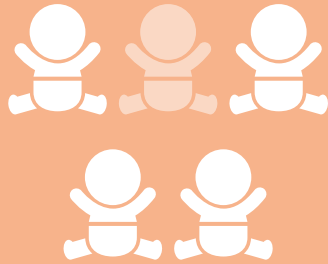




RHODE ISLAND
BIRTH DEFECTS
DATA BOOKLET
2021



Birth defects cause about **one in five** infant deaths in Rhode Island and the United States.



BIRTH DEFECTS SURVEILLANCE IN RHODE ISLAND

Birth defects are structural abnormalities that affect the development of organs and tissues of an infant or child. These abnormalities may be identified during pregnancy, at birth, or following birth. Possible causes or contributing factors of birth defects include genetics, environmental pollutants, occupational hazards, diet, medications, and personal behaviors.

Early recognition of, and response to, birth defects often prevents more serious effects. A birth defects surveillance and information system is essential for the development of programs and policies that can reduce birth defects and infant mortality. At the Rhode Island Department of Health (RIDOH), the Rhode Island Birth Defects Program (RIBDP) maintains this surveillance system. The RIBDP identifies newborns with birth defects; assures that these children receive appropriate preventive, specialty, and other healthcare services; and monitors trends over time. All information collected by the RIBDP is confidential and is protected under state and federal privacy laws.

All healthcare providers are mandated, by regulation, to report cases of birth defects identified among children up to age five, to the RIBDP. The reporting of birth defects cases helps the RIBDP assure that these children receive appropriate services and referrals on a timely basis and helps identify children who were not diagnosed with a birth defect at the time of birth. The RIBDP also works with all five birthing hospitals in Rhode Island to capture birth defects diagnosed at birth using hospital discharge data.

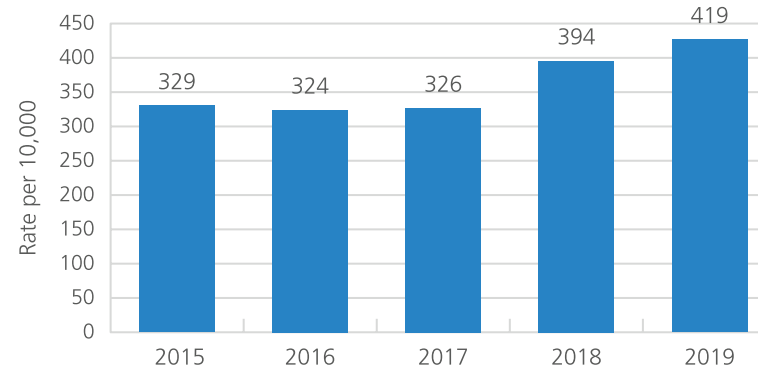
Birth defects cases include children born to Rhode Islanders, from birth to age five, and are identified using the 10th clinical modification of the International Classification of Diseases (ICD 10-CM) codes for diagnoses. The RIBDP confirms the accuracy of reported birth defects diagnoses through chart review and follows birth defects surveillance guidelines developed by the National Birth Defects Prevention Network (NBDPN).

Identification of Cases During the Newborn Period

From 2015 to 2019, the birth defects rate increased by 27%, from 329 per 10,000 live births in 2015 to 419 per 10,000 live births in 2019 (Figure 1).



FIGURE 1: Prevalence of Birth Defects Cases, Rhode Island, 2015-2019



Source: Rhode Island Birth Defects Program

The overall prevalence rate of birth defects from 2015 to 2019 was 358 cases per 10,000 live births (Table 1). Cardiovascular defects were the most common type of defect (148 per 10,000). Other common birth defects in Rhode Island include those related to musculoskeletal (116 per 10,000) and genitourinary (109 per 10,000) systems.

TABLE 1: Cases and Prevalence of Birth Defects by Body System, Rhode Island, 2015-2019

BIRTH DEFECT	NUMBER OF CASES	RATE (PER 10,000 LIVE BIRTHS)
Cardiovascular	782	148
Musculoskeletal	614	116
Genitourinary	579	109
Central Nervous System	158	30
Gastrointestinal	127	24
Chromosomal	119	22
Eye Ear Face Neck	78	15
Orofacial	73	14
Respiratory	43	8
All birth defects	2,636	498
All birth defects cases	1,893	358

Source: Rhode Island Birth Defects Program

Note: All birth defects represent all birth defects diagnosed in Rhode Island from 2015-2019. All birth defects cases represent the total number of Rhode Island babies born from 2015-2019 with at least one diagnosed birth defect. One case can have multiple birth defects.

Before or during early pregnancy, women should **avoid getting overheated**, should treat fevers promptly, and take 400 micrograms (mcg) of folic acid every day to prevent neural tube defects.



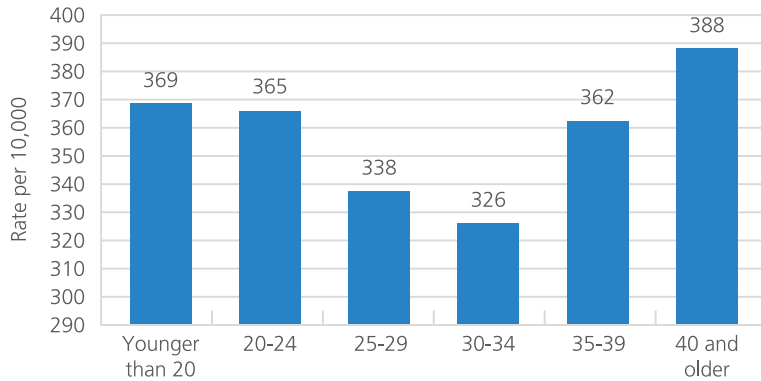
Before women become pregnant, they should maintain a healthy weight, quit smoking, and **talk to their doctor about medication they are taking** that may increase the risk of having a baby with congenital heart defects.



Maternal Risk Factors

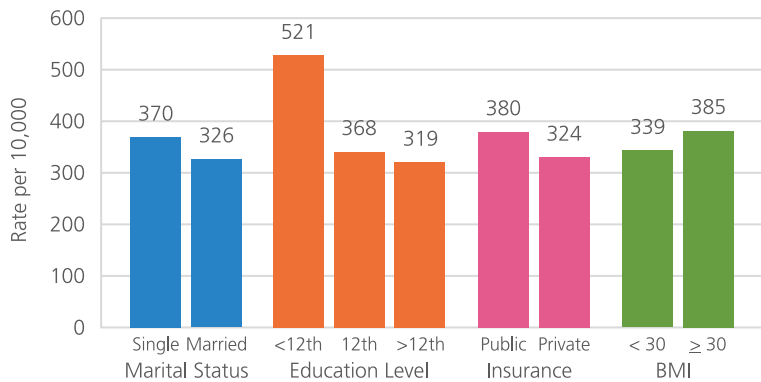
Babies born to women with certain maternal characteristics were at a higher risk of having a birth defect (Figures 2 and 3). From 2015 to 2019, women who were unmarried, had less than a high school education, were insured through public programs (such as Rite Care and Medicaid), were 40 and older, and had a pre-pregnancy body mass index (BMI) greater than or equal to 30 were more likely to have a baby born with a birth defect.

FIGURE 2: Prevalence of Birth Defects by Maternal Age Group, Rhode Island, 2015-2019



Source: Rhode Island Birth Defects Program

FIGURE 3: Prevalence of Birth Defects by Selected Maternal Characteristics Rhode Island, 2015-2019

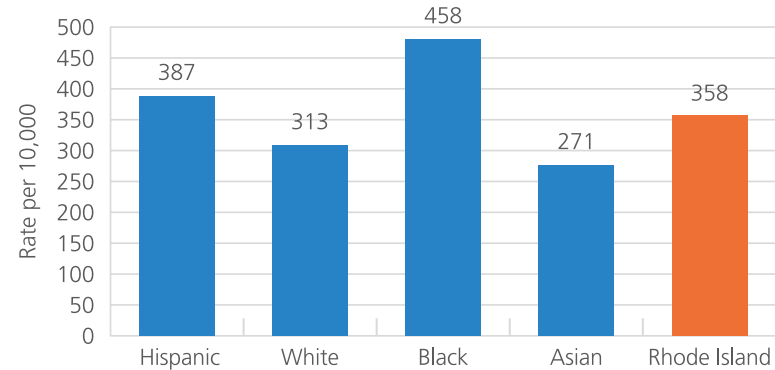


Source: Rhode Island Birth Defects Program

Racial/Ethnic and Geographic Disparities

Birth defects prevalence also varied by race/ethnicity (Figure 4) and by geography (Figure 5). During 2015-2019, non-Hispanic Blacks/African Americans and Hispanics had higher birth defects rates than non-Hispanic Whites and non-Hispanic Asians. The birth defects prevalence rate for non-Hispanic Asians and non-Hispanic Whites was lower than the Rhode Island prevalence rate for this timeframe.

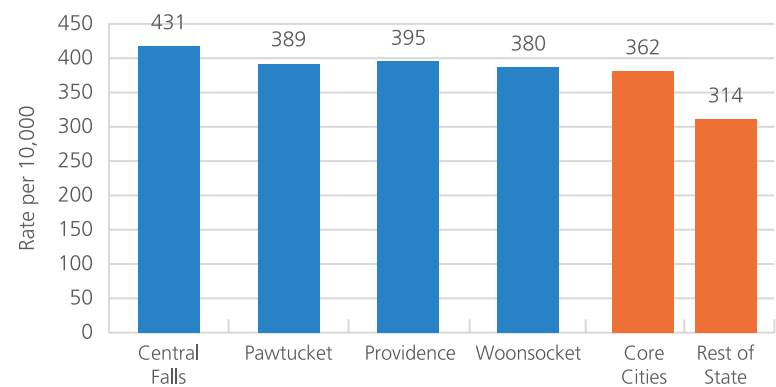
FIGURE 4: Prevalence of Birth Defects by Race/Ethnicity, Rhode Island, 2015-2019



Source: Rhode Island Birth Defects Program

Babies born to women who live in core cities where the poverty level is higher than 25% (Central Falls, Pawtucket, Providence, and Woonsocket) were about 1.2 times more likely to have a birth defect than babies born to residents living in the rest of the state (Figure 5).

FIGURE 5: Prevalence of Birth Defects by Selected Geographic Areas, Rhode Island, 2015-2019



Source: Rhode Island Birth Defects Program

Women should **avoid getting infections while pregnant** by practicing good hygiene, getting vaccinated, prevent insect bites, and wash fruits and vegetables before eating them.



There is **no known safe amount, no safe time, and no safe type** of alcohol to drink during pregnancy.



Critical Congenital Heart Defects

Critical congenital heart defects (CCHD) are a range of 12 heart defects that can cause serious, life threatening symptoms (see Table 2 for list). CCHD may require intervention and, commonly, surgery within the first days of a newborn’s life. These birth defects can involve abnormal heart rhythms and structural heart problems, including abnormal or absent chambers, holes in the heart, abnormal connections, and abnormal functioning. Babies who are not diagnosed or treated soon after birth are at high risk of death and disabilities later in life. To aid in early detection of birth defects, newborn pulse oximetry screening can help identify CCHD before symptoms appear. By identifying CCHD among newborns early, the appropriate care and treatment can be provided.

Based on a recommendation from the US Health and Human Services (HHS) Secretary’s Advisory Committee on Heritable Disorders in Newborns and Children (SACHDNC), CCHD was added to the newborn screening panel in Rhode Island in 2015, requiring birthing facilities to perform a pulse oximetry screening within 24 hours of the birth of every newborn in its care. The case counts of the 12 birth defects associated with CCHD from 2015 to 2019 are listed in Table 2.

TABLE 2: Cases of Critical Congenital Heart Defects, Rhode Island, 2015-2019

CCHD	CASES
Tetralogy of Fallot	21
Coarctation of aorta	18
Pulmonary valve atresia (with intact septum)	15
Hypoplastic left heart syndrome	10
Double outlet right ventricle	9
Transposition of great arteries	9
Total anomalous pulmonary venous return	4
Single ventricle	3
Ebstein's anomaly	2
Truncus arteriosus	1
Tricuspid valve atresia	0
Interrupted aortic arch	0
All critical congenital heart defects	92
All CCHD cases	84

Source: Rhode Island Birth Defects Program

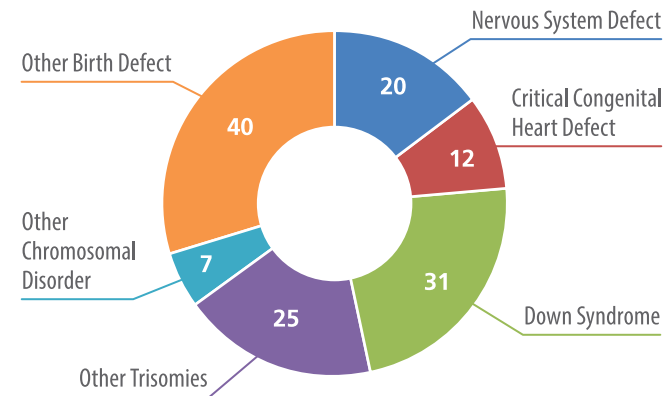
Note: All critical congenital heart defects represent all critical congenital heart defects diagnosed in Rhode Island from 2015-2019. All CCHD cases represent the total number of Rhode Island babies born from 2015-2019 with at least one diagnosed CCHD. One case may have multiple CCHDs.

Prenatal Data

Since 2008, the RIBDP has been collecting birth defects cases identified during the prenatal period through collaborating laboratories and prenatal clinics, including the Cytogenetics Testing Laboratory, Prenatal and Special Testing Laboratory at Women & Infants Hospital, and the Fetal Treatment Program at Hasbro Children’s Hospital. About four to six percent of birth defects are identified prenatally. Prenatal case identification improves the prevalence estimate of certain birth defects by detecting cases not found at newborn discharge.

From 2015 to 2019, there were 135 birth defects among the 104 birth defects cases that were identified prenatally. Chromosomal abnormalities (including Down syndrome and other trisomies; n =56) accounted for approximately 41% of all cases (Figure 6). Specifically, Down syndrome was identified in 23% of all prenatally ascertained cases (n = 31). The largest proportion of prenatally ascertained cases were among women age 35 or older (n = 55), accounting for 53% of prenatally identified birth defects cases (Figure 7).

FIGURE 6: Prenatally Ascertained Birth Defects*, By Type, Rhode Island, 2015-2019



*Data include fetal losses (spontaneous fetal deaths and induced terminations).

Source: Rhode Island Birth Defects Program

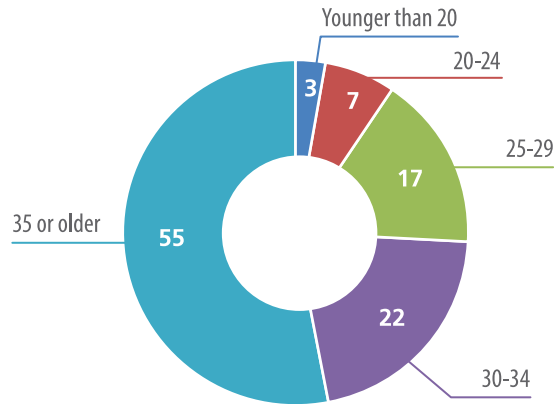
The use of **marijuana** and other drugs during pregnancy **can lead to preterm birth and birth defects.**



Smoking during pregnancy **increases the chances of premature birth, certain birth defects, and infant death.**



FIGURE 7: Prenatally Ascertained Birth Defects*, By Maternal Age, Rhode Island, 2015-2019



*Data include fetal losses (spontaneous fetal deaths and induced terminations).

Source: Rhode Island Birth Defects Program

Service Assessments

A priority of the RIBDP is to assure that children with birth defects receive appropriate and timely preventive, specialty, and other healthcare services. The RIBDP, in collaboration with the Rhode Island Parent Information Network (RIPIN), employs a Parent Consultant who conducts service assessments with families who have children up to age five with specific birth defects to determine whether these children have received appropriate referrals and services on a timely basis. The Parent Consultant meets with families at pediatric and specialty care practices that serve children with birth defects or mails forms to those families who cannot be interviewed in a practice. Follow-up service assessments are conducted to ensure continuity of referrals and services until the child is five years old.

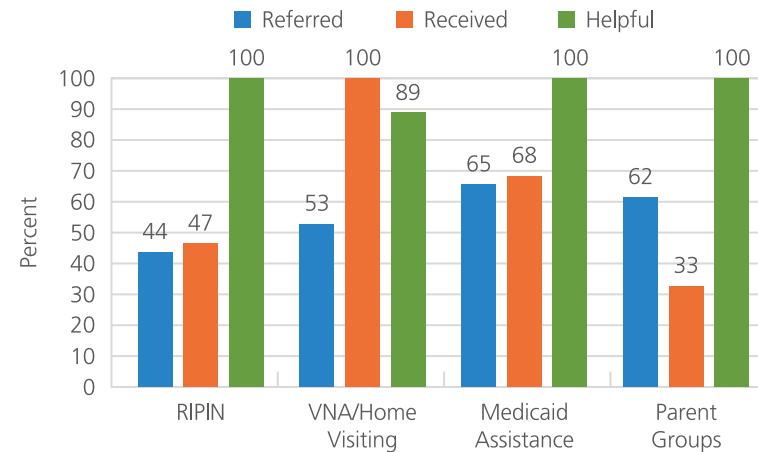
Service assessments help the RIBDP determine what services and referrals were provided to children based on the national guidelines for specific conditions. The RIBDP has conducted more than 1100 new and follow-up service assessments to date.

Service assessments are currently conducted with families of children who have Down syndrome, spina bifida, craniofacial defects, critical congenital heart defects, abdominal wall defects, hearing loss, and microcephaly or other central nervous system conditions.

The RIBDP recently evaluated educational, developmental, and family support service referrals reported by families of children with Down syndrome. From 2016 to 2020, 34 families completed service assessments for children with Down syndrome, and 29 families (85%) completed more than one assessment. Most initial assessments were completed at a clinic while most repeat assessments were completed through mail.

For family support services for families of children with Down syndrome (Figure 8), the highest referral rates were for Medicaid assistance (65%) and parent groups (62%). There were fewer referrals to the Visiting Nurses Association (VNA)/Home visiting (53%) and RIPIN (44%). Families were more likely to receive VNA/Home visiting (100%) and Medicaid assistance (68%) once referred. Of all families who received support services, 100% of families found RIPIN, Medicaid assistance, and parent groups helpful.

FIGURE 8: Family Support Service Referral and Receipt by Families of Children with Down Syndrome, Rhode Island, 2016-2020



Source: Rhode Island Birth Defects Program

Families eligible for developmental and educational services were most likely to be referred to Early Intervention (88%). Families were least likely to receive referrals to special education (40% of children age three or older) (Figure 9). Those referred to the Children's Neurodevelopment Center (CNDC) at Hasbro Children's Hospital and Early Intervention were very likely to receive these services (96% and 80% respectively). Overall, families who received educational and developmental services found them helpful.

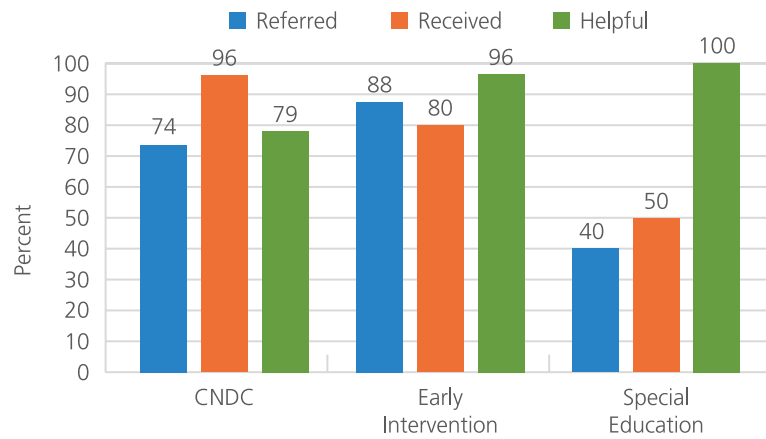
Proper management of chronic conditions, such as diabetes, can help prevent birth defects and other poor outcomes.



In the United States, a baby is born with a birth defect every 4.5 minutes.



FIGURE 9: Educational and Developmental Support Service Referral and Receipt by Families of Children with Down Syndrome, Rhode Island, 2016-2020



Source: Rhode Island Birth Defects Program

Prevention Activities

Although not all causes of birth defects are known, there are many things a woman can do before and during pregnancy to reduce the risk of having a baby with a birth defect. These include getting routine prenatal check-ups; taking folic acid supplements before and during pregnancy; avoiding tobacco, alcohol, and other harmful substances; eating a healthy diet; getting appropriate levels of exercise; preventing exposure to chemicals; and managing existing medical conditions (diabetes, epilepsy, and high blood pressure).

The RIBDP works with RIDOH's Family Planning Program to purchase and distribute multivitamins with folic acid to uninsured women who receive a negative pregnancy test at family planning clinics. Additionally, the program collaborates with the Women's Facility at the Rhode Island Department of Corrections to distribute multivitamins with folic acid to women who are incarcerated.

Data Dissemination and Partnerships

Sharing data and information on birth defects with healthcare providers, policy makers, community organizations, families, and other stakeholders can increase awareness of birth defects and lead to program enhancements and policy development. The RIBDP uses a multi-pronged approach to data dissemination, including maintaining an up-to-date website; publishing studies in peer-reviewed journals; presenting information at state, local, and national meetings; and sponsoring grand rounds at local hospitals.

The RIBDP participates in collaborative studies with the National Birth Defects Prevention Network (NBDPN) to gain a better understanding of specific birth defects and their causes. The RIBDP also works in partnership with its Advisory Council, which includes representatives from Women & Infants Hospital, Hasbro Children's Hospital, the March of Dimes, and RIPIN. The Advisory Council provides guidance to the RIBDP in the development and implementation of its surveillance, prevention, service assurance, and information dissemination strategies.





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health.ri.gov/birthdefects