Data Brief ■ No. 5 ■ February 2021

Birth Defects

Delaware Profile 2010-2017

The data brief provides an overview of birth defects, one of the leading causes of infant mortality in Delaware. It uses the birth defects registry dataset and birth certificate data. The Birth Defects Registry (BDR) was established in 1997, by the Delaware Division of Public Health (DPH); data are collected through medical record review of each potential case by trained BDR abstractors with ascertainment of prevalence for approximately 100 specified birth defects [1]. The Delaware BDR abstractors review medical records on every child who is born in Delaware to a Delaware resident, and who has a reported potential birth defect. Cases are identified from birth hospitals' records, reports from maternal fetal medicine specialists, and discharge records. Case ascertainment of birth defects is consistent with recommendations from the Centers for Disease Control and Prevention (CDC), and from the National Birth Defects Prevention Network (NBDPN).

Overview

As per the most recent data available, approximately three to five percent of births are affected by a birth defect, which are congenital structural and/or genetic conditions [2]. One in five infant deaths in the U.S. are due to birth defects, and birth defects are a significant contributor to lifelong disabilities with inpatient-related costs averaging $23 billion in the U.S. [3]. While environmental factors recognized as teratogens play a role in birth defects, determining a specific cause for a birth defect is often difficult [4-10]. In birth defect etiology, genetic factors are the most common causes of birth defects. One population-based study on the etiology and clinical manifestation of birth defects, found that of the 5,000 cases reviewed, approximately 80% of the cases were due to an 'unknown etiology,' and only 20% could be assigned a 'definite cause' [9]. The risk for birth defects falls in modifiable and nonmodifiable (i.e., age, family history, previous child with a birth defect) category [8, 10]. Identification of modifiable risk factors can be particularly challenging due to ascertainment techniques

Importance

One in every 33 babies (about 3% of all babies) born in the United States each year are affected by a birth defect. A total of 2,948 infants were diagnosed with a birth defect during 2010-2017 in Delaware.

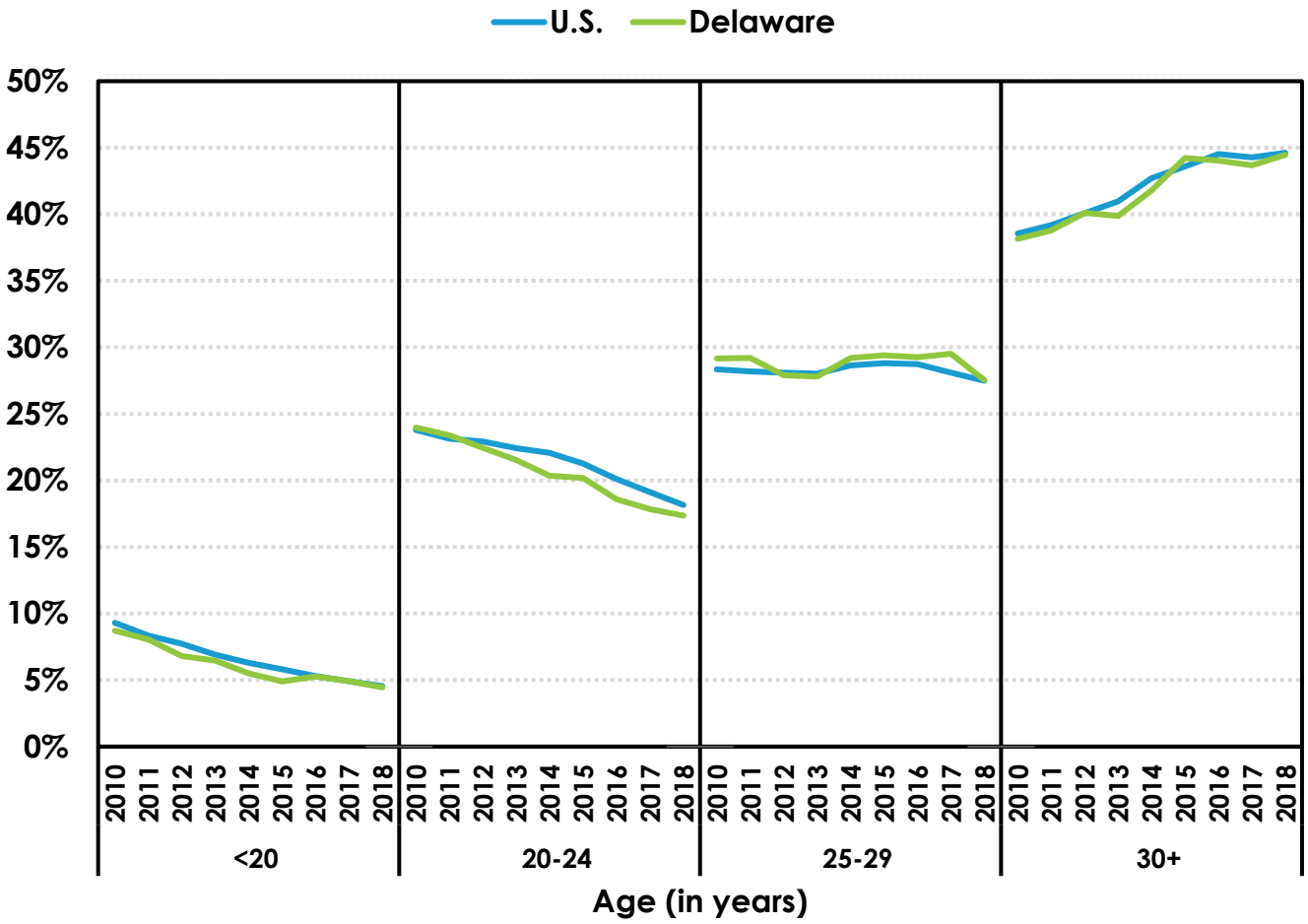
Key findings

- The overall 2010-2017 birth defect prevalence rate in Delaware was 33.4 per 1,000 live births (95% CI: 32.3-34.6) or 3.3 percent and similar to the U.S.
- Of the 2,948 infants who were diagnosed with a birth defect, 2,448 infants (83%) had a single birth defect and one in six (n = 500; 17%) infants with a birth defect had multiple birth defects.
- Birth defects were more common in males (54%) as compared to females (46%). Ventricular septal defect was the most common birth defect in Delaware during 2010-2017.
- Women 35 and older had 20% greater odds of delivering an infant with a birth defect. White non-Hispanic women had 30% greater odds of delivering an infant with a birth defect, and women with no prenatal care had 50% greater odds of delivering an infant with a birth defect.

as most birth defects develop during the first trimester and it is difficult to ascertain early pregnancy exposures [8] during organogenesis (i.e., phase of embryonic development).

There is no national surveillance data registry for birth defects. Thirty-nine states including Delaware have their own independent registries either funded by the Centers for Disease Control and Prevention (CDC) and/or local state agencies that contribute data to the CDC and the National Birth Defects Prevention Network [NBPDN]. Although the prevalence of major birth defects has been stable, some birth defects have shown an increase from 2010-2014 [2].

Figure 1. Percentage of births by age-groups, U.S. and Delaware, 2010-2018



Source: Delaware Department of Health and Social Services, Division of Public Health, Delaware Health Statistics Center, 2010-2018

Figure 1 above provides an overview of U.S. and Delaware births by maternal age. While the proportion of all births to women less than 25 years old has generally declined over the past decade, the proportion of births among women older than 30 has increased and among women 25-29 years of age has remained relatively stable (see figure 1).

Birth defects ascertainment in Delaware

The Birth Defects Registry (BDR) was established in 1997, by the Delaware Division of Public Health (DPH); data are collected through medical record review of each potential case by trained BDR abstractors with ascertainment of prevalence for approximately 100 specified birth defects [1]. The Delaware BDR abstractors review medical records on every child who is born in Delaware to a Delaware resident, and who has a suspected birth defect. Cases are identified using birth hospitals' records, reports from maternal fetal medicine specialists, and discharge records up to one year of age. Case ascertainment of birth defects is consistent with CDC and from the NBDPN [1].

The ascertainment methods for the number of birth defects in Delaware have changed since it was first established in 1997 based on emerging science and technology, recommendations from NBDPN, and resources available. However, the major birth defects reportable to the national registry have remained relatively stable. Not all states who report to the NBDPN use active surveillance methods such as medical chart abstraction and validation to compile the list of core birth defects. Many states utilize passive surveillance methods (i.e., administrative databases) such as the Birth Certificate (BC) and Hospital Discharge (HDD) data as their primary source. The NBDPN annually publishes state-specific birth defect counts and prevalence estimates for 47 major birth defects that cover wide array of organ systems [11] with inherent challenges incumbent in all of these methods.

Appendix 1 provides the current taxonomy of the birth defects ascertained in Delaware by organ systems as well as whether they are structural (i.e., physical abnormalities in the development of body parts including the skeleton and organs), functional (i.e., affecting the function and development of many parts of the body or abnormalities in the systems that run the body like the neurological, immune and endocrine systems), and/or inborn errors of metabolism, endocrine, hemoglobinopathies.

The major birth defects typically reported [11] are shown in the table 1 and the national prevalence estimates for some of the major birth defects derived from 2010-2014 analysis by Mai et al. [2] are also shown. Although no national prevalence estimates for ventricular septal defect (i.e., VSD) are available it is among the most common heart defect. The other common birth defects include: i) clubfoot (1 in 593 births); ii) Trisomy 21or Down Syndrome (1 in 707 births); iii) Pulmonary valve atresia and stenosis (1 in 1,052); iv) cleft lip with cleft palate (1 in 1,563); v) cleft lip alone (without cleft palate 1 in 1,687); vi) coarctation of aorta (1 in 1,795); vii) atrioventricular septal defect (AVSD 1 in 1,853); viii) limb deficiencies (1 in 1,943); and ix) gastroschisis (1 in 1,953). Deletion 22 q11.2 and Turner syndrome are some of the rare birth defects for which prevalence estimates are not available.

Keeping view of the most common birth defects and the prevalence it is important to interpret the prevalence of birth defects in Delaware rates. Birth defect rates are typically reported as prevalence rates [12] per 1,000 or 10,000 live births.

Table 1. Major birth defects by organ systems reported to National Birth Defects Prevention Network

Birth Defect	National Prevalence Estimates
1. Central nervous system	
Anencephaly	1 in every 4,647 births
Encephalocele	1 in every 10,502 births
Holoprosencephaly	Not estimated
Spina bifida without anencephaly	1 in every 2,758 births
2. Eye	
Anophthalmia/microphthalmia	1 in every 5,243 births
Congenital cataract	Not estimated
3. Ear	
Anotia/microtia	Not estimated
4. Cardiovascular	
4a. Primary critical congenital heart defects	
Common truncus	1 in every 15,696 births
Hypoplastic left heart syndrome	1 in every 3,841 births
Pulmonary valve atresia and stenosis	1 in every 1,052 births
Pulmonary valve atresia	1 in every 7,104 births
Tetralogy of Fallot	1 in every 2,171 births
Total anomalous pulmonary venous connection	1 in every 7,809 births
Transposition of the great arteries	1 in every 2,695 births
Dextro-transposition of great arteries	1 in every 3,413 births
4b. Secondary critical congenital heart defects	
Coarctation of aorta	1 in every 1,795 births
Double outlet right ventricle	1 in every 5,997 births
Ebstein anomaly	1 in every 13,047 births
Interrupted aortic arch	1 in every 16,066 births
Single ventricle	1 in every 13,351 births
Tricuspid valve atresia and stenosis	1 in every 5,938 births
4c. Other congenital heart defects	
Aortic valve stenosis	Not estimated
Atrial septal defect	Not estimated
Atrioventricular septal defect	1 in every 1,859 births
Ventricular septal defect	Not estimated
5. Orofacial	
Choanal atresia	Not estimated
Cleft lip alone (without cleft palate)	1 in every 2,807 births
Cleft lip with cleft palate	1 in every 1,563 births
Cleft palate alone (without cleft lip)	1 in every 1,687 births
6. Gastrointestinal	
Biliary atresia	Not estimated
Esophageal atresia/tracheoesophageal fistula	1 in every 4,144 births

Table 1 contd/.

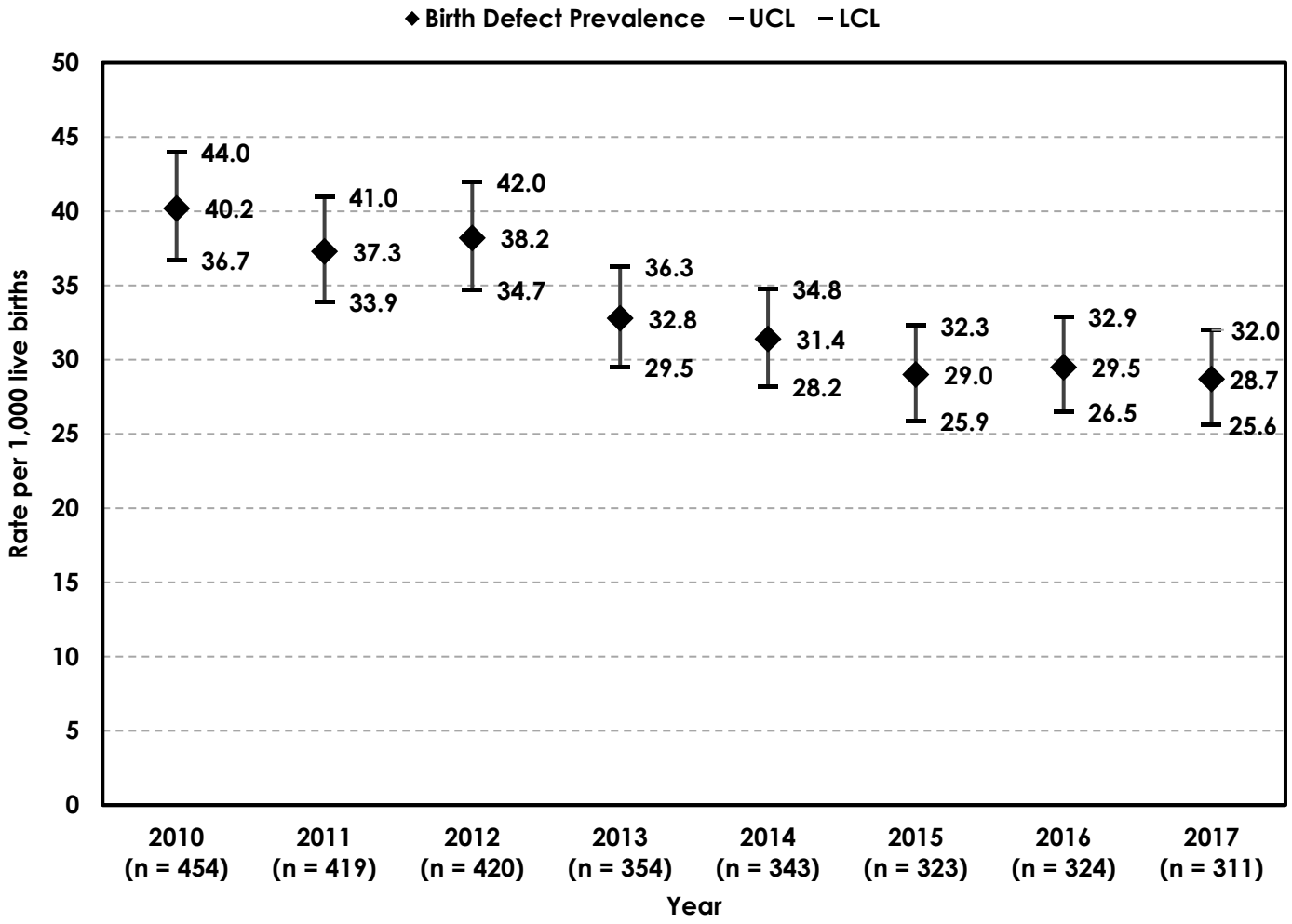
Birth Defect	National Prevalence Estimates
Rectal and large intestinal atresia/stenosis	1 in every 2,242 births
Small intestinal atresia/stenosis	Not estimated
7. Genitourinary	
Bladder exstrophy	Not estimated
Cloacal exstrophy	Not estimated
Congenital posterior urethral valves	Not estimated
Hypospadias	Not estimated
Renal agenesis/hypoplasia	Not estimated
8. Musculoskeletal	
Clubfoot	1 in every 593 births
Diaphragmatic hernia	1 in every 3,591 births
Gastroschisis	1 in every 1,953 births
Limb deficiencies (reduction defects)	1 in every 1,943 births
Omphalocele	1 in every 4,175 births
9. Chromosomal	
Deletion 22 q11.2	Not estimated
Turner syndrome	Not estimated
Trisomy 13 (Patau syndrome)	1 in every 7,409 births
Trisomy 18 (Edwards syndrome)	1 in every 3,315 births
Trisomy 21 (Down syndrome)	1 in every 707 births

Notes: Birth defects typically reported to NBPDN are from Salemi et al. (2019), National estimates are from Centers for Disease Control and Prevention available at https://www.cdc.gov/ncbddd/birthdefects/data.html from 2010-2014 analysis of birth defects by Mai et al. (2019).

Figure 2 displays the annual birth defect prevalence rates in Delaware between 2010 to 2017. While it seems that the birth defect rates have declined during 2010 to 2017 time-period from 40.2 per 1,000 live births in 2010 to 28.7 in 2017, the decline is specific to the number of birth defect cases ascertained during 2010-2017. The differences in the number of birth defects are reflective of changes in reportable conditions by NBDPN, emerging science for addition or deletion of specific birth defects but not necessarily related to the core birth defects.

During 2010-2017, there were 2,948 infants who were diagnosed with a birth defect. Of the 2,948 infants who were diagnosed with birth defects, 1,607 (54.5%) were males, and 1,341 (45.5%) were females. Of the 2,948 infants with birth defects, 2,833 (96.1%) were live births, followed by 53 (1.8%) infant deaths, 52 (1.8%) fetal deaths, and 10 (0.3%) terminations. The overall 2010-2017 birth defect prevalence rate in Delaware was 33.4 per 1,000 live births (95% CI: 32.3-34.6) or 3.3 percent. According the CDC, three percent of all live births have a birth defect [3] and Delaware's birth defect rate is similar to the U.S.

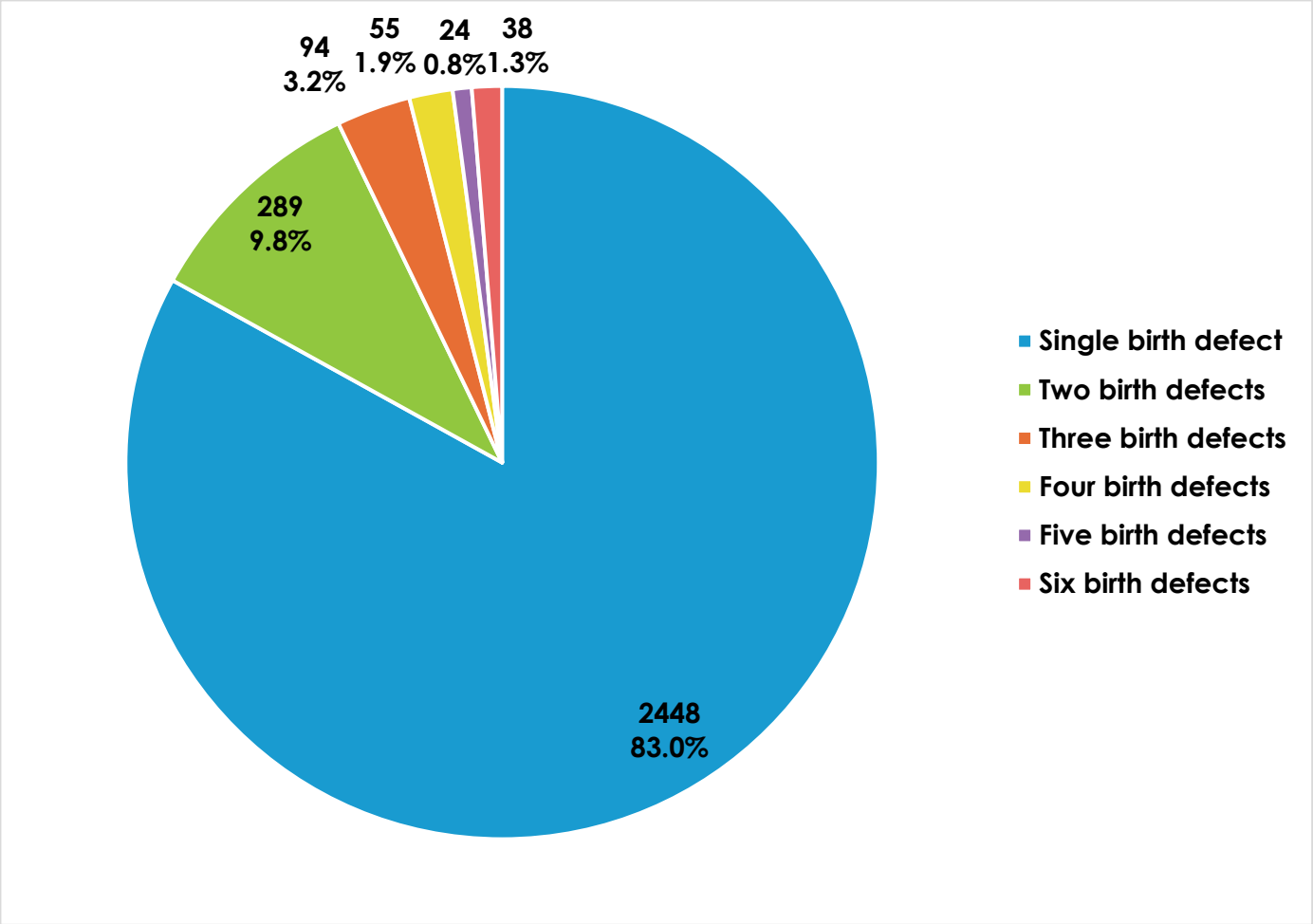
Figure 2. Birth defect prevalence in Delaware with 95% Confidence Intervals, 2010-2017



Source: Delaware Health and Social Services, Division of Public Health, Delaware Health Statistics Center, and Birth Defects Registry Data, 2010-2017.
Notes: Rates are presented with 95% confidence intervals; UCL = upper confidence limit; LCL = lower confidence limit. In birth defects surveillance and prevalence studies prevalence rates are calculated per 1000 live births. Birth defect cases are numerators and all live births comprise the denominator (i.e., a ratio instead of a rate).

Figure 3 displays the total number of birth defects in an infant in Delaware during 2010-2017. Of the 2,948 infants who were diagnosed with a birth defect, 2,448 infants (83.0%) had a single birth defect. Although majority of the infants had a single birth defect, one in ten (n = 289; 9.8%) infants had two birth defects, and about seven percent (n = 211) had three or more birth defects. In Delaware, one in six infants with birth defects had multiple birth defects (n = 500; 17.0%). While age was not associated with multiple birth defects, race and ethnicity was associated with multiple birth defects. Of the 500 infants with multiple birth defects (i.e., 2 or more), Hispanic infants shared a disproportionate burden (n = 88 or 23.6%; 88/372) as compared to all other race and ethnic groups (n = 410 or 16.0%; 410/2,558).

Figure 3. Total number and proportion of birth defects in an infant in Delaware, 2010-2017

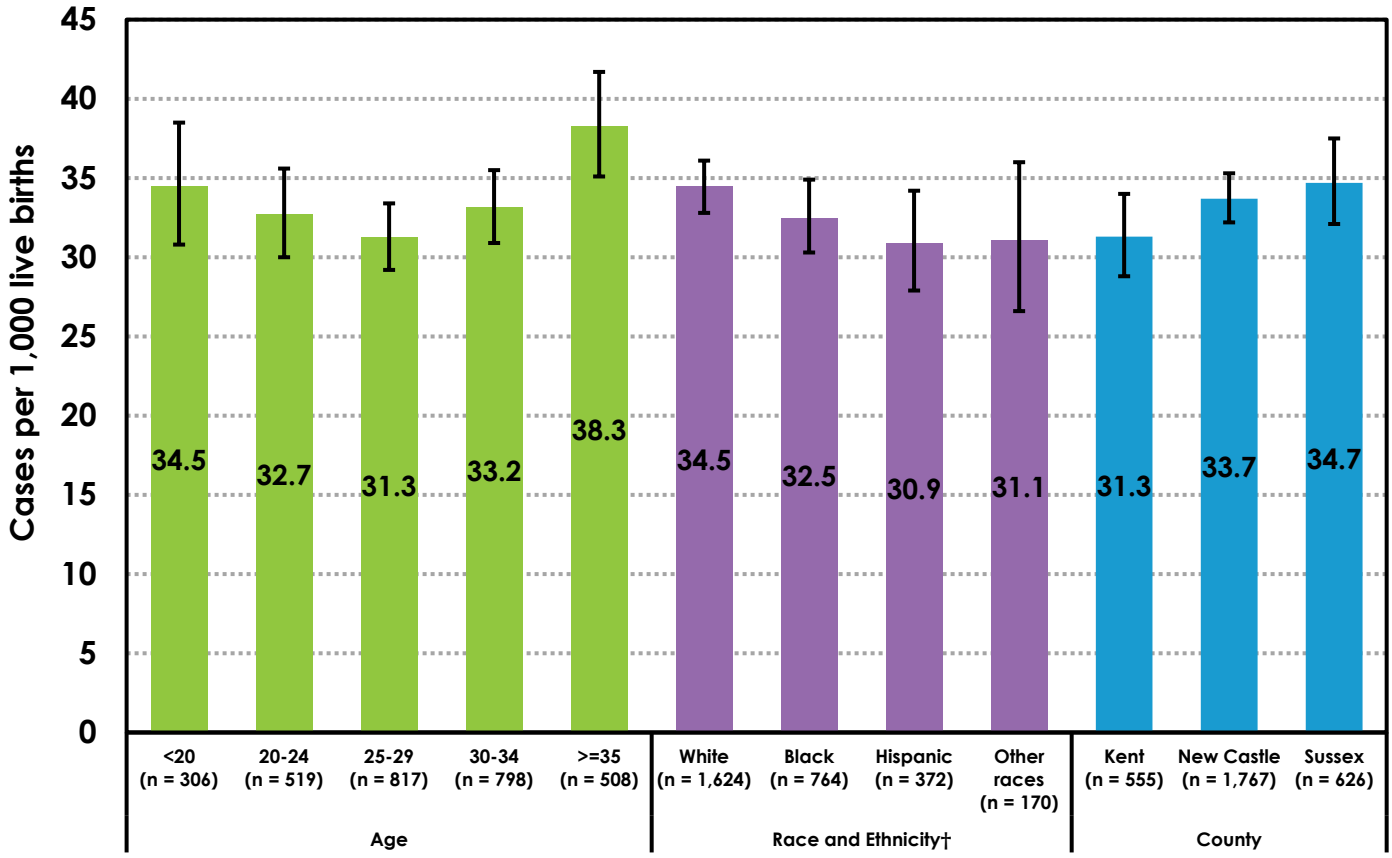


Source: Delaware Health and Social Services, Division of Public Health, Delaware Health Statistics Center, and Birth Defects Registry Data, 2010-2017.

Figure 4 displays overall birth defect rates by mother's age, race and ethnicity, and county of residence for 2010-2017. The rates of birth defects were higher at the extremes of maternal age. For instance, the prevalence of birth defects in infants among mothers: was 34.5 (95% CI: 30.8-38.5) who were less 20 years of age; 32.7 (95% CI: 30.0-35.6) who were 20-24 years of age; 31.3 (95% CI: 29.2-33.4) who were 25-29 years of age; 33.2 (95% CI: 30.9-35.5) who were 30-34 years of age; and 38.3 (95% CI: 35.1-41.7) per 1,000 live births who were 35 and older.

With regards to maternal race and ethnicity, prevalence of birth defects was higher among White (non-Hispanic) 34.5 (95% CI: 32.8-36.1), followed by Black (non-Hispanic) 32.5 (95% CI: 30.3-34.9), followed by other races 31.1 (95% CI: 26.6-36.0) that include Asian (non-Hispanic), American Indians and Alaska Natives, Native Hawaiian and Pacific Islanders, and multiple races. Hispanics had the lowest prevalence rates of birth defects 30.9 (95% CI: 27.9-34.2) per 1,000 live births. Prevalence of birth defects was highest in Sussex county 34.7 (95% CI: 32.1-37.5), followed New Castle county 33.7 (95% CI: 32.2-35.3), and Kent county 31.3 (95% CI: 28.8-34.0) per 1,000 live births. Appendix 2 provides birth defect rates in Delaware by zip code tabulation areas (ZCTAs).

Figure 4. Overall birth defect prevalence rates stratified by maternal age, maternal race and ethnicity, and county of residence in Delaware with 95% Confidence Intervals, 2010-2017



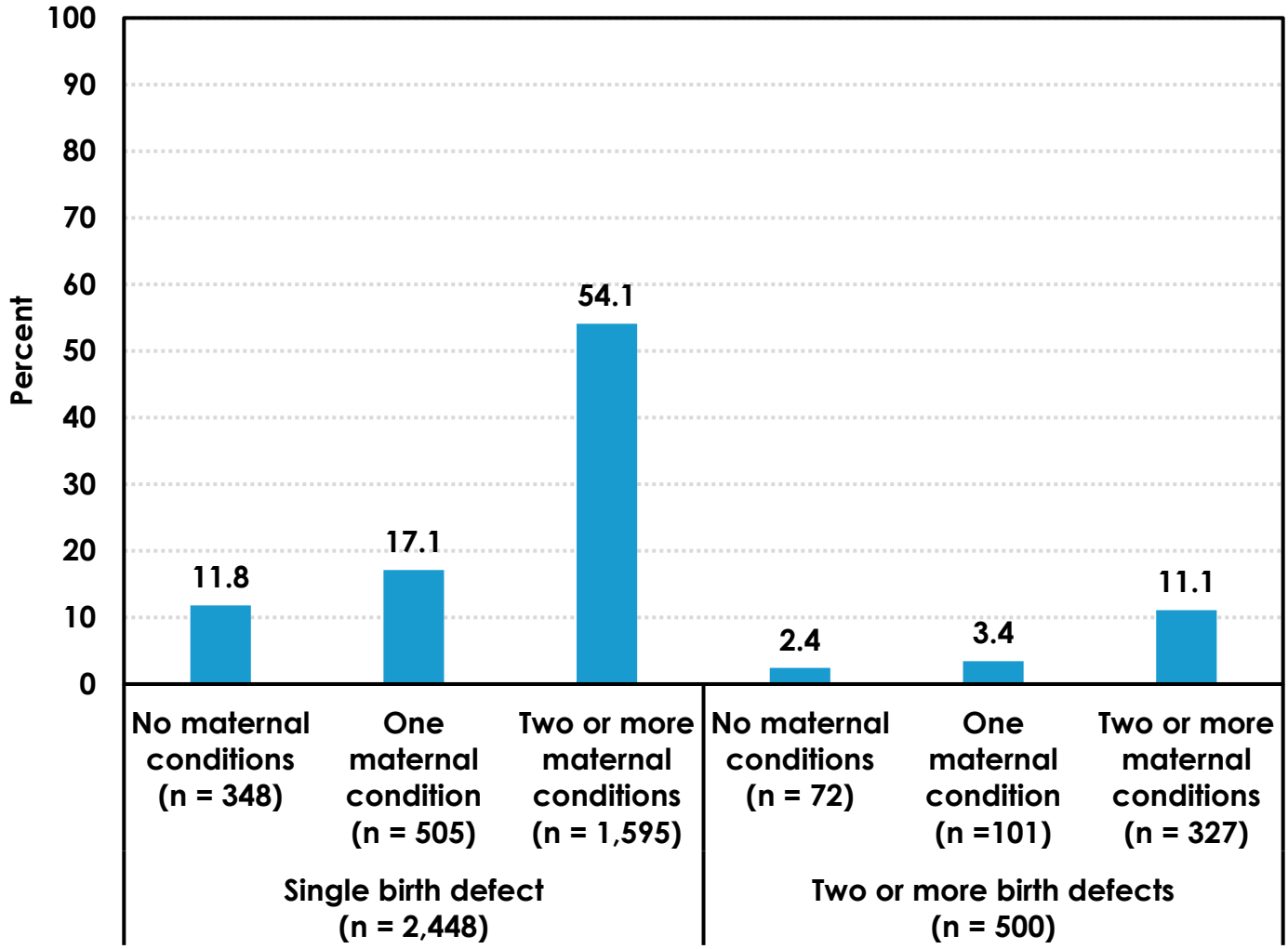
Source: Delaware Health and Social Services, Division of Public Health, Delaware Health Statistics Center, and Birth Defects Registry Data, 2010-2017.
Notes: Rates are presented with 95% confidence intervals; UCL = upper confidence limit; LCL = lower confidence limit. In birth defects surveillance and prevalence studies prevalence rates are calculated per 1000 live births. Birth defect cases are numerators and all live births comprise the denominator (i.e., a ratio instead of a rate).
†White, Black, Other categories of race exclude Hispanic ethnicity

The birth defect registry captures maternal conditions of women who delivered an infant with a birth defect that are available and documented on the medical record. These conditions may include any surgical procedures performed during delivery, chronic conditions such as hypertension, diabetes, depression, psychiatric disorders (i.e., bi-polar disorders, schizophrenia, etcetera.), thyroid disorders, heart disease, lupus, and obesity, conditions originating during pregnancy such as pregnancy-induced hypertension, gestational diabetes, substance use related conditions such as alcohol use, tobacco use, and illicit drug use, and other conditions.

Figure 5 displays maternal conditions of women who delivered an infant with a birth defect. It is evident that irrespective of the number of birth defects, most women had two or more maternal conditions documented in their medical record. For instance, of the 2,448 infants with a single birth

defect, 2,100 (i.e., 505 + 1,595) cases had one or more maternal conditions documented and 348 cases had no maternal conditions documented in their maternal record. Similarly, of the 500 cases with two or more birth defects, 428 (i.e., 101 + 327) cases had one or more maternal conditions documented and 72 cases had no maternal conditions documented in their maternal record. In total, 420 cases (348 + 72 or ~14%) had no maternal conditions documented and 2,528 (i.e, 2,100 + 428 or ~86%) cases had documented maternal conditions.

Figure 5. Number and percentage of maternal conditions among women who delivered an infant with birth defects in Delaware, 2010-2017



Source: Delaware Health and Social Services, Division of Public Health, Delaware Health Statistics Center, and Birth Defects Registry Data, 2010-2017.

Excluding 724 cases (28.6%) that documented surgical intervention/surgery, the most commonly documented medical conditions included: 1) obesity (n = 299; 11.8%); 2) alcohol use (n = 256; 10.1%); 3) depression (n = 171; 6.8%); 4) tobacco use (n = 171; 6.8%); 5) gestational diabetes (n = 136; 5.4%); 6) pregnancy-induced hypertension (n = 112; 4.4%); 7) illicit drugs (n = 99; 3.9%); 8) chronic hypertension (n = 90; 3.6%); 9) placenta previa (n = 83; 3.3%); 10) hypothyroidism (n = 56; 2.2%). These 10 conditions accounted for 58% of all maternal conditions. One in four women (25% = 11.8% obesity + 5.4% gestational diabetes + 4.4% pregnancy induced hypertension + 3.6% chronic hypertension) had obesity-related morbidities. Similarly, one in five women (20% = 10.1% alcohol use + 6.8% tobacco use + 3.9% illicit drugs) who delivered infants with a birth defect had documented alcohol, tobacco, or other drugs (ATOD) in their medical record.

Common birth defects in Delaware 2010-2017

The following analyses focuses on 2,448 infants with a single birth defect (see figure 3) that account for over 80 percent of the reported birth defect cases. Table 2 presents the top 20 birth defects in Delaware during 2010-2017 with counts, percent, and the prevalence estimates with 95% confidence intervals. Of the 2,448 single birth defects, the top 20 birth defects accounted for 74 percent (1,808) of all birth defects.

Of the top 20 birth defects, 11 birth defects are part of the NBDPN major birth defect categories: 1) ventricular septal defect; 2) hypospadias; 3) clubfoot; 4) atrial septal defect; 5) trisomy; 6) gastroschisis; 7) pulmonary valve atresia and stenosis; 8) single gene disorder; 9) renal agenesis/hypoplasia; 10) cleft lip with cleft palate; and 11) cleft palate alone without cleft lip. The remaining nine: 1) developmental hip dysplasia; 2) sickle cell disease; 3) cystic dysplastic kidneys; 4) congenital hypothyroidism; 5) pyloric stenosis; 6) craniosynostosis; 7) microcephalus; 8) other specified anomalies and syndromes; and 9) obstructive genitourinary defect are captured as part of Delaware's birth defect registry. The following paragraphs discuss the top 10 birth defects in Delaware and compares it to available NBDPN estimates.

During 2010-2017 there were 481 cases of ventricular septal defect (VSD) identified among Delaware infants. Ventricular septal defect, a congenital heart defect, was one of the top 20 birth defects that accounted for 20 percent (481/2,448) of single birth defects with an overall prevalence of 5.5 per 1,000 live births (95% CI: 5.0-6.0). While there is currently no national estimate for VSDs, the prevalence of VSDs has been generally higher in Delaware with one previous study estimating VSDs at 8.3 per 1,000 live births or 83.4 per 10,000 live births during 2007-2010 [1]. The study also noted that Delaware includes all types of VSDs and when small muscular VSDs are excluded the prevalence is similar to other states and ranges between 1.6 to 70.0 per 10,000 live births [1].

There were 331 cases of hypospadias identified among Delaware male infants that accounted for 14 percent (331/2,448) of the top 20 single birth defects with an overall prevalence of 3.8 per 1,000 live births (95% CI: 3.4-4.2). The prevalence of hypospadias is similar to the North America prevalence estimate of 3.4 per 1000 live births or 34.2 per 10,000 live births [13].

Developmental hip dysplasia or developmental dysplasia of the hip (DDH) was diagnosed among 161 (6.7%) Delaware infants having a singular birth defect with an overall prevalence of 1.9 per 1,000 live births (95% CI: 1.6-2.2), slightly higher as compared to the estimates of 1 in 1,000 live births reported by Shaw et al. [14].

Clubfoot was among the top four birth defects with a total of 99 cases (4.0%) with an overall prevalence of 1.1 per 1,000 live births (95% CI: 0.9-1.4). The Delaware prevalence for clubfoot was similar to that of the U.S. — 1.3 per 1,000 live births [15].

Sickle cell disease (SCD) was among the top five birth defects with a total of 93 cases (3.8%) with an overall prevalence of 1.1 per 1,000 live births (95% CI: 0.9-1.3). According to the National Center on Birth Defects and Development Disorders (NCBDD) at the CDC, SCD is a genetic condition and is present at birth and inherited when a child receives two sickle cell genes, one from each parent. It is unlike sickle cell trait (SCT), which is not a disease [16].

Atrial septal defect (ASD), not the same as atrioventricular septal defect (AVSD or endocardial cushion effect) was among the top six birth defects with a total of 89 cases (3.6%) with an overall prevalence of 1.0 per 1,000 live birth (95% CI: 0.8-1.2). While there is no current national estimate for ASD, the Delaware's ASD prevalence is similar to the worldwide prevalence of 1.6 [17].

Cystic dysplastic kidneys (i.e., malformed kidney with cysts) was diagnosed among 71 infants having a singular birth defect (2.9%) in Delaware during 2010-2017 with an overall prevalence of 0.8 per 1,000 live births (95% CI: 0.6-1.0). According to the National Institute of Diabetes and Digestive and Kidney Diseases (NIDDK), about one in 4,000 infants are affected by kidney dysplasia [18].

Trisomy which is a chromosomal anomaly and includes trisomy 13 (i.e. Patau's syndrome), trisomy 18 (i.e., Edward's syndrome), and trisomy 21 (i.e., Down syndrome) was the eighth top birth defect in Delaware and was diagnosed among 66 infants having a singular birth defect (2.7%) with an overall prevalence of 0.7 per 1,000 live births (95% CI: 0.6-1.0). Of the 66 cases, 63 cases were specific to Down syndrome, one case was specific to Patau's syndrome, and two cases were specific to Edward's syndrome. The national estimate for Down syndrome is about one in 700 births [12] slightly higher as compared to Delaware.

Gastroschisis was the ninth top condition in Delaware and was diagnosed among 44 infants having a singular birth defect (1.8%) with an overall prevalence of 0.5 per 1,000 live births (95% CI: 0.4-0.7) during 2010-2017. Delaware's prevalence of gastroschisis was similar to the national prevalence which is estimated at 5.4 per 10,000 live births or 0.5 per 1,000 live births [12].

Congenital hypothyroidism (CHT) is a common endocrine abnormality and was the 10th top birth defect. While estimates suggest that two to five percent of CHT is inherited and there is a genetic component, most often the cause of CHT is the shortage of iodine in diet [19]. As per national estimates, CHT impacts one in 2,000 to 4,000 live births. During 2010-2017, there were a total of 43 cases (1.8%) of infants diagnosed with CHT with an overall prevalence of 0.5 per 1,000 live birth (95% CI: 0.4-0.7) and Delaware's prevalence was similar to the national prevalence.

Table 2. Top 20 birth defects by category, count, and prevalence in Delaware, 2010-2017

Birth defect	Birth defect category	Count (%)	Prevalence (95 % CI)
1. Ventricular septal defect	Other congenital heart defects	481 (19.7%)	5.5 (5.0-6.0)
2. Hypospadias	Genitourinary	331 (13.5%)	3.8 (3.4-4.2)
3. Developmental hip dysplasia*	Musculoskeletal	164 (6.7%)	1.9 (1.6-2.2)
4. Clubfoot	Musculoskeletal	99 (4.0%)	1.1 (0.9-1.4)
5. Sickle cell disease*	Hemoglobinopathies	93 (3.8%)	1.1 (0.9-1.3)
6. Atrial septal defect	Other congenital heart defects	89 (3.6%)	1.0 (0.8-1.2)
7. Cystic dysplastic kidneys*	Genitourinary	71 (2.9%)	0.8 (0.6-1.0)
8. Trisomy	Chromosomal	66 (2.7%)	0.7 (0.6-1.0)
9. Gastroschisis	Musculoskeletal	44 (1.8%)	0.5 (0.4-0.7)
10. Congenital hypothyroidism*	Endocrine, Metabolic, Immunodeficiency	43 (1.8%)	0.5 (0.4-0.7)
11. Pyloric stenosis*	Genitourinary	42 (1.7%)	0.5 (0.3-0.6)
12. Pulmonary valve atresia and stenosis	Primary critical congenital heart defects	41 (1.7%)	0.5 (0.3-0.6)
13. Single gene disorder	Single gene disorder	40 (1.6%)	0.5 (0.3-0.6)
14. Craniosynostosis*	Musculoskeletal	38 (1.6%)	0.4 (0.3-0.6)
15. Microcephalus*	Central nervous system	35 (1.4%)	0.4 (0.3-0.6)
16. Renal agenesis/hypoplasia	Genitourinary	31 (1.3%)	0.4 (0.2-0.5)
17. Other specified anomalies and syndromes*	Other birth defects	28 (1.1%)	0.3 (0.2-0.5)
18. Obstructive genitourinary defect*	Genitourinary	26 (1.1%)	0.3 (0.2-0.4)
19. Cleft lip with cleft palate	Orofacial	25 (1.0%)	0.3 (0.2-0.4)
20. Cleft palate alone (without cleft lip)	Orofacial	21 (0.9%)	0.2 (0.1-0.4)
Top 20 total†		**1,808 (73.9%)**	**20.5 (19.6-21.5)**
Single birth defects§		**2,448 (83.0%)**	**27.8 (26.7-28.9**
All birth defects (includes single and multiple)		**2,948 (100%)**	**33.4 (32.3-34.6)**

Source: Delaware Health and Social Services, Division of Public Health, Delaware Health Statistics Center, and Birth Defects Registry Data, 2010-2017.

Notes: Prevalence estimates are based on overall counts of birth defects during 2010-2017 per 1,000 live births. 95% confidence intervals (CI) are based on Poisson distribution.

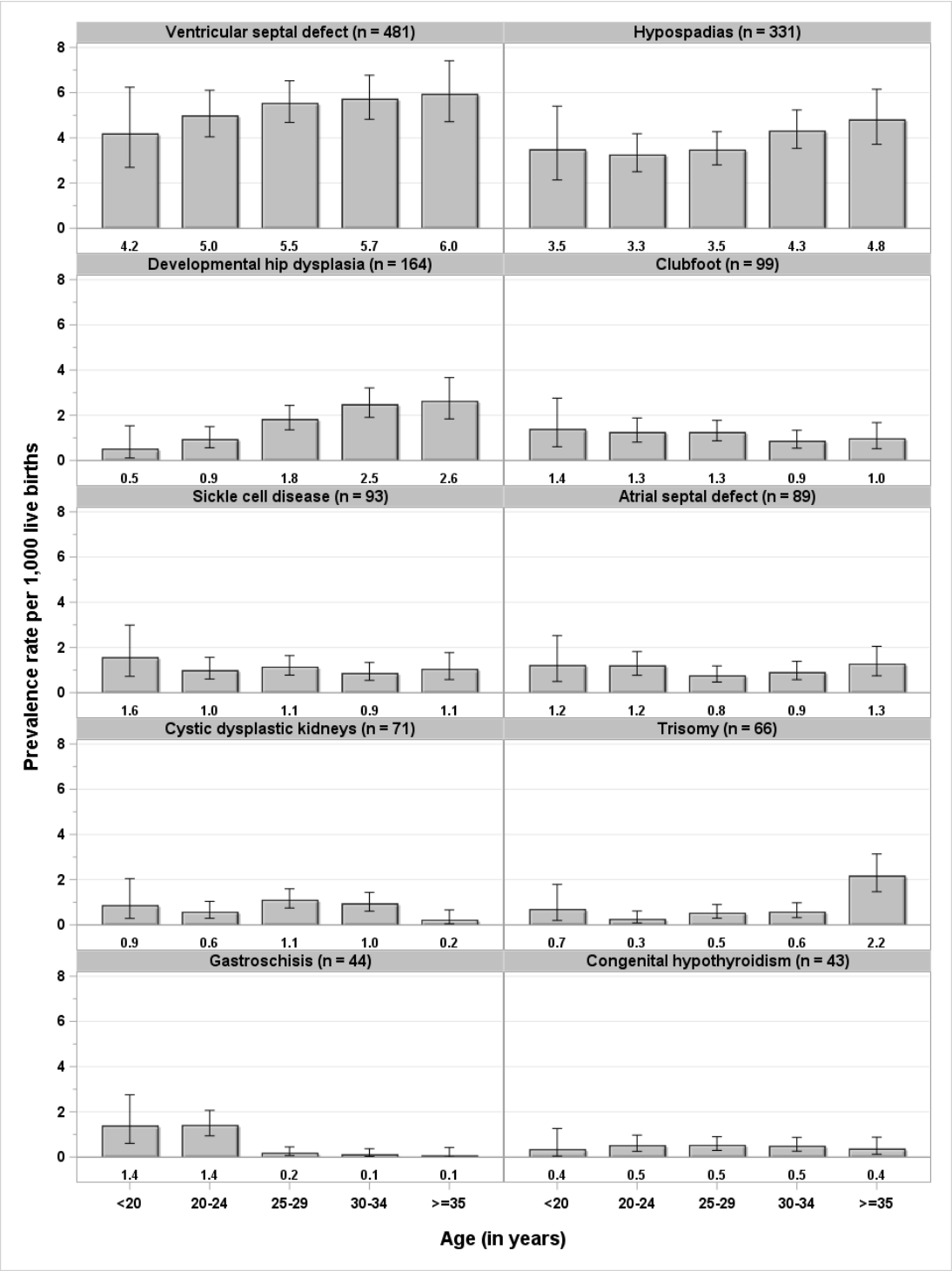
*Not part of the National Birth Defects Prevention Network (NBDPN) and CDC major birth defect category.

†Top 20 birth defects are based on single birth defects (1,808/2,448).

§Single birth defects (2,448/2,948).

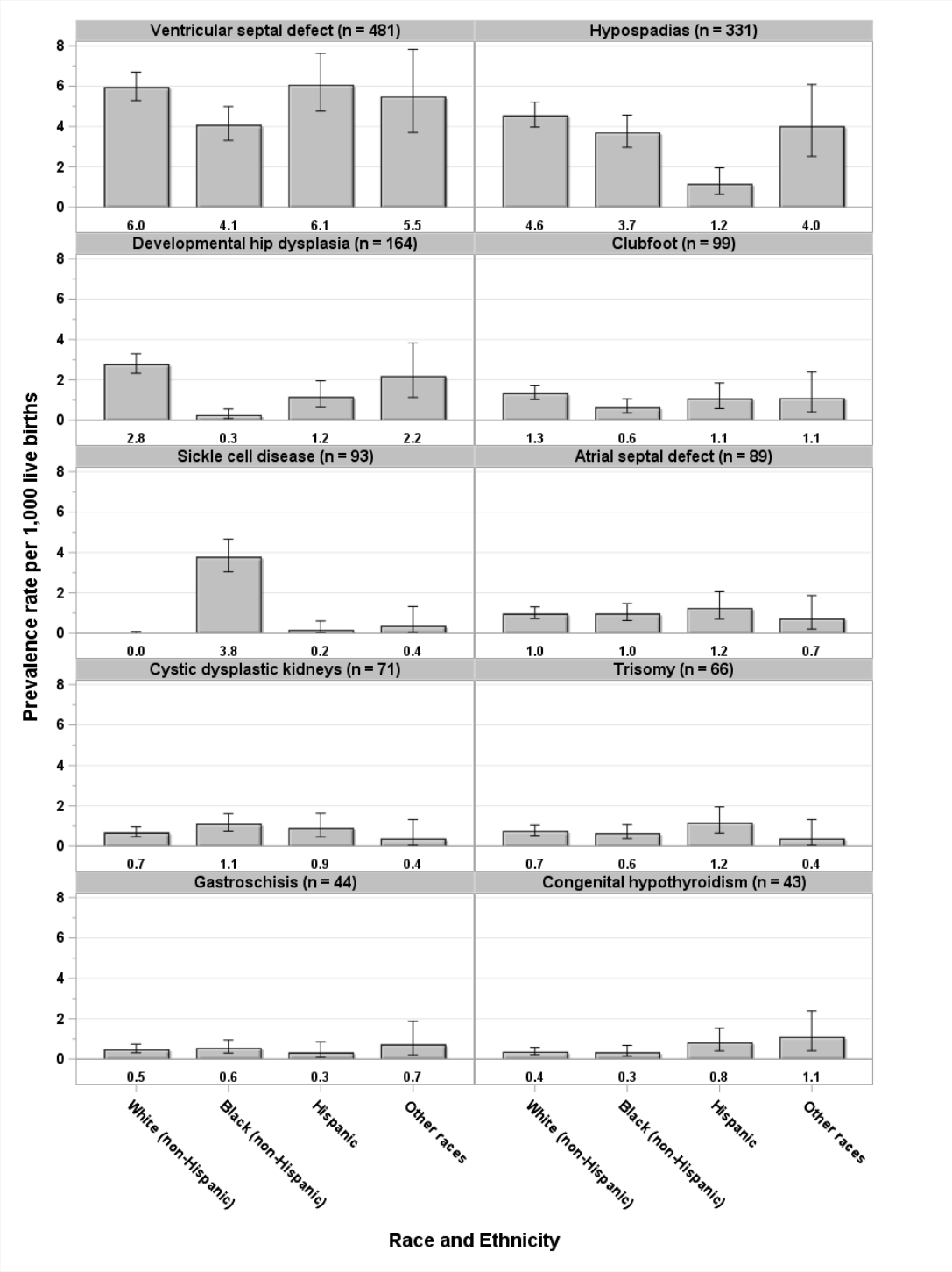
To better understand the characteristics of the infants diagnosed with birth defects, the top 10 birth defects in Delaware were stratified by maternal age, race and ethnicity, and county of residence.

Figure 6. Top 10 most prevalent in Delaware stratified by maternal age with 95% confidence intervals, 2010-2017



Source: Delaware Health and Social Services, Division of Public Health, Delaware Health Statistics Center, and Birth Defects Registry Data, 2010-2017.
Notes: Prevalence rates are based on overall counts of birth defects during 2010-2017 per 1,000 live births. 95% confidence intervals (CI) are based on Poisson distribution.

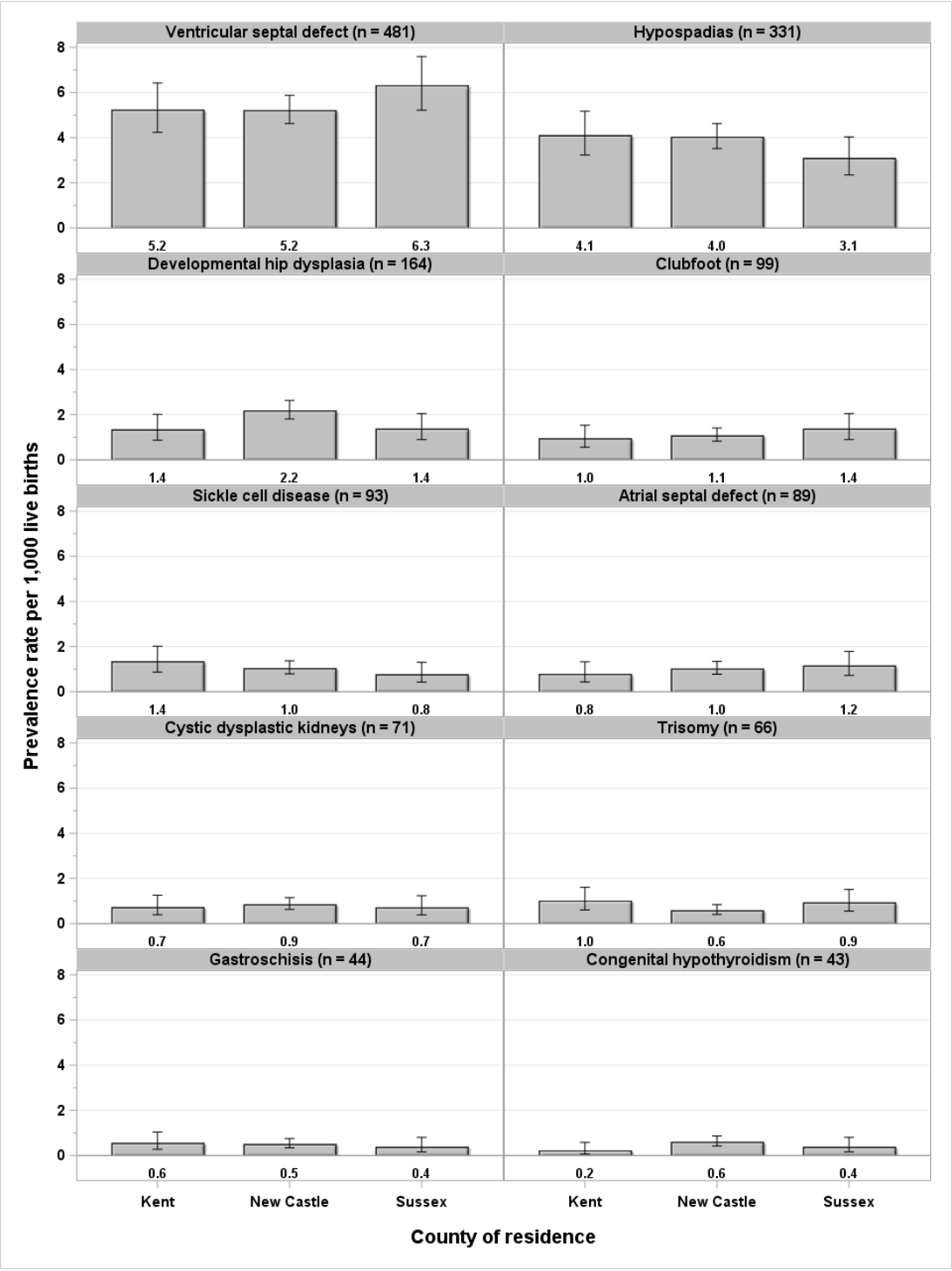
Figure 7. Top 10 most prevalent in Delaware stratified by maternal race and ethnicity with 95% confidence intervals, 2010-2017



Source: Delaware Health and Social Services, Division of Public Health, Delaware Health Statistics Center, and Birth Defects Registry Data, 2010-2017.

Notes: Prevalence rates are based on overall counts of birth defects during 2010-2017 per 1,000 live births. 95% confidence intervals (CI) are based on Poisson distribution.

Figure 8. Top 10 most prevalent in Delaware stratified by county of residence with 95% confidence intervals, 2010-2017



Source: Delaware Health and Social Services, Division of Public Health, Delaware Health Statistics Center, and Birth Defects Registry Data, 2010-2017.
Notes: Prevalence rates are based on overall counts of birth defects during 2010-2017 per 1,000 live births. 95% confidence intervals (CI) are based on Poisson distribution.

As per the figures 6, 7, and 8 the top 10 birth defects varied by maternal age, race and ethnicity, and county of residence. For example, rates of VSD, hypospadias, and DDH increased with maternal age. However, this pattern was not consistent for all birth defects. Trisomy rates were highest among mothers who were 35 and older (2.2 per 1,000 live births; 95% CI: 1.5-3.1) per 1,000 live births, while gastroschisis rates were highest among mothers who were less than 25 years old (1.4 per 1,000 live births; 95% CI: 0.9-2.0). With regards to race and ethnicity, rates of VSD, DDH, and clubfoot were lowest among African Americans, while prevalence rate of SCD was highest (3.8 per 1,000 live births; 95% CI: 3.0-4.7). Among Hispanics, prevalence rate of trisomy was highest (1.2 per 1,000 live births; 95% CI: 0.6-2.0) and the rate of hypospadias was lowest (1.2 per 1,000 live births; 95% CI: 0.6-2.0). Although there was no discernible pattern with regards to county of residence, VSD prevalence rates were higher in Sussex county (6.3 per 1,000 live births; 95% CI: 5.2-7.6) and DDH rates were higher in Kent county (2.2 per 1,000 live births; 95% CI: 1.8-2.6). The next section describes the characteristics of infants with and without a birth defect using a linked dataset of birth defects registry and birth certificates that excluded fetal deaths and terminations. The dataset for 2010-2017 included 81,287 hospital births with 2,784 birth defects and 78,503 no birth defects [20].

Characteristics of infants with and without birth defects in Delaware 2010-2017

Table 3 displays the maternal and infant characteristics of infants with and without birth defects in Delaware during 2010-2017. There were statistically significant differences in maternal age, race and ethnicity, cigarette use during pregnancy, trimester of prenatal care of mothers who delivered with and without birth defects. For instance, mothers who delivered infants with birth defects were slightly older (17.6%) as compared to mothers who delivered infants without birth defects (14.7%). There was a large percentage of White (non-Hispanic) mothers who delivered infants with birth defects (56.8%) as compared to those without birth defects (52.8%). The percentage of mothers who smoked during pregnancy who delivered an infant with a birth defect (12.2%) was slightly higher as compared to those who did not deliver an infant with a birth defect (11.1%). There was a slightly higher percentage of mothers who delivered an infant with a birth defect without prenatal care (3.7%) as compared to mothers who delivered an infant without a birth defect (2.5%). Although there were statistically significant differences in maternal characteristics of mothers who delivered infants with a birth defect, these differences were not large. Mothers with multiple gestation (i.e., twin pregnancy or higher) had a slightly larger proportion of infants with birth defects (4.1%) as compared to those mothers who delivered an infant without a birth defect (3.2%).

However, with regards to infant outcomes such as birth weight, preterm birth, small for gestational age (e.g., intrauterine growth restriction), and infant death the differences were large and statistically significant. For instance, it is well-established that infants with birth defects are more likely to be growth restricted and as such are premature, have low birth weight, and have higher mortality rates as compared to infants without birth defects [21-22]. In Delaware one in five infants with a birth defect was low birth weight (20.5% vs. 8.3%), was born premature (20.1% vs. 9.4%), and was small for gestation (20.2% vs. 12.3%). Consequently, infants with birth defects also had higher rates of mortality (3.5%) as compared to infants without a birth defect (0.6%).

Table 3. Maternal and infant characteristics of infants with and without birth defects in Delaware, 2010-2017

Maternal and infant characteristics	Birth Defect (N = 81,287)	
	Yes (n = 2,784) Number (%)	No (n = 78,503) Number (%)
Maternal age (in years)		
< 20	176 (6.3)	5,314 (6.8)
20-24	611 (21.9)	17,269 (22)
25-29	762 (27.4)	23,249 (29.6)
30-34	746 (26.8)	21,118 (26.9)
35 or more	489 (17.6)	11,553 (14.7)
Maternal education		
< 9 years of schooling	144 (5.2)	4,268 (5.4)
9 – 11 years of schooling	329 (11.8)	10,004 (12.7)
High school graduate	753 (27)	20,512 (26.1)
1 – 3 years of college	742 (26.7)	20,772 (26.5)
College graduate (> 3 years)	800 (28.7)	22,508 (28.7)
Unknown/missing	16 (0.6)	439 (0.6)
Maternal race and ethnicity		
White (non-Hispanic)	1581 (56.8)	40,993 (52.2)
Black (non-Hispanic)	722 (25.9)	21,472 (27.4)
Hispanic	337 (12.1)	11,180 (14.2)
Other races (non-Hispanic)	144 (5.2)	4,829 (6.2)
Unknown/missing	0 (0.0)	29 (0.0)
Maternal county of residence		
Kent	534 (19.2)	16,141 (20.6)
New Castle	1,689 (60.7)	46,561 (59.3)
Sussex	561 (20.2)	15,801 (20.1)
Cigarette use during pregnancy		
Yes	341 (12.2)	8,716 (11.1)
No	2,440 (87.6)	69,764 (88.9)
Unknown/missing	3 (0.1)	23.0 (0.0)
Trimester of prenatal care initiation		
No prenatal care	102 (3.7)	1,936 (2.5)
First trimester	2142 (76.9)	58,786 (74.9)
Second trimester	392 (14.1)	13,009 (16.6)
Third trimester	95 (3.4)	3,438 (4.4)
Unknown/missing	53 (1.9)	1,334 (1.7)
Pre-pregnancy BMI (kg/m^2)*		
Underweight (<18.5)	119 (4.3)	3,286 (4.2)
Normal weight (18.5 to <25.0)	1126 (40.4)	32,868 (41.9)
Overweight (25.0 to <30.0)	708 (25.4)	20,131 (25.6)
Obese (> = 30.0)	772 (27.7)	20,880 (26.6)
Unknown/missing	59 (2.1)	1,338 (1.7)

Table 3 contd/.

Maternal and infant characteristics	Birth Defect (N = 81,287)	
	Yes (n = 2,784) Number (%)	**No (n = 78,503) Number (%)**
Plurality†		
Singleton	2,669 (96.9)	75,987 (96.8)
Multiple	115 (4.1)	2,516 (3.2)
Birth weight		
Low (<2,500 grams)	572 (20.5)	65,31 (8.3)
Normal (>=2,500 grams)	2,212 (79.5)	71,970 (91.7)
Unknown/missing	0 (0.0)	2 (0.0)
Gestational age		
Preterm birth (< 37 weeks)	560 (20.1)	7418 (9.4)
Term birth (37 weeks or more)	2,224 (79.9)	71020 (90.5)
Unknown/missing	0 (0.0)	65 (0.1)
Size for gestational age		
Small	563 (20.2)	9,691 (12.3)
Appropriate	1,883 (67.6)	58,371 (74.4)
Large	292 (10.5)	8,882 (11.3)
Unknown/missing	46 (1.7)	1,559 (2.0)
Infant Death (before first birthday)		
Yes	98 (3.5)	444 (0.6)
No	2,686 (96.5)	78,059 (99.4)

Source: Delaware Health and Social Services, Division of Public Health, Delaware Health Statistics Center, and Birth Defects Registry Data, 2010-2017.

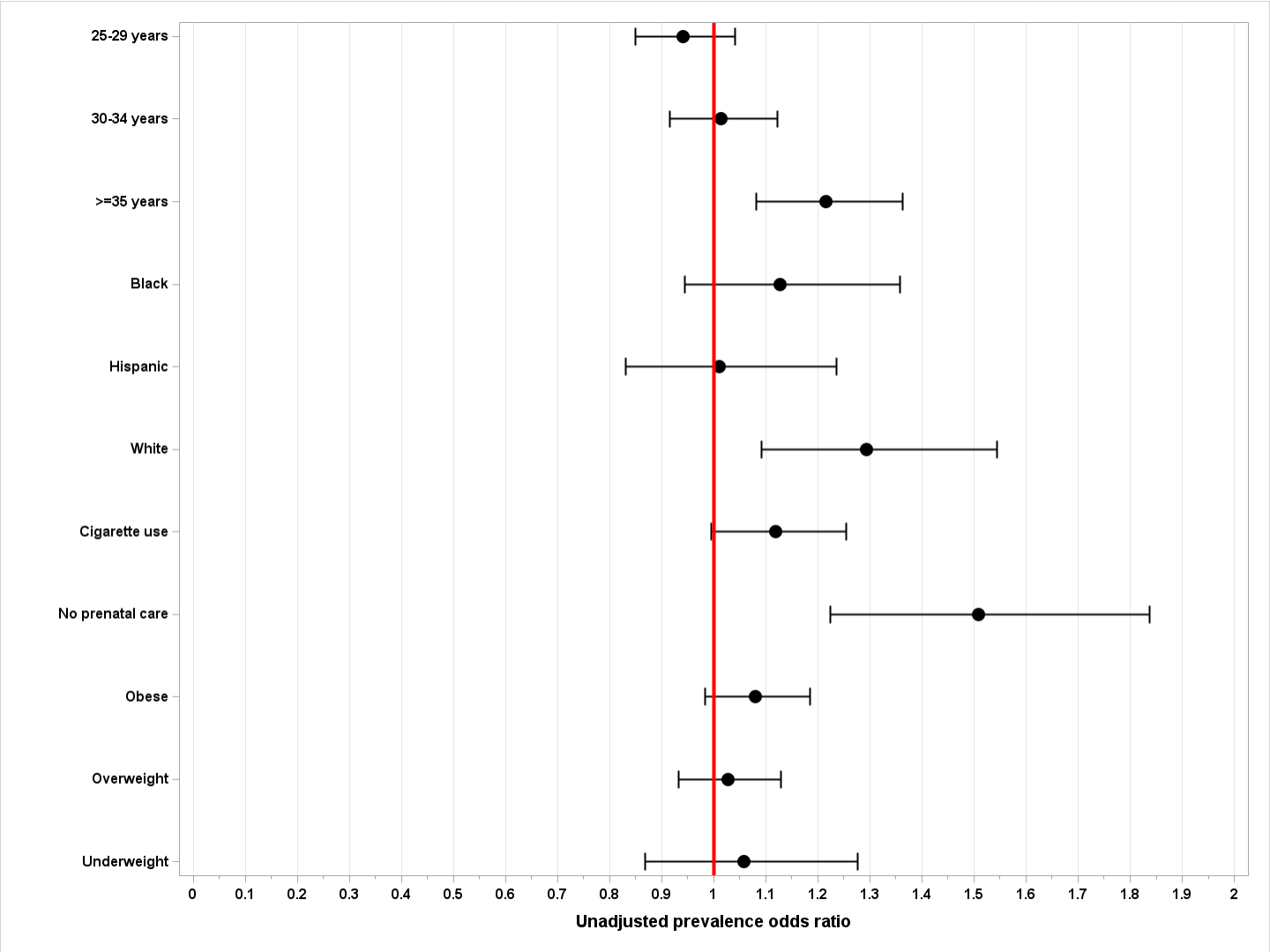
Notes: Linked birth defects registry and birth certificate data

†Plurality is the number of fetuses delivered live or dead at any time in the pregnancy regardless of gestational age, or if the fetuses were delivered at different dates in the pregnancy.

*BMI, or body mass index, is defined as the body mass divided by the square of the body height, expressed in mass in kilograms and height in meters.

To assess the factors associated with birth defects, unadjusted prevalence odds ratios (POR) were estimated for maternal age, race and ethnicity, cigarette use, timing of prenatal care, and pre-pregnancy BMI were estimated. Figure 9 indicated that mothers who were 35 years or older had a 20% greater odds of delivering an infant with a birth defect as compared to mothers less than 25 years of age (POR = 1.2; 95% CI: 1.1-1.3); White non-Hispanic mothers had a 30% greater odds of delivering infant with a birth defect as compared to other races (POR = 1.3; 95% CI: 1.1-1.5); and mothers with no prenatal care had a 50% greater odds of delivering an infant with a birth defect as compared to mothers with prenatal care (POR = 1.5; 95% CI: 1.2-1.8).

Figure 9. Unadjusted prevalence odds ratios of birth defects by key maternal demographic characteristics in Delaware, 2010-2017 with 95% confidence intervals



Source: Delaware Health and Social Services, Division of Public Health, Delaware Health Statistics Center, and Birth Defects Registry Data, 2010-2017.
Notes: Linked birth defects registry and birth certificate data. Unadjusted prevalence odds ratios calculated using reference categories for <25 years for maternal age; other race for maternal race and ethnicity; cigarette use vs. no use; no prenatal care vs. some care; normal weight for pre-pregnancy BMI, or body mass index.

Results from this data brief suggests that with the exception of VSDs, the prevalence of most of the birth defects are similar to the nation. Prevalence of birth defects was higher among older women, among White (non-Hispanic), and those who did not receive prenatal care. While, in majority of the cases the causes of birth defects are unknown, birth defects are known to have a significant impact on infant morbidity and mortality.

Endnotes

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Appendix 1. Typology of birth defects reportable and ascertained in Delaware, 2010-2017

Birth Defects	Structural (i.e.,physical abnormalities in the development of body parts including the skeleton and organs)	Functional or developmental (i.e., affecting the function and development of many parts of the body or abnormalities in the systems that run the body like the neurological, immune and endocrine systems)	Inborn errors of metabolism, endocrine, hemoglobinopathies
Congenital Anomalies of Central Nervous System			
Anencephalus	X		
Spina bifida without anencephalus	X		
Encephalocele	X		
Microcephalus	X		
Holoprosencephaly	X		
Lissencephaly	X		
Congenital Anomalies of the Eye			
Anophthalmia/microphthalmia	X		
Congenital cataract	X		
Coloboma	X		
Congenital Anomalies of the Ear			
Anotia/microtia	X		
Congenital Anomalies of the Cardiovascular System			
Common truncus (include truncus arteriosus)	X		

Birth Defects	Structural (i.e.,physical abnormalities in the development of body parts including the skeleton and organs)	Functional or developmental (i.e., affecting the function and development of many parts of the body or abnormalities in the systems that run the body like the neurological, immune and endocrine systems)	Inborn errors of metabolism, endocrine, hemoglobinopathies
Transposition of the great arteries	X		
Double outlet right ventricle	X		
Tetralogy of Fallot	X		
Single ventricle	X		
Ventricular septal defect (exclude "inlet" type, code as AVSD)	X		
Atrial septal defect (exclude "primum" type, code as AVSD)	X		
Atrioventricular septal defect (include Endocardial cushion defect, AV canal defect)	X		
Pulmonary valve atresia and stenosis (excludes dysplasia; supra- and sub-valvular stenosis)	X		
Tricuspid valve atresia and stenosis (excludes dysplasia)	X		
Ebstein anomaly	X		
Aortic valve stenosis (excludes supra- and sub-valvular stenosis)	X		

Birth Defects	Structural (i.e.,physical abnormalities in the development of body parts including the skeleton and organs)	Functional or developmental (i.e., affecting the function and development of many parts of the body or abnormalities in the systems that run the body like the neurological, immune and endocrine systems)	Inborn errors of metabolism, endocrine, hemoglobinopathies
Hypoplastic left heart syndrome	X		
Coarctation of the aorta	X		
Interrupted aortic arch	X		
Total anomalous pulmonary venous connection (includes return and drainage)	X		
Congenital anomalies of the Respiratory System			
Congenital pulmonary airway malformation (CPAM) (includes Congenital cystic lung, sequestrations)	X		
Lung agenesis	X		
Lung sequestration	X		
Congenital Anomalies of the Orofacial Area			
Choanal atresia	X		
Cleft palate alone (without cleft lip)	X		
Cleft lip alone (without cleft palate)	X		
Cleft lip with cleft palate	X		

Birth Defects	Structural (i.e.,physical abnormalities in the development of body parts including the skeleton and organs)	Functional or developmental (i.e., affecting the function and development of many parts of the body or abnormalities in the systems that run the body like the neurological, immune and endocrine systems)	Inborn errors of metabolism, endocrine, hemoglobinopathies
Congenital Anomalies of the Gastrointestinal Tract			
Esophageal atresia/tracheoesophageal fistula	X		
Small intestinal atresia/stenosis	X		
Rectal and large intestinal atresia/stenosis (include imperforate anus)	X		
Hirschsprung's disease	X		
Biliary atresia	X		
Congenital Anomalies of the Genitourinary System			
Cloacal exstrophy	X		
Hypospadias	X		
Renal agenesis/hypoplasia (include Potters)	X		
Cystic/dysplastic kidneys (Include multicystic dysplastic kidneys)	X		
Bladder exstrophy	X		
Congenital posterior urethral valves	X		

Birth Defects	Structural (i.e.,physical abnormalities in the development of body parts including the skeleton and organs)	Functional or developmental (i.e., affecting the function and development of many parts of the body or abnormalities in the systems that run the body like the neurological, immune and endocrine systems)	Inborn errors of metabolism, endocrine, hemoglobinopathies
Congenital Anomalies of the Musculoskeletal Regions			
Developmental hip dysplasia	X		
Clubfoot	X		
Limb deficiencies (reduction defects)	X		
Craniosynostosis	X		
Anomalies of skull and face bones (Include non-syndromic defects, e.g. Pierre Robin anomaly, etc)	X		
Diaphragmatic hernia	X		
Omphalocele	X		
Gastroschisis	X		
Chromosomal Anomalies			
Trisomy 21 (Down syndrome)	X	X	
Trisomy 13	X	X	
Trisomy 18	X	X	
Deletion 22q11.2 (includes Velo-cardio-facial syndrome; DiGeorge)	X	X	

Birth Defects	Structural (i.e.,physical abnormalities in the development of body parts including the skeleton and organs)	Functional or developmental (i.e., affecting the function and development of many parts of the body or abnormalities in the systems that run the body like the neurological, immune and endocrine systems)	Inborn errors of metabolism, endocrine, hemoglobinopathies
Other autosomal deletions (include microdeletions, Cri du chat, etc.)	X	X	
Other conditions due to autosomal anomalies (duplications, Trisomies other than 21,13,18)	X	X	
Turner syndrome (include gonadal dysgenesis)	X	X	
Other conditions due to sex chromosome anomalies (include XYY, XXX, Klinefelter syndrome, etc)	X	X	
Other conditions due to chromosome anomalies (e.g. Triploidy, Tetraploidy)	X	X	
Other and Unspecified Congenital Anomalies			
Situs inversus	X		
Other specified anomalies and syndromes			
(e.g. Goldenhar, VACTERL Association, Prader-Willi, Amniotic band disruption complex, etc.)	X	X	

Birth Defects	Structural (i.e.,physical abnormalities in the development of body parts including the skeleton and organs)	Functional or developmental (i.e., affecting the function and development of many parts of the body or abnormalities in the systems that run the body like the neurological, immune and endocrine systems)	Inborn errors of metabolism, endocrine, hemoglobinopathies
Single Gene Disorders			
(e.g. Noonan, deLange, Beckwith-Weidemann, Rubinstein-Taybi, TAR, Marfan, Cockayne, Lowe, Polysyndactyly, Charge, Coffin-Lowry, Apert, Crouzon, Treacher Collins)		X	X
Endocrine, Metabolic, Immunodeficiency			
Congenital hypothyroidism			X
Congenital adrenal hyperplasia			X
Phenylketonuria			X
Hyperphenylalaninemia			X
Tyrosinemia			X
Maple Syrup Urine Disease			X
3-Methylcrotonyl-CoA Carboxylase Deficiency (3-MCC)			X
Propionic Acidemia			X
Multiple Carboxylase Deficiency			X
Isovaleric Acidemia			X
Beta-Ketothiolase Deficiency			X

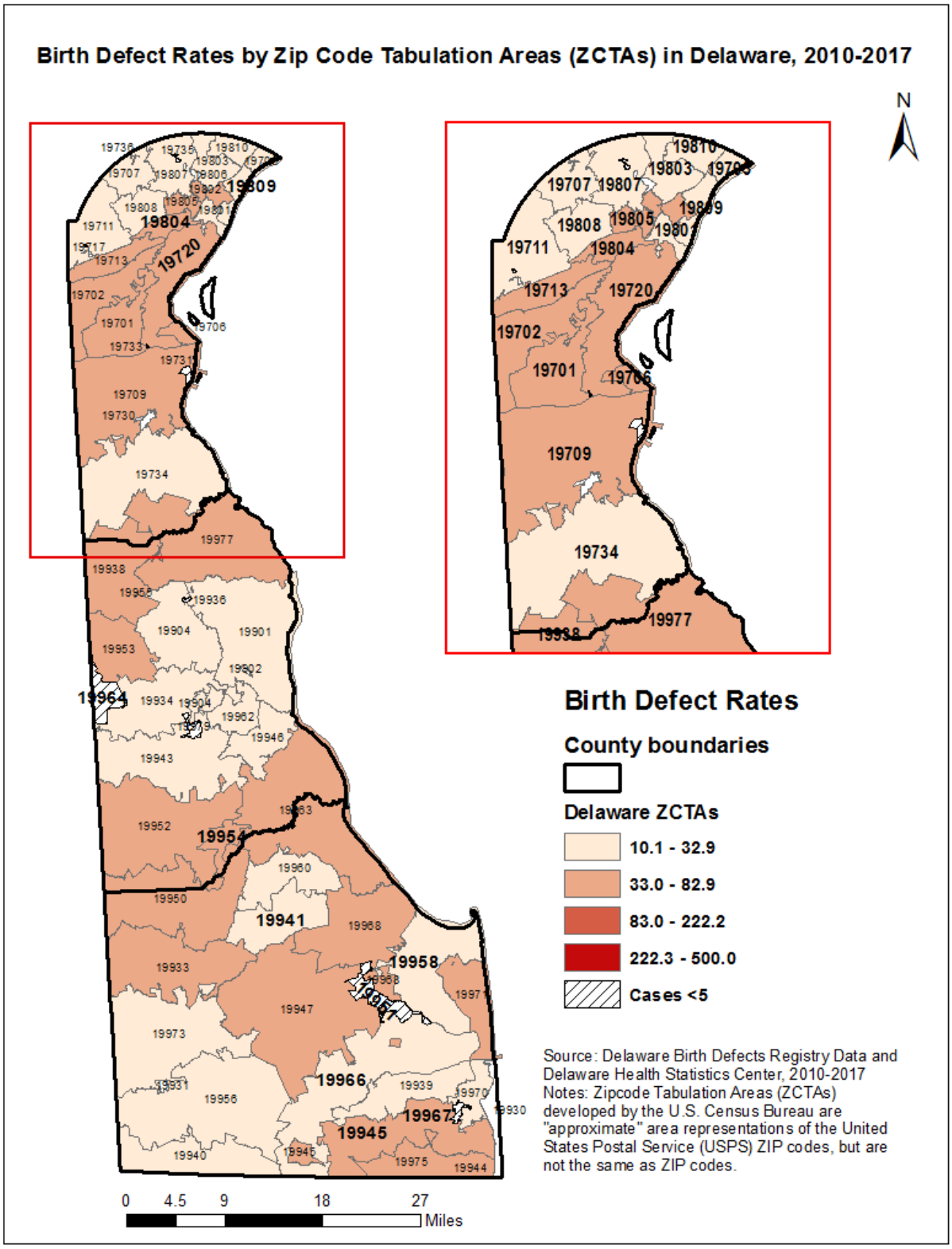
Birth Defects	Structural (i.e.,physical abnormalities in the development of body parts including the skeleton and organs)	Functional or developmental (i.e., affecting the function and development of many parts of the body or abnormalities in the systems that run the body like the neurological, immune and endocrine systems)	Inborn errors of metabolism, endocrine, hemoglobinopathies
Homocystinuria			X
Hypermethioninemia			X
Citrullinemia			X
Argininemia			X
Arginine Lyase Deficiency			X
3OH-3-Methylglutaryl CoA Lyase Deficiency			X
Other Urea Cycle Disorders			X
Methylmalonic Acidemia			X
Glutaric Aciduria I			X
Other Specified Disorders of Amino Acid Metabolism			X
Other Aminoacidopathies			X
Galactosemia (Galactose-1-phosphate uridyl transferase (GALT) deficiency; Galactokinase deficiency; Epimerase deficiency; Other Galactosemias)			X
Cystic Fibrosis			X
Biotinidase Deficiency (total or partial)			X

Birth Defects	Structural (i.e.,physical abnormalities in the development of body parts including the skeleton and organs)	Functional or developmental (i.e., affecting the function and development of many parts of the body or abnormalities in the systems that run the body like the neurological, immune and endocrine systems)	Inborn errors of metabolism, endocrine, hemoglobinopathies
2-Methylbutyryl-CoA Dehydrogenase Deficiency			X
Isobutyryl-CoA-Dehydrogenase Deficiency			X
Cobalamin Metabolic Defect			X
Carnitine Uptake Deficiency			X
Carnitine/Acylcarnitine Translocase Deficiency			X
Medium Chain Acyl-CoA Dehydrogenase Deficiency (MCAD)			X
Long-chain 3-hydroxyacyl-CoA Dehydrogenase Deficiency (LCHAD)			X
Short-chain Acyl-CoA Dehydrogenase Deficiency (SCAD)			X
Long-chain Acyl-CoA Dehydrogenase Deficiency (LCAD)			X
Very Long-chain Acyl-CoA Dehydrogenase Defiency (VLCAD)			X
Glutaric Acidemia II (Multiple CoA Dehydrogenase Deficiency)			X

Birth Defects	Structural (i.e.,physical abnormalities in the development of body parts including the skeleton and organs)	Functional or developmental (i.e., affecting the function and development of many parts of the body or abnormalities in the systems that run the body like the neurological, immune and endocrine systems)	Inborn errors of metabolism, endocrine, hemoglobinopathies
Carnitine Palmitoyl Transferase Deficiency II			X
Other disorders of fatty acid oxidation			X
Disorders of mitochondrial metabolism			X
Other specified disorders of metabolism			X
Severe Combined Immunodeficiency (SCID)-many forms			X
Other Immunodeficiency disorders			X
Hemoglobinopathies			
Sickle Cell Disease			X
Other hemoglobinopathies			X

Source: Delaware Department of Health and Social Services, Division of Public Health, Delaware Birth Defects Registry

Appendix 2. Birth defect rates by Zip Code Tabulation Areas (ZCTAs) in Delaware, 2010-2017



Mission — Protect and Promote the Health of all People in Delaware

Vision — Healthy People in Healthy Communities

Core Values — Integrity–Respect–Participation–Accountability–Teamwork–Excellence

Division of Public Health
Family Health Systems Section
Jesse S. Cooper Building
417 Federal Street | Dover, DE 19901
Phone: 302-744-4553 | Fax: 302-739-6653
www.dhss.delaware.gov/dhss/dph

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