Genomics England | Generation Study

Participant Information Sheet

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Contents

Introduction

Scan for more information

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.

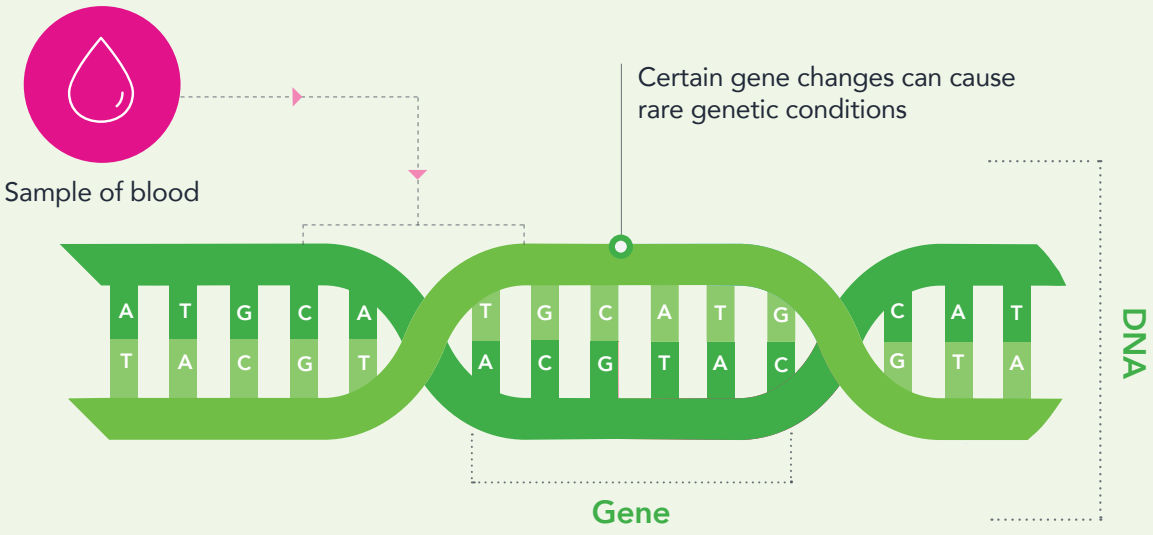
This study is free and optional. This sheet has lots of information to help you decide if you'd like you and your baby to take part. You should talk to your healthcare team and family about this decision. To sign up, talk to a member of the study team.

This study is run by Genomics England, a company owned by the UK Government Department of Health and Social Care. Our research looks at new ways genetics can affect our health. We're partnering with the NHS to do this. The Health Research Authority has approved this study. They have made sure it's ethical and legally sound.

This study investigates the genomes of newborn babies to see if we can find and treat genetic conditions early. A genome is a person's entire genetic sequence — their body's instruction manual. Genomes are made up of a chemical called DNA, and contain thousands of genes. Genes tell our bodies how to grow and develop.

Looking into someone's genome can give us information. That's because changes in a genome can lead to health problems, including rare conditions. To study someone's genome, we take a DNA sample. It usually comes from a few drops of blood. From that, we create a digital file of their genome. This process is called genetic sequencing.

To learn more about genetic sequencing, go to www.genomicsengland.co.uk/genomic-medicine/understanding-genomics



Goals of the study

Our research has two main goals:

01 Find rare genetic conditions early, so affected babies can get treatment fast

02 Learn more about genes and health, so we can improve testing and treatment for genetic conditions in the future

This study will help us understand how this kind of testing could work for babies, families, and the NHS. By taking part, you could find out early about a possible genetic condition in your baby. You'd also join a community of families helping to support research about genes and health. Joining the study could also help future generations of children with genetic conditions get a healthier start to life.

People from all backgrounds are welcome to join this study. But people from Black, Asian, and minority ethnic communities are under-represented in this kind of research. If you are a member of these communities, taking part could make the study more diverse — and help to improve genetic testing for everyone in the future.

Who can join the study?

As a part of this study, we look at your antenatal records. We also look at your baby's healthcare records over time. Because of this, there are a few things we require in order to take part.

To be eligible for the study, you need to:

- Be pregnant
- Have an NHS number
- Be 16+ years old
- Not be having twins, triplets, etc
- Not be a surrogate or planning to give the baby up for adoption
- Be registered with a GP in England

The study team will check these again with you when you consent to join.

Before you decide to join the study, it's important you speak with your family or people who 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 also talk to your baby's other parent, if you're in contact. You should both agree on joining the study.

Differences from NHS care

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. It's an NHS service that has already been well-researched.

The Generation Study is different from the newborn blood spot screening. This study looks for genetic changes that can cause around 200 rare conditions.

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 this study should not replace standard NHS care for you or your baby. If you are worried about a genetic condition in your family, you should talk to your midwife or GP.

To learn more about the newborn blood spot test, visit www.nhs.uk/conditions/baby/newborn-screening/blood-spot-test/

Benefits and risks of being a part of the study

Like any medical research, there are pros and cons to joining this study. We want to make sure you have all the information you need to make a decision.

Benefits

Your baby could get early genetic testing. They will get tested for 200+ rare genetic conditions. It's rare but possible that we'll identify a condition early. We'll share this result as soon as possible. They could get early treatment which may help reduce their symptoms or stop them from becoming ill.

You could help us try to improve treatment for genetic conditions. Taking part helps researchers learn more about the link between genes and health. Their work could help develop new treatments. It could also help us predict or diagnose conditions more quickly in the future.

Risks

Your baby could feel uncomfortable when we collect the sample. If we are not able to collect a sample from the umbilical cord, we will need to prick the baby's heel with a tiny needle. This could cause them temporary discomfort.

Your baby could get an incorrect result. 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.

You could feel uncertain about results. It might be stressful to wait for results. If we suspect your baby has a condition, they'd need to have follow up appointments with the NHS. This could take some time. Because these conditions are rare, there might not be a lot of information available.

You and your baby could be identified through the data. In rare circumstances, a researcher could match your names to your data. We have safeguards in place which make this very unlikely.

How the study works

This is a long-term study. It starts while you are pregnant and continues until your child is around 16. During this time, we'll send you updates about the study.

Step 1

Sign up during pregnancy

Contact the study team to sign you up during your pregnancy. This can be in-person during a hospital appointment or over the phone. They'll help you understand the study and answer your questions. Next, they will ask you some basic questions and confirm your contact details. After that, you'll receive an email welcoming you to the study.

Step 2

Sample collected shortly after birth

An NHS practitioner will collect a blood sample shortly after birth. If you give birth at home, your midwife can collect a blood sample or we'll schedule a hospital appointment for this. We'll use this sample to analyse your baby's DNA.

Step 3

Receive results in a few months

99% of babies will not have any of the gene changes we look for. If we do not suspect a condition, we'll let you know by email or letter about 2 months after your baby is born. If we suspect a condition, an NHS specialist will call you as soon as possible. They'll talk you through the next steps, including any further tests needed.

Step 4

Sample and data stored and used for research

We'll safely store your baby's sample, a digital file of their DNA, and your antenatal data. We'll also get regular updates from your baby's healthcare record. We keep this data so that we can learn more about finding and treating genetic conditions over time. Approved researchers will study this data to learn more about genes and health. Your baby's identity will not be visible to them.

Step 5

Ongoing contact about the study

We'll contact you from time to time to update you about the study. We may ask for feedback or if you'd like to take part in other research. When your child is around 16 years old, we'll ask them if they'd like to stay in the study.

Conditions we test for

Scan to learn more about these conditions

This study tests newborn babies for 200+ 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.

The conditions we test for:

Usually appear in the first few years of life

Can be improved if caught early

Have treatment through the NHS in England

Types of conditions

The conditions we test for vary in how common they are, what their symptoms may be, and how they can be treated. Some conditions are well-known, like Cystic Fibrosis. Others are more rare, like Barth Syndrome. Each condition has different symptoms. However, they can all cause someone to get sicker than the average child.

Treatment for conditions

All the conditions we test for have treatment through the NHS in England. Some conditions can be easily treated, like taking a vitamin every day. Other conditions have a more intensive treatment plan. For example, Severe Combined Immunodeficiency (SCID) is treated with a stem cell transplant.

Keep in mind

A few of the conditions we test for do not have immediate symptoms. Here is an example to consider.

Your baby is diagnosed with a genetic condition after extra tests following a result from the Generation Study. You meet with the specialist team at a hospital. They explain that although your baby seems well now, they will likely start to have symptoms within two years. Once that happens, your child will start getting treatment in the NHS.

Until then, your baby will need check-ups every six months. You will need to monitor your baby for symptoms, and this could be worrying. You get support from the specialist team and genetic counsellors.

Finding this condition early can help to be prepared to start treatment as early as possible. However, some families would prefer not to know about the condition until symptoms are showing.

We recommend considering this example carefully before you join the study.

Collecting the sample

An NHS practitioner will collect a blood sample. They will ask your permission before they do this. If you give birth in hospital, this will happen shortly after your baby is born.

Blood from the umbilical cord: A small amount of blood will be drawn from the umbilical cord. This is possible with delayed cord clamping. This will not hurt you or your baby.

If we cannot collect an umbilical cord sample, we'll do a heel prick. This involves pricking your baby's heel to collect a few drops of blood. This could be uncomfortable for your baby for a moment. We recommend cuddling or feeding your baby to help with this.

In rare cases, collecting a sample might be more complicated, and the doctor treating your baby will be able to decide how to proceed.

If you give birth at home

If you give birth at home, your midwife can collect a blood sample or we'll schedule a hospital appointment to collect a sample shortly after your baby is born.

If we cannot collect a sample

There may be other reasons why we cannot collect samples. For example, this could happen if you give birth in a different NHS Trust, or the birth has complications. This is rare. If this happens, you and your baby cannot join the study. We will send you an email or letter to confirm this.

If you do not want us to collect a sample

If you decide you do not want us to collect a sample from your baby, that's fine. Let the NHS practitioner know. This will not affect you or your baby's healthcare in any way. If this happens, you and your baby cannot join the study. We will send you an email or letter to confirm this.

After the sample is collected

The hospital team will label the sample with a unique code number. This allows us to keep track of them without using your baby's name. Only the hospital team and Genomics England will be able to link the sample to your baby.

Next, the hospital team send the sample to external companies. These companies extract and sequence DNA. They are commissioned by Genomics England. They cannot access your baby's personal details.

After your baby's sample has been collected, you do not need to do anything else to remain in the study.

Getting results

When we analyse your baby's DNA, we look for changes in their genes. These changes are known to cause 200+ rare genetic conditions.

Test results

There are two different test results: no condition suspected, or condition suspected. The way we contact you depends on the result.

How we contact you: by email or letter, a few months after birth

No condition suspected

Most babies will get this result — about 99 in 100. It means we did not find any of the gene changes known to cause the genetic conditions in this study.

This result does not mean your baby will never get sick. There are many other health conditions they could get. And although it's unlikely, they could still get one of the conditions we tested for.

We'll send a copy of this result to your GP. You can always talk to them about the result. You should also talk to them if you are worried about your baby's health or your family history.

How we contact you: by phone, a few weeks after birth

Condition suspected

A very small number of babies in this study will get this result — about 1 in 100. It means we found one of the gene changes linked to one of the genetic conditions in our study.

If we suspect your baby has a condition, we will share this with a specialist team in the NHS. We will also share your contact details with them, including your baby's name and personal details. It will be someone new to you, so that your baby can get fast and specialised care. Your baby may already be showing symptoms of the condition at this point, and could already be receiving care.

There is a small chance we won't be able to complete the test or provide you with results. This means that you and your baby won't be participants in the study. If this happens, we will let you know by letter.

Phone call from the NHS: The specialist team will call you as soon as possible in the weeks after birth. They will also contact your baby's GP. They'll arrange an appointment with you and your baby to discuss the result and next steps. The study team can cover the travel costs of this appointment if you need it.

Follow up tests: The specialist team will likely arrange more tests to confirm a diagnosis. This may include a blood or urine sample, a scan, or other types of tests. You may be asked to give a sample too, so that we can understand how your baby inherited these gene changes.

Diagnosis: If the follow up tests show that your baby has a genetic condition, the specialist team will discuss this with you and provide support.

Treatment plan: Every genetic condition in this study has a treatment plan in the NHS. The specialist team will explain what the plan for your baby is.

Feedback: The specialist team will share information about your baby with us. This helps us check how they are doing, and enables us to understand the accuracy of the test. This includes your baby's name and personal details.

If a condition is suspected, here's what we expect to happen.

Dealing with uncertainty

We only test for conditions the NHS knows how to diagnose and treat. But if we suspect your baby has a condition, there might be some uncertainty.

Possible uncertainties:

- **Incorrect diagnosis:** There's a small chance we get the result wrong, and your baby does not have the condition.
- **Unclear diagnosis:** There's a small chance that follow up tests cannot confirm or disprove a diagnosis.
- **Delayed diagnosis:** It could take many tests before the condition is confirmed.
- **Unclear symptoms:** It could be difficult to know when or if your baby will start having symptoms.
- **Effect on family:** Because these conditions are genetic, other members of your family could be impacted by the result.

These possible uncertainties can be worrying. We and the specialist team can connect you with more information, counselling, and support groups.

Scan to learn more about results

Keep in mind

Although unlikely, it's possible that your baby could get an incorrect result at first. Here is an example to consider:

When your baby is 3 weeks old, you find out that they are suspected to have a genetic condition.

Your baby's doctor schedules some extra tests. In the meantime, you read about the condition and become worried.

A few weeks later, the results come back.

Your baby does not have the condition after all.

The specialist explains that the Generation Study result was not a diagnosis, and on rare occasions, it can be incorrect.

This situation can happen with other tests too, like the NHS newborn blood spot test. It can be a confusing time for families.

We recommend considering this example carefully before you join the study.

Data and access

During the study, we'll securely store you and your baby's data. Keeping it safe and confidential is our top priority. Healthcare researchers, who go through a rigorous approval process, will access the data for projects related to genes and health.

Data we store

It's our legal and ethical duty to take care of samples and data. We have a strong record of keeping data safe in other studies.

We keep the following data:

Contact details for you and your baby: This helps us keep in touch. This will not be kept where researchers can access it.

Your baby's DNA: We store this as a digital file.

Your antenatal record: This is maternity data which includes details about the pregnancy, labour, and birth, including any stays in hospitals during your pregnancy.

Regular updates from your baby's healthcare record: This could include information from the NHS and other medical organisations such as medical test results or information about an illness.

The antenatal and health data are collected from NHS England and other organisations listed at www.genomicsengland.co.uk/privacy-policy/.

Where the data is stored

We store your baby's genetic and healthcare data—and your antenatal data—in a secure database called the National Genomic Research Library. This is a library where genetic and health data from thousands of people is accessed for research. We manage the library, and approve researchers from around the world to access it. The library is held in secure data centres in the UK. We use industry-standard security to make sure only approved researchers can access the library.

You can learn more about the type of data and research in the library at:
www.genomicsengland.co.uk/patients-participants/data

Who can access the data

Only Genomics England and approved healthcare researchers can access the data. We never share this data with insurers or marketers.

Only select individuals at Genomics England can access data about you and your baby's identity and contact details. We only share this information with your baby's GP and NHS specialist team. We do this when we have the result from the test.

We will also review your baby's health data throughout their childhood so we can better understand how the test works, and to determine whether it makes sense to make this type of test more widely available in the future.

Approved healthcare researchers will study data in the library, including your baby's data. They could come from hospitals, universities, charities, or healthcare companies like pharmaceutical companies. They will use the data to learn more about genes and health, discover new conditions, and create new treatments.

These researchers cannot see personal information, like name and contact details. However, we cannot guarantee that your data will never be indirectly linked to you or your baby. For example, if your baby has an exceptionally rare condition, it may be possible to work out that their data belongs to them. We have strict penalties for anyone who tries to identify or misuse this data.

Your baby's data, along with thousands of other people's data, may be used to:

- **Help researchers distinguish between harmful and harmless genetic changes.**
  This is important for accurately diagnosing and developing treatments for genetic conditions.
- **Aid in cancer research.**
  By looking at data with and without certain changes, researchers can find new cancer-related genes and develop therapies that benefit many cancer patients.
- **Help improve the accuracy of genomic screening tests.**
  By helping us better identify people who are at-risk sooner, treatments can be started earlier which may lead to better outcomes for those with conditions.

How healthcare researchers are approved

All researchers who access the data are working on healthcare projects. New research proposals are approved by an independent Access Review Committee. This committee includes clinical experts, scientists, and NHS patients already in the library. Every researcher signs a code of good practice and completes data protection training.

You can see the approval process for all researchers on the Genomics England website at www.genomicsengland.co.uk/patients-participants/data

What we do with leftover samples

If any samples are left over from your baby's test, we'll store them in a secure biobank in the UK. Each sample is identified with a unique code. This protects your baby's identity.

These samples may be used again for approved healthcare research. If this happened, the research would be related to genes and health. It would need to be approved by an independent Access Review Committee before going ahead.

Keep in mind

One type of approved researcher who can access data is from private pharmaceutical companies. Here is an example to consider:

A private pharmaceutical company is making a new drug for children with a rare disease.

You hear that they used data from the Generation Study during their research. You gave consent for your baby to join the study five years ago, and so this might include your baby's digital data and their stored blood sample.

They could not see any details that could identify your baby. And like any researcher, they followed a strict approval process.

This kind of research can help advance medicine — but some people might be uncomfortable with it.

We recommend considering this example carefully before you join the study.

Ongoing contact

We will contact you from time to time throughout your baby's childhood—typically no more than a couple times per year. This could be by email or letter. We'll use the contact details you gave us.

We may contact you to:

- Share news and updates about the study
- Ask for more samples or information
- Ask for feedback on the study
- Invite you to join further research or other similar studies

Anything we ask is optional. You can say no to any requests we make.

New findings in your child's DNA

A researcher might find something related to your baby's health during the study. This is very rare, but possible. If this happens, we will work with the NHS to contact you. We will only do this if it relates to a serious and treatable condition, or if we already know your child has a condition.

If you withdraw from the study or unsubscribe from all contact, we will not contact you with this information.

Contacting us with concerns

Contact the study team at your NHS Trust or Genomics England if you have concerns about the study. Find contact details on the website www.generationstudy.co.uk/contact.

If you remain unhappy and wish to complain formally, you can find more information on the NHS complaints procedure here: www.england.nhs.uk/contact-us/feedback-and-complaints/complaint/complaining-to-nhse/

Unsubscribing from all contact

You can ask us to stop contacting you about the study. We will keep your baby's data for research but will no longer contact you. You will not receive further updates or requests from us.

How to unsubscribe: Visit www.generationstudy.co.uk/unsubscribe

If you unsubscribe before we've shared the result of your baby's test, we will still contact you to share the result. If your baby has a condition suspected result, we will refer them to the NHS for confirmatory testing and clinical care.

Withdrawing from the study

You can change your mind about taking part at any time and you don't need to tell us why.

Withdrawing before the sample is collected

How to withdraw: Contact the study team or tell the NHS practitioner

You can change your mind about having the sample collected. Tell your midwife or healthcare team before your baby is born. You can decide this after you give birth, too. The NHS practitioner will ask your permission before collecting the sample, and you can say no. If you withdraw before the sample is collected, your baby will not join the study.

Withdrawing after the sample is collected

How to withdraw: Visit www.generationstudy.co.uk/withdraw

You can change your mind about being in the study after the sample is collected. If the data has already been stored in the National Genomic Research Library, or it is already involved in research, we cannot stop this. But we can make sure no new research is done, and no more healthcare information is collected. We will also destroy any leftover samples.

If you withdraw before we've shared the result of your baby's test, we will still contact you to share the result. If your baby has a condition suspected result, we will refer them to the NHS for confirmatory testing and clinical care. Your baby's data and sample will not be stored or used for research.

If your child would like to withdraw

As your child grows up, they will be able to decide for themselves if they still want to be a part of the study. They can contact us to withdraw. You can help them do this.

When your child is around 16 years old, we will contact them to check if they'd still like to be a part of the study. If we cannot contact them, after a reasonable number of attempts we will withdraw them from the study.

Data protection, safety, and security

Scan to see our privacy notice

How your information is used

The General Data Protection Regulation (GDPR) and the UK Data Protection Act of 2018 govern how we process and use your personal data.

You can find out more about how we use your information by:

- Visiting www.hra.nhs.uk/patientdataandresearch
- Reading our leaflet available at participating hospitals
- Contacting your study team
- Sending an email to generationstudy@genomicsengland.co.uk
- Contacting us on 0808 281 9535
- Contacting our Information Officer, using the details provided above

Accessing your data

You have the right to request what data we hold about you. Please note that your data subject rights may be limited due to the purposes of the research and that any such request to exercise data subject rights will be reviewed by our Data Protection Officer.

Safeguarding

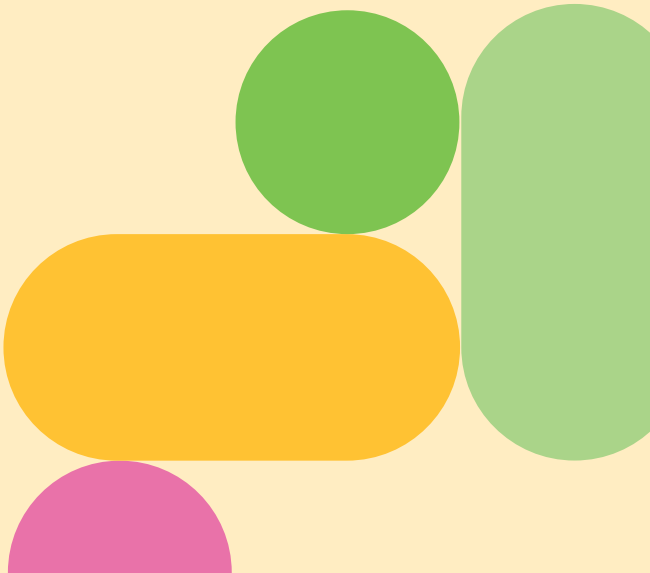
During the study, it's possible that we learn about a safety issue affecting you or your baby. If this happens, we will work with the study team to manage this.

If you suffer study-related harm

Although it is very unlikely that you will experience physical harm as a result of your study participation, we have insurance that covers injuries in certain circumstances. Please contact us at generationstudy@genomicsengland.co.uk for more details.

General enquiries

If you have any further questions about the study, you may contact Genomics England at generationstudy@genomicsengland.co.uk or 0808 281 9535.

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