



RHODE ISLAND BIRTH DEFECTS PROGRAM

BIRTH DEFECTS DATA BOOK 2012

RHODE ISLAND DEPARTMENT OF HEALTH



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. Of these, 8,000 (6.7%) die during the first year of life. Many babies who survive beyond their first year have childhood illness and disability.

Birth defects cause about one in five infant deaths in the United States and Rhode Island. During 2010, of the 79 infants who died in Rhode Island, 15 of the deaths (19.0%) were attributed to a birth defect. This represents an increase in the proportion of infant deaths from birth defects since 2006, when 12 (16.2%) of the 74 Rhode Island infant deaths resulted from a birth defect. It also represents a 38% increase in the rate of infant mortality from birth defects. This rate rose from 97.0 per 100,000 live births in 2006 to 134.3 per 100,000 live births in 2010.

Many preterm infants (born before 37 weeks gestation) have birth defects. In Rhode Island, 97 (9.3%) of the 1,036 preterm babies born in 2010 had a birth defect. Preterm birth is the leading cause of infant death in Rhode Island. In 2010, more than one quarter (26.6%) of Rhode Island's 79 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 this 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 2008-2010, Rhode Island's hospital discharge database identified 1,590 newborns with at least one birth defect. The total hospital cost for these newborns was about \$102 million, with an average cost per newborn of \$64,403. This amounts to more than six times the cost for a newborn with no birth defects (\$10,441). The average length of stay for a newborn with a birth defect (10.3 days) was nearly three times higher than that for a newborn without a birth defect (3.7 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 (9.2 days) was over three times higher than the average length of stay for children without birth defects (2.8 days). The average cost per day for children with birth defects was \$57,124 compared to \$15,020 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 TOTAL ADJUSTED HOSPITAL CHARGES FOR NEWBORNS BY SELECTED BIRTH DEFECTS, RHODE ISLAND AND UNITED STATES

BIRTH DEFECT	RHODE ISLAND: 2008-2011			UNITED STATES: 2003		
	CASES	LENGTH OF STAY (DAYS)	MEAN TOTAL COST (\$)	CASES	LENGTH OF STAY (DAYS)	MEAN TOTAL COST (\$)
Spina bifida	2	8	25,291	1,136	15	65,942
Microcephaly	15	8	18,500	NA	NA	NA
Hydrocephaly	8	29	92,184	NA	NA	NA
Cleft lip with and without cleft palate	17	3	2,899	3,486	6	15,397
Cleft palate	12	5	4,925	2,187	10	33,387
Rectal and large intestinal atresia/stenosis	5	26	86,719	1,604	17	75,220
Esophageal atresia/tracheoesophageal fistula	4	18	58,180	NA	NA	NA
Gastroschisis	4	18	117,496	NA	41	155,620
Down syndrome	9	8	3,062	5,036	11	38,745

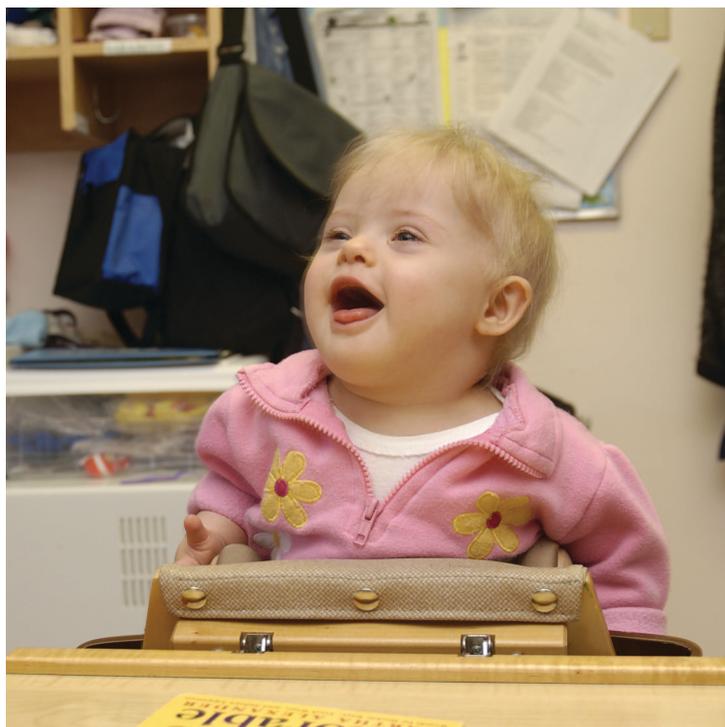
Notes: Costs adjusted using hospital cost-to-charge ratios. Mean total cost refers to the average cost per case across all maternity hospitals.
Sources: Rhode Island Hospital Discharge Database, Rhode Island Department of Health; Centers for Disease Control and Prevention. Hospital stays, hospital charges, and in-hospital deaths among infants with selected birth defects—United States, 2003. MMWR Morb Mortal Wkly Rep 2007;56(2):25-9.

PUBLIC HEALTH SURVEILLANCE: RHODE ISLAND BIRTH DEFECTS PROGRAM

Early recognition of and response to birth defects often prevents more serious health problems. An active birth defects surveillance and information system is essential to develop programs and policies to reduce both the incidence of 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; detect morbidity (disease) and mortality (death) trends; and help 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 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.

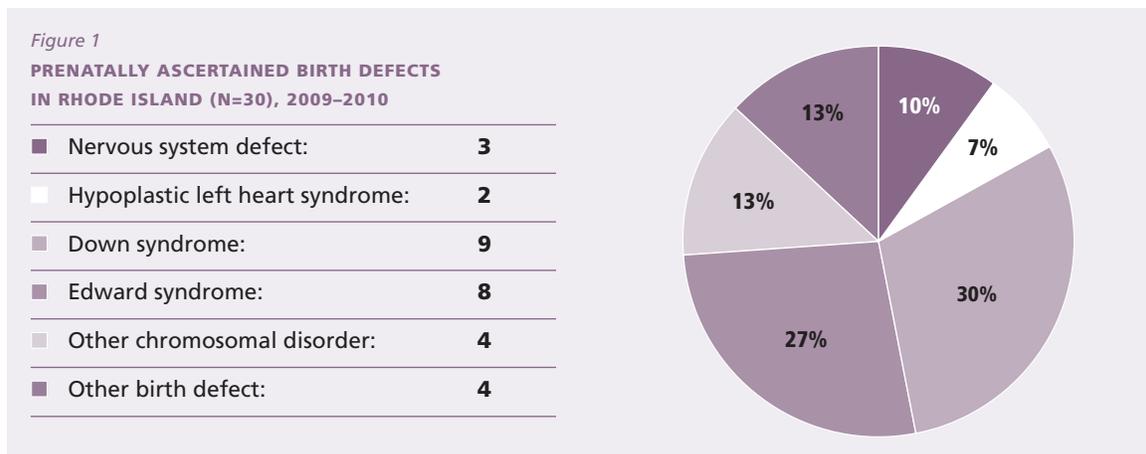
CASE IDENTIFICATION AND DATA

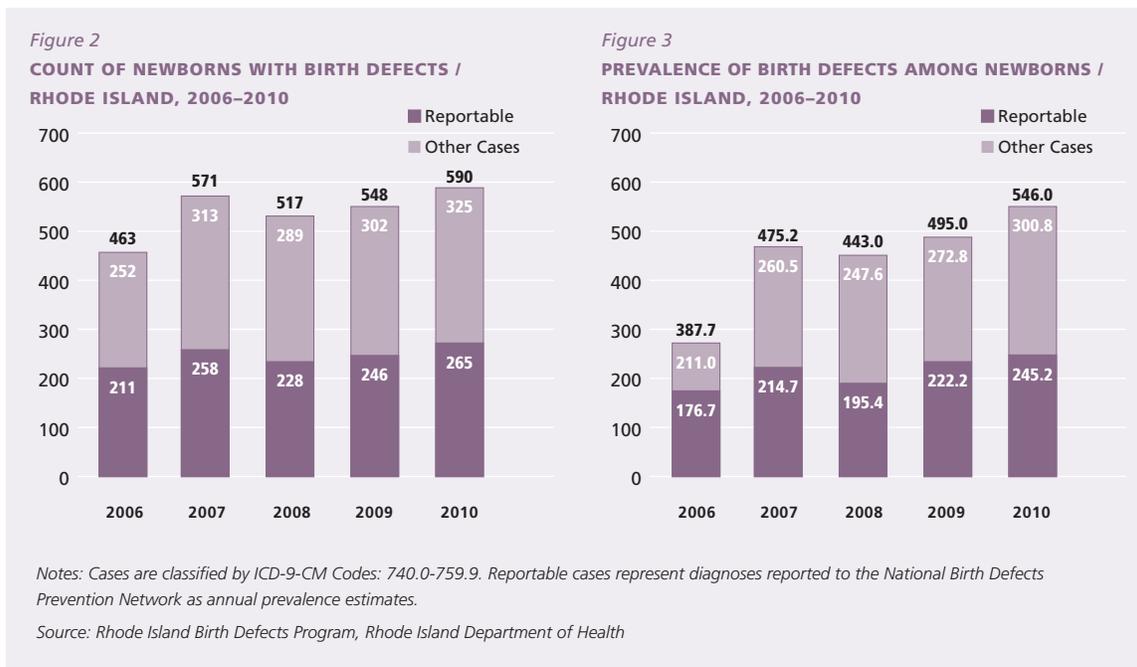
The RIBDP uses hospital discharge data (HDD) as the primary source for capturing birth defects data in Rhode Island. The RIBDP works with all seven maternity hospitals to collect discharge information. The RIBDP also collects information from specialty clinics, such as the Children’s Neurodevelopment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

Birth defects cases 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 updated its birth defects case definition in 2008 to exclude certain minor congenital anomalies and to reflect birth defects surveillance guidelines developed by the National Birth Defects Prevention Network. Previous data have been adjusted to fit this current case definition for comparable data analyses.

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 Genetics Testing Center and Fetal Treatment Program at Women and Infants 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 the time of newborn discharge. Figure 1 shows the percentages of birth defects diagnoses ascertained in the prenatal stages for the 2009-2010 period in Rhode Island. Nearly one third of all prenatally ascertained cases (n=30) were diagnosed with Down syndrome. The RIBDP plans to continue expanding routine prenatal surveillance with prenatal data partners to better understand birth defects prevalence and trends in Rhode Island.





Data

Figures 2 and 3 show the overall count and prevalence of birth defects in Rhode Island over the five-year period from 2006 to 2010. The totals are grouped by all birth defects and those reported to the CDC and National Birth Defects Prevention Network. During this period, the rate of birth defects in Rhode Island increased by 40.8% from 387.7 per 10,000 live births in 2006 to 546.0 per 10,000 in 2010. The low birth defects rate in 2006 (387.7 per 10,000) is unusual, as there were no changes in methods used to identify birth defects or in the trends of other birth outcomes such as low birth weight, preterm birth, and infant mortality.

Table 2 shows the number and prevalence of selected birth defects among Rhode Island residents during 2006-2010, organized by organ system. Cardiovascular defects (those relating to the heart) are the most common type of defects (188 per 10,000 live births). Ventricular septal defects and patent ductus arteriosus represent the highest proportion of these cases.

Other common birth defects in Rhode Island include those related to musculoskeletal (129 per 10,000) and genitourinary (142 per 10,000) systems. Among these groups of defects, hypospadias and epispadias (39 per 10,000) and obstructive genitourinary defect (30 per 10,000) are most frequent, but rates for these conditions have remained stable.

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 valve atresia, tetralogy of Fallot, total anomalous pulmonary venous return, complete transposition

Table 2

SELECTED BIRTH DEFECTS, RHODE ISLAND, 2006-2010

BIRTH DEFECT	NUMBER	RATE (PER 10,000 LIVE BIRTHS)	BIRTH DEFECT	NUMBER	RATE (PER 10,000 LIVE BIRTHS)
Central Nervous System	100	16.8	Gastrointestinal	308	51.9
Anencephalus	3	0.5	Esophageal atresia / tracheoesophageal fistula	16	2.7
Spina bifida	14	2.4	Pyloric stenosis	4	0.7
Encephalocele	1	0.2	Rectal and large intestinal atresia / stenosis	21	3.5
Microcephalus	18	3.0	Hirschsprung's disease	4	0.7
Hydrocephalus	29	4.9	Biliary atresia	1	0.2
Eye	76	12.8	Genitourinary	845	142.4
Microphthalmos	3	0.5	Hypospadias and epispadias	230	38.8
Congenital cataract	3	0.5	Bladder extrophy	1	0.2
Ear	55	9.3	Renal agenesis / hypoplasia	7	1.2
Anotia / Microtia	6	1.0	Obstructive genitourinary defect	177	29.8
Cardiovascular	1117	188.2	Musculoskeletal	766	129.1
Common truncus	1	0.2	Congenital hip dislocation	44	7.4
Complete transposition of great arteries	17	2.9	Clubfoot	77	13.0
Tetralogy of Fallot	19	3.2	Reduction deformity, upper limbs	9	1.5
Ventricular septal defect	205	34.5	Reduction deformity, lower limbs	12	2.0
Atrial septal defect	196	33.0	Diaphragmatic hernia	15	2.5
Atrioventricular septal defect	12	2.0	Gastroschisis	23	3.9
Pulmonary valve atresia / stenosis	34	5.7	Omphalocele	11	1.9
Tricuspid valve atresia / stenosis	2	0.3	Chromosomal	141	23.8
Ebstein's anomaly	2	0.3	Down syndrome	80	13.5
Aortic valve stenosis	5	0.8	Trisomy 13	10	1.7
Hypoplastic left heart syndrome	10	1.7	Trisomy 18	18	3.0
Patent ductus arteriosus	191	32.2	Other birth defects / conditions	83	14.0
Coarctation of aorta	11	1.9	All birth defects	3678	
Respiratory	107	18.0	All birth defect cases	2726	459.3
Choanal atresia	3	0.5			
Orofacial	80	13.5			
Cleft palate	35	5.9			
Cleft lip w/ and w/o cleft palate	45	7.6			

*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 2006-2010. The bolded "All birth defect cases" row represents the total number of Rhode Island babies born between 2006 and 2010 with at least one diagnosed birth defect.

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



of the great arteries, tricuspid valve atresia, and truncus arteriosus. CCHD can involve abnormal heart rhythms and structural heart problems including abnormal or absent chambers, holes in the heart, abnormal connections, and abnormal functioning. CCHD may require intervention and most commonly surgery within the first days of a newborn's life. 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.

The RIBDP is collaborating with the Rhode Island Newborn Screening Program, New England Genetics Collaborative, other New England states, and participating maternity hospitals in a grant-funded study to evaluate the use of pulse oximetry screening to detect CCHD in Rhode Island. The RIBDP is closely monitoring the seven CCHD conditions. HEALTH staff also participate on the Rhode Island Newborn Screening Advisory Committee CCHD Subcommittee to discuss the study's methods, provider education, and related topics.

Table 3 lists the counts and prevalence rates of the seven CCHD birth defects. These represent 2006-2010 data.

Table 3

**PREVALENCE OF BIRTH DEFECTS ASSOCIATED WITH CRITICAL CONGENITAL HEART DEFECTS
RHODE ISLAND, 2006-2010**

CONDITION	NUMBER	RATE/10,000 LIVE BIRTHS (95% CI)
Hypoplastic left heart syndrome	9	1.6 (0.7-3.0)
Tetralogy of Fallot	22	3.8 (2.4-5.8)
Total anomalous pulmonary venous return	4	0.7 (0.2-1.8)
Complete transposition of great arteries	9	1.6 (0.7-3.0)
Tricuspid valve atresia	3	0.5 (0.1-1.5)
Truncus arteriosus	2	0.3 (0.0-1.3)
Pulmonary valve atresia	9	1.6 (0.7-3.0)

Note: Confidence intervals (CI) represent the range of probable values and reflect the "stability" of an estimate. 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.

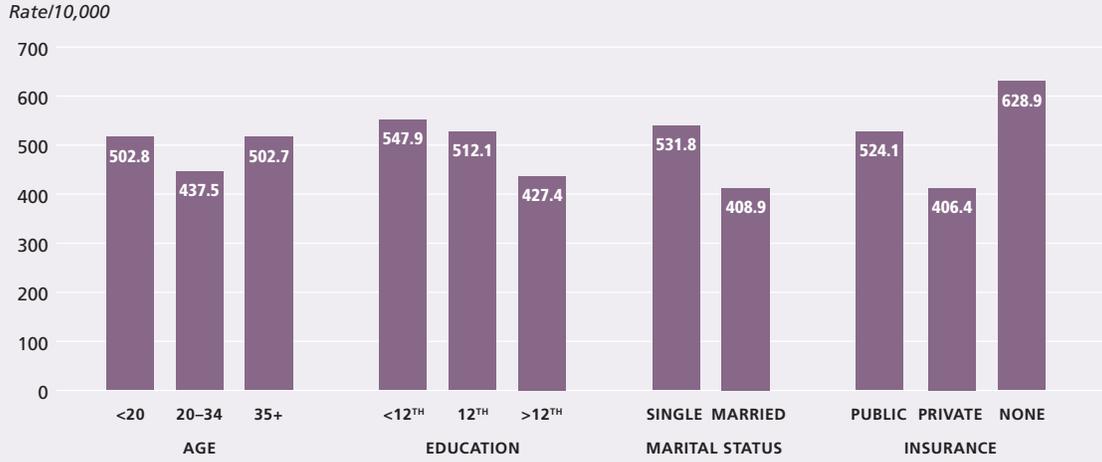
There is no known safe amount, no safe time, and no safe type of alcohol to drink during pregnancy.

Maternal Characteristics

Babies born to older women (aged 35 or older), younger women (aged less than 20), 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 (Figure 4). During 2006-2010, the birth defects prevalence rate among women aged less than 20 or 35 or older was 503 (per 10,000 live births) compared to 438 among women aged 20-34. The birth defects rate among women with less than a 12th grade education (548 per 10,000 live births) or with a 12th grade education (512) was higher than the rate among women with more education (427). Similarly, single women were more likely to have a baby with a birth defect (532 per 10,000 live births) than married women (409). Women insured through public programs such as RItE Care and Medicaid were also more likely to have a baby born with a birth defect (524 per 10,000 live births) than women insured through commercial or private providers such as Blue Cross & Blue Shield of Rhode Island or UnitedHealthcare (406). Although a small number of Rhode Island women did not have health insurance at the time of delivery (about 1%), the risk for having a baby with a birth defect was higher among this population (629 per 10,000 live births) than the combined rate for women with public or private health insurance (465).

Figure 4

BIRTH DEFECTS PREVALENCE BY SELECTED MATERNAL CHARACTERISTICS, RHODE ISLAND, 2006–2010

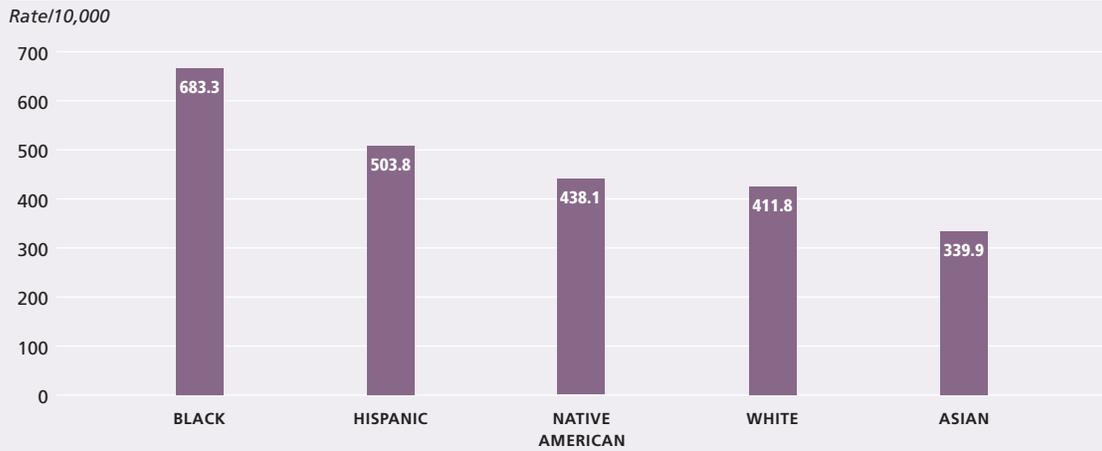


Source: Rhode Island Birth Defects Program, Rhode Island Department of Health

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

Figure 5

BIRTH DEFECTS RATES BY RACE / ETHNICITY, RHODE ISLAND, 2006–2010



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

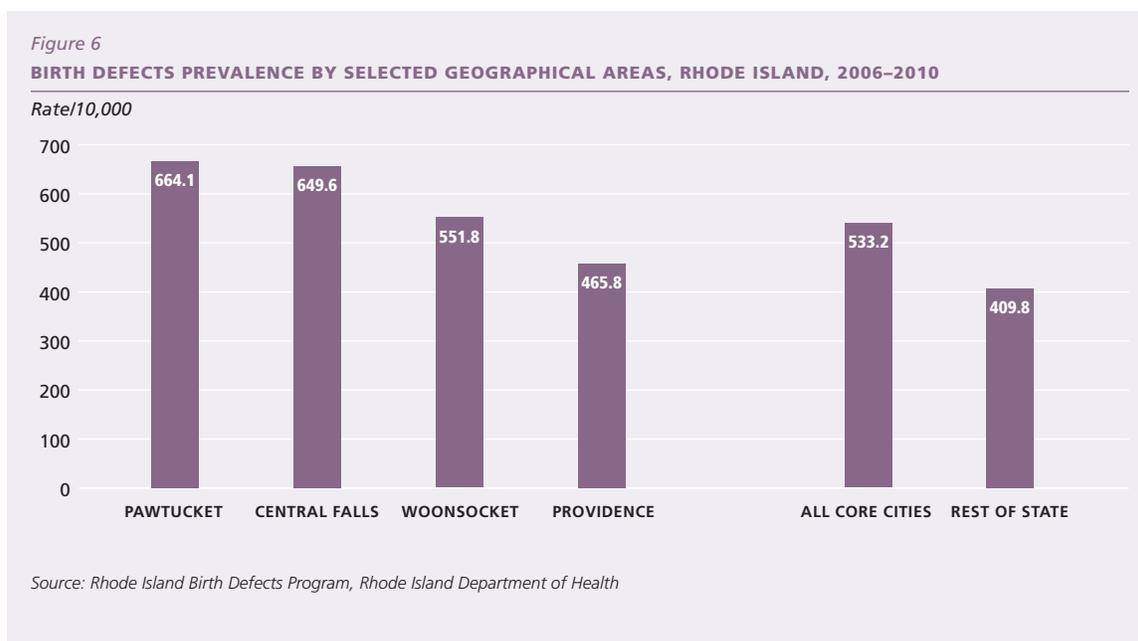
Racial/Ethnic and Geographic Disparities

Birth defects prevalence varies by race/ethnicity (Figure 5) and geographical residence (Figure 6). During 2006-2010, the average birth defects prevalence rate among Black/African Americans (683 per 10,000 live births) was 1.7 times the rate for Whites (412). Those of Hispanic ethnicity had the second highest rate (504 per 10,000 live births), which was 1.2 times higher than the rate for Whites. During this period, Asian women were least likely to have a baby born with a birth defect (340 per 10,000 live births).

Babies born to residents of core cities where the child poverty level is greater than 25% (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 6). Pawtucket and Central Falls had the highest birth defects rates in the state (664 and 650 per 10,000 live births, respectively). 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.

Mapping Rates of Birth Defects

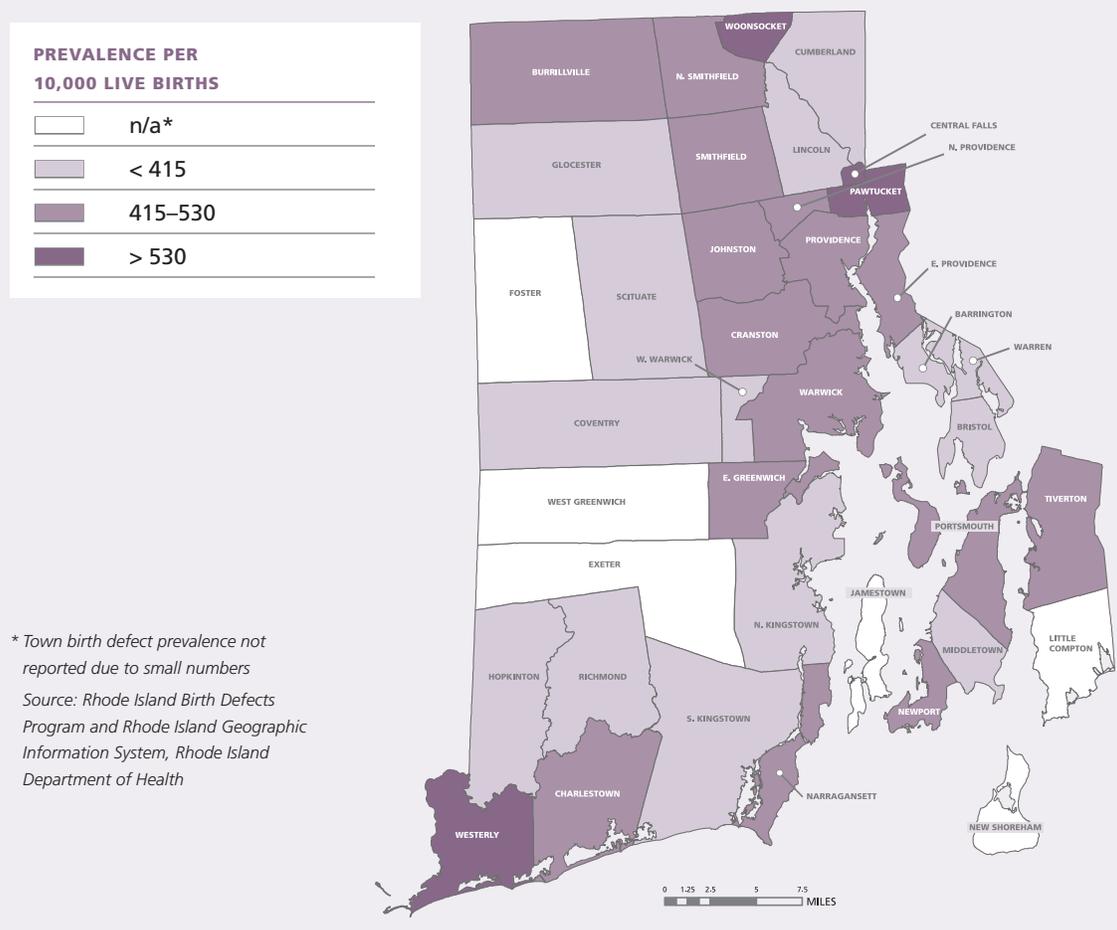
Geospatial analysis allows us to follow trends and detect clusters of birth defects in Rhode Island. The map on the next page illustrates birth defects prevalence rates during 2006-2010 by city/town in Rhode Island. To address small sample sizes in some cities and towns, the RIBDP has implemented data suppression rules. Prevalence rates for towns with a case count less than 15 or a live birth population less than 200 during the 2006-2010 period are not reported. This includes the towns of Exeter, Foster, Jamestown, Little Compton, New Shoreham, and West Greenwich.



The map shows a majority of cities and towns with a prevalence rate between 415 and 530 births with a birth defect per 10,000 live births, a common range for birth defects rates. As mentioned previously, Central Falls and Pawtucket form an urban area of high birth defects prevalence in Rhode Island. Southwestern Rhode Island towns such as Westerly show a marked increase in birth defects prevalence. This may be attributed to different diagnostic methods or hospital coding procedures.

Figure 7

BIRTH DEFECTS PREVALENCE BY CITIES / TOWNS, RHODE ISLAND, 2006-2010



SERVICE ASSESSMENT 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 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

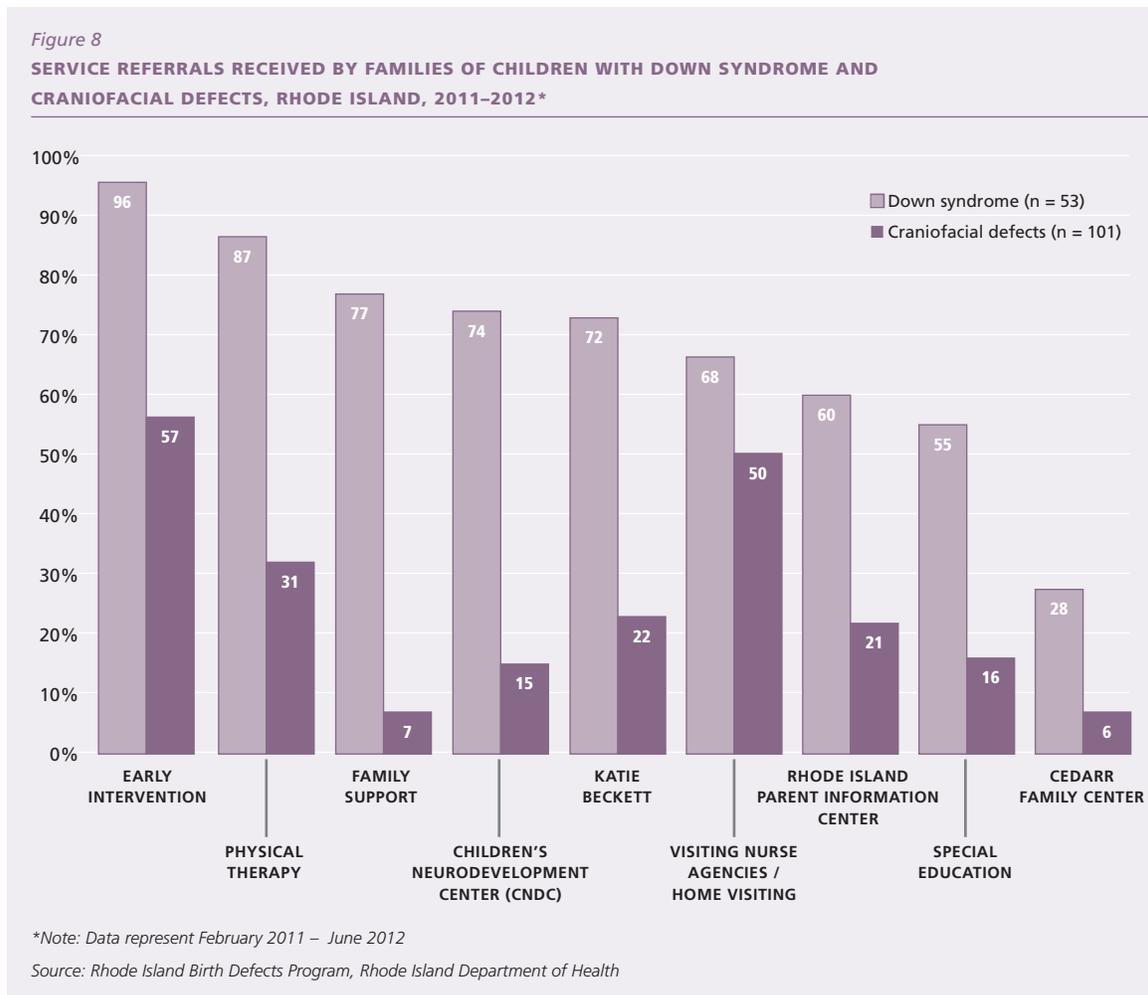
timely basis. The Family Resource Specialist meets with families at pediatric and specialty care practices that serve children with birth defects such as the Children's Neurodevelopment Center (CNDC) at Hasbro Children's Hospital. The RIBDP also works with the Family Resource Specialist to mail service assessment surveys to additional families of children with birth defects. The surveys are conducted with families of children, newborn to five years of age, to determine what services and referrals children received based on the national guidelines for specific conditions. Specifically, the survey asks about services such as medical tests and procedures, developmental and educational



services, and parent supports. Currently, the RIBDP conducts these surveys with families of children with Down syndrome, spina bifida, club foot, and craniofacial abnormalities. The RIBDP plans to include Critical Congenital Heart Defects (CCHD) in the near future.

Figure 8 shows the number of service assessment surveys completed by families during February 2011 through June 2012. The data summarize the number of completed surveys of families of children with Down syndrome (n=53) and craniofacial defects (n=101) and the percent of those 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 rate of referrals for children with Down syndrome (96%), while the referral rate for children with craniofacial conditions was 57%. These differences reflect the variation in needs children with these conditions may have. Compared to referrals for early intervention services, the referral rate to Special Education was much lower, with only 55% of children with Down syndrome and 16% of children with craniofacial conditions receiving these referrals. These lower rates may be due partially to age requirements, as Special Education serves children age three and above. Differences also exist in referral rates for financial support services. Families of children Down syndrome were more likely to receive referrals to the Katie Beckett Program (72%) compared to families of children with craniofacial conditions (22%). 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, more than three-quarters (77%) of families of children with Down syndrome received referrals to family support services compared to only 7% of families of children with craniofacial conditions.

As of October 2012, the RIBDP received 222 surveys completed during 2011-2012 by families of children with the four conditions (Down syndrome, spina bifida, club foot and craniofacial anomalies). The surveys include a comments section, where many families noted that although they were referred to services, they still face financial, social, and educational challenges. 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 inadequate and said they needed help to navigate health and educational systems. Many families did report 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.





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, women can take a number of steps 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 is currently working with various public health programs on birth defects awareness and risk-reduction activities. For example, in collaboration with Rhode Island's Family Planning Program, the RIBDP 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) financed this effort.

Additionally, the RIBDP worked with the Rhode Island Women, Infants, and Children (WIC) Program to distribute multivitamins containing 400 micrograms of folic acid to WIC participants at selected WIC sites during 2011. This initiative was conducted



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

in collaboration with the New England Birth Defects Consortium (NEBDC). The NEBDC made about 1,000 bottles of multivitamins available for distribution in each New England state through funding from the New England Genetics Collaborative. The target population for the project was postpartum and breastfeeding WIC participants. In addition to the vitamins, the WIC Program received kits including implementation instructions for WIC coordinators, folic acid education materials, and surveys to evaluate this intervention. Evaluation included pre- and post-surveys, with the initial (pre) survey conducted when the vitamins were distributed and the post-survey conducted three to six months after the initial distribution. WIC participants received a second three-month supply of multivitamins after completing the post-survey. In addition to the multivitamins and nutrition education, the WIC participants received educational materials that included brochures produced by the CDC and March of Dimes. All completed surveys were sent to the NEBDC regional coordinating center, which provided the states with their data. Among all respondents who completed a pre-survey, 152 (30.0%) completed and returned a post-survey. Data from the post-survey indicate that 108 (71.0%) of the 152 respondents were taking multivitamins; of this group, 59 (54.6%) had reported in the pre-survey that they were not taking multivitamins.

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.

Each January, the RIBDP works with its Advisory Council to plan and coordinate pediatric grand rounds at Rhode Island Hospital 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 is extended to two hours to include 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 on topics related to birth defects are held throughout January. Past topics have included spina bifida, fetal alcohol syndrome, Down syndrome, craniofacial anomalies, and hearing loss.

PARTNERSHIPS

In addition to sponsoring pediatric grand rounds, 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, Hasbro Children's Hospital, the Rhode Island Chapter of the March of Dimes, the Rhode Island Parent Information Network, and Rhode Island KIDS COUNT.

RESOURCES

RHODE ISLAND

March of Dimes Rhode Island: www.marchofdimes.com/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: 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: 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): www.dhs.ri.gov/ChildrenwithSpecialNeeds/EarlyInterventionProgram/tabid/839/default.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 Healthy Mothers Healthy Babies Coalition: 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
79 Plain Street, 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 genetic 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 preconception evaluations

Children's Neurodevelopment Center

Hasbro Children's Hospital
593 Eddy Street, Providence, RI 02903
Phone: 401-444-5685
Fax: 401-444-6115

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 02905
Phone: 401-453-7652
Fax: 401-453-7547.
Offers testing for cytogenetics, molecular cytogenetics, and molecular genetics

Prenatal & Special Testing Laboratory
Women & Infants Hospital
70 Elm Street, 2nd Floor, Providence, RI 02903
Phone: 888-AFP-MOMS
www.womenandinfants.org/Services/Medical-Screening-and-Special-Testing.cfm
Provides prenatal alpha-fetoprotein (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, Guam, and internationally.

American Academy of Pediatrics (AAP): 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.

Birth Defect Research for Children, Inc.: www.birthdefects.org

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

National Birth Defects Prevention Network (NBDPN): 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): 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 to improve health and prevent disease and disability. Below are links to 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: www.cdc.gov/reproductivehealth/MaternalInfantHealth
- Office of Genomics and Public Health: www.cdc.gov/genomics

National Dissemination Center for Children with Disabilities: www.nichcy.org

Provides information on disabilities in children and youth; programs and services for infants, children, and youth with disabilities; the Individuals with Disabilities Education Act (IDEA), the nation's special education law; No Child Left Behind, the nation's general education law; and research-based information on effective practices for children with disabilities.

National Organization on Fetal Alcohol Syndrome: 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: www.nsgc.org

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

Organization of Teratology Information Specialists (OTIS): www.otispregnancy.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.

Smiles: 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: www.sbaa.org

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

The Teratology Society: 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 to conduct worldwide surveillance and research, prevent birth defects, and improve 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, upper or lower limbs	Deformity of the arms or legs, in which one or both arms or legs 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.

ACKNOWLEDGEMENTS

RHODE ISLAND BIRTH DEFECTS ADVISORY COUNCIL

MARCH OF DIMES, RHODE ISLAND CHAPTER

WOMEN AND INFANTS HOSPITAL

RHODE ISLAND HOSPITAL

HASBRO CHILDREN'S HOSPITAL

RHODE ISLAND KIDS COUNT

RHODE ISLAND PARENT INFORMATION NETWORK (RIPIN)

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