

**University of North Carolina-Chapel Hill
Consent/Assent to Participate in a Research Study and Parental
Permission for the Participation of a Minor Child**

IRB Study # 08-1511
Consent Form Version Date: 11-22-10

Title of Study: *The Fragile X Newborn Screening Study: Part 1*

Principal Investigator: Debra Skinner/Don Bailey
UNC-Chapel Hill Department: FPG Child Development Institute
UNC-Chapel Hill Phone number: 919-966-4571
Email Address: skinner@mail.fpg.unc.edu
Co-Investigators: Don Bailey, PhD, Education, and RTI International
Arlene Davis, JD, RN, Social Medicine
Cynthia Powell, MD, Pediatrics
Myra Roche, MS, CGC, Pediatrics

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Study Contact telephone number: 1-877-207-5540
Study Contact email: fxnewborn@mail.fpg.unc.edu

What are some general things you should know about research studies?

You are being asked to take part in a research study. To join the study is voluntary. You may refuse to join, or you may withdraw your consent to be in the study, and withdraw your permission to include your baby, for any reason.

Research studies are designed to obtain new knowledge that may help other people in the future. You and your baby may not receive any direct benefit from being in the research study. There also may be risks to being in research studies.

Deciding not to be in the study or leaving the study before it is done will not affect your relationship with the researcher, your health care provider, or the University of North Carolina-Chapel Hill. If you are a patient with an illness, you do not have to be in the research study in order to receive health care.

Details about this study are discussed below. It is important that you understand this information so that you can make an informed choice about being in this research study. You will be given a copy of this form. You should ask the researchers named above, or staff members who may assist them, any questions you have about this study at any time.

**The Fragile X Newborn
Screening Study**

FPG Child Development Institute
CB# 8180
UNC-Chapel Hill
105 Smith Level Road
Chapel Hill, NC 27599-8180
ph 1.877.207.5540
www.fxnewborn.org



What is the purpose of this study?

North Carolina screens all newborn babies for certain medical conditions because a few babies look healthy, but have rare and serious problems. If these problems are caught early, the babies may be helped by early treatment. The purpose of this study is to screen for fragile X syndrome (FXS), the leading inherited cause of mental retardation. FXS is a genetic condition that is NOT included in your baby's routine newborn screening. Being in this study means that your baby will have an extra test that is not part of the state newborn screening program.

FXS is caused by changes in a single gene that can be inherited in families. Different changes in the gene can cause different problems. There is a wide range of symptoms. Some people have severe learning problems and delays in their development; some are mildly affected, and some have no problems. Boys who have FXS usually have moderate to severe mental retardation and other problems with learning, language, and behavior. Some also show signs of autism. Girls who have fragile X may not seem affected at all, or show only mild symptoms, but others show signs of mental retardation and anxiety.

FXS is inherited from a parent who is a "carrier" of the gene. A carrier is someone who has a change in the gene but is usually not affected. Either the mother or the father can be a carrier. Most people who are carriers do not know it. The experimental screening test is intended to identify newborns who carry FXS and newborns who have FXS.

You are being asked to participate in this study because your baby was born at UNC Hospitals and will be screened under the state's screening program. Study participation is voluntary and confidential. Our study brochure has information on this study, newborn screening, and the genetic condition (FXS) being studied.

The FXS screening in this study is experimental and it is not a diagnostic test. We cannot guarantee that the screening will find all cases of FXS. The study is not intended to find all genetic causes of mental retardation, developmental delay, or medical problems.

Currently there is no cure or medical treatment for FXS, but this study may help us learn more about new ways to help babies with FXS and their families. It may also help provide information about whether North Carolina should offer newborn screening for FXS to all parents.

Are there any reasons you should not be in this study?

You should not be in this study if you do not want to know if your baby has the gene change for FXS.

How many people will take part in this study?

This research study plans to enroll about 15,000 mothers and their newborn babies at UNC Hospitals for the experimental newborn screening procedure for fragile X syndrome.

How long will your part in this study last?

If the screening does not indicate a need for further FXS testing, your participation in the screening study will end when you receive a letter with the result of the screening test, about 4-8 weeks after you agree to the FXS screen.

If the screening indicates a need for further testing to look for the FXS gene change, we will arrange a confirmatory test, genetic counseling and genetic medical assessments that last about 3-4 hours. This follow-up process may take 3-4 months to complete, but will not cost you anything except your time.

Families of identified babies and some other families will be invited to take part in an interview, or in a 12-month study. Families will be given additional information about those studies, and give their consent separately.

What will happen if you take part in this first part of the study?

Before you leave the hospital, a little blood will be taken from your baby's heel (a blood spot) for North Carolina's routine newborn screening which is required by state law. If you decide to participate in this study, an extra blood spot will be taken and used for the FXS screening. This bloodspot will be kept by the lab doing the testing. It will only be used to look for and study the FXS gene change and will not be used for other research. Once the researchers can no longer learn anything about FX from this sample, it will be destroyed.

The FXS screening process can take about 4-8 weeks. You should receive a letter giving the results of the screening within 4-8 weeks from the time you and your baby leave the hospital. If the screening shows a need for more testing, it does not necessarily mean your baby has FXS. The screening looks for the fragile X gene change, but cannot diagnose it by itself. That takes specialized (diagnostic) testing and a clinical genetics evaluation. For a few babies in the study, the screening will show the need for more testing and evaluation. If these are needed, we will provide them without cost to you.

What if my baby needs more testing in the study?

If screening shows a need for more testing, the genetic counselor (a specialist in discussing genetic conditions with families) on the research team will contact you. She will discuss the screening results and tell you about the part of the study for babies who get more testing. We will arrange a visit with a medical geneticist (a doctor with special training in genetic conditions) and the genetic counselor for more discussion. If you agree, we will draw blood from your baby for diagnostic testing at that visit.

Invitations to other studies

Within the next 3 months, some mothers will be asked if they would like to be interviewed on the telephone about their experiences being invited to be in this screening study. Participation in the interviews is voluntary and we will give those 30 mothers more information about the **telephone interview study** before we ask them to decide.

During the 5 years of the study, we will invite all mothers of babies who are diagnosed as carriers of FXS or as having FXS to join a year-long study on how families adapt to FXS. We will also invite the same number of mothers whose babies do not have FXS to be in this additional year-long study. We expect to include a total of 45 to 90 mothers whose babies were diagnosed as carriers or having FXS, and 45-90 mothers of babies who were not. Participation in this additional year-long **study of mothers and their babies** is voluntary and we will give those mothers more information before we ask them to decide.

What are the possible benefits from being in this screening study?

Research is designed to benefit society by gaining new knowledge. We do not know if you and your baby will benefit from participation in the screening study. Benefits of being in the screening study may include Learning more about newborn screening and about fragile X syndrome

Benefits to babies identified as having FXS or being a carrier may include:

- The diagnostic testing, genetic counseling, clinical evaluation, and referrals provided by the study.
- Eligibility for certain learning and therapeutic services that may be helpful to the baby.

- Alerting parents to the future health or learning problems the baby may have.
- Preparing parents to raise a child with special needs.

Benefits to parents of babies identified as having FXS or being a carrier may include:

- Saving financial and emotional costs of many trips to doctors to find out the cause of their child's problems.
- Getting the early intervention services the child may need.
- Information that they and their relatives may be at risk for having children with fragile X syndrome
- Getting support services which may be helpful to them

What are the possible risks or discomforts involved with being in this study?

Typically the screening poses no physical risk to the baby because the extra bloodspot can be obtained at the same time as the heel prick for the routine newborn screening.

There may be a small emotional risk; you might worry about whether the baby's screening in the study will show a need for more testing. However, this is not different from wondering what the routine screening for many conditions, which is done for all babies, might show.

If the baby is found to have fragile X or be a carrier of the FX gene, there are other possible risks:

- Learning that one's baby has fragile X may bring about feelings of grief, a sense of loss, worry, or anxiety. It may cause some stress in the family.
- Learning that one's baby has fragile X may cause parents to worry about what the condition will mean for the baby and family.
- If a baby has the gene change for FXS, it means that one of the parents also has the gene change. Parents may feel bad about passing on that gene change. They may worry that other children or family members need testing.
- There may be some stigma associated with learning there is an inherited genetic condition in one's family.
- Even though there are laws to protect genetic information, you may worry about how others could use this information. Discrimination against your family or child by insurance companies, employers or schools, could be possible. We have many protections in place to make sure that it will be extremely unlikely for others to find out this information unless you authorize it.
- Finally, although unlikely, the study screening result could be wrong. It may show that babies need more testing when they do not, or it may not find babies with FXS. If your baby has health or developmental problems later, he or she may need diagnostic testing for FXS.

What if we learn about new findings or information during the study?

You will be given any new information gained during the course of the study that might affect your willingness to continue your participation.

How will your privacy be protected?

We will keep your information confidential and secure. Information about you and your baby, and your baby's research blood spot, are linked to your name only through a special study ID number. This link will be kept in a secure place where only the researchers have access to it. The link will be kept in a secure place until all the research is completed. When researchers can no longer learn

anything more about the gene change involved in fragile X syndrome from these blood spots, the link will be destroyed.

The blood spot will be used for screening for FXS, for making sure the test is accurate, and to study the gene change for FXS. Your baby's blood spot will not be used for other research purposes.

Published study results and presentations of our findings to other scientists and physicians will not identify you or your baby.

This signed consent form will be kept in a locked filing cabinet in a locked office, apart from all study data.

Although very unlikely, there may be times when federal or state law requires the disclosure of research information, including personal information. UNC-CH will take steps allowable by law to protect the privacy of personal information. In some cases, your study information could be reviewed by representatives of the University, research sponsors, or government agencies for purposes such as quality control or safety.

A Federal law called the Genetic Information Nondiscrimination Act (GINA) generally makes it illegal for health insurance companies, group health plans, and most employers to discriminate against you based on your genetic information. GINA does not protect you against genetic discrimination by companies that sell life insurance, disability insurance, or long-term care insurance. GINA also does not protect you against discrimination based on an already-diagnosed genetic condition or disease.

What if you want to stop before your part in the study is complete?

You can withdraw from this study at any time, without penalty. You can leave the study at any time by calling the study number.

What else do I need to know about the study?

- Your decision to join the study or not will not change anything about your medical care or that of your baby. It will not change anything about your ability to get diagnostic testing for these genetic conditions on your own.
- There are **no costs to you** in participating in the study. The screening for fragile X and the follow-up confirmatory test will be provided at no charge to you. If follow up testing is needed, a trip to UNC Hospitals for this may mean missed work or child care costs. Costs for missed work or child care for these follow up visits are not reimbursed by the study, but the study will pay for transportation and parking.
- You will not receive anything for being in this part of the screening study. Mothers who are invited to participate in the other related studies will be given subject payments in appreciation for their time.

What if you are a UNC student or a UNC employee?

Participation is voluntary and you may leave the study at any time, just as described above. Participation will not give you special consideration. If you are a student, participation will not affect your class standing or grades. If you are an employee, participating is not part of your job and refusing will not affect your job.

What if you have questions about this study?

You have the right to ask, and have answered, any questions you may have about this research. If you have questions, you should contact the researchers listed on the first page of this form.

What if you have questions about your rights as a research subject?

All research on human volunteers is reviewed by a committee that works to protect your rights and welfare. If you have questions or concerns about your rights as a research subject you may contact, anonymously if you wish, the Institutional Review Board at 919-966-3113 or by email to IRB_subjects@unc.edu.

After you have made your decision, we will ask you a few questions. This will only take about 3 minutes of your time. We will ask you about things such as your age, how many children you have had, and your reasons for deciding to allow, or not to allow, your baby to be screened.

Title of Study: *The Fragile X Newborn Screening Study*

Principal Investigator: Debra Skinner/Don Bailey

Subjects' Agreement:

I have read the information provided above. I have asked all the questions I have at this time. I voluntarily agree to participate in this research study.

Signature of Parent

Date

Printed Name of Parent

Signature of Parent

Date

Printed Name of Parent

Signature of Person Obtaining Consent/Assent

Date

Printed Name of Person Obtaining Consent/Assent

Please read the statement below and initial it if you agree:

_____ The researchers may talk with another person about the baby's screening testing results. I have named that person on the "contact information" sheet attached to this form.