Tennessee Birth Defects Data Report 2012-2017

Tennessee Department of Health | December 2020

TN Department of Health

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Executive Summary

Birth defects are common, costly, and critical. The Tennessee Birth Defects Surveillance System (TNBDSS) estimates that between 2012 and 2017, over 68 million dollars was spent on hospitalizations related to critical congenital heart defects alone.[1,2] Between 2012 and 2017, there were 16,704 babies (approximately 2,784 babies per year) diagnosed with birth defects in Tennessee. The most commonly reported 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 account for 21% of the infant deaths in Tennessee, 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 population-based surveillance report provides details on the prevalence of 47 major birth defects and fetal alcohol syndrome for Tennessee infants born in the years 2012 through 2017. This report also includes specific information about birth defect rates by socio-demographics characteristics, 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.
- Maternal health behaviors such as smoking and chronic health conditions such as diabetes and hypertension are associated with an increased risk of specific birth defects.

1. Arth AC, Tinker SC, Simeone RM, Ailes EC, 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 category in Table 2 of Arth *et al*. The median cost was then multiplied by the number of defects, provided in Table 9 of this report, of the corresponding diagnoses included in the authors' organ system definition.

- Certain types of birth defects, especially chromosomal defects, were more common among babies who were born to mothers aged 35 years old and greater.
- The highest rates of birth defects were found in Shelby County, Sullivan County, and the Northeast region.
- The highest rates of infant mortality due to a birth defect were found in Shelby County, the Upper Cumberland region, and the Northeast region.
- Non-Hispanic Black babies had the highest prevalence of birth defects among maternal racial/ethnic groups.

Key Prevention Messages:

- Birth defects surveillance programs play a key role in efforts to prevent birth defects.
- 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 months 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 healthcare providers to discuss any medication use, routine vaccinations that are given before and during pregnancy, and ways to prevent infections.
- It is recommended that women plan and space pregnancies at least 18 months apart.

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.[3] 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 2012 and 2017, over 800 thousand dollars was spent on hospitalizations due to ear-related birth defects, over 7.8 million was spent on orofacial birth defect hospitalizations, 8.3 million was spent on hospitalizations related to chromosomal birth defects, and over 68 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 mothers, and certain maternal infections.

3. 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.

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[4] and fetal alcohol syndrome for Tennessee infants born in the years 2012 through 2017. TNBDSS selected which birth defects to study based on national surveillance recommendations. 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, age, and maternal health characteristics, such as pre-pregnancy diabetes and smoking during pregnancy. Special attention is given to selected birth defects of public health significance. This report provides education on birth defects prevention, highlights current prevention efforts, 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 the genitourinary defects such as hypospadias, which affects only males. Certain types of birth defects, especially chromosomal defects, were more common among babies who were born to mothers aged 35 years old and greater. Non-Hispanic Blacks had the highest prevalence of birth defects among maternal racial/ethnic groups. The highest prevalence of birth defects was found in Unicoi County. Higher prevalence of birth defects is also noted among infants of women with a 12th grade education or less and women on Medicaid compared to private insurance. Additionally, babies born to mothers with pre-pregnancy diabetes or obesity are at increased risk for cardiovascular, central nervous system, genitourinary, musculoskeletal, and orofacial birth defects compared to babies born to mothers who did not have diabetes or had a normal weight prior to pregnancy.

4. 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.

These findings also underscore the impact of social and maternal health factors such as education, income, environment, and prevalence of chronic disease on health outcomes.

Data Sources and Limitation

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, demographics, and street-level geography 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. For infant death cases, demographic, geographic, and risk factor information are obtained from the death certificate data 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 data 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 previous 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, which is a congenital heart defect, and hypospadias, a common genitourinary defect in males. 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.

Until recently, surveillance was conducted passively, 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[5] 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.

Monitoring birth defects is essential to ensure timely referral to services and enhance care coordination for affected children in Tennessee. Following the confirmation of a reported neurologic birth defect, referrals are made by TDH to the Tennessee Early Intervention System (TEIS), Children's Special Services (CSS), and Family Voices of Tennessee. 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.

5. 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.

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. 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.

In addition to connecting families to needed services and monitoring the occurrence of birth defects and patterns or trends, TNBDSS contributes to research conducted by the CDC and the National Birth Defects Prevention Network. Through collaboration with national partners, TNBDSS aims to better understand the causes of birth defects and identify strategies for reducing birth defects.

Tennessee Birth Data

In Tennessee, an average of 80,820 live births occurred to resident mothers annually during the years 2012 through 2017. During this time frame, approximately 67% of all infants born were Non-Hispanic White and 20% were Non-Hispanic Black (Table 1).

Table 1. Live Births by Maternal Race/Ethnicity, Tennessee, 2012-2017

Year	Total	Non-Hispanic White	Non-Hispanic Black	Hispanic
2012	80,202	54,018	16,462	6,977
2013	79,954	54,251	16,764	6,850
2014	81,609	55,345	16,927	6,982
2015	81,374	54,621	16,571	7,260
2016	80,755	53,725	16,212	7,628
2017	81,024	53,472	16,403	7,676

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.

Birth Defects Prevalence in Tennessee, 2012-2017

Table 9 in Appendix B shows the case numbers and rates for the 47 major birth defects[6] by organ system and fetal alcohol syndrome. Between January 2012 and December 2017, there were 16,704 babies diagnosed with birth defects. In addition, there were 59 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 (22,515) over this time period is higher. Out of the 22,515 defects, 13,690 were cardiovascular defects which represent 60% of the total. The genitourinary system, with 3,356 defects, is the second most-affected organ system (almost 15% of total defects). The largest single birth defect in Tennessee is atrial septal defect with a count of 8,589, followed by hypospadias (n=2,654) and ventricular septal defect (n=2,461). By identifying the most common birth defects and most commonly affected organ systems, targeted prevention efforts can be developed based on known risk factors for particular birth defects.

Birth Defects in Tennessee versus the United States

Table 2 shows prevalence data for 30 birth defects. This table features Tennessee data from 2012-2017 for the selected birth defects, while national rates are listed for these same birth defects from the years 2010-2014. National estimates are based on pooled data from 39 state birth defects surveillance programs. Of the 30 birth defects listed, rates in Tennessee are higher for 22 of the birth defects. When examining the cardiovascular birth defects, in particular, there are notable differences between rates in Tennessee and the United States. The rate for Double Outlet Right Ventricle is approximately 2 times higher in Tennessee than the national average. In addition, rates for Single Ventricle and Coarctation of the Aorta are 1.8 times and 1.5 times, respectively, higher than the national rates. It is important to identify birth defects with rates that are higher than the national average as these warrant particular attention.

6. 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.

Table 2. Frequency of Selected Birth Defects for the U.S. and Tennessee.				
Birth Defect	**Tennessee 2012-2017**		**United States[1] 2010-2014**	
	Rate[2]	**Frequency[3]**	**Rate**	**Frequency**
Central Nervous System				
Anencephaly	1.71	1 in 5,842	2.15	1 in 4,647
Encephalocele	1.40	1 in 7,131	0.95	1 in 10,502
Spina bifida without anencephaly	4.80	1 in 2,081	3.63	1 in 2,758
Eye				
Anophthalmia/microphthalmia	1.32	1 in 7,577	1.91	1 in 5,243
Cardiovascular				
Coarctation of the aorta	8.41	1 in 1,189	5.57	1 in 1,795
Common truncus	0.97	1 in 10,317	0.64	1 in 15,696
Double outlet right ventricle	3.28	1 in 3,050	1.67	1 in 5,997
Ebstein anomaly	1.63	1 in 6,138	0.77	1 in 13,047
Hypoplastic left heart syndrome	3.75	1 in 2,664	2.60	1 in 3,841
Interrupted aortic arch	3.40	1 in 2,939	0.62	1 in 16,066
Pulmonary valve atresia and stenosis	10.56	1 in 947	9.51	1 in 1,052
Single ventricle	1.36	1 in 7,347	0.75	1 in 13,351
Tetralogy of Fallot	6.17	1 in 1,622	4.61	1 in 2,171
Total anomalous pulmonary venous connection	1.30	1 in 7,697	1.28	1 in 7,809
Transposition of great arteries	4.93	1 in 2,029	3.71	1 in 2,695
Tricuspid valve atresia and stenosis	1.18	1 in 8,507	1.68	1 in 5,938
Atrioventricular septal defect	5.86	1 in 1,707	5.38	1 in 1,859
Orofacial				
Cleft lip with cleft palate	6.70	1 in 1,492	6.40	1 in 1,563
Cleft lip alone (without cleft palate)	2.76	1 in 3,619	3.56	1 in 2,807
Cleft palate alone (without cleft lip)	6.41	1 in 1,559	5.93	1 in 1,687
Gastrointestinal				
Esophageal atresia/tracheoesophageal fistula	3.28	1 in 3,050	2.41	1 in 4,144
Rectal and large intestinal atresia/stenosis	5.20	1 in 1,924	4.46	1 in 2,242
Musculoskeletal				
Clubfoot	19.76	1 in 506	16.87	1 in 593
Diaphragmatic hernia	4.10	1 in 2,437	2.79	1 in 3,591
Gastroschisis[4]	5.28	1 in 1,894	4.94	1 in 2,025
Limb deficiencies (reduction defects)	4.27	1 in 2,343	5.15	1 in 1,943
Omphalocele	2.68	1 in 3,730	2.40	1 in 4,175

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Chromosomal[4]				
Trisomy 13	1.15	1 in 8,659	1.49	1 in 6,717
Trisomy 18	1.94	1 in 5,159	3.43	1 in 2,918
Trisomy 21 (Down syndrome)	14.62	1 in 684	15.74	1 in 635

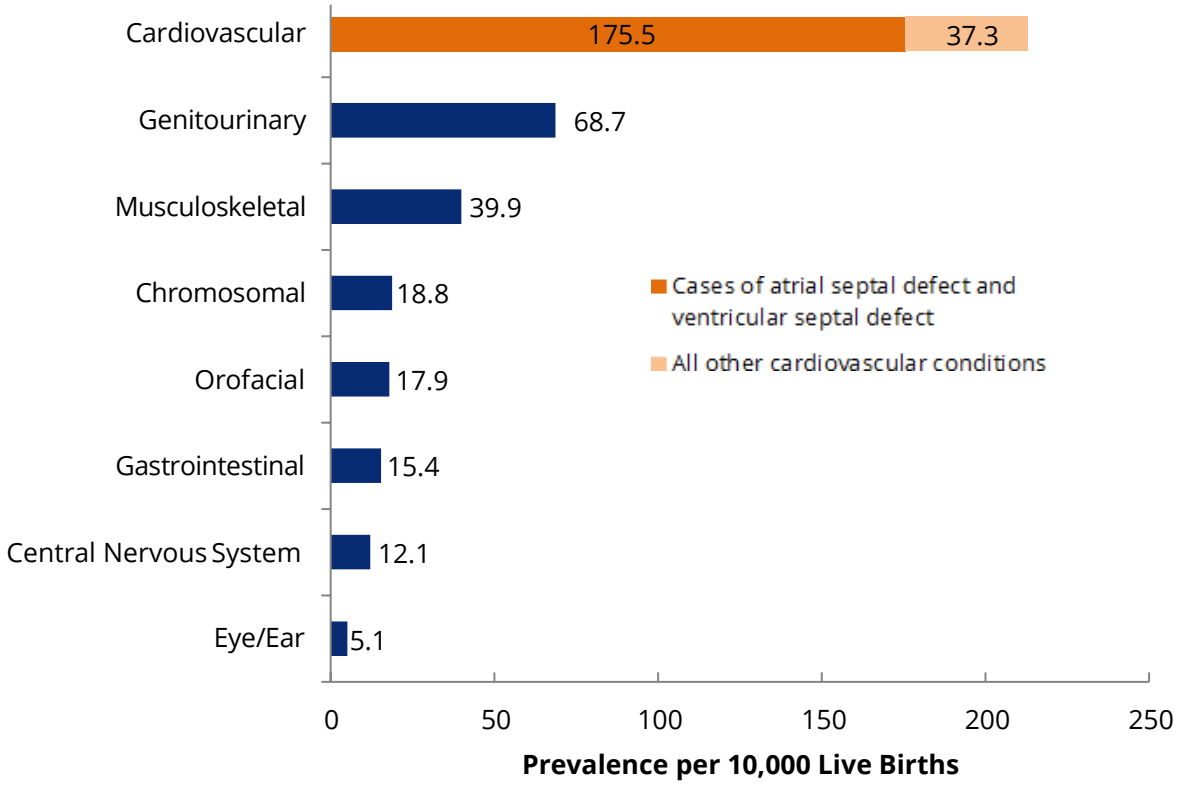
1. National estimates based on pooled data from 39 state birth defects surveillance programs. Estimates were standardized to the racial and ethnic distribution of the United States live birth population from 2010 through 2014. See full paper: Mai CT, Isenburg JL, Canfield MA, et al. National population-based estimates for major birth defects, 2010–2014. *Birth Defects Research.* 2019;111(18),1420-1435. https://doi.org/10.1002/bdr2.1589
2. Rate per 10,000 live births.
3. Estimated frequency of occurrence in a given number of live births.
4. Estimates for gastroschisis and the three chromosomal birth defects were standardized to the United States maternal age distribution.

Prevalence of Major Birth Defects by Organ System

Figure 1 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 212.8 per 10,000 live births. As seen in Figure 1, the most common cardiovascular birth defects are atrial septal defects (ASD) and ventricular septal defects, (VSD) with a combined rate of 175.5 per 10,000 live births compared to a rate of 37.3 per 10,000 live births for all other cardiovascular birth defects. An ASD is a hole in the wall (septum) that divides the two upper chambers of the heart.[7] ASDs are common (accounting for 10-15% of congenital heart defects) and often spontaneously resolve during infancy or early childhood.[8] A VSD is a hole in the septum that separates the two lower chambers (ventricles) of the heart.[9] 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.[10] Most small VSDs spontaneously close during the first two years of life. 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, these rates of cardiovascular birth defects were calculated separately from other cardiovascular birth defects.

7. 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.
8. 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.
9. 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.
10. 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. Accessed September 20,2020.

Figure 1. Prevalence of Major Birth Defects by Organ System, Tennessee, 2012-2017



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

Infant Mortality Data and Birth Defects in Tennessee

During the period 2012-2017, there was an average of 575 infant deaths per year (Table 3). While the total infant mortality rate decreased from 2012-2013, the overall rate increased from 2013-2017. Infant deaths do not impact all races equally. Non-Hispanic Black infants have an average mortality rate of over 2 times higher than Non-Hispanic White infants and nearly 2.5 times higher than Hispanic infants.

Table 3. All Infant Deaths by Maternal Race/Ethnicity, Tennessee, 2012-2017

	Total		Non-Hispanic White		Non-Hispanic Black		Hispanic	
Year	Number	Rate[1]	Number	Rate	Number	Rate	Number	Rate
2012	579	7.2	337	6.2	200	12.1	26	3.7
2013	543	6.8	295	5.4	195	11.6	40	5.8
2014	562	6.9	302	5.5	211	12.5	28	4.0
2015	569	7.0	331	6.1	184	11.1	33	4.5
2016	597	7.4	347	6.5	195	12.0	38	5.0
2017	597	7.4	322	6.0	211	12.9	50	6.5

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.*

Table 4 shows the ten leading causes of infant deaths in Tennessee between 2012 and 2017. Birth defects were the leading cause of all infant deaths (21%), followed by preterm birth/low birthweight (14%).

Table 4. Leading Causes of Infant Death, Tennessee, 2012-2017

Rank	Cause of Death	Number of Deaths	Percent of Deaths
1	Birth defects	740	21
2	Preterm birth and low birthweight	495	14
3	Accidents	240	7
4	Sudden infant death syndrome (SIDS)	164	5
5	Maternal complications of pregnancy	126	4
6	Complications of placenta, cord, and membranes	93	3
7	Bacterial sepsis of newborn	85	2
8	Atelectasis (partial lung collapse)	83	2
9	Diseases of the circulatory system	78	2
10	Respiratory distress of newborn	66	2
--	All other causes	1,277	37
--	*All Causes*	*3,447*	*100*

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

Table 5 examines the top two causes of infant deaths, birth defects and prematurity/low birth weight, more closely. Among infants whose primary cause of death was a birth defect, 58% were also born premature (< 37 weeks). Preterm delivery often exacerbates the medical complications faced by infants born with major birth defects. Depending on the type of defect, infants born preterm may be at significantly greater risk of mortality compared to their counterparts delivered at term.[11] This pattern of preterm infants experiencing increased mortality has been demonstrated for neural tube defects,[12] congenital diaphragmatic hernia,[13] and congenital heart defects.[14]

Table 5. Co-Occurrence of Two Leading Causes of Infant Death, Tennessee, 2012-2017

Cause of Infant Death[1]		
	Born Preterm (<37 weeks)	Born Full Term (37+ weeks)
Birth Defect	58%	42%
	Major Birth Defect Present	No Major Birth Defect Present
Preterm birth and low birthweight	3%	97%

1. Represents underlying cause of death recorded on infant's death certificate.
Note: All birth defect or preterm birth/low birthweight related deaths with corresponding birth data available were included. This represented 98 percent of all birth defect deaths and 98 percent of preterm birth/low birthweight deaths.
Data Source: Tennessee Department of Health, Office of Vital Records and Statistics; Tennessee Department of Health, Tennessee Birth Defects Registry.

11. 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.
12. 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.
13. 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.
14. Tanner K, Sabrine N, Wren C. Cardiovascular malformations among preterm infants. *Pediatrics.* 2005; 116(6): e833–e838.

Table 6 provides more detail about the gestational ages of infants who died due to birth defects. Among the infants who died due to birth defects, 42% were full-term (37+ weeks) and 26% were late-preterm (34 to <37 weeks).

Table 6. Gestational age at birth of infant deaths caused by birth defects, Tennessee, 2012-2017

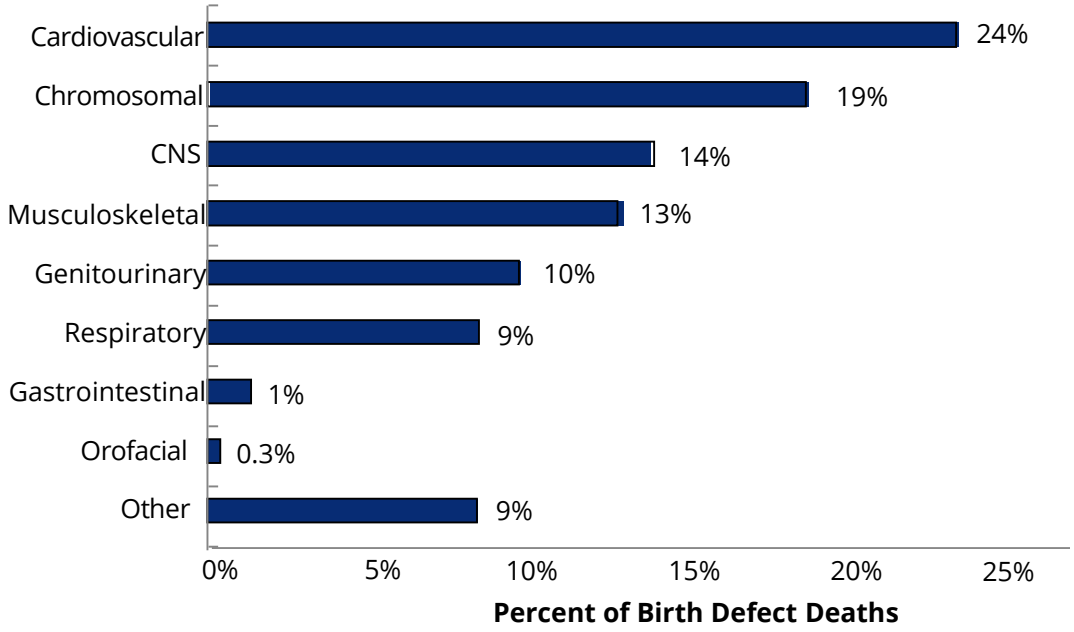
Gestational Age at Birth	Percent of Cases
<28 weeks	10
28-<32 weeks	12
32-<34 weeks	11
34-<37 weeks	26
37+ weeks	42

Note: All birth defect related deaths with corresponding birth data available were included. This represented 99 percent of all birth defect related deaths.

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

Figure 2 demonstrates birth defect deaths by the type of defect. Heart defects were the leading cause of birth defect deaths (24% of birth defect deaths), followed by chromosomal (19%) and central nervous system (14%) defects. These categories may not be mutually exclusive (i.e., chromosomal birth defects may be the cause of cardiovascular and other birth defects).

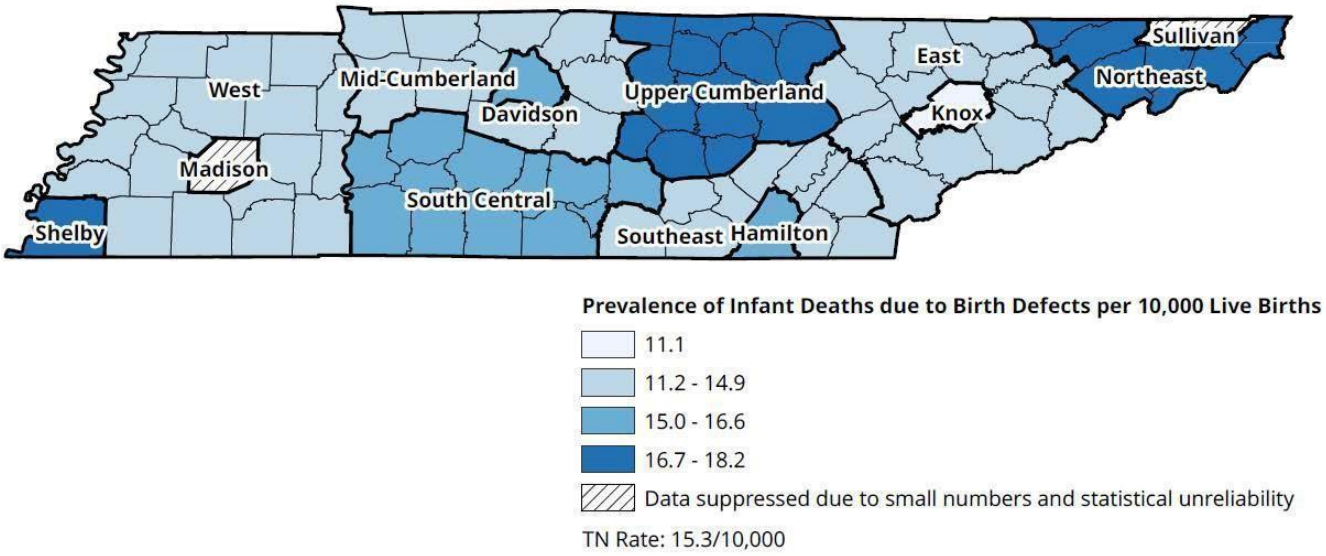
Figure 2. Birth Defect Deaths by Type of Defect, Tennessee, 2012-2017



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

Figure 3 depicts the rate of infant deaths due to a birth defect by maternal region of residence. The overall prevalence of infant mortality due to a birth defect for Tennessee is 15.3 per 10,000 live births. The highest number of infant deaths due to birth defects occurred in Shelby County. The highest rates were seen in the Northeast and Upper Cumberland regions, however it must be noted that these regions have smaller populations. Given that the differences between the rates are generally small the differences between the regions should be interpreted with caution.

Figure 3. Prevalence of Infant Deaths due to Birth Defects by Region, Tennessee, 2012-2017



Data Source: Tennessee Department of Health, Division of Vital Records & Statistics, Death Statistical File, 2012-2017.

Each Tennessee Birth Defects Report will highlight a specific birth defect, with the goal of providing information on causes, risk factors, and opportunities for prevention. This year's report features spina bifida.

Birth Defect Highlight: Spina Bifida

Image from the Centers for Disease Control and Prevention, National Center on Birth Defects and Developmental

What is Spina Bifida?

Spina bifida is a birth defect that occurs when the backbone does not develop properly during pregnancy. This may leave the spinal cord and nerves exposed and lead to spinal damage. This defect occurs during the first few weeks of pregnancy, often before a woman knows she's pregnant. Spina bifida may be diagnosed during pregnancy or after birth. Management and treatment of spina bifida depends on the severity of the diagnosis, but surgical repair may be needed.

Occurrence in the United States and Tennessee

The national rate for spina bifida is 3.63 per 10,000 live births (1 in every 2,758 live births is born with spina bifida). In Tennessee, the rate of babies born with spina bifida is higher – 4.80 per 10,000 live births (1 in every 2,081 live births is born with spina bifida).

Causes and Risk Factors

Similar to other birth defects, spina bifida is thought to be caused by a combination of genetic changes and other factors, such as environmental exposures or certain medications taken during pregnancy.

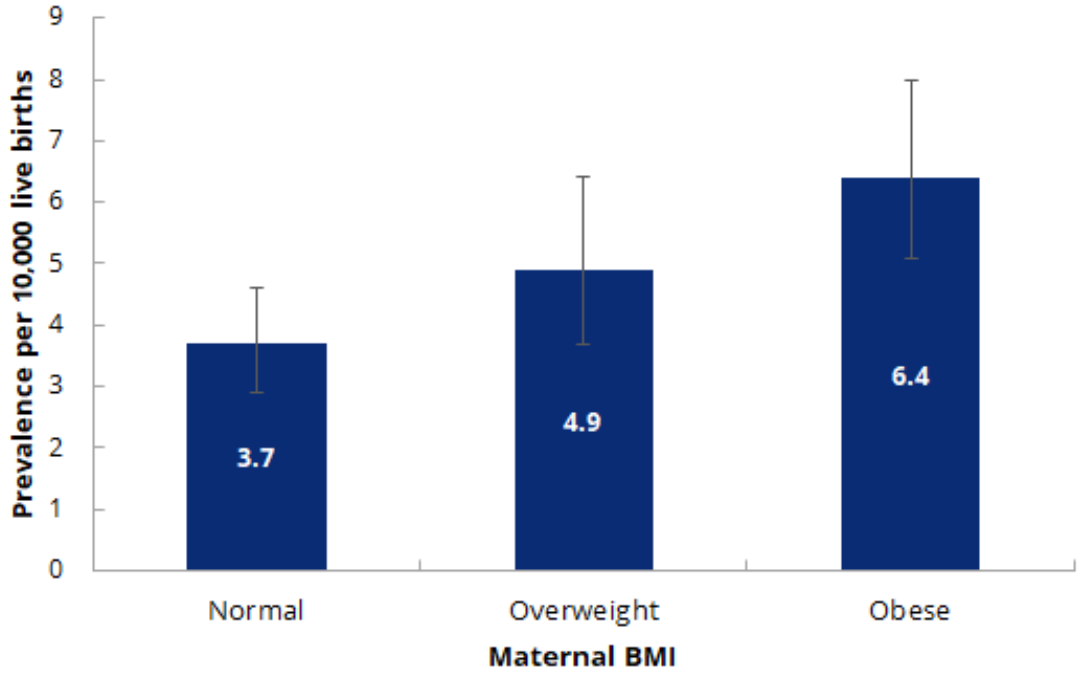
Research shows that pregnant women with low folate levels are more likely to have a baby born with spina bifida. The CDC recommends that women who are pregnant, or likely to become pregnant, should take 400 mcg of folic acid daily. Survey results from the Tennessee Pregnancy Risk Assessment Monitoring System (PRAMS) indicate that 56% of Tennessee mothers did not take a daily folic acid supplement prior to pregnancy.[15] In addition, it is especially important for women of child-bearing age to take a daily supplement as more than half of all pregnancies in Tennessee are unplanned.[15]

Medical conditions present prior to pregnancy, such as obesity or uncontrolled diabetes, are also shown to increase the risk of giving birth to a baby with spina bifida. Between 2014 and 2018, 1% of women who gave birth in Tennessee had pre-pregnancy diabetes, while 27% were considered obese.[16] Figure 4 shows the rate of spina bifida by maternal weight prior to pregnancy. There is a statistically significant difference in the rate of spina bifida between women considered obese and women of normal weight.

15. Tennessee Department of Health, Division of Population Health Assessment, Tennessee Pregnancy Risk Assessment Monitoring System (PRAMS), 2018.
16. Tennessee Department of Health, Division of Vital Records & Statistics, Birth Statistical File, 2014-2018.

Birth Defect Highlight: Spina Bifida

Figure 4. Prevalence of Spina Bifida by Maternal Pre-Pregnancy BMI, Tennessee, 2012-2017



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

Tennessee Department of Health Resources

- **Tennessee Women, Infants, and Children (WIC) Program** provides supplemental food assistance and nutrition education to low-income pregnant, postpartum, and breastfeeding women, infants, and children until age five. More information can be found at: https://www.tn.gov/health/health-program-areas/fhw/wic.html
- **The National Diabetes Prevention Program** is an evidence-based lifestyle change program for preventing type-2 diabetes. More information can be found at: https://www.tn.gov/health/health-program-areas/mch-diabetes/d/diabetes-prevention-program.html

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 and older, women with ≤ 12th grade education, and women on Medicaid have a higher rate of birth defects (Figures 5 and 9). 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 (Figures 8 and 10). 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 certain types of birth defects, with advanced maternal age (defined as women who are 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 5 shows the overall prevalence of birth defects by maternal age group in Tennessee. During 2012-2017, the birth defects prevalence rates were highest among women 40 and older (512.0 per 10,000 live births), followed by women aged 35-39 (396.0 per 10,000 live births) and women less than 20 years old (371.0 per 10,000 live births).

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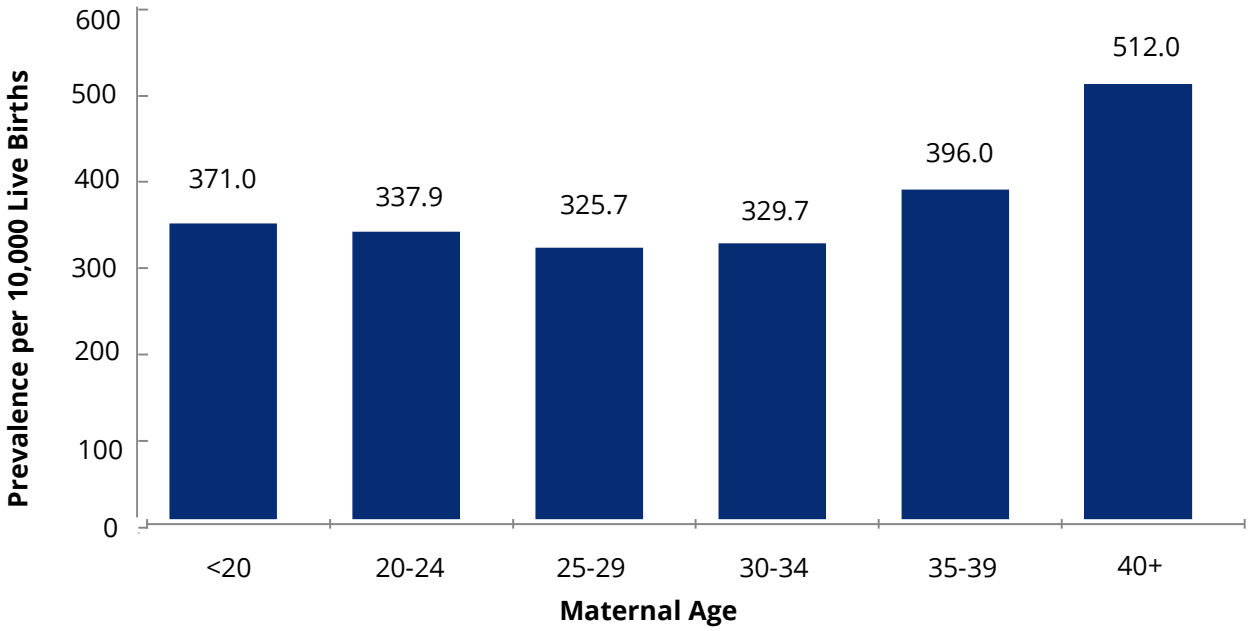
17. Canfield MA, Mai CT, Wang Y, O'Halloran M, Marengo LK, Olney RS, et al. The Association Between Race/Ethnicity and Major Birth Defects in the United States, 1999 - 2007. *Am J Public Health*. 2014;104(9): e14–e23.
18. National Center on Birth Defects and Developmental Disabilities, Centers for Disease 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.

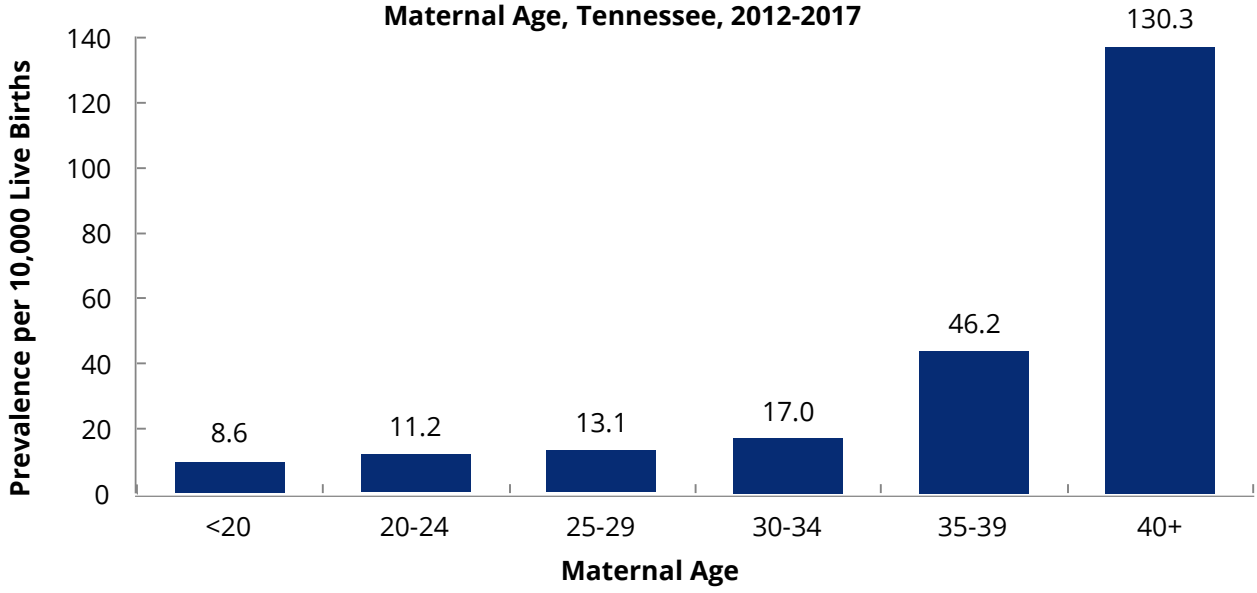
Figure 5. Prevalence of Major Birth Defects by Maternal Age, Tennessee, 2012-2017



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

Figure 6 and Figure 7 further illustrate the significant role that maternal age plays in birth defect occurrence. Figure 6 demonstrates that children born to mothers aged 40 years and above 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.

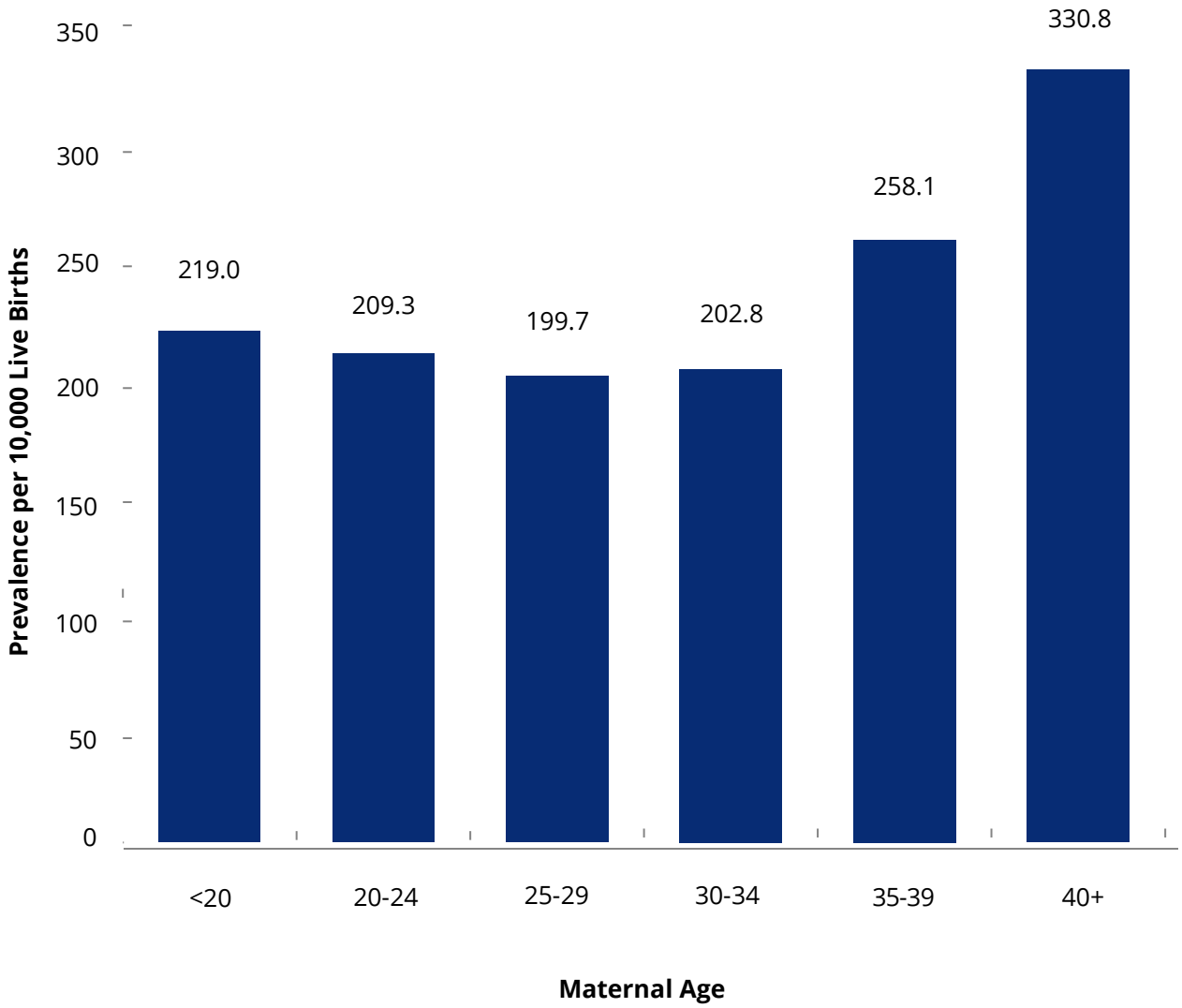
Figure 6. Prevalence of Chromosomal Major Birth Defects by Maternal Age, Tennessee, 2012-2017



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

Figure 7 illustrates that the rate of cardiovascular birth defects is higher for women 35 years old or greater, as compared to younger mothers. The rate of cardiovascular birth defects among women aged 40 years old or greater (330.8 per 10,000 live births) is about 1.7 times the average rate for babies born to mothers in the age group 25-29 (199.7 per 10,000 live births).

Figure 7. Prevalence of Cardiovascular Major Birth Defects by Maternal Age, Tennessee, 2012-2017

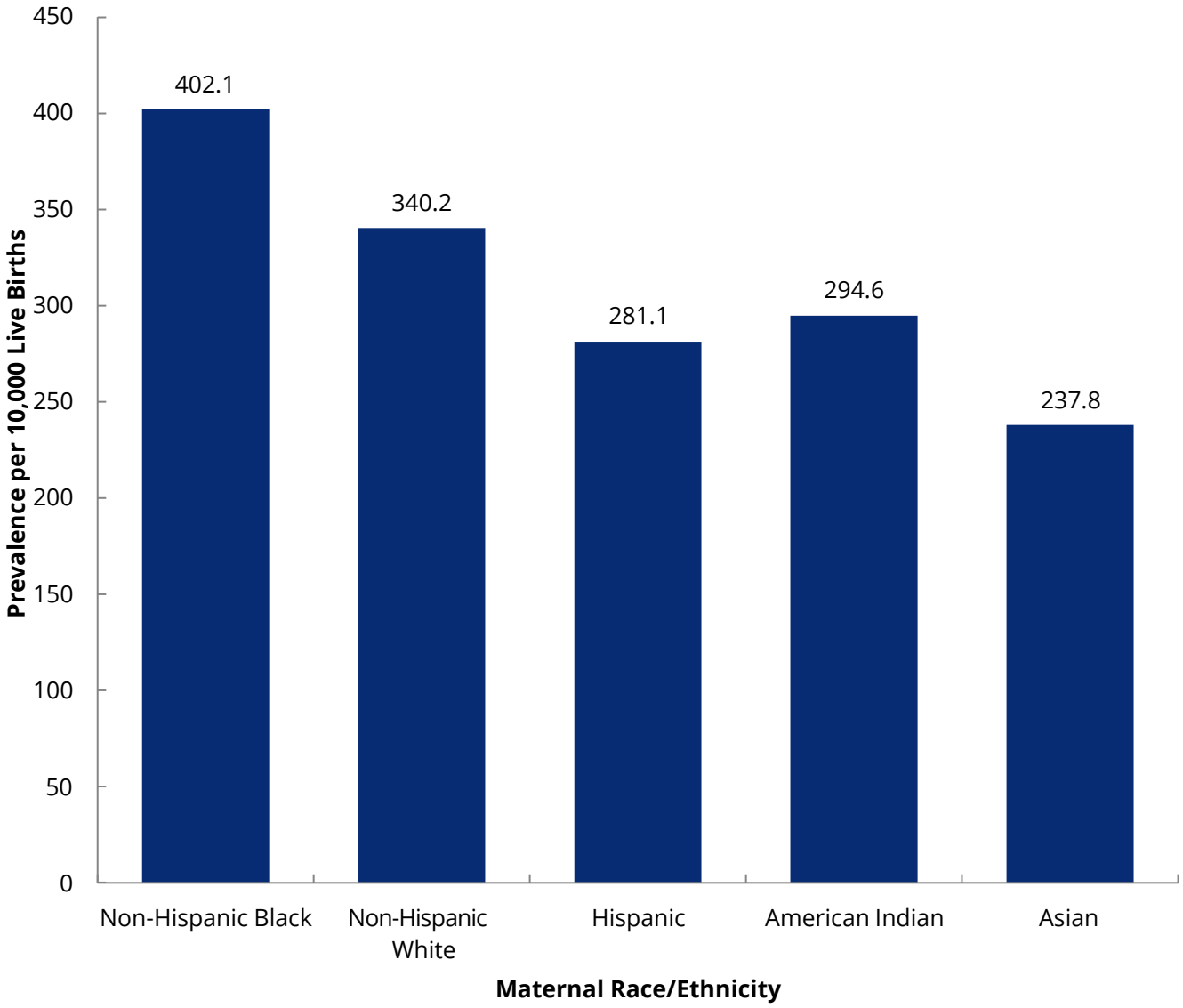


Data Source: Tennessee Department of Health, Tennessee Birth Defects Registry

Maternal Race and Ethnicity

Birth defects prevalence rates are highest for Non-Hispanic Blacks (402.1 per 10,000 live births), followed by Non-Hispanic Whites and Hispanic women (Figure 8). The lowest prevalence rate for birth defects (237.8 per 10,000 live births) was seen among babies born to Asian women.

Figure 8. Prevalence of Major Birth Defects by Maternal Race/Ethnicity, Tennessee, 2012-2017

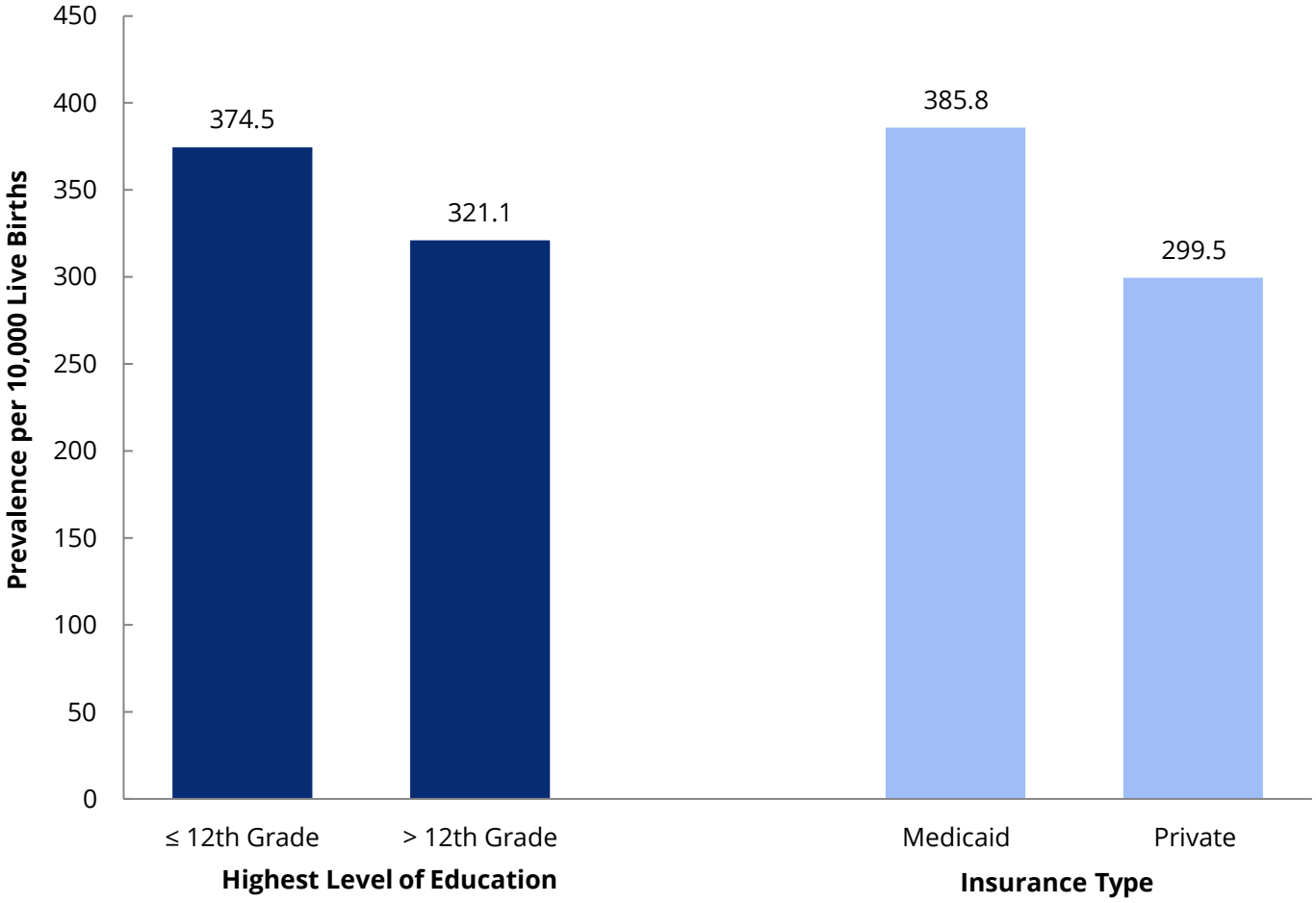


Data Source: Tennessee Department of Health, Tennessee Birth Defects Registry

Other Maternal Characteristics

Birth defect prevalence rates also differed by education and insurance type. Figure 9 shows that birth defect prevalence is higher among babies born to women with ≤ 12 grade education and women on Medicaid. These differences highlight the influence of social determinants of health (such as education and income levels) on health outcomes.

Figure 9. Prevalence of Major Birth Defects by Selected Maternal Characteristics, Tennessee, 2012-2017

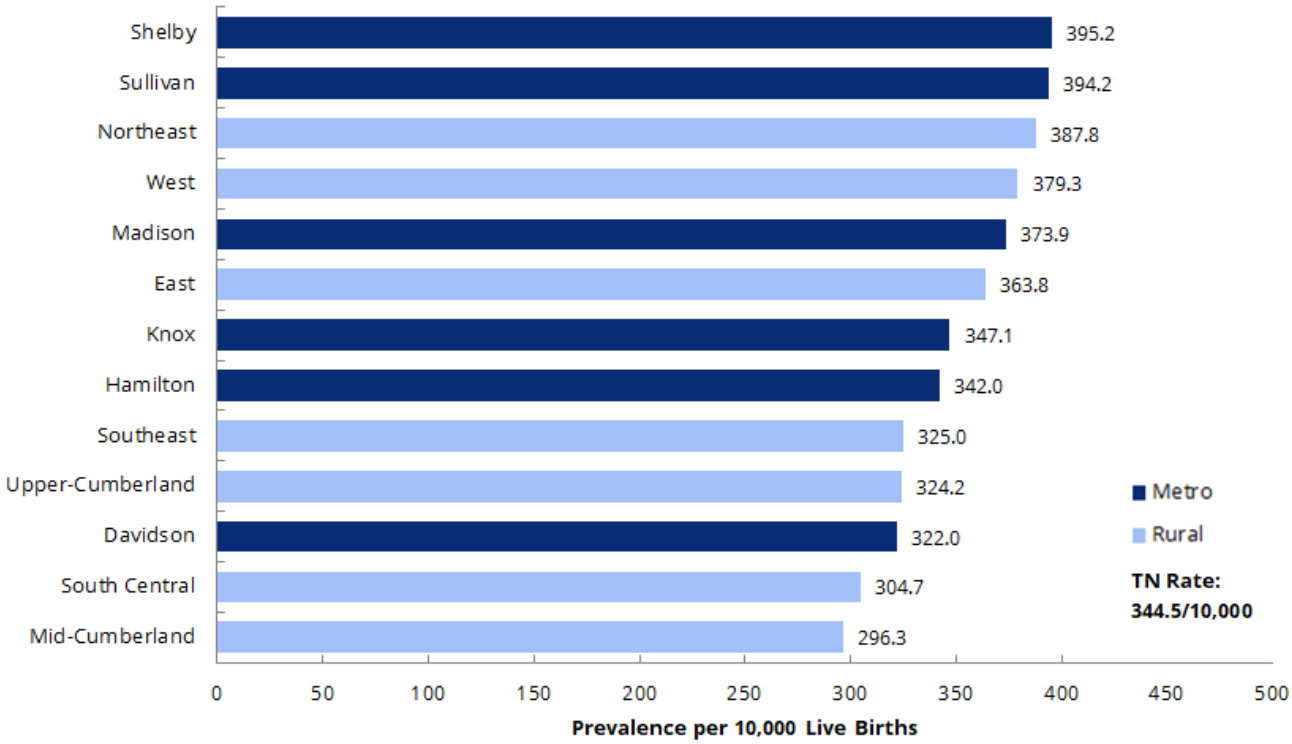


Data Source: Tennessee Department of Health, Tennessee Birth Defects Registry.

Birth Defects by Mother's Residence

Birth defects prevalence rates also differ by mother's residence. Figure 10 shows that birth defects prevalence rates are highest for those living in Shelby County, followed by Sullivan County, the Northeast and West regions.

Figure 10. Prevalence of Major Birth Defects by Maternal Region of Residence, Tennessee, 2012-2017

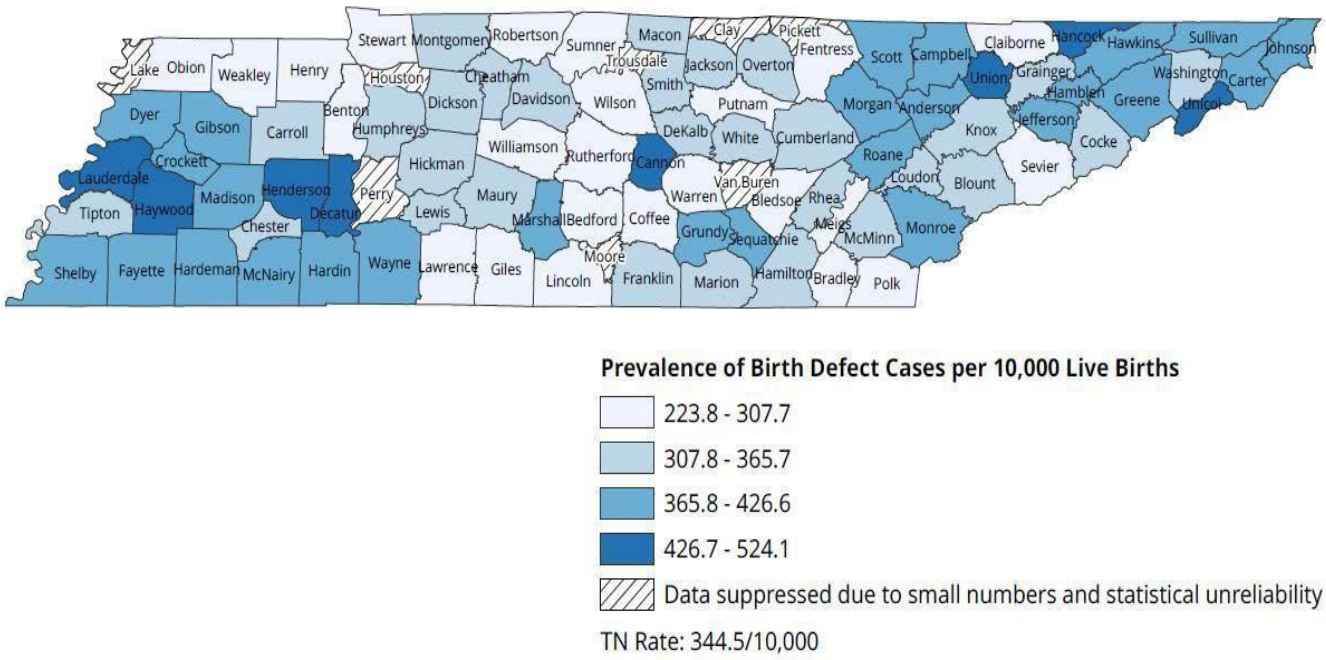


Data Source: Tennessee Department of Health, Tennessee Birth Defects Registry.

Birth Defect Rates by County

Figure 11 features birth defects rates by maternal county of residence. There are variations from one county to another in terms of specific defect rates. These differences may reflect underlying differences in the population and variations in risk factors. For instance, the overall defect rate is 524.1 per 10,000 live births in Unicoi County compared to 223.8 in Polk County. Given that the numbers are generally small the differences in rates should be interpreted with caution. Overall regions calculated to have the highest rates of birth defects include: Shelby County, Sullivan County, and the Northeast region.

Figure 11. Prevalence of Major Birth Defects by County, Tennessee, 2012-2017



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 7 illustrates therelative risk of birth defects by specific 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 8 demonstrates the prevalence of major 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 almost 1.9 times the risk of birth defects, while infants born to mothers with gestational hypertension had almost 1.4 times the risk of birthdefects 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 more than 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 about 1.5 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 7 presents crude relative risks not adjusted for other factors such as maternal age and race that could contribute to the observed associations.

Table 7. Relative Risk of Birth Defects by Maternal Health Characteristics, Tennessee, 2012-2017[1]

Maternal Health Characteristic		Relative Risk of Birth Defects[2]	95% CI[3]
Diabetes	Pre-existing	3.00	2.77-3.26
	Gestational	1.40	1.32-1.48
	None	Reference	–
Hypertension	Pre-existing	1.89	1.76-2.03
	Gestational	1.39	1.32-1.47
	None	Reference	–
Pregnancy Smoking Status[4]	Smoker	1.24	1.19-1.29
	Non-Smoker	Reference	–
BMI	Underweight	1.14	1.06-1.23
	Normal	Reference	–
	Overweight	1.08	1.04-1.12
	Obese	1.30	1.25-1.34
Prenatal Care[5]	Inadequate	1.47	1.41-1.55
	Intermediate	1.15	1.08-1.23
	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 2.77 and 3.26. 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 8. 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, 2012-2017.

Organ System	Pre-Pregnancy Diabetes			Gestational Diabetes			No Diabetes		
	Cases	Rate[1]	Relative Rate[2]	Cases	Rate	Relative Rate	Cases	Rate	Relative Rate
Cardiovascular	413	757.4	3.8	896	315.9	1.6	9010	199.7	Reference
Major Cardiovascular[3]	66	121.0	3.4	149	52.5	1.5	1596	35.4	Reference
Central Nervous System	22	40.3	3.5	34	12.0	1.0	521	11.6	Reference
Genitourinary	77	141.2	2.1	231	81.4	1.2	3020	67.0	Reference
Musculoskeletal	42	77.0	2.0	127	44.8	1.1	1762	39.1	Reference
Orofacial	25	45.8	2.6	52	18.3	1.0	790	17.5	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-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.

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. Accessed September 20, 2020.

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.

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, lowfat dairy, and lean proteins

Avoid harmful substances

- Avoid smoking
- Avoid drinking alcohol
- Avoid drugs such as opioids, marijuana, cocaine, methamphetamines, and other "street" drugs
- 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 (flu vaccine and Tdap vaccine), and after pregnancy
- 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

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 9. Cases and Prevalence of Major Birth Defects and Fetal Alcohol Syndrome, Tennessee, 2012-2017

Birth Defect	Number[1]	Rate[2]	95% CI[3]
Central Nervous System			
Anencephaly	83	1.7	1.4-2.1
Encephalocele	68	1.4	1.1-1.8
Holoprosencephaly	232	4.8	4.2-5.4
Spina bifida without anencephaly	233	4.8	4.2-5.4
Total Central Nervous System Cases	585	12.1	11.1-13.0
Total Central Nervous System Defects	616	12.7	11.7-13.7
Eye/Ear			
Anophthalmia/microthalmia	64	1.3	1.0-1.7
Anotia/microtia	71	1.5	1.1-1.8
Congenital cataract	122	2.5	2.1-3.0
Total Eye and Ear Cases	246	5.1	4.4-5.7
Total Eye and Ear Defects	257	5.3	4.7-5.9
Cardiovascular			
Aortic valve stenosis	80	1.6	1.3-2.1
Atrial septal defect	8,589	177.1	173.3-180.9
Atrioventricular septal defect (Endocardial cushion defect)	284	5.9	5.2-6.5
Coarctation of the aorta	408	8.4	7.6-9.2
Common truncus (truncus arteriosus or TA)	47	1.0	0.7-1.3
Double outlet right ventricle (DORV)	159	3.3	2.8-3.8
Ebstein anomaly	79	1.6	1.3-2.0
Hypoplastic left heart syndrome	182	3.8	3.2-4.3
Interrupted aortic arch (IAA)	165	3.4	2.9-3.9
Pulmonary valve atresia and stenosis	512	10.6	9.6-11.5
Single Ventricle	66	1.4	1.1-1.7
Tetralogy of Fallot (TOF)	299	6.2	5.5-6.9
Total anomalous pulmonary venous connection (TAPVC)	63	1.3	1.0-1.7
Transposition of the great arteries (TGA)	239	4.9	4.3-5.6
Tricuspid valve atresia and stenosis	57	1.2	0.9-1.5
Ventricular septal defect	2,461	50.8	48.7-52.8
Total Cardiovascular Cases	10,321	212.8	208.7-217.0
Total Cardiovascular Defects	13,690	282.3	277.5-275.5
Orofacial			
Choanal atresia	101	2.1	1.7-2.5
Cleft lip with cleft palate	325	6.7	6.0-7.4
Cleft lip alone (without cleft palate)	134	2.8	2.3-3.2
Cleft palate alone (without cleft lip)	311	6.4	5.7-7.1
Total Orofacial Cases	867	17.9	16.7-19.1
Total Orofacial Defects	871	18.0	16.8-19.2

Continued on next page

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

Birth Defect	Number	Rate	95% CI
Gastrointestinal			
Biliary atresia	148	3.1	2.6-3.5
Esophageal atresia/tracheoesophageal fistula	159	3.3	2.8-3.8
Rectal and large intestinal atresia/stenosis	252	5.2	4.6-5.8
Small intestinal atresia/stenosis	250	5.2	4.5-5.7
Total Gastrointestinal Cases	749	15.4	14.3-16.6
Total Gastrointestinal Defects	809	16.7	15.5-17.8
Genitourinary			
Bladder exstrophy	12	0.2	0.1-0.4
Cloacal exstrophy	371	2.1	1.7-2.5
Congenital Posterior Urethral Valves	80	3.2	2.6-4.0
Hypospadias	2,654	106.8	102.7-110.9
Renal agenesis/hypoplasia	325	6.7	6.0-7.4
Total Genitourinary Cases	3,330	68.7	66.3-71.0
Total Genitourinary Defects	3,356	69.2	66.9-71.6
Musculoskeletal			
Clubfoot	958	19.8	18.5-21.0
Diaphragmatic hernia	199	4.1	3.5-4.7
Gastroschisis	256	5.3	4.6-5.9
Limb deficiencies (reduction defects)	207	4.3	3.7-4.9
Omphalocele	130	2.7	2.2-3.1
Craniosynostosis[4]	249	15.4	13.5-17.3
Total Musculoskeletal Cases	1,934	39.9	38.1-41.7
Total Musculoskeletal Defects	1,999	41.2	39.4-43.0
Chromosomal			
Deletion 22q11.2	12	0.2	0.1-0.4
Trisomy 13	56	1.2	0.9-1.5
Trisomy 18	94	1.9	1.6-2.4
Trisomy 21 (Down syndrome)	709	14.6	13.5-15.7
Turner syndrome	46	1.9	1.4-2.6
Total Chromosomal Cases	911	18.8	17.6-20.0
Total Chromosomal Defects	917	18.9	17.7-20.1
Total Birth Defects Cases	16,704	344.5	339.2-349.8
Total Birth Defects	22,515	464.3	458.1-470.5
Fetal Alcohol Syndrome[5]	59	1.2	0.9-1.6

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 10,321 infants were diagnosed with cardiovascular birth defects, but amongst these 10,321 infants, there were 13,690 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 only 2015 and 2016. Prior to 2015, craniosynostosis cases could not be identified. Rate is calculated using live births from 2015 and 2016.
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:
https://vkc.mc.vanderbilt.edu/assets/files/resources/DS%20Guide%20for%20Parents.pdf
Understand Your Patient's Diagnosis of Down Syndrome:
https://vkc.mc.vanderbilt.edu/assets/files/resources/DS%20Guide%20for%20Doctors.pdf
Baby & Me - Tobacco Free™:
https://www.tn.gov/health/health-program-areas/fhw/baby-me-tobacco-free.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

National Resources

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/
MotherToBaby:
https://mothertobaby.org/

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