NHS
Central and South
Genomics

Generation Study

The Generation Study is a long-term research study. We want to understand if we can improve how we diagnose and treat genetic conditions by looking at the DNA of newborn babies. This study is run by Genomics England, a company owned by the UK Government Department of Health and Social Care. We're partnering with the NHS to do this. The Health Research Authority has approved this study, they have made sure it is ethical and legally sound.

Our research has two main goals. To find rare genetic conditions early, so affected babies can get treatment fast and to learn more about genes and health, so we improve testing and treatment for genetic conditions in the future.

The NHS already offers newborn blood spot screening, the heel prick test, to all babies when they're 5 days old. This tests for 9 rare, treatable conditions. The Generation Study is different from the newborn blood spot screening. This study tests newborn babies for 200 plus rare genetic conditions. It is unlikely that your baby will have a condition, but if they do, finding it early could help improve their health and life. It's important to know that this study is for research. It's not a standard NHS service. This means we do not know how well our approach will work. That's why the study should not replace standard NHS care for you or your baby.

The conditions we test for usually appear in the first few years of life, can be improved if caught early, and can be treated through the NHS in England. Some conditions have simple treatments, like taking a daily vitamin. Others are treated more intensively, with therapies and medication.

An NHS practitioner will collect a blood sample shortly after birth. They will ask your permission before they do this. A small amount of blood will be drawn from the umbilical cord. This will not hurt you or your baby. If we cannot collect an umbilical cord sample, we'll do a heel prick. This could feel uncomfortable for your baby for a moment.

We use the samples to turn your baby's DNA, Their genome, into a digital file. Then we look for changes in certain genes that might suggest a baby has a rare condition. Any baby could have these even if there's no family history.
We'll contact you by phone or email within a few months with results. 99% of babies will not have any of the genetic conditions we look for. This does not mean your baby will never get sick. There are many other health conditions they could get. And although its unlikely, they could still get one of the conditions we test for.

If we suspect a condition, an NHS specialist will call you as soon as possible. The result is not a diagnosis and must be confirmed by further tests.
As part of the study, we look at your antenatal records. We also look at your baby's health records over time. This is a long-term study. It starts while you are pregnant and continues until your child is around 16.

We'll safely store your baby's sample, a digital file of their DNA, and your antenatal data in a secure database called the National Genomic Research Library. Approved researchers will study this data to learn more about genes and health. Your baby's identity will not be visible to them.

By taking part, you could find out early about a possible genetic condition in your baby. Joining the study could also help future generations of children with genetic conditions get a healthier start to life.

Before you decide to join the study, it's important you speak with your family or people that help make decisions for your baby. This is because people related to your baby, or who care for your baby, could be affected by the result. You should talk to your baby's other parent, if you're in contact. You should both agree on joining the study. You can change your mind about taking part at any time and you don't need to tell us why.

Like any medical research, there are pros and cons to joining this study.

Risks: This study is for research and is not a diagnosis. This means there is a small chance we could get your baby's result wrong. It may be stressful to wait for the results.

Benefits: your baby could get early genetic testing, and early treatment may help reduce symptoms or stop them from becoming ill.

People from all backgrounds are welcome to join the study. Taking part could make the study more diverse and help to improve genetic testing for everyone in the future.

This study is free and optional. To join the study, you need to give your consent. Contact the study team to sign you up during your pregnancy. They'll help you understand the study and answer your questions.

If you want to find out more about the study or if you want to sign up to take part, we can arrange an appointment with an interpreter. Talk to your midwife or the team delivering the study at your hospital, or visit our website www.generationstudy.co.uk

Please indicate if you would like to know more about the study:

Yes - I am interested in taking part in the study and would like to find out more.
[We can arrange an appointment for you with an interpreter]

No - I am not interested in taking part in this study