organizations (MCOs) and other agencies, transition planning, medical equipment, assistive technology, supplies, prescription assistance, therapies, screening/evaluations, and support services for families (medical and non-medical needs)

FAMILY VOICES OF TENNESSEE

- A program of the TN Disability Coalition
- Families provide free emotional and educational support to other families of children with special healthcare needs, chronic illnesses or disabilities
- Connects families to community resources and peer support
- Shares first-hand experience and tips
- Makes referrals to outside agencies
- Provides assistance in navigating the healthcare system
- Connects parents/families in peer networks

Next Steps

This report reveals **major disparities in birth defects** overall prevalence and **infant mortality due to a birth defect**. Race, education level, income level, and geography of residence are only a few examples of where these disparities persist. The findings outlined in this report present opportunities for future directions for not only TNBDSS, but a variety of community partners and decision makers.

The main goals of the Tennessee Department of Health are to **increase access** and **prevention**. By addressing these identified disparities and ensuring access to resources and care for those most at-risk, we can help to promote the best outcomes possible for all Tennessee families.

Future Directions for TNBDSS

- Address the connection between preconception health and birth defects.
- Inform supportive services of county-level birth defect prevalence for program planning purposes.
- Prioritize those populations identified as most at risk for having a baby with a birth defect.
- Implement additional activities outlined in the enhanced surveillance cooperative agreement including improving data quality, ensuring access to supportive services, program evaluation, and dissemination.

Recommendations for Partners and Decision Makers

Community and Statewide Agencies

- Ensure programs related to preconception health and healthy pregnancy are accessible to residents in identified high-risk areas and include targeted linguistically-appropriate media.
- Programs related to supportive services for families with a child with a birth defect could utilize the maps included in this report to ensure adequate coverage and outreach in identified high-burden areas.
- Prioritize efforts to improve access to care in counties designated as Maternity Care Deserts, including:
  - partnering with existing programs that offer alternative care options, such as telehealth.
  - exploring opportunities for community-based and payor-based care management programs to provide for specific needs such as remote patient monitoring and transportation.

Clinical and Hospital Systems

- Ensure appropriate funding and staff access to translation services.
- Offer staff implicit bias training.
- Prioritize implementing access to prenatal and maternal care practitioners in counties designated as a Maternity Care Desert.

Healthcare Providers

- Ensure proper utilization of available translation services.
- Ensure referrals to supportive services for families of babies born with a birth defect.

Legislators

- Prioritize funding for financial incentives for healthcare systems and practitioners to provide access in designated Maternity Care Deserts.

Success Stories

Tennessee is making great strides in year 3 to accomplish the activities outlined in the **5-year CDC Cooperative Agreement** to enhance state birth defects surveillance.

Improved Data Quality: TNBDSS has expanded its data sources for faster and more robust case finding, case agreement, and case verification. TNBDSS is partnering with other TDH programs for secure data sharing to ensure program alignment.

Timeliness: TNBDSS continues to gain access to Electronic Health Records in key birthing facilities across the state for faster case verification.

Collaboration: TNBDSS is partnering with TDH's Environmental Epidemiology Program on its new Environmental Public Health Tracking initiative. This initiative will connect information on various environmental exposures to birth defects data to help identify any potential associations.

Appendix I: Birth Defects by Organ System, 2016-2020

Birth Defect	Number	Frequency	Prevalence (per 10K births)
Chromosomal			
Deletion 22q11.2	24	1 in 16,735	0.6
Trisomy 13	50	1 in 8,033	1.2
Trisomy 18	104	1 in 3,862	2.6
Trisomy 21 (Down syndrome)	603	1 in 666	15.0
Turner syndrome	48	1 in 4,093	2.4
Total Chromosomal Cases	812	1 in 495	20.2
Cardiovascular			
Critical congenital heart disease (CCHD) conditions			
Coarctation of the aorta	333	1 in 1,206	8.8
Common truncus (truncus arteriosus)	38	1 in 10,569	0.9
Double outlet right ventricle (DORV)	134	1 in 2,997	3.3
Ebstein anomaly	65	1 in 6,179	1.6
Hypoplastic left heart syndrome	149	1 in 2,696	3.7
Interrupted aortic arch (IAA)	62	1 in 6,478	1.5
Pulmonary valve atresia and stenosis	390	1 in 1,030	9.7
Tetralogy of Fallot (TOF)	255	1 in 1,575	6.3
Total anomalous pulmonary venous connection (TAPVC)	46	1 in 8,731	1.1
Transposition of the great arteries (TGA)	150	1 in 2,678	3.7
Tricuspid valve atresia and stenosis	48	1 in 8,367	1.2
Single ventricle	62	1 in 6,478	1.5
Other cardiovascular conditions			
Aortic valve stenosis	70	1 in 5,738	1.7
Atrial septal defect	8,713	1 in 46	216.9
Atrioventricular septal defect	266	1 in 1,510	6.6
Ventricular septal defect	2,090	1 in 192	52.0
Total Cardiovascular Cases	10,053	1 in 40	250.3
Orofacial			
Choanal atresia	97	1 in 4,141	2.4
Cleft lip with cleft palate	314	1 in 1,279	7.8
Cleft lip alone (without cleft palate)	137	1 in 2,932	3.4
Cleft palate alone (without cleft lip)	338	1 in 1,188	8.4
Total Orofacial Cases	879	1 in 457	21.9

Appendix I: Birth Defects by Organ System, 2016-2020

Birth Defect	Number	Frequency	Prevalence (per 10K births)
Eye/Ear			
Anophthalmia/microphthalmia	58	1 in 6,925	1.4
Anotia/microtia	88	1 in 4,564	2.2
Congenital cataract	104	1 in 3,862	2.6
Total Eye and Ear Cases	237	1 in 1,695	5.9
Gastrointestinal			
Biliary atresia	211	1 in 1,903	5.3
Esophageal atresia/tracheoesophageal fistula	124	1 in 3,239	3.1
Rectal and large intestinal atresia/stenosis	207	1 in 1,940	5.2
Small intestinal atresia/stenosis	195	1 in 2,060	4.9
Total Gastrointestinal Cases	683	1 in 588	17.0
Genitourinary			
Bladder exstrophy	12	1 in 33,469	0.3
Cloacal exstrophy	5	1 in 80,326	0.1
Congenital posterior urethral valves	64	1 in 3,206	3.1
Hypospadias	2,287	1 in 90	111.5
Renal agenesis/hypoplasia	304	1 in 1,321	7.6
Total Genitourinary Cases	2,651	1 in 152	66.0
Central Nervous System			
Anencephaly	74	1 in 5,464	1.8
Encephalocele	65	1 in 6,179	1.6
Holoprosencephaly	57	1 in 7,046	1.4
Spina bifida without anencephaly	187	1 in 2,148	4.7
Total Central Nervous System Cases	371	1 in 1,038	9.2
Musculoskeletal			
Clubfoot	1,004	1 in 400	25.0
Diaphragmatic hernia	179	1 in 2,244	4.5
Gastroschisis	171	1 in 2,349	4.3
Reduction deficits (extremities)	174	1 in 2,308	4.3
Omphalocele	116	1 in 3,462	2.9
Craniosynostosis	658	1 in 610	16.4
Total Musculoskeletal Cases	2,238	1 in 495	55.7
Total Birth Defect Cases	9,837	1 in 41	244.9

Appendix II: Definitions

Anencephaly: Congenital absence of the skull, with cerebral hemispheres completely missing or reduced to small masses attached to the base of the skull. Anencephaly is not compatible with life.

Anophthalmia: A developmental defect characterized by complete absence of the eyes, or by the presence of vestigial eyes.

Anotia: A congenital absence of one or both ears.

Aortic valve stenosis: A cardiac anomaly characterized by a narrowing or stricture of the aortic valve.

Atresia: Absence or closure of a normal opening.

Atrial Septal Defect (ASD): A congenital cardiac malformation in which there are one or several openings in the atrial septum (wall between the right and left atria).

Atrioventricular Septal Defect (AVSD): A congenital heart defect in which there are holes between the chambers of the right and left sides of the heart, and the valves that control the flow of blood between these chambers may not be formed correctly.

Biliary atresia: A congenital absence or underdevelopment of one or more of the ducts in the biliary tract.

Bladder exstrophy: Incomplete closure of the anterior wall of the bladder and the abdominal cavity. The upper urinary tract is generally normal. Often associated with anorectal and genital malformations.

Choanal atresia or stenosis: A congenital anomaly in which a bony or membranous formation blocks the passageway between the nose and the pharynx.

Chromosome abnormalities: A major group of genetic diseases in which alterations of chromosome number or structure occur and are observable by microscope.

Cleft lip: The congenital failure of the fetal components of the lip to fuse or join, forming a groove or fissure in the lip. Infants with this condition can have difficulty feeding and may use assistive devices for feeding. This condition is corrected when the infant can tolerate surgery.

Cleft palate: The congenital failure of the palate to fuse properly forming a grooved depression or fissure in the roof of the mouth. This defect varies in degree of severity. The fissure can extend into the hard and soft palate and into the nasal cavities. Infants with this condition have difficulty feeding, and may use assistive devices for feeding. Surgical correction is begun as soon as possible. Children with cleft palates are at high risk for hearing problems due to ear infections.

Cloacal exstrophy: The condition where some internal organs that are normally in the lower abdomen are exposed externally. Additionally, some of these organs may not have developed correctly and the lower parts of the reproductive, urinary and intestinal tracts may not be completely formed.

Clubfoot: A condition in which the foot is turned inward, often so severely that the bottom of the foot faces sideways or even upward.

Coarctation of the Aorta (COA): Localized narrowing of the aorta. This condition can vary from mild to severe.

Common Truncus Arteriosus: A congenital heart defect in which the common arterial trunk fails to divide into pulmonary artery and aorta.

Appendix II: Definitions

Congenital cataract: When the lens of the eye is cloudy instead of clear at birth, making it hard to see.
Congenital Diaphragmatic Hernia (CDH): A failure of the diaphragm to form completely, leaving a hole. Abdominal organs can protrude through the hole into the chest cavity and interfere with development of the heart and lungs. Usually life-threatening and requires emergent surgery.
Craniosynostosis: The condition in which the bones in a skull join too early. This happens before the brain is fully formed.
Deletion 22q11.2 (DiGeorge syndrome): The chromosomal abnormality characterized by a missing chromosome 22. This deletion results in the poor development of several body systems.
Double Outlet Right Ventricle (DORV): A congenital heart defect in which the body's main artery (aorta) and the lung artery (pulmonary artery) do not connect to the usual areas in the heart. Sometimes these blood vessels also are reversed from their usual positions. This condition can lead to heart failure and poor growth**.**
Down syndrome (Trisomy 21): The chromosomal abnormality characterized by an extra copy of chromosome 21. In rare cases this syndrome is caused by translocation. Many of these infants have congenital heart disease.
Ebstein anomaly: A congenital heart defect in which the tricuspid valve is displaced downward into the right ventricle.
Esophageal atresia (EA): An incomplete formation of the esophagus. Usually a surgical emergency. Frequently associated with a Tracheoesophageal Fistula.
Folic acid deficiency: A lack of folic acid in the mother's diet which may lead to an increased risk for neural tube defects.
Gastroschisis: A congenital opening of the abdominal wall with protrusion of the intestines. This condition is surgically treated.
Hypoplastic Left Heart Syndrome (HLHS): Atresia, or a marked hypoplasia, of the aortic valve, atresia or marked hypoplasia of the mitral valve, with hypoplasia of the ascending aorta and underdevelopment of the left ventricle.
Hypospadias: A congenital defect in which the urinary meatus (urinary outlet) is on the underside of the penis or on the perineum (area between the genitals and anus). The urinary sphincters are not defective so incontinence does not occur. The condition may be surgically corrected if needed for cosmetic, urologic, or reproductive reasons.
Interrupted Aortic Arch (IAA): A congenital heart defect in which the aorta doesn't form completely - part of the aortic arch is missing, leaving a gap or interruption.
Microphthalmia: The congenital abnormal smallness of one or both eyes. Can occur in the presence of other ocular defects.
Microtia: A small or maldeveloped external ear and atretic or stenotic external auditory canal.
Multiple congenital anomalies: Term used to describe the presence of more than one anomaly at birth.
Neural Tube Defect (NTD): A defect resulting from failure of the neural tube to close in the first month of pregnancy. The major conditions include anencephaly, spina bifida, and encephalocele.

Appendix II: Definitions

Obstructive genitourinary defect: Stenosis or atresia of the urinary tract at any level. Severity of the defect depends largely upon the level of the obstruction. Urine accumulates behind the obstruction.
Omphalocele: The protrusion of intestines into the umbilicus. The defect is usually closed surgically soon after birth.
Patent Foramen Ovale (PFO): A hole between the left and right atria (upper chambers) of the heart. This hole exists in everyone before birth, but most often closes shortly after being born. PFO is what the hole is called when it fails to close naturally after a baby is born.
Pulmonary valve atresia or stenosis: Failure of formation of the pulmonary valve or a narrowing or obstruction of the pulmonary valve, resulting in obstruction of blood flow from the right ventricle to the pulmonary artery.
Reduction deficits (extremities): The congenital absence of a portion of the lower or upper limbs. There are two general types of defect, transverse and longitudinal. Transverse defects appear like amputations with the complete or partial absence of the arm or leg. Longitudinal defects are missing rays of the limb and may involve the preaxial (thumb or big toe side) or central parts of the arm or leg.
Renal agenesis or dysgenesis: The failure, or deviation, of embryonic development of the kidney.
Renal hypoplasia: A condition where part of a kidney does not fully develop in the womb. The kidney may be only slightly smaller than usual or it may be tiny.
Single ventricle: A congenital heart defect which involves having only one ventricle that is large enough or strong enough to pump blood effectively. This condition often requires surgical correction and with advancements in technology, can now lead to survival well into adulthood.
Spina bifida: An incomplete closure of the vertebral spine (usually posterior) through which spinal cord tissue or membranes (meninges) covering the spine herniate.
Small intestinal atresia or stenosis: A narrowing or incomplete formation of the small intestine obstructing movement through the digestive tract.
Tetralogy of Fallot (TOF): The simultaneous presence of a ventricular septal defect, pulmonic stenosis, a malposition aorta that overrides the ventricular septum, and right ventricular hypertrophy.
Total Anomalous Pulmonary Venous Connection (TAPVC): A congenital heart defect in which the lung blood vessels, called the pulmonary veins, attach to the wrong places in the heart. Surgery is almost always indicated in early childhood, with lifelong health checkups and monitoring.
Tracheoesophageal fistula (TEF): An abnormal connection between the esophagus and trachea.
Transposition of the Great Arteries (TGA): A congenital malformation in which the aorta arises from the right ventricle and the pulmonary artery from the left ventricle (opposite of normal), so that the venous return from the peripheral circulation is recirculated without being oxygenated in the lungs. Immediate surgical correction is needed. When this is not associated with other cardiac defects, and not corrected, the condition is fatal.

Appendix II: Definitions

Tricuspid valve atresia or stenosis: A congenital cardiac condition characterized by the absence or constriction of the tricuspid valve.
Trisomy 13 (Patau syndrome): The chromosomal abnormality caused by an extra chromosome 13. Characterized by impaired midline facial development, cleft lip and palate, polydactyly (the condition of having more than 5 fingers or toes on each limb) and severe intellectual disabilities. Most infants do not survive beyond 6 months of life.
Trisomy 18 (Edwards syndrome): The chromosomal abnormality caused by an extra copy of chromosome 18. It is characterized by intellectual disabilities, growth restriction, low-set ears, skull malformation and short digits. Survival for more than a few months is rare.
Trisomy 21 (T21): See Down Syndrome.
Turner syndrome: The chromosomal abnormality caused by a missing or partially missing X chromosome (sex chromosomes). This condition only affects females and can cause a variety of medical and developmental problems.
Ventricular Septal Defect (VSD): A congenital cardiac malformation in which there are one or several openings in the ventricular system (Muscular and fibrous wall between the right and left ventricle or right and left lower chambers of the heart).

To learn more, visit TNBDSS:
https://www.tn.gov/health/BirthDefectsInfo
on the Tennessee Department of Health's website.

TIPS FOR A HEALTHY PREGNANCY FOR WOMEN AND THEIR PARTNERS

- Talk to your healthcare professional about reproductive life planning
- Plan and space pregnancies at least 18 months apart
- Discuss family history of medical conditions with your partner and your healthcare professional
- See your healthcare professional regularly and early in pregnancy
  - Discuss your risks and how they should be managed
  - Discuss which foods should be avoided
  - Discuss your options for genetic screening
  - Discuss any medication use with your healthcare professional (prescription, over-the-counter, and supplements)
  - Before stopping or changing medications, discuss with your healthcare professional
  - Standard recommended Folic Acid dose for most pregnancies is 400mcg at least one month prior to pregnancy
    - Discuss any additional Folic Acid needs (such as higher doses and/or starting earlier)
- Prevent and/or manage chronic health conditions, such as diabetes and hypertension (both men and women)
  - Ask your provider if taking a daily aspirin is necessary
- Strive to reach and maintain a healthy weight*
- Be physically active every day
- Brush your teeth daily and see a dentist at least once during your pregnancy
- Remain current with vaccinations, including flu, Tdap, and COVID-19
- Avoid infections and seek medical care for any suspected illness, including sexually transmitted infections
- Avoid harmful substances, such as nicotine-containing products, alcohol, and drugs (opioids, marijuana, cocaine, methamphetamines, and other "illegal" drugs)
- Seek cessation support for pre-existing substance use disorders
- Be aware/avoid potentially harmful exposures at work and home
- Report any concerning symptoms to your healthcare provider ASAP (including fever, swelling, headaches, heartburn, blurred vision, etc.)
- Find tips to cope with stress and changing emotions and learn the signs of depression & anxiety (National Maternal Mental Health Hotline is available 24/7 by calling 1-833-943-5746)
- Partners should also seek mental health support, both during pregnancy and postpartum (Tennessee Statewide Crisis Line is available 24/7 by calling 1-855-274-7471)
- For ANY thoughts of suicide or mental health emergency, call 988

*Although current industry standards use BMI as a measure of health, TNBDSS recognizes its limitations and encourages families to discuss with their healthcare provider.

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