Data Brief ■ No. 2 ■ December 2022

Birth Defects

Delaware Profile 2010-2019

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 3% to 5% percent of births are affected by a birth defect, which are congenital structural, functional, 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) categories [8, 10]. Identification of modifiable risk factors can be particularly challenging due to ascertainment techniques 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).

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 3,612 infants were diagnosed with a birth defect during 2010-2019 in Delaware.

Key findings

- The overall 2010-2019 birth defect prevalence rate in Delaware was 33.1 per 1,000 live births (95% CI: 32.1-34.2) or 3.3 percent and similar to the U.S.
- Of the 3,612 infants who were diagnosed with a birth defect, 3,059 infants (85%) had a single birth defect and one in six (n = 553; 15%) infants with a birth defect had multiple birth defects.
- Birth defects were more common in males (55%) as compared to females (45%). Ventricular septal defect was the most common birth defect in Delaware during 2010-2019.
- 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. Women with no prenatal care had 40% greater odds of delivering an infant with a birth defect. Women who conceived through use of assistive reproductive technology had 40% greater odds of delivering an infant with a birth defect.

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 in the U.S. have shown an increase from 2010-2014 [2].

Birth defects ascertainment in Delaware

The Birth Defects Registry (BDR) was established in 1997, by the Delaware Department of Health and Social Services, 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 recommendations of the CDC and 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 sources. 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, and hemoglobinopathies.

Table 1 shows the major birth defects typically reported [11] and the national prevalence estimates for some of the major birth defects derived from 2010-2014 analysis by Mai et al. [2]. 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: clubfoot (1 in 593 births); Trisomy 21or Down Syndrome (1 in 707 births); pulmonary valve atresia and stenosis (1 in 1,052); cleft lip with cleft palate (1 in 1,563); cleft lip alone (without cleft palate 1 in 1,687); coarctation of aorta (1 in 1,795); atrioventricular septal defect (AVSD 1 in 1,853); limb deficiencies (1 in 1,943); and 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. 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

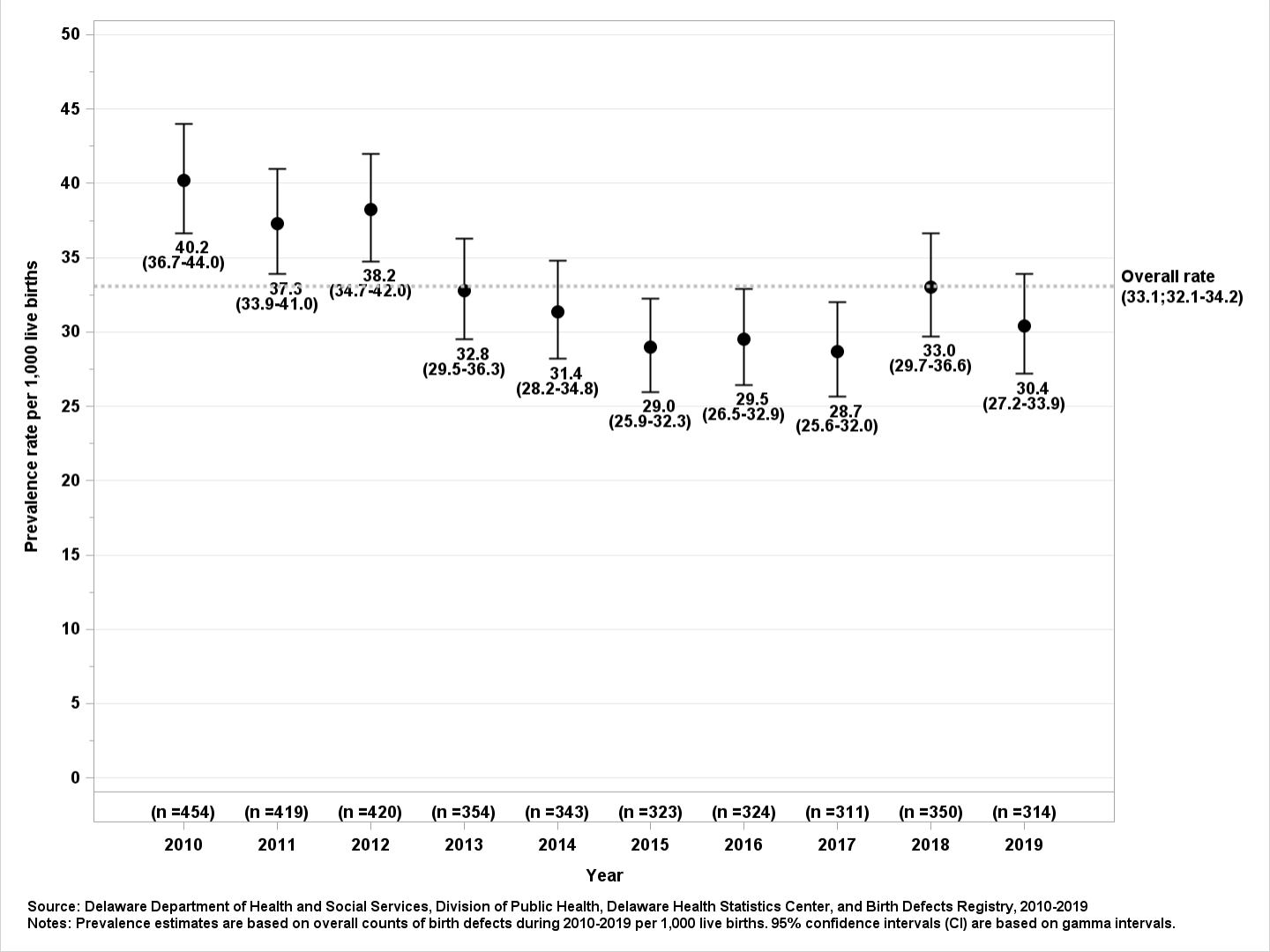
Notes: Birth defects typically reported to NBPDN are from Salemi et al. (2019),
Source: National estimates are from Centers for Disease Control and Prevention available at https://www.cdc.gov/ncbddd/birthdefects/data.html from 2010-2014 analysis of birth defects by Mai et al. (2019).

Birth defects in Delaware, 2010-2019

From 2010 to 2019, 3,612 infants were diagnosed with a birth defect in Delaware and 1,980 (54.8%) were males, and 1,632 (45.2%) were females. Of the 3,612 infants with birth defects, 3,478 (96.3%) were live births, followed by 53 (1.5%) infant deaths, 63 (1.7%) fetal deaths, and 18 (0.5%) terminations. Of the 3,612 infants who were diagnosed with a birth defect, 3,059 infants (84.7%) had a single birth defect, 315 (8.7%) infants had two birth defects, and 238 (7%) had three or more birth defects. The overall 2010-2019 birth defect prevalence rate in Delaware was 33.1 per 1,000 live births (95% CI: 32.1-34.2) or 3.3 percent. According to 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 1 displays the annual birth defect prevalence rates in Delaware between 2010 to 2019. While it seems that the birth defect rates have declined during 2010 to 2019 time-period from 40.2 per 1,000 live births in 2010 to 30.4 in 2019, the decline is specific to the number of birth defect cases ascertained during 2010-2019. The differences in the number of birth defects are reflective of changes in reportable conditions by NBDPN, and emerging science for addition or deletion of specific birth defects but not necessarily related to the major reportable birth defects.

Figure 1. Birth defect prevalence per 1,000 live births in Delaware, 2010-2019

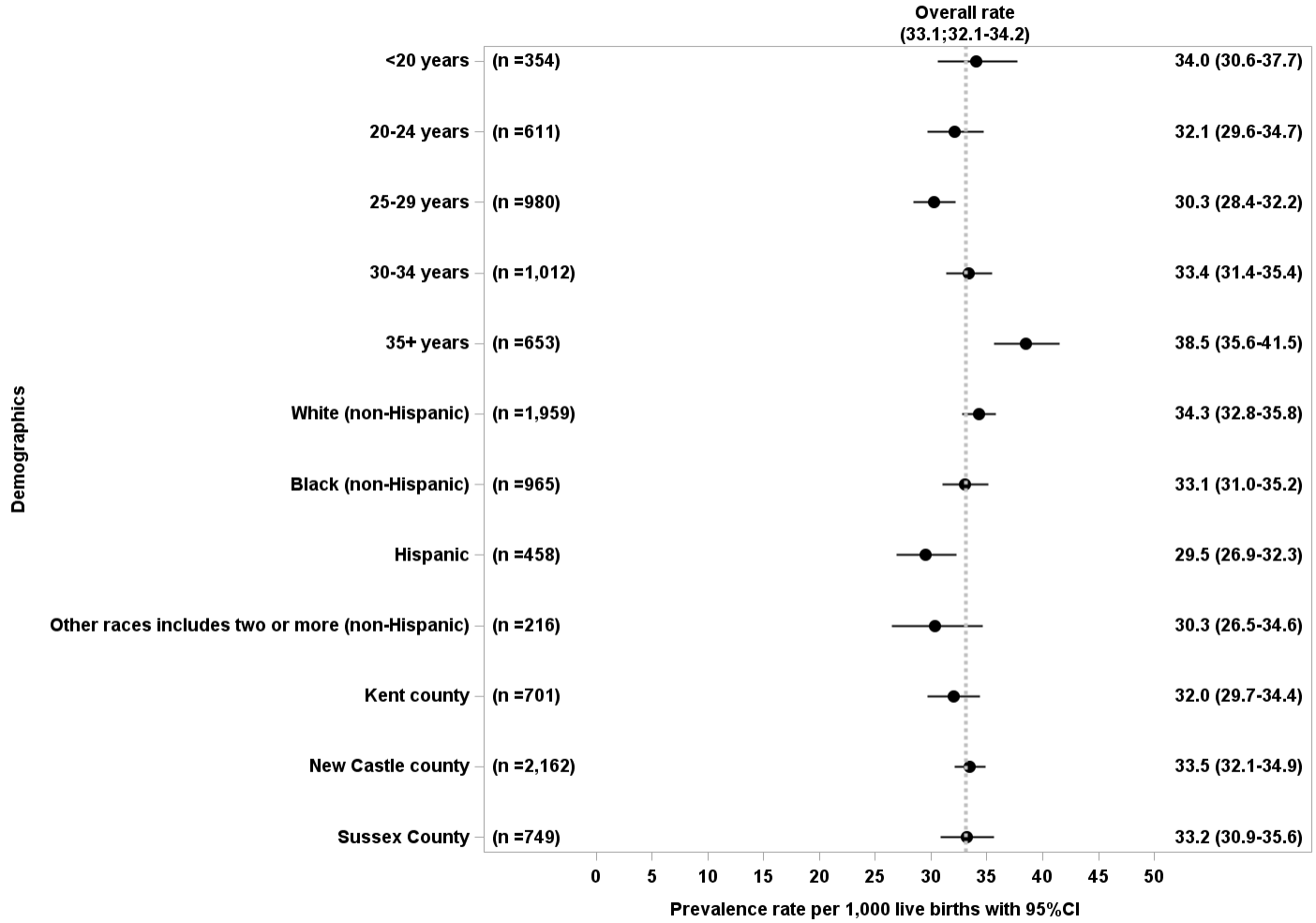


Source: Delaware Department of Health and Social Services, Division of Public Health, Delaware Health Statistics Center, and Birth Defects Registry, 2010-2019
Notes: Prevalence estimates are based on overall counts of birth defects during 2010-2019 per 1,000 live births. 95% confidence intervals (CI) are based on gamma intervals.

Figure 2 displays the birth defect prevalence rates by maternal age, race and ethnicity, and county of residence. During 2010-2019 time-period, birth defect prevalence rate was highest among women 35 and older with 38.5 (95%CI: 35.6-41.5) per 1,000 live births, followed by White (non-Hispanic) women with 34.3 (95%CI: 32.8-35.8), Black (non-Hispanic) women with 33.1 (95%CI: 31.0-35.2), other races that include two or more races (i.e., American Indian/Alaska Native, Asian, Native Hawaiian/Pacific Islander) with 30.3 (95%CI: 26.5-34.5), and Hispanic women with 29.5 (95%CI: 26.9-32.3). Birth defect prevalence rates were similar in all three counties: New Castle County, rate of 33.5 (95%CI: 32.1-34.9); Sussex County, rate of 33.2 (30.9-35.6); and Kent County, rate of 32.0 (95%CI: 29.7-34.4) per 1,000 live births.

Although the majority (n = 3,059) of infants had a single birth defect, about 15% (n = 553) of infants had multiple defects (i.e., two or more defects). The proportion of multiple defects varied by race and ethnicity. Of infants with any birth defects, 21% of Hispanic, 16% of Black (non-Hispanic), 16% other races, and 14% of White (non-Hispanic) infants had two or more birth defects. Adjusting for race and ethnic categories, Hispanic infants had 1.7 (95%CI: 1.3-2.2) times greater odds of having multiple birth defects as compared to White (non-Hispanic) infants.

Figure 2. Birth defect prevalence per 1,000 live births by key demographics in Delaware, 2010-2019



Source: Delaware Department of Health and Social Services, Division of Public Health, Delaware Health Statistics Center, and Birth Defects Registry, 2010-2019
Notes: Prevalence estimates are based on overall counts of birth defects during 2010-2019 per 1,000 live births. 95% confidence intervals (CI) are based on gamma intervals.

The Delaware BDR captures the maternal conditions available and documented on the medical record of women who delivered an infant with a birth defect. These conditions may include any gynecological surgical procedures performed prior to the index pregnancy, chronic conditions such as hypertension, diabetes, depression, psychiatric disorders (i.e., bi-polar disorders, schizophrenia, etcetera.), thyroid disorders, heart disease, lupus, obesity, and 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. About 15% (n = 527) did not have a documented maternal condition, 20% (n = 749) had one maternal condition, and the majority (n = 2,336; 65%) had two or more maternal conditions documented.

Common or core birth defects in Delaware 2010-2019

These “core” defects are part of NBDPN list of organ systems and exclude categories such as developmental hip dysplasia, sickle cell disease, microcephaly, cystic fibrosis, congenital hypothyroidism, other single gene

disorders that fall into the "recommended" and/or extended categories. The Delaware BDR captures a wide range of birth defects and may differ from the list of defects from the NBDPN (Table 1).

Table 2 presents the number and percentage of infants with a single defect or two or more birth defects and whether the birth defects are part of the NBPDN core list. Of the 3,059 Delaware infants with a single birth defect, 62% (1,901/3,059) had a birth defect in the NBPDN core list, and 38% (1,158/3,059) infants with a single birth defect had a defect not on the NBDPN core list. Similarly, of the 553 infants with two or more birth defects, 85% (473/553) of infants had a birth defect on the NBDPN core list, and about 15% (80/553) of infants with two or more birth defects had a defect not on the NBDPN core list.

Table 2. Number and percentage of infants with single or multiple defects in Delaware, 2010-2019

Birth defect	Birth defect		Total
	NBDPN Core	Other	
Single defect	1,901 (80.1%)	1,158 (93.5%)	3,059 (84.7%)
Two or more defects	473 (19.9%)	80 (6.5%)	553 (15.3%)
Total	2,374 (100%)	1,238 (100%)	3,612 (100%)

Source: Delaware Health and Social Services, Division of Public Health, Delaware Health Statistics Center, and Birth Defects Registry Data, 2010-2019.
Notes: Column percentages unless otherwise noted.

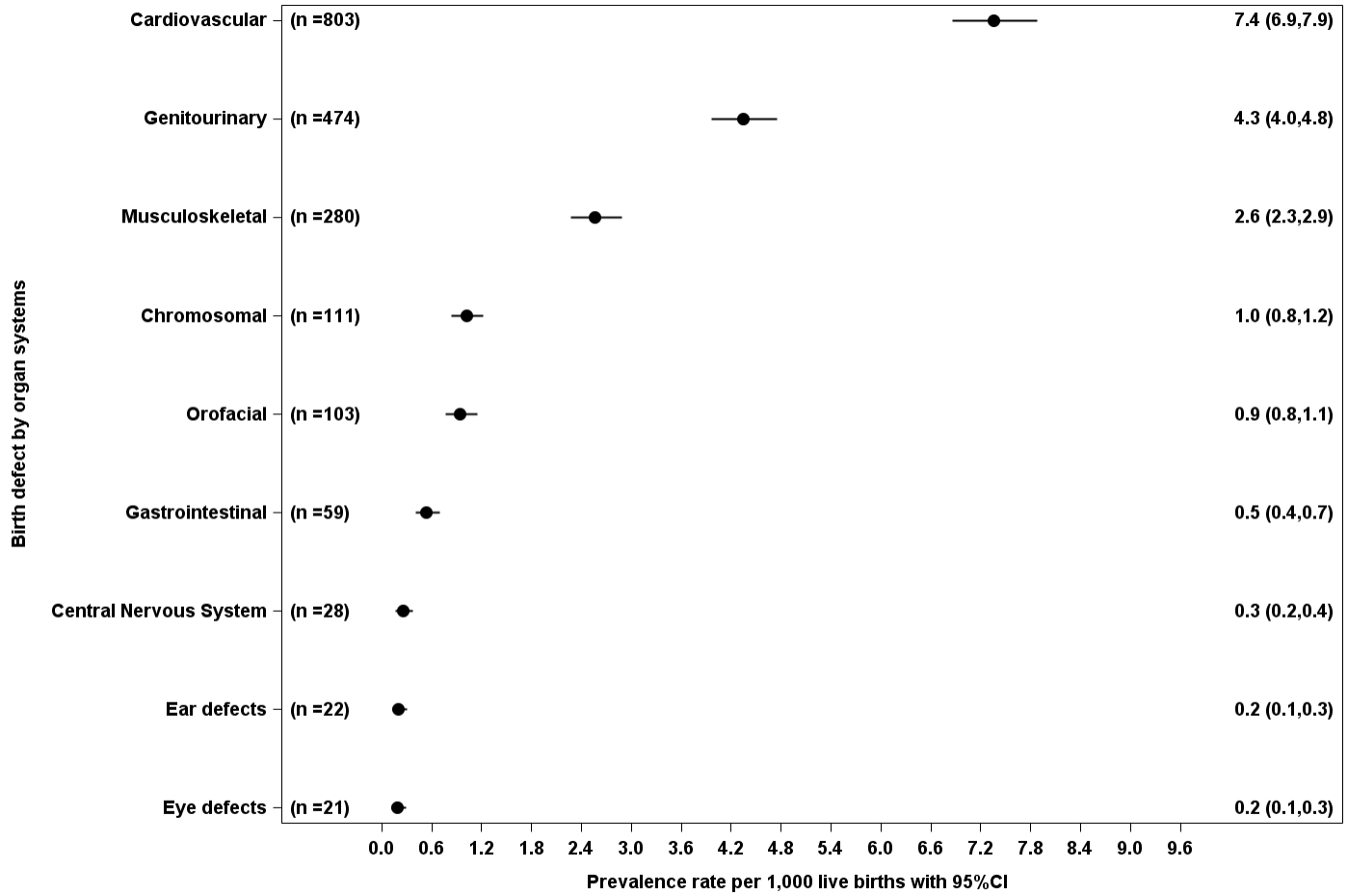
Figure 3 displays the prevalence estimates of single birth defects in the NBPDN core (1,901) by major organ systems diagnosed in infants in Delaware during 2010-2019. During 2010-2019, birth defects that are part of the cardiovascular system had the highest prevalence rate with 7.4 (95%CI: 6.9-7.9) per 1,000 live births. The birth defects in the cardiovascular system include primary (i.e., common truncus, hypoplastic left heart syndrome, pulmonary valve atresia and stenosis, pulmonary valve atresia, tetralogy of Fallot etcetera); secondary (i.e., coarctation of aorta, double outlet right ventricle, Ebstein anomaly etcetera); and other congenital heart defects (i.e., aortic valve stenosis, atrial septal defect, atrioventricular septal defect, ventricular septal defect).

Similarly, birth defects that are part of the genitourinary system had the second highest prevalence rate with 4.3 (95%CI: 4.0-4.8) per 1,000 live births. Some of the birth defects in the genitourinary system include bladder exstrophy, cloacal exstrophy, congenital posterior urethral valves, hypospadias, and renal agenesis or hypoplasia.

Birth defects that are part of the musculoskeletal organ system had the third highest prevalence rate with 2.6 (95%CI: 2.3,2.9) per 1,000 live births during 2010-2019. The birth defects in this category include clubfoot, diaphragmatic hernia, gastroschisis, limb deficiencies (reduction defects), omphalocele etcetera.

Finally, birth defects that are chromosomal had the fourth highest prevalence rate with 1.0 (0.8-1.2) per 1,000 live births. These include deletion 22 q11.2, Turner syndrome, Trisomy 13 (Patau syndrome), Trisomy 18 (Edward's syndrome), and Trisomy 21 (Down syndrome).

Figure 3. Core single birth defect* prevalence per 1,000 live births by organ systems in Delaware, 2010-2019



Source: Delaware Department of Health and Social Services, Division of Public Health, Delaware Health Statistics Center, and Birth Defects Registry, 2010-2019
Notes: Prevalence estimates are based on overall counts of birth defects during 2010-2019 per 1,000 live births. 95% confidence intervals (CI) are based on gamma intervals.
*Single birth defect across all diagnoses (dx) categories.

Table 3 presents the top 20 birth defects in Delaware during 2010-2019 with total counts, percent, and the prevalence estimates. Of the 3,059 single birth defects, the top 20 birth defects accounted for 74% (2,271) 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) renal agenesis/hypoplasia; 9) cleft lip with cleft palate; 10) limb deficiencies; 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) single gene disorder; and 9) other specified anomalies and syndromes are captured as part of Delaware's birth defect registry. It is important to note that reporting of pyloric stenosis stopped after 2012 birth cohort.

Table 3. Top 20 birth defects by category, count, and prevalence in Delaware, 2010-2019

Birth defect	Birth defect category	Count (%)	Prevalence (95 % CI)
1. Ventricular septal defect	Other congenital heart defects	595 (19.5%)	5.5 (5.0-5.9)
2. Hypospadias	Genitourinary	405 (13.2%)	3.7 (3.4-4.1)
3. Developmental hip dysplasia*	Musculoskeletal	211 (6.9%)	1.9 (1.7-2.2)
4. Clubfoot	Musculoskeletal	122 (4.0%)	1.1 (0.9-1.3)
5. Atrial septal defect	Other congenital heart defects	118 (3.9%)	1.1 (0.9-1.3)
6. Sickle cell disease*	Hemoglobinopathies	112 (3.7%)	1.0 (0.8-1.2)
7. Trisomy	Chromosomal	93 (3.0%)	0.9 (0.7-1.0)
8. Cystic dysplastic kidneys*	Genitourinary	89 (2.9%)	0.8 (0.7-1.0)
9. Single gene disorder*	Single gene disorder	73 (2.4%)	0.7 (0.5-0.8)
10. Congenital hypothyroidism*	Endocrine, Metabolic, Immunodeficiency	53 (1.7%)	0.5 (0.4-0.6)
11. Pulmonary valve atresia and stenosis	Primary critical congenital heart defects	53 (1.7%)	0.5 (0.4-0.6)
12. Craniosynostosis*	Musculoskeletal	48 (1.6%)	0.4 (0.3-0.6)
13. Gastroschisis	Musculoskeletal	48 (1.6%)	0.4 (0.3-0.6)
14. Microcephalus*	Central nervous system	43 (1.4%)	0.4 (0.3-0.5)
15. Pyloric stenosis*	Gastrointestinal	42 (1.4%)	0.4 (0.3-0.5)
16. Renal agenesis/hypoplasia	Genitourinary	41 (1.3%)	0.4 (0.3-0.5)
17. Other specified anomalies and syndromes*	Other birth defects	39 (1.3%)	0.4 (0.3-0.5)
18. Cleft lip with cleft palate	Orofacial	29 (0.9%)	0.3 (0.2-0.4)
19. Limb deficiencies (reduction defects)*	Musculoskeletal	29 (0.9%)	0.3 (0.2-0.4)
20. Cleft palate alone (without cleft lip)	Orofacial	28 (0.9%)	0.3 (0.2-0.4)
Top 20 total†		**2,271 (74.2%)**	**20.8 (20.0-21.7)**
Single birth defects§		**3,059 (84.7%)**	**28.0 (27.1-29.0)**
All birth defects (includes single and multiple)		**3,612 (100%)**	**33.1 (32.1-34.2)**

Source: Delaware Health and Social Services, Division of Public Health, Delaware Health Statistics Center, and Birth Defects Registry Data, 2010-2019.

Notes: Prevalence estimates are based on overall counts of birth defects during 2010-2019 per 1,000 live births. 95% confidence intervals (CI) are based on gamma intervals.

*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 (2,271/3,059).

§Single birth defects (3,059/3,612).

The following paragraphs discuss the top 10 birth defects of all core single birth defects in Delaware and compares each to available NBDPN estimates. During 2010-2019 there were 595 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% (595/3,059) of single birth defect with an overall prevalence of 5.5 (95% CI: 5.0-5.9) per 1,000 live births. 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 405 cases of hypospadias identified among Delaware male infants that accounted for 13% (405/3,059) of the top 20 single birth defects with an overall prevalence of 3.7 (95% CI: 3.4-4.2) per 1,000 live births. 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 211 (6.9%) Delaware infants having a singular birth defect with an overall prevalence of 1.9 per 1,000 live births (95% CI: 1.7-2.2), 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 122 cases (4.0%) and an overall prevalence of 1.1 (95% CI: 0.9-1.4) per 1,000 live births. The Delaware prevalence for clubfoot was similar to that of the U.S, which is 1.3 per 1,000 live births [15].

Atrial septal defect (ASD), not the same as atrioventricular septal defect (AVSD or endocardial cushion defect), was among the top five birth defects with a total of 118 cases (3.9%) and an overall prevalence of 1.1 per 1,000 live births (95% CI: 0.9-1.3). While there is no current national estimate for ASD, the Delaware's ASD prevalence is similar to the worldwide prevalence of 1.6 [17].

Sickle cell disease (SCD) or hemoglobinopathies was diagnosed among 112 infants (3.7%) and had an overall prevalence of 1.0 (95% CI: 0.9-1.3) per 1,000 live births. 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].

Trisomy 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 seventh top birth defect in Delaware and was diagnosed among 93 infants having a singular birth defect (3.0%) with an overall prevalence of 0.9 (95% CI: 0.7-1.0) per 1,000 live births. Of the 93 cases, 88 cases were specific to Down syndrome, four cases were specific to Edward's syndrome, and one case was specific to Patau's syndrome. The national estimate for Down syndrome is about one in 700 births [12], slightly higher than Delaware.

Cystic dysplastic kidneys (i.e., malformed kidney with cysts) was diagnosed among 89 infants having a singular birth defect (2.9%) in Delaware during 2010-2019 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].

Single gene disorders are when a single gene mutation occur and affect any aspect or function or structure, may include common single gene disorders (e.g., hemoglobin disorders, cystic fibrosis, oculo-cutaneous albinism) or "rare" gene disorders [19]. These disorders may be classified into different categories: dominant, recessive, and X-linked [19]. The prevalence of single gene disorders is extremely rare, as they affect 1% [19]. While no national estimate is available, during 2010-2019, the prevalence of single gene disorder was 0.7 (95%CI: 0.5-0.8) per 1,000 live births in Delaware.

Congenital hypothyroidism (CHT) is a common endocrine abnormality and was the tenth top birth defect. Some estimates suggest that 2% to 5% of CHT is inherited and there is a genetic component [20]. As per national estimates, CHT impacts one in 2,000 to 4,000 live births. During 2010-2019, 53 cases (1.7%) of infants with CHT with an overall prevalence of 0.5 per 1,000 live births (95% CI: 0.4-0.7) in Delaware, and Delaware's prevalence was similar to the national prevalence.

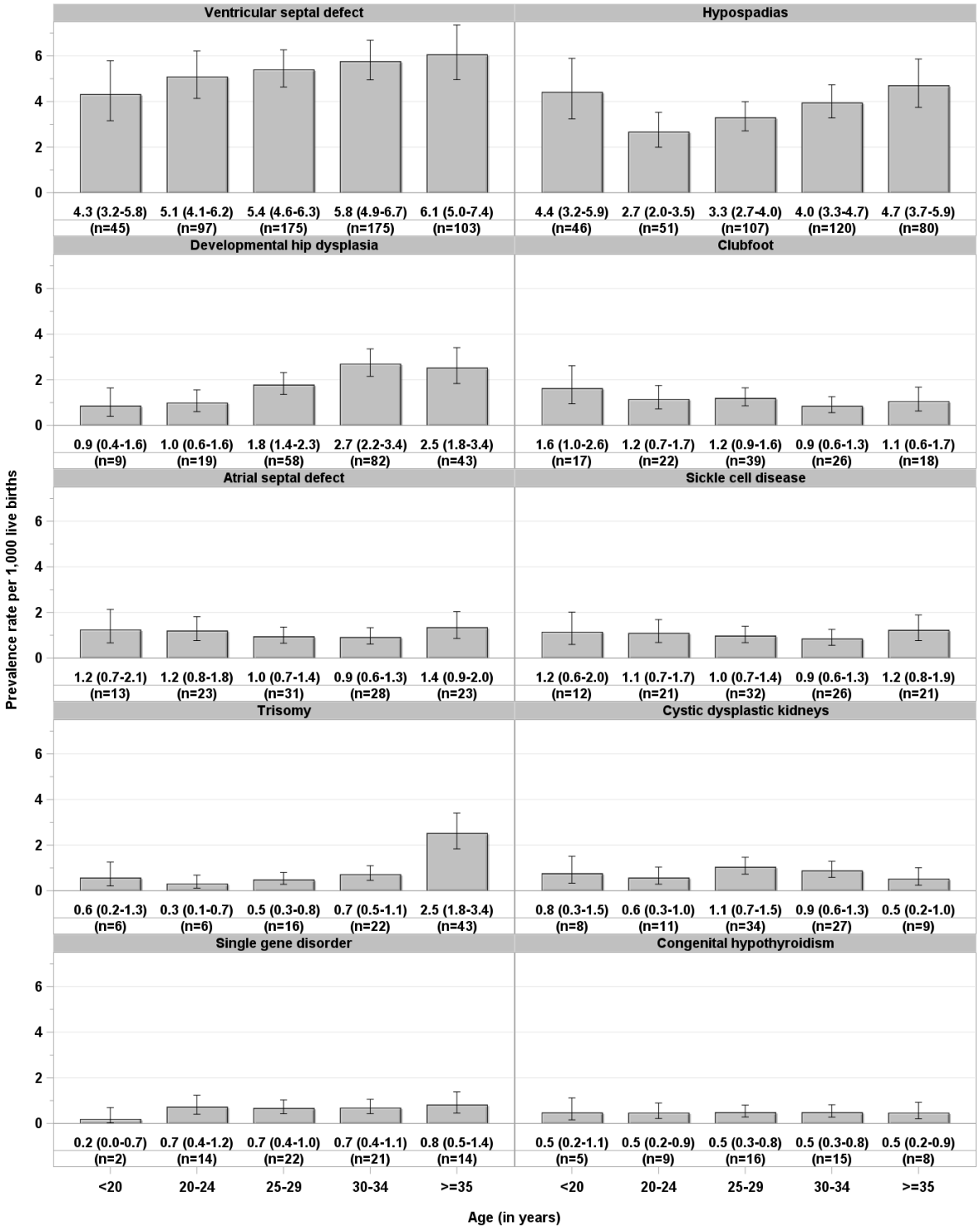
National prevalence estimates for birth defects vary by maternal age, and maternal race and ethnicity [2, 21]; for some birth defects, maternal race and ethnicity specific prevalence rates are available for comparison based on 2012-2016 data [21]. Figures 4 through 6 display the Delaware prevalence rates per 1,000 live births for the top 10 birth defects stratified by maternal age, race and ethnicity, and county of residence for 2010-2019.

For example, rates of VSD, hypospadias, and DDH increased with maternal age consistent with studies that also find older maternal age as a risk factor [21, 22]. However, this pattern was not consistent for all birth defects. Trisomy rates were highest among mothers who were 35 and older (2.5 per 1,000 live births; 95% CI: 1.8-3.4), while birth defect rates for clubfoot were highest among mothers who were less than 25 years old (1.6 per 1,000 live births; 95% CI: 1.0-2.6). Older maternal age is also found to be associated with trisomy [23].

With regards to maternal race and ethnicity, rates of VSD, DDH, and clubfoot were lowest among Black (non-Hispanic) people, and the prevalence rate of cystic dysplastic kidney (1.1 per 1,000 live births; 95% CI: 0.7-1.5) and sickle cell disease (3.7 per 1,000 live births; 95% CI: 3.0-4.7). was highest in this group. Among Hispanic people, prevalence rate of trisomy was highest (1.2 per 1,000 live births; 95% CI: 0.9-2.1) and the rate of hypospadias was lowest (1.3 per 1,000 live births; 95% CI: 0.8-2.0). The prevalence rate of VSD (6.1 per 1,000 live births; 95% CI: 5.5-6.7) and DDH (2.8 per 1,000 live births; 95% CI: 2.4-3.3) was highest among White (non-Hispanic) persons. The estimates stratified by maternal race and ethnicity using 2012-2016 national data [21] show similar patterns, where VSD was higher in White (non-Hispanic), trisomy was higher among Hispanics.

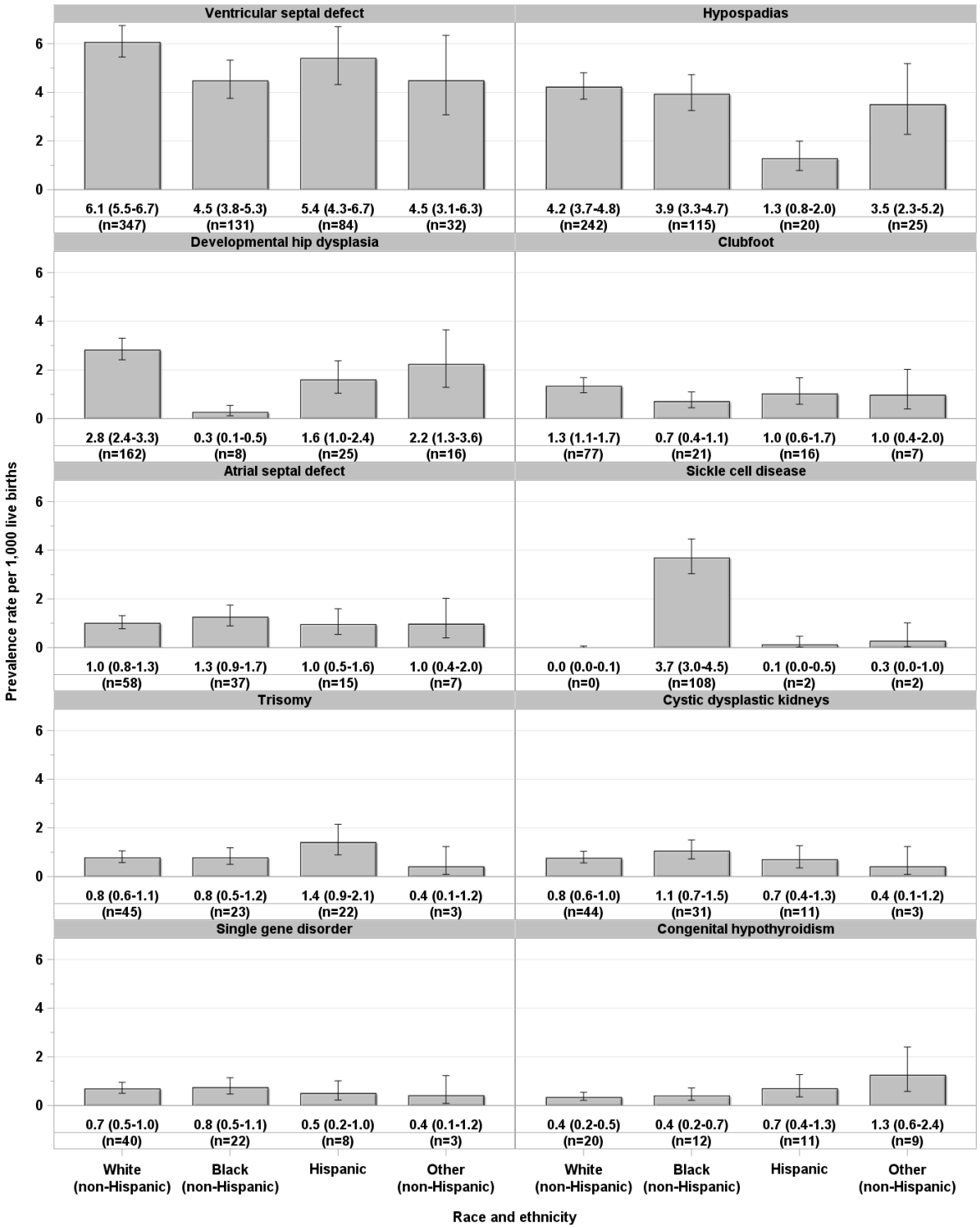
Although there was no discernible pattern with regards to county of residence, VSD prevalence rates were higher in Sussex County (5.8 per 1,000 live births; 95% CI: 4.9-6.9) and DDH rates were higher in New Castle County (2.3 per 1,000 live births; 95% CI: 2.0-2.7).

Figure 4. Prevalence rates of top 10 birth defects* stratified by maternal age in Delaware, 2010-2019



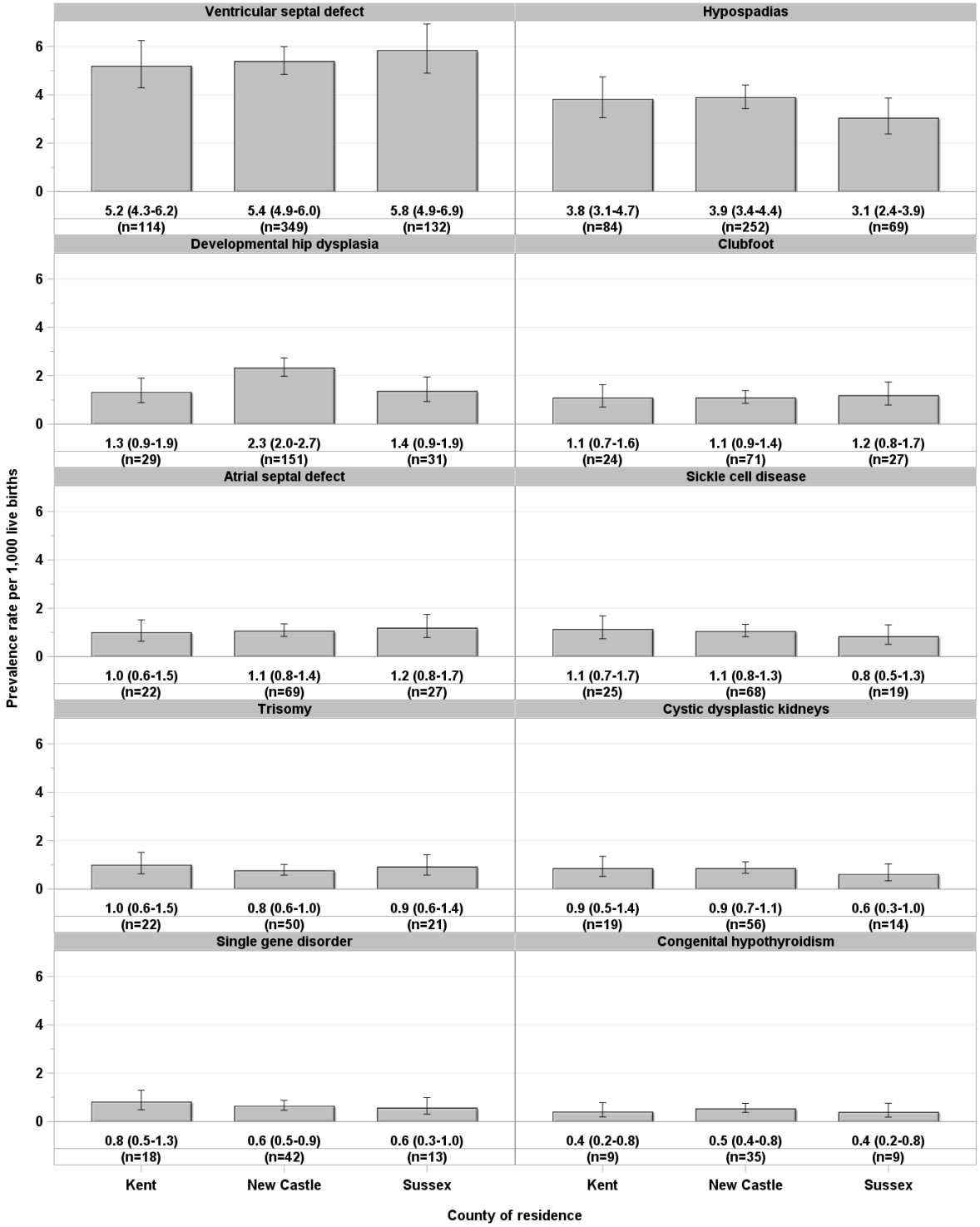
Source: Delaware Department of Health and Social Services, Division of Public Health, Delaware Health Statistics Center, and Birth Defects Registry, 2010-2020
Notes: Prevalence estimates are based on overall counts of birth defects during 2010-2019 per 1,000 live births. 95% confidence intervals (CI) are based on gamma intervals.
***Single birth defect across all diagnoses (dx) categories.**

Figure 5. Prevalence rates of top 10 birth defects* stratified by maternal race and ethnicity in Delaware, 2010-2019



Source: Delaware Department of Health and Social Services, Division of Public Health, Delaware Health Statistics Center, and Birth Defects Registry, 2010-2020
Notes: Prevalence estimates are based on overall counts of birth defects during 2010-2019 per 1,000 live births. 95% confidence intervals (CI) are based on gamma intervals.
***Single birth defect across all diagnoses (dx) categories.**

Figure 6. Prevalence rates of top 10 birth defects* stratified by county of residence in Delaware, 2010-2019



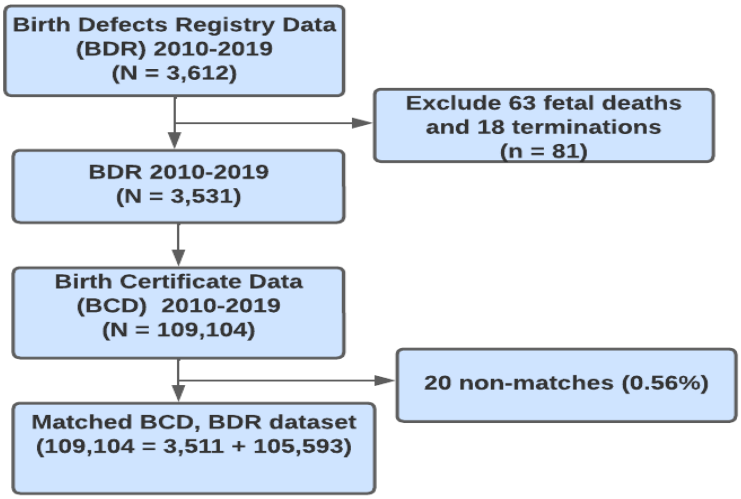
Source: Delaware Department of Health and Social Services, Division of Public Health, Delaware Health Statistics Center, and Birth Defects Registry, 2010-2020

Notes: Prevalence estimates are based on overall counts of birth defects during 2010-2019 per 1,000 live births. 95% confidence intervals (CI) are based on gamma intervals.

***Single birth defect across all diagnoses (dx) categories.**

The next section describes the characteristics of infants with and without a birth defect using a linked dataset. The dataset for 2010-2019 included 109,104 hospital births with 3,511 birth defects and 105,593 without birth defects and excluded fetal deaths and terminations (see Figure 7).

Figure 7. Matched birth defects registry and birth certificated data in Delaware, 2010-2019



Characteristics of infants with and without birth defects in Delaware 2010-2019

Table 4 displays the maternal and infant characteristics of infants with and without birth defects in Delaware during 2010-2019. Of the 109,104 infants, 3,511 (3.2%) infants had a birth defect and 105,593 (96.8%) did not have a birth defect. There were differences in maternal and infant characteristics.

Table 4. Maternal and infant characteristics of infants with and without birth defects in Delaware, 2010-2019

Maternal and infant characteristics	Total	Birth Defects, Number (%)	
		Birth defect	No Birth defect
Total	**109,104**	**3,511 (3.2%)**	**105,593 (96.8%)**
Parity*			
Missing/Unknown	144 (0.1%)	3 (0.1%)	141 (0.1%)
Nulliparous	42,755 (39.2%)	1,483 (42.2%)	41,272 (39.1%)
Multiparous	66,205 (60.7%)	2,025 (57.7%)	64,180 (60.8%)
Plurality†			
Singleton	105,615 (96.8%)	3,376 (96.1%)	102,239 (96.8%)
Two or more	3,489 (3.2%)	135 (3.9%)	3,354 (3.2%)
Maternal age (mean [SD])	28.4 (±5.8)	28.8 (±6.1)	28.4 (±5.8)
Maternal age (in years)			
< 20	6,661 (6.1%)	209 (6.0%)	6,452 (6.1%)
20-24	22,774 (20.9%)	734 (20.9%)	22,040 (20.9%)
25-29	32,371 (29.7%)	940 (26.8%)	31,431 (29.8%)
30-34	30,330 (27.8%)	984 (28.0%)	29,346 (27.8%)
35 or more	16,968 (15.6%)	644 (18.3%)	16,324 (15.5%)

Table 4 cont'd/.

Maternal and infant characteristics	Total	Birth Defects, Number (%)	
		Birth defect	No Birth defect
Maternal education			
Missing/Unknown	727 (0.7%)	23 (0.7%)	704 (0.7%)
< 9 years of schooling	5,785 (5.3%)	180 (5.1%)	5,605 (5.3%)
9 – 11 years of schooling	12,688 (11.6%)	397 (11.3%)	12,291 (11.6%)
High school graduate	28,005 (25.7%)	949 (27.0%)	27,056 (25.6%)
1 – 3 years of college	28,853 (26.4%)	938 (26.7%)	27,915 (26.4%)
College graduate (> 3 years)	33,046 (30.3%)	1,024 (29.2%)	32,022 (30.3%)
Maternal race and ethnicity			
Missing/Unknown	131 (0.1%)	1 (0.0%)	130 (0.1%)
White (non-Hispanic)	57,141 (52.4%)	1,958 (55.8%)	55,183 (52.3%)
Black (non-Hispanic)	29,195 (26.8%)	928 (26.4%)	28,267 (26.8%)
Hispanic	15,517 (14.2%)	438 (12.5%)	15,079 (14.3%)
Other races (non-Hispanic)	7,120 (6.5%)	186 (5.3%)	6,934 (6.6%)
Payor of birth			
Missing/Unknown	527 (0.5%)	3 (0.1%)	524 (0.5%)
Medicaid	50,404 (46.2%)	1,642 (46.8%)	48,762 (46.2%)
Private	52,921 (48.5%)	1,704 (48.5%)	51,217 (48.5%)
Self-pay	2,053 (1.9%)	58 (1.7%)	1,995 (1.9%)
Other	3,199 (2.9%)	104 (3.0%)	524 (0.5%)
Trimester of prenatal care initiation			
Missing/Unknown	3,277 (3.0%)	85 (2.4%)	3,192 (3.0%)
No prenatal care	2,523 (2.3%)	117 (3.3%)	2,406 (2.3%)
First trimester	80,132 (73.4%)	2,650 (75.5%)	77,482 (73.4%)
Second trimester	18,295 (16.8%)	526 (15.0%)	17,769 (16.8%)
Third trimester	3,277 (3.0%)	133 (3.8%)	4,744 (4.5%)
Pre-pregnancy Body Mass Index (BMI kg/m^2)[§]			
Missing/Unknown	1,843 (1.7%)	65 (1.9%)	1,778 (1.7%)
Underweight (<18.5)	5,740 (5.3%)	147 (4.2%)	5,593 (5.3%)
Normal weight (18.5 to <25.0)	44,284 (40.6%)	1,394 (39.7%)	42,890 (40.6%)
Overweight (25.0 to <30.0)	27,817 (25.5%)	902 (25.7%)	26,915 (25.5%)
Obese (> = 30.0)	29,420 (27.0%)	1,003 (28.6%)	28,417 (26.9%)
Cigarette use during pregnancy			
Missing/unknown	244 (0.2%)	6 (0.2%)	238 (0.2%)
Yes	11,332 (10.4%)	406 (11.6%)	10,926 (10.3%)
No	97,528 (89.4%)	3,099 (88.3%)	94,429 (89.4%)
Assistive reproductive technology (ART)[¶]			
Yes	1,721 (1.6%)	76 (2.2%)	1,645 (1.6%)
No	107,383 (98.4%)	3,435 (97.8%)	103,948 (98.4%)

Table 4 cont'd/.

Maternal and infant characteristics	Total	Birth Defects, Number (%)	
		Birth defect	No Birth defect
Infant sex			
Male	55,672 (51.0%)	1,929 (54.9%)	53,743 (50.9%)
Female	53,432 (48.9%)	1,582 (45.1%)	51,850 (49.1%)
Low birth weight (<2,500 grams)			
Missing/unknown	83 (0.1%)	3 (0.1%)	80 (0.1%)
Yes	9,595 (8.8%)	715 (20.4%)	8,880 (8.4%)
No	99,426 (91.1%)	2,793 (79.5%)	96,633 (91.5%)
Preterm birth (<37 weeks gestation)			
Missing/unknown	138 (0.1%)	3 (0.1%)	135 (0.1%)
Yes	10,725 (9.8%)	695 (19.8%)	10,030 (9.5%)
No	98,241 (90.0%)	2,813 (80.1%)	95,428 (90.4%)
Small for gestational age**			
Missing/unknown	1,810 (1.7%)	51 (1.5%)	1,759 (1.7%)
Yes	13,269 (12.2%)	677 (19.3%)	12,592 (11.9%)
No	94,025 (86.2%)	2,783 (79.3%)	91,242 (86.4%)

Source: Delaware Health and Social Services, Division of Public Health, Delaware Health Statistics Center, and Birth Defects Registry Data, 2010-2019.

Notes: Linked birth defects registry and birth certificate data

*Parity is the number of pregnancies reaching 20 weeks and 0 days of gestation or beyond, regardless of the number of fetuses or outcomes.

†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.

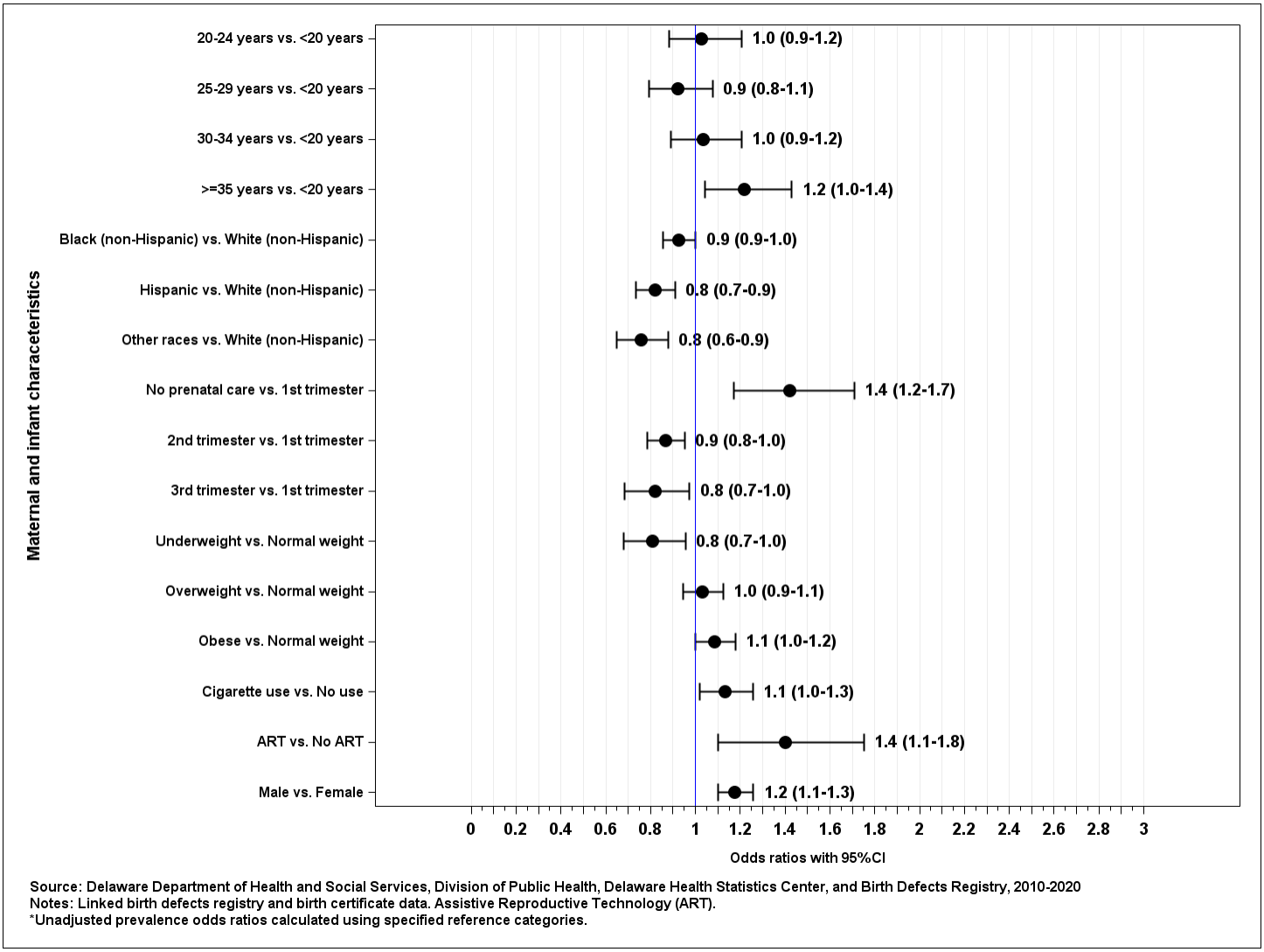
§BMI, or body mass index, is defined as the body mass divided by the square of the body height, expressed in mass in kilograms and height in meters.

¶Assistive reproductive technology (ART) includes all fertility treatments in which either eggs or embryos are surgically removed from a woman's ovaries and combined them with sperm in the laboratory and return them to the woman's body or donating them to another woman (See: https://www.cdc.gov/art/whatis.html#:~:text=According%20to%20this%20definition%2C%20ART,donating%20them%20to%20another%20woman).

**Small for gestation term is used for infants who are smaller than usual amount for the number of weeks of pregnancy. Typically, these infants have birth weights below the 10th percentile or smaller than other babies of the same gestational age.

The percentage of birth defects was higher among nulliparous women (42.2% vs. 39.1%). Women who delivered infants with a birth defect were older (*M* = 28.8 years; *SD* = ±6.1) as compared to women who did not deliver an infant with a birth defect (*M* = 28.4 years; *SD* = ±5.8); for instance, the percentage of women 35 and older were 18.3% in the birth defects group as compared 15.3% in the non-birth defects group. As compared to the women in the non-birth defects group, the percentage of women who delivered an infant with a birth defect was higher among White (non-Hispanic) women (55.8% vs. 52.3%) and lower in Hispanic women (12.5% vs. 14.5%). The percentage of birth defects was also higher among women without prenatal care (3.3% vs. 2.3% in non-birth defects group) and among women with higher BMI (i.e., obese 28.6% vs. 26.9% in non-birth defects group). Birth defects were also higher among women who conceived through use of assistive reproductive technology (2.2% vs. 1.6% in non-birth defects group). Higher percentage of male infants had birth defects (54.9%). As compared to infants with no birth defects, infants with birth defect were low birth weight (20.4% vs. 8.4%), premature (19.8% vs. 9.5%), and small for gestational age (19.3% vs. 11.9%). Figure 7 presents the unadjusted prevalence odds ratios for key maternal demographic characteristics. Women 35 and older, women without prenatal care, women with BMI >=30, women who conceived through use of ART, and women who delivered a male infant had greater odds of a birth defect.

Figure 7. Unadjusted prevalence odds ratios of birth defects by key maternal demographic characteristics with 95% confidence intervals, Delaware, 2010-2019



Source: Delaware Department of Health and Social Services, Division of Public Health, Delaware Health Statistics Center, and Birth Defects Registry, 2010-2020
Notes: Linked birth defects registry and birth certificate data. Assistive Reproductive Technology (ART).
*Unadjusted prevalence odds ratios calculated using specified reference categories.

Conclusion

During 2010-2019, there were 3,612 infants diagnosed with birth defects in Delaware with an overall prevalence rate of 33.1 per 1,000 live births. Alternatively, 1 in 3 infants in Delaware had a birth defect with ventricular septal defect as the most common birth defect. Results from this data brief suggests that except for 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. Although 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.

References

1. Acheson A, Vaidy A, Stomieroski K, et al. Surveillance of ventricular septal defects in Delaware. *Birth Defects Research Part A*: Clinical and Molecular Teratology. 2016;106:888-893.

2. Mai CT, Isenburg JL, Canfield MA, et al. National population-based estimates for major birth defects, 2010-2014. *Birth Defects Res*. 2019;111(18):1420-1435. doi:10.1002/bdr2.158.

3. Centers for Disease Control and Prevention. Available at: https://www.cdc.gov/ncbddd/birthdefects/features/kf-hospital-stays-costs-birthdefects-2013.html. Accessed 2020-06-18.

4. Naeye RL, Blanc W, Leblanc W, Khatamee MA. Fetal complications of maternal heroin addiction: abnormal growth, infections, and episodes of stress. *J Pediatr*. 1973;83(6):1055–1061pmid:4757521

5. Källén B, Borg N, Reis M. The use of central nervous system active drugs during pregnancy. *Pharmaceuticals* (Basel). 2013; 6(10):1221–1286pmid:24275849.

6. Holbrook BD, Rayburn WF. Teratogenic risks from exposure to illicit drugs. *Obstetrics and gynecology clinics of North America*. 2014;41:229-239.

7. Oliveira CIF, Fett-Conte AC. Birth defects: Risk factors and consequences. *J Pediatr Genet*. 2013;2:85-90.

8. Tinker SC, Gilboa S, Reefhuis J, Jenkins MM, Schaeffer M, Moore CA. *Challenges in studying modifiable risk factors for birth defects*. Curr Epodemiol Rep. 2015;2:23-30.

9. Feldkamp ML, Carey JC, Byrne JLB, Krikov S, Botto LD. Etiology and clinical presentation of birth defects: population based study. *BMJ*. 2019;357:j2249 http://dx.doi.org/10.1136/bmj.j2249

10. Berard A. Zhao JP, Sheehy, O. Antidepressant use during pregnancy and the risk of major congenital malformations in a cohort of depressed pregnant women: an updated analysis of the Quebec Pregnancy Cohort. *BMJ Open*. 2019;7:e013372. doi:10.1136/bmjopen-2016-013372.

11. Salemi JL, Tanner JP, Kirby RS, Cragan JD. The impact of the ICD-9-CM to ICD-10-CM transition on the prevalence of birth defects among infant hospitalizations in the United States. *Birth Defects Res*. 2019;111(18):1365-1379. doi:10.1002/bdr2.1578

12. Mason CA, Kirby RS, Sever LE, Langlois PH. Prevalence is the preferred measure of frequency of birth defects. *Birth Defects Res A Clin Mol Teratol*. 2005;73(10):690-692. doi:10.1002/bdra.20211

13. van der Horst HJ, de Wall LL. Hypospadias, all there is to know [published correction appears in Eur J Pediatr. 2019 Oct;176(10):1443]. *Eur J Pediatr*. 2019;176(4):435-441. doi:10.1007/s00431-017-2864-5

14. Shaw BA, Segal LS; SECTION ON ORTHOPAEDICS. Evaluation and Referral for Developmental Dysplasia of the Hip in Infants. *Pediatrics*. 2016;138(6):e20163107. doi:10.1542/peds.2016-3107

15. Ansar A, Rahman AE, Romero L, et al. Systematic review and meta-analysis of global birth prevalence of clubfoot: a study protocol. *BMJ Open*. 2018;8(3):e019246. Published 2018 Mar 6. doi:10.1136/bmjopen-2019-019246

16. Centers for Disease Control and Prevention. Available at: https://www.cdc.gov/ncbddd/sicklecell/documents/SCD-factsheet_SCD-Pregnancy.pdf. Accessed 2020-06-18.

17. van der Linde D, Konings EE, Slager MA, et al. Birth prevalence of congenital heart disease worldwide: a systematic review and meta-analysis. *J Am Coll Cardiol*. 2011;58(21):2241-2247. doi:10.1016/j.jacc.2011.08.025

18. National Institute of Diabetes and Digestive and Kidney Diseases. Available at: https://www.niddk.nih.gov/health-information/kidney-disease/children/kidney-dysplasia

19. Blencowe H, Moorthie S, Petrou M, et al. Rare single gene disorders: estimating baseline prevalence and outcomes worldwide. *J Community Genet*. 2018;9(4):397-406. doi:10.1007/s12687-018-0376-2

20. MedlinePlus (2020). Congenital hypothyroidism. Available at: https://medlineplus.gov/genetics/condition/congenital-hypothyroidism/#references. Accessed 2020-10-20.

21. Stallings EB, Isenburg JL, Short TD, et al. Population-based birth defects data in the United States, 2012-2016: A focus on abdominal wall defects. *Birth Defects Res*. 2019;111(18):1436-1447. doi:10.1002/bdr2.1607

22. Chan A, McCaul KA, Cundy PJ, Haan EA, Byron-Scott R. Perinatal risk factors for developmental dysplasia of the hip. *Arch Dis Child Fetal Neonatal Ed*. 1997;76(2):F94-F100. doi:10.1136/fn.76.2.f94

23. Best KE, Rankin J. Is advanced maternal age a risk factor for congenital heart disease?. *Birth Defects Res A Clin Mol Teratol*. 2016;106(6):461-467. doi:10.1002/bdra.23507

24. Hussaini KS, Drummond D, E Bartoshesky L, et al. Assessing the relationship between neonatal abstinence syndrome and birth defects in Delaware. *Birth Defects Res.* 2021;113(2):144-151. doi:10.1002/bdr2.1811 Epub 2020 Sep 30.

25. Rasmussen SA, Moore CA, Paulozzi LJ, Rhodenhiser EP. Risk for birth defects among premature infants: a population-based study. *J Pediatr*. 2001;138(5):668-673. doi:10.1067/mpd.2001.112249

26. Honein MA, Kirby RS, Meyer RE, et al. The association between major birth defects and preterm birth. *Matern Child Health J*. 2009;13(2):164-175. doi:10.1007/s10995-008-0348-y

Appendix 1. Typology of birth defects reportable and ascertained in Delaware, 2010-2019

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		
Transposition of the great arteries	X		
Double outlet right ventricle	X		

Appendix 1 cont'd/.

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
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		
Hypoplastic left heart syndrome	X		
Coarctation of the aorta	X		
Interrupted aortic arch	X		

Appendix 1 cont'd/.

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
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		
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		

Appendix 1 cont'd/.

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		

Appendix 1 cont'd/.

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	

Appendix 1 cont'd/.

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	
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

Appendix 1 cont'd/.

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
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
Homocystinuria			X
Hypermethioninemia			X
Citrullinemia			X
Argininemia			X
Arginine Lyase Deficiency			X
3OH-3-Methylglutaryl CoA Lyase Deficiency			X
Other Urea Cycle Disorders			X

Appendix 1 cont'd/.

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
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
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

Appendix 1 cont'd/.

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
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
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

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

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