



# NEWBORN SCREENING ACCURACY PROJECT

Helping to ensure accurate testing for early detection of genetic diseases

Every state in the United States screens newborn babies for many serious but treatable genetic diseases. This screening can make the difference between life and death for some newborns, and helps those with genetic diseases get treatments they need to avoid life-long health problems.

You can help ensure that newborn screening is accurate and that babies are diagnosed and treated early.



With a simple blood test given shortly after birth, all babies born in the U.S. are screened for a range of genetic diseases. This early testing means that babies who have any of these diseases can be diagnosed quickly and begin treatment before serious health problems start.

It is important that newborn screening tests are accurate so that babies can get the correct diagnoses and treatments. The Newborn Screening Accuracy (NBSA) Project, a Sequoia Foundation program funded by The Centers for Disease Control and Prevention (CDC), helps CDC ensure that newborn screening tests are accurate. The CDC is working with the Sequoia Foundation and specialty care clinics to collect blood samples from children and adults who have already been diagnosed with a disease detectable during the newborn period. The blood samples are used to create samples to test (“quality assurance

testing materials”) that laboratories can use to make sure their screening tests are accurate. The samples can also be used to develop new screening tests so even more babies can benefit.

## Why should I participate in the NBSA Project?

Participating in this project helps ensure that newborn screening tests can accurately find disease-causing variants (also called mutations) in babies with genetic diseases. Early and accurate testing allows doctors to diagnose and treat babies with genetic diseases right away, often before serious health problems happen.

You have been asked to participate in the NBSA Project because you have a genetic disease, or you are the parent of a child with one of the genetic diseases that is found through newborn screening, or for which there may someday be a screening test.



## What happens if I decide to participate?

If you want to participate, at your next regular appointment, clinic staff will go over the NBSA Project with you and answer any questions you have. You and/or your child will be asked to sign a consent form and your clinic will take a small blood sample – about 3 tablespoons for adults, and about 1 to 2 tablespoons for children. This should only take about 10 to 15 minutes. **You will get a Certificate of Appreciation from the CDC and a \$50 gift card for participating.**



## How many times will I and/or my child need to give a blood sample?

You and/or your child will only have to give 1 blood sample.

## Can other family members participate?

If you are the parent of a child with a genetic disease and you also carry the same disease-causing variant, you can also participate. If any of your other children carry the disease-causing variant, they too can participate.

## What happens to the blood samples?

Your clinic will send the blood samples to the CDC. The CDC will test the sample and use part of it to create the test samples, called “dried blood spots,” that will be used to make sure newborn screening tests are accurate.

The other part of the sample will be used to develop cell lines, which means it will be used to grow your white blood cells in the laboratory. The cell lines allow the CDC to make many dried blood spots from just one small blood sample. The CDC will store the materials and send them out to newborn screening laboratories or use them internally to develop new tests. The sample won't be used for any other purpose.

## How is my information protected?

Only your clinic will know that you have participated. Your clinic will send the sample to the CDC without any information about you. The sample will be identified only by a number, not by your name or your child's name. There will be no way for anyone to tell who donated the sample.

## What if I decide not to participate?

You do not have to participate in this project. If you decide to participate, you can still change your mind at any time.

## Where can I learn more about the NBSA project?

For more information about the NBSA Project, talk to your clinic or go to [www.nbsaproject.org](http://www.nbsaproject.org).