## Why Do We Need a Copy of Your Child's Medical Record?

To have confidence that our data are correct and to be sure that our findings are accepted by doctors and others, we need to document the exact diagnoses for all of the infants in our study. We need to know the precise medical terminology so we can make sure we correctly classify each diagnosis. Even when parents know the exact term, scientific standards require that we can also document the diagnosis from the child's medical record. The same applies to children who do not have birth defects—we need to have confirmation from the child's medical record to meet strict scientific standards about the conditions we are studying. Some conditions are complicated and the medical terms can be confusing. Various tests, surgical reports, echocardiograms, x-rays or even lab results help us code medical conditions correctly.

It can often take a while to diagnose some conditions, and a baby may be seen at more than one health care facility during his/her first year of life. For example, a baby with a heart defect may be transferred right away to a hospital for intensive care or a baby with a cleft lip may have the lip surgically repaired several weeks after birth. This is why it's important for us to review records from all hospitals where the baby was seen.

The medical record authorization form you complete and return allows us to obtain a copy your child's record . This authorization is only valid for a specific time period after birth. All information that would identify you or your child is removed when we receive the record. We value the trust you place in us and we follow strict rules for protecting your privacy and confidentiality. If you have any questions about this, please call us.


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Spring 2009

## Hirschsprung Disease Research

Hirschsprung disease is a congenital condition in which certain nerve cells are missing from a part of the large bowel. The absence of these nerves can cause intestinal blockage. If the condition is severe, the newborn may fail to pass meconium or stool and may vomit.

Hirschsprung disease occurs in about 1 in 5000 births, and causes about 25% of all cases of newborn intestinal obstruction. It occurs in males more often than in females, and occurs more often in conditions such as Down syndrome.

Dr. Robert Heuckeroth, a pediatric gastroenterologist at Washington University in St. Louis, has been studying Hirschsprung disease in his laboratory for many years. Together, we are learning more about genetic and non-genetic factors (such as nutrition) that may be important in the development of the intestinal tract. Using cheek cell samples and the interview information collected in our study, we hope our research will lead us to better understand and perhaps even help prevent Hirschsprung disease. □


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BOSTON UNIVERSITY

# Research News

## A Word of Thanks from our Principal Investigator

We would like to express our deepest appreciation to each of the families who participated in the study. We know how busy family life can be and we value the time and effort you took to be part of our research. Your generosity has been what has helped us interview over 37,000 women in the past 32 years. Thank you!

A major goal of our study is to learn more about the safety of medications taken by pregnant women. Though women take a wide variety of medications in pregnancy, we know very little about how those medicines might affect the infant. This lack of information can make a pregnant woman terribly anxious about whether a needed medicine is safe for her baby.

To learn as much as we can, our study focuses not only on prescription medicines, but also on over-the-counter medicines, vitamins, and herbal products. Results of our research, which are published in major medical journals, have supported the safety of some products and the risks of others, and as we interview more and more women about their pregnancies, we will have even greater opportunities to answer important questions.

All of us who have devoted our careers to this important public health effort recognize that the success of this research effort depends directly on the tens of thousands of women who have contributed their experiences to the study. They have repeatedly told us that they participate in the study for one simple reason—to improve the health of women and babies in the future. We believe that our study's contributions over the years clearly show that their participation really does make a difference! □

**Allen A. Mitchell, MD**

## Study Results

Over the years, we have published over 100 articles in medical journals. It would be impossible to list all the study findings here, but we want to share highlights of a few of them in this newsletter to give you an idea of how your participation helps other families. For more detailed information about our study results, please visit our website at www.slone.bu.edu/phis. □

## Study Nuts and Bolts

Women who participate in this study come from the metropolitan areas of Boston, Philadelphia, and San Diego, as well as the state of Rhode Island, Delaware, upstate New York and southern New Hampshire. They are mothers of babies with a wide range of birth defects, mothers of babies with no birth problems at all, and women who have had a pregnancy loss.

After completing the study interview, each woman is asked to sign and return a medical record release form. This is very important, because it helps us to be sure that the information we gather about the medical diagnoses of each baby is complete and accurate and that the results of our study are correct. If mothers are interested, they are also invited to provide cheek swab samples. These are used to study biologic differences in families which might increase or decrease the risk of a birth defect or other pregnancy complication.

All information we receive is kept strictly confidential. In addition, we remove all names, addresses, phone numbers and any other identifying information from all study data, including all medical records and cheek swab samples we receive. Preserving your confidentiality is a priority for us. □

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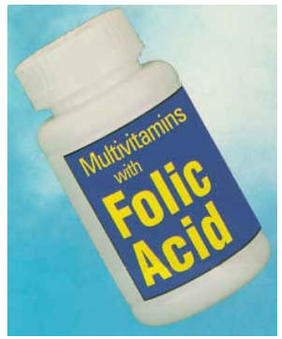
## The Folic Acid Story

When our study began 32 years ago, we didn't think that any medicine or vitamin could actually *reduce* the risk of a birth defect. To our delight, we were wrong! Researchers had suggested that folic acid taken around the time a woman became pregnant might reduce the risk of a baby being born with neural tube defects, such as spina bifida. We studied this possibility carefully in our own study data, and in 1993 we found that women who took a multivitamin containing folic acid around the time they became pregnant reduced the risk of neural tube defects by about half—a dramatic effect, and one that has been shown in most other studies as well. In addition, our study was the first to show that the amount of folic acid (0.4mg or 400 mcg) contained in a standard multivitamin was enough to produce this effect.

Because of the clear benefit of folic acid in reducing risks of neural tube defects, we have studied whether it might reduce the risks of other birth defects as well, and we found that it might also lower risks for heart defects, cleft lip and palate, and urinary tract defects. It's now recommended that women who might become pregnant make sure they take enough folic acid (0.4mg) each day, either by eating lots of foods that contain folic acid or by taking a daily multivitamin. Getting enough folic acid from a normal diet can be difficult, so the government now requires that this vitamin be added to most flour, corn meal, pasta, and breakfast cereals.

When we looked at the dietary information women provided us during their interviews, we found that eating flour and cereal grains fortified with folic acid still didn't provide enough folic acid for most women. These results reinforced expert advice that the best way to get enough folic acid to reduce the risk of certain birth defects is to take a daily multivitamin that contains folic acid. Since folic acid has this benefit only if it's taken around the time of conception, and since about half of all pregnancies are unplanned, it's important for all women of childbearing potential to be taking a daily vitamin that contains folic acid.

Next, we looked at our data to see how many women knew about the need for folic acid and how many took a folic acid-containing multivitamin around the time of conception. By the late 1990's, we found that half the women we interviewed knew that folic acid could help prevent birth defects. In recent years our data show that almost 40% of women take a multivitamin containing folic acid. This means that 60% of women are not taking folic acid. Women with lower incomes and less education, along with women who hadn't planned on becoming pregnant, were less likely to know about the benefit of folic acid and were also less likely to be taking it. This information is helping to improve public education efforts designed to increase the number of women taking folic acid around the time of pregnancy. □




## Risk Factors for Persistent Pulmonary Hypertension of the Newborn

Persistent pulmonary hypertension of the newborn (PPHN) is a rare but very serious condition in which a newborn baby has unusually high blood pressure in the arteries of the lung. While advances have reduced deaths and other complications in these babies, it would be helpful to know before delivery which babies might be at risk for PPHN so medical care could be made available immediately—and it would be even better if we could predict factors that might reduce the risk of developing PPHN in the first place.

In a major study of PPHN, we found that babies with this condition were more often delivered by Cesarean section, were unusually large, and more often born to mothers who were overweight or had diabetes or asthma. We also found that PPHN was more common among mothers who took certain antidepressant medications (called "SSRIs") in the second half of pregnancy. These factors may not cause PPHN, but knowing them can help doctors anticipate which women might have a higher risk of delivering a baby with PPHN. □



## Vaccines Given in Pregnancy

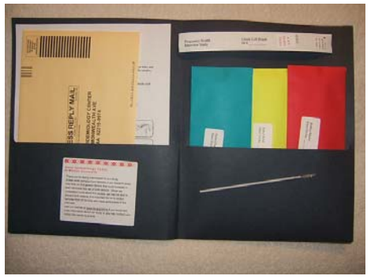
Vaccines provide protection against many serious illnesses. Important new vaccines have been developed in the last few years. Some, such as flu vaccine, are recommended for use in pregnancy by federal health officials, while other vaccines that aren't needed in pregnancy may be given to women who don't yet know they're pregnant. Not surprisingly, many pregnant women avoid vaccinations in pregnancy because of concerns of possible harm to the fetus.

A new focus of our study is to provide badly-needed information on the safety of various vaccines that may be given to pregnant women. As an important first step, we've shown that our study could identify women who received flu vaccine, which is not only the most common vaccine given in pregnancy, but also one that is important to protect against influenza in pregnancy—a disease which can be particularly severe in pregnant women.

Our study has already provided hard-to-come-by information on how many women are following the recommendation to receive a flu vaccine while pregnant. To date, we have learned that 1 in 4 women in our study receive flu vaccine. Thanks to the vaccine releases provided by our subjects, we were able to confirm the specific type and timing of flu vaccine given for most pregnant women.

We will continue to collect more information on vaccine use and safety in pregnancy; thank you for helping us to learn more about this important public health issue. □

## Vanderbilt University Collaboration Helps Unravel Mystery of Congenital Heart Disease

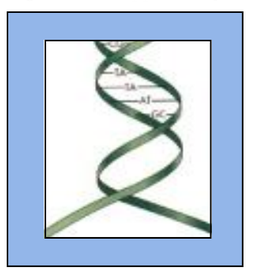


Congenital heart disease (CHD) is the most common cause of infant deaths, and the most common type of CHD is a hole in the dividing wall of the heart (known as a septal defect). We know that the development of the heart is influenced by genes that are inherited from both parents. Although certain genes are already known to cause CHD, many others have yet to be identified, and we are working with other experts in this important effort.

A new technology called "microarray analysis" allows researchers to identify over one million of an individual's genes at the same time. It's like doing a million genetic tests all at once. By comparing the genetic pattern of a child with CHD to his or her parents' patterns, researchers can determine whether the child has missing genes or extra copies of genes—exactly the kinds of genetic problems that can disturb the normal process of heart development.

Our group at the Slone Epidemiology Center at Boston University is working closely with Thomas Morgan, M.D., a medical geneticist and heart disease researcher at Vanderbilt University. Dr. Morgan is using a state-of-the-art system to find genes that are involved in heart development, and the results of his studies could shed new light on how the human heart develops, and what can go wrong when genetic problems are identified. This information may lead to new medications to help children with congenital heart disease. In addition, this powerful new genetic technology may eventually be applied to children with all types of birth defects.

The Slone Center continues to collect cheek cell swabs on patients and their parents to further the kinds of research that will help us understand human development and birth defects. Parents should keep in mind that results from any one study need to be confirmed by other studies, so it can take many years before research findings actually are ready to be applied in clinical care. However, we can only develop new understanding of genetic effects on birth defects if parents provide buccal swab samples, which allow us to take the first steps in this very important new and promising research opportunity. □







Pregnancy Health Interview Study