

National Birth Defects Prevention Study Participation Update

The NBDPS is well on its way. To date, the Centers have interviewed over 15,000 women and have collected cheek cell samples from over 5,500 families. Scientists are starting to look at the data to find some answers to questions about the causes of birth defects. We have just added two new Centers. These Centers, located in North Carolina and Utah, have progressed rapidly and are now interviewing women.



North Carolina Center for Birth Defects Research and Prevention

The North Carolina Center is a joint effort between the University of North Carolina, School of Public Health at Chapel Hill (UNC) and the North Carolina Division of Public Health (NCDPH). The Birth Defects Monitoring Program within the NCDPH tracks infants born with birth defects through its statewide system. UNC is known world-wide as a leader in epidemiologic research and training. This partnership will enrich the birth defects projects. The Center staff has expertise in the study of embryology, teratology, and medical genetics.



Utah Center for Birth Defects Research and Prevention

The Utah Center is part of the Utah Department of Health's Birth Defect Network (UBDN). It is made up of staff from the Utah Department of Health, the University of Utah Department of Pediatrics and Utah State University. The Center has expertise in genetic and environmental risk factors.

The UBDN was founded in 1994. It covers all births to women who are residents of Utah; UBDN staff visits all hospitals in Utah. The mission of the UBDN is to monitor birth defects in the state, determine risk factors and prevention strategies, and assist families and their providers in preventing disabilities. The UBDN provides referral services to families as needed. The UBDN also supplies the Center with birth defects data.



Utah researchers in a meeting to discuss the study

NBDPS News is a periodic newsletter of the Centers for Birth Defects Research and Prevention.

The purpose of this newsletter is to inform the public of the Centers' activities and current news about birth defects.

IN THIS ISSUE...

Page	Content
1	NBDPS Update
2 - 3	Center-specific Research Studies
3, 6	Research Plans for the NBDPS Data
4	Recurrence Risk of Birth Defects
5	A Mom's Thoughts on Having a Baby with a Birth Defect
6 - 7	Resources on Birth Defects and Related Topics
8	Directory of the Centers for Birth Defects Research and Prevention

Center-specific Research Studies

In the 2002 newsletter, we included studies from four Centers. In this issue, we are featuring local studies from four other Centers.



Arkansas: The Importance of Folic Acid in Preventing Birth Defects

The U.S. Public Health Service recommends that all women who could become pregnant get 400 µg (0.4 mg) of folic acid every day. It is important that women take this vitamin before and during pregnancy. This could prevent up to 70% of some types of serious birth defects.

Many women have heard about folic acid, but do not know if they should take it. They report that they would take folic acid if their doctor recommends it. A study at the Arkansas Center is being done to find out if this is true. We are studying the influence of doctors and nurses who tell women to take folic acid daily.

In this study, women patients were given facts about folic acid as part of their routine doctor's visits. The patients received information in two ways. One group was given pamphlets on folic acid. The other group was told by their doctor and a clinic nurse to take folic acid.

So far, we have found that many women do not know about the link between folic acid and birth defects. Women who do not want to have children are hesitant to take this B-vitamin. We also found that a very brief message by the doctor almost doubled the percentage of women taking folic acid regularly from 38% to 66%. It is reassuring to know that doctors who spend just a little time sharing the message about folic acid can have a very big impact on their patients. To find out more about the study results, call 1-877-662-4567.



Massachusetts: Drug Use in Pregnancy

In the 1960s, the drug thalidomide was found to cause missing limbs in children of pregnant users. Since then, there has been concern about other drug use in pregnancy. About 85% of women in the U.S. take at least one drug during pregnancy. Some women have conditions that require them to take medications. But, the most commonly used drugs during pregnancy are over-the-counter products for pain, headache, coughs, colds, flu, and allergies. Unfortunately, the risks and safety of most drugs in pregnancy have not been clearly defined. Researchers at Boston University are studying all types of drugs in pregnancy. These include prescription, over-the-counter products, and herbs.

A recent study looked at a group of drugs that interfere with the body's ability to properly use folate (one of the B vitamins). We found early pregnancy use of these drugs doubled the risks of neural tube defects, oral clefts, and heart defects. Another recent study looked at use of erythromycin (using this antibiotic in newborn babies had earlier been found to increase the risk of pyloric stenosis). We found that late pregnancy use of this antibiotic was not higher in mothers of babies with pyloric stenosis. We will continue to study the risks and safety of drug use in pregnancy. In particular, we will look at genetic factors that alter the metabolism of certain drugs that might affect birth defect risks. To find out more about the study results, call 1-888-302-2101.



New York: Evaluating a Screening Test for Heart Defects in Infants

Detecting and treating infants with heart defects have improved a great deal over the last 15 years. But these defects still cause large numbers of deaths and illnesses. Some of the heart defects are not caught right away in the first few days after birth but may become life threatening as the infant adjusts to life outside the womb. One study found that 1 in 10 infants with heart defects who died in the first year did not have a diagnosis of a defect before death. This increases to 1 in 4 for those infants who died during the first week of life.

Many newborns are going home within 48 hours. If clinical signs appear a few days after birth, many of these

-- continued on page 3

Center-specific Research Studies

-- continued from page 2

babies will already be at home and their symptoms might not be detected. A test, called pulse oximetry, might be used to screen infants in the early days to detect any problems before they become life threatening. This is a simple test that checks the amount of oxygen in the baby's body. It has been shown that normal term infants have oxygen levels within a narrow range. Babies with some types of heart defects have lower oxygen levels. This test might be used to promptly detect infants with undiagnosed heart defects. If problems can be detected early enough, then we can get the infants into treatment earlier and possibly reduce the death rate. We conducted a study to find out the usefulness of the test. So far, we have screened 11,281 newborns, and found out 3 had a serious heart defect. We found that 75% of infants who screened positive had a heart defect, while almost 100% of those who screened negative did not have a heart defect. These are promising results, and we hope to do further studies.

Reference: Koppel RI, Druschel CM, Carter T, Goldberg BE, Mehta PN, Talwar R, Bierman FZ. Effectiveness of pulse oximetry screening for congenital heart disease in asymptomatic newborns. *Pediatrics* 2003;111(3):451-455.



Texas: Survey of Texas Women's Awareness of Birth Defects Prevention

Every four years, Texas scientists conduct the Texas Women's Health Survey. This is a short phone survey of health knowledge, thoughts and behaviors. The questions are related to the prevention of birth defects. In 1997, we surveyed 1,200 Texas women. In 2001, we asked another 1,200 women the same questions.

During each time period, we asked women about their folic acid knowledge and intake of vitamins and cereals with folic acid. We also asked about prenatal care and birth defects beliefs. Women also told us about their experience and awareness of Fetal Alcohol Syndrome. After completing the survey, we gave the women a packet of materials on birth defects or a phone number to call for more information.

During 2003, we looked at the results from both time periods. We saw changes in Texas women's knowledge and awareness. Some of these changes can help reduce the risk of birth defects. Awareness of folic acid increased from 66% to 78% among women in the survey. However, only 33% of women from each time period reported

taking a vitamin with folic acid every day. We are concerned about the gap between the awareness and use of folic acid. This shows that more work is needed to prevent birth defects. By repeating the survey every few years, we can track changes in these trends. To find out more about the study results, call 1-888-844-4633.



Research Plans for the NBDPS Data

Now that the Centers have several years of data, researchers can start to analyze the combined data from all the Centers. Some defects are more common than others. The first projects will start with the common defects because there are enough complete interviews. Some common defects are cleft lip, cleft palate, some types of heart defects, and hypospadias (a defect of the penis).

Past studies have shown us that a birth defect is usually caused by a combination of many factors. For example, a certain type of gene plus smoking plus intake of a medication might occur together. And, these exposures combined might cause a birth defect. Very few single exposures alone are thought to be enough to cause a birth defect.

So, where do we start? In the NBDPS, we are looking at exposures during pregnancy between two groups. We are comparing the information from interviews with women who had babies or pregnancies affected by a birth defect to those who had babies born without a birth defect. There are many types of exposures that we are considering. Examples of exposures that might occur during pregnancy are: infections, smoking, use of a medication, and intake of certain nutrients. Some exposures might make it more likely to have a baby with a certain type of birth defect. Others might make it less likely. And, some exposures might not have any connection with a birth defect.

-- continued on page 6

Recurrence Risk of Birth Defects: "Will I have another child with a birth defect?"

When a child is born with a birth defect, parents are busy dealing with the present. They are usually not thinking about future childbearing and the chance of having another child with a birth defect. They're asking basic questions such as "What is this defect? What went wrong?" If the baby is sick, the most important thing is taking care of the baby. But, later on, a parent may start thinking about other issues.

Parents may want to know when the birth defect occurred during pregnancy. Of course, they are also probably wondering what caused it. Some common things they may wonder about are diet, illnesses or medications. The NBDPS is trying to answer some of those questions.

Parents often ask whether a birth defect is common or rare. They may ask if it is "as rare as a lightning strike," or "something you see every week at the hospital." Health care providers know the frequency of most birth defects. For example, cleft palate occurs in about 1 out of every 2,000 newborns.

It is natural for parents to look beyond the present and start to wonder about their child's future. At some point, parents may wonder whether they could have another child with a birth defect. We call this "recurrence risk."

What factors affect the chance of a birth defect happening again?

- The specific type of birth defect
- Whether the exact same (or similar) defect is present in a parent or sibling
- Whether there is only one defect or there are other defects present
- Whether a child has a specific "syndrome." This refers to a group of defects or features that occur together.

The parents or the child's primary care doctor may want the child to be seen by a specialist such as a clinical geneticist, a doctor who specializes in birth defects and genetic disorders. This specialist can give parents more complete information about their child's birth defects. In order to provide precise recurrence risk, we need to have complete information. For example, the recurrence risk for heart defects can vary depending on

the situation. If a child was said to have a:

- Heart defect, the chance of having another child with a heart defect would be about 2–4%. This can be written also as 2–4 out of 100, or 1–2 out of 50.
- Very rare heart defect such as a single ventricle, then the risk is even lower (less than 1%).
- Heart defect which is part of a syndrome such as the Smith-Lemli-Opitz syndrome, then the chance of having a child with a heart defect depends on the chance of having a child with the syndrome. In this case, the risk is 25% (1 out of 4).
- Heart defect called truncus, and the child and mother both had the chromosome defect called 22q11 deletion, then the risk of having another child with a heart defect would be based on the 22q11 deletion. The risk for this family would be 50% (1 out of 2).

In general, a woman who has had a child with a birth defect can take steps to reduce the risk of it happening again. First, she should meet with her health care provider to plan the next pregnancy. The health care provider may also want her to talk to a genetic counselor or a clinical geneticist.

A genetic consultation would include many things related to the child's birth defect. This discussion would go over the family history in great detail and carefully review the previous pregnancy. Then, the woman would receive information about the future pregnancy, such as choices for possible testing and recurrence risk.

Second, all women of childbearing age should get 400 µg (0.4 mg) of folic acid every day. However, for women who had a previous child with some types of neural tube defect, the recommended dose of folic acid is higher (4.0 mg); this is ten times the usual amount. To get this amount, they need to talk to a doctor.

Having a child with a birth defect has a different impact on every family. Making a decision about having more children is a personal one. In general, the first step is to get as much information as possible from your health care providers.

NOTE: Readers should not think that this brief information is medical advice. Please talk to your doctor for questions about yourself.

A Mom's Thoughts on Having a Baby with a Birth Defect

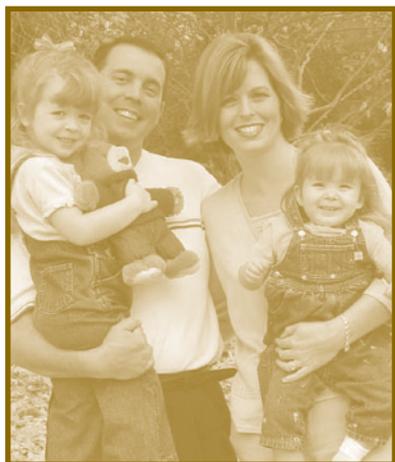
By Julie Mayberry

When I am pregnant, I am a fanatic about doing everything I can to ensure the healthiest start in life for my baby. I read every pregnancy book and magazine. I stop drinking caffeine. I don't color my hair or take hot baths. I eat the healthiest meals and I take 400 micrograms of the B vitamin folic acid every day. Research has shown 400 micrograms of folic acid taken every day, beginning three months before pregnancy and during pregnancy, will reduce the chances of having a child with a neural tube defect, like spina bifida, by 50–70 percent. In my case, it didn't.

At 19 weeks of my second pregnancy, my husband and I were excited because we were having an ultrasound to learn the sex of our second child. We never imagined the test would reveal anything more dramatic. We were told our baby would be born with spina bifida. (Spina bifida occurs when the neural tube that protects the spinal cord does not close properly. This is typically by the 29th day of the pregnancy. Affected children have varying degrees of lower body paralysis and bladder and bowel problems, among other side effects.)

My first thought when I heard the news was “This is my fault.” I was taking folic acid; however, at the time I got pregnant, I had just lost my job. My world was turned upside down and I was not in a routine. I think I forgot to take my vitamin for a few days.

Every doctor I have talked with has told me missing a few doses probably would not have made a difference in the health of my baby. Regardless, it would have made a difference in my emotional state.



A Mother's Responsibility

See, I wish that I definitely knew I had taken at least 400 micrograms of folic acid every day. Then I would know I had done everything I could to bring the healthiest baby into this world. As it stands now, I will always feel a guilt I would never want any other woman to feel.

That is why it is so important for any woman of childbearing years, whether intending to get pregnant or not, to take a daily vitamin fortified with folic acid. In Arkansas, the March of Dimes has conducted studies that show 75 percent of women in childbearing years know about folic acid, but only 31 percent take it daily. I hope those women who don't take the supplement never hear the same words from their ultrasound technician that I did.

I'll be honest. There was a great deal of mourning after I was told the news. I had to realize that my daughter may never be able to walk, have children, or live on her own. But I was reminded by my father-in-law that Helen Keller faced numerous challenges too, and look how many people she inspired.

My second daughter was born Dec. 26, 2001. She is now a toddler and does have movement in her legs. We have hope that she will walk one day, but she will likely need some help in doing so. She has numerous other medical side effects and spends a great deal of time at doctors' offices and at therapy.

Do I wish she did not have spina bifida and could run and play like other toddlers? Yes. But would I change who she is? No. She is beautiful and perfect in every way to me. She has been such a blessing to our family. God chose my husband and me to take care of this special child for a reason. Maybe it's so I would write this column.

Julie Mayberry is a former Arkansas television news anchor. She and her husband are also publishers of two newspapers, The East Ender and The Spirit of Saline County. They live in Arkansas with their children.

Editor's note: This article has been published in the *Little Rock Family Magazine*. The author has allowed us to reprint it here with her name. The privacy of study participants is important to us. We will not name anyone in the study in any report or publication unless we have been given permission to do so.

Research Plans for the NBDPS Data

-- continued from page 3

Here are a few areas where the Centers will first focus their research efforts.

- **Smoking and birth defects**

One research project is assessing smoking during pregnancy and the birth defect hypospadias. Another project is looking at cleft lip and cleft palate and exposure to second hand smoke during pregnancy. We do not know the results of these studies yet. One should not assume that because we are studying certain exposures that it means we know there is a link. We do not know whether or not they are related, and that is why we are studying them.

- **Folic acid and birth defects**

Many studies have shown that folic acid, a B vitamin, lowers the chance of having a baby with spina bifida or anencephaly. These are serious birth defects of the brain and spinal cord. Researchers in our study are looking at the impact of folic acid intake on other birth defects.

We will start with cleft lip, cleft palate, and some types of heart defects. We want to know if consuming a little or a lot of folic acid will affect a woman's chance of having a baby with one of these defects.

- **Fertility treatments and birth defects**

The use of fertility treatments has become much more common in the United States in the past 10 years. We are comparing women who used fertility treatments with those who did not. We want to know if any types of birth defects occur more often or less often than expected among these women. This information will be useful to those who want to use fertility treatments in the future.

We look forward to sharing the results of these projects in future issues of this newsletter. Please be sure to let us know about any change of address if you would like to continue to receive these newsletters.



Resources on Birth Defects and Related Topics

The resources are provided for your information only. These links do not constitute an endorsement of these organizations or their programs by the Centers, and none should be inferred. The Centers are not responsible for the content found at these web sites.

Air Lifeline coordinates free air transportation for people in need.

Phone: (877) AIR-LIFE

Web site: <http://www.airlifeline.org/a1/servlet/visit>

The **Family Voices** web site provides information for friends and families of children with special needs.

Address: 3411 Candelaria NE, Suite M, Albuquerque, NM 87107

Phone: (888) 835-5669 or (505) 872-4774

E-mail: kidshealth@familyvoices.org

Web site: www.familyvoices.org

The **Internet Resources for Special Children**

web site has useful resources and links with a focus on children with disabilities and other health-related disorders.

Web site: www.irsc.org



The **Alliance of Genetic Support Groups** web site has resources on genetics. It also has a toll-free helpline for questions on genetics and genetic resources.

Address: 4301 Connecticut Ave. NW, Suite 404, Washington, DC 20008-2304

Phone: (800) 336-GENE or (202)966-5557

E-mail: info@geneticalliance.org

Web site: www.geneticalliance.org

The **Compassionate Friends** web site provides grief support after the loss of a child.

Address: P. O. Box 3696, Oak Brook, IL 60522

Phone: (630) 990-0010 or (877)969-0010 - toll free

Web site: www.thecompassionatefriends.org

--continued on page 7

Resources on Birth Defects and Related Topics -- continued from page 6

MUMS National Parent to Parent matches parents who have a child with a disorder, medical condition, mental or emotional disorder or rare diagnosis with other parents whose children have the same or similar condition.

Address: 150 Custer Court, Green Bay, Wisconsin 54301-1243

Phone: (877) 336-5333 or (920) 336-5333

E-mail: mums@netnet.nett

Web site: www.netnet.net/mums

The **Anencephaly Support Foundation** web site has information for families who have had a baby born with anencephaly.

Web site: www.asfhelp.com

The **Cleft Palate Foundation** web site provides a lot of information for patients and families on cleft lip and cleft palate. The web site has materials in Spanish.

Address: 104 South Estes Drive, Suite 204, Chapel Hill, NC 27514 USA

Phone: (919) 933-9044

E-mail: info@cleftline.org

Web site: www.cleftline.org

The "SMILES" web site has information on the needs of children with cleft lip, cleft palate and craniofacial deformities.

Web site: www.cleft.org

The **Chromosome 18 Registry & Research Society** web site has resources on chromosome 18 abnormalities.

Address: 6302 Fox Head, San Antonio, TX 78247

Phone: (210) 657-4968

E-mail: Office@Chromosome18.org

Web site: http://www.chromosome18.org/

The **Spina Bifida Association of America (SBAA)** web site has a wealth of information on spina bifida. SBAA works to prevent spina bifida and to enhance the lives of all affected. The web site has materials in Spanish.

Address: 4590 MacArthur Blvd., NW, Suite 250, Washington, DC 20007-4226

Phone: (800) 621-3141

E-mail: sbaa@sbaa.org

Web site: www.sbaa.org

The **Association of Congenital Diaphragmatic Hernia Research, Advocacy, and Support (CHERUBS)** web site has useful information on diaphragmatic hernia.

Web site: www.cherubs-cdh.org

Web sites Related To Health:

The **Nutrition.gov** web site has easy access to over 100 health and nutrition related web pages sponsored by federal agencies. Find out about food, diet, fitness, and diseases. There is a section on government resources and reports.

Web site: www.nutrition.gov

The **Combined Health Information Database (CHID)** web site allows the computer user to type in a birth defect and it will show available literature.

Web site: CHID.nih.gov

The **Healthfinder.gov** website also allows the computer user to type in a birth defect and it will show available literature. The literature is available both in English and Spanish.

Web site: www.healthfinder.gov

The **MAGIC Foundation** web site provides information on syndromes.

Web site: www.magicfoundation.org



CENTERS FOR BIRTH DEFECTS RESEARCH AND PREVENTION

ARKANSAS

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CALIFORNIA

Dr. Gary Shaw
California Birth Defects Monitoring Program
Phone: (510) 549-4155
E-mail: gsh@cbdmp.org
Internet: www.cbdmp.org

GEORGIA/CDC

Dr. Margaret Honein
Centers for Disease Control and Prevention
Phone: (404) 498-4315
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Internet: www.cdc.gov/ncbddd/bd

IOWA

Dr. Paul Romitti
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E-mail: paul-romitti@uiowa.edu
Internet: www.public-health.uiowa.edu/birthdefects

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NORTH CAROLINA

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Internet: www.schs.state.nc.us/SCHS/about/programs/bdmp.html

TEXAS

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Internet: www.tdh.state.tx.us/tbdmd/index.htm

UTAH

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If you no longer wish to receive this newsletter or would like to update your address, please let us know by calling toll-free at 1-800-814-1186 or emailing gpe@cbdmp.org.