

*Frequently Asked Questions about*

# DNA Sequencing and Newborn Screening

New technologies can help improve screening and diagnostic testing. Sequencing is a new technology that has gotten a lot of attention in recent years. This sheet of Frequently Asked Questions (FAQs) describes how this technology may be used in newborn screening.

## WHAT IS NEWBORN SCREENING?

*Newborn screening* is a state public health service that aims to screen all babies for certain serious conditions at birth. For those babies with these conditions, it allows health professionals to start treatment before some of the harmful effects happen.

Newborn screening starts with a blood test. During the blood test, which is sometimes called a heel stick, the baby's heel will be pricked to collect a small sample of blood. The baby's blood sample is then sent to the lab. Other tests are done right in the nursery - testing oxygen levels to look for heart problems and a hearing test. For more information regarding newborn screening, visit [BabysFirstTest.org](http://BabysFirstTest.org). This FAQ just discusses how sequencing might be used for the part of newborn screening that looks at the baby's blood.

## WHAT IS A GENOME?

The genome has all the genetic material of a living thing, known as deoxyribonucleic acid, or DNA. The way DNA groups together ([genes](#)) provides instructions for building the proteins that make up your body. It contains information about how a living thing will look and function. It is the informational blueprint for your body.

## WHAT IS DNA SEQUENCING?

*DNA sequencing* is a laboratory method that looks at the DNA in a person's cells to identify changes in the DNA. Sequencing is usually done on a single gene when there is concern that a change in that gene might be causing a health problem in a person. Sometimes sequencing looks at all, or larger parts, of the DNA in a person, not just one gene. If sequencing is done on specific parts of the DNA called exons, it is called "Whole Exome Sequencing". When it is done on all of the DNA, it is called "Whole Genome Sequencing".

## WHAT KINDS OF INFORMATION COULD "WHOLE GENOME" OR "WHOLE EXOME" SEQUENCING PROVIDE?

Since certain changes or misspellings in the sequence of letters that make up DNA may cause genetic conditions, genome or exome sequencing may help to identify these changes.

## HOW IS DNA SEQUENCING USED NOW IN NEWBORN SCREENING PROGRAMS?

In some states, sequencing of a specific gene is done when a baby's initial newborn screening results show something abnormal. DNA sequencing in these cases is only looking at one or two genes related to certain conditions and NOT a newborn's "whole genome".

## IS MY BABY GETTING WHOLE GENOME OR WHOLE EXOME SEQUENCING AS PART OF THE NEWBORN SCREENING TEST?

No. Whole genome or whole exome sequencing is not part of current state newborn screening programs. To learn more about your state newborn screening program, visit: <http://babysfirsttest.org/newborn-screening/states>.

## WILL WHOLE GENOME OR WHOLE EXOME SEQUENCING REPLACE NEWBORN SCREENING?

Not at this time. DNA sequencing is separate from the initial newborn screening test and is only used by some states if the initial test is out-of-range. It is difficult to predict whether this new technology will ever replace traditional newborn screening. Keep in mind that whole genome or whole exome sequencing is currently only in a research phase.

The National Institutes of Health (NIH) started a research program in 2013 looking at the use of whole genome or whole exome sequencing in newborns. The NIH research program is taking place at four hospitals: Rady Children's Hospital, San Diego, CA; University of California San Francisco; University of North Carolina at Chapel Hill; and Brigham and Women's Hospital, Boston, MA. However, (genomic or genome) sequencing of newborns is only done with the parents' consent. The overall goal of these programs is to learn about the possible benefits and challenges of genome sequencing in newborns and to better understand how it can help diagnose conditions and how to communicate results of genome sequencing to health professionals and parents.

For more information on these projects, visit: <http://www.nih.gov/news/health/sep2013/nhgri-04.htm>