



**RHODE ISLAND
BIRTH DEFECTS DATA BOOK
2014**



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 to birth defects include genetic (inherited) factors, environmental pollutants, occupational hazards, dietary factors, 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, 1 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 survive beyond the first year experience childhood illness and disability.

Birth defects cause about one in five infant deaths in the United States and Rhode Island. However, of the 64 infants who died in Rhode Island during 2012, only 4 of the deaths (6.3%) were attributed to a birth defect. This represents a 59% decrease in the proportion of infant deaths resulting from a birth defect since 2008, when 11 (16.1%) of the 68 infant deaths resulted from a birth defect. The low count in 2012 may be an aberration, since provisional 2013 data indicate there were 14 (16.2%) infant deaths related to a birth defect, similar to previous years.

Many preterm infants (born before 37 weeks gestation) have birth defects. In Rhode Island, 90 (8.9%) of the 1,012 preterm babies born in 2012 had a birth defect. Preterm birth is the leading cause of infant death in Rhode Island. In 2012, 27 (42.2%) of the 64 infant deaths were attributed to prematurity.

Birth defects have serious economic costs

In addition to the emotional impact that families of children with birth defects 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 (HEALTH) studies the costs of selected birth defects using national surveillance guidelines based on the severity and frequency of the birth defect. Hospital discharge data provide unadjusted crude total and specific hospital costs for newborn admissions. The Rhode Island analysis shows the total unadjusted cost for all newborn hospital admissions with at least one diagnosed birth defect and the total unadjusted cost by selected birth defects.



During 2010-2012, Rhode Island's hospital discharge database identified 1,390 newborns with at least one birth defect. The total hospital cost for these newborns was approximately \$108 million, with an average cost per newborn of \$77,990. This amounts to more than six times the cost for a newborn with no birth defects (\$12,480). The average length of stay for a newborn with birth defects (10.6 days) was nearly three times higher than that for a newborn without a birth defect (3.8 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 (6.1 days) was twice as high as the average length of stay for children without birth defects (2.9 days). The average cost per day for children with birth defects was \$42,837 compared to \$18,406 per day for children without 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) between Rhode Island and the United States for selected birth defects.

TABLE 1: MEAN HOSPITAL COSTS FOR NEWBORNS BY SELECTED BIRTH DEFECTS, RHODE ISLAND AND UNITED STATES

Birth Defect	RHODE ISLAND: 2010-2012			UNITED STATES: 2003		
	Cases	Length of Stay (Days)	Mean Total (\$)	Cases	Length of Stay (Days)	Mean Total (\$)
Spina bifida	3	14	122,800	1,136	15	65,942
Microcephaly	14	4	14,019	NA	NA	NA
Hydrocephaly	3	3	17,199	NA	NA	NA
Cleft lip w/ and w/o cleft palate	18	3	8,263	3,486	6	15,397
Cleft palate	6	10	32,837	2,187	10	33,387
Rectal and large intestinal atresia/stenosis	3	23	76,124	1,604	17	75,220
Esophageal atresia/tracheoesophageal fistula	3	11	108,529	NA	NA	NA
Gastroschisis	10	42	379,843	NA	41	155,620
Down syndrome	23	5	21,310	5,036	11	38,745

Notes: Costs adjusted using hospital cost-to-charge ratios. Mean total cost refers to the average cost per case across all maternity hospitals. NA = not available.

Sources: Rhode Island Hospital Discharge Database, Rhode Island Department of Health; Centers for Disease Control and Prevention. "Hospital stays, hospital charges, and in-hospital deaths among infants with selected birth defects"; United States, 2003. MMWR, 2007;56(2):25-9.



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 began developing a birth defects information system in 2000, with funding from the Centers for Disease Control and Prevention (CDC). The Rhode Island Birth Defects Program (RIBDP) is located at the Rhode Island Department of Health (HEALTH), 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 kept confidential and is protected under state and federal privacy laws.

During 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 Health appointed the Rhode Island Birth Defects Advisory Council to advise HEALTH on the establishment and implementation of the system and 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 HEALTH develop the surveillance system and analyze and disseminate information. These groups are represented on the Advisory Council. The RIBDP also solicits input directly via surveys, focus groups, and interviews.



CASE IDENTIFICATION AND DATA

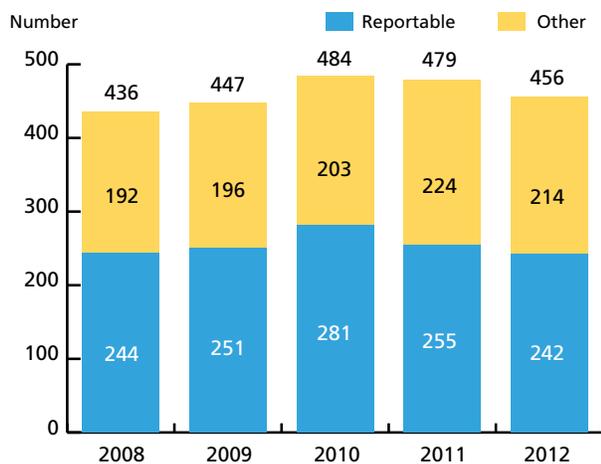
The RIBDP uses hospital discharge data (HDD) as the primary source for capturing birth defects data in Rhode Island. The RIBDP works with all six maternity hospitals to collect discharge information. The RIBDP also collects information from specialty clinics, such as the Children’s Neurodevelopment Center (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 limited to children born to Rhode Island residents, who are aged birth to five. They are identified using diagnoses coded by the 9th Clinical Modification of the International Classification of Diseases (ICD 9-CM) and include 740-759.9 and 760.71 codes. The RIBDP confirms the accuracy of birth defects diagnoses through chart review of birth defects cases. The RIBDP has identified exclusion criteria to omit certain minor congenital anomalies and focus on more relevant conditions for data analysis and service assurance. The RIBDP has updated its birth defects case definition to exclude certain minor congenital anomalies and to reflect birth defects surveillance guidelines developed by the National Birth Defects Prevention Network (see Appendix 1). Previous data have been adjusted to fit this current case definition for comparable data analyses.

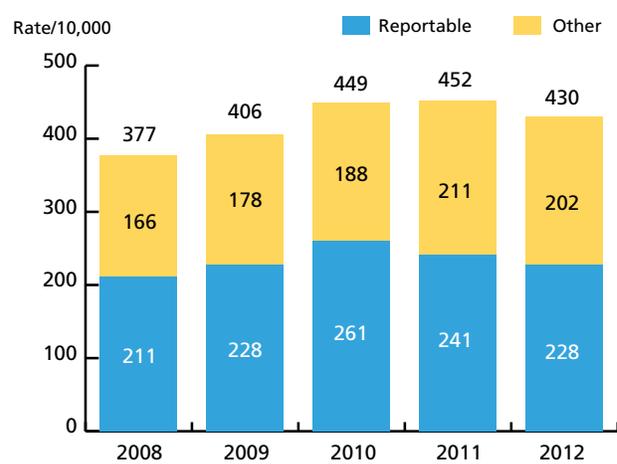
Identification of Cases During the Newborn Period

Figures 1 and 2 show the overall count and prevalence of birth defects in Rhode Island over the five-year period from 2008 through 2012. The totals are grouped by all birth defects and those reported to the CDC and National Birth Defects Prevention Network (NBDPN). During this period, the rate of birth defects in Rhode Island increased by 14.2% from 376.4 per 10,000 live births in 2008 to 429.8 per 10,000 in 2012, after adjusting for the updated birth defects case definition. There was also an increase in the prevalence of cases with the 46 conditions that are reported to the NBDPN. In 2008, the prevalence of these cases was 210.6 per

**FIGURE 1. BIRTH DEFECTS CASES
RHODE ISLAND, 2008-2012**



**FIGURE 2. BIRTH DEFECTS PREVALENCE
RHODE ISLAND, 2008-2012**



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

10,000 live births (n = 244), and by 2012, the prevalence rose slightly to 228.1 (n = 242), representing an 8.3% increase. The prevalence rate for all birth defects peaked during 2010 (448.8) and 2011 (451.8).

Table 2 shows the number and prevalence of selected birth defects among Rhode Island residents during 2008-2012, organized by organ system. Genitourinary defects are the most common type of defects (155.4 per 10,000). Hypospadias and epispadias represent the highest proportion of these cases.

Other common birth defects in Rhode Island include those related to cardiovascular (148.7 per 10,000) and musculoskeletal (135.9 per 10,000) systems. Among cardiovascular defects (those related to the heart), ventricular septal defects (42.5) and patent ductus arteriosus (39.0) are most frequent, but rates for these conditions have remained stable.

TABLE 2. SELECTED BIRTH DEFECTS, RHODE ISLAND, 2008-2012

BIRTH DEFECT	NUMBER	RATE (PER 10,000 LIVE BIRTHS)	BIRTH DEFECT	NUMBER	RATE (PER 10,000 LIVE BIRTHS)
Central Nervous System	112	20.4	Orofacial	63	11.5
Hydrocephaly	33	6.0	Cleft lip with and without cleft palate	42	7.7
Microcephaly	33	6.0	Cleft palate	25	4.6
Spina Bifida	24	4.4	Gastrointestinal	139	25.3
Anencephaly	6	1.1	Rectal and large intestinal atresia/stenosis	17	3.1
Encephalocele	4	0.7	Esophageal atresia/ tracheoesophageal fistula	12	2.2
Eye/Ear/Face/Neck	95	17.3	Hirschsprung's disease	9	1.6
Congenital cataract	6	1.1	Biliary atresia	4	0.7
Anophthalmos/ Microphthalmos	5	0.9	Genitourinary	853	155.4
Anotia / Microtia	3	0.5	Hypospadias and epispadias	235	42.8
Cardiovascular	816	148.7	Obstructive genitourinary defect	188	34.3
Ventricular septal defect	233	42.5	Renal agenesis / hypoplasia	2	0.4
Patent ductus arteriosus	214	39.0	Bladder extrophy	1	0.2
Atrial septal defect	166	30.2	Musculoskeletal	746	135.9
Pulmonary valve atresia and stenosis	32	5.8	Clubfoot	87	15.9
Tetralogy of Fallot	17	3.1	Gastroschisis	24	4.4
Transportation of great vessels	15	2.7	Omphalocele	15	2.7
Hypoplastic left heart syndrome	13	2.4	Reduction deformity, lower limbs	13	2.4
Coarctation of aorta	10	1.8	Diaphragmatic hernia	12	2.2
Atrioventricular septal defect	9	1.6	Reduction deformity, upper limbs	10	1.8
Aortic valve stenosis	7	1.3	Chromosomal	110	20.0
Ebstein's anomaly	2	0.4	Down syndrome	89	16.2
Tricuspid valve atresia	2	0.4	Edward syndrome	16	2.9
Respiratory	28	5.1	Patau syndrome	9	1.6
Choanal atresia	1	0.2	All birth defects	3,053	556.3
			All birth defect cases	2,291	417.4

*Notes: 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 from 2008-2012. The bolded "All birth defects cases" row represents the total number of Rhode Island babies born between 2008 and 2012 with at least one diagnosed birth defect. Source: Rhode Island Birth Defects Program, Rhode Island Department of Health

Reportable Birth Defects

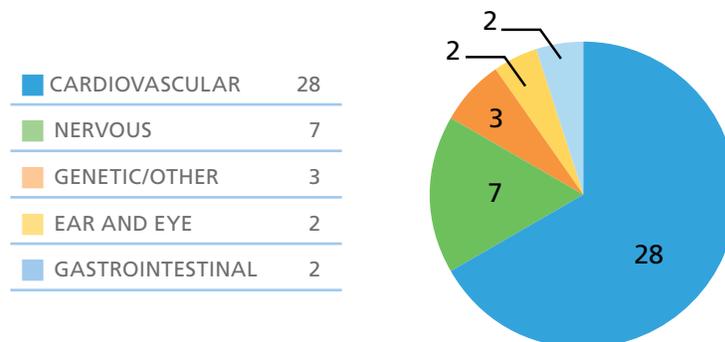
In 2005, regulations were enacted mandating all healthcare providers to report cases of birth defects identified among children up to age five to HEALTH. The reporting of birth defects cases helps the RIBDP assure that these children receive appropriate services and referrals on a timely basis, and helps identify children who were not diagnosed with a birth defect at the time of birth. In 2011, the RIBDP worked with KIDSNET, HEALTH’s integrated child information system, to build a reporting component that would allow pediatric providers to report birth defects cases electronically. Since then, RIBDP staff and KIDSNET provider liaisons have been training pediatric providers and their office staff to report birth defects using the web-based reporting system in KIDSNET.

Figure 3 shows data on 32 cases of birth defects reported by providers through KIDSNET, during 2012-2014. Among these cases, 19 (59%) had not previously been identified by the RIBDP. The body system with the most reported defects was the cardiovascular system (those relating to the heart). Septal heart defects (n = 7) were the most common cardiovascular defects reported by providers.

In addition to these cases, healthcare providers (audiologists) reported 28 cases of deafness or general hearing loss through KIDSNET since September 2011. These data demonstrate the value of the birth defects case reporting process via KIDSNET, as many children are diagnosed with birth defects after birth and these data were not previously available to the RIBDP.



**FIGURE 3. PROVIDER-REPORTED BIRTH DEFECTS BY BODY SYSTEM
RHODE ISLAND, 2012-2014**



Notes: These data represent 32 children born during 2004 – 2014, some of whom may have more than one birth defect.
Source: Rhode Island Birth Defects Program, Rhode Island Department of Health

Identification of Cases during the Prenatal Period

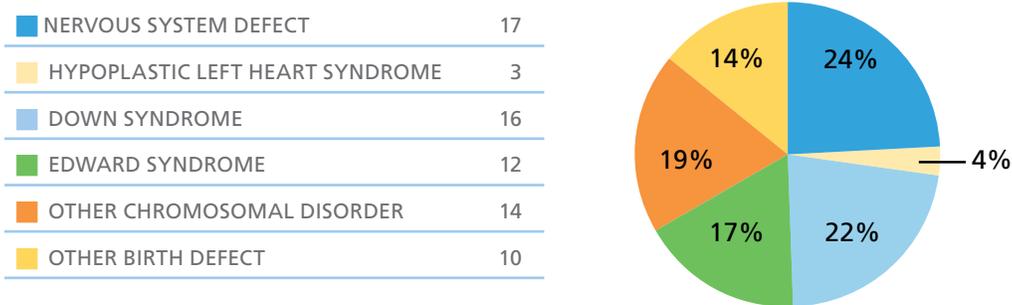
In 2008, the RIBDP began collecting birth defects cases identified during the prenatal period from collaborating laboratories and prenatal clinics such as the Cytogenetics Testing Laboratory and Prenatal and Special Testing Laboratory at Women and Infants Hospital of Rhode Island and the Fetal Treatment Program based at Hasbro Children’s Hospital. About four to six percent of birth defects are identified



prenatally. Prenatal case finding improves the prevalence estimate of certain birth defects by detecting cases not found at newborn discharge.

Figure 4 shows the percentages of birth defects diagnoses ascertained in the prenatal stages during 2009-2012 in Rhode Island. Nearly one quarter (n = 16) of all prenatally ascertained cases were diagnosed with Down syndrome. Prenatally ascertained case data are included in the Rhode Island birth defects prevalence. The RIBDP plans to establish routine prenatal surveillance with our prenatal data partners to better understand birth defects prevalence and trends in Rhode Island.

FIGURE 4. PRENATALLY ASCERTAINED BIRTH DEFECTS IN RHODE ISLAND (N=72), 2009-2012



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

Critical Congenital Heart Defects

Critical congenital heart defects (CCHD) are a range of seven heart defects that can cause serious, life-threatening symptoms. These defects include hypoplastic left heart syndrome, pulmonary atresia (with intact septum), tetralogy of Fallot, total anomalous pulmonary venous return, complete transposition of great arteries, tricuspid atresia, and truncus arteriosus. CCHD may require intervention and most 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 on in life.

In 2010, the US Health and Human Services (HHS) Secretary's Advisory Committee on Heritable Disorders in Newborns and Children (SACHDNC) recommended adding CCHD to the uniform newborn screening panel. The goal is to identify CCHD among newborns before symptoms appear by conducting pulse oximetry screening, which measures oxygenation in the blood. A failed screen resulted by low blood oxygenation saturation (< 90%) is likely to indicate the presence of CCHD. By identifying CCHD among newborns early, the appropriate special care and treatment can be provided.

The RIBDP is collaborating with the Rhode Island Newborn Screening Program and participating maternity hospitals in a grant-funded pilot study to evaluate the use of pulse oximetry screening to detect critical congenital heart defects in Rhode Island. Current data from the pilot study (as of September 2014) show that over 16,000 newborns were screened, resulting in a 99.6% screening rate. The high screening rate is primarily due to CCHD being diagnosed during the prenatal period and by physical examination at birth. Staff members are also participating on the Rhode Island Newborn Screening Advisory Committee's CCHD Subcommittee to discuss the study's methods, provider education, and other related topics.

The counts and prevalence rates of the seven birth defects associated with CCHD are listed in Table 3 and represent 2008-2012 data.

**TABLE 3. PREVALENCE OF BIRTH DEFECTS ASSOCIATED WITH CRITICAL CONGENITAL HEART DEFECTS
RHODE ISLAND, 2008-2012**

CONDITION	COUNT	RATE/10,000
Hypoplastic left heart syndrome	9	1.6
Tetralogy of Fallot	16	2.9
Total anomalous pulmonary venous return	2	0.4
Complete transposition of great arteries (d-TGA)	13	2.4
Tricuspid valve atresia	2	0.4
Common truncus	2	0.4
Pulmonary valve atresia (with intact septum)	9	1.6

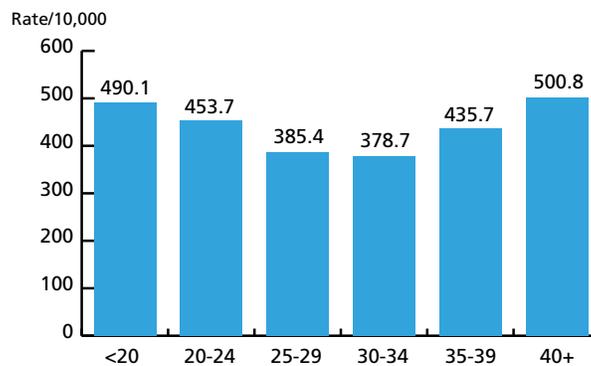
Notes: Numbers and rates in this table may be a subset of those conditions listed in Table 2 and may not match counts in Table 2.
Source: Rhode Island Birth Defects Program, Rhode Island Department of Health

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

Maternal Characteristics

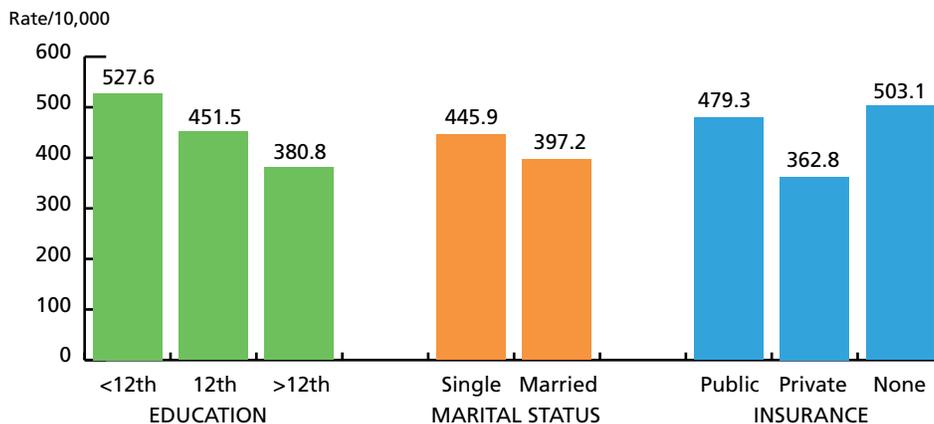
Babies born to women age 40 or older, women with less than a 12th grade education, single women, and women with publicly funded health insurance or no health insurance are at a higher risk for birth defects (Figures 5 and 6). During 2008-2012, the birth defects prevalence rate among women aged 40 or older was 501 per 10,000 live births compared to 385 among women aged 25-29. The birth defects rate among women with less than a 12th grade education (528 per 10,000 live births) or with a 12th grade education (452) was higher than the rate among women with more education (381). Similarly, single women were more likely to have a baby with a birth defect (446) than married women (397). Women insured through public programs such as RIte Care and Medicaid (479) were also more likely to have a baby born with a birth defect than women insured through commercial or private providers such as Blue Cross or United Healthcare (363).

**FIGURE 5. BIRTH DEFECTS PREVALENCE BY MATERNAL AGE GROUPS
RHODE ISLAND, 2008-2012**



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

**FIGURE 6. BIRTH DEFECTS PREVALENCE BY SELECTED MATERNAL CHARACTERISTICS
RHODE ISLAND, 2008-2012**



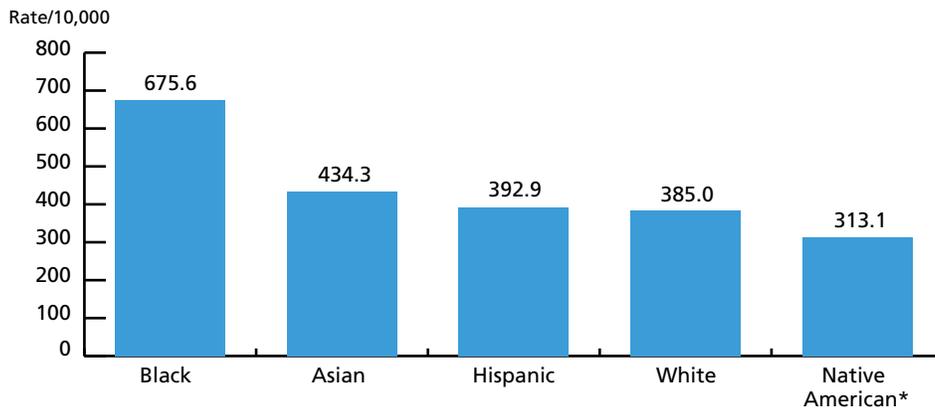
Source: Rhode Island Birth Defects Program, Rhode Island Department of Health

Racial/Ethnic and Geographic Disparities

Birth defects prevalence varies by race/ethnicity (Figure 7) and geographical residence (Figure 8). During 2008-2012, the average birth defects prevalence rate among Black/African Americans (676 per 10,000 live births) was 1.8 times the rate for Whites (385). Asians had the second highest rate (434), which was 13% higher than the rate for Whites. The birth defects prevalence rate among those of Hispanic ethnicity (393) was close to the rate for Whites. These trends were also the same for the subset of 46 conditions that are reported to the National Birth Defects Prevention Network.

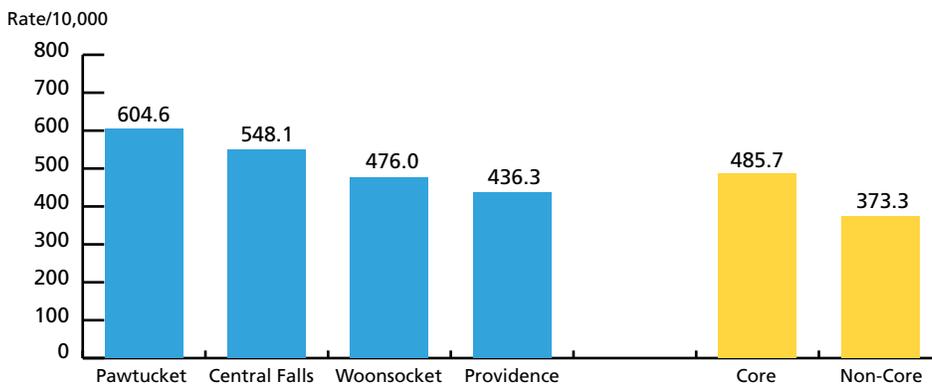
Babies born to residents of core cities where the poverty level is greater than 15% (Central Falls, Pawtucket, Providence, and Woonsocket) were about 1.3 times more likely to have a birth defect than those born to residents living in the rest of the state (Figure 8). Pawtucket (605) and Central Falls (548) 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, and poverty compared to the rest of the state.

**FIGURE 7. BIRTH DEFECTS PREVALENCE BY RACE/ETHNICITY
RHODE ISLAND, 2008-2012**



*Notes: Data for Native Americans reflect small numbers and may not be reliable.
Source: Rhode Island Birth Defects Program, Rhode Island Department of Health

**FIGURE 8. BIRTH DEFECTS PREVALENCE BY SELECTED GEOGRAPHIC AREAS
RHODE ISLAND, 2008-2012**



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



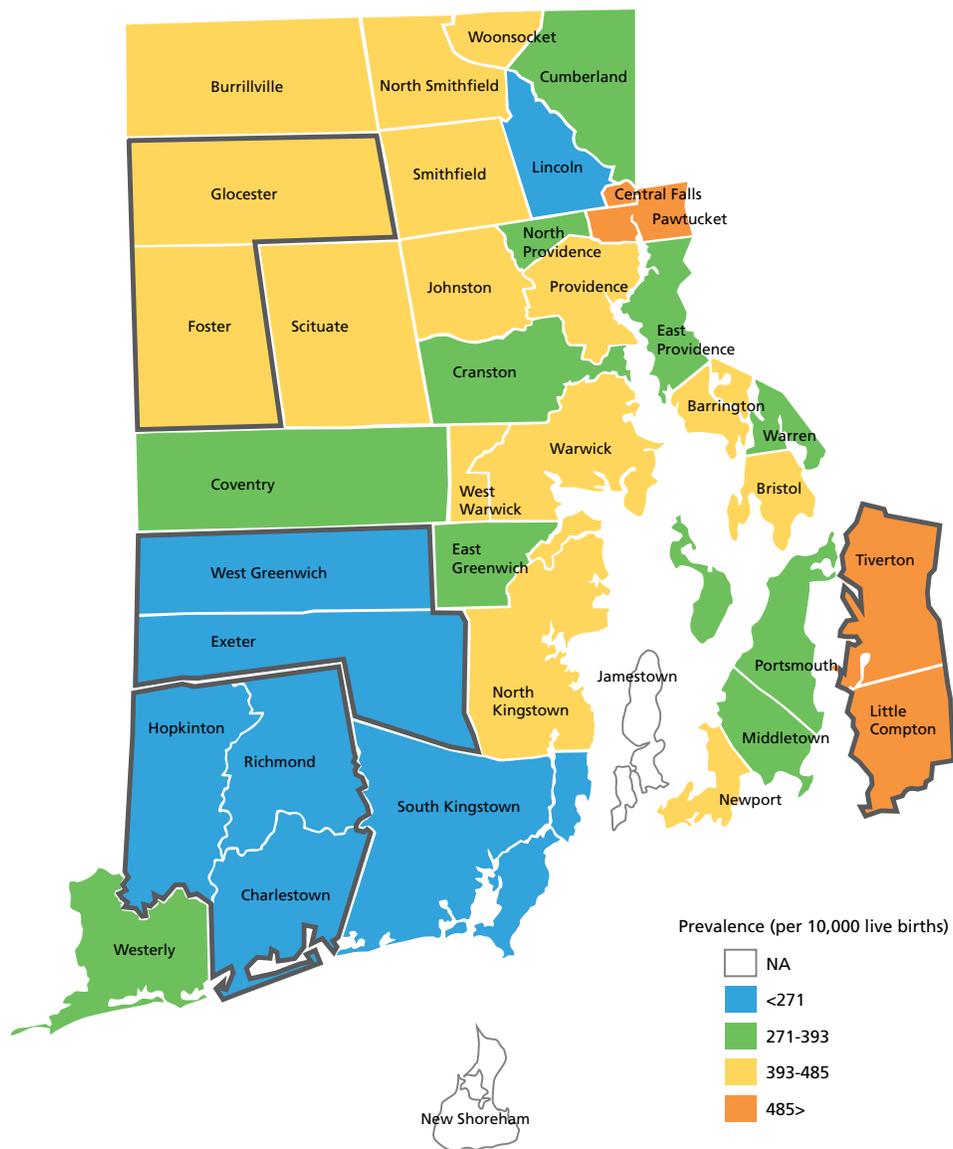
Smoking during pregnancy increases the chances of premature birth, certain birth defects, and infant death.

Mapping Rates of Birth Defects

Geospatial analysis allows us to follow trends and detect clusters of birth defects in Rhode Island. The map on the following page illustrates birth defects prevalence rates during 2008-2012 by city/town 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 2008-2012 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.

The map (Figure 9) shows a majority of cities/towns with a prevalence rate between 393 and 485 birth defects per 10,000 live births, a range of prevalence that is common for birth defects rates. As mentioned previously, Central Falls and Pawtucket form an urban area of high birth defects prevalence in Rhode Island. The region of Little Compton and Tiverton had a notably high prevalence (579 per 10,000 live births). This may be due to more mothers in this region seeking high risk prenatal and delivery care at Women and Infants Hospital, the regional perinatal center. Women at lower risk for poor birth outcomes are more likely to deliver at nearby hospitals over the border in Southeastern Massachusetts, thereby lowering the denominator of Rhode Island resident births.

FIGURE 9. BIRTH DEFECTS PREVALENCE BY CITIES, TOWNS, AND REGIONS RHODE ISLAND, 2008-2012



*Notes: NA= Not available: Town birth defect prevalence not reported due to small numbers. Note: Due to small live birth numbers, the towns of Foster-Glocester, Charlestown-Richmond-Hopkinton, Coventry-West Greenwich, and Tiverton-Little Compton were combined to report aggregated prevalence rates.
 Source: Rhode Island Birth Defects Program and Rhode Island Geographic Information System, Rhode Island

SERVICE ASSESSMENT AND ASSURANCE

A priority goal of the RIBDP is to assure that children with birth defects receive appropriate and timely preventative, specialty, and other healthcare services. The RIBDP, in collaboration with the Rhode Island Parent Information Network (RIPIN), employs a Family Resource Specialist (parent consultant) who interviews and conducts service assessments with families who have children with specific birth defects to determine whether their children have received appropriate referrals and services on a timely basis. The Family Resource Specialist meets with families at pediatric and specialty care practices that service children with birth defects such as the Children's Neurodevelopment Center (CNDC) at Hasbro Children's Hospital. The RIBDP also works with the Family Resource Specialist to mail service assessment survey forms to additional families of children with birth defects. The service assessment surveys are conducted with families of children, newborn to five years of age, 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 services such as medical tests and procedures, developmental and educational services, and parent supports. Currently, service assessments are conducted with families of children with Down syndrome, spina bifida, club foot, craniofacial defects and critical congenital heart defects (CCHD). The RIBDP plans to expand this service assurance process in 2015 to include abdominal wall defects.

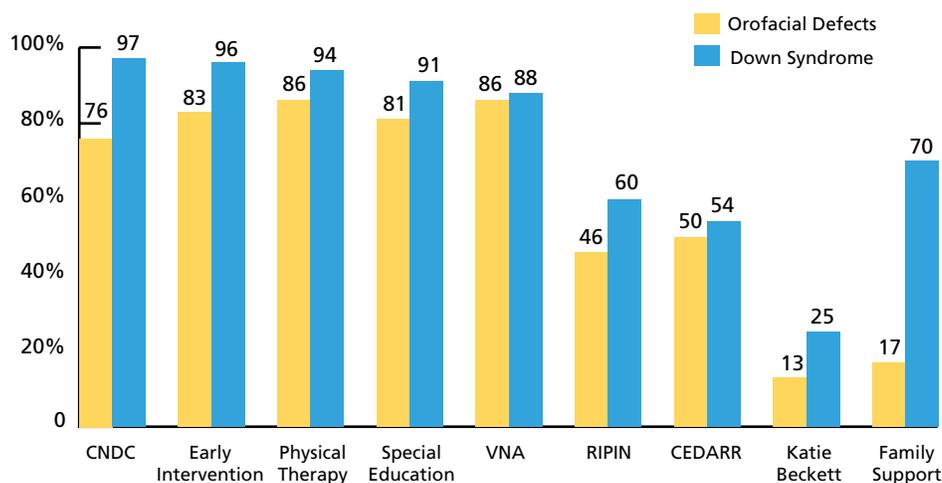


Figure 10 shows the percentage of service assessment forms completed by families during February 2011 through November 2014. The data summarize the percentage of families of children with Down syndrome (n=80) and orofacial defects (n=152) who received referrals to appropriate medical and social services. The Early Intervention Program (EI), which provides developmental services to children aged birth to three, had the highest percentage of referrals for children with Down syndrome (96%), while the referral percentage for children with orofacial defects was 83%. These differences reflect the variation in needs children with these conditions may require. The percentage of referrals to special education was higher among children with Down syndrome (91%) and orofacial defects (81%) during 2011-2014 than the percentage of referrals for these children reported in the 2012 birth defects data book (55% for children with Down syndrome and 16% for children with orofacial defects during 2011-2012). These higher percentages reflect the increased number of repeat assessments (n = 135) conducted from 2012-2014; some families who were re-surveyed had children who aged out of EI at age 3 and into the Special Education system, which serves children age 3 and older. There were also differences in referral percentages for financial support

services. Families of children with Down syndrome were nearly twice as likely to receive referrals to the Katie Beckett Program (25%) compared to families of children with orofacial defects (13%). The Katie Beckett Program is a Rhode Island medical assistance coverage group that allows very disabled and/or medically complex children to be cared for at home instead of at a hospital or institution. Similarly, 70% of families of children with Down syndrome received referrals to family support services (Rhode Island Parent Information Network) compared to only 17% among families of children with orofacial defects.

As of November 2014, the RIBDP received 343 assessment forms completed during 2011-2014 by families of children with the five conditions: Down syndrome (n = 92), spina bifida (n = 37), club foot (n = 48), craniofacial defects (n = 153), and CCHD (n = 13). The service assessment forms include a “comments” 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 reported that services previously provided or recommended by their child’s treatment team were not provided by their school districts. Additionally, families found financial options to be inadequate and said that they needed help to navigate health and educational systems. Many families reported that the Early Intervention teams provided support in transitioning their children to the school system. 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.

FIGURE 10. PERCENTAGE OF FAMILIES OF CHILDREN WITH DOWN SYNDROME AND OROFACIAL DEFECTS WHO RECEIVED SERVICE REFERRALS, RHODE ISLAND, 2011-2014



Notes: Orofacial defects include cleft lip and/or palate.
 Source: Rhode Island Birth Defects Program, Rhode Island Department of Health

Healthy eating and exercise during pregnancy can benefit both the mother and baby with proper nutrition to maintain a healthy weight.

REDUCING THE RISK OF BIRTH DEFECTS

Although not all causes of birth defects are known, there are a number of steps a woman can take 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 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 (e.g., diabetes, epilepsy, and high blood pressure). Specific tips on 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 initiated local case-control studies to support national birth defects research, which found associations between risk factors and certain birth defects. In Rhode Island, pre-pregnant obese women were linked with birth outcomes related to conotruncal heart defects. Also, women who smoked during pregnancy were at risk for birth outcomes resulting in pulmonary stenosis and clubfoot. The RIBDP is working on a study that examines the relationship between diabetes (Type II and gestational) and birth defects.

The RIBDP is currently working with various public health programs on birth defects awareness and risk reduction activities. For example, in collaboration with the Rhode Island Department of Health's Family Planning Program, the RIBDP has financed the purchase and distribution of free multivitamins with folic acid to uninsured women who receive a negative pregnancy test at family planning clinics. 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. Funds awarded by the Centers for Disease Control and Prevention (CDC) have financed this effort.

It is recommended that women take 400 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. The RIBDP posts information online (www.health.ri.gov/birthdefects), publishes studies in peer-reviewed journals, presents information at state, local, and national meetings, and sponsors pediatric grand rounds.

The RIBDP works with its Advisory Council to plan and coordinate pediatric 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 Rhode Island Chapter of the March of Dimes. The format of these grand rounds includes a keynote speaker and a discussion panel usually made up 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, three additional grand rounds are dedicated to topics related to birth defects and are held throughout January. Past topics presented at these grand rounds have included spina bifida, fetal alcohol syndrome, Down syndrome, hearing loss, craniofacial defects, and gastroschisis.



PARTNERSHIPS

In addition to sponsoring pediatric grand rounds, the RIBDP has participated in or is currently participating 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 is also fortunate to work in partnership with its Advisory Council, which includes representatives from Women and Infants Hospital of Rhode Island, Hasbro Children's Hospital, the Rhode Island Chapter of the March of Dimes, the Rhode Island Parent Information Network, and Rhode Island KIDS COUNT.

APPENDIX 1: RHODE ISLAND BIRTH DEFECTS CASE DEFINITION

RHODE ISLAND BIRTH DEFECTS PROGRAM CASE DEFINITION

A Rhode Island birth defects case is a live birth to a Rhode Island resident and includes children up to the age of five, diagnosed with one or more of the ICD-9 codes listed below:

146, 147, 155, 181, 183, 186, 189, 190, 191, 194, 195, 299, 369, 389, 740-759.9, 760.71.

Additionally, prenatally ascertained outcomes (terminations and fetal deaths greater than 20 weeks of gestation) are included.

The following conditions are excluded:

- A live birth in Rhode Island with an out-of-state maternal residence (set aside for New England Birth Defects Consortium)
- Patent ductus arteriosus (747.0) with a gestational age less than 36 weeks that is not coupled with another birth defect, or a case with a gestational age less than 36 weeks that received prostaglandins
- Cryptochidism less than 1 year old that is not coupled with another birth defect
- Other conditions below:

Condition	ICD-9 code
Lipoma	214
Skin neoplasms	216
Hemangioma	228.01
Dacryostenosis (tear duct obstruction)	743.65
Ear pit	744.1
(Peripheral) pulmonic stenosis	747.31
Laryngomalacia/tracheomalacia	748.3
Ankyloglossia	750.0
Pyloric stenosis	750.5
Embryonic cyst	752.41
Imperforate hymen	752.42
Hydronephrosis (except postnatal diagnosis)	753.29
Flat foot	754.61
Hip dysplasia, hip laxity	755.63
Anomalies of skull, face, and bones (except craniosynostosis)	756.0
Port wine stain/hemangioma	757.32
Mongolian spot	757.33
Birthmark	757.39
Accessory nipple	757.6

APPENDIX 2: RESOURCES

RHODE ISLAND:

March of Dimes Rhode Island:

<http://www.marchofdimes.org/rhodeisland>

Helps moms have full-term pregnancies and research 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>

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 Healthy Mothers Healthy Babies Coalition:

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

A partnership of individuals and professional, voluntary, and government organizations devoted to improving the well-being of mothers and babies in Rhode Island through education and advocacy.

Genetic Counseling & Medical Genetics Services

Prenatal Diagnostic Center

Women & Infants Hospital

Bay Tower Medical Center

101 Plain Street, 6th Floor

Providence, RI 02903

Phone: 401-453-7510

Fax: 401-453-7517

Offers screening, diagnostic and genetic counseling during pregnancy

Genetic Counseling Center

Rhode Island Hospital

593 Eddy Street

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.

Greystone / Radiologic Associates

235 Plain Street
Providence, RI 02905
Phone: 401-272-8510
Fax: 401-272-0315

Provides genetic counseling, prenatal testing and diagnosis, and pre-conception evaluations.

Children's Neurodevelopment Center

Hasbro Children's Hospital
335R Prairie Avenue
Providence, RI 02905
Phone: 401-444-5685
Fax: 401-444-6115

<http://www.hasbrochildrenshospital.org/childrens-neurodevelopment>

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

Laboratories**Genetics Laboratory - Division of Genetics**

Women & Infants Hospital
70 Elm Street, 3rd Floor
Providence, RI 02903
Phone: 401-453-7652
Fax: 401-453-7547.

Offers testing for cytogenetics, molecular cytogenetics, and molecular genetics.

Prenatal & Special Testing Laboratory: <http://www.womenandinfants.org>

Women & Infants Hospital
70 Elm Street, 2nd Floor
Providence, RI 02903
Phone: 888-AFP-MOMS
Provides prenatal AFP analysis

NATIONAL:**American Academy of Family Physicians (AAFP)**

www.aafp.org

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, D.C., Puerto Rico, the Virgin Islands, and Guam, and internationally.

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 Heart Association: <http://www.heart.org>

Provides information and resources on congenital heart defects.

Birth Defect Research for Children, Inc.: <http://www.birthdefects.org>

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

Mother to Baby: <http://www.mothertobaby.org>

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

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 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 birth defects, to improve health promotion and prevent disease and disability. Below are 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 Organization on Fetal Alcohol Syndrome: <http://www.nofas.org>

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

National Society of Genetic Counselors: <http://www.nsgc.org>

Works to promote the genetic counseling profession as a recognized and integral part of healthcare delivery, education, research, and public policy.

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://www.sbaa.org>

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

Teratology Society: <http://www.teratology.org>

Provides research, authoritative information, education, and training related to birth defects and other disorders of developmental origin.

INTERNATIONAL:

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.
Anophthalmia	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.
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 those on 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.
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 21st 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.

Epispadias	The displacement of the opening of the urethra on top of and further away from the tip of the penis.
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.
Hydrocephalus	A buildup of fluid inside the skull that lead 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	The displacement of the opening of the urethra underneath and farther away from the tip of the penis.
Infant	A child up to 1 year (12 months) of age.
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 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. This causes severe skull and facial deformation and developmental delays. Some of these include heart defects, brain defects, and cleft lip palate.
Trisomy 18 (Edwards)	When an infant has three copies of chromosome 18. This 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.

Acknowledgment:

We thank the members of the Rhode Island Birth Defects Advisory Council for their ongoing support and guidance.

Funding for this publication is from a cooperative agreement with the Centers for Disease Control and Prevention (Grant Number: 5U50DD000612-05).



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