



# Centers for Birth Defects Research and Prevention



U.S. DEPARTMENT OF HEALTH AND HUMAN SERVICES  
Centers for Disease Control and Prevention





**CENTERS FOR  
BIRTH DEFECTS  
RESEARCH &  
PREVENTION**



## FOREWORD

For more than two decades, birth defects have been the leading cause of infant death in the United States. Although more babies with birth defects now survive, many still suffer from illnesses, lifelong disabilities, and social challenges. Because the causes of most birth defects are still unknown, the first step in preventing them is to identify their causes. To achieve this goal, the Centers for Disease Control and Prevention (CDC), the Nation's prevention agency, established the Centers for Birth Defects Research and Prevention in 1996. These Centers comprise some of the Nation's leading birth defects researchers—including those working at CDC as well as at universities and State health agencies. Through unparalleled collaborative efforts, these researchers explore the causes of and risk factors for birth defects. CDC, through its National Center for Environmental Health and other staff, is very pleased to be a coordinating partner in this important public health effort to uncover the causes of birth defects.

The Centers are conducting the National Birth Defects Prevention Study, one of the largest studies of the causes of birth defects ever done, and are also conducting local or regional birth defects research, which is essential for evaluating regional-specific causes of birth defects and addressing local concerns. By building a strong foundation in research, the Centers can identify causes of birth defects and develop effective prevention strategies for families and communities.

A handwritten signature in blue ink that reads "Jeffrey P. Koplan".

Jeffrey P. Koplan, M.D., M.P.H.  
Director  
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The human and economic toll of birth defects is significant and tragic—there are more than 150,000 infants born with serious birth defects in America each year. However, with the creation of the Centers for Birth Defects Research and Prevention (CBDRP) in 1996 and with the adoption of the Birth Defects Prevention Act in 1998, Congress has taken momentous steps forward in the effort to better understand and prevent birth defects in America. The Act, which calls for a National Birth Defects Prevention Study will, for the first time in our history, mobilize America to collect, analyze, and report statistics on 30 different birth defects. These activities will help us understand the root causes of many birth defects. The value of this information will be extremely significant.

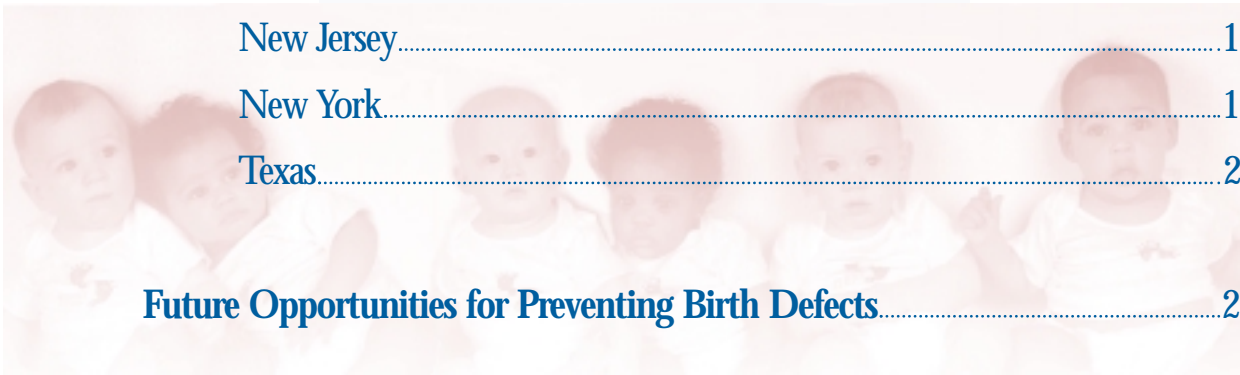
The March of Dimes, dedicated to the prevention of birth defects, is committed to continuing to work closely with CDC and CBDRP. We welcome CBDRP, as well as the National Birth Defects Prevention Network, and all state birth defects surveillance systems to our common struggle. The March of Dimes supported the creation of this booklet because it is an important source of information about birth defects prevention research for policy makers, health care professionals, and the public.

Jennifer L. Howse, Ph.D.  
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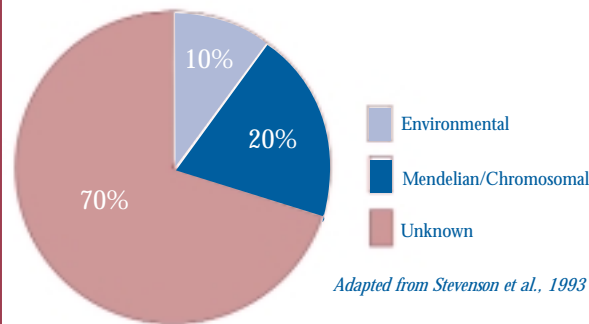


# Introduction

## The Need for Birth Defects Research

**B**irth 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 identifying their causes; however, the causes of about 70% of all birth defects are still 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

**T**o 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 on preventing birth defects. Currently, CDC has established centers in Arkansas, California, Iowa, Massachusetts, New Jersey, New York, and Texas, awarding each center \$800,000 per year for 5 years. The centers were established in states with existing birth defect programs that had nationally recognized expertise in birth defects surveillance and research. CDC's National Center for Environmental Health both coordinates CBDRP and participates in the National Birth Defects Prevention Study (NBDPS) as the eighth study site (Figure 2).

Figure 2: Centers for Birth Defects Research and Prevention



## CBDPR Activities

The establishment of CBDPR has increased the capability of seven 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 many birth defects from occurring. Specifically, CBDPR is 1) participating in NBDPS, 2) conducting center-specific research projects, and 3) enhancing each of its state birth defect surveillance systems.

### *NBDPS*

NBDPS is one of the largest studies 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.

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

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

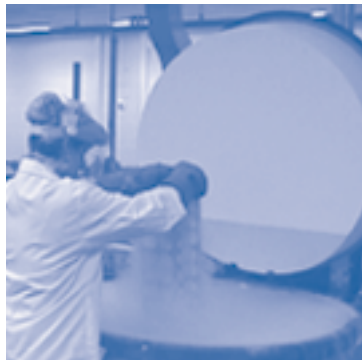
System Affected	Birth Defects Studied
Cardiovascular	Conotruncal heart defects, septal heart defects, obstructive heart defects, atrioventricular septal defects, single ventricle, anomalous pulmonary venous return, heterotaxy, Ebstein malformation
Central nervous system	Neural tube defects, hydrocephaly, holoprosencephaly, Dandy-Walker malformation
Eye	Anophthalmia/microphthalmia, congenital cataracts, glaucoma
Ear	Anotia/microtia
Orofacial	Choanal atresia, cleft lip, cleft palate
Gastrointestinal	Esophageal atresia and tracheoesophageal fistula, intestinal atresia, biliary atresia
Genitourinary	Renal agenesis (bilateral), hypospadias (2nd or 3rd degree)
Musculoskeletal	Diaphragmatic hernia, gastroschisis, omphalocele, limb deficiency, craniosynostosis, bladder exstrophy, cloacal exstrophy, sacral agenesis/caudal regression
Non-system-specific	Amniotic band sequence

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. Collectively, CBDRP will conduct an estimated 12,000 interviews over a 5-year period.



Third, CBDRP is collecting cheek cells from the infants and their parents in order 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 increase the risk of or cause a particular birth defect. A portion of the DNA that is collected from the families will be 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. CBDRP is also using state-of-the-art technology for in-depth studies of the role of specific genetic variants. CBDRP's 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.



# Centers for Birth Defects Research and Prevention

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# Profiles of the Centers for Birth Defects Research & Prevention

## Centers for Disease Control & Prevention

The Birth Defects and Pediatric Genetics Branch of the National Center for Environmental Health (NCEH), 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 multivitamins, diet, medication, maternal health conditions, smoking and alcohol, genetic variation, and disinfection by-products in drinking water.

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

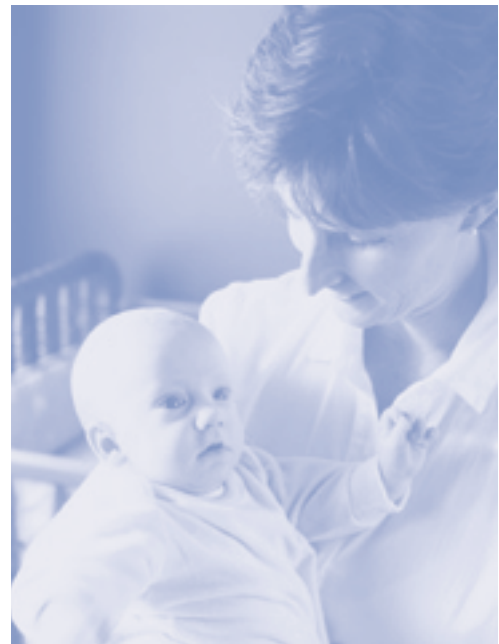
- Working on a campaign with the National Council on Folic Acid to educate women of reproductive age about the need to consume folic acid to reduce the risk of giving birth to children with neural tube defects.



- Evaluating, with Beijing Medical University, the effectiveness of folic acid in reducing the incidence of neural tube defects 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 defect surveillance programs and funding states to promote the use of surveillance data for prevention and intervention activities.

## CDC's Partners

- Battelle Memorial Institute
- Environmental Health Laboratory, NCEH, CDC
- Georgia Department of Human Resources
- March of Dimes Birth Defects Foundation



# Arkansas

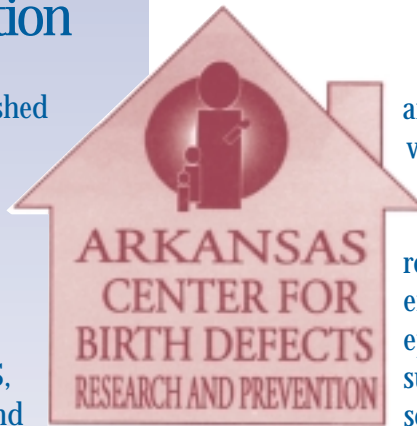
## Center for Birth Defects Research and Prevention

The Arkansas Center was established in 1997 at the University of Arkansas for Medical Sciences (UAMS) in Little Rock. The Arkansas Center is a collaboration between the Arkansas Reproductive Health Monitoring System (ARHMS), the Department of Pediatrics at UAMS, the Arkansas Department of Health, and the Arkansas Children's Hospital. Its mission is to reduce the prevalence of birth defects and their economic, social, and psychological impact through surveillance, research, and prevention.



Founded in 1980, ARHMS is one of the oldest active birth defect surveillance systems in the United States. ARHMS staff members visit all of the 83 hospitals in Arkansas that provide obstetrical or pediatric care. Arkansas is predominately rural and has the fifth highest poverty rate in the nation. Specialized health services, such as clinical genetic services, are available only in Little Rock or at satellite clinics affiliated with the Arkansas Children's Hospital.

ARHMS plays a key role in the Center's daily operations. Since the establishment of the Arkansas Center, ARHMS has expanded from a surveillance system with a small staff to a research



and prevention organization with clinicians and scientists.

These professionals focus on three interdisciplinary research themes: 1) gene-environment interactions, 2) epidemiology and surveillance, and 3) health services research and community interventions.

Each research group uses a multidisciplinary approach to advance its understanding of the causes of birth defects and their impact on Arkansas and the nation. Clinicians and experts in biochemistry, nutrition, molecular biology, biostatistics, health economics, health services research, and epidemiology combine their skills to advance the Center's research agenda. The Center's partnership with the Arkansas Department of Health facilitates collaboration with public health professionals in maternal and child health, nutrition, health education, and health statistics.

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:

- Studying the interaction of diet- and folate-dependent metabolic pathways on major structural birth defects.
- Investigating health-care utilization and medical costs for children with birth defects.
- Developing a guideline for the evaluation of birth defect surveillance systems.



- Conducting a project to study knowledge and intake of folic acid among women in the Lower Mississippi River Delta.
- Performing intensive intervention efforts targeted at Arkansas women who have had a pregnancy affected by a neural tube defect.

## Arkansas Center's Partners

- Arkansas chapter of the March of Dimes Birth Defects Foundation
- Arkansas Folic Acid Coalition
- National Center for Toxicological Research, United States Food and Drug Administration
- United States Department of Agriculture Delta Nutrition Intervention Research Initiative
- Arkansas Children's Hospital Research Institute

# California

## Center for Birth Defects Research and Prevention

Established in 1996, the California Center is based in the California Birth Defects Monitoring Program (CBDMP) in Emeryville. 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 is utilizing CBDMP's experience in collecting biological samples; investigating gene-environment interaction; and studying heart defects, neural tube defects, and cleft lip and palate. California's large and racially diverse population puts the California Center in a unique position to identify risk factors for birth defects among Hispanics and Asians. 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 it is enabling 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 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 infection contribute to heart or limb birth defects.
- Studying the association between specific birth defects and drinking water contaminants.
- Analyzing data from case-control interview studies to examine the association between environmental exposures and neural tube defects in Mexico and California.

## California Center's Partners

- Children's Hospital, Oakland, California
- Genetic Disease Branch, California  
Department of Health Services
- State Department of Health, Baja  
California, Mexico
- University of Iowa
- University of Nebraska



# Iowa

## Center for Birth Defects Research and Prevention

In 1996, the Iowa Center was established in the University of Iowa's College of Medicine in Iowa City. Its mission is to identify genetic and environmental risk factors for birth defects and to conduct educational activities for the prevention of birth defects. The Iowa Center is a collaboration between the Iowa Birth Defects Registry (IBDR), the University of Iowa Survey and Sampling Center (ISSC), and the University of Iowa's Department of Pediatrics. IBDR is a collaborative effort that began in 1983 between the University of Iowa and the Iowa Department of Public Health. IBDR conducts active, statewide surveillance of birth defects and is providing the Iowa Center with its surveillance data. ISSC recruits study subjects, conducts telephone interviews, and collects the biologic specimens required for the Center's research. The University of Iowa's Department of Pediatrics provides expertise in mutation detection, characterization of genetic variations on large populations, and gene identification and characterization.



Members of the Center's staff have considerable expertise in investigating genes, environmental factors, and potential gene-environment interactions that contribute to the development of orofacial clefts. They also have experience in applying geographic information systems (GIS) to studies of adverse birth outcomes.

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

- Examining the association between compounds in drinking water, particularly trihalomethanes and nitrates, and adverse birth outcomes.
- Studying the prevalence of major birth defects among offspring of Persian Gulf War veterans.
- Applying GIS to IBDR data to help plan efforts to prevent infant mortality and to assess prenatal care patterns in Iowa.
- Investigating genes, environmental exposures and their interactions as risk factors for orofacial clefts.
- Educating Iowa's women about preventing neural tube birth defects by taking folic acid, developing a model for disseminating prevention information, and developing methods to evaluate the effectiveness of the prevention efforts.



## Iowa Center's Partners

- Agency for Toxic Substances and Disease Registry
- Iowa Birth Defects Advisory Committee
- Iowa Newborn Screening Program
- Iowa Regional Genetic Consultation Services
- Iowa Department of Public Health
- Naval Health Research Center
- Iowa chapters of the March of Dimes Birth Defects Foundation



# 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 Unit (SEU), and Brigham and Women's Hospital's Active Malformation Surveillance Program (BWH). The 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 33 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 monitoring system that is statewide and population-based. The Massachusetts Center has been collecting data on birth defects in the eastern part of the state since October 1997 and throughout the entire state since September 1998.

SEU and BWH bring to the Massachusetts Center collaboration more than 20 years of combined experience in birth defects research. The Massachusetts Center will also draw on the expertise of and foster communication among the region's strong network of clinicians and researchers. The Center's areas of expertise include surveillance methodology; pediatric, reproductive, and social epidemiology; heart



defects and drug teratogenicity research; and experience conducting health service needs assessment.

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

- Studying the association of over-the-counter medications and birth defects.
- Studying the association of antiepileptic drugs and birth defects.
- Examining the association of socioeconomic status 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.
- Studying the relationship between substances involved in folic acid metabolism and neural tube defects.
- Developing methods of birth defect classification and surveillance.

## Massachusetts Center's Partners

- Genetics and Teratology Unit, Massachusetts General Hospital
- Massachusetts chapters of the March of Dimes Birth Defects Foundation
- Office of Health Statistics and Research and Office of Environmental Health Assessment, Massachusetts Department of Public Health



# New Jersey

## Center for Birth Defects Research and Prevention

In 1997, the New Jersey Center was established as part of the Birth Defects Monitoring Program (BDMP) of the State Department of Health and Senior Services (DHSS) in Trenton, New Jersey. The mission of BDMP is to identify children with birth defects and other special health needs in order to 1) provide the state with information for epidemiologic studies, health planning, research, and evaluation, and 2) plan for and provide children who have birth defects with early and easy access to services. The New Jersey Center uses the New Jersey Birth Defects Registry to identify eligible cases for NBDPS. The Registry, which was established in 1928, is the oldest state registry in the country and identifies nearly 5,000 newborns each year who have a birth defect and another 5,000 who have other special health needs. All registered children are then linked to Special Child Case Management Services, another arm of the DHSS Division of Family Health Services.

New Jersey is an ideal setting for a center because it is the most densely populated state in the U.S. and is ethnically and racially diverse. The Center's location within DHSS has been advantageous for both the Center and BDMP. Other divisions and offices of the DHSS and DHSS' partners have all helped by creating awareness and support for the Center's programs within the medical community and the public. The formal and informal networks with state maternity hospitals and county case managers have helped facilitate data collection. New Jersey Center staff and collaborators have expertise in areas such as environmental health, fetal medicine, endocrine disruptors, and HIV/AIDS among pregnant women.



The New Jersey Center, in turn, has improved the accuracy of the Registry information and heightened the importance of the Registry in the medical community. It has enabled BDMP to 1) develop a birth defects research agenda, building on the database that has been carefully developed over the years, and 2) establish collaborations among a variety of people and agencies with interest in birth defects.



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

- Studying the link between endocrine disruptors in the environment and male reproductive anomalies.
- Studying the effects of maternal use of HIV medications on their offspring.
- Establishing a Fetal Abnormalities Registry to determine more accurately the occurrence of birth defects.
- Studying demographic differences, particularly race and ethnicity, in the distribution of birth defects.



- Studying geographic clustering of heart defects.
- Examining the role of maternal hypercoagulability in birth defects.

## New Jersey Center's Partners

- Children's Hospital of New Jersey at Newark  
Beth Israel Medical Center
- Division of AIDS in the New Jersey  
Department of Health and Senior Services
- Eagleton Institute of Politics at Rutgers, the  
State University
- University of Medicine and Dentistry of New  
Jersey, Robert Wood Johnson Medical School
- University of Medicine and Dentistry of New  
Jersey, New Jersey Medical School
- New Jersey chapters of the March of Dimes  
Birth Defects Foundation
- New Jersey Maternal Child Health and  
Perinatal Consortia



# New York

## 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 Wadsworth Center for Laboratories and Research. 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 defect 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 Wadsworth Center houses the Department of Health's



comprehensive public health laboratory, which combines basic and applied research and education in biomedical and environmental sciences. The New York Center's areas of expertise include geographic information systems (GIS), occupational exposure studies, environmental epidemiology, folate metabolism, mouse genetics, 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 folate derivatives and other factors involved in neural tube defects.
- Studying the role of genes and gene-environment interactions in limb development and oral clefts.
- 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 the relationship between asthma medication and heart defects.
- Studying the effects of maternal use of HIV medications on their offspring.
- Improving surveillance of neural tube defects and fetal alcohol syndrome cases.
- Establishing a registry for hemangiomas to enable researchers to conduct studies of causes and treatments.
- Studying the relationship between mutations in androgen receptor genes and hypospadias.

## New York Center's Partners

- Division of Genetics, Children's Hospital of Buffalo
- Eastman Dental Center, University of Rochester
- New York chapters of the March of Dimes Birth Defects Foundation
- Pediatric Otolaryngology Department, Children's Hospital of Buffalo
- Regional Medical Genetics Center, Westchester, NY
- Pregnancy Risk Network of New York



# Texas

## Birth Defects Research Center

In 1996, the Texas Center was established as a part of the Texas Birth Defects Monitoring Division (TBDMD) of the Texas Department of Health in Austin. The Center's mission is to conduct research studies to understand the causes of specific birth defects and to enhance the surveillance and prevention of birth defects.



330,000 each year, and identifies cases of birth defects. Children identified through the Registry are referred to appropriate medical and community services. Cases identified in four public health regions are eligible to participate in the National Birth Defects Prevention Study (NBDPS).

TBDMD 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 higher than expected rate of neural tube defects 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 TBDMD.

Since 1994, TBDMD has maintained the Texas Birth Defects Registry, a statewide, population-based birth defects surveillance system. Through multiple sources of information, the Registry monitors all births in Texas, approximately



Establishing a Texas Center has enabled TBDMD to use the registry data to conduct case-control studies to describe causes of birth defects due to variations in ethnicity, genetics, and geography. It has also enabled TBDMD to partner with universities to study factors that are unique to Texas and may be associated with birth defects, such as environmental exposures due to poorly regulated industries along the Texas-Mexico border. The 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 metabolic, genetic, and environmental factors associated with certain birth defects of the brain and spinal cord.
- Studying the relationship between chemicals formed during water chlorination and several types of birth defects.



- Examining survival patterns of infants with birth defects.
- Studying the feasibility of implementing a statewide strategy for preventing birth defects among women who have previously had a child with birth defects.
- Evaluating and increasing awareness among health care providers that consuming folic acid can help prevent neural tube defects, and integrating education about folic acid into standards of care for women of childbearing age.
- Periodically conducting a telephone survey to examine women's knowledge, attitudes, and practices related to the use of vitamins containing folic acid to prevent neural tube defects.
- Studying the blood levels of folic acid among nonpregnant Texas women.

## Texas Center's Partners

- Texas A& M University, Public Policy Research Institute
- Texas chapters of the March of Dimes Birth Defects Foundation
- University of Medicine and Dentistry of New Jersey, Robert Wood Johnson Medical School
- University of Nebraska Medical Center, Center for Molecular Genetics
- University of Texas School of Public Health in Houston
- University of Texas Health Science Center at San Antonio



# Future Opportunities for Preventing Birth Defects

The launching 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 the causes of birth defects and prevent the tragedy that occurs for 150,000 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

In order 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 begun developing standards for birth defects surveillance that will be used to guide new surveillance systems and to improve existing ones. Surveillance data are the foundation on which research and prevention activities are based.

## Research

The data collected through NBDPS will 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 mapping of all human genes through the Human Genome Project will be particularly useful to researchers studying variations in genes that play a role in birth defects. CBDRP is currently funded to carry out NBDPS, but additional resources will be needed to support the study of gene-environment interactions and data analysis. CBDRP could also collect environmental samples, such as water, soil, or air, and study the samples for contaminants or changes in the environment that may be linked to birth defects. CBDRP is also interested in collecting and analyzing 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.



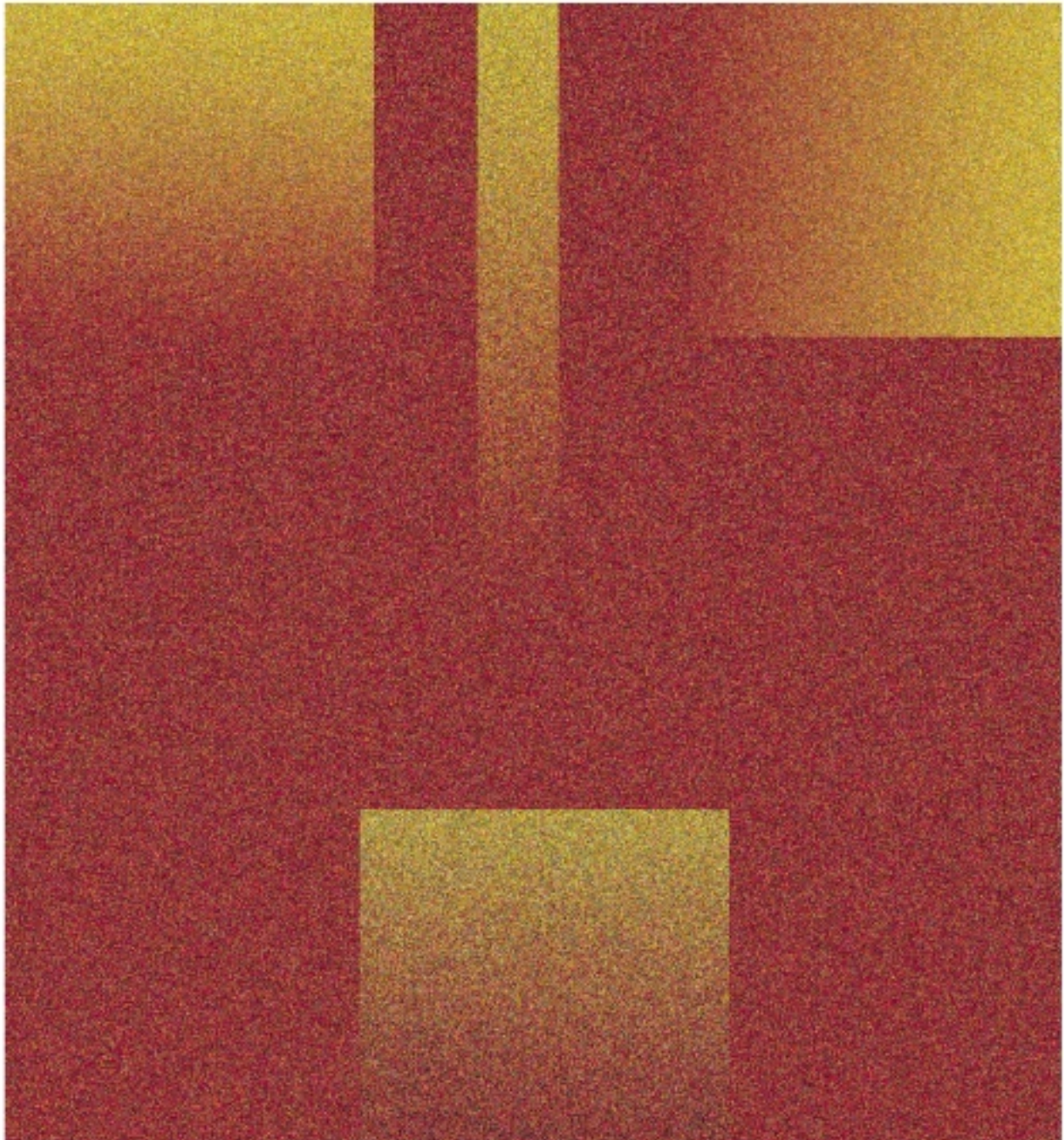
## Prevention

Ultimately, surveillance and research activities are translated into concrete strategies to prevent birth defects. In 1992, with solid 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 CBDRP's work is to share its 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 defect prevention activities. As CBDRP uncovers the causes of birth defects and gains more knowledge about how to prevent birth defects, this knowledge will be imparted to various audiences, including legislators, health professionals, and the public.







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