Epidemiology of Birth Defects in Rhode Island, 2018–2020

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In Rhode Island, approximately 375 infants are born with a birth defect every year.[1] A birth defect is a structural abnormality present at birth that affects the development of organs and tissues of an infant. It may be identified during pregnancy, at birth, or following birth. Although not all birth defects can be prevented, common causes include environmental pollutants, certain medical conditions and medications, infections, personal behaviors, and genetics.[1,2] Depending on the severity of the birth defect, there may be serious consequences for the infant, requiring medical, educational, or developmental interventions.[2] Infants with birth defects account for approximately 20% of infant deaths each year both nationally[3] and in Rhode Island.[4] Having a birth defects surveillance system is important in the early identification of infants and children with birth defects to prevent more serious effects throughout their life.

Rhode Island's birth defects surveillance system, located in the Rhode Island Department of Health's (RIDOH) Birth Defects Program (RIBDP), tracks birth defects, identifies children who may require additional support services, and assists in developing policies to reduce birth defects and infant mortality. Providers are mandated by regulation (216-RICR-10-10-3) to report birth defects in children up to the age of five who are diagnosed with a birth defect. Reporting occurs mainly through automated discharge reporting from birth hospitals and from provider reports.

The RIBDP routinely analyzes its surveillance data to examine trends and risk factors for birth defects. The results of these analyses are used to inform Rhode Island prevention efforts and policy. From 2015 to 2019, the prevalence rate of birth defects in Rhode Island increased by 27%.[1] Prevalence remained stable from 2015 to 2017, with the largest increase (20%) occurring from 2017 to 2018 and remaining elevated from 2018 to 2019.[1] More recent Rhode Island data for 2020 are now available. The RIBDP examined the epidemiology of birth defects cases from 2018 to 2020 to determine overall prevalence, prevalence of maternal characteristics of cases, and frequently impacted body systems to better characterize populations affected by birth defects in Rhode Island.

METHODS

All RI maternity hospitals report newborns diagnosed with a 10th Clinical Modification of the International Classification of Diseases (ICD-10) 'Q' code at discharge from the birth hospital to the RIBDP on a regular basis. The RIBDP staff confirm the accuracy of reported birth defects diagnoses through chart review of reported cases. A birth defects case is a newborn diagnosed with a birth defect ICD-10 'Q' code after chart review and had a Rhode Island maternal residence at time of birth. Minor birth defects were excluded from our case definition to focus on more relevant conditions for data analysis (case definition is available at https://health.ri.gov/publications/databooks/2018BirthDefects.pdf). Prevalence rates were calculated for 2016 to 2020 to examine overall prevalence trends over time. Cases were limited to newborns born from 2018 to 2020, which is the most recent full year of data available and the period of recent increases in prevalence rates.

To obtain infant and maternal information, birth defects cases were linked to birth certificate data from RIDOH's Center for Vital Records. Maternal characteristics used in analyses included city/town of residence, education, insurance, education level, pre-pregnancy weight and height, and race/ethnicity. These characteristics were known to be generally associated with birth defects in Rhode Island from previous analyses[1,5,6] and were self-reported by the mother on the birth certificate. To calculate the overall prevalence rate, birth certificate data for live births were used as the denominator for each birth year in the cohort. Birth certificate data were then stratified by maternal characteristic to provide denominators to calculate prevalence rates for birth defects by these characteristics.

City/town of residence was defined as core or non-core. A core city (Central Falls, Pawtucket, Providence, and Woonsocket) had a poverty level higher than 25%. Combined race and ethnicity categories were classified as follows: non-Hispanic White, non-Hispanic Black or African American, non-Hispanic Asian, and Hispanic (all races). Those with unknown ethnicity and who were non-Hispanic with an unknown or other race were not included in race/ethnicity rates. Additionally, American Indian/Alaskan Native and Native Hawaiian/Pacific Islander race categories were not included due to RIDPH's small numbers policy.[7] Body mass index (BMI) was calculated using pre-pregnancy height (inches) and weight (pounds) from birth certificate data (BMI=weight/height^2 x 703). Obesity was defined as having a BMI greater than or equal to 30 kg/cm^2.

To determine prevalence rates by body system, birth defects were also grouped into major body systems using ICD-10 'Q' codes. Some newborns had multiple birth defects and as these birth defects often affected different body systems, this analysis used all birth defects reported when determining counts by body system.

SAS Version 9.4 software was used for data cleaning and frequency calculations (SAS Institute, Cary, NC). Prevalence rates were calculated using Microsoft Excel (Microsoft Office 365, Version 2008).

RESULTS

Birth defects prevalence increased 24% from 2016 to 2020 in Rhode Island (**Figure 1**). From 2018 to 2020, there were 1,247 birth defects cases reported to the RIBDP and 30,755 Rhode Island resident births. The overall prevalence rate of birth defects from 2018 to 2020 was 405 cases per 10,000 live births (**Table 1**). Two percent of birth defects cases were unable to be linked with the birth certificate file to obtain maternal characteristic information. **Table 1** shows the overall prevalence of birth defects by year, along with prevalence rates per 10,000 live births by selected maternal characteristics. Prevalence rates were highest for each respective maternal characteristic in cases with a maternal age at delivery of 35 years of age and older, maternal residence in a core city, maternal non-Hispanic black race/ethnicity, public insurance, and a maternal education level of less than 12th grade completed. Prevalence rates were similar when stratifying cases by maternal marital status and obesity status.

Twenty-six percent of newborns (n=324) had more than one birth defect diagnosed (range: 1 to 9 birth defects). **Table 2** shows the prevalence rates of birth defects grouped by body system. Cardiovascular system birth defects were the most prevalent among cases (171 cases per 10,000 live births), followed by the genitourinary (157 cases per 10,000 live births) and musculoskeletal systems (123 cases per 10,000 live births). Birth defects affecting the orofacial and respiratory body systems were least frequently seen.

Figure 1. Birth Defects Prevalence Rates, Rhode Island, 2016–2020

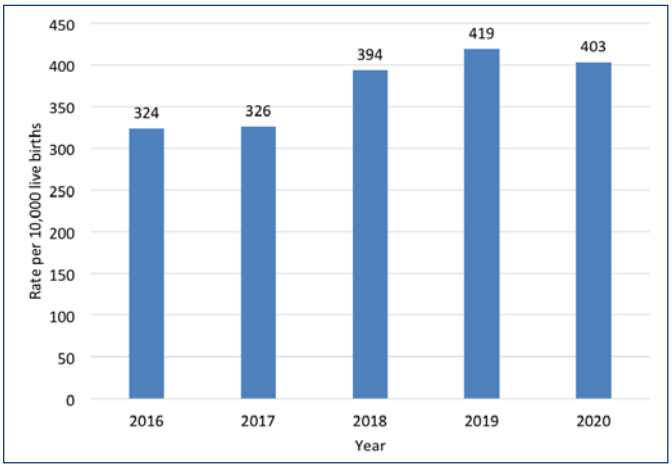

Table 1. Birth Defect Prevalence Rates by Maternal Characteristics, Rhode Island, 2018–2020

	Counts	Prevalence rate per 10,000 live births
Overall	**1247**	**405**
Maternal Age		
<25	231	402
25-34	701	380
35 and older	284	432
City/Town		
Core*	486	393
Non-Core	691	376
Race/Ethnicity		
Non-Hispanic White	612	358
Non-Hispanic Black	136	496
Non-Hispanic Asian	53	364
Hispanic	368	430
Marital Status		
Single	533	396
Married	674	393
Insurance		
Public	721	418
Private	487	390
Education		
<12th grade	185	527
12th grade	271	434
>12th grade	722	372
Obese (BMI greater than or equal to 30)		
Yes	310	403
No	844	394

*A core city (Central Falls, Pawtucket, Providence, and Woonsocket) has a poverty level higher than 25%.

Table 2. Birth Defect Prevalence Rates by Body System, Rhode Island, 2018–2020

Birth Defect	ICD-10 'Q' Codes	Counts (n)	Rate per 10,000 live births
Cardiovascular	Q20-Q28	526	171
Genitourinary	Q50-Q64	484	157
Musculoskeletal	Q65-Q79	380	123
Central Nervous System	Q00-Q07	103	33
Chromosomal	Q90-Q99	83	27
Gastrointestinal	Q38-Q45	62	20
Eye/Ear/Face/Neck	Q10-Q18	56	18
Orofacial	Q35-Q37	35	11
Respiratory	Q30-Q34	19	6

DISCUSSION

Rhode Island birth defects prevalence rates began to increase in 2018. It is uncertain if this increase is seen nationally or in other states, as there are many factors complicating this comparison. Birth defects reporting to the Centers for Disease Control and Prevention (CDC) is not required. The National Birth Defects Prevention Network (NBDPN) has surveillance guidelines that only include 47 major birth defects, while Rhode Island's case definition is inclusive of most birth defects. Additionally, there is variation in the surveillance methods used by states and variations in maternal characteristics by state, which would affect the rates produced. The NBDPN only publishes state-level, not national, prevalence estimates for the 47 birth defects with 2014 to 2018 being the most recent years available, by race/ethnicity, maternal age, and infant sex. Because of Rhode Island's size and case counts, the RIBDP can conduct statewide birth defects surveillance and have more current data, which is important to identify trends over time.

Consistent with previous years, newborns born to mothers residing in a core city, having a maternal education level less than 12th grade, identifying with a non-Hispanic Black or African American race/ethnicity, and having public insurance had higher birth defects prevalence rates. Rhode Island is beginning to see shifts in the most prevalent maternal characteristics of newborns diagnosed with birth defects. Our current prevalence rates are similar when looking at marital status and are now highest in the maternal age group of 35 years of age and older. The most recent five-year period (2013–2017) not including the birth cohorts in our analyses suggested birth defects prevalence for each respective characteristic was highest in maternal age groups of under 25 and newborns born to women with a marital status of single.[8] Based on our findings, RIBDP will continue to monitor trends in prevalence rates to assist in informing our outreach efforts and existing birth defects-related policies. It is possible that shifts in maternal characteristics trends are related to the decreasing birth population size over time and changes in the characteristics of the birth population. The RIBDP will also work with partners in RIDOH's Health Equity Institute to determine if there are any different outreach strategies that can be used to reach these populations in outreach and prevention efforts.

Additionally, previous Rhode Island case control studies suggested that Rhode Island women who were obese prior to pregnancy had an increased risk of having a baby with a birth defect[5,6], while the prevalence rates for 2018 to 2020 did not vary when examining by maternal obesity status. The RIBDP plans to conduct an updated case control study with these more recent data to determine if the trends observed previously for maternal obesity still hold for recent years.

Compared to previous years, the cardiovascular system continues to have the highest prevalence rate among body systems.[8] Although the genitourinary and musculoskeletal systems have also had higher rates than other body systems in previous years, the genitourinary system has the second highest prevalence from 2018 to 2020, whereas from 2013 to 2017, it was the third highest prevalence.[8] The RIBDP will work with its Birth Defects Advisory Council to determine if there is a true increase in prevalence in the genitourinary body system which should be explored or if it could be due to any changing diagnostic practices in recent years.

With an increase in the prevalence rate of birth defects in Rhode Island in recent years, it is important to more frequently analyze data to examine trends in prevalence over time. In addition to the typical five-year timeframes, the RIBDP will also conduct analyses by two- or three-year timeframes to help better identify changes in prevalence rates by maternal characteristics. Future analyses will focus on prevalence rates for certain maternal characteristics by body systems to further focus birth defects awareness efforts, as was previously done with maternal obesity.[5,6]

It is possible the recent increase in birth defects prevalence is due to an unknown risk factor, as not all birth defects have known causes or can be prevented.[1,2] There are some modifiable risk factors, such as pre-pregnancy weight, the RIBDP can focus on to help reduce birth defects prevalence in Rhode Island. As pre-term birth and low birthweight have also been tied to birth defects[4], the RIBP will study how these are related to receipt of prenatal care. Additionally, there may be environmental risk factors, which are not included in birth certificate data, that are contributing to increasing prevalence. The RIBDP will continue to examine prevalence of maternal characteristics in its birth defects case data to help focus future outreach and prevention efforts in Rhode Island.

LIMITATIONS

All maternal characteristics were self-reported on the birth certificate, which may over- or underestimate some characteristics, such as BMI, in the birth population and birth defects cases.

References

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