

# Centers for Birth Defects Research and Prevention

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U.S. Department of Health and Human Services  
Centers for Disease Control and Prevention

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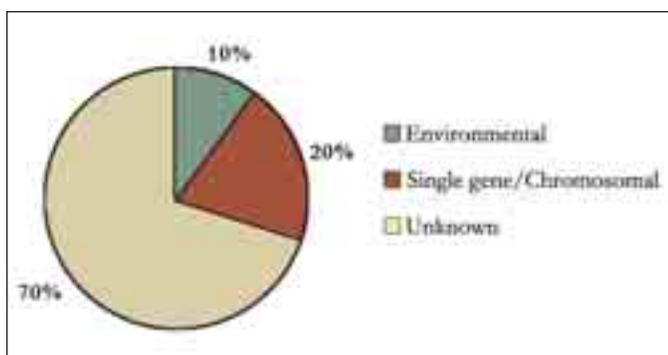


# INTRODUCTION

## The Need for Birth Defects Research

Birth defects are the leading cause of infant death in the United States, accounting for more than 20% of all infant deaths. Birth defects also contribute substantially to illness and long-term disability. The first step in preventing birth defects is to identify their causes. However, the causes of about 70% of all birth defects remain unknown (Figure 1).

Figure 1: Causes of Birth Defects



Because many types of birth defects occur at such low rates, it is difficult to conduct studies that include enough children with specific birth defects to identify their causes. A collaborative national effort is essential to establish a study group large enough to determine what environmental, genetic, and behavioral factors cause or contribute to specific birth defects.

## Establishment of the Centers for Birth Defects Research and Prevention

To help reduce birth defects among U.S. babies, in 1996, Congress directed the Centers for Disease Control and Prevention (CDC) to establish the Centers for Birth Defects Research and Prevention (CBDRP). This directive was formalized with the passage of the Birth Defects Prevention Act of 1998 (Public Law 105-168). This act authorized CDC to 1) collect, analyze, and make available data on birth defects; 2) operate regional centers that will conduct applied epidemiologic research for the prevention of birth defects; and 3) provide the public with information about preventing birth defects. Currently, CDC has established centers in Arkansas, California, Iowa, Massachusetts, New York, North Carolina, Texas, and Utah. These states have existing birth defects programs with nationally recognized expertise in birth defects surveillance and research. CDC coordinates the CBDRP and participates in the National Birth Defects Prevention Study (NBDPS) as the ninth study site (Figure 2).

Figure 2: Centers for Birth Defects Research and Prevention



## CBDPR Activities

The establishment of CBDPR has increased the capability of the state programs to carry out research that will increase our understanding of the causes of birth defects and provide information that can be used to prevent birth defects. The centers are 1) participating in NBDPS, 2) conducting center-specific research projects, and 3) enhancing their state birth defects surveillance systems.

## NBDPS

The National Birth Defects Prevention Study (NBDPS) is the largest population-based study ever conducted on the causes of birth defects. It will provide information about environmental and genetic factors that contribute to birth defects and will serve as a mechanism for identifying new factors that are harmful to developing babies. The study is comprised of three components. First, through existing surveillance systems, CBDPR is identifying and collecting information on "cases"—infants who have any of the major birth defects listed in Table 1.



Table 1: Birth Defects Studied in the National Birth Defects Prevention Study by System Affected

System Affected	Birth Defects Studied
Cardiovascular	Anomalous pulmonary venous return, conotruncal heart defects, Ebstein malformation, heterotaxy, hypoplastic left heart syndrome, obstructive heart defects, septal heart defects, single ventricle
Central nervous system	Anencephaly, Dandy-Walker malformation, encephalocele, holoprosencephaly, hydrocephaly, spina bifida
Eye	Anophthalmia/microphthalmia, congenital cataracts, glaucoma
Ear	Anotia/microtia
Orofacial	Choanal atresia, cleft lip, cleft palate
Gastrointestinal	Biliary atresia, esophageal atresia and tracheoesophageal fistula, intestinal atresia
Genitourinary	Hypospadias (2nd or 3rd degree), renal agenesis (bilateral)
Musculoskeletal	Bladder exstrophy, cloacal exstrophy, craniosynostosis, diaphragmatic hernia, gastroschisis, limb deficiency, omphalocele, sacral agenesis/caudal regression
Non-system-specific	Amniotic band sequence

Clinical geneticists at each center are reviewing and classifying the clinical information for each case. The clinical information about cases eligible for the study is being stored in a central database. CBDRP is also identifying "control" infants who do not have birth defects. Information gathered about the cases and controls will be compared to identify factors that increase the risk for or protect against birth defects.

Second, CBDRP is interviewing the infants' mothers using a computer-assisted telephone interview. Interviews include questions about pregnancy and medical history, occupational and environmental exposures, lifestyle, diet, and medication use.

Third, CBDRP is collecting cheek cells from the infants and their parents to identify genetic factors. Cheek cells are collected by brushing a swab across the inside wall of the mouth. Researchers will study the DNA (genetic material) from these cheek cells to identify whether certain genes cause or increase the risk of a particular birth defect. A portion of the DNA that is collected from the families is stored in a specimen bank at CDC.



The information gathered from the interviews, combined with the availability of DNA, will provide an invaluable resource for the study of genetic susceptibility to environmental exposures for a broad range of carefully classified birth defects. The unprecedented statistical power from this collaborative study will enable scientists to study the epidemiology of some rare birth defects for the first time, and the compiled data and banked DNA will facilitate future research as new hypotheses and improved technologies emerge.



## Center-Specific Birth Defects Research Studies

CBDRP is also conducting studies of birth defects that are of local or regional interest. Issues of interest include nutritional, environmental, and behavioral factors involved in birth defects; gene-environment interactions that increase or decrease risk for birth defects; financial and other costs of birth defects; and primary prevention of birth defects. The centers are also using state-of-the-art technology for in-depth studies of the role of specific genetic variants. Center-specific research projects are described in the individual center profiles.

## Birth Defects Surveillance

Each center maintains a population-based birth defects surveillance system. Data from surveillance systems are used to detect trends in birth defects and suggest areas for further study. Surveillance information can also be used to identify epidemiologic factors associated with birth defects, to address community concerns about the environment's effects on birth outcomes, and to evaluate screening and prevention programs. CBDRP's surveillance activities are described in the individual center profiles.



# PROFILES OF THE CENTERS FOR BIRTH DEFECTS RESEARCH AND PREVENTION

## Centers for Disease Control and Prevention

The Division of Birth Defects and Developmental Disabilities in the Centers for Disease Control and Prevention (CDC) monitors and conducts epidemiologic studies of birth defects and assists state and local health agencies, nonprofit organizations, international agencies, and others in preventing these conditions. Located in Atlanta, Georgia, CDC provides both technical and administrative oversight to the Centers for Birth Defects Research and Prevention (CBDRP) and participates in the National Birth Defects Prevention Study (NBDPS) as the Georgia study site.

Since 1967, CDC has been monitoring the occurrence of serious birth defects and genetic diseases in Atlanta through the Metropolitan Atlanta Congenital Defects Program (MACDP) surveillance system. MACDP provides continuous data collection on all major structural birth defects and serves as a model for other state surveillance systems. It also provides the Georgia study site with cases for NBDPS and the diagnostic information for classification of cases.

The establishment of the Georgia study site has enabled CDC to strengthen its research agenda, particularly in gene-environment interaction studies, and to maximize the use of CDC staff members' expertise and their participation in various birth defects research projects.



NBDPS's design, methodology, and data collection instruments were developed by committees composed of CBDRP collaborators. CDC coordinates these committees and maintains a repository for all NBDPS data. In addition to identifying the causes of birth defects, CDC's goal is to strengthen the capacity of CBDRP to carry out research by mentoring and training young scientists in birth defects surveillance, research, and prevention. CDC encourages collaboration among CBDRP's researchers and provides guidance on epidemiologic research development.



CDC brings many years of experience and expertise to birth defects surveillance, research, and prevention. It has a variety of experts—epidemiologists, pediatricians, clinical geneticists, statisticians, and behavioral scientists—working on numerous genetic and environmental epidemiologic studies related to birth defects. CDC is currently studying the association between birth defects and factors such as use of multivitamins, diet, use of medication, maternal health conditions, maternal smoking and alcohol use, genetic variation, and fertility treatments.



In addition to NBDPS, CDC conducts other research projects and activities, including the following:

- Working with the National Council on Folic Acid to educate women of reproductive age about the need to consume folic acid to reduce the risk of giving birth to children with neural tube defects. Low literacy materials have been developed to reach a critical at-risk audience.
- Collaborating with Beijing Medical University to identify birth defects risk factors and implement prevention efforts in two areas of the People's Republic of China.
- Helping to establish and coordinate the National Birth Defects Prevention Network (NBDPN), a network of individuals working at national, state, and local levels on birth defects surveillance, research, and prevention. NBDPN develops surveillance standards, designs prevention activities, and compiles and shares its data.
- Providing technical assistance to states that are developing birth defects surveillance programs and funding states to promote the use of surveillance data for prevention and intervention activities.

## CDC's Partners

- Molecular Genetics Laboratory, National Center for Environmental Health, CDC
- Georgia Department of Human Resources
- March of Dimes Birth Defects Foundation
- Battelle Memorial Institute

For more information about CDC activities, visit their website at [www.cdc.gov/ncbddd](http://www.cdc.gov/ncbddd).

# ARKANSAS

## Center for Birth Defects Research and Prevention

The Arkansas Center is a multi-disciplinary collaboration representing the Department of Pediatrics at the University of Arkansas for Medical Sciences (UAMS), the Arkansas Department of Health, and the Arkansas Children's Hospital Research Institute. Established in 1997, the mission of the Arkansas Center is to reduce the prevalence of birth defects in Arkansas and the nation, as well as to decrease the economic, social, and psychological impact of birth defects through surveillance, research, and prevention. Essential funding support has come from the Centers for Disease Control and Prevention (CDC), the National Institute of Child Health and Human Development (NICHD), the Arkansas Children's Hospital Research Institute (ACHRI), the Arkansas Biosciences Institute, the Arkansas state legislature, and the Pamela D. Stephens Endowed Chair in Birth Defects Research.

To accomplish its goals, the Arkansas Center uses data from the Arkansas Reproductive Health Monitoring System (ARHMS) - the state's birth defects surveillance system. Founded in 1980, ARHMS is one of the oldest active case ascertainment and highly regarded birth defects surveillance systems in the United States.



Since the establishment of the Arkansas Center, ARHMS has expanded from a surveillance system with a small staff to a multidisciplinary research and prevention team. Clinicians specialized in pediatrics, neonatology, pediatric cardiology, and clinical dysmorphology work with experts in human genetics, biostatistics, biochemistry, nutrition, developmental biology, health economics, health services research, and genetic epidemiology to advance the center's research agenda. The Arkansas Center's partnership with the Arkansas Department of Health facilitates collaboration with public health professionals in maternal and child health and health education.

The Arkansas Center's capacity to perform genetic studies was greatly enhanced in 2002 with support from the Arkansas Biosciences Institute, through the establishment of a state-of-the-art Genomics Laboratory. The Arkansas Biosciences Institute, founded through state appropriations from national tobacco settlement revenues, stimulates important research into areas of health affected by the use of tobacco products. The Genomics Laboratory plays a key role in all of the major studies currently underway at the Arkansas Center.

## Arkansas Center's Partners

The Arkansas Center serves as the lead agency for the Arkansas Folic Acid Coalition, which encourages Arkansas women of childbearing age to take a multivitamin containing 400 micrograms of folic acid daily and to eat foods rich in folate to help prevent neural tube defects such as spina bifida.

Through continued support of sponsors and study participants and collaborative efforts, the Arkansas Center will work to combine surveillance, research and prevention to help diminish the consequences of birth defects and help make a difference in the lives of children and families.

In addition to participating in the National Birth Defects Prevention Study (NBDPS), the Arkansas Center is conducting other research projects and activities, including the following:

- Genetic epidemiology of birth defects
- Biochemical genetics of birth defects
- Evaluation of primary prevention of birth defects
- Economic evaluation of birth defects
- Health care cost and utilization studies of birth defects

- Arkansas Children's Hospital
- Arkansas Children's Hospital Research Institute
- Arkansas Department of Health
- Arkansas Reproductive Genetics Program at UAMS
- Arkansas Biosciences Institute
- Arkansas Folic Acid Coalition
- Arkansas Chapter of the March of Dimes Birth Defects Foundation

For more information about the Arkansas Center, visit their website at [arbirthdefectsresearch.uams.edu](http://arbirthdefectsresearch.uams.edu).



## Center for Birth Defects Research and Prevention

Established in 1996, the California Center is based in the California Birth Defects Monitoring Program (CBDMP) in Berkeley. CBDMP is publicly funded and jointly administered by the California Department of Health Services and the March of Dimes Birth Defects Foundation. Since 1982, CBDMP has maintained a population-based registry and has conducted a number of case-control studies to advance its mission to identify the causes of birth defects.

The registry provides the California Center with cases for inclusion in the National Birth Defects Prevention Study (NBDPS) and complete diagnostic information for case classification. The California Center uses CBDMP's experience in collecting biological samples; investigating gene-environment interactions; and studying heart defects, neural tube defects, and cleft lip and palate. California's large and racially diverse population puts this Center in a unique position to identify risk factors for birth defects among Hispanic and Asian individuals. To encourage implementation of birth defects prevention activities, CBDMP's communications staff will widely publicize the Center's findings to public health professionals and the public.



CALIFORNIA  
BIRTH DEFECTS  
MONITORING  
PROGRAM

Federal funding not only supports the center's participation in NBDPS, but enables CBDMP to pursue important questions from earlier research, such as why Hispanics are at higher risk for neural tube defects, why folic acid is not as beneficial to Hispanics in preventing neural tube defects, and what role certain genes and nutritional factors play in the risk for birth defects.

In addition to participating in NBDPS, CBDMP is conducting other research projects, including the following:

- Assessing a wide range of risk factors—maternal diet, weight, acculturation, life-event stress, socioeconomic status, and family history—and their relationship to neural tube defects, cleft lip and palate, and selected heart defects.
- Studying the interaction between pre-pregnancy obesity and neural tube defects.
- Investigating whether genes and markers of inflammation contribute to heart, limb, or other birth defects.
- Investigating nutrient parameters in sera collected from women during mid-pregnancy among women who delivered infants with versus without birth defects.

## California Center's Partners

- Children's Hospital Oakland Research Institute, Oakland, California
- Genetic Disease Branch, California Department of Health Services
- University of Iowa
- Texas A&M University

For more information about the California Center, visit their website at [www.cbdmp.org](http://www.cbdmp.org).



CALIF  
BIRTH  
MONITORING  
PROGRAM

## Center for Birth Defects Research and Prevention

The Iowa Center, established in 1996, is a collaborative effort between the Colleges of Medicine and Public Health in the University of Iowa in Iowa City. The mission of the Iowa Center is to identify genetic and environmental risk factors for birth defects and to conduct educational activities to prevent birth defects. The Iowa Registry for Congenital and Inherited Disorders, located within the College of Public Health, conducts active, statewide surveillance of birth defects to identify cases for the National Birth Defects Prevention Study (NBDPS). The Reproductive Molecular Epidemiology Research and Education Program, located within the College of Public Health, recruits study subjects, conducts telephone interviews, and collects biologic specimens required for the center's research as well as conducts data analysis. The Department of Pediatrics in the College of Medicine provides expertise in mutation detection, characterization of genetic variations on large populations, gene identification and characterization and data analysis.

Iowa is a predominantly rural population, and the Iowa Registry for Congenital and Inherited Disorders is the only active case ascertainment birth defects surveillance system in the Midwest. This, along with state and university infrastructure support for a number of complementary projects, such as ground water surveillance, positions the Iowa Center well to investigate risk factors among rural populations that may affect fetal development.



Iowa Center staff have considerable expertise in investigating genes and environmental factors that contribute to the development of birth defects. They also have experience in applying geographic information systems (GIS) to studies of adverse birth outcomes.



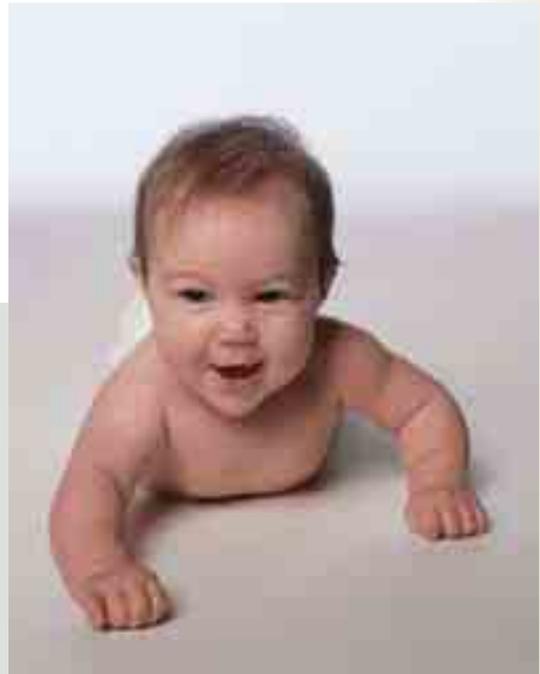
In addition to participating in NBDPS, the Iowa Center is conducting the following research projects:

- Examining the association between compounds in drinking water and adverse birth outcomes.
- Examining the association between exposure to agricultural chemicals and birth defects.

- Investigating genes, environmental exposures, and their interactions as risk factors for orofacial clefts.
- Investigating the effects of fertility treatments on the risk of birth defects.

## Iowa Center's Partners

- Iowa Center for Congenital and Inherited Disorders
- Iowa Regional Genetic Consultation Services
- Iowa Department of Public Health
- Iowa Chapter of the March of Dimes Birth Defects Foundation
- University of Iowa Center for Health Effects of Environmental Contamination



For more information about the Iowa Center, visit their website at [www.public-health.uiowa.edu/ircid](http://www.public-health.uiowa.edu/ircid).



# MASSACHUSETTS

## Center for Birth Defects Research and Prevention

Established in 1996, the Massachusetts Center is a collaboration between the Massachusetts Department of Public Health's Bureau of Family and Community Health (MDPH), Boston University's Slone Epidemiology Center (SEC), and the Active Malformation Surveillance Program at Brigham and Women's Hospital (BWH). The Massachusetts Center's mission is to support surveillance, research, and dissemination of information aimed at preventing birth defects.

State law has mandated the reporting of birth defects to MDPH for the past 40 years. While different public health programs have been collecting birth defects data during this time, administrative review of hospital discharge data and vital records has been the primary method of collecting data since the early '90s. The establishment of the Massachusetts Center enabled expansion of the monitoring program to a more active case finding, statewide, and population-based system. The Massachusetts Center has been actively collecting data on birth defects in the eastern part of the state since October 1997 and throughout the entire state since September 1998.

MDPH, coordinator of the Massachusetts Center, has a long history of experience in surveillance, maternal and child health needs assessment and program evaluation, and perinatal epidemiology. SEC and BWH bring to this collaboration more than 50 years of combined experience in birth defects research. The Massachusetts Center also draws on the expertise of and fosters communication among the region's strong network of clinicians and researchers.



The Massachusetts Center's areas of expertise include surveillance and research methodology; pediatric, reproductive, and social epidemiology; heart defects, congenital diaphragmatic hernia, limb reduction defects and orofacial clefts; teratology; drug research; and health service needs assessment.

In addition to participating in the National Birth Defects Prevention Study (NBDPS), the Massachusetts Center is conducting the following research activities:

- Studying the association of over-the-counter medications and herbal preparations on the risk of birth defects.
- Investigating the pharmacogenetic determinants of human birth defects.



- Examining the association of antiepileptic drugs and birth defects.
- Evaluating the risk for birth defects among infants of diabetic mothers.
- Analyzing the impact of prenatal diagnosis on the prevalence of birth defects, such as cardiovascular defects.
- Determining patterns of multivitamin use during pregnancy.
- Developing methods of birth defects classification and surveillance.

## Massachusetts Center's Partners

- Genetics and Teratology Unit, Massachusetts General Hospital
- Massachusetts Chapter of the March of Dimes Birth Defects Foundation
- Department of Maternal and Child Health and  
Department of Epidemiology, Boston University School of Public Health
- Department of Medical Genetics, Boston University School of Medicine
- Office of Health Statistics and Research and Office of Environmental  
Health Assessment, Massachusetts Department of Public Health

For more information about the Massachusetts Center, visit their website at [www.mass.gov/birthdefectscenter](http://www.mass.gov/birthdefectscenter).



# NORTH CAROLINA

## Center for Birth Defects Research and Prevention

The North Carolina Center, established in 2002, is a collaborative effort between the Department of Epidemiology at the University of North Carolina School of Public Health at Chapel Hill (UNC) and the North Carolina Birth Defects Monitoring Program (NCBDMP) in the Division of Public Health, North Carolina Department of Health and Human Services. The center's mission is to conduct epidemiologic research into the causes of birth defects, and to promote the use of research findings to enhance public health education and prevention efforts.

The UNC Department of Epidemiology is internationally recognized as a leader in epidemiologic research and training. As a part of its commitment to supporting state and local public health programs, the Department of Epidemiology and the School of Public Health as a whole have developed strong ties with public health agencies in North Carolina. The North Carolina Center's affiliation in a school of public health also provides a unique opportunity for training masters and doctoral students interested in pursuing careers in birth defects epidemiology and prevention.

Legislatively mandated in 1995, the NCBDMP maintains a statewide, population-based registry covering more than 110,000 births annually. The NCBDMP collaborates with UNC and other universities and agencies to develop a strong and effective working relationship involving teaching, research, program evaluations, and other public health activities.



Establishing the North Carolina Center has enabled the state to enhance an already strong multiagency collaboration that has been developed over the years. Such collaboration also brings to the North Carolina Center a considerable depth of expertise upon which to build a strong research program, including epidemiology, surveillance, embryology, teratology, and medical genetics.

In addition to participating in the National Birth Defects Prevention Study, the North Carolina Center is conducting other research projects and activities, including the following:

- Evaluating gene-environment interactions and the risk of selected birth defects including neural tube defects, oral clefts, and heart defects.
- Examining the temporal/geographic patterns and risk factors for gastroschisis.
- Investigating parental occupational and environmental exposures and the risk of birth defects.



- Evaluating folic acid education programs in relation to the prevalence of neural tube defects and intake of folic acid.
- Assessing potential barriers related to access and use of health services among children with birth defects and their families.

## North Carolina Center's Partners

- Duke University Medical Center
- University of North Carolina Center for Maternal and Infant Health
- North Carolina Chapters of the March of Dimes Birth Defects Foundation
- North Carolina Folic Acid Council
- Women's and Children's Health Section, Division of Public Health, NCDHHS

For more information about the North Carolina Center, visit their website at [www.schs.state.nc.us/SCHS/bdmp](http://www.schs.state.nc.us/SCHS/bdmp).



## Center for Birth Defects Research and Prevention

In 1996, the New York Center was established as part of the New York State Department of Health's Congenital Malformations Registry (CMR) and the State University of New York at Albany School of Public Health (SUNY SPH). The New York Center's mission is to refine existing surveillance activities and to develop partnerships for conducting birth defects prevention research.

CMR was established on October 1, 1982, partly as a response to the Department of Health's study of environmental contamination at Love Canal. The study showed that birth certificate data were inadequate for identifying the types of birth defects that were occurring and where they occurred. CMR was established to fill this need for data on birth defects. It is one of the largest birth defects registries in the nation, covering a racially and ethnically diverse population with approximately 270,000 births annually. The New York Center draws its cases for the National Birth Defects Prevention Study (NBDPS) from two areas: 1) eight counties in western New York that are also the focus of fetal alcohol syndrome surveillance; and 2) the lower Hudson Valley Region, which covers seven counties and is also the focus of neural tube defect surveillance.



Establishing the New York Center has enabled CMR to develop partnerships that strengthen its work and has provided resources to build its research agenda, particularly in the area of gene regulation and birth defects. The SUNY SPH includes faculty with expertise in environmental assessment and socio-economic factors and the new Genomics Institute. The New York Center's areas of expertise include geographic information systems (GIS), occupational exposure studies, environmental epidemiology, folate metabolism, and studies of the relationship between socioeconomic status and birth defects.



In addition to participating in NBDPS, the New York Center is conducting other research projects and activities, including the following:

- Studying maternal chronic diseases/medications and birth defects.
- Studying the role of common exposures, such as the effects of caffeine and alcohol on heart defects, including genes and gene-environment interactions.
- Examining quality-of-care issues for children with birth defects, which includes supporting the development of clinical guidelines for doctors to assist in the complex task of properly diagnosing children with birth defects.
- Studying screening methods to identify children with congenital heart malformations before hospital discharge.
- Examining trends in abdominal wall defects and factors in infant survival.
- Improving surveillance of neural tube defects and fetal alcohol syndrome.
- Studying parental occupational exposures and birth defects.

## New York Center's Partners

- Division of Genetics, Children's Hospital of Buffalo
- Department of Epidemiology at the University of Alabama at Birmingham
- New York Chapters of the March of Dimes Birth Defects Foundation
- SUNY Albany School of Public Health
- Regional Medical Genetics Center, Westchester, NY



For more information about the New York Center, visit their website at [www.health.state.ny.us/nysdoh/cmr/cmrhome.htm](http://www.health.state.ny.us/nysdoh/cmr/cmrhome.htm).

## Center for Birth Defects Research and Prevention



In 1996, the Texas Center was established as a part of the Birth Defects Epidemiology and Surveillance Branch (BDES) of the Texas Department of State Health Services in Austin. The Texas Center's mission is to conduct research studies to understand the causes of specific birth defects.

BDES was established in 1993 as the result of an unusual cluster of anencephaly cases (a type of neural tube defect) that occurred in Brownsville, Texas. Epidemiologic investigations revealed a rate of neural tube defects higher than expected among children born to Hispanic mothers living in South Texas. In recognition that epidemiologic resources are routinely needed to investigate birth defects clusters, the Texas State Legislature passed the Texas Birth Defects Act in 1993, which authorized the establishment of BDES.



Since 1994, BDES has maintained the Texas Birth Defects Registry, a population-based birth defects surveillance system, which is now statewide. Through multiple sources of information, the Registry monitors all births in Texas, approximately 370,000 each year, and identifies cases of birth defects. Children identified through the Registry are referred to appropriate medical and community services.

The Texas Center is in a unique position to contribute to our understanding of what causes birth defects, especially because of the 1,200-mile shared border with Mexico. Health disparities between Texans living along the border with Mexico and those living in non-border areas have long been a concern for public health officials, as well as for those who live and work in the border counties. The majority of border residents are Hispanic and data from this area can be compared with other populations to isolate geographic versus ethnic factors. Since 1997, the Texas Center has contributed information about birth defects cases as well as from healthy "control" families in border counties to the National Birth Defects Prevention Study (NBDPS). The study area for Texas is currently the area known as the Lower Rio Grande Valley, which encompasses Gulf Coast industrial cities such as Corpus Christi, as well as Cameron County, which has experienced some of the country's highest neural tube defect rates and is at the southern-most point in the continental United States.

The Texas Center's staff and collaborators have expertise in the epidemiology of neural tube defects and their associated risk factors, demographic risk factors for birth defects, epidemiology, survey research, and molecular genetics.

In addition to participating in NBDPS, the Texas Center is conducting other research projects and activities, including the following:

- Studying the interaction of metabolic, genetic, and environmental risk factors for certain birth defects of the brain and spinal cord.
- Examining the link between neural tube defects and risk factors such as maternal diabetes, obesity, and dieting behaviors.
- Studying the link between birth defects and certain environmental factors, such as hazardous waste sites, air pollution, pesticides, and water disinfection byproducts.
- Conducting and analyzing results from a telephone survey to examine women's knowledge, attitudes, and practices related to the prevention of birth defects, including folic acid supplementation and alcohol use.
- Analyzing the patterns and risk factors associated with oral clefts and clubfoot in Texas.

## Texas Center's Partners

- Texas A&M University Public Policy Research Institute
- Texas A&M University System Health Science Center
  - Institute of Biotechnology
  - School of Rural Public Health
- University of Texas Health Science Center at Houston
  - University of Texas School of Public Health at Houston
  - University of Texas Medical School at Houston
- Baylor College of Medicine
- University of Texas Southwestern Medical Center at Dallas
- Texas State University - San Marcos
- Texas Office for the Prevention of Developmental Disabilities
- Texas Department of State Health Services, Health Screening Branch

For more information about the Texas Center, visit their website at [www.dshs.state.tx.us/birthdefects](http://www.dshs.state.tx.us/birthdefects).



## Center for Birth Defects Research and Prevention

The Utah Center, established in 2002, is based within the Utah Birth Defect Network (UBDN). UBDN is a program in the Bureau of Children with Special Health Care Needs, Utah Department of Health, and functions collaboratively with the University of Utah Health Sciences Center, Department of Pediatrics. UBDN seeks to prevent birth defects and secondary disabilities among affected children through monitoring, research, education and outreach.

UBDN is a comprehensive statewide surveillance program of major structural malformations among all pregnancy outcomes (live births, stillbirths, terminations and miscarriages). In 1994, UBDN began as a neural tube defect surveillance project. A year later, orofacial clefts and common trisomies were added. In 1999, the program expanded to include all major birth defects.

The Utah Center's staff and collaborators have considerable expertise in molecular and clinical genetics, particularly with respect to heart and limb abnormalities, and epidemiological studies investigating environmental risk factors.

Utah's population census for 2000 was more than 2.2 million with a birth rate 40% higher than the U.S. birth rate; its annual birth frequency is approximately 50,000. Most (77%) of the state's population resides along the urban Wasatch mountain range representing 4 of the 29 counties, with Ogden in the north, Salt Lake City in the middle, and Provo in the south.



In addition to its participation in the National Birth Defects Prevention Study, UBDN is involved in several other projects, including the following:

- Investigating the genetics of heart defects associated with disturbances of left-right asymmetry.
- Investigating nutrient biomarkers and genetics of orofacial clefts in collaboration with Utah State University.
- Evaluating awareness, knowledge and consumption of folic acid among women in Utah using the Behavioral Risk Factor Surveillance System.
- Evaluating health care providers' awareness, knowledge and education of their patients about folic acid.
- Tracking the occurrence of neural tube defects and facilitating recurrence prevention.

- Educating women of childbearing age and health care providers about folic acid.
- Providing statewide birth defect outreach and education to families and health care providers, through information about particular birth defects, medical information, or parent support groups.

## Utah Center for Birth Defects

### Utah Center's Partners

- Division of Medical Genetics, Department of Pediatrics, University of Utah Health Sciences Center
- Division of Pediatric Cardiology, Department of Pediatrics, University of Utah Health Sciences Center
- Division of Maternal-Fetal Medicine, Department of Obstetrics and Gynecology, University of Utah Health Sciences Center
- Primary Children's Medical Center
- Utah State University
- Utah Chapter of the March of Dimes Birth Defects Foundation
- Utah Folic Acid Coalition

For more information about the Utah Center, visit their website at [www.health.utah.gov/birthdefect](http://www.health.utah.gov/birthdefect).



# FUTURE OPPORTUNITIES FOR PREVENTING BIRTH DEFECTS

The establishment of the Centers for Birth Defects Research and Prevention (CBDRP) and the National Birth Defects Prevention Study (NBDPS) is just the beginning of a national effort to broaden and intensify the search for causes of birth defects and to prevent the tragedy that occurs for 120,000 American families each year. With the commitment of more resources, birth defects surveillance could be conducted in more states; factors in our environment that interact with our genes to cause birth defects could be identified; and effective prevention strategies could be developed so that more women can have healthy babies.

## Surveillance

To prevent birth defects, it is essential to know what types of birth defects are occurring, how often they are occurring, and where they are occurring. Currently, three-quarters of the states have a system for tracking birth defects; however, their methods and data sources vary considerably, making it difficult to compile and compare the data. The National Birth Defects Prevention Network has developed guidelines covering the fundamental aspects of developing, implementing, and conducting surveillance for birth defects and using the resulting data. They provide a way of improving the quality of birth defects surveillance data. Surveillance data are the foundation on which research and prevention activities are based.



## Research

The data collected through NBDPS offer an unprecedented opportunity to study the role of gene-environment interactions that may cause birth defects. Certain genes may make a developing fetus more susceptible to environmental exposures that result in birth defects. The rapid expansion in our understanding of the human genome and the technological advances of the past few years have greatly enhanced the potential for the DNA samples collected in



NBDPS. CBDRP is currently funded to carry out the basic elements of NBDPS, but additional resources are needed to support the full potential of the study to analyze the valuable data already collected and identify gene-environment interactions. The NBDPS data also offer the opportunity for more research about the risks of frequently used medications during pregnancy. NBDPS results can provide valuable data about which medications may cause birth defects and which may be safe during pregnancy.

NBDPS provides an outstanding foundation on which we can potentially build with fruitful follow-up studies such as reviewing the types of services received by affected families and long-term outcomes such as special education and speech therapy; supplementing the study interview with a detailed family history questionnaire to identify genetic factors associated with certain birth defects; collecting environmental samples, such as water, soil, or air, and studying the samples for contaminants or changes in the environment that may be linked to birth defects; and collecting and analyzing additional biological samples, such as blood or urine, for biomarkers (indicators) of exposure to toxic substances. All of these research avenues—genetic mapping, environmental sampling, and study of exposure biomarkers—are essential for identifying the very complex factors and mechanisms that lead to birth defects.



## Prevention

Ultimately, surveillance and research activities are translated into concrete strategies to prevent birth defects. In 1992, with evidence from epidemiologic research studies, the U.S. Public Health Service recommended that all women of childbearing age consume 400 micrograms (400mcg or 0.4mg) of folic acid daily to reduce the risk (up to 70%) of having a pregnancy affected by a neural tube defect. This has spurred prevention activities at local and national levels to promote the folic acid message.



The culmination of CDBRP's work is to share research data with various groups and individuals to assist in developing legislation, recommendations, messages, activities, and programs for preventing birth defects. The data can also be used to assess the effectiveness of birth defects prevention activities. As the centers uncover the causes of birth defects and gain more knowledge about how to prevent birth defects, this knowledge will be imparted to various audiences, including legislators, health professionals, and the public.



Centers  
for  
Birth Defects  
Research  
and  
Prevention

