

About the National Birth Defects Prevention Study

One in every 33 babies in the United States is born with a birth defect. Birth defects are one of the leading causes of death in the first year of life and can affect how a child's body looks, works, or both. While the causes of some birth defects are known, we do not know what causes most of them. The Centers for Disease Control and Prevention (CDC) is working to find answers through

the National Birth Defects Prevention Study (NBDPS). This study looks at risk factors and potential causes of birth defects. Understanding the potential causes of birth defects can lead to recommendations, policy planning, and services to help prevent them. The study is a key first step toward helping bring us to a day when fewer babies are born with birth defects.

What is the National Birth Defects Prevention Study? NBDPS is the largest U.S. study looking at potential risk factors and causes of birth defects. NBDPS studies birth defects that affect the structure of various parts of the body, such as the heart, spine, brain, face, stomach, and intestines. For example, some commonly-known birth defects such as cleft lip, spina bifida, and heart defects are included in the study.

How does the study work? Researchers talk to women who have had pregnancies or babies affected by birth defects as well as mothers of babies with no birth defects about their pregnancy experience and health. Cheek cell samples are also collected from families to examine the role that genes play in the health of a baby, especially those that interact with the environment.

What states are involved and why include more than one site in the study? CDC funds the study and collects data along with researchers in other sites across the country. Participating sites include: Arkansas, California, Georgia (CDC), Iowa, Massachusetts, New York, North Carolina, Texas, and Utah. The New Jersey site has also been part of the study. Because many types of birth defects occur at such low rates, it is difficult to conduct studies that include enough children with specific birth defects. NBDPS gives us a unique opportunity to create a study group large enough to determine what environmental, genetic, and behavioral factors cause or contribute to specific birth defects.

What will the researchers do with the results? Results from the study will teach us more about the potential factors that might raise or lower the risk of having a baby with birth defects. When we know the causes of birth defects, we can develop public health policies and raise awareness of the things parents and potential parents can do to prevent them.

Is the study confidential? The names, identities, interview answers, and genetic test results of study participants are private and confidential. This information is kept under lock and key and will never be given to any person outside of the study, including insurance companies or other government agencies. No participant's personal information will ever be included in any study report or publication.

Why is this study so important? Understanding the causes of birth defects can help us prevent them. The size and scope of this study will provide important clues to help us in our journey to ensure every child is born with the best health possible.



What You Need to Know About Participating

How are study participants selected? Because birth defects are a serious public health concern, state law allows each study site to collect information on babies affected by a birth defect. This is how most women are identified in the study. Women whose babies do not have birth defects are chosen randomly from a group of women who gave birth in the same year according to state birth records. These families are contacted by phone to find out if they want to participate in the study.

Where is the study conducted? Participation in the study is convenient; it requires a phone interview and collection of cheek cell samples. This can all be done in the privacy of one's home.

How does it work? Site researchers conduct a 60-minute phone interview with selected women to learn about things like their pregnancy experience, physical environment, lifestyle, family background, health, and diet. All study participants are asked the same questions in the same way. After the telephone interview, participants will be sent a cheek cell sample kit; they will be given instructions on how to take the sample(s) and send it to the site. Once the cheek cell samples are mailed, the participant's involvement in the study is done.

How is DNA collected? Genetic material, or DNA, is collected from parents and babies through a simple cheek cell swab process that can be done at home and securely mailed to the study site. Cheek cells are easily collected by brushing a swab across the inside wall of the mouth.

What does CDC do with the collected samples? Researchers study the collected cheek cell samples to see whether certain genes cause or increase the risk of a certain birth defect. Researchers also look at how genes may interact with a woman's environment and other factors gathered during the interview.

How long are the samples kept? Cheek cell samples are kept in laboratories at each of the study sites in a secure place without participants' names attached. Some cheek cell samples will be studied shortly after they are collected; others will be stored in a specimen bank for future use. These samples are crucial to helping us figure out the causes of birth defects and how to prevent them, so they will be kept for use unless participants request that their sample be removed and destroyed.

Will participants receive their results from the cheek cell samples? No, for most findings they will not receive their test results. Instead, the data are pooled together and the findings are published in medical journals. Because birth defects are of great interest, findings are often covered in the news as well. They may also be used in health education materials. We will also publish study findings in a newsletter that we send to women who took part in the study. This newsletter will inform families of any major findings, especially if the results of a study might be useful for family members who are interested in genetic testing.