The Importance of Birth Defects Research:
A Personal Message by Dr. Marjorie Royle, New Jersey Center, National Birth Defects Prevention Study

I am a research psychologist and the stepmother of a child with a birth defect. When I heard about the National Birth Defects Prevention Study, I jumped at the chance to work on it. I saw it as a way to combine my personal and professional lives. After I started, I learned what an important study it is. For years to come, people will talk about what was learned from this study about the causes of birth defects and how to prevent them.

For the first time ever, researchers will be able to study possible causes of specific defects. Some of the common defects such as cleft palates have been studied before. Even for these, the size of the study sample will allow researchers to answer questions about the way different factors work together to cause or to prevent the defects, questions they have never been able to study before.

One thing I’ve learned already is that the process by which two cells develop into a baby is a long, amazing one, with many places where something can go wrong. When something does go wrong, parents often ask what they did to cause it. Many of us remember when drugs such as thalidomide and DES that were taken by pregnant women for medical problems resulted in damage to their babies. But in most cases, the causes of birth defects are a lot more involved than that. We’re just beginning to learn that some people have particular genes that, if they are passed on to the baby, may cause problems, but only if other factors are present or absent, such as something in the environment, or not enough of a particular nutrient in the mother’s diet. During a pregnancy, some time periods are more critical than others. Something will affect the baby’s development at one time, but have no effect a few weeks later. With so many factors, no one thing by itself caused the defect, but many things may have influenced it.

Very early in my research career I learned that research takes longer than you thought it would, and good research takes even longer. So be patient with us. We would love to have some answers for you in this first newsletter, but we don't. We will continue to gather information from mothers for several more years. Researchers from different Centers already have planned 48 different studies using this information, and some of those studies will be starting soon. I hope that 10 years from now, researchers will still be using this information to answer new questions. In the meantime, we’re working as hard and as fast as we know how to get some answers.

Dr. McGehee of the Arkansas Center is one of the researchers studying birth defects.
The miracle of life...fortunately, most babies are born healthy. Yet, one in every 33 babies is born with a birth defect. We know that certain genetic and environmental factors cause about 30% of birth defects. Scientists cannot explain the causes of the other 70%. In the past, it has been hard to identify the causes of birth defects because many are rare. However, in 1996 and 1997, eight Centers for Birth Defects Research and Prevention were created to combine efforts to find the causes. These Centers are located in Arkansas, California, Georgia, Iowa, Massachusetts, New Jersey, New York, and Texas. The Centers draw their participants from a pool of 482,000 births each year.

National Birth Defects Prevention Study

The eight Centers are participating in the National Birth Defects Prevention Study (NBDPS). This study is one of the largest ever conducted on the causes of birth defects. The Centers will interview more than 16,000 women. These women are mothers who have had babies or pregnancies affected by birth defects as well as mothers of babies with no birth defects. To date, the Centers have completed over 6,000 interviews. Mothers are also asked to collect cheek cells from their families. These cheek cells will be used for genetic studies.

The Centers' leading scientists will use the information from the NBDPS to help explain why certain birth defects occur. These include defects of the heart, brain and spine, limb, eye, ear, muscle, skeleton, urinary tract, and abdomen. Some research questions include:

- Does smoking, diet, tap water, or some medicines cause birth defects?
- Does taking multivitamins help prevent some types of birth defects?
- Do factors in the environment interact with our genes to cause birth defects?

Prevention

As the Centers learn more about how to prevent birth defects, we will share our findings with the public and with other scientists. Studies like ours found a link between the B vitamin "folic acid" and defects of the baby's brain and spine. Folic acid could help prevent up to 70% of these defects. We now recommend that all women who can become pregnant get 400 micrograms (400 mcg or 0.4 mg) of folic acid every day.

Your participation and continued support are vital to the Centers' work to identify causes of birth defects. We will keep you updated on the progress of the Centers' activities.
Profiles of the Centers for Birth Defects Research and Prevention

Georgia Center, CDC

The Centers for Disease Control and Prevention (CDC) is the Georgia study site for the National Birth Defects Prevention Study. It also coordinates the Centers’ activities. CDC brings many years of experience to birth defects research. Since 1967, scientists and doctors at CDC have carried out studies related to birth defects. These studies look at links between birth defects and vitamins, tap water, diet, smoking, alcohol, medications, and genes.

Arkansas Center

The Arkansas Center is based at the University of Arkansas for Medical Sciences (UAMS) in Little Rock. The Arkansas Center is made up of staff from 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 social and psychological impact through surveillance, research and prevention. ARHMS was founded in 1980. It is one of the oldest state birth defect systems in the United States. Its staff visits all the hospitals in Arkansas that have health services for pregnant women and their babies. Special health services are mainly found in Little Rock or at clinics related to the Arkansas Children's Hospital.

The Center has three research teams working to find the causes of birth defects. These teams focus on gene-environment interactions, epidemiology and surveillance, and health services research.

California Center

The California Center is based within the California Birth Defects Monitoring Program (CBDMP) in Oakland. Since 1982, CBDMP has maintained a birth defects registry and has conducted studies to identify the causes of birth defects. California’s diverse population enables the Center to study risk factors for birth defects among Hispanics and Asians. Some of their research questions are:

- Why do Hispanics have a higher risk for defects of the brain and spine?
- Why is folic acid not as helpful to Hispanics in preventing birth defects?
- What role do genes and nutrition play in birth defects risk?

Iowa Center

The Iowa Center is based in the Colleges of Medicine and Public Health at the University of Iowa in Iowa City. The Iowa Center is made up of staff from the Iowa Birth Defects Registry, the Iowa Survey and Sampling Center, and the University of Iowa Department of Pediatrics.

The Registry monitors the rate of birth defects. It also provides the Center with birth defects data. The Survey and Sampling Center recruits study subjects, conducts telephone interviews, collects cheek cell samples and conducts analyses of environmental data. Scientists in the Department of Pediatrics conduct analyses of genetic data.

Massachusetts Center

The Massachusetts Center is made up of staff from the Massachusetts Department of Public Health’s Bureau of Family and Community Health, Boston University’s Slone Epidemiology Unit, and Brigham and Women’s Hospital’s Active Malformation Surveillance Program.

The Center’s staff has more than 20 years of combined experience in birth defects research. Its areas of expertise are surveillance techniques, heart defects and drug research, and health service assessment.
New Jersey Center

The New Jersey Center is part of the State Department of Health and Senior Services in Trenton. The Center uses the New Jersey Birth Defects Registry to identify babies with birth defects for the study. This registry was created in 1928 and is the oldest state registry in the country. Each year the registry identifies about 5,000 babies with birth defects and 5,000 babies with other special health needs. All registered children are referred to Special Child Case Management Services.

New Jersey is the most densely populated state in the United States. It is ethnically and racially diverse. The Center staff’s areas of expertise include environmental health, fetal medicine, endocrine disruptors, and HIV/AIDS among pregnant women.

New York Center

The New York Center is part of the New York State Department of Health’s Congenital Malformations Registry and the Wadsworth Center for Laboratories and Research. This registry is one of the largest birth defect registries in the nation. It covers about 270,000 births each year.

The Center staff’s areas of expertise include geographic information systems, exposure studies, environmental research, folate metabolism, genetics, and social factors associated with birth defects.

Texas Center

The Texas Center is located within the Texas Department of Health in Austin. The Department of Health founded the Texas Birth Defects Registry in 1994. This registry covers all births in Texas or about 330,000 births each year. The Center uses the registry data to carry out different studies. These studies often look at the causes of birth defects associated with ethnicity, genetics, and environment. For example, one study is looking at how genetics, metabolism, and the environment may cause certain birth defects of the brain and spinal cord.

The Center staff’s areas of expertise include research on defects of the brain and spine, demographic factors related to birth defects, survey research, and genetics.
What Steps Can Women Take for Healthier Babies?

While the cause of most birth defects is unknown, a woman can increase her chance of having a healthy baby. Many birth defects happen very early in pregnancy, sometimes before a woman even knows she is pregnant. Remember that about half of all pregnancies are unplanned. Therefore, every woman should:

- Take a multivitamin that has 400 mcg of folic acid in it every day.
- Have regular medical check-ups.
- Talk to her health care provider about any medical problems and medicine use (both over-the-counter and prescription). Ask about avoiding any substances at work or at home that might be harmful to a developing baby.
- Keep vaccinations (shots) up-to-date.
- Eat a healthy, balanced diet.
- Avoid eating raw or under cooked meat.
- Avoid alcohol, tobacco, and street drugs.

While pregnant:

- Keep up these healthy habits.
- Get early prenatal care and go to every appointment.
- Avoid using hot tubs and saunas.
- Have x-rays done only if necessary.

Information on Birth Defects

The March of Dimes Birth Defects Foundation is a national agency dedicated to preventing birth defects, low birth weight, and infant death. Its website links, "health library" and "resource center," have a wealth of information. You can get a free copy of "Mama--Your Guide to a Healthy Pregnancy."
Toll-free number: (888) 663-4637
Internet: www.modimes.org

CDC's Division of Birth Defects, Child Development, and Disability and Health site provides information on CDC's birth defects and childhood disabilities activities.
Toll-free number: (888) 232-6789
Internet: www.cdc.gov/nceh/cddh

National Organization for Rare Disorders, Inc. (NORD) is a group of more than 140 voluntary agencies serving people with rare disorders and disabilities. This site has an A to Z list of rare disorders.
Toll-free number: (800) 999-6673
Internet: www.rarediseases.org

Referral and Support Sources

The Alliance of Genetic Support Groups was founded to help individuals and families affected by genetic disorders. The site has a search directory by genetic condition, organization name, and available services.
Toll-free number: (800) 336-GENE
Internet: www.geneticalliance.org

The National Information Center for Children and Youth with Disabilities provides information and referral on disabilities and related issues.
Toll-free number: (800) 695-0285
Internet: http://www.kidsource.com/NICHCY/index.html

Spina Bifida Association of America (SBAA) works to prevent spina bifida and improves the lives of all affected. This site has a lot of general information about spina bifida.
Toll-free number: (800) 621-3141
Internet: www.sbaa.org

The Cleft Palate Foundation site has a wealth of information for parents of babies with cleft lip and cleft palate.
Toll-free number: (800) 24-CLEFT
Internet: www.cleft.com/cpf/cpffrm.html
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