



RHODE ISLAND BIRTH DEFECTS PROGRAM

BIRTH DEFECTS DATA BOOK 2010

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 of birth defects include genetics (inherited factors), environmental pollutants, occupational hazards, diet, medications, and personal behaviors.

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

## WHY STUDY BIRTH DEFECTS?

Each year in the United States, 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 do survive beyond the first year experience childhood illness and disability.

Early recognition and response to birth defects often prevents more serious health consequences. 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.

Approximately 20% of all infant deaths in the United States are due to birth defects. In Rhode Island, this proportion is lower. Among the 71 infants who died in Rhode Island during 2008, 11 (15.5%) of the deaths were related to a birth defect. This proportion represents a decrease from 2004, when 16 (23.5%) of the 68 deaths among Rhode Island infants were associated with a birth defect. During 2004-2008, the birth defects-related infant mortality rate decreased by 27%, from 125 deaths per 100,000 births in 2004 to 91 per 100,000 in 2008.

Many infants who are born prior to 37 weeks gestation, or preterm, also have birth defects. A study conducted by the National Birth Defects Prevention Network showed that birth defects were more than twice as common among preterm births compared with term births, and that 8% of preterm infants had a birth defect.\* In Rhode



Island, 93 (6.9%) of the 1,339 babies born preterm in 2008 had a birth defect. Preterm births are also the leading cause of infant deaths in Rhode Island. In 2008, 19 (26.8%) of the 71 infant deaths were due to prematurity.

\* Source: Honein, MA, Kirby, RS, Meyer, RE, et al. (2009). Association between birth defects and preterm birth. *Maternal Child Health J* 13:164–175. doi: 10.1007/s10995-008-0348-y





## RHODE ISLAND BIRTH DEFECTS PROGRAM: A PROGRESS REPORT

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 Center for Health Data and Analysis. The RIBDP was created to identify newborns with birth defects, ensure that they 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 (RI General Law 23-13.3) requiring the development and implementation of a birth defects reporting, surveillance, and information system. This system will describe the occurrence of birth defects in children up to age five, detect trends of morbidity and mortality, and help ensure that 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 the Department on the establishment and implementation of the system and recommend a list of birth defects to be reported to the surveillance system. It is critical that state agencies, healthcare services providers, community organizations, parents, and other key stakeholders provide input not only for the development of the surveillance system, but also for issues regarding information dissemination and analysis.

## 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 Care New England to collect discharge audits from Women and Infants Hospital and Kent County Hospital, which represent about 80% of all birth defects cases in the state. The RIBDP also collects information from specialty clinics, such as the Child Neurodevelopment Center 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.

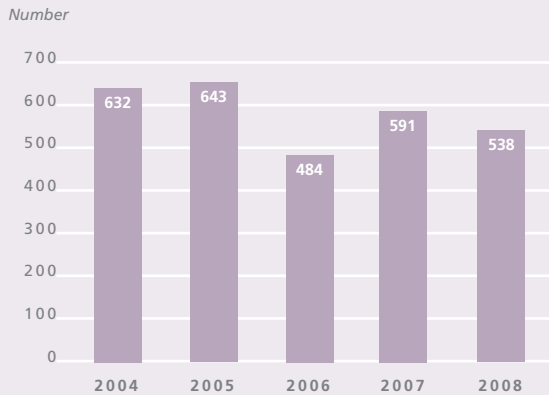
Birth defects cases are identified using diagnoses coded by the 9th Clinical Modification of the International Classification of Diseases (ICD 9-CM) and include 740-759.9 and 760.71 codes. Accuracy of birth defects diagnoses is confirmed through chart review of birth defects cases at Women and Infants and Kent County hospitals. 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.

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

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. Prenatal case finding expands birth defects ascertainment and improves the prevalence estimate of certain birth defects by detecting cases not found at the time of newborn discharge. Currently, prenatal case data are provisional and are not included in the Rhode Island birth defects prevalence reporting. The RIBDP plans to establish routine prenatal surveillance with existing and future prenatal data partners to better understand birth defects prevalence and trends in Rhode Island.

Figures 1 and 2 (on the following page) show the overall count and prevalence of birth defects in Rhode Island over the five-year period from 2004 to 2008. During this period, the rate of birth defects in Rhode Island decreased by 10.7%, from 516 per 10,000 live births in 2004 to 461 per 10,000 in 2008. A three year moving average indicates a 6.2% decrease in overall birth defects prevalence from 2004 to 2008. It should be noted that the low birth defects rate in 2006 (405 per 10,000) is an aberration, as there were no changes in ascertainment methodologies or in the trends of other birth outcomes, such as low birth weight, preterm births, and infant mortality.

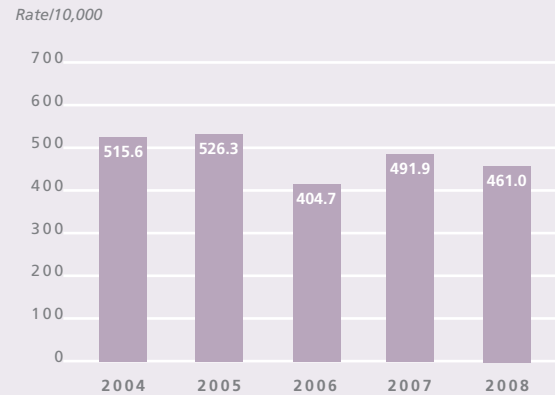
**Figure 1**  
**COUNT OF NEWBORNS WITH BIRTH DEFECTS\***  
**RHODE ISLAND, 2004–2008**



\*ICD-9-CM Codes: 740.0-759.9

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

**Figure 2**  
**PREVALENCE OF BIRTH DEFECTS AMONG**  
**NEWBORNS, RHODE ISLAND, 2004–2008**



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

Table 1 (on the following page) shows the number and prevalence of selected birth defects among Rhode Island residents during 2004-2008, organized by organ system. Cardiovascular defects (those relating to the heart) are the most common type of defects (192.3 per 10,000 live births). Ventricular septal defects and pulmonary valve atresia and stenosis represent the highest proportion of these cases. Medical record reviews conducted during 2008-2009 indicate that atrial septal defects (ASD) share an ICD-9 code (745.5) with patent foramen ovale (PFO), a condition which is not considered a birth defect. Since these two conditions were not distinguished during earlier chart reviews, the prevalence of atrial septal defects may be overestimated for the five-year period.

Other common birth defects in Rhode Island include those related to musculoskeletal (140.9 per 10,000) and genitourinary (138.4) systems. Among these groups of defects, hypospadias and epispadias (35.6 per 10,000) and obstructive genitourinary defect (26.8) are most frequent, but rates for these conditions have remained stable.



Table 1

**SELECTED BIRTH DEFECTS, RHODE ISLAND, 2004–2008**

BIRTH DEFECT	NUMBER	RATE PER 10,000 LIVE BIRTHS
<b><i>Central Nervous System</i></b>	<b>112</b>	<b>18.6</b>
SPINA BIFIDA	14	2.3
ANENCEPHALY	1	0.2
ENCEPHALOCELE	3	0.5
HYDROCEPHALY	28	4.7
<b><i>Eye, Ear, Face, Neck</i></b>	<b>155</b>	<b>22.5</b>
CONGENITAL CATARACT	2	0.3
ANOPHTHALMOS AND MICROPTHALMUS	3	0.5
ANOTIA/MICROTIA	5	0.8
<b><i>Cardiovascular</i></b>	<b>1156</b>	<b>192.3</b>
TRANSPOSITION OF GREAT VESSELS	13	2.2
TETRALOGY OF FALLOT	25	4.2
VENTRICULAR SEPTAL DEFECT	213	35.4
ATRIAL SEPTAL DEFECT	215	35.8
ATRIOVENTRICULAR SEPTAL DEFECT	9	1.5
PULMONARY VALVE ATRESIA/STENOSIS	78	13.0
TRICUSPID VALVE ATRESIA	5	0.8
AORTIC VALVE STENOSIS	0	0.0
HYPOPLASTIC LEFT HEART SYNDROME	5	0.8
COARCTATION OF AORTA	10	1.7
<b><i>Respiratory</i></b>	<b>95</b>	<b>15.8</b>
CHOANAL ATRESIA	4	0.7
<b><i>Orofacial</i></b>	<b>83</b>	<b>13.8</b>
CLEFT LIP WITH AND WITHOUT CLEFT PALATE	45	7.5
CLEFT PALATE	38	6.3
<b><i>Gastrointestinal</i></b>	<b>252</b>	<b>41.9</b>
ESOPHAGEAL ATRESIA/TRACHEOSOPHAGEAL FISTULA	14	2.3
RECTAL AND LARGE INTESTINAL ATRESIA/STENOSIS	21	3.5
HIRSHSPRUNG'S DISEASE	5	0.8
<b><i>Genitourinary</i></b>	<b>832</b>	<b>138.4</b>
HYPOSPADIAS AND EPISPADIAS	214	35.6
RENAL AGENESIS/HYPOPLASIA	11	1.8
OBSTRUCTIVE GENITOURINARY DEFECT	161	26.8
<b><i>Musculoskeletal</i></b>	<b>847</b>	<b>140.9</b>
CLUB FOOT	64	10.6
REDUCTION DEFORMITY, UPPER LIMBS	16	2.7
REDUCTION DEFORMITY, LOWER LIMBS	7	1.2
DIAPHRAGMATIC HERNIA	15	2.5
GASTROSCHISIS	34	5.7
OMPHALOCELE	7	1.2
<b><i>Chromosomal</i></b>	<b>152</b>	<b>25.3</b>
DOWN SYNDROME	79	13.1
PATAU SYNDROME	5	0.8
EDWARD SYNDROME	12	2.0
<b><i>All Birth Defects</i></b>	<b>3651</b>	<b>607.3</b>
<b><i>All Birth Defect Cases</i></b>	<b>2888</b>	<b>480.4</b>

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

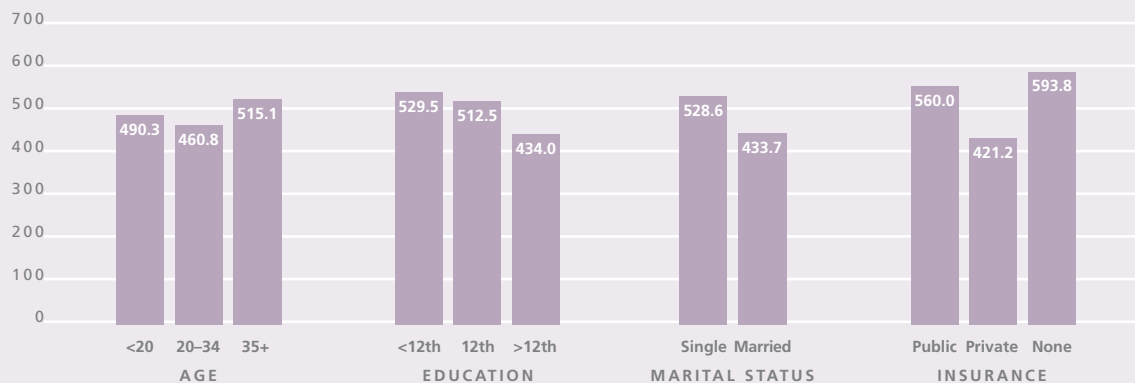
\* Numbers and rates of body system represent total diagnosed birth defects associated with that body system

Figure 3

## BIRTH DEFECTS PREVALENCE BY SELECTED MATERNAL CHARACTERISTICS

RHODE ISLAND, 2004–2008

Rate/10,000



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.*

## MATERNAL CHARACTERISTICS

Babies born to older women (aged 35 or older), women with less than a 12th grade education, single women, and women with publicly funded health insurance or no health insurance are at a higher risk for birth defects (Figure 3). During 2004–2008, the birth defects prevalence rate among women aged 35 or older was 515 (per 10,000 live births), compared to 461 among women aged 20–34. The birth defects rate among women with less than a 12th grade education (530) or with a 12th grade education (513) was higher than the rate among women with higher education levels (434). Similarly, single women (529) were more likely to have a baby with a birth defect than married women (434). Women insured through public programs, such as RIte Care and Medicaid (560), were also more likely to have a baby born with a birth defect than married women insured through commercial or private providers, such as Blue Cross or United Healthcare (421). It should be noted that although a small number of Rhode Island women did not have health insurance at the time of delivery (approximately 1%), the risk for having a baby with a birth defect was highest among this population (594) compared to the population who had private insurance (421).

## RACE AND ETHNICITY AND GEOGRAPHIC DISPARITIES

Birth defects prevalence varies by race and ethnicity (Figure 4) and geographical residence (Figure 5). During 2004-2008, the average birth defects prevalence rate among Blacks and African Americans (632 per 10,000 live births) and Hispanics (620) was 1.3-1.4 times the rate for Whites (467). During this period, Asian women (398) were least likely to have a baby born with a birth defect.

Babies born to residents of core cities where the child poverty level is greater than 15% (Central Falls, Newport, Pawtucket, Providence, West Warwick, and Woonsocket) were about 1.2 times more likely to have a birth defect than those born to residents living in the rest of the state. Compared to the other core cities, the birth defects rate in West Warwick was the lowest (363) and was below the statewide rate (480). Pawtucket (650) and Central Falls (633) had 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.

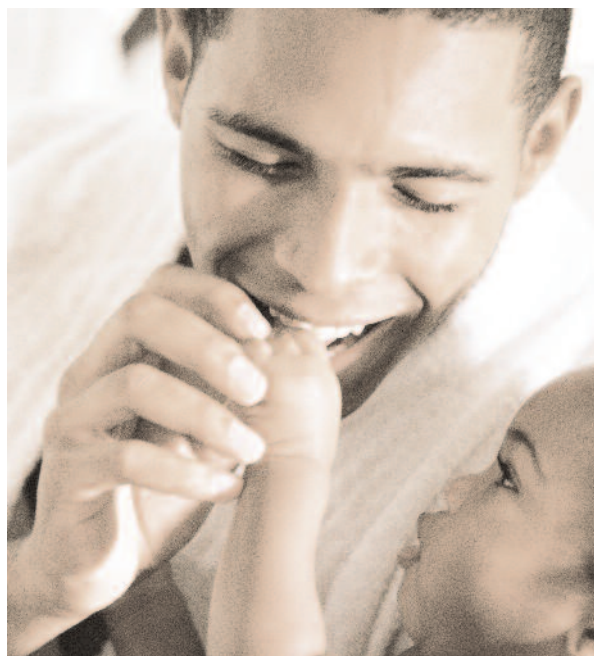
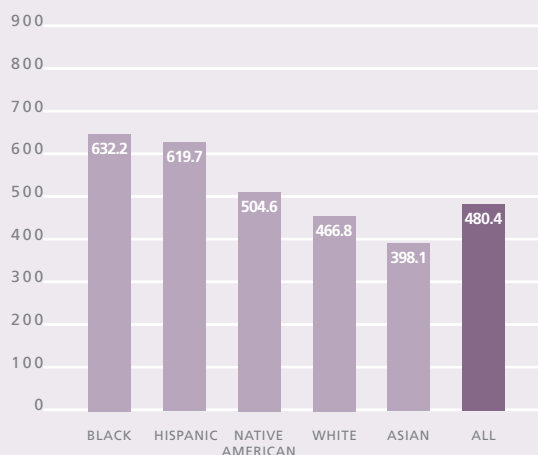


Figure 4

### BIRTH DEFECTS PREVALENCE BY RACE/ETHNICITY RHODE ISLAND, 2004-2008

Rate/10,000

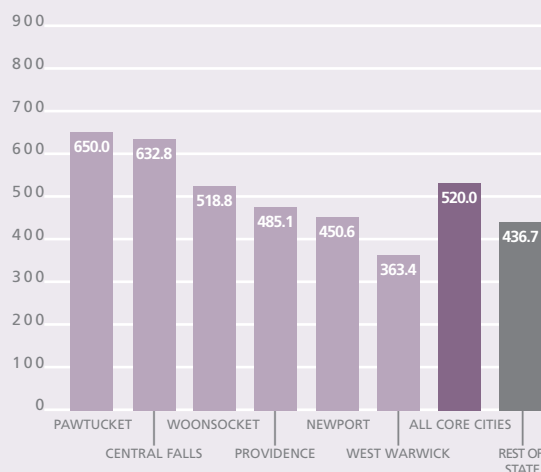


Source: Rhode Island Birth Defects Program, Rhode Island Department of Health

Figure 5

### BIRTH DEFECTS PREVALENCE IN THE CORE CITIES, RHODE ISLAND, 2004-2008

Rate/10,000



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



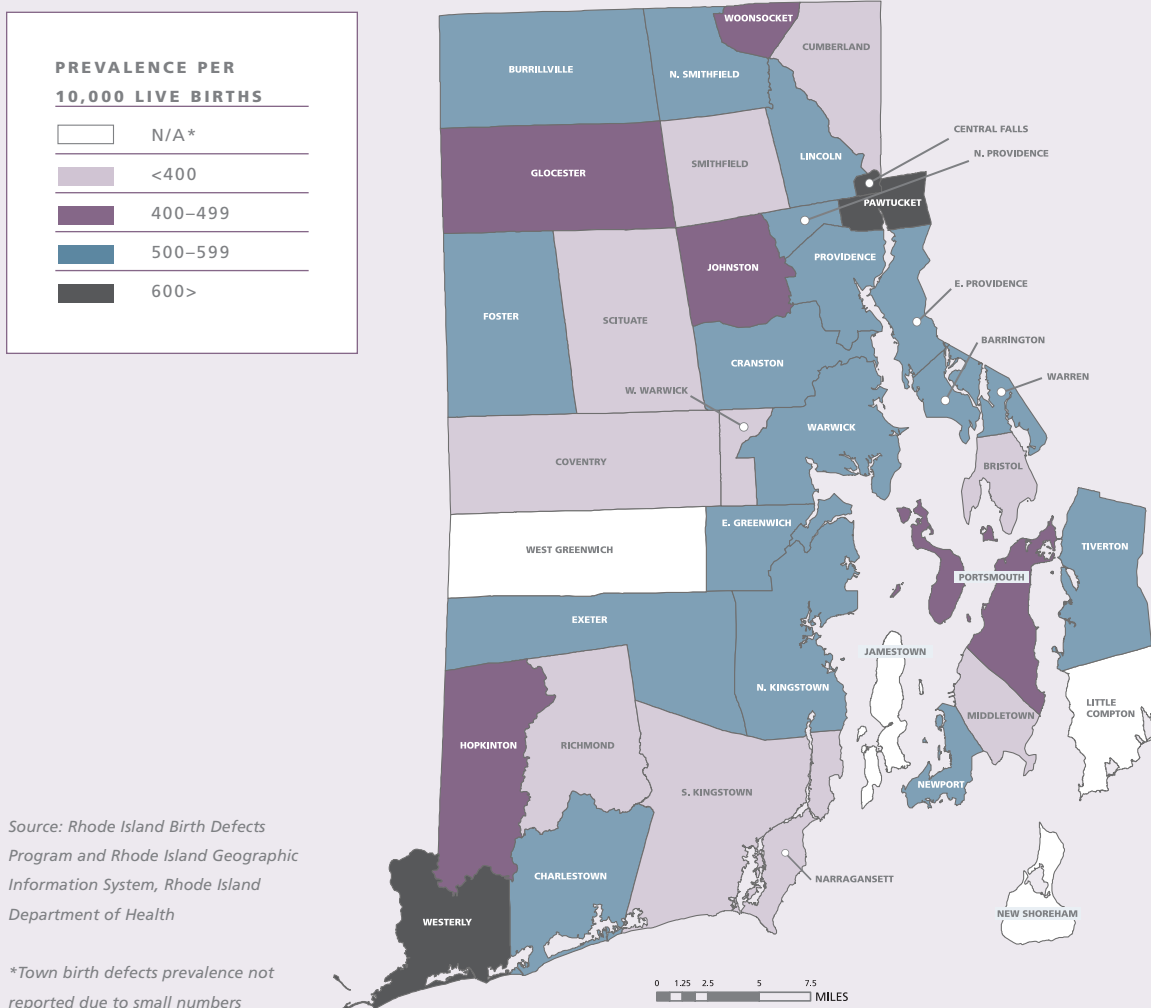
## MAPPING RATES OF BIRTH DEFECTS

Geospatial analysis allows us to follow trends and detect clusters of birth defects in Rhode Island. The following map illustrates birth defects prevalence rates during 2004-2008 by city/town in Rhode Island. To address sample size, 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 for the 2004-2008 period are not reported. This includes the towns of Jamestown, West Greenwich, Little Compton, and New Shoreham.

The map shows a majority of cities/towns with a prevalence rate between 400 and 600 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. 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 6

### BIRTH DEFECTS PREVALENCE BY CITIES/TOWNS, RHODE ISLAND, 2004-2008



## SENTINEL CONDITIONS

The Birth Defects Advisory Council identified a set of conditions (Table 2) for targeted outreach and follow-up to help ensure that families of children with these conditions are provided with appropriate services, referrals, information, and resources. The selection was based on criteria that included number of children affected, timeliness of identification, condition severity, service availability, resource intensity, recurrence risk, and availability of prevention strategies.

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

Table 2

**RHODE ISLAND BIRTH DEFECTS PROGRAM, SENTINEL CONDITIONS LIST**

CONDITION	ICD-9-CM CODE
NEURAL TUBE DEFECTS	
» ANENCEPHALY	740.0-740.1
» SPINA BIFIDA	741.0-741.9
EYE	
» ANOPHTHALMIA/MICROPTALMIA	743.0, 743.1
» CONGENITAL CATARACT	743.30-743.34
» ANIRIDIA	743.45
CONGENITAL HEART DISEASE	745-747
CLEFT LIP/PALATE	749
ESOPHAGEAL ATRESIA/STENOSIS	750.3
ATRESIA/STENOSIS OF LARGE INTESTINE, RECTUM, AND ANAL CANAL	751.2
GENITAL ANOMALIES	752
RENAL ANOMALIES	753.0-753.1
REDUCTION DEFORMITIES OF LIMBS	755.20-755.39
CONGENITAL DIAPHRAGMATIC HERNIA	756.6
GASTROSCHISIS/OMPHALOCELE	756.79
CHROMOSOMAL ANOMALIES	758
EAR	
» ANOTIA/MICROTIA	744.01, 744.23
» HEARING LOSS	(referral via RI Hearing Assessment Program)
LUNG	
» CONGENITAL CYSTIC LUNG	748.4
» AGENESIS, HYPOPLASIA, AND DYSPLASIA OF LUNG	748.5
CONGENITAL TUMORS	140-239
<b>DEVELOPMENTAL CONDITIONS</b>	<b>ICD-9-CM CODE</b>
AUTISM	299.0

## SERVICE ASSESSMENT AND ASSURANCE

A priority goal of the RIDBP is to assure that children with birth defects receive appropriate and timely preventive, specialty, and other healthcare services. The RIDBP, in collaboration with the Rhode Island Parent Information Network (RIPIN), employs a Family Resource Specialist (parent consultant) who interviews parents of children with specific conditions to determine whether their children have received appropriate referrals and services on a timely basis. The Family Resource Specialist meets with parents at pediatric practices that serve a high volume of children with birth defects, such as the Children's Neurodevelopment Center (CNDC) at Hasbro Children's Hospital. The RIDBP will also work with RIPIN to mail service assessment forms to additional parents of children with birth defects and conduct telephone follow-up with non-respondents. The service assessment form was designed to determine what services and referrals were provided to children and at what age they were provided, based on national guidelines for specific conditions. Specifically, the assessment asks about services such as pediatric visits, nutritional support, clinical screenings, chromosomal tests, and genetic counseling, as well as about referrals to developmental and educational services and to family support groups. Currently, this service assurance process is conducted among families of children with Down syndrome, cleft lip/palate, and clubfoot. The RIDBP plans to expand this service assurance process to all sentinel birth defects in the near future.



## ECONOMIC COSTS OF BIRTH DEFECTS

In addition to the emotional impact that families of children with birth defects often experience, birth defects also have financial implications for families, the healthcare system, and society. Therefore, it is important to understand the economic burden of birth defects in order to help drive prevention activities and policy decisions.

The RIBDP has begun to study 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 reports the total unadjusted burden of cost for all newborn hospital admissions with at least one diagnosed birth defect and the total unadjusted burden of cost by selected birth defects.

During 2004-2008, 2,723 newborns were identified with at least one birth defect via Rhode Island's hospital discharge database. The total hospital cost for these children was approximately \$140 million, with an average cost per child of \$51,380. In 2008, the average hospital cost per child with a birth defect (\$58,672) was more than seven times the cost for a child with no birth defects (\$7,876).

Unadjusted hospital costs do not account for the differences of expenses incurred among differing maternity hospitals and service delivery plans. The RIBDP will begin exploring analyses based on adjustments to hospital costs. The table below compares the number of cases, lengths of stay, and mean hospital charges between the United States and Rhode Island for selected birth defects.

Table 3

**MEAN TOTAL HOSPITAL CHARGES FOR NEWBORNS BY SELECTED BIRTH DEFECTS  
RHODE ISLAND, 2004-2008**

BIRTH DEFECT	NUMBER OF US CASES	LENGTH OF STAY (DAYS)	US 2003* MEAN (\$)	NUMBER OF RI CASES	LENGTH OF STAY (DAYS)	RI 2004-08** MEAN (\$)
SPINA BIFIDA	1,136	15	65,942	9	31	108,614
MICROCEPHALY	–		–	12	12	54,294
HYDROCEPHALY	–		–	30	33	185,521
CLEFT LIP WITH AND WITHOUT CLEFT PALATE	3,486	6	15,397	44	5	16,778
CLEFT PALATE	2,187	10	33,387	38	13	56,834
RECTAL & LARGE INTESTINAL ATRESIA/STENOSIS	1,604	17	75,220	19	17	84,533
CLUBFOOT	–		–	61	10	57,535
GASTROSCHISIS	1,419	41	155,620	36	34	216,909
DOWN SYNDROME	5,036	11	38,745	69	8	38,624
EDWARD SYNDROME	576	10	39,547	9	8	59,655

Sources:

\*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.

\*\*Rhode Island Hospital Discharge Database, Rhode Island Department of Health



## REDUCING THE RISKS OF BIRTH DEFECTS

Although not all causes of birth defects are known, there are a number of steps a woman can take before and during pregnancy to reduce the risk of having a baby with a birth defect. These include getting routine prenatal check-ups, taking folic acid before and during pregnancy, avoiding tobacco, alcohol, and other substances, eating a healthy diet, getting appropriate levels of exercise, preventing exposure to chemicals, and managing existing medical conditions (e.g. diabetes, epilepsy, and high blood pressure). Specific tips on having a healthy pregnancy and improving birth outcomes are included throughout this data book.

The RIBDP is currently working with various public health programs on birth defects awareness and risk reduction activities. For example, in collaboration with the Women's Health Screening and Referral Program, the RIBDP will finance the purchase and distribution of free multivitamins with folic acid to uninsured women who receive negative pregnancy tests at family planning clinics. Uninsured women with positive pregnancy tests are enrolled in the state's Medicaid managed care program, Rite Care, and will receive prenatal vitamins at their first prenatal visit.

The RIBDP is also collaborating with other public health programs, including the Initiative for a Healthy Weight, Tobacco Control Program, Diabetes Prevention and Control Program, First Connections (Home Visiting) Program, and Office of Minority Health to disseminate birth defects prevention messages to at-risk populations.

## INFORMATION FOR EDUCATION AND DECISION MAKING

Sharing data and information on birth defects with healthcare providers, policy makers, community organizations, families, and other stakeholders is the means to increasing awareness of birth defects and can lead to program enhancements and policy development. The RIBDP uses a multi-pronged approach to data dissemination, including posting information on the RIBDP web page ([www.health.ri.gov/family/birthdefects](http://www.health.ri.gov/family/birthdefects)), sponsoring pediatric grand rounds, presenting information at state, local, and national meetings, and publishing printed materials.

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 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.

Past topics presented at these grand rounds have included spina bifida, fetal alcohol syndrome, and Down syndrome. During January 2010, in addition to the extended grand rounds focused on hearing loss, three additional grand rounds related to birth defects were held throughout the month. The topics were genetic treatment using RNA interference, the genetics of autism, and craniofacial anomalies.

## COLLABORATIVE RESEARCH: NATIONAL AND INTERNATIONAL STUDIES

The RIBDP has participated in, or is currently participating in, a variety of national and international collaborative studies to gain a better understanding of specific birth defects. Examples of these collaborative research studies, their organizational sponsors, and their purposes are described in Table 4. 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.

Table 4

**COLLABORATIVE RESEARCH STUDIES**

STUDY/PROJECT	SPONSOR	PURPOSE/RESEARCH QUESTIONS
International Database on Craniofacial Anomalies (IDCFA)	World Health Organization (WHO)	To collect and disseminate data on craniofacial anomalies (oral clefts) to stimulate research for the development of prevention strategies and a better understanding of the characteristics associated with these anomalies
Preterm Births and Birth Defects	National Birth Defects Prevention Network (NBDPN)	To determine what percentage of preterm and low birth weight births have birth defects and whether there is an increased rate of preterm delivery for infants born with birth defects
Neural Tube Defects and Infant Mortality	National Birth Defects Prevention Network (NBDPN)	To examine the impact of folic acid fortification on neural tube defect-specific infant mortality, whether a decline in neural tube defect-specific infant mortality has contributed to the decline in overall infant mortality, and if the severity of neural tube defects has changed over time
Gastroschisis/Ventral Wall Defects	National Birth Defects Prevention Network (NBDPN)	To determine the prevalence and trends of specific ventral wall defects in the United States
National Prevalence Estimates	National Birth Defects Prevention Network (NBDPN)	To monitor trends and determine differences between states and regions
Clubfoot	National Birth Defects Prevention Network (NBDPN)	To estimate the prevalence of club foot and investigate its risk factors

# RESOURCES

## RHODE ISLAND:

**March of Dimes Rhode Island:** [www.marchofdimes.com/rhodeisland](http://www.marchofdimes.com/rhodeisland)

The mission of the March of Dimes is to improve the health of babies by preventing birth defects and infant mortality through research, community services, education, and advocacy.

**Rhode Island Parent Information Network (RIPIN) and Family Voices of Rhode Island:** [www.ripin.org](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:** [www.dssri.org](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:** [www.dhs.ri.gov/ChildrenwithSpecialNeeds/EarlyInterventionProgram](http://www.dhs.ri.gov/ChildrenwithSpecialNeeds/EarlyInterventionProgram)

Provides developmental testing and support services for families with children younger than the age of three

**Rhode Island Healthy Mothers, Healthy Babies Coalition:** [www.hmhb.org](http://www.hmhb.org)

A partnership of individuals, 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

### ***Greystone / Radiologic Associates***

Obstetrician Geneticist: Kathryn McGowan, MD, MS  
235 Plain Street, Providence, RI 02905  
Phone: 401-272-8510  
Fax: 401-272-0315  
Provides genetic counseling, prenatal testing and diagnosis, and pre-conception evaluations

### ***Genetic Counseling Center***

Director: Dianne Abuelo, MD  
Rhode Island Hospital  
593 Eddy Street, Providence, RI 02903  
Phone: 401-444-8361  
Fax: 401-444-3288  
Provides genetics counseling and diagnostic services for children, adults, and families with histories of birth defects or genetic disorders

### ***Children's Neurodevelopment Center***

Hasbro Children's Hospital  
593 Eddy Street, Providence, RI 02903  
Phone: 401-444-5685  
Fax: 401-444-6115  
[www.lifespan.org/hch/Services/neuro\\_cntr/](http://www.lifespan.org/hch/Services/neuro_cntr/)  
Provides interdisciplinary, comprehensive care for children with developmental and learning problems

## Laboratories

### ***Genetics Laboratory - Division of Genetics***

Director: Umadevi Tantravahi, PhD  
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***

Director: Jacob Canick, PhD  
Women & Infants Hospital  
70 Elm Street, 2nd Floor, Providence, RI 02903  
Phone: 888-AFP-MOMS  
[www.womenandinfants.org](http://www.womenandinfants.org)  
Provides prenatal AFP analysis

## **NATIONAL:**

### **American Academy of Pediatrics: [www.aap.org](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. The website contains information regarding the Academy's many programs, activities, policy statements, practice guidelines, publications, and other child health resources.

### **American Academy of Family Physicians: [www.aafp.org](http://www.aafp.org)**

The American Academy of Family Physicians is a medical specialty society that represents more than 93,700 physicians and medical students.

### **Birth Defect Research for Children, Inc. (BDRC): [www.birthdefects.org](http://www.birthdefects.org)**

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

### **Centers for Disease Control & Prevention (CDC): Maternal and Infant Health:**

**[www.cdc.gov/reproductivehealth/MaternalInfantHealth](http://www.cdc.gov/reproductivehealth/MaternalInfantHealth)**

To better understand the burden of maternal complications and mortality, and to decrease disparities among populations at risk of death and complications from pregnancy, the Division of Reproductive Health supports national and state-based surveillance systems to monitor trends and investigate health issues; conducts epidemiologic, behavioral, demographic, and health services research; and works with partners to translate research findings into healthcare practice, public health policy, and health promotion strategies.

### **CDC National Center on Birth Defects and Developmental Disabilities (NCBDDD): [www.cdc.gov/ncbddd/bd/default.htm](http://www.cdc.gov/ncbddd/bd/default.htm)**

NCBDDD is dedicated to helping people live to the fullest. Much of the work of NCBDDD focuses on protecting people who are especially vulnerable to health risks – babies, children, people with blood disorders, and people with disabilities.

### **CDC Office of Genetics and Disease Prevention: [www.cdc.gov/genomics](http://www.cdc.gov/genomics)**

The CDC Office of Genetics and Disease Prevention uses genomic knowledge to improve the lives and health of all people by integrating genomics into public health research, policy, and programs.

### **National Birth Defects Prevention Network (NBDPN): [www.nbdpn.org](http://www.nbdpn.org)**

NBDPN is 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 Center for Biotechnology Information (NCBI): [www.ncbi.nlm.nih.gov/sites/GeneTests?db=GeneTests](http://www.ncbi.nlm.nih.gov/sites/GeneTests?db=GeneTests)**

NCBI advances science and health by providing access to biomedical and genomic information. By providing current, authoritative information on genetic testing and its use in diagnosis, management, and genetic counseling, NCBI promotes the appropriate use of genetic services in patient care and personal decision-making.

### **National Healthy Mothers, Healthy Babies Coalition: [www.hmhb.org](http://www.hmhb.org)**

The National Healthy Mothers, Healthy Babies Coalition works to improve the health and safety of mothers, babies, and families through education and collaborative partnerships of public and private organizations.

### **National Information Center for Children and Youth with Disabilities (NICHCY)**

NICHCY provides information on disabilities in children and youth; programs and services for infants, children, and youth with disabilities; 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 (NOFAS): [www.nofas.org](http://www.nofas.org)**

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



**National Society of Genetic Counselors (NSGC): [www.nsgc.org](http://www.nsgc.org)**

The NSGC 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 Services (OTIS): [www.otispregnancy.org](http://www.otispregnancy.org)**

OTIS studies the effects that drugs, medications, chemicals, and other exposures may have on the fetus. It also provides resources for medical consultation on prenatal exposures and fact sheets about exposures that are known to cause birth defects.

**Share Your Story: [www.shareyourstory.org](http://www.shareyourstory.org)**

Share is an online community for parents of babies born prematurely or who have spent time in a neonatal intensive care unit (NICU). Brought to you by the March of Dimes, Share is intended to offer parents a safe place to talk about their experiences and gain support from each other.

**Smiles: [www.cleft.org](http://www.cleft.org)**

"SMILES" is 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 (SBA): [www.spinabifidaassociation.org](http://www.spinabifidaassociation.org)**

SBA promotes the prevention of spina bifida and enhances the lives of all affected.

**Teratology Society: [www.teratology.org](http://www.teratology.org)**

A scientific organization to study the causes and biological processes leading to abnormal development and birth defects

**INTERNATIONAL:****Newlife Foundation for Disabled Children: [www.newlifecharity.co.uk](http://www.newlifecharity.co.uk)**

Newlife gives practical support for disabled children throughout the UK, cares for the carers, funds medical research, creates awareness, and campaigns for change.

**International Clearinghouse for Birth Defects Surveillance and Research: [www.icbdsr.org](http://www.icbdsr.org)**

The mission of the International Clearinghouse for Birth Defects Surveillance and Research is to bring 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.

**International Birth Defects Information System (IBIS): [www.birth-defects.org](http://www.birth-defects.org)**

IBIS is a multi-lingual website dedicated to promoting better care and prevention of birth defects through information sharing.



# GLOSSARY

<b>Anencephalus</b>	A neural tube defect that is fatal and involves the absence of most of the brain and/or spinal cord
<b>Anophthalmia</b>	Congenital absence of the eyes
<b>Anotia</b>	Congenital absence of the ear
<b>Aortic Valve Stenosis</b>	A heart defect involving the aorta. It's a narrowing of the valve between the left ventricle (lower chamber) of the heart and the major vessel carrying blood from the heart. This condition can be repaired surgically in some cases.
<b>Atresia</b>	Absence or closure of a tubular structure
<b>Atrial Septal Defect</b>	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.
<b>Atrioventricular Defect</b>	A hole or malformation in the connective tissue that divides the right and left chambers of the heart, between either the ventricles (lower chambers) or the atria (upper chambers)
<b>Choanal Atresia</b>	An abnormal membranous formation that blocks the opening at the back of the nostrils, restricting breathing through the nose
<b>Cleft Lip with &amp; without Cleft Palate</b>	An incomplete development of the lip. Sometimes, it extends into the roof of the mouth (hard and soft palate).
<b>Cleft Palate without Cleft Lip</b>	A partial or complete split in the palate (roof of the mouth), occurring without a split in the lip
<b>Club Foot</b>	A condition where the foot is turned to the side. It may even appear that the top of the foot is where the bottom should be. The involved foot, calf, and leg are smaller and shorter than the normal side.
<b>Coarctation of Aorta</b>	The narrowing of the aorta, the main blood vessel carrying blood from the heart to the rest of the body. It causes abnormal pressure in the heart during contractions.
<b>Congenital Cataract</b>	A condition where the capsule or lens of the eye is obscured. This opacity might cause vision impairments or blindness.
<b>Congenital Disorder</b>	A medical condition that is present at birth but may be recognized before birth
<b>Diaphragmatic Hernia</b>	The absence of, or a defect of, the membrane between the chest cavity and the abdomen, allowing protrusion of organs such as the intestines into the chest and interfering with the development of the heart and lungs
<b>Down Syndrome</b>	Also called Trisomy 21, this disorder is caused by the presence of an extra 21st chromosome, causing developmental disability, distinctive physical features, and short stature.
<b>Encephalocele</b>	A gap or hole in the skull that usually causes a protrusion of brain tissue
<b>Esophageal Atresia/ Tracheoesophageal Fistula</b>	Abnormal closure or abnormal holes with the esophagus or trachea (windpipe), or unconnected upper and lower ends of the esophagus
<b>Fetal Alcohol Syndrome</b>	The sum total of the damage done to the child before birth as a result of the mother drinking alcohol during pregnancy. Fetal alcohol syndrome (FAS) always involves brain damage, impaired growth, and head and face abnormalities.
<b>Gastroschisis</b>	A protrusion of the digestive organs to the outside of the body through a hole in the abdominal wall
<b>Genetic</b>	Having to do with heredity and variation in organisms

<b>Hirschsprung's Disease</b>	Congenital enlarged colon due to absent nerves in the wall of the colon
<b>Hydrocephalus</b>	Accumulation of fluid within the spaces of the brain
<b>Hypoplastic Left Heart Syndrome</b>	An incomplete development of the left chambers of the heart and one of the most life-threatening heart defects
<b>Hypospadias and Epispadias</b>	The abnormal development of the tube carrying urine from the bladder to the outside of the body (urethra), causing the urinary opening to be misplaced on the upper surface of the penis, or where the urethra opens into the vagina
<b>Infant</b>	A child up to one year (12 months) of age
<b>Microphthalmia</b>	Congenital smallness of the eye
<b>Microtia</b>	A small, abnormally shaped, or absent external ear. It can occur on one side only (unilateral) or on both sides (bilateral).
<b>Mortality Rate</b>	Number of deaths in a year in a given population
<b>Obstructive Genitourinary Defect</b>	Stenosis or atresia of the urinary tract. This malformation clogs or narrows the passageways of the genital or urinary tracts (such as the urethra).
<b>Omphalocele</b>	Congenital opening of the abdominal wall with protrusion of the abdominal contents. Can be distinguished from gastroschisis by location within the umbilical ring.
<b>Pulmonary Valve Atresia &amp; Stenosis</b>	Abnormal closure, absence, or narrowing of the duct that opens into the pulmonary artery, the vessel that carries blood to the lungs
<b>Rectal &amp; Large Intestinal Atresia/Stenosis</b>	Abnormal closure, absence, or narrowing of the duct or passageway of the digestive tract in the region of the rectum or large intestine
<b>Reduction Deformity, Upper or Lower limbs</b>	Deformity of the arms or legs, in which one or both arms or legs are missing or shortened
<b>Renal Agenesis/Hypoplasia</b>	A defect where the kidney was formed incompletely or is absent
<b>Spina Bifida</b>	A defect in which the spinal neural tube closed imperfectly, so part of the spinal cord may protrude. This condition often results in neurological disorders.
<b>Stenosis</b>	Abnormal narrowing of a tubular structure
<b>Tetralogy of Fallot</b>	Complex malformation of the heart characterized by a hole in the wall between the ventricles, a misplacement of the origin of the aorta, a narrowing of the pulmonary artery, and the enlargement of the right ventricle
<b>Transposition of Great Arteries</b>	A defect in which the main blood vessels leading from the heart are reversed. The aorta arises from the right ventricle and the pulmonary artery arises from the left ventricle.
<b>Tricuspid Valve Atresia</b>	Congenital absence or closure of the three-segmented valve of the heart that normally keeps blood in the right ventricle from flowing backward into the right atrium
<b>Trisomy 13 (Patau)</b>	The condition of having three copies of chromosome 13, causing severe skull and facial deformation and developmental delays. Some cases include heart defects, brain defects, and cleft lip/palate.
<b>Trisomy 18 (Edwards)</b>	The condition of having three copies of chromosome 18, causing developmental issues associated with life-threatening medical complications in the early months and years of life
<b>Ventricular Septal Defect</b>	A malformation or perforation in the wall between the ventricles, or lower chambers of the heart, allowing a mixing of oxygenated and unoxygenated blood

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