



RHODE ISLAND BIRTH DEFECTS PROGRAM

BIRTH DEFECTS DATA BOOK 2008

RHODE ISLAND DEPARTMENT OF HEALTH



## WHAT ARE BIRTH DEFECTS?

Birth defects are structural abnormalities that affect the development of organs and tissues of an infant or child. These abnormalities may be identified during pregnancy, at birth or following birth. Possible causes or contributing factors to birth defects include genetic (inherited factors), environmental pollutants, occupational hazards, dietary factors, medications, and personal behaviors.

## WHY STUDY BIRTH DEFECTS?

Each year in the United States, 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 experience childhood illness and disability.

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.

Approximately 20% of all infant deaths in the United States are due to birth defects. Compared to the United States, a slightly higher proportion of Rhode Island infant deaths (nearly one in four) were a result of a birth defect. Of the 71 infants who died in Rhode Island during 2006, 10 of the deaths (14.1%) were attributed to a birth defect. During 2002-2006, the proportion of infant deaths resulting from a birth defect and the birth defects specific infant mortality rate decreased. Specifically, in 2002, 22 of the 91 deaths (24.2%) among Rhode Island infants resulted from a birth defect, yielding a birth defects specific infant mortality rate of 170.6 per 100,000 live births. Since then, the Rhode Island birth defects specific infant mortality rate decreased by more than half (52.6%) to 80.9 in 2006.

Many infants who are born preterm (prior to 37 weeks gestation) also have birth defects. Research is currently being conducted nationally to determine the association of preterm births and birth defects. In Rhode Island, 104 (6.7%) of the 1,550 babies born preterm in 2006, had a birth defect. Preterm births are also the leading cause of infant deaths in Rhode Island. Of the 71 infant deaths in 2006, 27 (38.0%) were attributed to prematurity.

# 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, Division of Community, Family Health and Equity. The Program 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 Program is kept confidential and is protected under State and Federal privacy laws.

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

The Rhode Island Birth Defects Advisory Council was appointed by the Director of Health 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 analyses.



## CASE ASCERTAINMENT AND DATA

The Rhode Island Birth Defects Program (RIBDP) has been using hospital discharge data to identify babies born with birth defects because it is the only data set that captures diagnoses coded by the International Classification of Diseases (ICD) system. This coding system, which is in its 9th Clinical Modification (ICD 9-CM) for births, provides more specificity on the type of birth defect. Although the birth certificate contains some information on birth defects, it does not capture all of them and does not include ICD codes. A higher number of babies with birth defects have been identified using the hospital discharge database than the birth certificate. For example, during 2000, 781 babies were identified with birth defects using hospital discharge data compared with 372 using birth certificate data.

In addition to collecting data through the hospital discharge database, RIBDP is also working with Women and Infants Hospital, Rhode Island Hospital, and Hasbro Children's Hospital to obtain additional cases of birth defects and information on services provided to families of children with birth defects.

Hospital discharge data indicate that during 2002-2006, among the 61,308 babies born to Rhode Island residents in Rhode Island maternity hospitals, 3,314 (5.4%) had at least one birth defect.

Figures 1 and 2 show that overall, the number and rate of birth defects in Rhode Island remained fairly stable between 2002-2005. However, during 2006, the number of cases of birth defects dropped to 562 (470 per 10,000), representing a 17.6% decline since 2002. This decline is not attributable to any significant changes in the prevalence of specific conditions, coding, and ascertainment procedures/methodologies or computer programming changes. Preliminary data for 2007 indicate that the number of cases of birth defects may be similar to the 2006 figure. The Birth Defects Program will continue to track these data and try to determine the factors contributing to the decline.



Figure 1  
NUMBER OF BABIES WITH BIRTH DEFECTS\*  
RHODE ISLAND, 2002-2006



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

Source: Hospital Discharge Database, Rhode Island Department of Health

Figure 2  
BIRTH DEFECTS PREVALENCE RATES,  
RHODE ISLAND, 2002-2006

Rate/10,000



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

Tables 1 and 2 show the number and rate of selected birth defects among Rhode Island residents by organ system. The most frequent birth defects are those related to the cardiovascular system. The most common types of heart defects include: ventricular and atrial septal defects, and pulmonary valve atresia or stenosis. One out of every 39 Rhode Island babies born in Rhode Island are born with a defect related to the cardiovascular system. Other common birth defects include hypospadias and club foot.

*One out of every 39 babies born  
in Rhode Island is born with a defect  
related to the cardiovascular system.*

Table 1

**BIRTH DEFECTS BY BODY SYSTEM, RHODE ISLAND, 2002-2006**

ORGAN SYSTEM	NUMBER	RATE PER 10,000
CARDIOVASCULAR	1233	201.1
MUSCULOSKELETAL/INTEGUMENTAL	1066	173.9
GENITOURINARY	869	141.7
GASTROINTESTINAL	291	47.5
EYE, EAR, FACE, AND NECK	193	31.5
CENTRAL NERVOUS SYSTEM	141	23.0
CHROMOSOMAL	103	16.8
RESPIRATORY	84	13.7



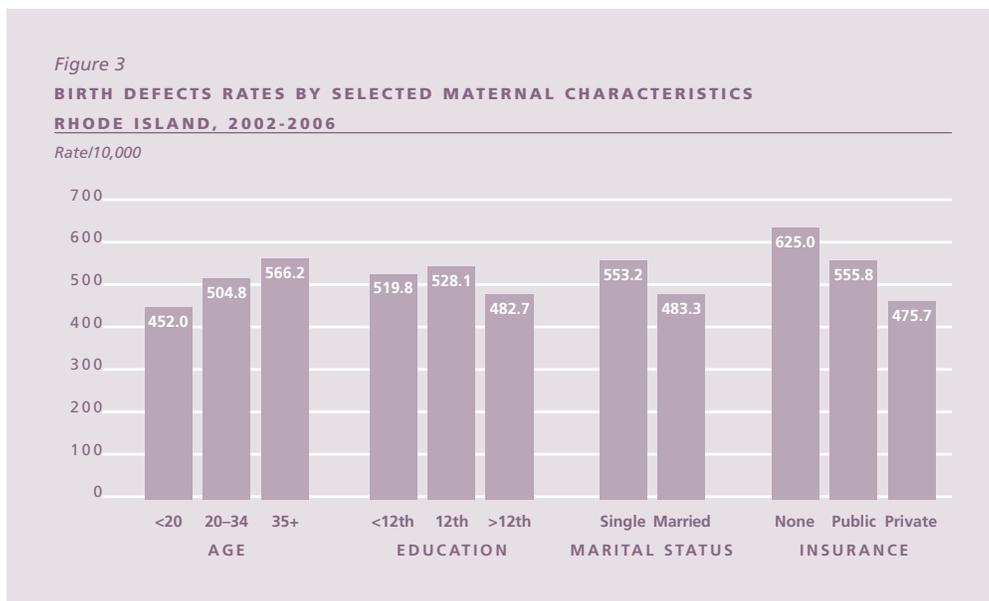


Table 2

**SELECTED BIRTH DEFECTS, RHODE ISLAND, 2002-2006**

BIRTH DEFECT	NUMBER	RATE PER 10,000 LIVE BIRTHS
<i>Central Nervous System</i>		
SPINA BIFIDA	18	2.9
ANENCEPHALY	2	0.3
ENCEPHALOCELE	4	0.7
HYDROCEPHALY	35	5.7
<i>Eye</i>		
CONGENITAL CATARACT	3	0.5
ANOPHTHALMOS AND MICROPHTHALMUS	3	0.5
<i>Ear</i>		
ANOTIA/MICROTIA	8	1.3
<i>Cardiovascular</i>		
TRANSPOSITION OF GREAT ARTERIES	15	2.4
TETRALOGY OF FALLOT	25	4.1
VENTRICULAR SEPTAL DEFECT	234	38.2
ATRIAL SEPTAL DEFECT	220	35.9
ENDOCARDIAL CUSHION DEFECT	13	2.1
PULMONARY VALVE ATRESIA/STENOSIS	101	16.5
TRICUSPID VALVE ATRESIA	5	0.8
AORTIC VALVE STENOSIS	3	0.5
HYPOPLASTIC LEFT HEART SYNDROME	8	1.3
COARCTATION OF AORTA	11	1.8
<i>Orofacial</i>		
CHOANAL ATRESIA	7	1.1
CLEFT LIP WITH AND WITHOUT CLEFT PALATE	51	8.3
CLEFT PALATE	29	4.7
<i>Gastrointestinal</i>		
ESOPHAGEAL ATRESIA/TRACHEOSOPHAGEAL FISTULA	6	1.0
RECTAL AND LARGE INTESTINAL ATRESIA/STENOSIS	20	3.3
HIRSHSPRUNG'S DISEASE	6	1.0
<i>Genitourinary</i>		
HYPOSPADIAS AND EPISPADIAS	212	34.6
RENAL AGENESIS/HYPOPLASIA	20	3.3
OBSTRUCTIVE GENITOURINARY DEFECT	184	30.0
<i>Musculoskeletal</i>		
CLUB FOOT	117	19.1
REDUCTION DEFORMITY, UPPER LIMBS	16	2.6
REDUCTION DEFORMITY, LOWER LIMBS	6	1.0
DIAPHRAGMATIC HERNIA	12	2.0
GASTROSCHISIS	35	5.7
OMPHALOCELE	3	0.5
<i>Chromosomal</i>		
DOWN SYNDROME	75	12.2
TRISOMY 13	35	0.5
TRISOMY 18	7	1.1
<b>ALL BIRTH DEFECTS</b>	<b>3314</b>	<b>540.5</b>

*Babies born to older women,  
women with less than a high school education, and  
women with publicly funded health insurance  
are at a higher risk for birth defects.*



## MATERNAL CHARACTERISTICS

Babies born to older women (aged 35 or older), women with less than a college 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 2002-2006, the birth defects rate among women aged 35 or older was 566.2 compared with 452.0 among women aged less than twenty. Similarly, the birth defects rate among women with a 12th grade (528.1) or lower education (519.8) was higher than the rate among women with more than a high school education (482.7). Single women (553.2) were also more likely to have a baby with a birth defect than married women (483.3). Women who were insured through public programs such as, RIte Care and Medicaid (555.8), were 1.2 times more likely to have a baby with a birth defect than women who were insured by commercial or private providers such as, Blue Cross and United HealthCare (475.7). It should be noted that although a small number of women did not have health insurance at the time of delivery (approximately 1% of all births), the risk for having a baby with a birth defect was highest among this population (625.0) compared to the populations that had health insurance.



## RACIAL/ETHNIC AND GEOGRAPHIC DISPARITIES

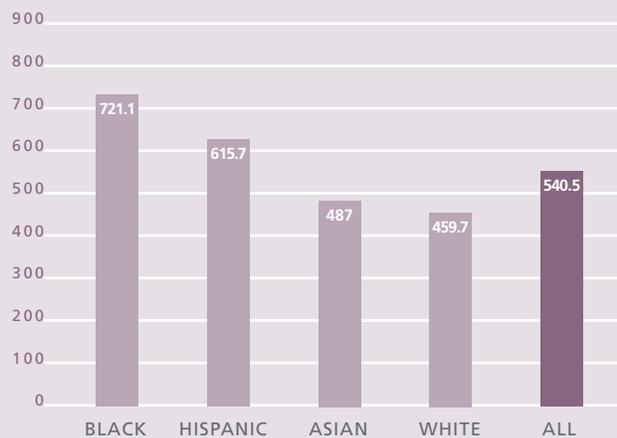
Birth defects rates vary by race/ethnicity and geographical area. During 2002-2006, the average birth defects rate among Black/African Americans was 721.1, 1.6 times the rate for Whites (459.7). Women of Hispanic/Latino ethnicity and Asians also had birth defects rates that were higher than the rate for Whites (615.7 and 486.7, respectively). See Figure 4.

Birth defects rates were also higher among those who reside in Rhode Island's six core cities (Central Falls, Newport, Pawtucket, Providence, West Warwick, and Woonsocket) than those who live in the rest of the state (Figure 5). Babies born to residents of the core cities were approximately 1.2 times more likely to have a birth defect than those born in the rest of the state (554.7 versus 475.5). Specifically, babies born to Newport residents had the highest birth defects rate, 934.1, nearly twice the rate for those residing in a non-core city (rest of the state).

Figure 4

### BIRTH DEFECTS RATES BY RACE/ETHNICITY RHODE ISLAND, 2002-2006

Rate/10,000



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

Figure 5

### BIRTH DEFECTS RATES BY SELECTED GEOGRAPHICAL AREAS RHODE ISLAND, 2002-2006

Rate/10,000



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

# MAPPING RATES OF BIRTH DEFECTS IN RHODE ISLAND

The map on the following page illustrates birth defects rates during 2002-2006 by city/town and region. Regions are denoted by orange lines and were created for those towns that had less than 500 live births (e.g., Charlestown, Exeter, Foster, Glocester, Hopkinton, Jamestown, Little Compton, New Shoreham, North Smithfield, Richmond, Scituate, and West Greenwich). These towns were aggregated with other towns in close proximity to increase the denominators and statistical significance of the data. Table 3 below shows the seven regions by towns, number of birth defect cases, total live births and birth defects rates.

It should be noted that some variations in birth defects rates among towns and/or regions may be attributed to different diagnostic and coding procedures used by hospitals. For example, minor skin anomalies and ankyloglossia (tongue-tie) appear to be coded more often by certain hospitals. The development of statewide standards for case definitions may help reduce the number of cases of these types of birth defects and provide consistency.

Table 3

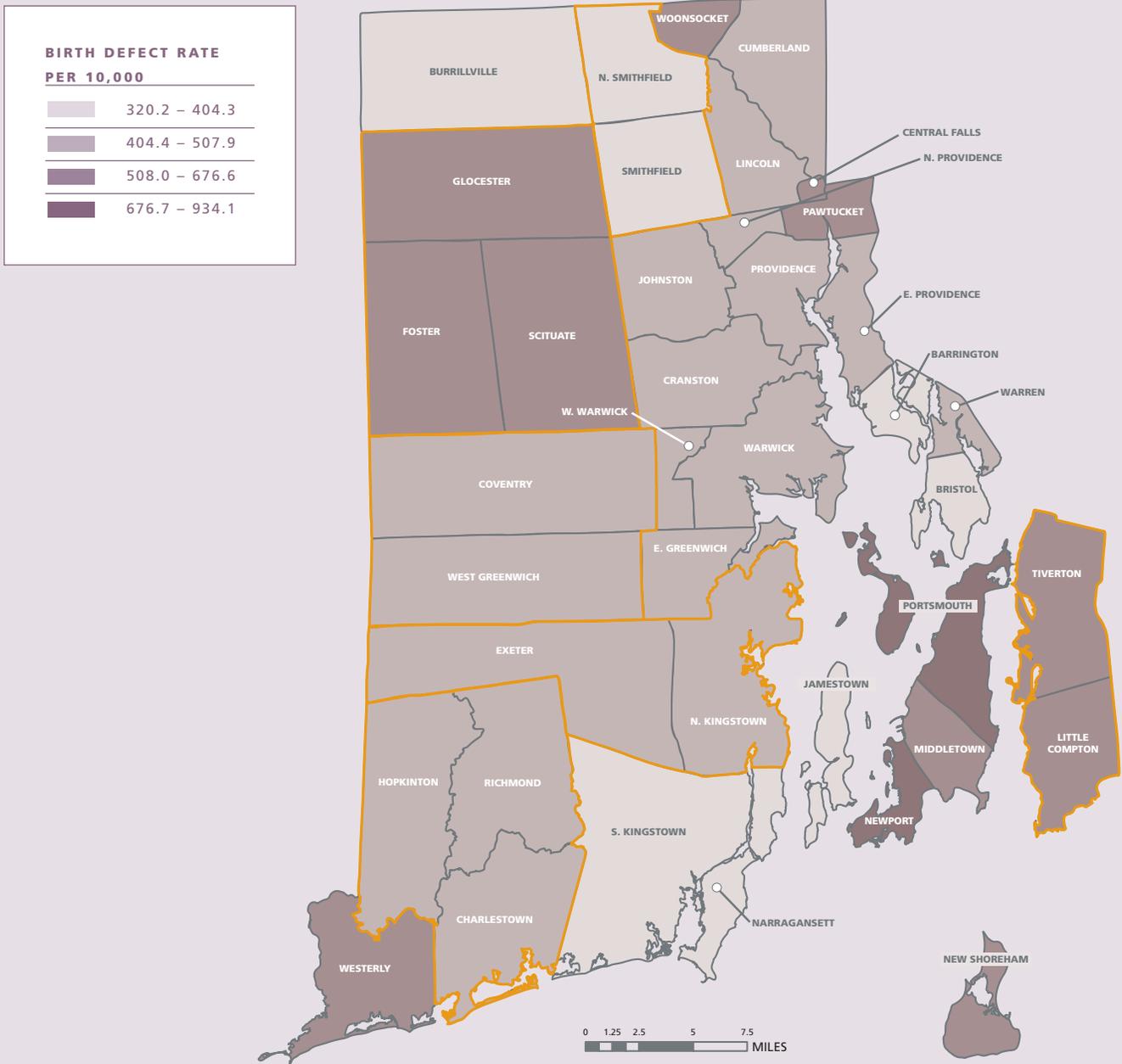
**BIRTH DEFECTS RATES BY REGIONS**

REGION	TOWN	BIRTH DEFECTS CASES	TOTAL LIVE BIRTHS	PREVALENCE (RATE/10,000 LIVE BIRTHS)
1	North Smithfield, Smithfield	43	1142	376.5
2	Glocester, Foster, Scituate	64	1080	592.6
3	Coventry, West Greenwich	97	2081	466.1
4	Exeter, North Kingstown	71	1613	440.2
5	Hopkinton, Richmond, Charlestown	57	1222	466.4
6	Tiverton, Little Compton, New Shoreham	31	554	559.6
7	Jamestown, Narragansett	29	800	362.5



*Babies born to residents of the core cities  
were approximately 1.2 times more likely  
to have a birth defect than those born  
in the rest of the state.*

RATES OF BIRTH DEFECTS BY CITIES/TOWNS AND REGIONS  
RHODE ISLAND, 2002-2006



## SENTINEL CONDITIONS

The Rhode Island Birth Defects Advisory Council identified a set of conditions (see Table 4) for targeted outreach and follow-up to help assure appropriate services and referrals are made. The selection was based on criteria that included the following: number of children affected; timeliness of identification; severity; service availability; resource intensity; recurrence risk; and availability of prevention strategies. The purpose of creating this list is to assure that families of children with these conditions are provided information and resources.



Table 4

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

CONDITION	ICD-9-CM CODE
<b>NEURAL TUBE DEFECTS</b>	
» ANENCEPHALY	740.0-740.1
» SPINA BIFIDA	741.0-741.9
<b>EYE</b>	
» ANOPHTHALMIA/MICROPHthalmIA	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
<b>EAR</b>	
» ANOTIA/MICROTIA	744.01, 744.23
» HEARING LOSS	(referral via RI Hearing Assessment Program)
<b>LUNG</b>	
» CONGENITAL CYSTIC LUNG	748.4
» AGENESIS, HYPOPLASIA AND DYSPLASIA OF LUNG	748.5
CONGENITAL TUMORS	140-239
<b>DEVELOPMENTAL CONDITIONS</b>	
AUTISM	299.0



## SERVICE ASSURANCE FOR CHILDREN WITH BIRTH DEFECTS

The Rhode Island Birth Defects Program (RIBDP) was created to identify newborns with birth defects; ensure that these children receive appropriate preventive, specialty, and other healthcare services; and monitor birth defects data trends. The RIBDP used a three-pronged approach that included quantitative and qualitative data to ensure that children with birth defects and their families receive the medical and social support services they need:

### 1) FAMILY FOCUS GROUPS

The RIBDP conducted four focus groups and five one-on-one interviews with parents of children with birth defects in order to obtain information about their experiences with the programs and services offered by the RIBDP and other Rhode Island healthcare resources. The research was designed to determine the degree and quality of support, treatment, and healthcare coverage parents and their children received. The research also gathered parents' opinions about the use of the phrase "birth defects" to inform a potential program name change.

Overall, parents' experiences with the diagnostic process, accessing healthcare services, and obtaining healthcare coverage and/or medical assistance were mixed. Some had very positive experiences, but most had encountered a variety of challenges. Their responses indicate the availability of a wide range of options for services and support, but a lack of information about and coordination between these services. Parents need consistent communication, smooth coordination, with providers and programs that provide individualized services and attention. Most parents were highly involved in their children's care and were willing advocates, but they expected and appreciated dedicated support from providers, and quality customer service from healthcare companies as well.

Many of the available support programs and services were invaluable for parents, particularly Early Intervention (EI), Rhode Island Parent Information Network (RIPIN), Children's Neurodevelopment Center (CNDC) and Comprehensive Evaluation,

Diagnosis, Assessment, Referral and Re-evaluation Family Centers (CEDARR). The most significant problem parents reported about the programs was that they did not learn about them sooner. They felt that more timely and consistent communication about the programs, both from providers and within the network of services, would be very helpful.

Finally, most parents agreed that renaming the Birth Defects Program would be a positive change. They recommended avoiding use of the word “defect,” and focusing on less clinical and loaded terms. They would also prefer terminology that includes the wide range of conditions that are considered “birth defects,” and that does not imply specific severities or types of conditions.

## 2) SERVICE ASSESSMENTS

In collaboration with the RI Birth Defects Advisory Council, the RIBDP developed service assessment tools based on national guidelines for pediatric primary care providers and families of children with Down syndrome to collect information on what and when services were provided to these children. The Advisory Council recommended piloting these tools for children with Down syndrome before expanding to the other RI sentinel conditions.

As a pilot, 50 families of children with Down syndrome were surveyed to determine the healthcare services and referrals their children received. This was done in collaboration with the Down Syndrome Society of Rhode Island and the Rhode Island Parent Information Network (RIPIN). The following results were obtained from the 32 (64%) families who responded to the survey:

*Medical Services/Testing:* The majority of the children in the survey visited the pediatrician and received many of the medical tests and services recommended by national guidelines, such as heart echocardiograms and chromosome tests. However, less than half of the families had received genetic counseling. Approximately, one in three children and/or their families received nutritional information and hearing tests; and less than one in five (<20%) received vision, thyroid, and celiac tests as recommended by the national guidelines. Finally, very few children in the survey had received dental exams.

*Parent Referrals to Support and Developmental Services:* The agencies or organizations that families in the survey were most likely be referred to were the Down Syndrome Society of Rhode Island (72%); visiting nurse associations (53%); and the Rhode Island Parent Information Network (41%).

*Developmental/Educational Services:* The majority of families had been referred for the following developmental and educational services: early intervention (88%), special education (88%), physical therapy (82%), speech therapy (75%), occupational therapy (66%), and Children’s Neurodevelopment Center (53%). Among children aged less than three, 53% received an Individualized Family Service Plan (IFSP) and among children aged 3 or older, 38% received an Individualized Educational Plan (IEP).

## 3) KIDSNET

The RIBDP is linked to Rhode Island’s integrated child health information system, KIDSNET, to determine whether children with birth defects have received appropriate



preventive and program services. Data indicate that in comparison to children without birth defects, children with birth defects are more likely to have been screened for lead poisoning (83.3% vs. 81.3%), be enrolled in Women, Infants and Children (WIC) (54.0% vs. 48.0%), receive home visits (43.1% vs. 39.2%) and be enrolled in the Early Intervention Program (26.7% vs. 8.0%). Nearly all (94.9%) of the children identified with Down syndrome were enrolled in the Early Intervention Program, 38.5% received a home visit and 33.3% were enrolled in WIC.

The RIBDP has found that using a multi-pronged approach provides a more complete representation of the degree to which children and families are receiving appropriate and timely medical and social support services. Collaboration with providers, families, and social support organizations is critical for data collection and service assessment.

## COLLABORATIVE RESEARCH: NATIONAL AND INTERNATIONAL STUDIES

The RIBDP has participated or is currently participating in a variety of national and international collaborative studies to gain a better understanding of specific birth defects, their trends and associated factors. Examples of these collaborative research studies, their organizational sponsor and purpose are described in Table 5.

Table 5

### 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.

## RESOURCES

Many children with birth defects are evaluated and followed at the Genetics Counseling and Child Development Centers at Rhode Island Hospital. Clinics are available for most birth defects, including meningomyelocele, Down syndrome, inborn errors of metabolism, and other syndromes. The Cardiology Clinic is a pediatric specialty clinic at Hasbro Children's Hospital. Rare conditions are referred to Boston specialists.

## ADDITIONAL INFORMATION: RELATED SOURCES AND WEBSITES:

### RHODE ISLAND

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

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), Family Voices of Rhode Island: [www.ripin.org](http://www.ripin.org)**

Provides information, training, support, and advocacy to parents seeking help for their children.

**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.state.ri.us/dhs/famchild/early\\_intervention.htm](http://www.dhs.state.ri.us/dhs/famchild/early_intervention.htm)**

Promotes the growth and development of infants and toddlers. Children who are under three years of age who have a developmental disability or delay in one or more areas may be eligible.

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

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

### Genetic Counseling & Medical Genetics Services

***Prenatal Diagnostic Center***

Women & Infants Hospital  
79 Plain Street  
Providence, RI 02903  
Phone: 401-453-7510  
Fax: 401-453-7517  
Offers screening, diagnostic, and genetic counseling during pregnancy

***Genetics Counseling Center***

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

***Greystone/Radiologic Associates***

235 Plain Street  
Providence, RI 02905  
Phone: 401-272-8510  
Fax: 401-272-0315  
Provides genetic counseling, prenatal testing and diagnosis and pre-conception evaluations.

**Children's Neurodevelopment Center: [www.lifespan.org/Services/ChildHealth/CDC](http://www.lifespan.org/Services/ChildHealth/CDC)**

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

## 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. Website contains information regarding the Academy's many programs, activities, policy statements, practice guidelines, publications, and other child health resources.

American Academy of Family Practitioners: [www.aafp.org](http://www.aafp.org)

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

### **Birth Defects Research for Children, Inc.: [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.

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

Provides general information on birth defects, surveillance, and key research findings and risk factors.

### **CDC National Center for Chronic Disease Prevention & Health Promotion: Maternal and Infant Health: [www.cdc.gov/nccdphp/drh](http://www.cdc.gov/nccdphp/drh)**

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

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.

### **GeneClinics: [www.geneclinics.org](http://www.geneclinics.org)**

By providing current, authoritative information on genetic testing and its use in diagnosis, management, and genetic counseling, GeneClinics promotes the appropriate use of genetic services in patient care and personal decision-making.

### **KidNeeds.com (information and resource center): [www.kidneeds.com](http://www.kidneeds.com)**

KidNeeds.com is a place where children with special needs, families, and others can find professional opinions on important topics, learn about public health policy and advocacy efforts, link to programs, services and connect with other caring families and friends, and purchase a wide range of products specifically tailored for children with special needs.

### **National Birth Defects Prevention Network: [www.nbdpn.org](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 Down Syndrome Society: [www.ndss.org](http://www.ndss.org)**

Presents comprehensive information on Down syndrome for families, healthcare professionals, educators, scientists, affiliates, parent support groups, and other organizations.

**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: [www.nichcy.org](http://www.nichcy.org)**

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: [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 those affected individuals and families.

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

Studies the effects that drugs, medications, chemicals, and other exposures may have on the fetus. 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: [www.sbaa.org](http://www.sbaa.org)**

Promotes the prevention of spina bifida and enhancing 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****The Birth Defects Foundation: [www.bdfcharity.co.uk](http://www.bdfcharity.co.uk)**

The BDF funds vital medical research to understand, prevent, combat, and treat inborn conditions; has a nurse service that comforts, informs, and supports families and those at risk; gives direct grants to affected families, funding vital equipment and assistance; and campaigns to raise the level of awareness helping people to act, prior to pregnancy, to reduce the risks.

**International Clearinghouse for Birth Defects Monitoring Systems: [www.icbd.org](http://www.icbd.org)**

Dedicated to the sharing of data, news, and views on congenital malformations monitoring, research, and prevention.

## 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>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>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>Congenital Disorder</b>	a medical condition that is present at birth but may be recognized before birth.
<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>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 and 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>	is a partial or complete split in the palate (roof of the mouth), occurring without a split in the lip.
<b>Club Foot</b>	babies born with this condition have their foot turned to the side and 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>Diaphragmatic Hernia</b>	the absence 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 mild to moderate mental retardation, 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>Endocardial Cushion 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>Esophageal Atresia/ Tracheoesophageal Fistula</b>	these defects involve abnormal closure, or abnormal holes within the esophagus, or within the trachea (windpipe), it can also occur that the upper and lower ends of the esophagus are not connected.
<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 of the abdominal wall.
<b>Genetic</b>	having to do with heredity and variation in organisms.
<b>Hypoplastic Left Heart Syndrome</b>	one of the most life-threatening heart defects, it is an incomplete development of the left chambers of the heart.
<b>Hypospadias and Epispadias</b>	the 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.
<b>Infant</b>	a child aged less than one year (less than 12 months) of age.
<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>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 and 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 imperfectly closed, so part of the spinal cord may protrude, this condition often resulting in neurological disorders.
<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/Trisomy 18</b>	the condition of having three copies of chromosome 13 or 18, conditions causing severe skull and facial deformation and mental retardation.
<b>Tetralogy of Fallot</b>	this complex malformation of the heart is 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>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.

**ACKNOWLEDGEMENTS**

RHODE ISLAND BIRTH DEFECTS ADVISORY COUNCIL  
MARCH OF DIMES, RHODE ISLAND CHAPTER  
WOMEN AND INFANTS HOSPITAL  
RHODE ISLAND HOSPITAL  
HASBRO CHILDREN'S HOSPITAL  
RHODE ISLAND KIDS COUNT  
RHODE ISLAND PARENT INFORMATION NETWORK (RIPIN)  
VNA CARE NEW ENGLAND

**FOR MORE INFORMATION CONTACT:**

SAMARA VINER-BROWN, MS, DIRECTOR, RHODE ISLAND BIRTH DEFECTS PROGRAM  
RHODE ISLAND DEPARTMENT OF HEALTH  
SAMARA.VINER-BROWN@HEALTH.RI.GOV

