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 of 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.

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 certain 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.

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 helped us to provide 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 retain 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.

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.

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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.

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 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 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.