

**RHODE ISLAND
BIRTH DEFECTS
DATA BOOK
2018**

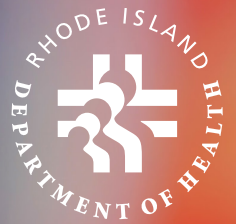


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INTRODUCTION

What are Birth Defects?

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 (inherited), environmental pollutants, occupational hazards, diet, medications, and personal behaviors.

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

Why Study Birth Defects?

Birth defects cause serious illness and death for many babies

Each year in the United States, one in 33 (about 120,000) babies are born with a birth defect, and of these, 8,000 (6.7%) die during the first year of life. Many babies who do survive beyond the first year experience childhood illness and disability.

Birth defects cause about one in five infant deaths in both the United States and in Rhode Island. Provisional data indicate that among the 123 infants who died in 2016 and 2017, 24 of the deaths (19.5%) were attributed to a birth defect. This represents a 103% increase in the proportion of infant deaths resulting from a birth defect since 2011-2012, when 13 (9.7%) of the 134 infant deaths resulted from a birth defect.

Many preterm infants (born before 37 weeks gestation) have birth defects. In Rhode Island, 63 (6.3%) of the 994 preterm babies born in 2016 had a birth defect. Preterm birth is the leading cause of infant death in Rhode Island. During the two-year period, 2016-2017, 38 (30.9%) of the 123 infant deaths were attributed to prematurity.

Birth defects have serious economic costs

In addition to the emotional impact of birth defects that families often experience, birth defects have financial implications for families, the healthcare system, and society. Understanding the economic burden can help drive prevention activities and policy decisions.

The Rhode Island Birth Defects Program (RIBDP) at the Rhode Island Department of Health (RIDOH) studies the costs of selected birth defects using national surveillance guidelines based on the severity and frequency of the birth defect. During 2014-2016, Rhode Island's hospital discharge database identified 1,735 newborns with at least one birth defect. The total unadjusted cost for newborn admissions with a birth defect diagnosis is \$207,004,289 with an average cost per newborn of \$119,311. This amounts to about 10 times the cost for a newborn with no birth defects (\$11,970). The average length of stay for a newborn with birth defects (14.8 days) was four times longer than that for a newborn without a birth defect (3.5 days).

Similarly, a comparison of hospitalization data for children (younger than age five) with birth defects compared to children without birth defects indicates that the average length of stay for children with birth defects (6.2 days) was approximately twice as long as the average length of stay for children without birth defects (2.7 days).

Hospital discharge data provide adjusted charges and specific hospital costs for newborn admissions. The Rhode Island analysis shows the total adjusted charges for all newborn hospital admissions with at least one diagnosed birth defect and the total costs by selected birth defects. Table 1 compares the number of cases, mean lengths of stay, and mean hospital charges per case (based on hospital cost-to-charge ratios) in Rhode Island for selected birth defects.

TABLE 1: MEAN TOTAL ADJUSTED HOSPITALIZATION CHARGES, BY SELECTED BIRTH DEFECTS, RHODE ISLAND, 2014-2016

Birth Defect	Number of Rhode Island Cases	Length of Stay (Days)	Adjusted Cost (\$)	Cost per day (\$)
Spina bifida	4	2	5,346	2,673
Microcephaly	3	4	13,552	3,388
Cleft lip with and without cleft palate	7	1	7,084	7,084
Rectal and large intestinal atresia/stenosis	7	6	13,945	2,324
Gastroschisis	1	49	69,305	1,414
Down syndrome	4	4	8,985	2,567

Note: Costs are based on hospital cost-to-charge ratios by fiscal year
 Sources: Rhode Island Hospital Discharge Database, Rhode Island Department of Health



PUBLIC HEALTH SURVEILLANCE: RHODE ISLAND BIRTH DEFECTS PROGRAM

Early recognition and response to birth defects often prevents more serious effects. An active birth defects surveillance and information system is essential for the development of programs and policies that can reduce birth defects and infant mortality.

Rhode Island developed a birth defects information system in 2000, funded by the Centers for Disease Control and Prevention (CDC). The RIBDP is housed within RIDOH's Center for Health Data and Analysis. The RIBDP was created to identify newborns with birth defects; assure that these children receive appropriate preventive, specialty, and other healthcare services; and monitor trends. All information collected by the RIBDP is confidential and is protected under state and federal privacy laws.

In 2003, the Rhode Island General Assembly enacted legislation (General Laws 23-13.3) requiring the development and implementation of a birth defects reporting, surveillance, and information system. This system describes the occurrence of birth defects in children up to age five; detects morbidity (disease) and mortality (death) trends; and helps assure children with birth defects receive services and treatment on a timely basis.

The Director of RIDOH created the Rhode Island Birth Defects Advisory Council to advise RIDOH on the establishment and implementation of the system and to recommend a list of reportable birth defects. It is critical that State agencies, healthcare service providers, community organizations, parents, and other key stakeholders provide input to help RIDOH develop the surveillance system and analyze and disseminate information. Stakeholders are represented on the Advisory Council. The RIBDP also solicits input directly via surveys, focus groups, and interviews.

Reportable Birth Defects

In 2005, regulations were enacted mandating all healthcare providers to report cases of birth defects identified among children, up to age five, to RIDOH. 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. In 2011, the RIBDP worked with KIDSNET, RIDOH's integrated child information system, to build a reporting component that would allow pediatric providers to report birth defects cases electronically. RIBDP staff and KIDSNET provider liaisons trained pediatric providers and office staff to report birth defects using the web-based KIDSNET reporting system.

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

CASE IDENTIFICATION AND DATA

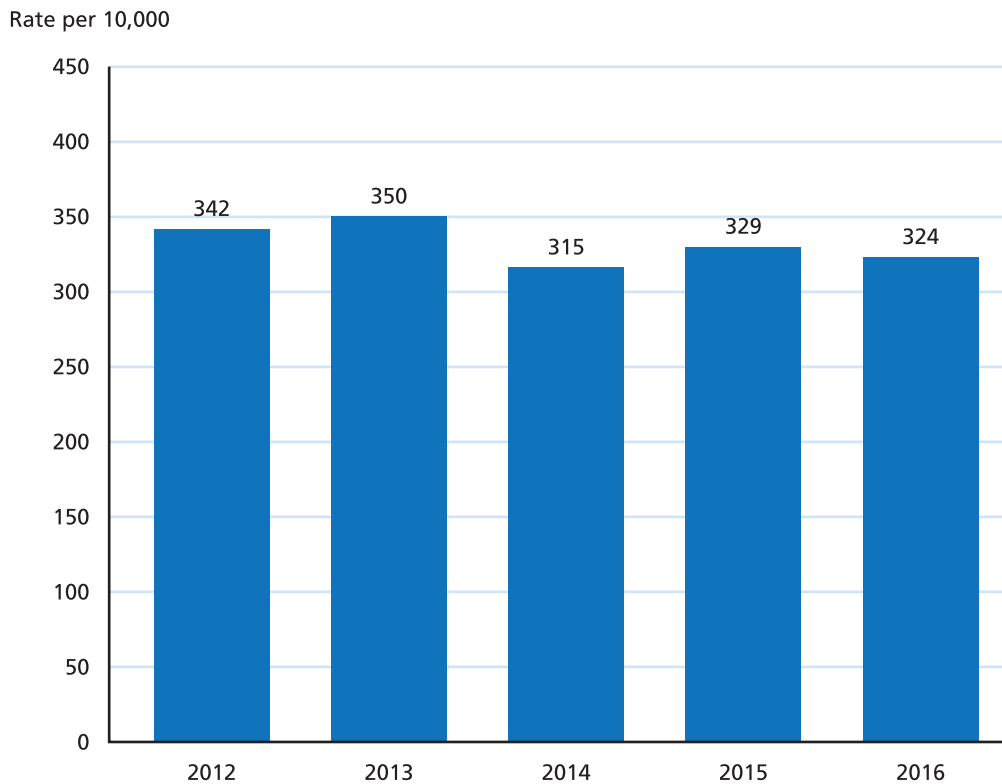
The RIBDP uses hospital discharge data as the primary source for capturing birth defects data in Rhode Island. The RIBDP works with all five maternity hospitals to collect discharge information. The RIBDP also collects information from specialty clinics, such as the Children’s Neurodevelopment Center (CNDC) at Rhode Island Hospital, to obtain additional cases and information on services provided to families of children with birth defects.

Birth defects cases are children born to Rhode Island residents, from birth up to age five, and are identified using diagnoses coded by the 10th clinical modification of the International Classification of Diseases (ICD 10-CM) and include all Q codes. The RIBDP confirms the accuracy of birth defects diagnoses through chart review. Exclusion criteria to omit certain minor congenital anomalies and focus on more relevant conditions for data analysis and service assurance were identified and are included in the RIBDP’s birth defects case definition (see Appendix 1). The RIBDP follows birth defects surveillance guidelines developed by the National Birth Defects Prevention Network. Previous data have been adjusted to fit the current case definition for comparable data analyses.

Identification of Cases During the Newborn Period

Figure 1 shows the overall prevalence of birth defects identified in newborns in Rhode Island from 2012 through 2016. During this period, the rate of birth defects in Rhode Island decreased by 5% from 342 per 10,000 live births in 2012 to 324 per 10,000 live births in 2016, after adjusting for the updated birth defects case definition.

FIGURE 1. PREVALENCE OF BIRTH DEFECTS CASES, RHODE ISLAND, 2012-2016



Source: Rhode Island Birth Defects Program, Rhode Island Department of Health

Table 2 shows the number and prevalence of selected birth defects among Rhode Island residents during 2012-2016, organized by organ system. Cardiovascular defects were the most common type of defect (129 per 10,000). Among these defects, ventricular septal defects represent the highest proportion of cases.

Other common birth defects in Rhode Island include those related to musculoskeletal (122 per 10,000) and genitourinary (87 per 10,000) systems. Among musculoskeletal defects, club foot is most common, but rates have remained stable over time. Within the genitourinary system, hypospadias represents more than half (60%) of those conditions and had a prevalence rate in males of 103 cases per 10,000.

TABLE 2. CASES AND PREVALENCE OF SELECTED BIRTH DEFECTS, RHODE ISLAND, 2012-2016

BIRTH DEFECT	NUMBER	RATE (PER 10,000 LIVE BIRTHS)	BIRTH DEFECT	NUMBER	RATE (PER 10,000 LIVE BIRTHS)
Central Nervous System	103	19	Orofacial	82	15
Microcephaly	31		Cleft lip with cleft palate	33	
Hydrocephaly	19		Cleft palate alone	26	
Spina bifida	16		Cleft lip alone	23	
Anencephaly	4		Gastrointestinal	115	21
Encephalocele	5		Small intestinal atresia/stenosis	24	
Holoprosencephaly	1		Rectal and large intestinal atresia/stenosis	20	
Eye/Ear/Face/Neck	47	9	Esophageal atresia/tracheoesophageal fistula	12	
Anophthalmos/Microphthalmos	9		Hirschsprung's disease	7	
Congenital cataract	9		Biliary atresia	1	
Anotia/Microtia	3		Genitourinary	471	87
Cardiovascular	698	129	Hypospadias	282	
Ventricular septal defect	233		Renal agenesis/hypoplasia	22	
Atrial septal defect	119		Congenital posterior urethral valves	7	
Pulmonary valve atresia and stenosis	27		Bladder extrophy	1	
Tetralogy of Fallot	15		Musculoskeletal	659	122
Coarctation of aorta	13		Clubfoot	77	
Atrioventricular septal defect	10		Gastroschisis	31	
Transportation of great arteries	6		Limb deficiencies	21	
Interrupted aortic arch	6		Craniosynostosis	15	
Tricuspid valve atresia	6		Diaphragmatic hernia	12	
Hypoplastic left heart syndrome	5		Omphalocele	10	
Aortic valve stenosis	4		Chromosomal	99	18
Ebstein's anomaly	3		Down syndrome	62	
Double outlet right ventricle	3		Edward syndrome	3	
Total anomalous pulmonary venous connection	3		Patau syndrome	3	
Single ventricle	3		Turner syndrome	2	
Truncus arteriosus	1		All birth defects	2,313	427
Respiratory	39	7	All birth defect cases	1,796	332
Choanal atresia	7				

Note: Numbers and rates in each bolded body system row represent total diagnosed birth defects associated with that body system. The bolded All birth defects row represents all birth defects diagnosed in Rhode Island during 2012-2016. The bolded All birth defects cases row represents the total number of Rhode Island babies born between 2012-2016 with at least one diagnosed birth defect.

Source: Rhode Island Birth Defects Program, Rhode Island Department of Health

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 3 for list). CCHD may require intervention and, commonly, surgery within the first days of a newborn’s life. These birth defects can involve abnormalities in rhythm of the heart 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. Newborn pulse oximetry screening, however, can help detect CCHD before symptoms appear. Identifying these newborns early helps them get appropriate care and treatment.

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. Conducting pulse oximetry screening, which measures oxygenation of the blood, allows for identification of newborns that may have CHHD before symptoms appear. A failed screen due to low blood oxygenation saturation (lower than 90%) is likely to indicate the presence of CCHD. By identifying CCHD among newborns early, the appropriate care and treatment can be provided.

The counts of the 12 birth defects associated with CCHD from 2012 to 2016 are listed in Table 3.

TABLE 3. CRITICAL CONGENITAL HEART DEFECTS, RHODE ISLAND, 2012-2016

CONDITION	COUNT
Tetralogy of Fallot	18
Coarctation of aorta	18
Transposition of great arteries	11
Pulmonary valve atresia (with intact septum)	9
Hypoplastic left heart syndrome	9
Tricuspid valve atresia	5
Ebstein’s anomaly	5
Double outlet right ventricle	4
Single ventricle	3
Total anomalous pulmonary venous return	3
Interrupted aortic arch	2
Truncus arteriosus	1

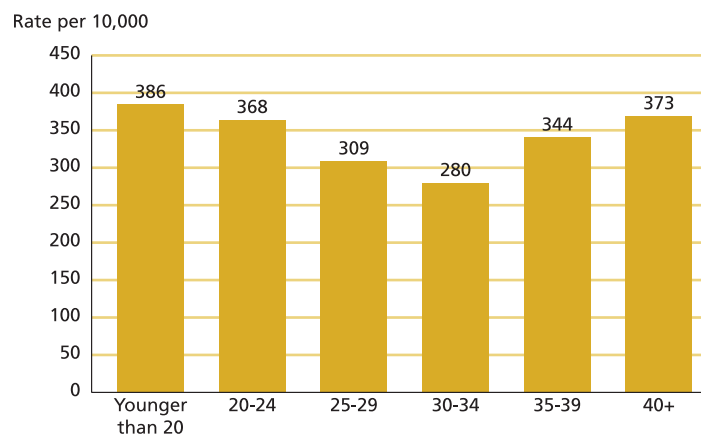
*Notes: Numbers and rates in this table may be a subset of those conditions listed in Table 2 and may not match counts in Table 2.
Source: Rhode Island Birth Defects Program, Rhode Island Department of Health*

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

Maternal Characteristics

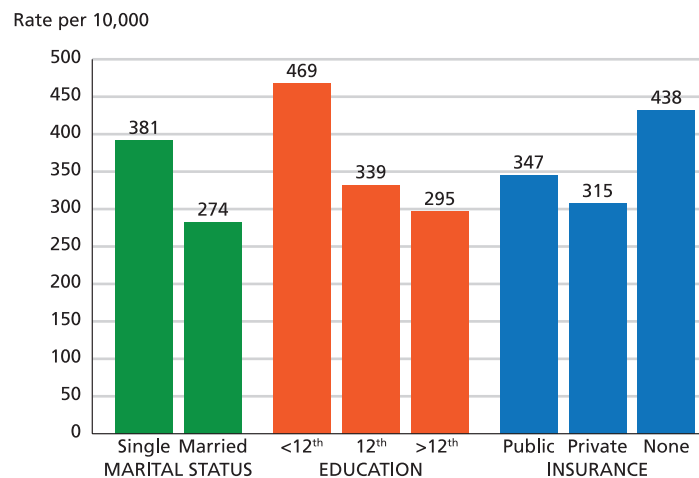
Babies born to women younger than 25 or age 40 and older, women with less than a high-school education, unmarried women, and women with publicly funded health insurance or no health insurance are at a higher risk for birth defects (Figures 2 and 3). During 2012-2016, the birth defects prevalence rate among women younger than 25 was 372 per 10,000 live births and the rate among women age 40 and older was 373 per 10,000 live births compared to lower rates among women age 25-34. The birth defects rate among women with less than a high school education (469) or with a high school education (339) was higher than the rate among women with more education (295). Similarly, unmarried women were more likely to have a baby with a birth defect (381) than married women (274). Women who did not have insurance (438) or were insured through public programs such as RItte Care and Medicaid (347) were more likely to have a baby born with a birth defect than women who were insured through private insurance (315).

FIGURE 2. PREVALENCE OF BIRTH DEFECTS, BY MATERNAL AGE GROUP, RHODE ISLAND, 2012-2016



Source: Rhode Island Birth Defects Program, Rhode Island Department of Health

FIGURE 3. PREVALENCE OF BIRTH DEFECTS, BY SELECTED MATERNAL CHARACTERISTICS, RHODE ISLAND, 2012-2016



Source: Rhode Island Birth Defects Program, Rhode Island Department of Health

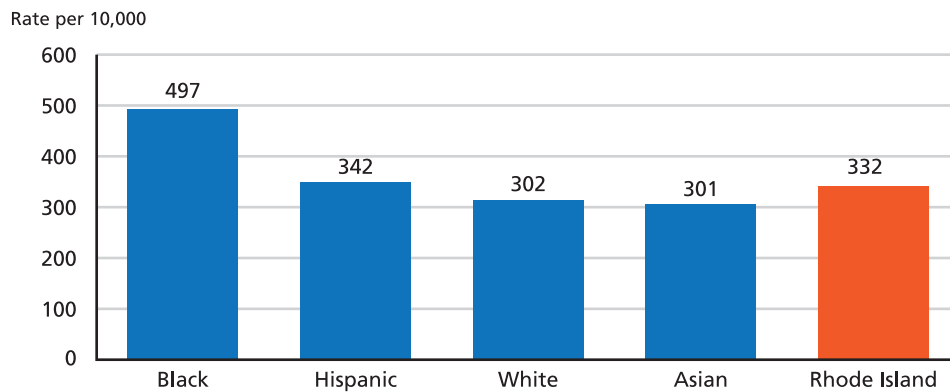
Smoking during pregnancy increases the chances of premature birth, certain birth defects, and infant death.

Racial/Ethnic and Geographic Disparities

Birth defects prevalence also varies by race/ethnicity (Figure 4) and by geography (Figure 5). During 2012-2016, the average birth defects prevalence rate among non-Hispanic Blacks/African Americans (497 per 10,000 live births) was 1.6 times the rate for non-Hispanic Whites (302). Hispanic/Latino ethnicity had the second highest rate (342), which was 13% higher than the rate for Whites. The birth defects prevalence rate among non-Hispanic Asians (301) was similar to the rate for non-Hispanic Whites.

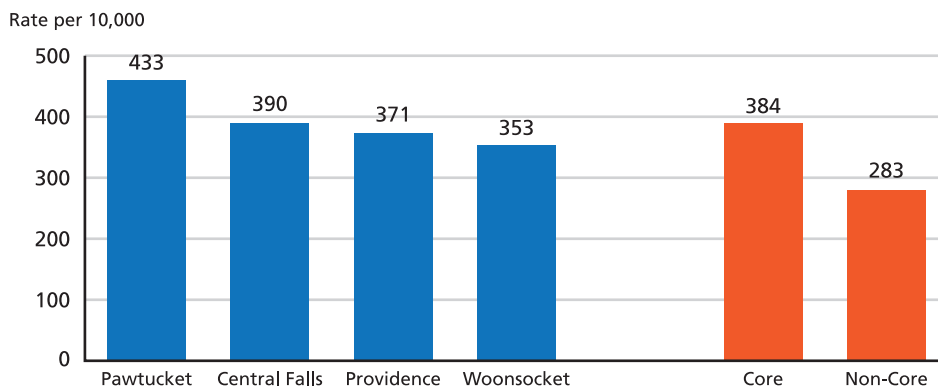
Babies born to residents of core cities where the poverty level is higher than 25% (Central Falls, Pawtucket, Providence, and Woonsocket) were about 1.4 times more likely to have a birth defect than babies born to residents living in the rest of the state (Figure 5). Pawtucket (433) and Central Falls (390) had two of the highest birth defects rates in the state. These two cities also have higher rates of teen pregnancy, low birth weight, late prenatal care (prenatal care started in third trimester or no prenatal care), and poverty compared to the rest of the state.

FIGURE 4. PREVALENCE OF BIRTH DEFECTS, BY RACE/ETHNICITY, RHODE ISLAND, 2012-2016



Source: Rhode Island Birth Defects Program, Rhode Island Department of Health

FIGURE 5. PREVALENCE OF BIRTH DEFECTS, BY SELECTED GEOGRAPHIC AREAS, RHODE ISLAND, 2012-2016



Source: Rhode Island Birth Defects Program, Rhode Island Department of Health

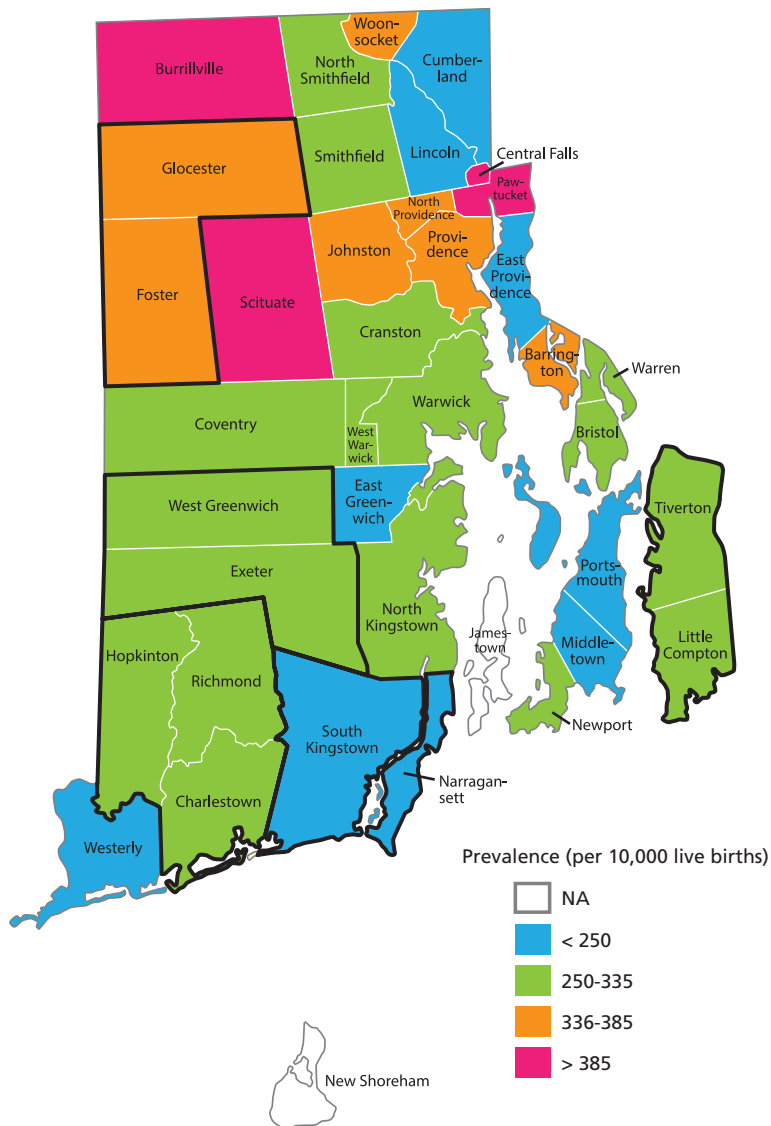
Mapping Rates of Birth Defects

Geospatial analysis allows us to follow trends and detect clusters of birth defects in Rhode Island. Birth defects prevalence rates from 2012-2016 were mapped by geographic area in Rhode Island. To address small sample sizes in some cities and towns, the RIBDP has implemented data suppression rules. Prevalence rates for towns with a case count less than 15 or a live birth population less than 200 during the 2012-2016 period are not reported. This includes the towns of Jamestown and New Shoreham. All other towns that did not pass the suppression rules alone for the five-year period were combined with other cities and towns into geographical regions that share proximity.

In 2012-2016, most cities/towns had a prevalence rate between 250 and 385 birth defects per 10,000 live births, a range that is common for birth defects rates (Figure 6). As mentioned previously, Central Falls and Pawtucket form an urban area of high birth defects prevalence in Rhode Island.

FIGURE 6. PREVALENCE OF BIRTH DEFECTS, BY GEOGRAPHIC AREAS, RHODE ISLAND, 2012-2016

Note: Map shown is not to scale or positional accuracy.



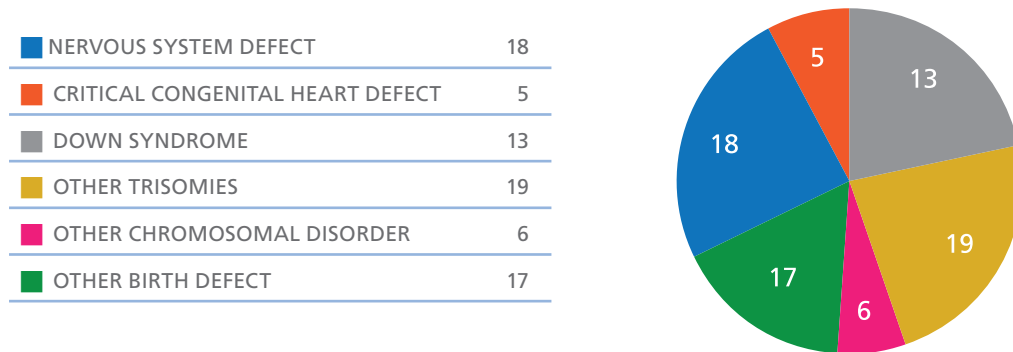
Source: Rhode Island Birth Defects Program and Rhode Island Geographic Information System, Rhode Island

Identification of cases during the prenatal period

In 2008, the RIBDP began collecting birth defects cases identified during the prenatal period from collaborating laboratories and prenatal clinics, such as the Cytogenetics Testing Laboratory and 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 ascertainment improves the prevalence estimate of certain birth defects by detecting cases not found at newborn discharge.

Figure 7 shows the percentages of birth defects diagnoses ascertained prenatally for the 2012-2016 period in Rhode Island. Among the 64 birth defects cases that were identified prenatally, chromosomal abnormalities (including Down syndrome and other trisomies; n = 38) account for almost half of cases. Specifically, Down syndrome was identified in 17% of all prenatally ascertained cases (n = 13). Figure 8 shows the maternal age distribution of prenatally ascertained birth defects cases. The largest proportion of prenatally ascertained cases were among women age 35 and older (n = 31), accounting for almost 50% of prenatal ascertained birth defects cases.

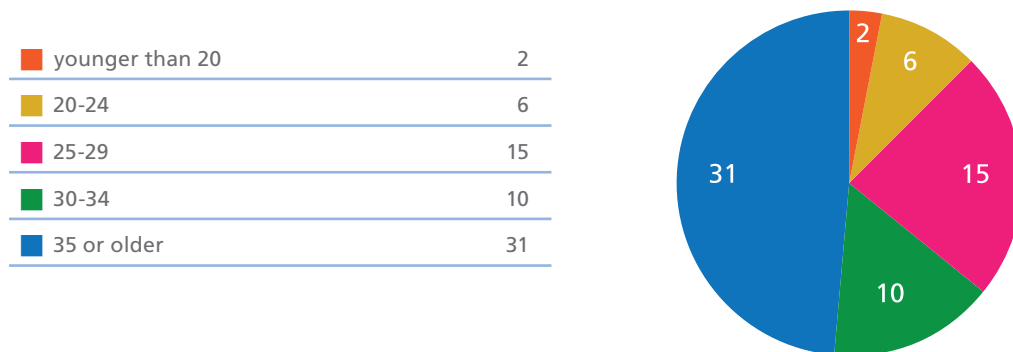
FIGURE 7. PRENATALLY ASCERTAINED BIRTH DEFECTS*, BY TYPE (N=78), RHODE ISLAND 2012-2016



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

Source: Rhode Island Birth Defects Program, Rhode Island Department of Health

FIGURE 8. PRENATALLY ASCERTAINED BIRTH DEFECTS*, BY MATERNAL AGE (N=64), RHODE ISLAND 2012-2016



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

Source: Rhode Island Birth Defects Program, Rhode Island Department of Health

Obesity before and during pregnancy increases the risk of several serious birth defects.

EMERGING ISSUES

Zika Virus

Zika virus (Zika) was first discovered in Uganda in 1947; however, human infection was not reported until 1952 in Nigeria. In Brazil, the spread of the virus grew to unexpected proportions, leading the World Health Organization (WHO) to declare a public health emergency in February 2016. Zika is primarily transmitted via mosquitoes from the Aedes family. Aside from mosquito transmission, Zika infection can be spread through sexual contact, blood transfusions, and from pregnant mothers to developing fetuses. Those infected with Zika may experience symptoms of fever, rash, joint pain, muscle pain, conjunctivitis, and/or headache; however, approximately 80% of infected people have no symptoms. In pregnant women, Zika presents an increased risk for more dangerous outcomes, including microcephaly and central nervous system defects in the developing fetus. Infants who are infected during the first trimester are at the greatest risk for developing Zika-related birth defects, including microcephaly, central nervous system defects, eye defects, and hearing loss.

The CDC is actively tracking the spread of Zika in the United States and rest of the world. Research regarding the link of Zika to other birth defects and anomalies is ongoing. The RIBDP participates in the CDC's Zika Birth Defects Surveillance System by monitoring all infants born in Rhode Island with birth defects associated with Zika that may or may not be related to Zika virus infection. The existing newborn and prenatal birth defects surveillance systems are used to rapidly collect information on all infants with these birth defects and report standard information to the CDC. Data for 2016 were recently published in a January 2018 *Morbidity and Mortality Weekly Report* highlighting the prevalence of birth defects potentially related to Zika virus infection in the United States (<https://www.cdc.gov/mmwr/volumes/67/wr/mm6703a2.htm>).

Substance Exposed Newborns

The number of substance exposed newborns (SEN) has increased in Rhode Island in the last 10 years. Neonatal abstinence syndrome (NAS) is a group of conditions that occur when a newborn exposed to certain types of drugs, including opioids, before birth experiences withdrawal. In Rhode Island, since 2006, the rate of NAS has increased to 91 newborns per 10,000, an increase of almost 150%. Currently, the RIBDP is working with the SEN Task Force's Data Group to explore how existing RIDOH program data can be used to conduct SEN surveillance.

HIV Antiretroviral Drug Safety Concerns and Neural Tube Defects

In May 2018, investigators studying the HIV antiretroviral drug Dolutegravir reported a potential safety issue related to neural tube defects in infants who were born to women taking this drug at the time of conception or early in the first trimester. The CDC, WHO, and other stakeholders are currently conducting more research into the safety and efficacy of Dolutegravir in women of child-bearing age. The RIBDP will continue to monitor the situation and work with RIDOH's Center for HIV, Hepatitis, STDs, and TB Epidemiology as needed.

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

SERVICE ASSESMENT AND ASSURANCE

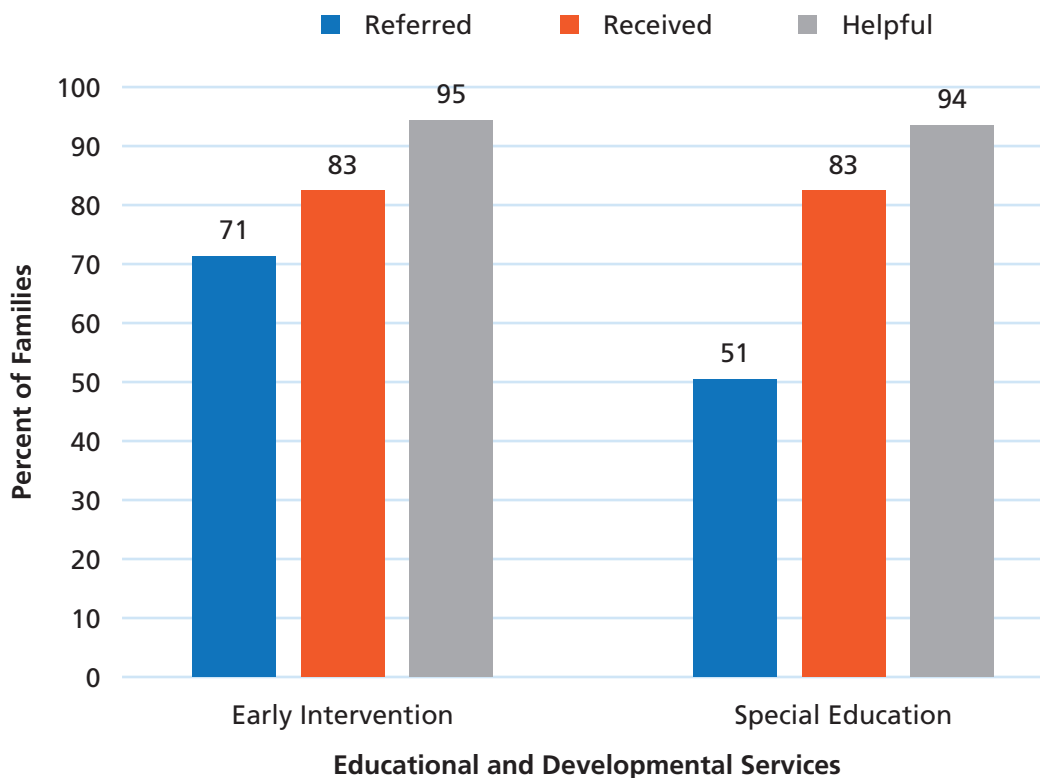
A priority goal of the RIBDP is to assure that children with birth defects receive appropriate and timely preventive, specialty, and other healthcare services. The RIBDP, in collaboration with the Rhode Island Parent Information Network (RIPIN), employs a Parent Consultant who interviews and conducts service assessments with families who have children with specific birth defects to determine whether the children have received appropriate referrals and services on a timely basis. The Parent Consultant meets with families at pediatric and specialty care practices, such as the Children's Neurodevelopment Center (CNDC) at Hasbro Children's Hospital, that service children with birth defects. The Parent Consultant also mails service assessment forms to additional families of children with birth defects that cannot be interviewed in a practice. The service assessment forms are used by families of children, newborn to age five, to determine what services and referrals were provided to the children based on the national guidelines for specific conditions. Specifically, the assessment forms ask about medical tests and procedures, developmental and educational services, and parent supports. Currently, service assessments are conducted with families of children who have Down syndrome, spina bifida, craniofacial defects, critical congenital heart defects (CCHD), abdominal wall defects, hearing loss, and microcephaly or other central nervous system conditions (CNS).

From 2011 through 2017, the RIBDP has received a total of 423 assessment forms completed by families of children with Down syndrome (n= 94), spina bifida (n=54), craniofacial anomalies (n =219), CCHD (n =24), abdominal wall defects (n =12), microcephaly/CNS conditions (n=9), and hearing loss (n=11). RIBDP also collected 356 repeat service assessments. The service assessment forms include a comment section where families have indicated that although they were referred to services, they still face challenges with financial, social, and educational issues associated with raising a child with a birth defect. Families found financial resources to be inadequate, the application process to be cumbersome, and the approval time prolonged. They also stated that they needed help from peer support and advocacy-assistance agencies to navigate health and educational systems. Some families thought the medical service needs of their children were not being met and chose to seek healthcare at facilities in other states. Families noted that physicians, organizations, and community supports helped their child's development, but that they would benefit from additional support through the referral, diagnosis, and treatment process. Many families reported that the Early Intervention (EI) teams provided support in transitioning their children to the school system. Parents reported that more local and active family support groups are needed for children with specific birth defects.

The RIBDP recently evaluated educational and developmental support service referrals reported by families of children with orofacial defects (cleft lip, cleft palate, or both; n=137) and craniosynostosis (n=76) born from 2006 to 2017. Families deemed eligible for developmental and educational services were referred to (EI) and special education (Figure 9). Approximately 71% of all children (n=213) were referred to EI, which provides developmental services to children from birth to age three, and 51% of children age three or older (42 of 83) were referred to special education, which provides educational and social services for children age three and older. Most families who received these services were highly satisfied.

Some infections a mother gets before or during pregnancy can hurt her and her baby.

FIGURE 9: EDUCATIONAL AND DEVELOPMENTAL SUPPORT SERVICES REFERRAL AND RECEIPT BY FAMILIES OF CHILDREN WITH CRANIOFACIAL DEFECTS, RHODE ISLAND, 2006-2017



REDUCING THE RISK OF BIRTH DEFECTS

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 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). Specific recommendations for having a healthy pregnancy and improving birth outcomes are included throughout this data book. To learn more, visit:

www.health.ri.gov/for/pregnantwomen and www.health.ri.gov/for/womenplanningpregnancy.

The RIBDP has conducted local case-control studies to support national birth defects research, which examined associations between risk factors and certain birth defects. In Rhode Island, women who were obese before pregnancy were linked with birth outcomes related to conotruncal heart defects (defects of outflow valve). Also, women who smoked during pregnancy were at risk for birth outcomes resulting in pulmonary stenosis and clubfoot.

The RIBDP continues to work with various public health programs on birth defects awareness and risk-reduction activities. For example, in collaboration with the Family Planning Program, using CDC funding, the RIBDP funds the purchase and distribution of free multivitamins that contain folic acid to uninsured women who receive a negative pregnancy test at a family planning clinic. Uninsured women with positive pregnancy tests are enrolled in the state's Medicaid managed-care program, RIte Care, and receive prenatal vitamins at their first prenatal visit. In 2017, approximately 914 multivitamins were distributed by seven family planning clinics through this program.



It is recommended that women take 400-800 micrograms (mcg) of folic acid every day, starting at least one month before getting pregnant.

INFORMATION FOR EDUCATION AND DECISION MAKING

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, which includes maintaining an up-to-date website (www.health.ri.gov/birthdefects); publishing studies in peer-reviewed journals; presenting information at state, local, and national meetings; and sponsoring grand rounds at Women & Infants and Rhode Island Hospitals.

The RIBDP works with its Advisory Council to plan and coordinate grand rounds at Rhode Island Hospital each January, in recognition of Birth Defects Awareness month. These birth defects grand rounds have been co-sponsored by the RIBDP, Rhode Island Hospital, and the March of Dimes Rhode Island/Southeastern Massachusetts market. The format of these grand rounds includes a keynote speaker and a discussion panel, usually comprised of families of children with birth defects. Community organizations and agencies that serve children with special needs are invited to share their materials before and after the grand rounds. In addition to the extended grand rounds, additional grand rounds and seminars at Rhode Island Hospital and Women & Infants Hospital are dedicated to topics related to birth defects and are held throughout the year. Topics have included spina bifida, fetal alcohol syndrome, Down syndrome, hearing loss, craniofacial anomalies, and gastroschisis.

PARTNERSHIPS

In addition to sponsoring grand rounds and other seminars, RIBDP has participated in a variety of national and international collaborative studies to gain a better understanding of specific birth defects. The National Birth Defects Prevention Network (NBDPN) initiated and coordinated most of these studies. In 2009, Rhode Island joined the other New England states to form the New England Birth Defects Consortium, whose mission is to improve services for infants and children in New England with birth defects by promoting regional collaboration through data sharing, research activities, prevention activities, and healthcare quality improvement.

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 Rhode Island/Southeastern Massachusetts market, and the RIPIN. The Advisory Council provides guidance to the RIBDP in the development and implementation of its surveillance, prevention, service assurance, and information dissemination strategies.

APPENDIX 1: RHODE ISLAND BIRTH DEFECTS PROGRAM CASE DEFINITION

A Rhode Island birth defects case is an unborn fetus (gestational age more than 10 weeks) or a child up to the age of five years diagnosed with a congenital anomaly and a Rhode Island maternal residence during a pregnancy loss, termination of pregnancy, or delivery. A congenital anomaly is defined as any condition diagnosed with an ICD-10 'Q' code.

Case Status

A birth defects case is considered a confirmed case following a verified clinical review of the congenital anomaly diagnosis.

A birth defects case is considered a probable case following an unverified clinical review of the congenital anomaly diagnosis or an unknown verification of the diagnosis after clinical review. Suspected congenital anomalies identified during pregnancy with maternal ICD-10 codes O35.0, O35.1, and O35.8 are considered probable cases until verification of a congenital anomaly is present.

Exclusionary Criteria

The following congenital anomalies are excluded as birth defects cases with no exception:

Tear duct obstruction

Ear pit

Preauricular sinus and cyst

Pulmonic stenosis

Laryngomalacia/tracheomalacia

Ankyloglossia

Pyloric stenosis

Embryonic cyst

Imperforate hymen

Cryptorchidism (diagnosed less than a year after birth)

Retractile testes

Hydronephrosis (except postnatal diagnosis)

Flat foot

Hip dysplasia, hip laxity

Port wine stain/hemangioma

Mongolian spot

Birthmark

Sacral dimple

Accessory nipple

Patent ductus arteriosus with a gestational age of less than 36 weeks that is not coupled with another birth defect, or a case with a gestational age of less than 36 weeks that received prostaglandins.

Reporting

All cases, regardless of confirmed status, are reported for surveillance.

APPENDIX 2: RESOURCES

Rhode Island Resources:

March of Dimes Rhode Island/Southeastern Massachusetts Market

<https://www.marchofdimes.org/index.aspx>

The March of Dimes helps women have full-term pregnancies and researches the problems that threaten the health of babies. This is done through community services, education, and advocacy.

Rhode Island Parent Information Network (RIPIN) and Family Voices of Rhode Island

<http://www.ripin.org>

Provides information, support, and training to help all Rhode Islanders become their own best advocate at school, in healthcare, and in all areas of life.

Down Syndrome Society of Rhode Island

<http://www.dssri.org>

A parent support organization dedicated to promoting the rights, dignity, and potential of all individuals with Down syndrome through advocacy, education, public awareness, and support.

Rhode Island Early Intervention Program (EI)

<http://www.eohhs.ri.gov/Consumer/FamilieswithChildren/EarlyIntervention.aspx>

Promotes the growth and development of infants and toddlers who have a developmental disability or delay in one or more areas; Children referred to EI receive a comprehensive developmental evaluation to determine if they are eligible.

Rhode Island Department of Health's Health Equity Institute

<http://www.health.ri.gov/programs/specialneeds>

Provides appropriate community-based systems of services for children and youth with special healthcare needs and their families.

Genetic Counseling and Medical Genetics Services

Prenatal Diagnostic Center

Women & Infants Hospital

101 Plain St., 6th Floor

Providence, RI 02903

Phone: 401-453-7510

Fax: 401-453-7517

Offers screening, diagnostic, and genetics counseling during pregnancy

Genetics Counseling Center

Hasbro Children's Hospital/Division of Human Genetics

2 Dudley St., Suite 460

Providence, RI 02903

Phone: 401-444-8361

Fax: 401-444-3288

Provides genetics counseling and diagnostic services for children, adults, and families with histories of birth defects or genetic disorders

Children's Neurodevelopment Center

Hasbro Children's Hospital

335 R Prairie Ave., Suite 2B

Providence, RI 02905

Phone: 401-444-5685

Fax: 401-444-6115

<https://www.lifespan.org/centers-services/childrens-neurodevelopment-center>

Provides interdisciplinary, comprehensive care for children with developmental and learning problems

Laboratories

Genetics Laboratory - Division of Genetics

Women & Infants Hospital

70 Elm St., 3rd Floor

Providence, RI 02903

Phone: 401-453-7652

Fax: 401-453-7547

Offers testing for cytogenetics, molecular cytogenetics, and molecular genetics

Prenatal and Special Testing Laboratory:

Women & Infants Hospital

70 Elm St., 2nd Floor

Providence, RI 02903

Phone: 401-453-7650

<http://www.womenandinfants.org/services/medical-screening/index.cfm>

Provides prenatal AFP analysis

National Resources and Associations

American Academy of Pediatrics (AAP)

<http://www.aap.org>.

An organization of 60,000 pediatricians committed to the attainment of optimal physical, mental, and social health and well-being for all infants, children, adolescents, and young adults. Website contains information regarding the Academy's many programs, activities, policy statements, practice guidelines, publications, and other child health resources.

American Academy of Family Practitioners (AAFP)

www.aafp.org

The AAFP is the national association of family doctors. It promotes and maintains high-quality standards for family doctors who are providing continuing comprehensive healthcare to the public. It is one of the largest national medical organizations, with more than 105,900 members in 50 states, Washington, D.C., Puerto Rico, the Virgin Islands, and Guam, as well as internationally.

Birth Defect Research for Children, Inc. (BDRC)

<http://www.birthdefects.org>

BDRC is a non-profit organization that provides parents and expectant parents with information about birth defects and support services for their children.

Centers for Disease Control and Prevention (CDC)

<http://www.cdc.gov>

The mission of the CDC is to develop resources for communities to protect their health. This national agency is made up of Centers that target specific health topics, such as congenital anomalies, to improve health promotion and prevent disease and disability. Links to CDC branches and offices that offer useful information and resources relevant to birth defects:

- National Center on Birth Defects and Developmental Disabilities (NCBDDD): www.cdc.gov/ncbddd
- Maternal and Infant Health: <http://www.cdc.gov/reproductivehealth/MaternalInfantHealth>
- Office of Genetics and Disease Prevention: www.cdc.gov/genomics

National Birth Defects Prevention Network (NBDPN)

<http://www.nbdpn.org/>

A national network of state and population-based programs for birth defects surveillance and research to assess the impact of birth defects upon children, families, and healthcare; to identify factors that can be used to develop primary prevention strategies; and to assist families and their providers in secondary disabilities prevention.

National Organization on Fetal Alcohol Syndrome (NOFAS)

<http://www.nofas.org>

NOFAS is dedicated to eliminating birth defects caused by alcohol consumption during pregnancy and to improving the quality of life for those affected individuals and families.

National Society of Genetic Counselors (NSGC)

<http://www.nsgc.org>

The NSGC works to promote the genetics counseling profession as a recognized and integral part of healthcare delivery, education, research, and public policy.

Organization of Teratology Information Services (OTIS)

<https://mothertobaby.org/>

OTIS is a non-profit organization made up of individual services (TIS) throughout North America. It is dedicated to providing accurate, evidence-based, clinical information to patients and healthcare professionals about exposures during pregnancy and lactation.

Smiles

<http://www.cleft.org>

A group of dedicated families who have developed a first-hand understanding of the needs of children with cleft lip, cleft palate, and craniofacial deformities.

Spina Bifida Association

<http://sbaa.org/>

Promotes the prevention of spina bifida and enhancing the lives of all affected.

Teratology Society

<http://www.teratology.org>

Teratology Society is the premier source for cutting-edge research and authoritative information related to birth defects and other disorders of developmental origin.

Zika Virus Information

<https://www.cdc.gov/zika/pregnancy/protect-yourself.html>

CDC website with resources for education and prevention from Zika infection, as well as how to follow up with your doctor. Additional information on Zika virus and pregnancy, pregnancy outcomes, birth defects and conditions related to Zika can be found at:

<https://www.cdc.gov/zika/pregnancy/index.html>.

International Associations**International Clearinghouse for Birth Defects Surveillance and Research**

<http://www.icbdsr.org>

Dedicated to bringing together birth defect programs from around the world with the aim of conducting worldwide surveillance and research to prevent birth defects and to ameliorate their consequences.



GLOSSARY

Anencephalus	Partial or complete absence of the brain or skull.
Anphthalmia	Lack of one or both eyes.
Anotia	Lack of the external (visible) ear.
Aortic valve stenosis	A heart defect involving the aorta, the main blood vessel carrying blood from the heart to the rest of the body. This condition involves a narrowing of the valve between the left ventricle (lower chamber) of the heart and the aorta. It can be repaired surgically in some cases.
Atrial septal defect	A hole (varies in size) in the wall of the heart between the right and left atrium, or the upper chambers. Also called ostium secundum defect.
Atrioventricular septal defect	A hole or abnormal shape in the connective tissue that divides the right and left chambers of the heart. This can occur between the ventricles (lower chambers) or the atria (upper chambers).
Congenital disorder	A medical condition that is present at birth but may be recognized before birth. Also called a birth defect. The conditions in this glossary are all congenital.
Congenital cataract	A clouding of the capsule or lens of the eye that is present at birth. This might cause vision problems or blindness.
Congenital posterior urethral valves	An abnormal congenital obstructing membrane that is located within the posterior male urethra; this valve is the most common cause of bladder outlet obstruction in male children.
Choanal atresia	A narrowing or blockage of the nasal airway by tissue. This causes difficulty breathing.
Cleft lip with and without cleft palate	When the lip does not completely develop. Sometimes, it extends into the palate (roof of the mouth).
Cleft palate without cleft lip	A partial or complete split in the palate (roof of the mouth) that happens without a split in the lip.
Club foot	Babies born with this condition have their foot turned to the side. It may even appear that the top of the foot is where the bottom should be. The involved foot, calf, and leg are smaller and shorter than the other side.
Coarctation of the aorta	The narrowing of the aorta, the main blood vessel carrying blood from the heart to the rest of the body.
Craniosynostosis	Premature closure of one or several connective tissue membranes that separate the bones of the developing skull.
Diaphragmatic hernia	The absence or a defect of the membrane between the chest cavity and the abdomen. This lets organs, such as the intestines, protrude into the chest. It also interferes with the development of the heart and lungs.
Down syndrome	A disorder caused by the presence of an extra 21 st chromosome. This causes developmental disability, distinctive physical features, and short stature. This condition is also called trisomy 21.
Encephalocele	A gap or hole in the skull that usually causes a sac-like protrusion of the brain and the membranes that cover it.
Esophageal atresia/ tracheoesophageal fistula	A condition in which the esophagus ends in a blind pouch and fails to connect with the stomach. Tracheoesophageal fistula is an abnormal communication between the esophagus and the trachea.
Fetal alcohol syndrome	The sum total of the damage done to the child before birth as a result of the mother drinking alcohol during pregnancy. This condition always involves brain damage, impaired growth, and head and face abnormalities.
Gastroschisis	When an infant's intestines stick out of the body through a defect on one side of the umbilical cord.
Genetic	Having to do with genes, heredity, and variation in living things.
Hirschsprung's disease	A blockage in the large intestine due to a lack of nerves in part of the bowel. This condition causes the bowel and abdomen (belly) to become swollen.
Holoprosencephaly	Structural brain anomaly that results from incomplete cleavage of the prosencephalon.
Hydrocephalus	A buildup of fluid inside the skull that leads to brain swelling.

Hypoplastic left heart syndrome	When the left chambers of the heart do not develop completely. This is one of the most life-threatening heart defects.
Hypospadias and Epispadias	Abnormal development of the tube carrying urine from the bladder to the outside of the body (urethra); the urinary opening is misplaced on the upper surface of the penis or where the urethra opens into the vagina.
Infant	A child up to one year (12 months) of age.
Microcephaly	A condition where the baby's head is much smaller than expected.
Microphthalmia	Smallness of the eye.
Microtia	A small, abnormally shaped external ear. It can occur on one side only (unilateral) or on both sides (bilateral).
Mortality rate	Number of deaths in a year in a given population.
Obstructive genitourinary defect	A narrowing or absence of a normal opening in the urinary tract that blocks the flow of urine at any place in the urinary tract from the kidney to the urethra.
Omphalocele	When an infant's intestines or other organs stick out of their abdominal cavity and are covered by a transparent sac.
Pulmonary valve atresia/stenosis	Abnormal closure or absence (atresia) or narrowing (stenosis) of the duct that opens into the pulmonary artery, the vessel that carries blood to the lungs.
Rectal and large intestinal atresia/stenosis	Abnormal closure, absence, or narrowing of the duct or passageway of the digestive tract in the rectum or large intestine.
Reduction deformity, or lower limbs	Deformity of the arms or legs in which one or both arms or legs upper are missing or shortened.
Renal agenesis/hypoplasia	A defect where the kidney was formed incompletely (hypoplasia) or is absent (agenesis).
Spina bifida	A defect in which the spinal neural tube is imperfectly closed. This can cause part of the spinal cord to stick out of the back. This condition often results in neurological (brain, spinal cord, and nerve) disorders.
Transposition of great arteries	A defect in which the main blood vessels leading from the heart (the aorta and the pulmonary artery) are reversed. This means there is less oxygen in the blood that is pumped from the heart to the rest of the body.
Tricuspid valve atresia	Absence or closure of one of the valves between two of the heart's chambers. This causes blood in the right ventricle (lower chamber) to flow backward into the right atrium (upper chamber), instead of flowing into the lungs to pick up oxygen.
Trisomy 13 (Patau)	When an infant has three copies of chromosome 13, it causes severe skull and facial deformation and developmental delays. Some of these include heart defects, brain defects, and cleft lip and/or palate.
Trisomy 18 (Edwards)	When an infant has three copies of chromosome 18, it can cause potentially life-threatening developmental and medical complications in the early months and years of life.
Tetralogy of Fallot	A heart defect that causes low oxygen levels in the blood. It typically includes four defects: a hole in the wall between the right and left ventricles (lower chambers of the heart), a misplaced aorta (the artery that carries oxygen-rich blood to the body), a narrowing of the pulmonary artery that carries blood from the heart to the lungs, and an enlarged right ventricle.
Ventricular septal defect	One or more holes in the wall between the ventricles, or lower chambers of the heart. This allows blood with oxygen to mix with blood that does not contain oxygen.

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