Tennessee Birth Defects Data Report 2014-2018

Tennessee Department of Health | December 2021

TN Department of Health

Table of Contents

Executive Summary

Birth defects are common, costly, and critical. The Tennessee Birth Defects Surveillance System (TNBDSS) estimates that between 2014 and 2018, over 52 million dollars was spent on hospitalizations related to critical congenital heart defects alone.[1,2] From 2014 and 2018, there were 14,874 babies (approximately 3,000 babies per year) diagnosed with birth defects in Tennessee. This translates to one in every 27 babies (about 4% of all babies) born with a birth defect. The most commonly identified birth defects during this period were atrial septal defect (ASD), a hole or opening in the heart, and hypospadias, a genitourinary defect that affects males. Birth defects accounted for 21% of infant deaths in Tennessee during this time frame, making it the leading cause of infant mortality.

The Tennessee Department of Health's Tennessee Birth Defects Surveillance System, as outlined in Tennessee Code Annotated § 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 guidance on prevention efforts; and, for children with specific neurologic birth defects, makes referrals for needed services, such as early intervention. This annual surveillance report provides details on the prevalence of 47 major birth defects and fetal alcohol syndrome for Tennessee infants born in the years 2014 through 2018. This report also includes specific information about birth defect rates by socio-demographic factors, known risk factors, region and county of residence.

Major findings from this report include:

- A higher prevalence of birth defects is noted among infants of women with a 12th grade education or less and women on Medicaid compared to private insurance.
- Smoking during pregnancy and maternal chronic health conditions such as diabetes, hypertension, and obesity are associated with an increased risk of birth defects.

[1] Arth AC, Tinker SC, Simeone RM, Ailes ED, Cragan JD, Grosse SD. Inpatient Hospitalization Costs Associated with Birth Defects Among Persons of All Ages – United States, 2013. MMWR Morb Mortal Wkly Rep. 2017;66(2):41-46.
[2] Cost estimates are based on the median cost by birth defect organ system in Table 2 of Arth et al. The median cost was then multiplied by the number of defects, provided in Table 7 of this report, of the corresponding diagnoses included in the authors' organ system definition.

- Certain types of birth defects, especially chromosomal defects, are more common among babies born to mothers 35 years of age and older.
- The highest rates of birth defects were found in Shelby County, West, and the Northeast regions.
- The highest rates of infant mortality due to a birth defect were found in Madison County, Shelby County, and the Northeast region.
- Non-Hispanic Black infants had the highest rate of birth defects across maternal racial/ethnic groups.

Key Prevention Messages:

- Birth defects surveillance programs play a key role in efforts to prevent birth defects by identifying factors that increase or decrease the risk of birth defects. Improved understanding of the risk factors for birth defects allows for the development of recommendations, policies, and services to help prevent them.
- Women should see their health care providers when planning a pregnancy and begin prenatal care as early as possible.
- Women of childbearing age should consume at least 400 micrograms of folic acid every day. Folic acid supplementation should begin at least one month before becoming pregnant.
- Preventing and managing chronic health conditions (like diabetes and high blood pressure) and adopting healthy behaviors before pregnancy can help prevent birth defects
- Harmful substances (such as alcohol, tobacco, marijuana, and illicit drugs) and certain medications should be avoided during pregnancy.
- It is important for women and their health care providers to discuss any routine vaccinations and medication use, including supplements and over-the-counter drugs, that are given before and during pregnancy.
- It is recommended that women plan and space pregnancies at least 18 months apart to reduce the risk of adverse outcomes such as preterm birth, low birthweight, uterine rupture, maternal anemia, and maternal mood disorders.[3]

[3] Conde-Agudelo A, Rosas-Bermúdez A, Kafury-Goeta AC. Birth spacing and risk of adverse perinatal outcomes: a meta-analysis. JAMA. 2006 Apr 19;295(15):1809-23.

Introduction

What are Birth Defects?

Birth defects 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).

Why Study Birth Defects?

According to the Centers for Disease Control and Prevention (CDC), an infant is born every four and half minutes with a birth defect in the United States. Nationally, about 120,000 babies (nearly one in 33 babies) are affected by birth defects each year.[4] Birth defects cause 1 in 5 infant deaths and contribute to life- long disability. In addition to the emotional impact on affected children and their families, birth defects have financial implications for families as they are often faced with missing work and subsequent wages due to medical care associated with birth defects. The cost of medical care associated with birth defects in Tennessee is substantial. For example, TNBDSS estimates that between 2014 and 2018, about 750 thousand dollars was spent on hospitalizations due to ear-related birth defects, over 7 million was spent on orofacial birth defect hospitalizations, 6.9 million was spent on hospitalizations related to chromosomal birth defects, and over 52.1 million dollars was spent on hospitalizations associated with critical congenital heart defects.[1,2]

Despite the prevalence and potential for significant morbidity and/or mortality, the underlying cause of most birth defects is largely unknown. This underscores the importance of birth defect surveillance, which can detect changes in the occurrence of birth defects and identify associations between exposures and birth defects. A birth defect surveillance program also plays a critical role in providing education about birth defects and risk factors, such as drinking alcohol during pregnancy, smoking during pregnancy, low blood folate levels, poorly controlled blood sugar levels in diabetic

[4] Centers for Disease Control and Prevention. Update on Overall Prevalence of Major Birth Defects—Atlanta, Georgia, 1978-2005. MMWR Morb Mortal Wkly Rep. 2008;57(1):1-5.

mothers, and certain maternal infections

Finally, real-time birth defect surveillance programs can ensure timely connection to key support services, such as early intervention, home visiting, care coordination, and parent support organizations.

About this Report

The Tennessee Birth Defects Data Report is a statewide population-based birth defects report prepared by the Tennessee Birth Defects Surveillance System (TNBDSS). This report provides details about the prevalence of 47 major birth defects[5] and fetal alcohol syndrome for Tennessee infants born in the years 2014 through 2018. TNBDSS selected which birth defects to study based on national surveillance recommendations from the National Birth Defects Prevention Network. This report also includes specific information about birth defect rates by socio-demographic characteristics and known risk factors.

Individual birth defect counts and rates are presented in tabular form for the state overall. Data are also broken down by maternal education, race/ethnicity (based on maternal self-report), age, and maternal health characteristics, such as pre-pregnancy diabetes and smoking during pregnancy. This report provides education on birth defects prevention, resources to help promote a healthy pregnancy, and suggests future directions.

In Tennessee, the most commonly reported birth defect was an atrial septal defect (ASD), a hole or opening in the upper chambers of the heart. Birth defect rates were generally equal for males and females, with the exception of certain conditions which affect only males (i.e., genitourinary defects like hypospadias and congenital posterior urethral valves) or females (i.e. Turner syndrome). Certain types of birth defects, especially chromosomal defects, were more common among babies who were born to mothers 35 years of age and older. Non-Hispanic Black infants had the highest rate of birth defects across maternal racial/ethnic groups, a statistically significant trend that has been consistent over the past ten-year time frame. Higher rates of birth defects were also identified among infants of women with a 12th grade

[5] Confirmed diagnostics include : (i) fetal death cases, (ii) linked infant death cases with maternal information from Tennessee birth statistics file, (iii) linked hospital discharged cases with maternal information from Tennessee birth statistics file. The linkage is essential for confirming that the mother was Tennessee resident at the time of delivery, especially in the case of diagnoses that happened after birth.

education or less and women on Medicaid compared to private insurance. Across the thirteen health department regions, Shelby County had the highest number of birth defects cases and the highest rate per 10,000 resident live births.

Chronic conditions including diabetes, hypertension and obesity were also associated with increased risk of birth defects, particularly for certain types of defects. Infants born to mothers with pre-pregnancy diabetes were approximately four times as likely to be born with a cardiovascular birth defect compared to infants born to mothers without diabetes. These findings underscore the importance of improving health before pregnancy and controlling chronic medical conditions as critical pieces of birth defects prevention.

Data Sources and Limitations

The primary data sources for this report are the Hospital Discharge Data System (HDDS) and the Birth, Death, and Fetal Death Statistical Data Systems, which are compiled, processed and stored by the Office of Population Health Assessment and the Office of Vital Records and Statistics. The HDDS contains admission-level records for all patients treated in Tennessee-licensed hospitals and their outpatient treatment and rehabilitation centers. TNBDSS uses these records to track the 47 major birth defects and fetal alcohol syndrome. Infants' HDDS records containing diagnostic codes corresponding to the tracked birth defects are extracted, compiled, and linked with their birth certificate records. The linkages provide validity checks and add information such as maternal risk factors and demographics that are not available in the HDDS. Diagnostic data are also obtained from the fetal death and death certificate data systems. For fetal death cases, demographic, geographic, and risk factor information are obtained from the fetal death certificate system. Together these sources provide statewide population-based birth defects surveillance for Tennessee.

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. Additional limitations of administrative data systems involve coding. Some of the diagnostic codes used in the HDDS correspond to both the major and minor variants of a given birth defect. The coding system used in the HDDS prior to October 2015, the International Classification of Diseases Revision 9 (ICD-9- CM), prevents distinguishing these differences for certain birth defects. This may have the effect of increasing rates

for some of the more common birth defects, such as atrial septal defect and hypospadias. Less systematically, there are simple coding errors that result in both non-cases being miscoded as having a birth defect and valid cases not being recorded as having a birth defect.

The Tennessee Birth Defects Surveillance System

According to Tennessee Code Annotated § 68-5-506, the Tennessee Department of Health (TDH) is responsible for maintaining "an ongoing program for birth defects monitoring state-wide." The goals of the birth defects registry are to report on incidence, prevalence and trends of birth defects; to provide information about potential environmental hazards associated with birth defects; to evaluate current prevention initiatives; and to provide families of children with birth defects information on public services.

The Tennessee Birth Defects Surveillance System (TNBDSS) utilizes passive surveillance, primarily using data from the Hospital Discharge Data System and the Birth, Death, and Fetal Death Statistical Data Systems. An opportunity to enhance surveillance emerged after Zika virus[6] surfaced as a public health threat in the United States. In 2016, the Tennessee Department of Health (TDH) was awarded an Epidemiology and Laboratory Capacity grant from the Centers for Disease Control and Prevention (CDC), which has supported enhanced surveillance for specific neurologic birth defects that have been associated with Zika virus and connection to care for affected infants and their families. In January 2017, healthcare provider reporting of 23 neurologic birth defects associated with Zika was mandated by the Tennessee Department of Health. All physicians, hospitals, laboratories, healthcare providers, and other persons knowing of or suspecting a reportable disease case are responsible for reporting it to the health department. The list of reportable birth defects and the link to the reporting website can be found in Appendix A. While this funding stream ended in July 2019, TDH continues to collect case reports on these specific neurologic birth defects in order to monitor cases and connect families to services. In May 2021, the

[6] Zika virus infection during pregnancy can cause microcephaly and other neurologic birth defects. To understand more about Zika virus infection, CDC established the US Zika Pregnancy Registry and is collaborating with state, tribal, local, and territorial health departments to collect information about pregnancy and infant outcomes following laboratory evidence of Zika virus infection during pregnancy. The data collected through this registry is used to update recommendations for clinical care, plan for services for pregnant women and families affected by Zika virus, and improve efforts to prevent Zika virus infection during pregnancy.

TNBDSS was awarded another CDC funding opportunity to perform enhanced birth defects surveillance on 26 specific birth defects. Tennessee was one of 10 states to be awarded with the five-year cooperative agreement DD21-2101: Advancing Population-Based Surveillance of Birth Defects.

The goal of this cooperative agreement is to: “strengthen the capacity of birth defects surveillance programs to respond to emerging threats to mothers and babies as a key component of preparedness; identify and address mechanisms contributing to health disparities; and improve health outcomes of affected populations.”

Activities that the TNBDSS will perform under this cooperative agreement include:

- Conduct enhanced surveillance of 26 birth defects using passive case-finding and case agreement with case verification via medical records for selected birth defects
- Implement activities to enhance the timing and accuracy of Critical Congenital Heart Defects diagnoses
- Develop and implement strategies to improve and monitor data quality
- Identify risk factors and disparities
- Assess trends and identify emerging threats
- Develop reports and other products for dissemination to state and community partners
- Assess current and implement new primary and secondary prevention strategies

Furthermore, monitoring birth defects is essential to ensure timely referral to services and enhance care coordination for affected children in Tennessee. TNBDSS will coordinate with the child’s healthcare provider and parent to conduct service referrals to Children’s Special Services (CSS), Tennessee Early Intervention Services (TEIS), and Family Voices, as needed. TNBDSS will also work to identify any gaps in supportive services for families of children with a birth defect and augment service referrals as appropriate.

The CSS program provides resources for medical and non-medical services for children with physical disabilities and special health care needs from birth to 21 years of age if certain diagnostic and financial eligibility criteria are met by the family in need. TEIS is a voluntary educational program for families with children from birth through two years old with disabilities or developmental delays that supports families

in promoting their child's optimal development, facilitates the child's participation in family and community activities, and encourages the active participation of families by embedding strategies into family routines. Family Voices of Tennessee, a program of the Tennessee Disability Coalition, provides emotional and educational support to the families of children with special healthcare needs, chronic illnesses or disabilities.

During the period 2014-2018, there was an average of 577 infant deaths per year (Table 1). The overall infant mortality rate for Tennessee increased from 2014-2016 and maintained at the elevated level in 2017 before decreasing in 2018; the decrease was not statistically significant.

Infant deaths do not impact all races equally. Non-Hispanic Black infants accounted for one in five (20%) of the total live births in Tennessee from 2014-2018, but about one in three (34%) of the infant deaths. The infant mortality rate for non-Hispanic Black infants was approximately twice that of non-Hispanic White infants over this time frame. Hispanic infants accounted for 9% of live births in Tennessee and just under 7% of infant deaths. The infant mortality rate for Hispanic infants was more comparable to that seen for non-Hispanic White infants and was actually the lowest of the three racial/ethnic groups shown for the period from 2014-2016, before increasing in 2017. The Tennessee trends mirror the disparities seen nationally, with infants of non-Hispanic Black women having the highest mortality rate.[7]

Table 1. Infant Deaths by Maternal Race/Ethnicity. Tennessee, 2014-2018

Year	Total		Non-Hispanic White		Non-Hispanic Black		Hispanic	
	Number	Rate	Number	Rate	Number	Rate	Number	Rate
2014	562	6.9	297	5.4	206	12.5	28	4.0
2015	569	7.0	325	6.0	178	11.1	33	4.5
2016	597	7.4	340	6.4	194	12.4	38	5.0
2017	597	7.4	316	6.0	204	12.9	50	6.5
2018	559	7.4	287	5.4	194	12.3	48	6.2

1. Rate per 1,000 Tennessee resident live births.
Note: Race/ethnicity categories do not sum to total as other and unknown categories are not shown.
Data Source: Tennessee Department of Health, Office of Vital Records and Statistics.

[7] Ely DM, Driscoll AK. Infant mortality in the United States, 2018: Data from the period linked birth/infant death file. National Vital Statistics Reports, vol 69 no 7. Hyattsville, MD: National Center for Health Statistics. 2020.

Table 2 shows the ten leading causes of infant deaths in Tennessee between 2014 and 2018. Birth defects were the leading cause of infant death (21%), followed by preterm birth/low birthweight (14%).

Table 2. Leading Causes of Infant Death, Tennessee, 2014-2018

Rank	Cause of Death	Number of Deaths	Percent of Deaths
1	Birth defects	618	21
2	Preterm birth and low birthweight	405	14
3	Accidents (unintentional injuries)	215	7
4	Sudden infant death syndrome (SIDS)	139	5
5	Maternal complications of pregnancy	100	3
6	Complications of placenta, cord, and membranes	88	3
7	Bacterial sepsis of newborn	75	3
8	Atelectasis (partial lung collapse)	70	2
9	Necrotizing enterocolitis of newborn	63	2
10	Diseases of the circulatory system	55	2
--	All other causes	1056	37
--	*All Causes*	*2884*	*100*

Data Source: Tennessee Department of Health, Office of Vital Records and Statistics.

Table 3 examines the top two causes of infant death more closely. Among infants whose cause of death was a birth defect, 56% were also born preterm. Preterm delivery often exacerbates the medical complications faced by infants born with birth defects. For some defects, infants born preterm may have greater risk of mortality compared to their counterparts delivered at term.[8] This pattern of preterm infants experiencing increased mortality has been demonstrated for neural tube defects,[9] congenital diaphragmatic hernia,[10] and heart defects.[11]

Table 3. Co-Occurrence of Two Leading Causes of Infant Death, Tennessee, 2014-2018

Causes of Infant Death[1]		
	Born Preterm (<37 weeks)	Born Full Term (37+ weeks)
Birth Defect	56%	44%
	Major Birth Defect Present	No Major Birth Defect Present
Preterm birth and low birthweight	4%	96%

1. Represents underlying cause of death recorded on infant's death certificate

Data Source: Tennessee Department of Health, Office of Vital Records and Statistics; Tennessee Birth Defects Registry.

[8] Honein MA, Kirby RS, Meyer RE, Xing J, Skerrette NI, Yiskiv N, et al. The association between major birth defects and preterm birth. Matern Child Health J. 2009;13:164–75.

[9] Davidoff MJ, Petrini J, Damus K, Russell RB, Mattison, D. Neural tube defect-specific infant mortality in the United States. Teratology. 2002; 66(Suppl 1): S17–S22.

[10] Cannon C, Dildy GA, Ward R, Varner MW, Dudley DJ. A population-based study of congenital diaphragmatic hernia in Utah: 1988–1994. Obstet Gynecol.1996; 87(6): 959–963.

[11] Tanner K, Sabrine N, Wren C. Cardiovascular malformations among preterm infants. Pediatrics. 2005; 116(6): e833–e838.

Table 4 provides more detail about the gestational ages of infants who died due to birth defects. Among the infants who died due to birth defects, 44% were full-term (37+ weeks) and 27% were late-preterm (34 to <37 weeks).

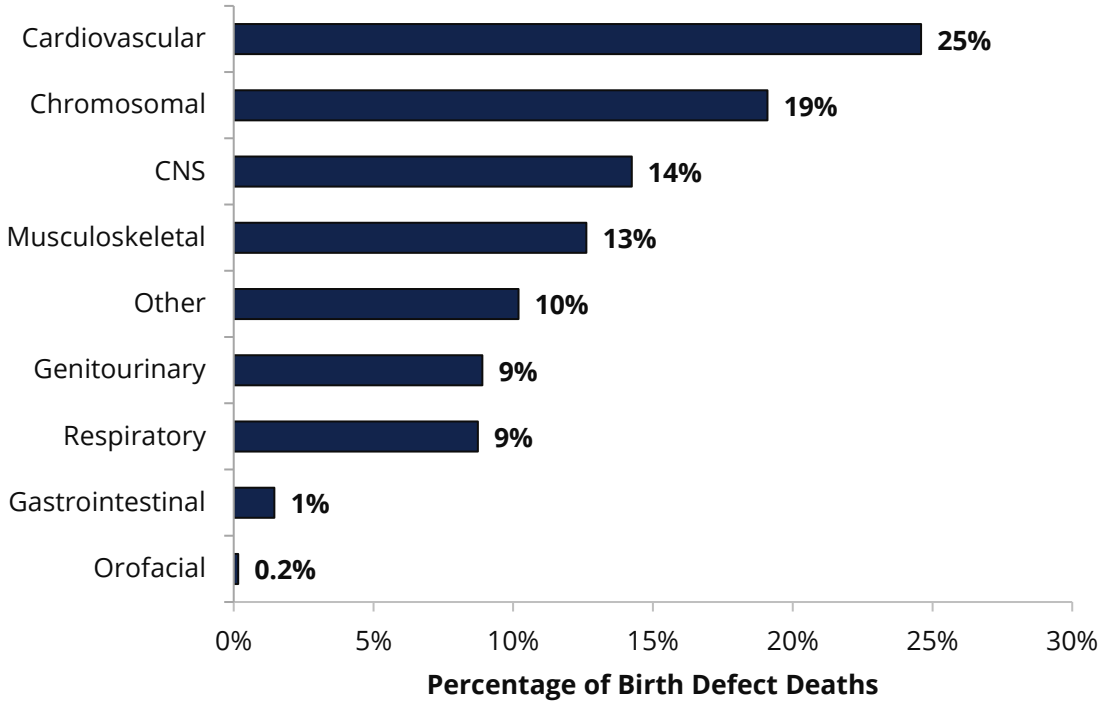
Table 4. Gestational age at birth of infant deaths caused by birth defects, Tennessee, 2014-2018

Gestational age at Birth	Percent of Cases
<28 weeks	10
28-<32 weeks	11
32-<34 weeks	9
34-<37 weeks	27
37+ weeks	44

Data Source: Tennessee Department of Health, Office of Vital Records and Statistics.

Figure 1 demonstrates birth defect deaths by the type of defect. Cardiovascular defects were the leading cause of birth defect deaths (25% of birth defect deaths), followed by chromosomal (19%) and central nervous system (14%) defects.

Figure 1. Birth Defect Deaths by Type of Defect, Tennessee, 2014-2018



Data Source: Tennessee Department of Health, Office of Vital Records and Statistics; Tennessee Birth Defects Registry.

Figure 2 provides more information about the age at death for infants who died due to birth defects. Over a third (36%) died within 24 hours of birth. A smaller proportion (30%) died later in the year, between 28 and 364 days.

Figure 2. Infant Deaths Due to Birth Defects by Age at Death, Tennessee, 2014-2018

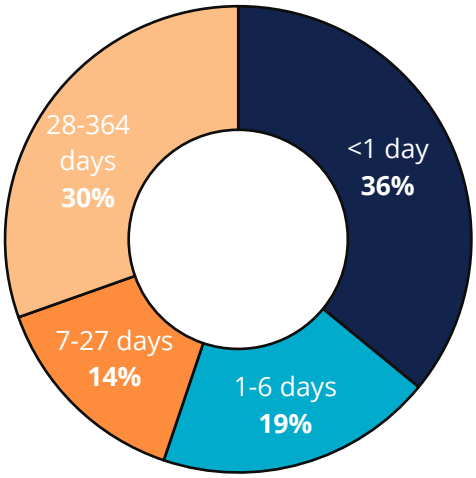
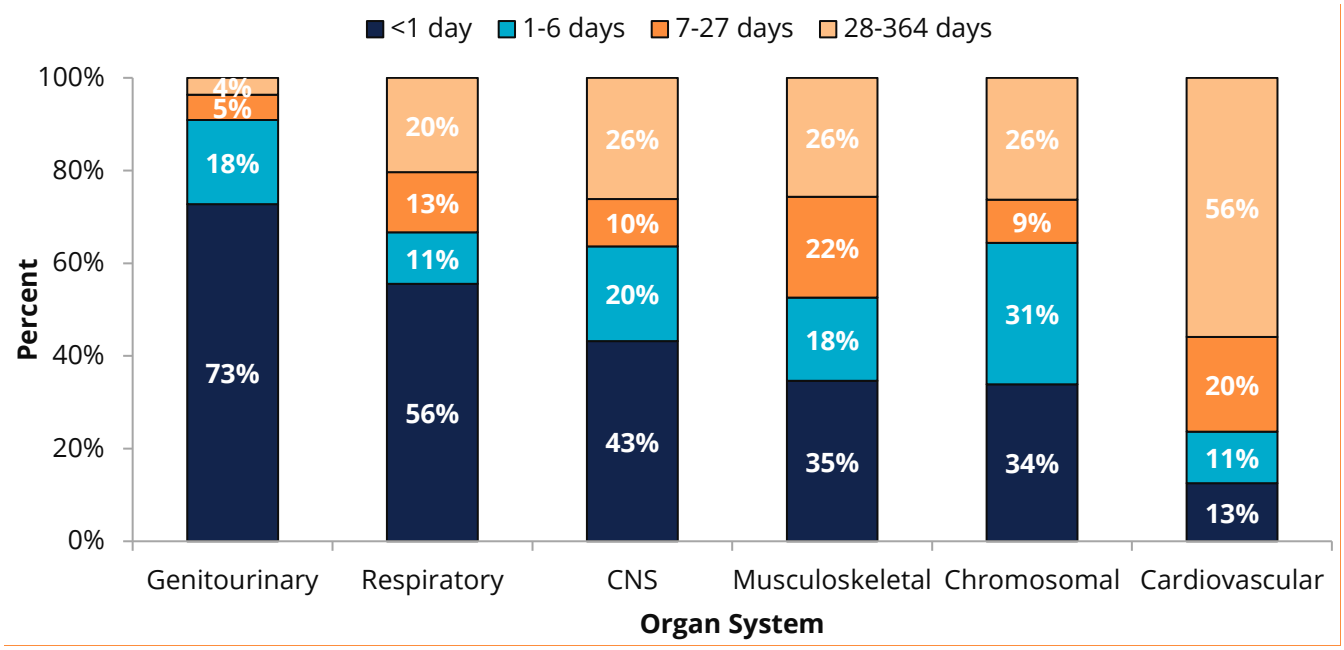


Figure 3 shows the variation in age at death across the types of birth defects. The vast majority (73%) of infant deaths due to genitourinary defects occurred within the first day, with just 4% involving older infants aged 28-364 days. Cardiovascular deaths were on the other end of the spectrum: most cases were older infants aged 28-364 days and just 13% occurred during the first day of life.

Figure 3. Infant Deaths Due to Birth Defects by Age at Death and Type, Tennessee, 2014-2018



Data Source: Tennessee Department of Health, Office of Vital Records and Statistics; Tennessee Birth Defects Registry.

Figures 4 and 5 depict the number and rate of infant deaths due to birth defects by health department region. Shelby County had the highest number of deaths (accounting for 19% of total deaths) and the second highest statewide rate. Madison County had the highest rate, but had a relatively low number of deaths, accounting for just 3% of Tennessee infant deaths due to birth defects. Shelby County and the Mid-Cumberland region have consistently had the highest annual number of birth defects deaths, while Madison and Shelby counties have historically had the highest rates. The rate for Sullivan County is suppressed as rates based on counts less than 11 are statistically unreliable.

Figure 4. Number Infant Deaths Due to Birth Defects by Region, Tennessee, 2014-2018

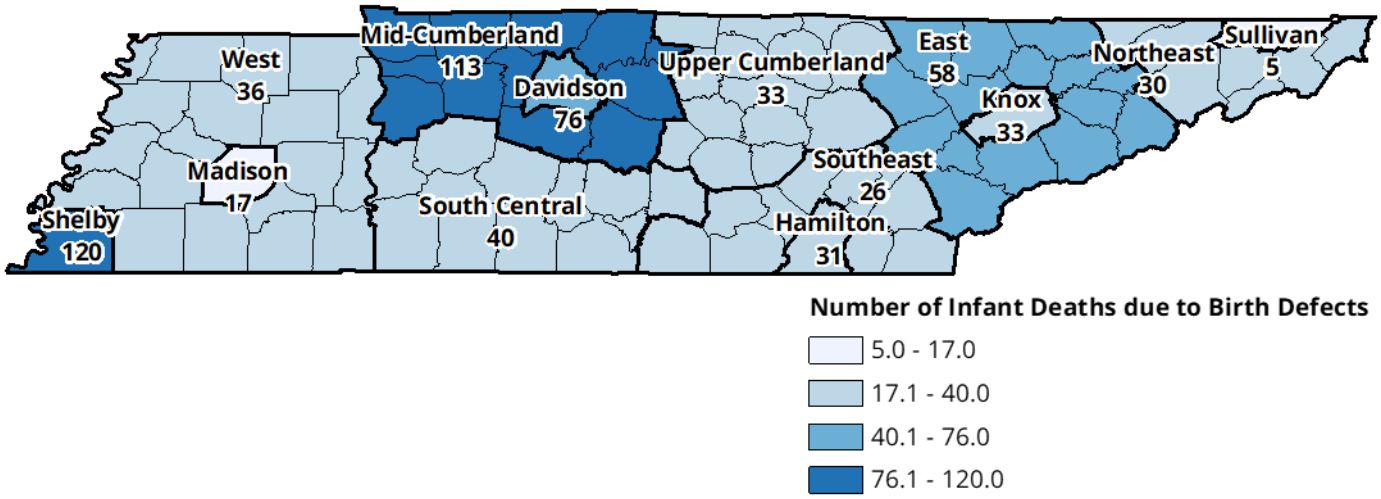
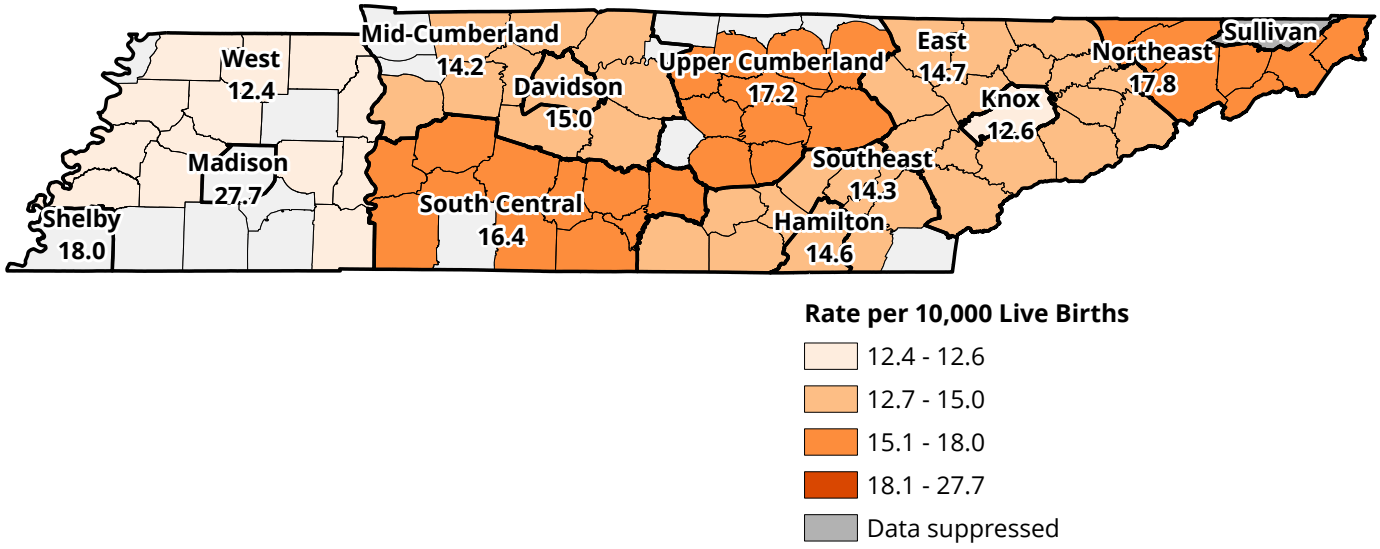


Figure 5. Rate Infant Deaths Due to Birth Defects by Region, Tennessee, 2014-2018



Data Source: Tennessee Department of Health, Office of Vital Records and Statistics; Tennessee Birth Defects Registry.

Birth Defects Prevalence in Tennessee

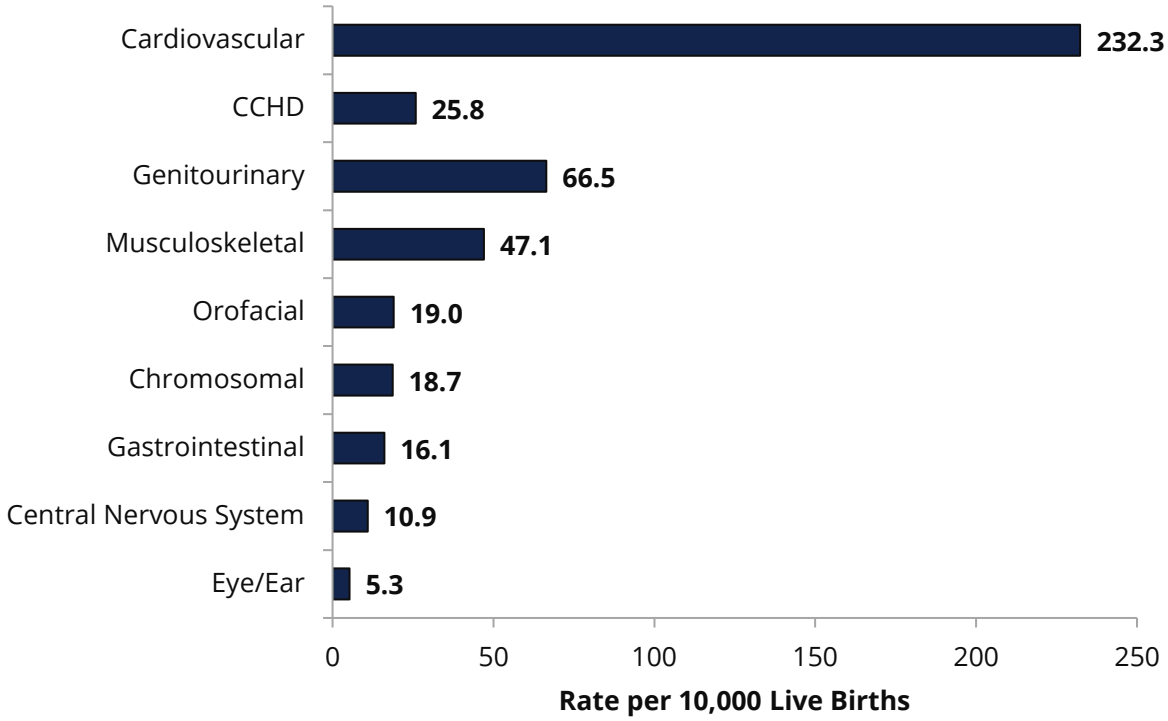
Table 7 in Appendix B shows the case numbers and rates for the 47 major birth defects by organ system and fetal alcohol syndrome. Between January 2014 and December 2018, there were 14,874 Tennessee babies diagnosed with birth defects. In addition, there were 40 infants identified with fetal alcohol syndrome during this time period. Because a baby may be diagnosed with more than one birth defect, the number of confirmed diagnosed birth defects (19,727) over this time period is higher. Out of the 19,727 defects, 12,109 were cardiovascular defects, which represent 61% of the total. The genitourinary system, with 2,714 defects, is the second most-affected organ system (almost 14% of total defects). The most common single birth defect in Tennessee is atrial septal defect with a total of 8,046 cases from 2014-2018, followed by hypospadias (n=2,263) and ventricular septal defect (n=2,006). By identifying the most common birth defects and most affected organ systems, targeted prevention efforts can be developed based on known risk factors for particular birth defects. TNBDSS also strives to identify conditions that are common in Tennessee relative to the nation as a whole, though this comparison is limited by the significant lag in the availability of nationwide estimates and the differences in surveillance methodology used across the states. Because of these limitations, the nationwide estimates are not provided for comparison within this report, though the most national recent data for the time frame 2010-2014 are available.[12]

Prevalence of Major Birth Defects by Organ System

Figure 6 shows the prevalence of birth defects by organ system. Cardiovascular system defects are the most commonly diagnosed birth defects in Tennessee, with an overall rate of 232.3 infants with a cardiovascular defect per 10,000 live births.

[12] Mai CT, Isenburg JL, Canfield MA, Meyer RE, Correa A, Alverson CJ, Lupo PJ, Riehle-Colarusso T, Cho SJ, Aggarwal D, Kirby RS; National Birth Defects Prevention Network. National population-based estimates for major birth defects, 2010-2014. Birth Defects Res. 2019 Nov 1;111(18):1420-1435.

Figure 6. Prevalence of Major Birth Defects by Organ System, Tennessee, 2014-2018



Data Source: Tennessee Department of Health, Tennessee Birth Defects Registry.

As seen in Figure 7, the most common cardiovascular birth defects are atrial septal defects (ASD) and ventricular septal defects. Of the 9,420 total infants with at least one cardiovascular defect, 84% had an ASD or VSD as their only cardiovascular diagnosis. An ASD is a hole in the wall (septum) that divides the two upper chambers of the heart.[13] ASDs often spontaneously resolve during infancy or early childhood.[14] A VSD is a hole in the septum that separates the two lower chambers (ventricles) of the heart.[15] VSDs can be classified by the size of the hole in the septum (small, medium, or large); the size of the defect influences which signs and symptoms, if any, are present.[16] Most small VSDs spontaneously close during the first two years of life.

[13] National Center on Birth Defects and Developmental Disabilities, Centers for Disease Control and Prevention. Facts about Atrial Septal Defect. https://www.cdc.gov/ncbddd/heartdefects/atrialseptaldefect.html. Updated November 12, 2019. Accessed September 20, 2020.

[14] Vick GW, Bezold LI. Isolated atrial septal defects (ASDs) in children: Classification, clinical features, and diagnosis. In: UpToDate, Post, TW (Ed), UpToDate, Waltham, MA, 2019.

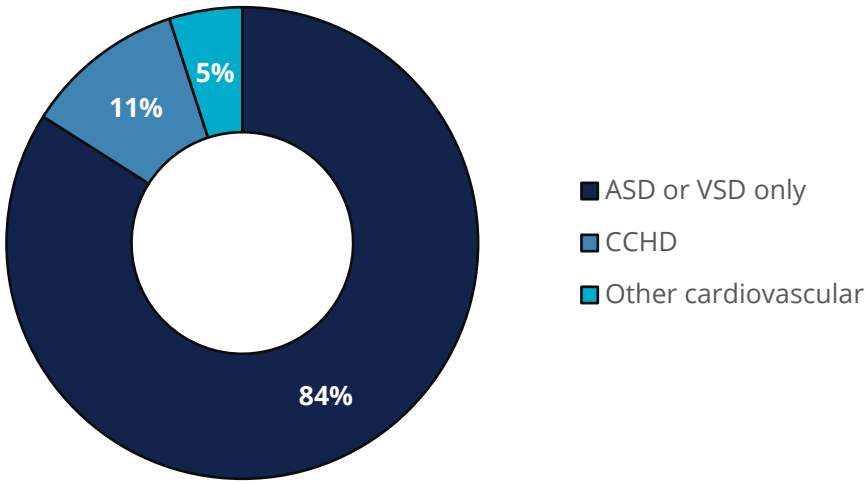
[15] Fulton DR, Saleeb S. Isolated ventricular septal defects in infants and children: Anatomy, clinical features, and diagnosis. In: UpToDate, Post, TW (Ed), UpToDate, Waltham, MA, 2019.

[16] National Center on Birth Defects and Developmental Disabilities, Centers for Disease Control and Prevention. Facts about Ventricular SeptalDefect.https://www.cdc.gov/ncbddd/heartdefects/ventricularseptaldefect.html. Updated November 12, 2019.

However, babies with large VSDs may have symptoms, such as shortness of breath, fast breathing, sweating, tiredness while feeding, or poor weight gain. As many ASDs and VSDs spontaneously close and are often less serious in nature than the other cardiovascular conditions monitored by TNBDSS, these defects are presented separately in Figure 7.

As also shown in Figure 7, 11% of the total cardiovascular cases identified from 2014-2018 were diagnosed with at least one condition classified as a critical congenital heart defect (CCHD). The twelve conditions included in this group (listed in Table 7) cause serious, life-threatening symptoms which require intervention within the first days or first year of life. The remaining 5% of cardiovascular cases had defects other than ASD or VSD that were not one of the twelve CCHDs. These included aortic valve stenosis and atrioventricular septal defects.

Figure 7. Total Cardiovascular Birth Defect Cases by Type of Condition, Tennessee, 2014-2018



Data Source: Tennessee Department of Health, Tennessee Birth Defects Registry.

Birth Defects by Socio-Demographic Factors

When examining the prevalence of birth defects, it is important to consider maternal socio-demographic and health factors. Advanced maternal age is a risk factor for certain birth defects. In addition, there are racial and ethnic differences in the occurrence of certain birth defects.[17] Some health behaviors (such as smoking, alcohol use, and drug use) and health conditions (such as diabetes and hypertension) are also associated with an increased risk of specific birth defects.[18]

In Tennessee, babies born to women 35 years of age and older, women with ≤ 12th grade education, and women on Medicaid have relatively high rates of birth defects. There are important racial/ethnic and geographical differences as well. Birth defect prevalence rates are highest for Non-Hispanic Blacks and for those living in Shelby County. Identifying these at-risk groups in Tennessee allows for the development of targeted prevention efforts, with the goal of reducing birth defects.

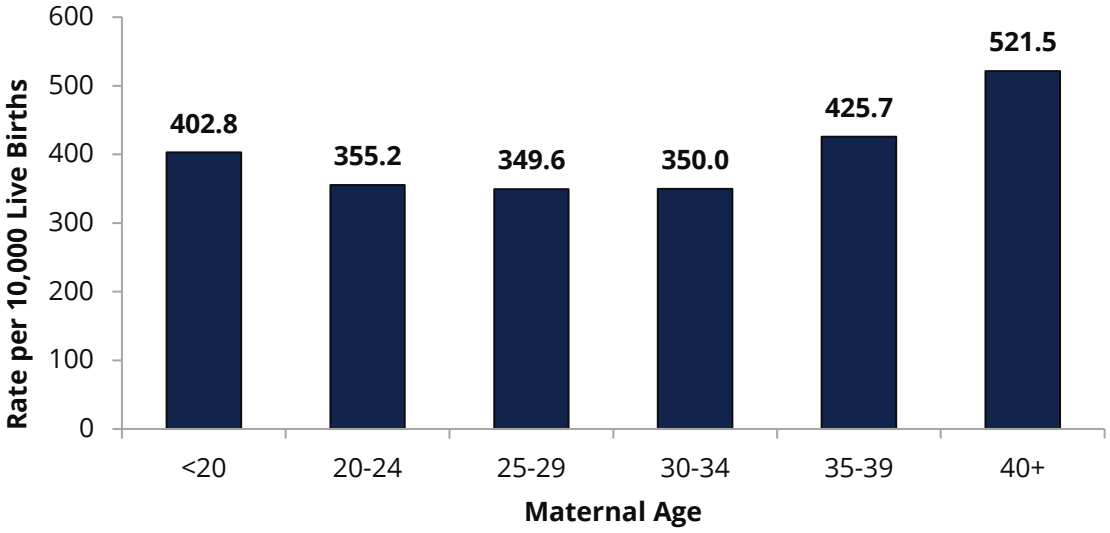
Maternal Age

Maternal age is a significant risk factor for some types of birth defects, with advanced maternal age (defined as 35 years old or older at the time of delivery) posing a higher risk for birth defects such as Trisomy 21 (Down Syndrome).[19] In contrast, women younger than 20 years old are more likely to have babies born with gastroschisis,[20] a birth defect of the abdominal wall, than older women.

Figure 8 shows the overall prevalence of birth defects by maternal age group in Tennessee. From 2014-2018, the rate of birth defects was highest among women 40 and older (521.5 per 10,000 live births), followed by women aged 35-39 (425.7 per 10,000 live births) and women less than 20 years old (402.8 per 10,000 live births).

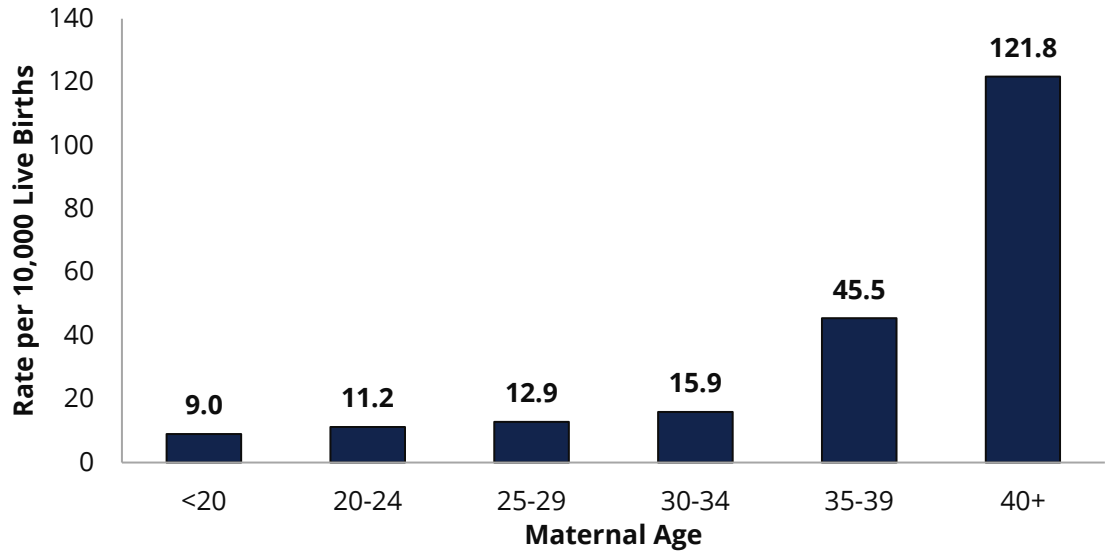
[17] Canfield MA, Mai CT, Wang Y, O'Halloran M, Marengo LK, Olney RS, et al. The Association Between Race/Ethnicity and Major Birth Defects in the United States, 1999 - 2007. Am J Public Health. 2014;104(9): e14–e23.
[18] National Center on Birth Defects and Developmental Disabilities, Centers for Disease Control and Prevention. Commit to Healthy Choices to Help Prevent Birth Defects. https://www.cdc.gov/ncbddd/birthdefects/prevention.html. Updated on December 5, 2019. Accessed on September 20, 2020.
[19] Allen EG, Freeman SB, Druschel C, Hobbs CA, O'Leary LA, Romitti, PA, et al. Maternal age and risk for trisomy 21 assessed by the origin of chromosome nondisjunction: a report from the Atlanta and National Down Syndrome Projects. Hum Genet. 2009;125(1):41-52.
[20] Jones AM, Isenburg J, Salemi JL, Arnold KE, Mai CT, Aggarwal D, et al. Increasing Prevalence of Gastroschisis—14 States, 1995-2012. MMWR morb Mortal Wkly Rep. 2016;65(2):23-26.

Figure 8. Rate of Birth Defects by Maternal Age, Tennessee, 2014-2018



Figures 9 and 10 further illustrate the significant role that maternal age plays in birth defect occurrence. Figure 9 demonstrates that infants born to mothers 40 years of age and older are more likely to have a chromosomal birth defect than those born to mothers in the other age groups. In this figure, chromosomal birth defects include Trisomy 21 and Trisomy 13, which are known to be associated with advanced maternal age, as well as Deletion 22q11.2 and Turner Syndrome, which are not traditionally associated with advanced maternal age.

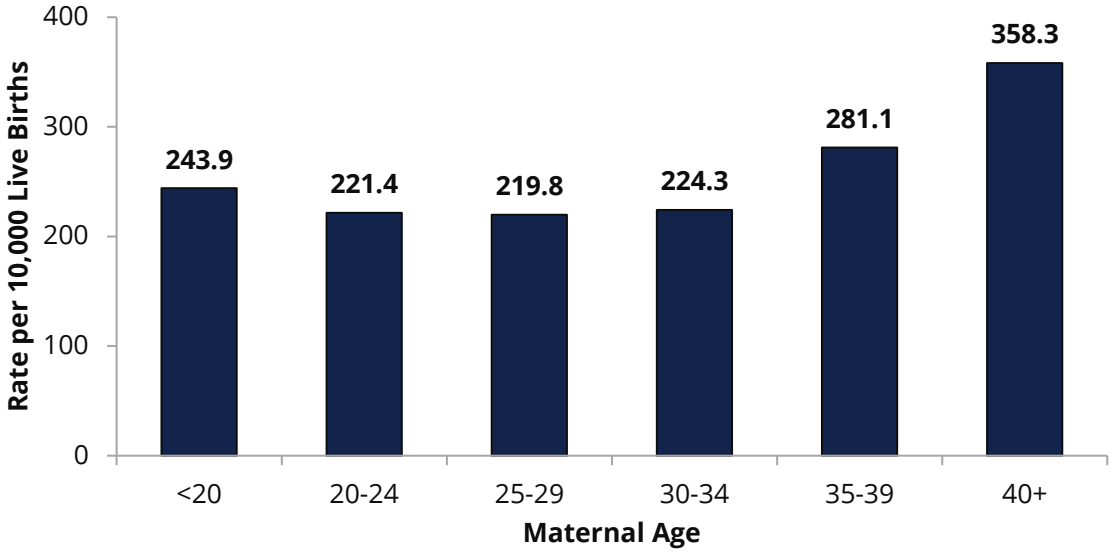
Figure 9. Rate of Chromosomal Birth Defects by Maternal Age, Tennessee, 2014-2018



Data Source: Tennessee Department of Health, Tennessee Birth Defects Registry.

Figure 10 illustrates that the rate of cardiovascular birth defects is higher for infants born to mothers aged 35 years and older as compared to infants born to younger mothers. The rate of cardiovascular birth defects for infants born to mothers 40 years of age and older (358.3 per 10,000 live births) was 1.6 times the rate for infants born to mothers in the age group 25-29 (219.8 per 10,000 live births).

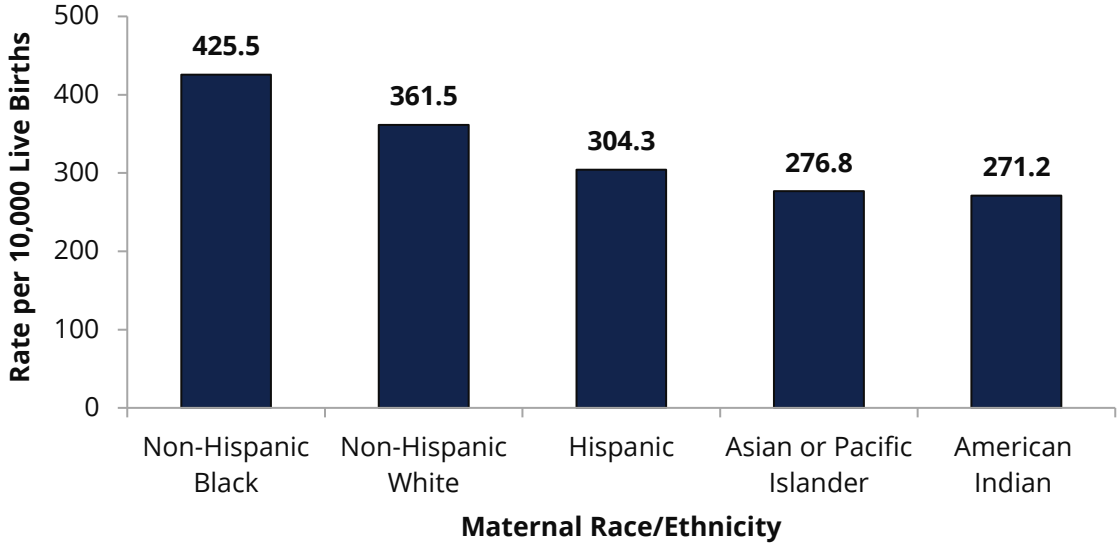
Figure 10. Rate of Cardiovascular Birth Defects by Maternal Age, Tennessee, 2014-2018



Maternal Race and Ethnicity

Birth defects rates are highest for non-Hispanic Blacks infants (425.5 per 10,000 live births), followed by non-Hispanic Whites and Hispanic infants (Figure 11).

Figure 11. Rate of Birth Defects by Maternal Race/Ethnicity, Tennessee, 2014-2018

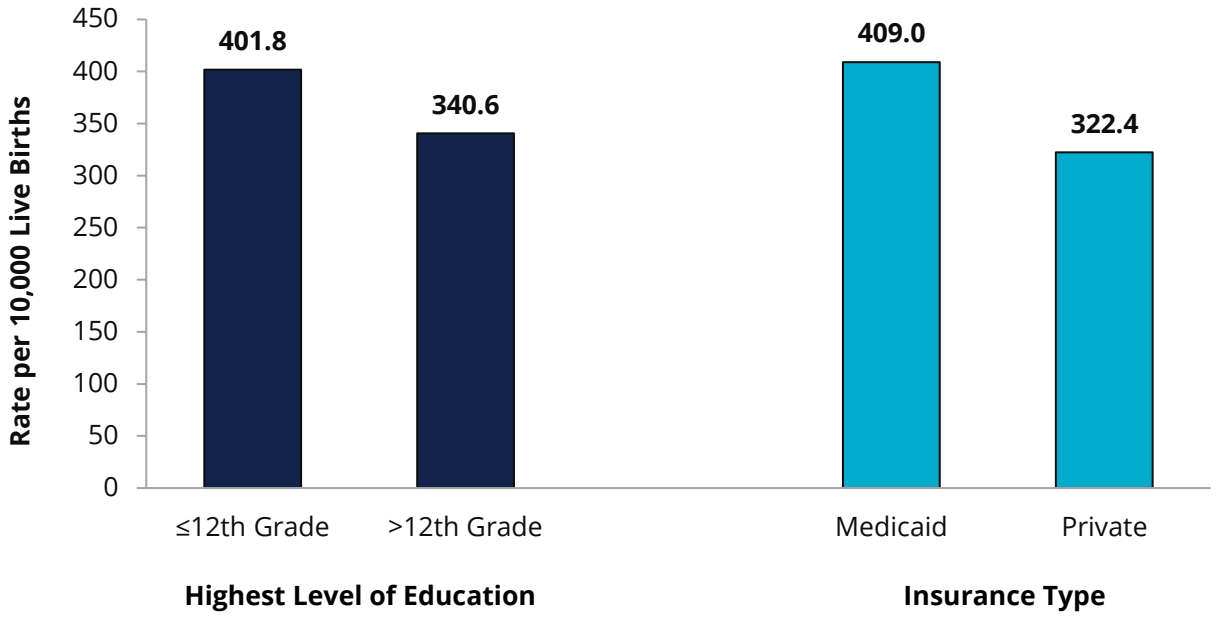


Data Source: Tennessee Department of Health, Tennessee Birth Defects Registry.

Other Maternal Characteristics

Birth defect prevalence rates also differed by education and insurance type. Figure 12 shows that birth defect prevalence was significantly higher among babies born to women with ≤ 12 grade education and women on Medicaid. These trends have remained consistent over time, with almost no change in the demonstrated disparities over the past ten-year time frame. The differences seen across these groups highlight the influence of social determinants of health (such as education and income levels) on health outcomes.

Figure 12. Rate of Birth Defects by Maternal Educational Attainment and Insurance Status, Tennessee, 2014-2018



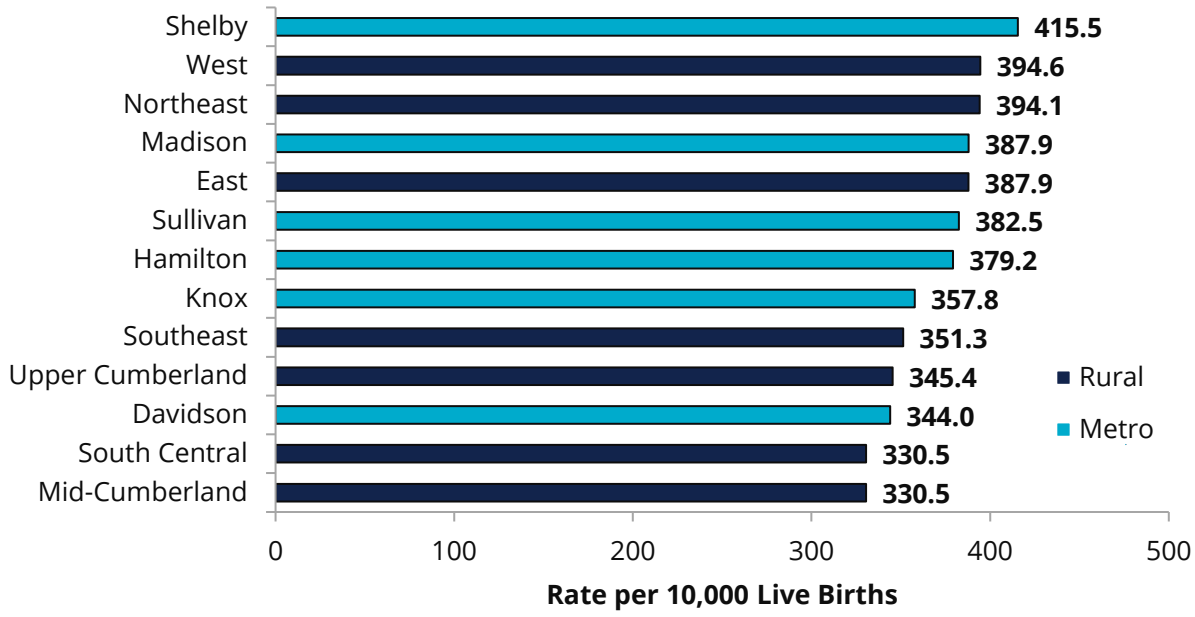
Data Source: Tennessee Department of Health, Tennessee Birth Defects Registry.

Birth Defects by Maternal Region of Residence

Birth defects prevalence rates also differ by mother's residence. Figure 13 shows that birth defects prevalence rates are highest for those living in Shelby County, followed by the West and Northeast regions. Historically, the Northeast region and neighboring Sullivan County had the highest rates of birth defects in Tennessee. In 2011, the Northeast region's rate of birth defects was significantly higher than that seen for any other region, and 1.6 times the rate for Tennessee overall. Most recently, however, the rates for the regions in the Western part of the state (Shelby County, the West region, and Madison County) have increased and begun to outrank the Northeastern regions as the most affected areas in Tennessee. The regions of Middle Tennessee (Mid-Cumberland, South Central, Davidson County, and Upper Cumberland) have consistently had the lowest rates of birth defects in the state.

These differences in birth defects prevalence may reflect the underlying variation in the pervasiveness of adverse social determinants of health seen across the state. Most recent census estimates based on 2015-2019 data place the percent of people living in poverty at 17.2% for Shelby and 17.6% for Madison, compared to 12.6% for Davidson. Residents of Davidson were also more likely to hold a Bachelor's degree or higher: 41.7% of adults in Davidson had a Bachelor's degree, compared to 31.6% for Shelby and 25.6% for Madison. These data provide important context when interpreting the below ranking and designing prevention strategies.

Figure 13. Rate of Birth Defects by Maternal Region of Residence, Tennessee, 2014-2018



Data Source: Tennessee Department of Health, Tennessee Birth Defects Registry.

Birth Defects by Maternal County of Residence

Figures 14 and 15 demonstrate the number and rate of birth defect cases by county. The highest case numbers were for the metro counties of Shelby (2,773 cases) and Davidson (1,743 cases). Figure 15 shows several areas with multiple high-rate counties clustered together including in the East (Morgan, Anderson, Union, and Campbell) and the Upper Cumberland regions (Pickett, Clay, and Jackson). Note that many of the high-rate counties have low case numbers. Pickett, the county with the highest rate, had only 13 cases in the five-year time frame. The rate for Moore County is suppressed as rates based on counts less than 11 are statistically unreliable.

Figure 14. Number of Birth Defect Cases by County, Tennessee, 2014-2018

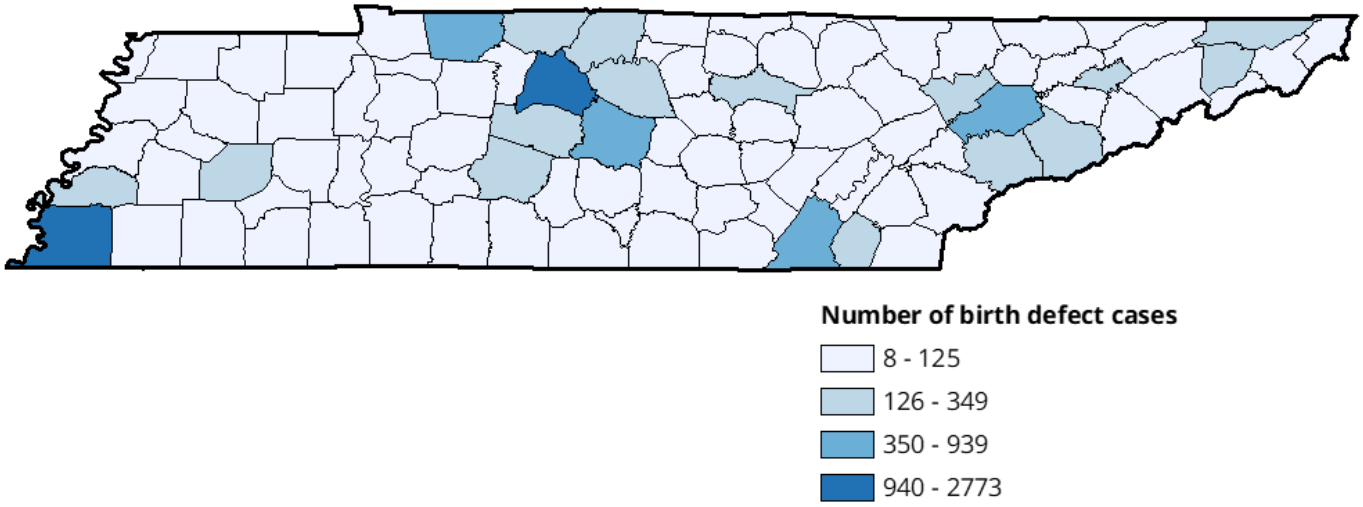
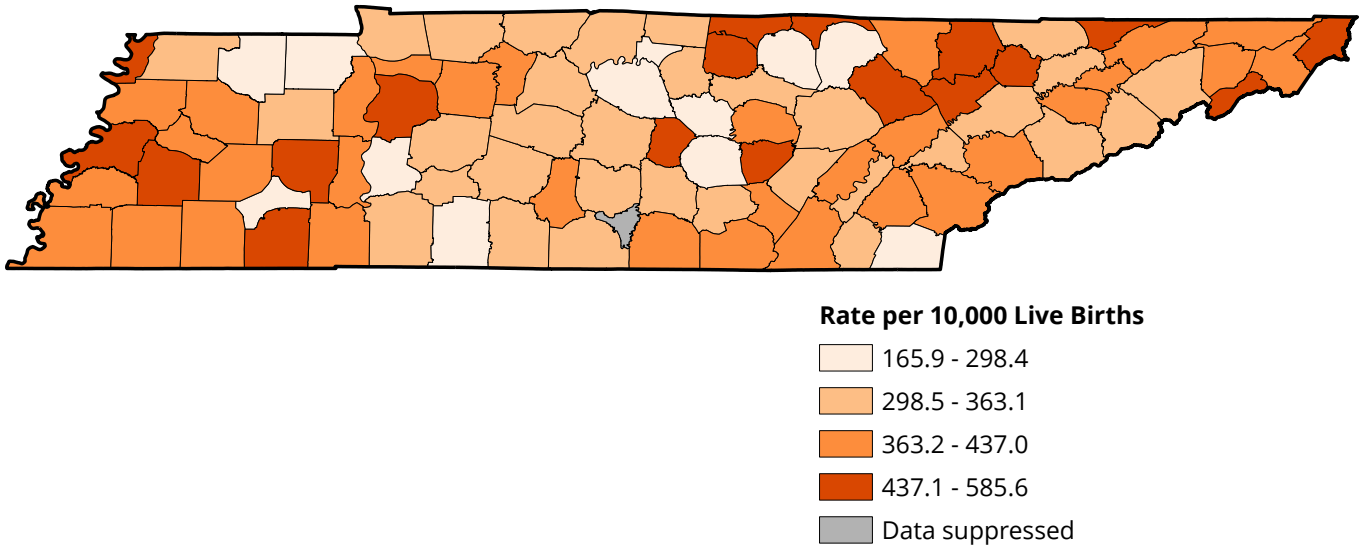


Figure 14. Number of Birth Defect Cases by County, Tennessee, 2014-2018



Data Source: Tennessee Department of Health, Tennessee Birth Defects Registry.

Maternal Health Factors

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. By identifying and analyzing these risk factors, targeted prevention efforts can be developed. Table 5 illustrates the relative risk of birth defects by maternal health factors: diabetes, hypertension (high blood pressure), smoking during pregnancy, body mass index (BMI), and level of prenatal care (based on the Kotelchuck index, which combines the timing of initiation of prenatal care with the number of prenatal visits). Infants born to mothers with pre-pregnancy diabetes had more than 3.0 times the risk of birth defects compared to infants born to mothers without diabetes. In addition, infants born to mothers with gestational diabetes had about 1.4 times the risk of birth defects compared to infants born to mothers without diabetes. Table 6 demonstrates the rate of birth defects by organ system in relation to the maternal diabetes status (no diabetes, pre-pregnancy diabetes, or gestational diabetes).

Maternal hypertension (pre-existing and gestational) was also associated with increased risk of birth defects. Infants born to mothers with pre-pregnancy hypertension had 1.9 times the risk of birth defects, while infants born to mothers with gestational hypertension had 1.4 times the risk of birth defects compared to infants born to mothers without hypertension.

Smoking during pregnancy was also associated with increased risk of birth defects; infants born to mothers who smoked during pregnancy had 1.2 times the risk of birth defects compared to infants born to mothers who did not smoke during pregnancy. An abnormal BMI (underweight, overweight or obese) was also associated with increased risk of birth defects; the highest relative risk of birth defects in this category was for infants born to mothers who were obese, compared to mothers with normal BMI. Finally, inadequate prenatal care was associated with increased risk of birth defects. Infants born to mothers with inadequate or intermediate prenatal care had 1.6 and 1.2 times the risk of birth defects, respectively, compared to mothers who received adequate prenatal care. All of these reported associations between maternal health factors and birth defects were found to be statistically significant, meaning that the differences are larger than would be expected by chance alone. It is important to

note, however, that Table 5 presents crude relative risks not adjusted for other factors such as maternal age and race that could contribute to the observed associations.

Table 5. Relative Risk of Birth Defects by Maternal Health Characteristics, Tennessee, 2014-2018[1]

Maternal Health Characteristic		Relative Risk of Birth Defects[2]	95% CI[3]
Diabetes	Pre-existing	3.03	2.79-3.29
	Gestational	1.37	1.30-1.46
	None	Reference	–
Hypertension	Pre-existing	1.89	1.76-2.03
	Gestational	1.42	1.34-1.50
	None	Reference	–
Pregnancy Smoking Status[4]	Smoker	1.24	1.18-1.29
	Non-Smoker	Reference	–
BMI	Underweight	1.17	1.08-1.27
	Normal	Reference	–
	Overweight	1.08	1.04-1.13
	Obese	1.31	1.26-1.36
Prenatal Care[5]	Inadequate	1.56	1.49-1.65
	Intermediate	1.15	1.07-1.23
	Adequate	Reference	–

1. Relative risks shown are crude, meaning they are not adjusted for any other factors that could contribute to the observed associations.
2. Compares the risk of birth defects in group exposed to a given maternal characteristic with the risk in the reference group for that category. Data interpretation example: infants born to mothers with pre- pregnancy diabetes had 3 times the risk of birth defects compared to infants born to mothers with no diabetes.
3. Can be interpreted as range that we are 95% confident contains the true relative risk for the population. Data interpretation example: we are 95% confident that the relative risk of birth defects in infants born to mothers with pre-pregnancy diabetes compared to infants born to mothers with no diabetes is between 2.79 and 3.29. Note that where 95% confidence interval does not include 1 (every instance shown), the difference is statistically significant.
4. Smokers defined as women who smoked during any trimester of pregnancy.
5. Prenatal care categories based on the Kotelchuck index, which combines the timing of initiation of prenatal care with the number of prenatal visits (adjusted for gestational age).

Data Source: Tennessee Department of Health, Tennessee Birth Defects Registry.

Table 6. Prevalence of Major Birth Defects by Organ System for Infants Born to Mothers with Pre- Pregnancy Diabetes, Mothers with Gestational Diabetes, and Mother with No Diabetes, 2014-2018.

Organ System	Pre-Pregnancy Diabetes			Gestational Diabetes			No Diabetes		
	Cases	Rate[1]	Relative Rate[2]	Cases	Rate	Relative Rate	Cases	Rate	Relative Rate
Cardiovascular	417	846.0	3.9	802	338.7	1.6	8198	217.5	Reference
CCHD	41	83.2	3.4	81	34.2	1.4	925	24.5	Reference
CNS	17	34.5	3.2	25	10.6	1.0	400	10.6	Reference
Gastrointestinal	21	42.6	2.8	49	20.7	1.3	582	15.4	Reference
Genitourinary	58	117.7	1.8	181	76.4	1.2	2456	65.2	Reference
Musculoskeletal	43	87.2	1.9	115	48.6	1.0	1749	46.4	Reference
Orofacial	23	46.7	2.5	41	17.3	0.9	708	18.8	Reference

1. Rate per 10,000 live births.
2. Compares the rate of birth defects for a given organ system amongst infants born to mothers with pre-pregnancy diabetes (or gestational diabetes) to the rate amongst infants born to mothers with no diabetes. Relative risks shown are crude, meaning they are not adjusted for any other factors that could contribute to the observed associations.
3. Excludes cases of atrial septal defect and ventricular septal defect.
Data Source: Tennessee Department of Health, Tennessee Birth Defects Registry.

Other Risk Factors

Although the causal mechanisms of most birth defects are not fully understood, there are known risk factors that increase the likelihood of giving birth to a baby with a birth defect. Drinking alcohol and smoking cigarettes during pregnancy are associated with increased risk of having a baby born with a birth defect. Babies born to mothers who smoke cigarettes are more likely to be born premature and low birth weight. They are also more likely to be born with cardiovascular, orofacial, gastrointestinal, and musculoskeletal birth defects. There is no amount of alcohol that is safe to drink during pregnancy. When a pregnant woman drinks, the alcohol in her system passes from mother to baby. Drinking can also cause fetal alcohol syndrome, which is a serious condition involving growth deficiencies, facial abnormalities, central nervous system impairment, and intellectual disabilities.

Some infections that a woman can get during pregnancy can be harmful to the developing baby and can even cause birth defects. For example, Zika virus infection during pregnancy can cause microcephaly and other neurologic birth defects. Fetal exposure to rubella, a vaccine preventable illness, increases the risk of a baby being born with congenital rubella syndrome, which affects the ear/eye and cardiovascular systems. Toxoplasmosis is caused by the parasite, Toxoplasma gondii. Babies born to women with a toxoplasmosis infection are at risk for hydrocephalus which affects the central nervous system. Likewise, babies born following in utero exposure to cytomegalovirus (CMV) may have long-term health problems, such as hearing loss, vision loss, microcephaly, seizures and developmental delay.

Although not all birth defects can be prevented, avoiding the known risk factors, managing chronic medical conditions, and committing to healthy choices can increase a woman's chance of having a healthy baby. Since most of the baby’s vital organs and systems are formed in the first four to eight weeks of gestation, (often before a woman knows she is pregnant), the best time to start preventing birth defects is before a woman becomes pregnant.

Occupational and environmental exposures such as radiation, certain chemicals, and strenuous physical labor may harm the health of mother and baby. 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.

The National Institute for Occupational Safety and Health recommends using personal protective equipment, avoiding skin contact with chemicals, washing hands before eating or drinking, reviewing all workplace material safety data sheets to learn about potential hazards, leaving contaminated clothing at work, showering with soap and water before leaving, and keeping street clothes separate from work clothes to prevent contamination.[21] These practices help prevent exposure of individuals and their familial contacts to hazardous chemicals.

Men should also become as healthy as possible prior to their partner becoming pregnant to increase the chance of having a healthy baby. Like women, men should also reach a healthy weight, keep chronic health conditions under control, and prevent or treat sexually transmitted diseases. Men should also avoid tobacco products, certain drugs, exposure to toxic substances and drinking too much alcohol. Furthermore, men should also strive to become mentally healthy and support their partners' health.

Preventing Birth Defects

Folic acid is a B-complex vitamin that is proven to be protective against neural tube defects such as anencephalus and spina bifida, which are defects of the central nervous system. It may also provide protection against other birth defects. To be fully effective, a woman needs to begin taking the recommended daily dose of 400 micrograms at least a full month before becoming pregnant and continue to take folic acid daily during pregnancy. For women who have had a baby with a neural tube defect in the past, the recommended daily dose of folic acid is higher. If a woman finds she is pregnant and has not been taking folic acid, it is best to start taking folic acid immediately and continue to do so thereafter.

A woman should see her medical provider when planning a pregnancy and start prenatal care as soon as she thinks that she is pregnant. A pregnant woman should work with her healthcare provider to keep chronic diseases (like diabetes) under control, avoid drinking alcohol, avoid smoking cigarettes and prevent infections as much as possible. Some easy steps to prevent infections include frequent hand-

[21] National Institute for Occupational Safety and Health. The Effects of Workplace Hazards on Female Reproductive Health. https://www.cdc.gov/niosh/docs/99-104/pdfs/99-104.pdf?id=10.26616/NIOSHPUB99104. Published February 1999.

washing, cooking meat until it is well done, and staying away from people who have an infection. Another way to prevent infections is to be up-to-date with recommended vaccines before, during, and after pregnancy. Vaccines such as the measles, mumps, rubella (MMR) vaccine, which are recommended in childhood, are critical to prevent congenital infections that can cause birth defects.

It is also important for people who the pregnant woman may come in contact with to be vaccinated so they don't expose the pregnant woman and her baby to vaccine-preventable diseases. Likewise, vaccines such as the influenza and Tdap vaccines are critical during pregnancy for the health of both mother and baby.

While there are still certain hereditary and genetic factors that cannot be avoided, there are many factors that public health staff, new mothers-to-be and health care providers can address together to reduce birth defect occurrences in infants born in Tennessee.

Future Directions

This report reveals disparities in birth defect rates associated with the Social Determinants of Health, including, but not limited to: race, education level, income level, and geography of residence. These findings present opportunities for future directions. According to its Tennessee Code Annotated, the charge of the TNBDSS is: "to evaluate the current prevention initiatives undertaken by the state, and to give guidance for improvement of these initiatives or for the addition of new prevention strategies."

To ensure that the at-risk populations identified in this report have equal access to state resources associated with healthy pregnancies and birth defects prevention, TNBDSS will provide identified programs with tailored recommendations based on these report findings. In addition, with funding provided by the recent cooperative agreement from the CDC, TNBDSS will work to develop new primary prevention strategies to address identified gaps in birth defects prevention. One example is to focus on educating key providers on the link between the importance of women's preconception health (including the control of chronic diseases like diabetes and hypertension, weight management, and substance cessation) and a healthy

pregnancy, as well as the resources available to address these issues through the Tennessee Department of Health. Provider education can lead to patient education, referrals, and resource recommendations. This would potentially help to prevent serious and costly birth defects such as Critical Congenital Heart Defects and neural tube defects.

By addressing the identified Social Determinants of Health indicated in this report and helping to ensure access to those identified as most at-risk for adverse birth outcomes, we can help to promote a healthy pregnancy for all Tennessee mothers.

Acknowledgement

The TNBDSS thanks the members of the Tennessee Birth Defects Registry Advisory Committee, and its dedicated community members, for their continued support and guidance.

Funding for this report is provided by a cooperative agreement with the Centers for Disease Control and Prevention (Grant Number: 6 NU50DD000103-01-01)

Tips for a Healthy Pregnancy

Before and during pregnancy

- Consume at least 400 micrograms (mcg) of folic acid every day
- See a healthcare professional regularly
- Plan and space pregnancies at least 18 months apart
- Prevent and treat medical conditions like diabetes and hypertension
- Strive to reach and maintain a healthy weight
- Be physically active
- Eat a healthy diet that includes fruits, vegetables, whole grains, low-fat dairy, and lean proteins

Avoid harmful substances

- Avoid smoking
- Avoid drinking alcohol
- Avoid drugs such as opioids, marijuana, cocaine, methamphetamines, and other “street” drugs and seek resources and cessation support for pre-existing addiction.
- Be aware of and avoid potentially harmful exposures at work and home

Talk to a healthcare provider about

- Getting a medical checkup
- Taking any medications, both prescription and over the counter
- Family history of medical conditions
- Vaccinations needed before, during, and after pregnancy, such as flu, Tdap, and COVID-19.
- Any upcoming travel (either domestically or abroad) to discuss prevention of infections abroad, vaccination requirements, and the potential need for medical care in transit and at your destination

Appendix A: Reportable Birth Defects in Tennessee

Reportable Birth Defects in Tennessee	
Birth Defect	**ICD-10-CM codes**
Brain abnormalities with and without microcephaly	
Confirmed or possible congenital microcephaly <3rd percentile	Q02
Intracranial calcifications	No specific code; may be included under Q04.8, Q04.9
Cerebral atrophy	No specific code; may be included under Q04.3
Abnormal cortical formation (e.g., polymicrogyria, lissencephaly, pachygyria, schizencephaly, gray matter heterotopia)	Q04.3, Q04.6, Q04.8
Corpus callosum abnormalities	Q04.0
Cerebellar abnormalities	No specific code; may be included under Q04.3
Porencephaly	Q04.6
Hydranencephaly	No specific code; should be included in Q04.3
Ventriculomegaly / hydrocephaly Mild or borderline Ventriculomegaly/enlargement of cerebral ventricles must have another qualifying defect to be reported.	Q03.0–Q03.9
Fetal brain disruption sequence (include: collapsed skull, overlapping sutures, prominent occipital bone, scalp rugae, etc.)	No specific code. This might be coded as microcephaly or another single brain malformation, or all the components that might be coded individually. Q02, Q04.8, Q04.9 Include the following abnormalities only if co-existing abnormalities of the brain have been diagnosed: Q67.4, Q75.8, Q75.9, Q82.8
Other major brain abnormalities, including intraventricular hemorrhage Include *in utero* IVH, only if an additional qualifying defect is present	Q04.0, Q04.3–Q04.9, Q07.00, Q07.02
Neural tube defects and other early brain malformations	
Anencephaly / Acrania	Q00.0–Q00.2
Encephalocele	Q01.0–Q01.9
Spina bifida	Q05.0–Q05.9, Q07.01, Q07.03
Holoprosencephaly / Arhinencephaly	Q04.1, Q04.2
Eye abnormalities	
Microphthalmia / Anophthalmia	Q11.0–Q11.2
Coloboma	Q12.2, Q13.0, Q14.1–Q14.8
Cataract	Q12.0
Intraocular calcifications	Q13.8, Q13.9, Q14.1–Q14.9
Chorioretinal anomalies involving the macula (e.g., chorioretinal atrophy and scarring, macular pallor, gross pigmentary mottling and retinal hemorrhage); excluding retinopathy of prematurity	No specific code. This might be coded under the affected part of the eye. Q14.1–Q14.9
Optic nerve atrophy, pallor, and other optic nerve abnormalities	Q14.2, H47.03
Consequences of central nervous system (CNS) dysfunction	

Congenital contractures (e.g., arthrogryposis, club foot, congenital hip dislocation/developmental dysplasia of the hip) **only** with associated brain abnormalities	Q65.0–Q65.9, Q66.0–Q66.9, Q68.8, Q74.3
Confirmed congenital deafness documented by postnatal testing	H90.0–H90.8, H90.A, H91.0-H91.9, Q16.0–Q16.9

Appendix B: Birth Defects by Organ System and Fetal Alcohol Syndrome

Table 7. Cases and Prevalence of Major Birth Defects and Fetal Alcohol Syndrome, Tennessee, 2014-2018

Birth Defect	Number[1]	Rate[2]	95% CI[3]
Central Nervous System			
Anencephaly	72	1.8	1.4-2.2
Encephalocele	58	1.4	1.1-1.8
Holoprosencephaly	145	3.6	3.0-4.2
Spina bifida without anencephaly	188	4.6	4.0-5.3
Total Central Nervous System Cases	442	10.9	9.9-11.9
Total Central Nervous System Defects	463	11.4	10.4-12.5
Eye/Ear			
Anophthalmia/microphthalmia	58	1.4	1.1-1.8
Anotia/microtia	66	1.6	1.3-2.1
Congenital cataract	99	2.4	2.0-3.0
Total Eye and Ear Cases	214	5.3	4.6-6.0
Total Eye and Ear Defects	223	5.5	4.8-6.2
Cardiovascular			
Critical congenital heart disease (CCHD) conditions			
Coarctation of the aorta	359	8.9	7.9-9.8
Common truncus (truncus arteriosus or TA)	36	0.9	0.6-1.2
Double outlet right ventricle (DORV)	129	3.2	2.6-3.7
Ebstein's anomaly	62	1.5	1.2-2.0
Hypoplastic left heart syndrome	149	3.7	3.1-4.3
Interrupted aortic arch (IAA)	79	1.9	1.5-2.4
Pulmonary valve atresia and stenosis	379	9.3	8.4-10.3
Tetralogy of Fallot (TOF)	263	6.5	5.7-7.3
Total anomalous pulmonary venous connection (TAPVC)	42	1.0	0.7-1.4
Transposition of the great arteries (TGA)	137	3.4	2.8-3.9
Tricuspid valve atresia and stenosis	50	1.2	0.9-1.6
Single Ventricle	59	1.5	1.1-1.9
Other cardiovascular conditions			
Aortic valve stenosis	74	1.8	1.4-2.3
Atrial septal defect	8,046	198.4	194.0-202.8
Atrioventricular septal defect (Endocardial cushion defect)	239	5.9	5.1-6.6
Ventricular septal defect	2,006	49.5	47.3-51.6
Total Cardiovascular Cases	9,420	232.3	227.6-237.1
Total Cardiovascular Defects	12,109	298.6	293.2-304.0
Orofacial			
Choanal atresia	95	2.3	1.9-2.9
Cleft lip with cleft palate	284	7.9	6.2-7.8

Continued on next page

Cleft lip alone (without cleft palate)	111	2.7	2.2-3.2
Cleft palate alone (without cleft lip)	288	7.1	6.3-7.9
Total Orofacial Cases	772	19.0	17.7-20.4
Total Orofacial Defects	778	19.2	17.8-20.5
Birth Defect	**Number**	**Rate**	**95% CI**
Gastrointestinal			
Biliary atresia	176	4.3	3.7-5.0
Esophageal atresia/tracheoesophageal fistula	112	2.8	2.3-3.3
Rectal and large intestinal atresia/stenosis	205	5.1	4.4-5.7
Small intestinal atresia/stenosis	209	5.2	4.5-5.9
Total Gastrointestinal Cases	653	16.1	14.9-17.3
Total Gastrointestinal Defects	702	17.3	16.0-18.6
Genitourinary			
Bladder exstrophy	14	0.3	0.2-0.6
Cloacal exstrophy	104	2.6	2.1-3.1
Congenital Posterior Urethral Valves	66	3.2	2.5-4.0
Hypospadias	2,263	109.1	104.6-113.7
Renal agenesis/hypoplasia	267	6.6	5.8-7.4
Total Genitourinary Cases	2,695	66.5	63.9-69.0
Total Genitourinary Defects	2,714	66.9	64.4-69.5
Musculoskeletal			
Clubfoot	892	22.0	20.6-23.4
Diaphragmatic hernia	170	4.2	3.6-4.8
Gastroschisis	202	5.0	4.3-5.7
Limb deficiencies (reduction defects)	176	4.3	3.7-5.0
Omphalocele	120	3.0	2.4-3.5
Craniosynostosis[4]	415	17.1	15.5-18.8
Total Musculoskeletal Cases	1,909	47.1	45.0-49.2
Total Musculoskeletal Defects	1,975	48.7	46.6-50.9
Chromosomal			
Deletion 22q11.2	12	0.3	0.2-0.5
Trisomy 13	49	1.2	0.9-1.6
Trisomy 18	79	1.9	1.5-2.4
Trisomy 21 (Down syndrome)	572	14.1	12.9-15.3
Turner syndrome	51	2.6	1.9-3.4
Total Chromosomal Cases	757	18.7	17.3-20.0
Total Chromosomal Defects	763	18.8	17.5-20.2
Total Birth Defects Cases	14,874	366.8	360.8-372.8
Total Birth Defects	19,727	486.5	479.5-493.4
Fetal Alcohol Syndrome[5]	40	1.0	0.7-1.3

Note: For each organ system, the total *cases* number represents the number of infants. The total *defects* number represents the full count of diagnosed birth defects. These numbers are not equivalent because one infant can potentially be diagnosed with more than one birth defect. For example, a total of 9,420 infants were diagnosed with cardiovascular birth defects, but amongst these 9,324 infants, there were 12,109 total cardiovascular defects.
1. Number includes cases born alive and fetal deaths.
2. Rate per 10,000 live births.
3. Can be interpreted as range that we are 95% confident contains the true incidence in the population. Confidence intervals for conditions with less than 100 cases are exact Poisson; otherwise confidence intervals are based on the normal approximation.
4. Includes cases from 2016-2018. Prior to 2016, craniosynostosis cases could not be identified. Rate is calculated using live births from 2016-2018.
5. Fetal alcohol syndrome cases are not included in the count for total birth defect cases/total birth defects.
Data Source: Tennessee Department of Health, Tennessee Birth Defects Registry

Appendix C: Resources

Tennessee Resources

Tennessee Department of Health:
https://www.tn.gov/health/health-program-areas/mch-cyshcn.html
Tennessee Medical Home:
https://www.tnaap.org/programs/tennessee-medical-home/tennessee-medical-home-overview
Family Voices of Tennessee:
https://www.familyvoicestn.org/
Support and Training for Exceptional Parents (STEP):
https://www.tnstep.org/
Kidcentral tn:
http://www.kidcentraltn.com
Disability Pathfinder:
http://vkc.mc.vanderbilt.edu/vkc/pathfinder/
Disability Rights Tennessee:
http://www.disabilityrightstn.org/
University of Tennessee Center for Developmental Disabilities:
https://www.uthsc.edu/bcdd/
Vanderbilt Kennedy Center:
https://vkc.mc.vanderbilt.edu/vkc/
Vanderbilt Consortium Leadership Education in Neurodevelopmental Disabilities (LEND):
https://www.etsu.edu/coe/efse/lend.php
Tennessee Early Intervention System (TEIS):
https://www.tn.gov/didd/for-consumers/tennessee-early-intervention-system-teis.html
Chattanooga Down Syndrome Society:
http://www.chattanoogadownsyndrome.org/

Clarksville Association Down Syndrome:
https://www.cadstn.org/
Down Syndrome Association of Middle Tennessee:
https://www.somethingextra.org/
Down Syndrome Association of West Tennessee:
https://dsawt.com/
Down Syndrome Awareness Group of East Tennessee:
https://dsagtn.org/
FRIENDS (Friends Reaching Inspiring Educating Neighbors about Down Syndrome):
http://dsfriends.net/
Understand Your Child's Diagnosis of Down Syndrome (for Parents):
https://vkc.mc.vanderbilt.edu/assets/files/resources/DS%20Guide%20for%20Parents.pdf
Understand Your Patient's Diagnosis of Down Syndrome (for Providers):
https://vkc.mc.vanderbilt.edu/assets/files/resources/DS%20Guide%20for%20Doctors.pdf

Tennessee Resources (Healthy Pregnancy)

Baby & Me - Tobacco Free™:
https://www.tn.gov/health/health-program-areas/fhw/baby-me-tobacco-free.html
Community Health Access and Navigation in Tennessee (CHANT):
https://tn.gov/health-program-areas/fhw/early-childhood-program/chant.html
Project Diabetes:
https://tn.gov/health/health-program-areas/mch-diabetes/project-diabetes.html
Tennessee Tobacco Quitline:
http://www.tnquitline.org
Take Charge of Your Diabetes:
https://ag.tennessee.edu/fcs/Pages/Health/TakeChargeOfYourDiabetes.aspx
Tennessee Women, Infants, and Children (WIC) Program:
https://www.tn.gov/health/health-program-areas/fhw/wic.html
Tennessee WIC Farmer's Market Nutrition Program:
https://www.tn.gov/content/tn/health/health-program-areas/fhw/farmers/wic-fmnp.html

National Resources (General)

CDC National Center on Birth Defects and Developmental Disabilities:
https://www.cdc.gov/ncbddd/birthdefects/index.html

CDC National Institute for Occupational Safety and Health:
https://www.cdc.gov/niosh/topics/repro/pregnancyjob.html/
CDC Men's Preconception Health:
https://www.cdc.gov/preconception/men.html
National Birth Defects Prevention Network:
https://www.nbdpn.org/
March of Dimes:
https://www.marchofdimes.org/

National Resources (Healthy Pregnancy)

American College of Obstetricians and Gynecologists (ACOG):
https://acog.org/womens-health/pregnancy
CDC Healthy Pregnancy:
https://www.cdc.gov/pregnancy.index.html
CDC Women's Preconception Health:
https://cdc.gov/preconception/overview.html
CDC Men's Preconception Health:
https://www.cdc.gov/preconception/men.html
Food and Drug Administration (FDA):
https://fda.gov/consumers/womens-health-topics/pregnancy
March of Dimes:
https://www.marchofdimes.org/
MotherToBaby:
https://mothertobaby.org/
MyPlate US Department of Agriculture:
https://myplate.gov/life-stages/pregnancy-and-breastfeeding
National Diabetes Prevention Program:
https://cdc.gov/diabetes/prevention/index/html
US Department of Health & Human Services:
https://womenshealth.gov/pregnancy

Appendix D: Birth Defects to be Monitored

CDC NOFO #DD21-2101: Advancing Population-Based Surveillance of Birth Defects and ICD-10-CM Codes

Critical Congenital Heart Disease (CCHD) Conditions	
Coarctation of the aorta*	Q25.1
Ebstein's anomaly*	Q22.5
Pulmonary atresia*	Q22.0
Tricuspid atresia*	Q22.4
Double outlet right ventricle (DORV)	Q20.1
Hypoplastic left heart syndrome (HLHS)	Q23.4
Interrupted aortic arch (IAA)	Q25.21
Single ventricle	Q20.4
Tetralogy of Fallot (TOF)	Q21.3
Total anomalous pulmonary venous connection (TAPVC)	Q26.2
Transposition of the great arteries (TGA)	Q20.3, Q20.5
Truncus arteriosis (TA)	Q20.0
Central Nervous System	
Anencephaly	Q00.0–Q00.1
Spina bifida without anencephalus	Q05.0–Q05.9, Q07.01, Q07.03
Eye/Ear	
Anophthalmia/ microphthalmia	Q11.0–Q11.2
Cardiovascular	
Atrioventricular septal defect (ASD)	Q21.2
Orofacial	
Cleft lip with cleft palate	Q37.0-Q37.9
Cleft lip alone (without cleft palate)	Q36.0-Q36.9
Cleft palate alone (without cleft lip)	Q35.1-Q35.9
Gastrointestinal	
Esophageal atresia/tracheoesophageal fistula	Q39.0-Q39.4
Small intestinal atresia/stenosis	Q41-Q41.9
Musculoskeletal	
Diaphragmatic hernia	Q79.0, Q79.1
Gastroschisis	Q79.3
Limb deficiencies (reduction defects)	Q71-Q71.9, Q72-Q72.9, Q73-Q73.8
Omphalocele	Q79.2
Chromosomal	
Trisomy 21 (Down Syndrome)	Q90-Q90.9

*Additional grant component will include monitoring of these additional Critical Congenital Heart Disease (CCHD) conditions.

Tennessee Department of Health Authorization Number. This electronic only publication was produced at no cost.