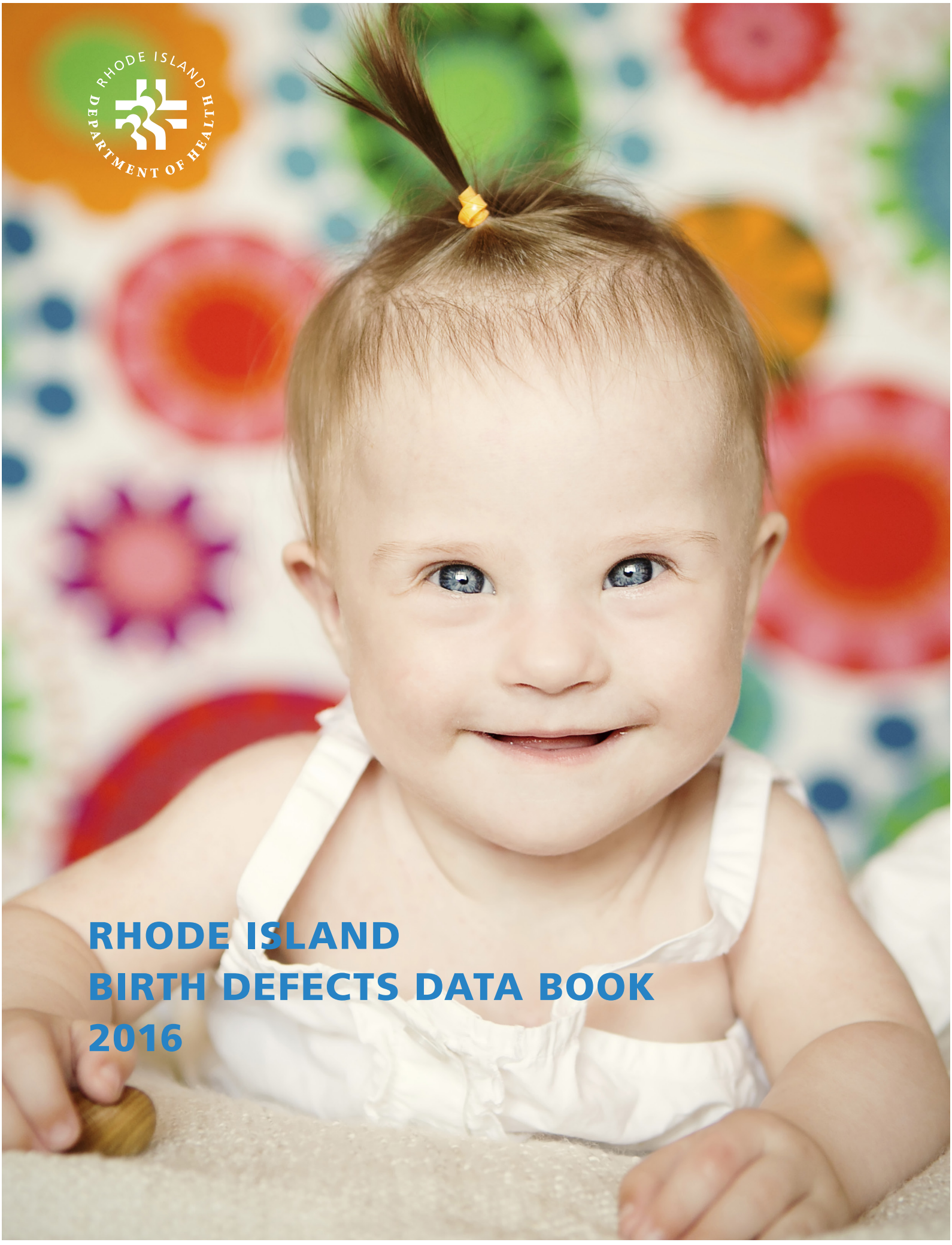


RHODE ISLAND BIRTH DEFECTS DATA BOOK 2016

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

Although birth defects cause about one in five deaths in the United States, they represent approximately 8% of infant deaths in Rhode Island. Provisional data indicate that among the 100 infants who died in 2014 and 2015, eight of the deaths (8.0%) were attributed to a birth defect. This represents a 37.5% decrease in the proportion of infant deaths resulting from a birth defect since 2010, when 10 (12.8%) of the 78 infant deaths resulted from a birth defect.

Many preterm infants (born before 37 weeks gestation) have birth defects. In Rhode Island, in 2014, 16 (1.8%) of the 903 preterm babies had a birth defect. Preterm birth is the leading cause of infant death in Rhode Island. During the two-year period, 2014-2015, 47 (47%) of the 100 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 (RIDOH) studies the costs of selected birth defects using national surveillance guidelines based on the severity and frequency of the birth defect. During 2012-2014, Rhode Island's hospital discharge database identified 1,390 newborns with at least one birth defect. The total unadjusted cost for newborn admissions with a birth defect diagnosis is $205,769,517 with an average cost per newborn of $148,036. This amounts to more than 12 times the cost for a newborn with no birth defects ($11,740). The average length of stay for a newborn with birth defects

(13.2 days) was nearly four 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 indicate that the average length of stay for children with birth defects (6.9 days) was twice as high as the average length of stay for children without birth defects (2.9 days).

Hospital discharge data provide adjusted charges and specific hospital costs for newborn admissions (see Table 1, page 4). The Rhode Island analysis shows the total adjusted charges for all newborn hospital admissions with a single diagnosed birth defect. Table 1 compares the number of cases, mean lengths of stay, and mean hospital charges per case (based on hospital cost-to-charge ratios) in Rhode Island for selected birth defects

TABLE 1: MEAN TOTAL ADJUSTED HOSPITALIZATION CHARGES BY SELECTED BIRTH DEFECTS, RHODE ISLAND, 2012-2014

Birth Defect	Cases	Length of Stay (Days)	Adjusted Cost	Cost per day
Spina bifida	3	10	48,449	4,845
Microcephaly	14	5	5,844	1,169
Hydrocephaly	6	34	125,455	3,690
Cleft lip w/ and w/o cleft palate	27	3	2,810	937
Cleft palate	11	11	17,165	1,560
Rectal and large intestinal atresia/stenosis	5	29	110,390	3,807
Gastroschisis	13	34	129,031	3,795
Down syndrome	13	4	4,372	1,093

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

PUBLIC HEALTH SURVEILLANCE: RHODE ISLAND BIRTH DEFECTS PROGRAM

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

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

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

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

Reportable Birth Defects

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

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

CASE IDENTIFICATION AND DATA

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

Birth defects cases are children born to Rhode Island residents, from birth up to age five, and are identified using diagnoses coded by the 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 from 2010 through 2014. 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 decreased by 5% from 356.1 per 10,000 live births in 2010 to 338.4 per 10,000 live births in 2014, after adjusting for the updated birth defects case definition. During this same period, the prevalence of cases with the 46 conditions that are reported to the CDC remained constant. In 2010, the prevalence of these cases was 174.3 per 10,000 live births (n = 188), and in 2014, the prevalence was 174.0 (n = 181).

FIGURE 1. BIRTH DEFECTS CASES, REPORTABLE AND OTHER CASES, RHODE ISLAND, 2010-2014

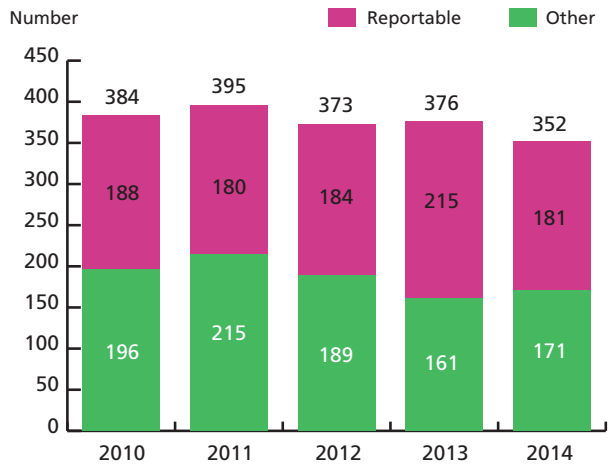
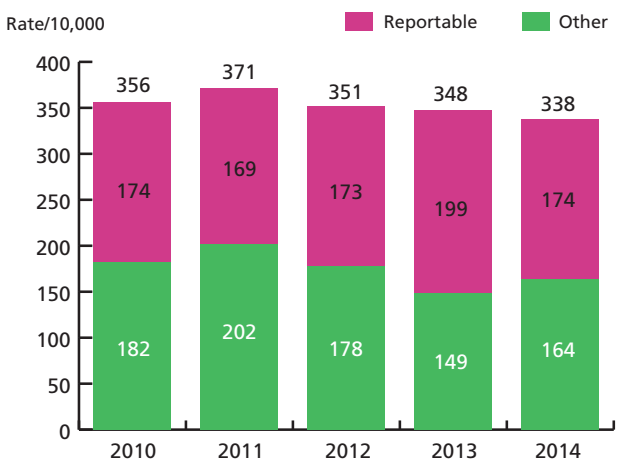


FIGURE 2. PREVALENCE OF BIRTH DEFECTS FOR REPORTABLE AND OTHER CASES, RHODE ISLAND, 2010-2014



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

Table 2 shows the number and prevalence of selected birth defects among Rhode Island residents during 2010-2014, organized by organ system. Musculoskeletal defects are the most common type of defects (131.94 per 10,000). Among these defects, club foot represents the highest proportion of these cases.

Other common birth defects in Rhode Island include those related to cardiovascular (131.5 per 10,000) and genitourinary (93.0 per 10,000) systems. Among cardiovascular defects, ventricular septal defects (41.5) and atria septal defects (23.3) are most common, but rates for these conditions have remained stable. Within the genitourinary system, hypospadias represents nearly half (48%) of those conditions and had a prevalence rate of 44.9.

TABLE 2. CASES AND PREVALENCE OF SELECTED BIRTH DEFECTS, RHODE ISLAND, 2010-2014

BIRTH DEFECT	NUMBER	RATE (PER 10,000 LIVE BIRTHS)
Central Nervous System	**112**	**20.4**
Hydrocephaly	17	
Microcephaly	31	
Spina Bifida	13	
Anencephaly	4	
Encephalocele	4	
Holoprosencephaly	3	
Eye/Ear/Face/Neck	**51**	**9.6**
Congenital cataract	6	
Anophthalmos/Microphthalmos	5	
Anotia / Microtia	5	
Cardiovascular	**700**	**131.5**
Ventricular septal defect	221	
Atrial septal defect	124	
Pulmonary valve atresia and stenosis	30	
Tetralogy of Fallot	12	
Transportation of great vessels	9	
Hypoplastic left heart syndrome	7	
Coarctation of aorta	11	
Atrioventricular septal defect	7	
Aortic valve stenosis	8	
Ebstein's anomaly	2	
Tricuspid valve atresia	5	
Truncus arteriosus	3	
Double outlet right ventricle	7	
Single ventricle	2	
Total anomalous pulmonary venous connection	3	
Interrupted aortic arch	1	
Respiratory	**33**	**6.2**
Choanal atresia	3	

BIRTH DEFECT	NUMBER	RATE (PER 10,000 LIVE BIRTHS)
Orofacial	**63**	**11.5**
Cleft lip alone	19	
Cleft palate alone	21	
Cleft lip/with cleft palate	27	
Gastrointestinal	**118**	**22.2**
Rectal and large intestinal atresia/stenosis	12	
Small intestinal atresia/stenosis	21	
Esophageal atresia/ tracheoesophageal fistula	3	
Hirshsprung's disease	1	
Biliary atresia	1	
Genitourinary	**495**	**93.0**
Hypospadias	239	
Renal agenesis / hypoplasia	18	
Congenital posterior urethral valves	4	
Bladder extrophy	2	
Musculoskeletal	**702**	**131.9**
Clubfoot	63	
Craniosynostosis	17	
Gastroschisis	21	
Omphalocele	10	
Limb deficiencies	15	
Diaphragmatic hernia	11	
Chromosomal	**91**	**17.1**
Down syndrome	63	
Edward syndrome	3	
Patau syndrome	5	
Turner syndrome	1	
All birth defects	**2,426**	**455.8**
All birth defect cases	**1,180**	**353.2**

*Note: Numbers and rates in each bolded body system row represent total diagnosed birth defects associated with that body system. The bolded "All birth defects" row represents all birth defects diagnosed in Rhode Island during 2010-1014. The bolded "All birth defects cases" row represents the total number of Rhode Island babies born between 20101-2014 with at least one diagnosed birth defect.
Source: Rhode Island Birth Defects Program, Rhode Island Department of Health

Identification of cases during the prenatal period

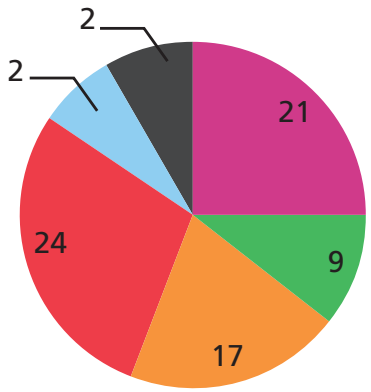
In 2008, the RIBDP began collecting birth defects cases identified during the prenatal period from collaborating laboratories and prenatal clinics such as the Cytogenetics Testing Laboratory and Prenatal and Special Testing Laboratory at Women & Infants Hospital and the Fetal Treatment Program at Hasbro Children's Hospital. About four to six percent of birth defects are identified prenatally. Prenatal case ascertainment improves the prevalence estimate of certain birth defects by detecting cases not found at newborn discharge.

Figure 3 shows the percentages of birth defects diagnoses ascertained in the prenatal stages for the 2010-2014 period in Rhode Island. Among the 84 birth defects cases that were identified prenatally, chromosomal abnormalities (including Down syndrome and other trisomies; n = 41) account for nearly half of the cases. Specifically, Down syndrome was identified in nearly 20% of all prenatally ascertained cases (n = 17). Figure 4 shows the maternal age distribution of prenatally ascertained birth defects cases. The largest proportion of prenatally ascertained cases were among women aged 35 and older (n = 37)).

The RIBDP plans to establish active prenatal surveillance with prenatal data partners to better understand birth defects prevalence and trends in Rhode Island.

FIGURE 3. PRENATALLY ASCERTAINED BIRTH DEFECTS CASES*, BY TYPE (N=84), RHODE ISLAND 2010-2014

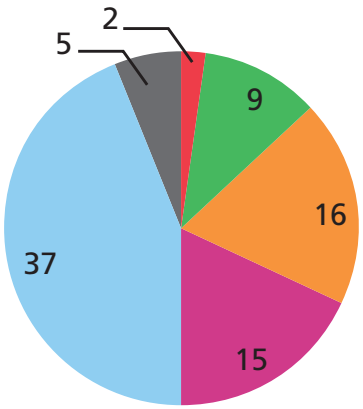
NERVOUS SYSTEM DEFECT	21
CRITICAL CONGENITAL HEART DEFECT	9
DOWN SYNDROME	17
OTHER TRISOMIES	24
OTHER CHROMOSOMAL DISORDER	6
OTHER BIRTH DEFECT	7



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

FIGURE 4. PRENATALLY ASCERTAINED BIRTH DEFECTS*, BY MATERNAL AGE (N=84), RHODE ISLAND 2010-2014

<20	2
20-24	9
25-29	16
30-34	15
35+	37
UNKNOWN	5



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

Critical Congenital Heart Defects

Critical congenital heart defects (CCHD) are a range of twelve heart defects that can cause serious, life-threatening symptoms (see Table 3 for list). CCHD may require intervention and, commonly, surgery within the first days of a newborn's life. These birth defects can involve abnormalities in rhythm of the heart and structural heart problems, including abnormal or absent chambers, holes in the heart, abnormal connections, and abnormal functioning. Babies who are not diagnosed or treated soon after birth are at high risk of death and disabilities later on 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.

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 screening panel. The goal was to identify CCHD among newborns before symptoms appear by conducting pulse oximetry screening, which measures oxygenation of the blood. A failed screen due to 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 has collaborated 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 CCHD in Rhode Island. Current data from the pilot study (as of September 2014) show that more than 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.

The counts of the 12 birth defects associated with CCHD are listed in Table 3 and represent 2010-2014 data.

TABLE 3. CRITICAL CONGENITAL HEART DEFECTS, RHODE ISLAND, 2010-2014

CONDITION	COUNT
Tetralogy of Fallot	15
Coarctation of aorta	14
Transposition of great vessels	10
Pulmonary valve atresia (with intact septum)	7
Tricuspid valve atresia	5
Hypoplastic left heart syndrome	5
Double outlet right ventricle	4
Single ventricle	4
Ebstein's anomaly	3
Total anomalous pulmonary venuous return	2
Truncus arteriosus	1
Interrupted aortic arch	1

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

Zika and Birth Defects

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

Microcephaly, a condition in which a newborn's head is abnormally small in comparison to other babies of the same age and sex, is a prominent birth defect associated with maternal infection of Zika. In April 2016, the CDC officially recognized Zika as the cause of microcephaly and other central nervous system defects. Research has shown that the Zika infects neural progenitor cells, precursor cells of the developing brain, impairing their growth. Infants who are infected during the first trimester are at the greatest risk for developing microcephaly, and infants who are not born with microcephaly, but are infected with Zika, can present other central nervous system defects, eye defects, and hearing loss.

The CDC is actively tracking the spread of Zika in the United States and rest of the world. They are also training healthcare professionals on how to identify the virus, doing public outreach about travel precautions, and safe behaviors, such as protected sex. Research regarding the link of Zika to other birth defects and anomalies is ongoing.

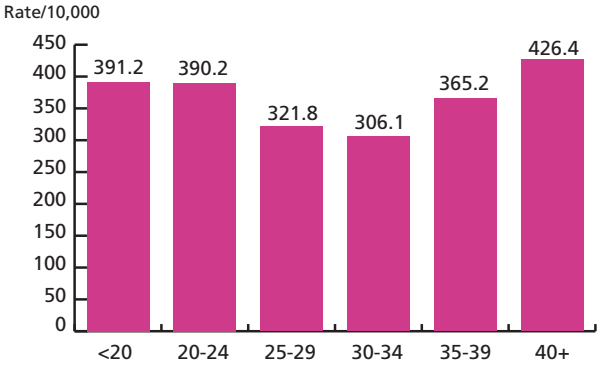
Currently, the RIBDP has implemented procedures to efficiently respond to cases of infants born with microcephaly and other central nervous system defects due to Zika. The RIBDP works in collaboration with other RIDOH programs and is a member of the RIDOH Zika Task Force. Additionally, RIBDP works closely with other state agencies, medical institutions, local organizations, and the media to address Zika-related issues in the state of Rhode Island. Specifically, the program has created an active surveillance system in collaboration with staff at the state's birthing hospitals to ensure rapid-case ascertainment of microcephaly and other central nervous system disorders upon newborn admission. As part of its proactive approach, the RIBDP will also be working with local obstetric providers to create an active surveillance system for infants who are diagnosed prenatally with conditions related to Zika, allowing for the program to follow up with these infants after birth. The RIBDP will work with providers in outpatient settings to capture microcephaly and other Zika-related conditions that may have been missed at birth. Lastly, Zika-related cases of microcephaly and central nervous system disorders have been added to the RIBDP's service assessment process, where a parent consultant will work with families of children born with these defects to ensure that they are receiving timely services and resources to adequately meet their children's needs.

Condoms can reduce the chance of getting Zika from sex.

Maternal Characteristics

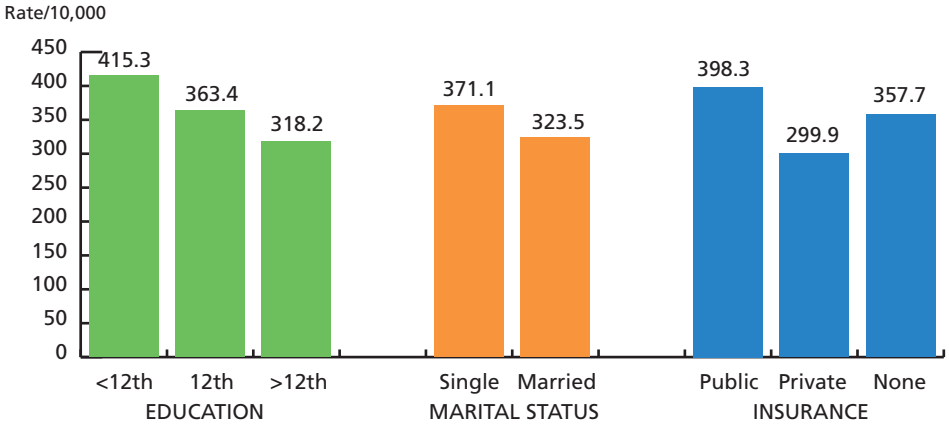
Babies born to women younger than 25, women 40 and older, women with less than a high-school education, unmarried women, and women with publicly funded health insurance or no health insurance are at a higher risk for birth defects (Figures 5 and 6). During 2010-2014, the birth defects prevalence rate among women younger than 25 was 390 per 10,000 live births and the rate among women age 40 and older was 426.4 per 10,000 live births compared to lower rates among women age 25-34. The birth defects rate among women with less than a high school education (415) or with a high school education (363) was higher than the rate among women with more education (318). Similarly, unmarried women were more likely to have a baby with a birth defect (371) than married women (323). Women who were insured through public programs such as RIte Care and Medicaid (398) were more likely to have a baby born with a birth defect than women who were insured through commercial or private providers (300).

FIGURE 5. PREVALENCE OF BIRTH DEFECTS BY MATERNAL AGE GROUP, RHODE ISLAND, 2010-2014



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

FIGURE 6. PREVALENCE OF BIRTH DEFECTS BY SELECTED MATERNAL CHARACTERISTICS, RHODE ISLAND, 2010-2014



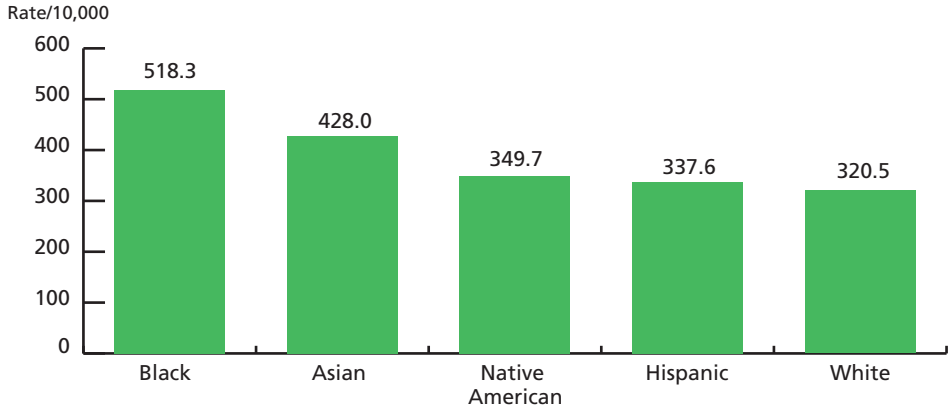
Source: Rhode Island Birth Defects Program, Rhode Island Department of Health

Racial/Ethnic and Geographic Disparities

Birth defects prevalence varies by race/ethnicity (Figure 7) and geographic residence (Figure 8). During 2010-2014, the average birth defects prevalence rate among Blacks/African Americans (518 per 10,000 live births) was 1.6 times the rate for Whites (321). Asians had the second highest rate (428), which was 33% higher than the rate for Whites. The birth defects prevalence rate among those of Hispanic/Latino ethnicity (338) 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.

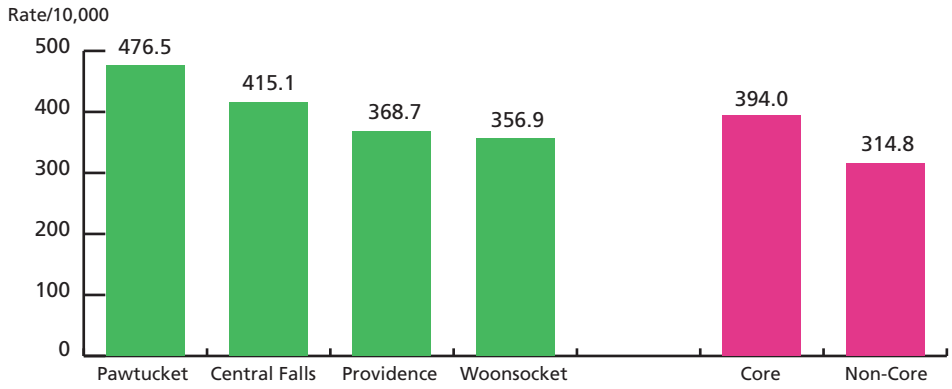
Babies born to residents of core cities where the poverty level is greater than 15% (Central Falls, Pawtucket, Providence, and Woonsocket) were about 1.3 times more likely to have a birth defect than babies born to residents living in the rest of the state (Figure 8). Pawtucket (476.5) and Central Falls (415) 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. PREVALENCE OF BIRTH DEFECTS BY RACE/ETHNICITY, RHODE ISLAND, 2010-2014



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

FIGURE 8. PREVALENCE OF BIRTH DEFECTS BY SELECTED GEOGRAPHIC AREAS, RHODE ISLAND, 2010-2014



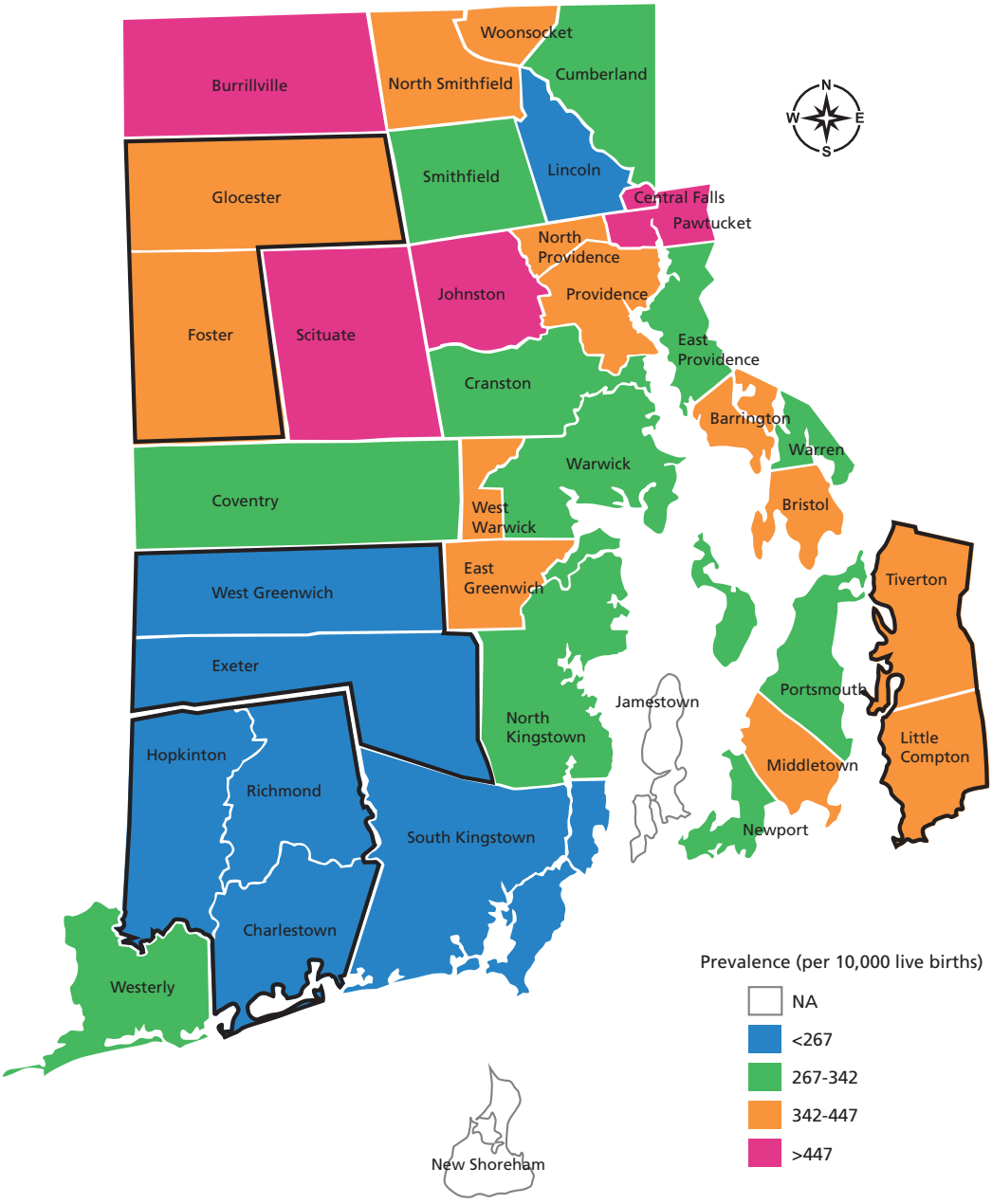
Source: Rhode Island Birth Defects Program, Rhode Island Department of Health

Mapping Rates of Birth Defects

Geospatial analysis allows us to follow trends and detect clusters of birth defects in Rhode Island. The map on the following page illustrates birth defects prevalence rates during 2010-2014 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 2010-2014 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, page 14) 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. PREVALENCE OF BIRTH DEFECTS BY GEOGRAPHIC AREAS, RHODE ISLAND, 2010-2014



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

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 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 the 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 send service assessment forms to additional families of children with birth defects. The service assessment forms are used by families of children, newborn to age five, to determine what services and referrals were provided to the children based on the national guidelines for specific conditions. Specifically, the assessment forms ask about medical tests and procedures, developmental and educational services, and parent supports. Currently, service assessments are conducted with families of children who have Down syndrome, spina bifida, craniofacial defects, critical congenital heart defects (CCHD), and abdominal wall defects (implemented in March 2016). In August 2016, the RIBDP expanded the service assessment process to include children with microcephaly or other central nervous system conditions (CNS) who tested either positive or negative for Zika-related viruses.

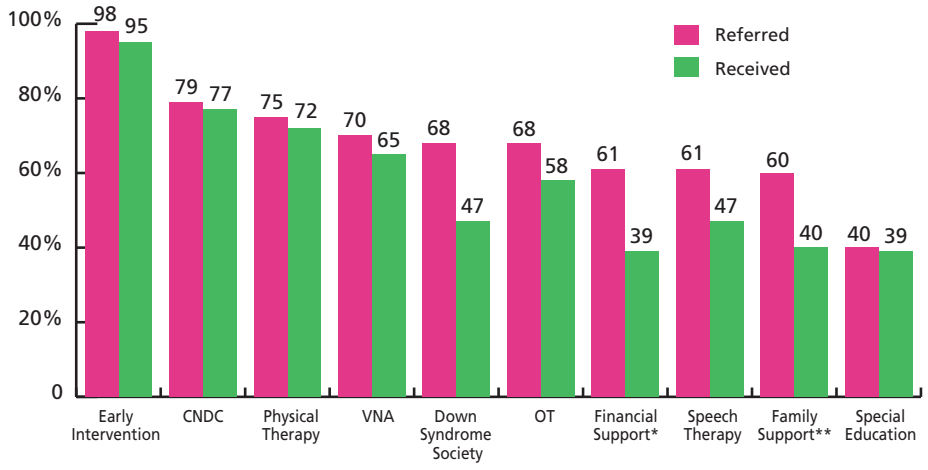
Smoking during pregnancy increases the chances of premature birth, certain birth defects, and infant death.

It should be noted that RIBDP service assessments for families of children with club foot were discontinued as of July 2016 because it was determined that most cases were treated very early and the children did not need additional services.

As of December 2016, RIBDP had received a cumulative total of 446 assessment forms completed from 2011 through 2016 by families of children with Down syndrome (n = 107), spina bifida (n = 49), club foot (n = 54), craniofacial anomalies (n = 209), CCHD (n = 19), abdominal wall defects (n = 8), and microcephaly/CNS conditions (n = 0). RIBDP also collected 192 repeat service assessments. The service assessment forms include a comment section where families have indicated that although they were referred to services, they still face challenges with financial, social, and educational issues associated with raising a child with a birth defect. Families reported that services previously provided or recommended by their child's treatment team were not provided by their school districts. Additionally, families found financial resources to be inadequate and said that they needed help from peer support and advocacy-assistance agencies to navigate health and educational systems. Some families thought the medical service needs of their children were not being met and chose to seek healthcare at facilities in other states. 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 summarizes the percentage of families of children with Down syndrome who received referrals to appropriate medical and social services. Nearly all (98%) of these families received referrals to the Early Intervention Program (EI), which provides developmental services to children from birth to age three, and 95% received EI services. A majority (79%) of these children with Down syndrome received a referral to Hasbro Children's Hospital's CNDC. However, families of children with Down syndrome were less likely to receive referrals for family support services (e.g., Rhode Island Parent Information Network) or for special education. A contributing factor to the latter is that special education services are for children age three and older.

FIGURE 10. SELECTED SUPPORT SERVICES, REFERRED FOR AND RECEIVED BY FAMILIES OF CHILDREN WITH DOWN SYNDROME, RHODE ISLAND, 2013-2016



*Includes Katie Beckett and Medicaid/SSI; **Includes RIPIN, Family Voices, and Sherlock Center
Source: Service Assessment Data, Rhode Island Birth Defects Program, Rhode Island Department of Health

REDUCING THE RISK OF BIRTH DEFECTS

Although not all causes of birth defects are known, there are a number of things a woman can do before and during pregnancy to reduce the risk of having a baby with a birth defect. These include getting routine prenatal check-ups; taking folic acid supplements before and during pregnancy; avoiding tobacco, alcohol and other substances; eating a healthy diet; getting appropriate levels of exercise; preventing exposure to chemicals; and managing existing medical conditions (diabetes, epilepsy, and high blood pressure). Specific recommendations for having a healthy pregnancy and improving birth outcomes are included throughout this data book. To learn more, visit **www.health.ri.gov/for/pregnantwomen** and **www.health.ri.gov/for/womenplanningpregnancy**

The RIBDP initiated local case-control studies to support national birth defects research, which found associations between risk factors and certain birth defects. In Rhode Island, women who were obese before pregnancy were linked with birth outcomes related to conotruncal heart defects. Also, women who smoked during pregnancy were at-risk for birth outcomes resulting in pulmonary stenosis and clubfoot. RIBDP is working on a study that examines the relationship between diabetes (Type II and gestational) and birth defects.

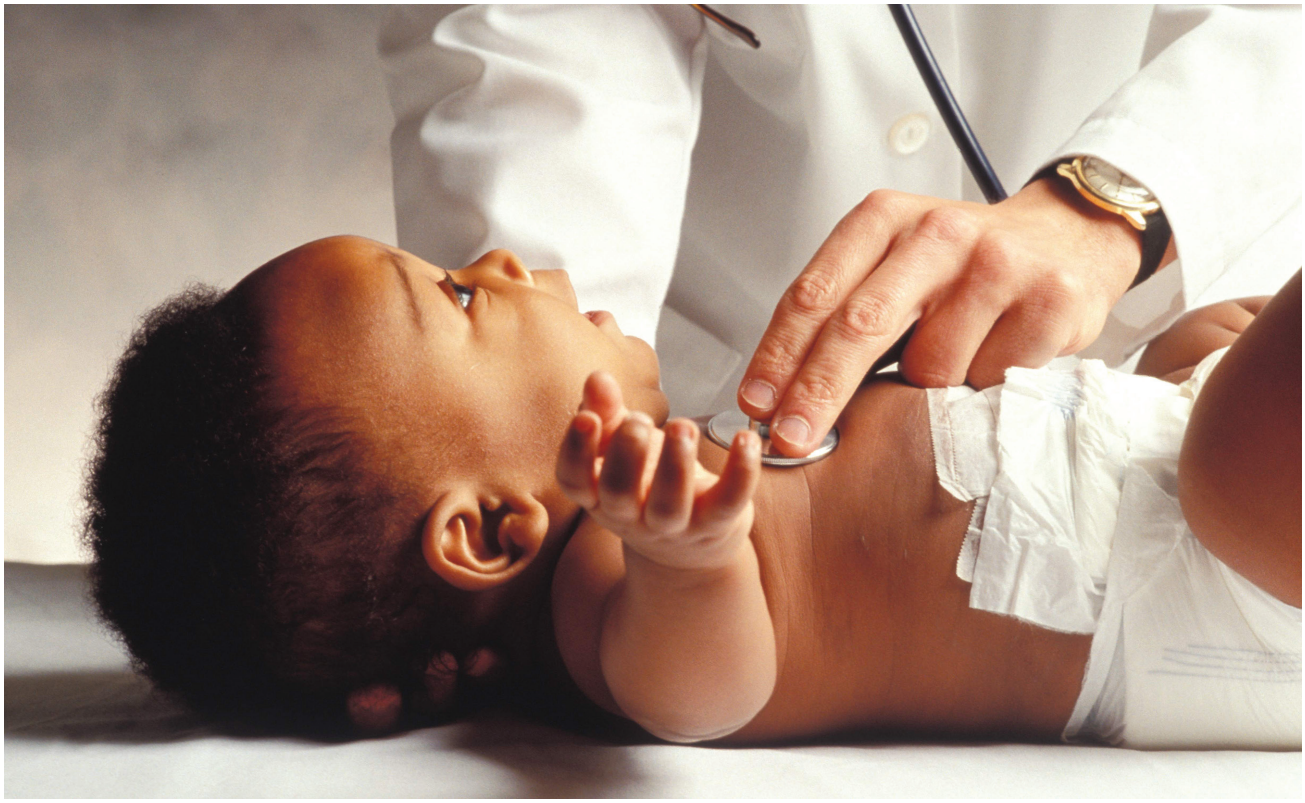
The RIBDP continues to work with various public health programs on birth defects awareness and risk-reduction activities. For example, in collaboration with the Family Planning Program, the RIBDP has funded the purchase and distribution of free multivitamins that contain folic acid to uninsured women who receive a negative pregnancy test at a family planning clinic. Uninsured women with positive pregnancy tests are enrolled in the state's Medicaid managed-care program, RIte Care, and receive prenatal vitamins at their first prenatal visit. Funds awarded by the Centers for Disease Control and Prevention (CDC) funded 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 comprised of families of children with birth defects. Community organizations and agencies that serve children with special needs are invited to share their materials before and after the grand rounds. In addition to the extended grand rounds, additional grand rounds are dedicated to topics related to birth defects and are held in January. Topics have included spina bifida, fetal alcohol syndrome, Down syndrome, hearing loss, craniofacial anomalies, and gastroschisis.



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 also works in partnership with its Advisory Council, which includes representatives from Women & Infants Hospital, Hasbro Children's Hospital, the Rhode Island Chapter of the March of Dimes, Rhode Island Parent Information Network, and Rhode Island KIDSCOUNT.

APPENDIX 1: RHODE ISLAND BIRTH DEFECTS PROGRAM CASE DEFINITION

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

Case Status

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

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

Exclusionary Criteria

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

Ear pit
Pulmonic stenosis
Laryngomalacia/tracheomalacia
Ankyloglossia
Pyloric stenosis
Embryonic cyst
Imperforate hymen
Cryptochidism (diagnosed less than a year after birth)
Retractile testes
Hydronephrosis (except postnatal diagnosis)
Flat foot
Hip dysplasia, hip laxity
Port wine stain/hemangioma
Mongolian spot
Birthmark
Accessory nipple
Patent ductus arteriosus with a gestational age of less than 36 weeks that is not coupled with another birth defect, or a case with a gestational age of less than 36 weeks that received prostaglandins

Reporting

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

APPENDIX 2: RESOURCES

RHODE ISLAND:

March of Dimes Rhode Island:
http://www.marchofdimes.org/rhodeisland
The March of Dimes helps women have full-term pregnancies and researches the problems that threaten the health of babies. This is done through community services, education, and advocacy.

Rhode Island Parent Information Network (RIPIN) and Family Voices of Rhode Island:
http://www.ripin.org
Provides information, support, and training to help all Rhode Islanders become their own best advocate at school, in healthcare, and in all areas of life.

Down Syndrome Society of Rhode Island:
http://www.dssri.org
A parent support organization dedicated to promoting the rights, dignity, and potential of all individuals with Down Syndrome through advocacy, education, public awareness, and support.

Rhode Island Early Intervention Program (EI):
http://www.eohhs.ri.gov/consumer/familieswithchildren/earlyintervention.aspx
Promotes the growth and development of infants and toddlers who have a developmental disability or delay in one or more areas. Children referred to EI receive a comprehensive developmental evaluation to determine if they are eligible.

Genetic Counseling & Medical Genetics Services

Prenatal Diagnostic Center
Women & Infants Hospital
101 Plain Street, 6th Floor
Providence, RI 02903
Phone: 401-453-7510
Fax: 401-453-7517
Offers screening, diagnostic and genetics counseling during pregnancy

Genetic Counseling Center
Hasbro Children's Hospital/Division of Human Genetics
2 Dudley Street, Suite 460
Providence, RI 02903
Phone: 401-444-8361
Fax: 401-444-3288
Provides genetics counseling and diagnostic services for children, adults, and families with histories of birth defects or genetic disorders.

Children's Neurodevelopment Center
http://www.lifespan.org/centers-services/childrens-neurodevelopment-center
Hasbro Children's Hospital
335R Prairie Avenue, Suite 2B
Providence, RI 02905
Phone: 401-444-5685
Fax: 401-444-6115
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:
Women & Infants Hospital
70 Elm Street, 2nd Floor
Providence, RI 02903
Phone: 888-AFP-MOMS
Provides prenatal AFP analysis

NATIONAL:

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

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.

Birth Defect Research for Children, Inc.(BDRC)
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.

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 congenital anomalies, 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 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 Information Center for Children and Youth with Disabilities (NICCYD)
http://www.nichcy.org
Provides information on disabilities in children and youth; programs and services for infants, children, and youth with disabilities; Individuals with Disabilities Education Act (IDEA), the nation's special education laws; 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
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
The NSGC works to promote the genetics counseling profession as a recognized and integral part of healthcare delivery, education, research, and public policy.

Organization of Teratology Information Services (OTIS)
http://www.mothertobaby.org
OTIS is a non-profit organization made up of individual services (TIS) throughout North America. It is dedicated to providing accurate, evidence-based, clinical information to patients and healthcare professionals about exposures during pregnancy and lactation.

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

Spina Bifida Association
http://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.

Zika Virus Information
https://www.cdc.gov/zika/pregnancy/protect-yourself.html
CDC website with resources for education and prevention from Zika infection, as well as how to follow up with your doctor. Additional information on Zika virus and pregnancy, pregnancy outcomes, birth defects and conditions related to Zika can be found at *https://www.cdc.gov/zika/pregnancy/index.html*

INTERNATIONAL:

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.
Congenital posterior urethral valves	An abnormal congenital obstructing membrane that i slocated within the posterior male urethra; this valve is the most common cause of bladder outlet obstruction in male children.
Choanal atresia	A narrowing or blockage of the nasal airway by tissue. This causes difficulty breathing.
Cleft lip with and without cleft palate	When the lip does not completely develop. Sometimes, it extends into the palate (roof of the mouth).
Cleft palate without cleft lip	A partial or complete split in the palate (roof of the mouth) that happens without a split in the lip.
Club foot	Babies born with this condition have their foot turned to the side. It may even appear that the top of the foot is where the bottom should be. The involved foot, calf, and leg are smaller and shorter than 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.
Craniosynostosis	Premature closure of one or several connective tissue membranes that separate the bones of the developing skull.
Diaphragmatic hernia	The absence or a defect of the membrane between the chest cavity and the abdomen. This lets organs such as the intestines protrude into the chest. It also interferes with the development of the heart and lungs.

Down syndrome	A disorder caused by the presence of an extra 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.
Esophageal atresia / tracheoesophageal fistula	A condition in which the esophagus ends in a blind pouch and fails to connect with the stomach. Tracheoesophageal fistula is an abnormal communication between the esophagus and the trachea.
Fetal alcohol syndrome	The sum total of the damage done to the child before birth as a result of the mother drinking alcohol during pregnancy. This condition always involves brain damage, impaired growth, and head and face abnormalities.
Gastroschisis	When an infant's intestines stick out of the body through a defect on one side of the umbilical cord.
Genetic	Having to do with genes, heredity, and variation in living things.
Hirschsprung's disease	A blockage in the large intestine due to a lack of nerves in part of the bowel. This condition causes the bowel and abdomen (belly) to become swollen.
Holoprosencephaly	Structural brain anomaly that results from incomplete cleavage of the prosencephalon
Hydrocephalus	A buildup of fluid inside the skull that 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 and Epispadias	Abnormal development of the tube carrying urine from the bladder to the outside of the body (urethra); the urinary opening is misplaced on the upper surface of the penis or where the urethra opens into the vagina
Infant	A child up to 1 year (12 months) of age.
Microcephaly	A condition where the baby's head is much smaller than expected.
Microphthalmia	Smallness of the eye.
Microtia	A small, abnormally shaped external ear. It can occur on one side only (unilateral) or on both sides (bilateral).
Mortality rate	Number of deaths in a year in a given population.
Obstructive genitourinary defect	A narrowing or absence of a normal opening in the urinary tract that blocks the flow of urine at any place in the urinary tract,from the kidney to the urethra.
Omphalocele	When an infant's intestines or other organs stick out of their abdominal cavity 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.
Triscupid 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.

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