STARTING IN A FEW MINUTES AT 15:30

The Generation Study: Genomics in action

26 February 2026

#iHVInsights

www.ihv.org.uk

iHV Insights

The Generation Study: Genomics in action

26 February 2026

#iHVInsights

www.ihv.org.uk

Welcome

Make the most of the opportunity

Enjoy the whole hour

Ask - type your questions to the panel as you think of them during the presentations – use the Q&A function:

- **Name of presenter** you would like to answer your question
- Brief **question**
- **Your name** (if you wish to do so)

Feedback
Participate in the poll or contact us at events@ihv.org.uk

Social media - #iHVInsights

Continue your CPD:

- Access recording of live session after the event on our website (no recording on other devices is allowed)
- Attendance at this event and engagement with the iHV reflection template will support you with demonstrating CPD activities that align to the 2022 NMC Standards of Proficiency for SCPHN Health Visiting.

The Institute of Health Visiting is a charity and centre of excellence for health visiting. Its core purpose is:
To improve outcomes for children and families and reduce health inequalities through strengthened health visiting services

Agenda

Welcome

- Sally Shillaker – Practice Development Lead - Genomics, Institute of Health Visiting

Guest Speakers

- Lindsay Ratan – Case Manager (Newborns), Genomics England
- Amy Clarkson – Generation Study Regional Results Coordinator (North East and Yorkshire), North East & Yorkshire Genomic Medicine Service
- Vicky and Joey Underhay – Parents of Freddie with lived experience
- Dawn Wickers – Interim Team Leader, Rotherham Children's Public Health Nursing Service – Family Health Division

Q&A with Speakers

Close and Evaluation

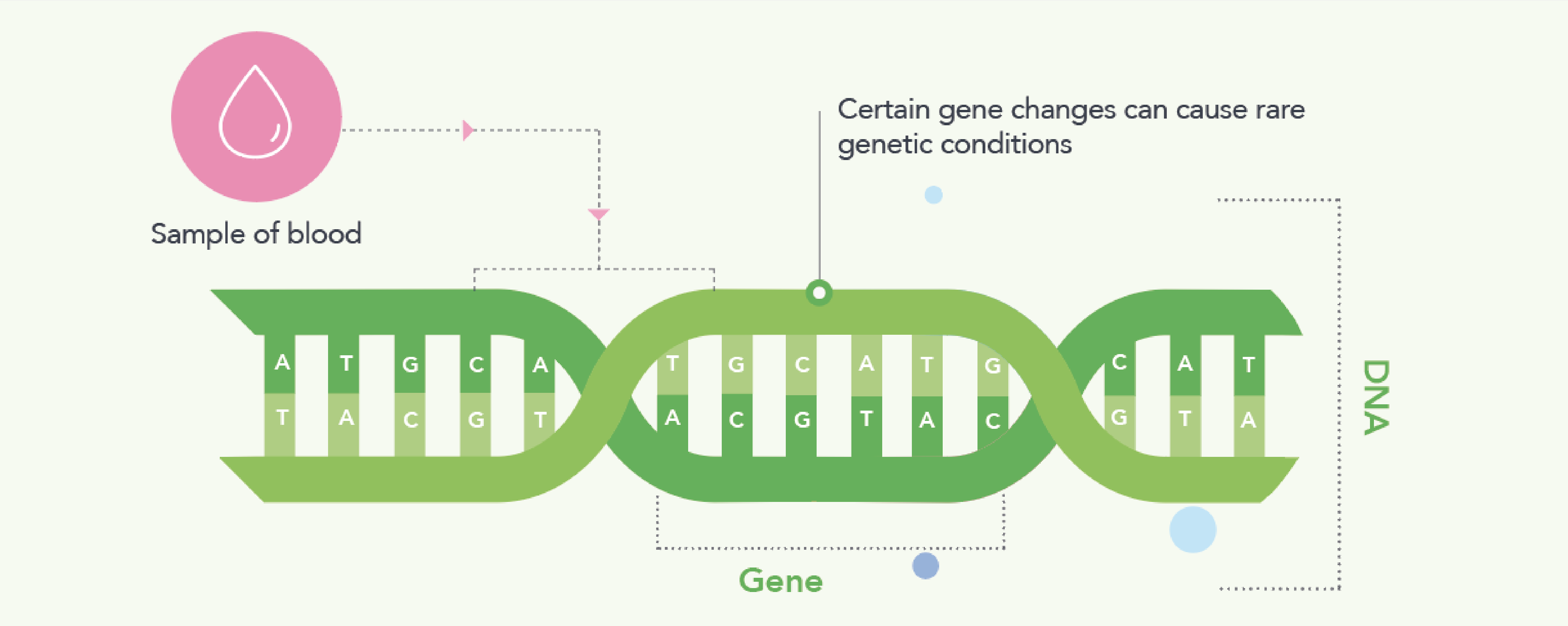
The Generation Study

- A long-term research study

- Aims to understand if we can improve how we diagnose and treat genetic conditions by looking at the DNA of newborn babies

- Involves collecting a blood sample after birth to read a baby's genome.

A refresher on genomics



Learn more: https://www.genomicseducation.hee.nhs.uk/education/core-concepts/what-is-genomics/

Our research study’s focus

Three parts | Approval has been received from Health Research Authority

** Key point: not just how each might be implemented, but whether they should be implemented.**

01

Evaluating the utility and feasibility of screening newborns for a larger number of childhood-onset rare genetic conditions in the NHS using whole genome sequencing

02

Understanding how babies’ genomic data could be used for discovery research, focusing on developing new treatments, and diagnostics for NHS patients

03

Exploring the potential risks, benefits, and broader implications of storing a baby’s genome over their lifetime

Eligibility

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

Next speaker

Lindsay Ratan

Case Manager (Newborns), Genomics England

Type your questions as you think of them during the presentations

Use the Zoom Q&A function

- **Name of presenter** you would like to answer your question
- Brief **question**
- **Your name** (if you wish to do so)

Genomics England | Generation Study

Generation Study

iHV Insights

Case Manager Presentation

Lindsay Ratan 26th Feb 2026

Version 1.0 11.09.24

Contents

Conditions we test for

200+ rare genetic conditions

- ✓ Usually appear in the first few years of life
- ✓ Can be improved if found early
- ✓ Have treatment through the NHS

Learn more:
https://www.genomicsengland.co.uk/initiatives/newborns/choosing-conditions

Version 1.0 11.09.24

This screening tests includes…

A full list of every condition tested for can be found on the Generation Study website.

- Conditions already tested for at birth (but through a different types of test) like Cystic Fibrosis
- Rarer conditions such as Barth Syndrome and Diamond Blackfan Anemia
- Hormonal conditions that affect growth and development, like genetic Hypothyroidism and Growth Hormone Deficiency.
- Blood conditions that affect red blood cells like Sickle Cell Disease of blood clotting like Hemophilia
- Immune system conditions that increase someone's risk of life-threatening infections, like Severe Combined Immune Deficiency (SCID).
- Metabolic conditions that affect the body's ability to process certain substances and remove toxins, like Phenylketonuria.

Version 1.0 11.09.24

Collecting a sample

Version 1.0 11.09.24

Sample types

Primary Sample

Cord blood

Hospital & Home Births

Secondary Sample

(if cord blood not collected)

Heel prick into capillary tube

Sample collected from an indwelling line (arterial/venous)

No repeat samples

Research study

As this is a research study, only one sample can be registered per participant.

If a sample fails, we cannot repeat the test with a heel prick.

Underscore this with teams so they understand the importance of good collection.

Version 1.0 11.09.24

This test uses genetics as a *screening tool*

This is new.

Because of this, some of the uncertainties and limitations are less familiar to healthcare professionals.

This can mean our expectations aren't always quite right

Genetics as screening tool

Expectation

As the Generation Study utilises 'whole genome sequencing (WGS),' it can find any genetic condition.

→

Reality

Although the study uses WGS, it will deliberately limit what it looks for, focussing on a select number of genes and conditions.

It will not only limit what genes are looked at, but importantly, the genetic changes (variants) within those genes.

Genetics as screening tool

Expectation

Finding a genetic variant gives you a definitive diagnosis of that condition.

Reality

The result from a research screening test is not a diagnosis.

This test is done at birth, typically before there are any symptoms. Further tests will have to be done to confirm a diagnosis of the suspected condition.

Genetics as screening tool

Expectation

If the test uses Whole Genome Sequencing technology and nothing is found, there is little further testing possible

→

Reality

This test is done as part of a research study.

We don't know how many babies with a 'no condition' suspected will still have a genetic condition.

If there are any concerns about a baby they should have all the normal NHS care and investigations.

Returning results

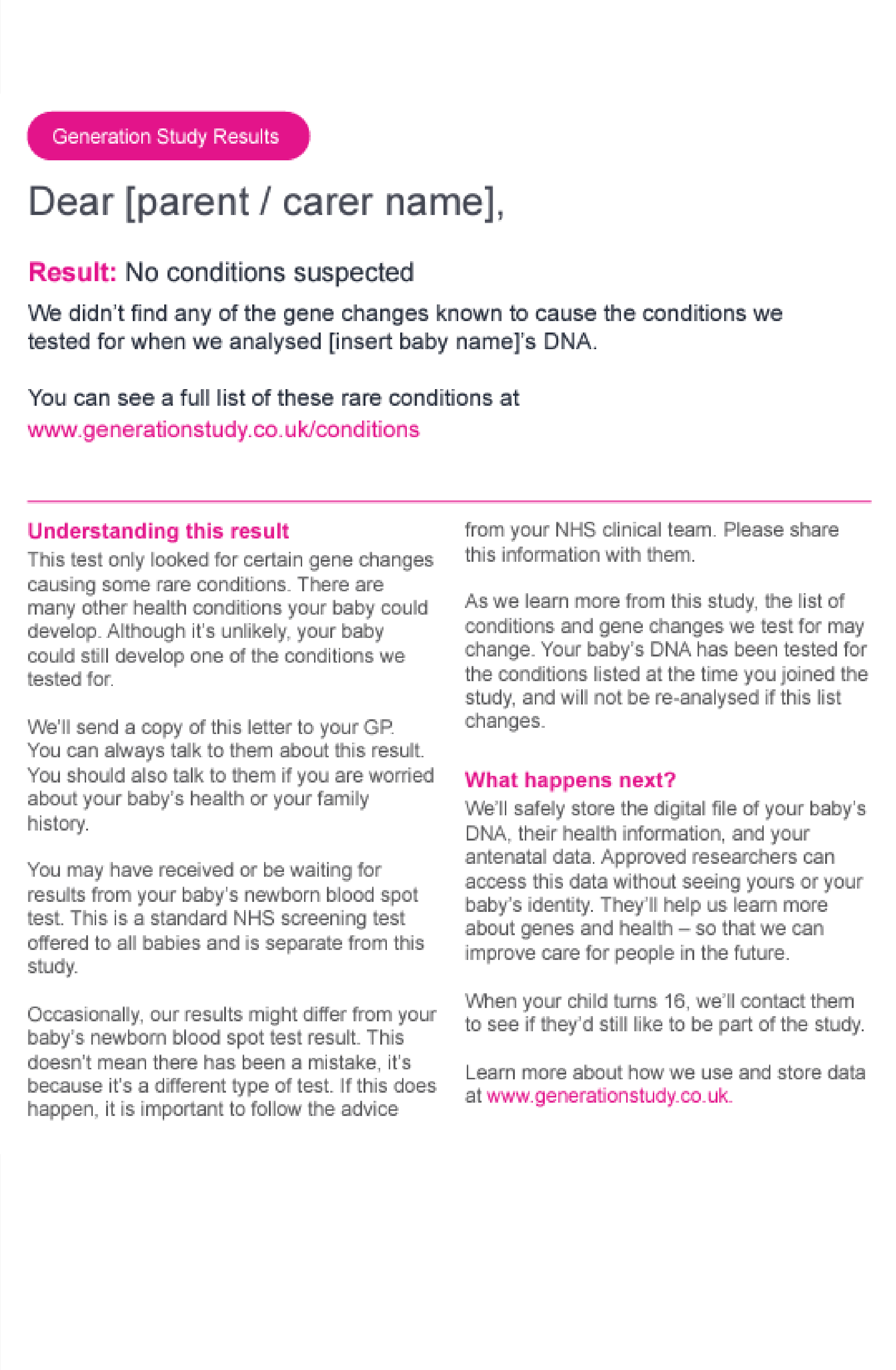
Communicating ‘no conditions suspected’:

- **~99% of babies**
- How: by email/letter from Genomics England, a few months after birth
- Copy also sent to the baby’s GP for their record
- Clarifies difference from NHS blood spot test and that this is not a ‘clean bill of health’
- Includes information about what to expect in the future as a study participant

Communicating ‘sample failure / no results returned’:

- How: by email/letter from Genomics England
- May occur if samples are not collected or the test could not be completed

Version 1.0 11.09.24



Generation Study Results

Dear [parent / carer name],

Result: No conditions suspected

We didn’t find any of the gene changes known to cause the conditions we tested for when we analysed [insert baby name]’s DNA.

You can see a full list of these rare conditions at www.generationstudy.co.uk/conditions

---

Understanding this result

This test only looked for certain gene changes causing some rare conditions. There are many other health conditions your baby could develop. Although it’s unlikely, your baby could still develop one of the conditions we tested for.

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

You may have received or be waiting for results from your baby’s newborn blood spot test. This is a standard NHS screening test offered to all babies and is separate from this study.

Occasionally, our results might differ from your baby’s newborn blood spot test result. This doesn’t mean there has been a mistake, it’s because it’s a different type of test. If this does happen, it is important to follow the advice from your NHS clinical team. Please share this information with them.

As we learn more from this study, the list of conditions and gene changes we test for may change. Your baby’s DNA has been tested for the conditions listed at the time you joined the study, and will not be re-analysed if this list changes.

What happens next?

We’ll safely store the digital file of your baby’s DNA, their health information, and your antenatal data. Approved researchers can access this data without seeing yours or your baby’s identity. They’ll help us learn more about genes and health – so that we can improve care for people in the future.

When your child turns 16, we’ll contact them to see if they’d still like to be part of the study.

Learn more about how we use and store data at www.generationstudy.co.uk.

Bioinformatics

- Bioinformatics is a way of processing information and data from the samples we collect.
- Bioinformaticians process data through applying algorithms that 'make sense' of the data.
- This allows us to identify if a baby has a genetic variant(s) that suggests they have a condition.
- We provide this information to clinicians so that further follow-up testing can establish a diagnosis – or a false positive in a small number of cases.

Variant classification

- The Generation Study will only look for specific variants within certain genes.
- Variants returned through the Generation Study will be class 4 or 5.
- This does not predict the severity of the condition. It is based on the probability that the finding is clinically relevant and linked to the condition.

Variant Class	Score 1	Score 2	Score 3	Score 4	Score 5
Interpretation	Not Pathogenic or of No clinical Significance	Likely Not Pathogenic or Little Clinical Significance	Uncertain	Likely Pathogenic	Pathogenic
Probability of being pathogenic	<0.001	<0.001 - 0.049	0.05 - 0.049	0.95 - 0.99	>0.99

Returning a Condition Suspected Result:

The Process

Version 1.0 11.09.24

Personnel involved in condition-suspected results

Case manager (Genomics England)

- Receives ‘condition suspected’ results from interpretation team
- Prepares result referral pack
- Relays result to regional results coordinator, along with other relevant information and documentation

Regional results coordinator (NHS GMSA)

- Maintains a list of relevant specialists within each NHS GMSA region
- Contacts specialist teams urgently to inform them of a ‘condition suspected’ result
- Follows up to ensure the result has been provided and to collect data on outcomes to support evaluation
- Links to study-funded genetic counselling support

Specialist clinical teams (NHS Trusts)

- Identified with the support of NHS GMSAs and NHS England specialised commissioning
- Communicates condition suspected results to the parents
- Facilitates confirmatory testing and onward care

Other healthcare professionals (NHS Trusts)

- May be involved in the return of results process (condition- and/or location-specific)
- Addresses any challenges in communicating results to families or arranging follow-up tests using a multi-disciplinary approach on a case-by-case basis

Patient organisations

- Provide an invaluable source of information and support
- Genomics England has worked closely with patient organisations via Genetic Alliance UK

Version 1.0 11.09.24

Result referral pack

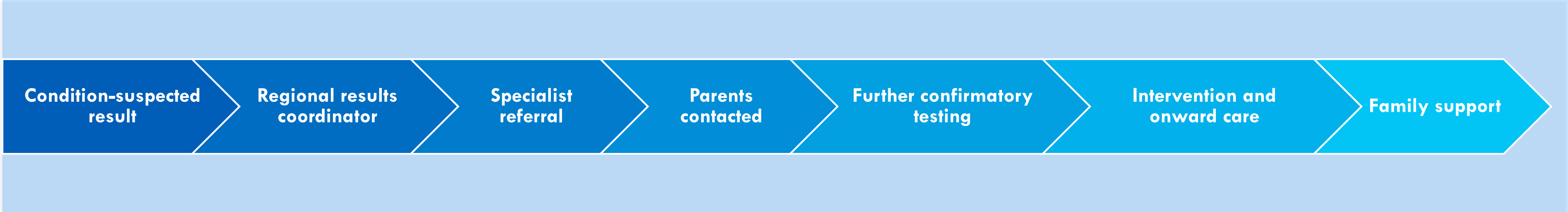
The regional results coordinator will contact the relevant specialist team following receipt of a condition suspected result.

Email to share result with clinician includes:

1. Genomic Research Report
2. Patient contact information
3. Result call guideline
4. Result follow-up email to parent
5. Confirmatory testing pathway
6. Condition information sheet
7. Clinician checklist for returning results
8. Link to diagnostic outcomes form for evaluation

Version 1.0 11.09.24

Providing support



Avenues of support:

- NHS specialist team (consultants, nurse specialists, psychologists, allied health professionals)
- Additional study-funded genetic counselling
- Patient organisations
- Genomics England Service Desk

Results that overlap with NHS blood spot test

The RCC will check result against NBS lab result.

If the result is the same (i.e. NBS is positive and Generation Study is 'condition suspected'), result will be returned to clinician already managing care for the family.

If the results are the same and the diagnosis is confirmed

- Result can go to parents
- Confirmatory testing pathway no longer needed

If NBS does not pick up a condition, but Generation Study does

- RRC pass result to clinical team the NBS would normally refer to
- Clinical team arrange testing and support for family

Contacts

For ALL queries and concerns:

Genomics England Service Desk:

Email: generationstudy@genomicsengland.co.uk

Tel: 0808 281 9535

Genomics England website:
www.genomicsengland.co.uk

Generation Study website:
www.generationstudy.co.uk

Version 1.0 11.09.24

Next speaker

Amy Clarkson

Generation Study Regional Results Coordinator (North East and Yorkshire), North East & Yorkshire Genomic Medicine Service

Type your questions as you think of them during the presentations

Use the Zoom Q&A function

- **Name of presenter** you would like to answer your question
- Brief **question**
- **Your name** (if you wish to do so)

Genomics England | Generation Study

NHS
North East and Yorkshire
Genomic Medicine Service

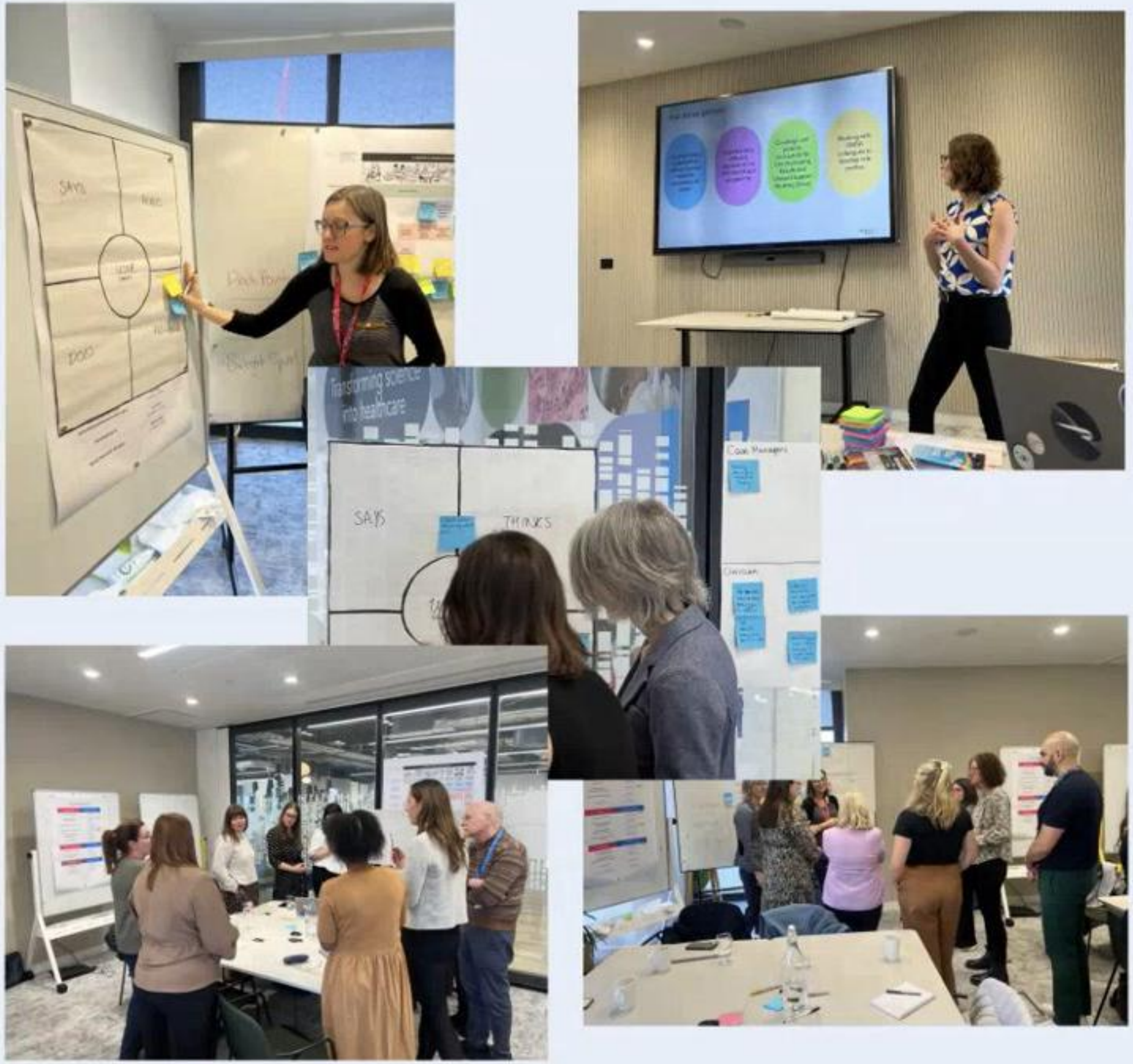
The Role of the Regional Results Coordinator

26 February 2026

Amy Clarkson

Regional Results Coordinator – North East and Yorkshire

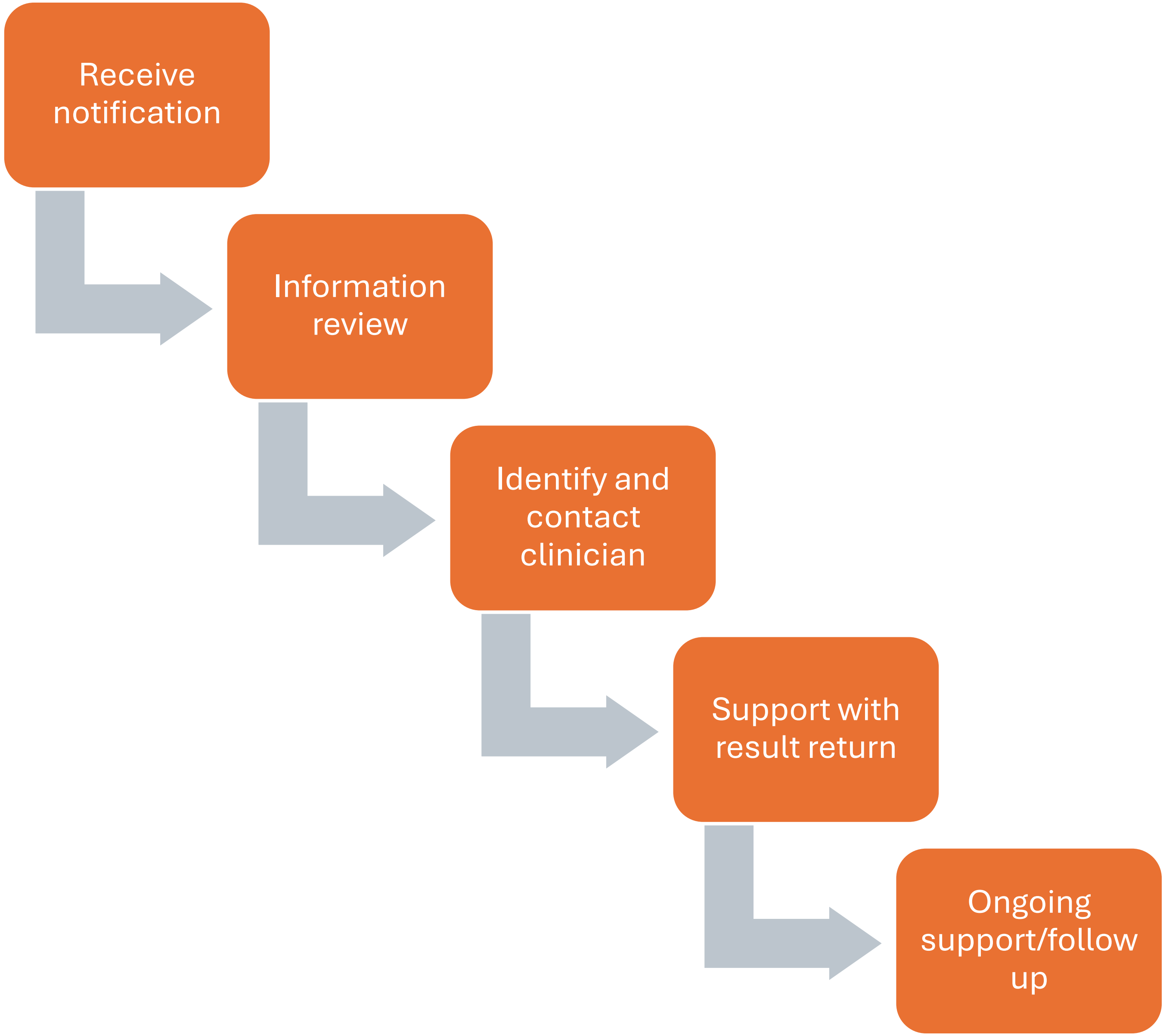
What is a Regional Results Coordinator?



Hereditary Retinoblastoma

- Retinoblastoma is a rare form of eye cancer
- Typically affects children <5
- Associated with the *RB1* gene

Condition Suspected Results


Receive notification

Information review

Identify and contact clinician

Support with result return

Ongoing support/follow up

Genomics England | Generation Study

NHS
North East and Yorkshire
Genomic Medicine Service

Angela Lightfoot and Amy Clarkson

nuth.neygenerationresults@nhs.net

Next speaker

Vicky and Joey Underhay

Parents with lived experience

Type your questions as you think of them during the presentations

Use the Zoom Q&A function

- **Name of presenter** you would like to answer your question
- Brief **question**
- **Your name** (if you wish to do so)

Next speaker

Dawn Wickers

Interim Team Leader, Rotherham Children's Public Health Nursing Service – Family Health Division

Type your questions as you think of them during the presentations

Use the Zoom Q&A function

- **Name of presenter** you would like to answer your question
- Brief **question**
- **Your name** (if you wish to do so)

Welcome and introductions

Dawn Wickers - Health Visitor for Freddie

Rotherham Children's Public Health Nursing Service

NHS
The Rotherham
NHS Foundation Trust

Background

- How I became involved with Freddie following his diagnosis.
- Understanding what this meant for Freddie?
- Liaising with specialist services and professionals.

Working together

- Understanding Freddie and his needs
- Building a therapeutic relationship
- Utilising my role as the family's HV to guide and support Freddie and his family

Conclusions and Reflections

- Personal awareness
- Professional reflection
- Shared learning and education

Thank you ☺

Rotherham Children's Public Health Nursing Service

NHS
The Rotherham
NHS Foundation Trust

Q&A

We will aim to answer as many questions as possible during the live session.

Use the Zoom Q&A function

- **Name of presenter** you would like to answer your question
- Brief **question**
- **Your name** (if you wish to do so)

Generation Study resources for health visitors

- The Generation Study – what is it and how does it work?

Helping participants make informed decisions

Participant Information Sheet

Leaflet

Participant Information Sheet

Leaflet

V1.0 IRAS number: 324562 Date: 31/03/2023

Genomics England | Generation Study

Free and optional

Place your local sticker here

Generation Study

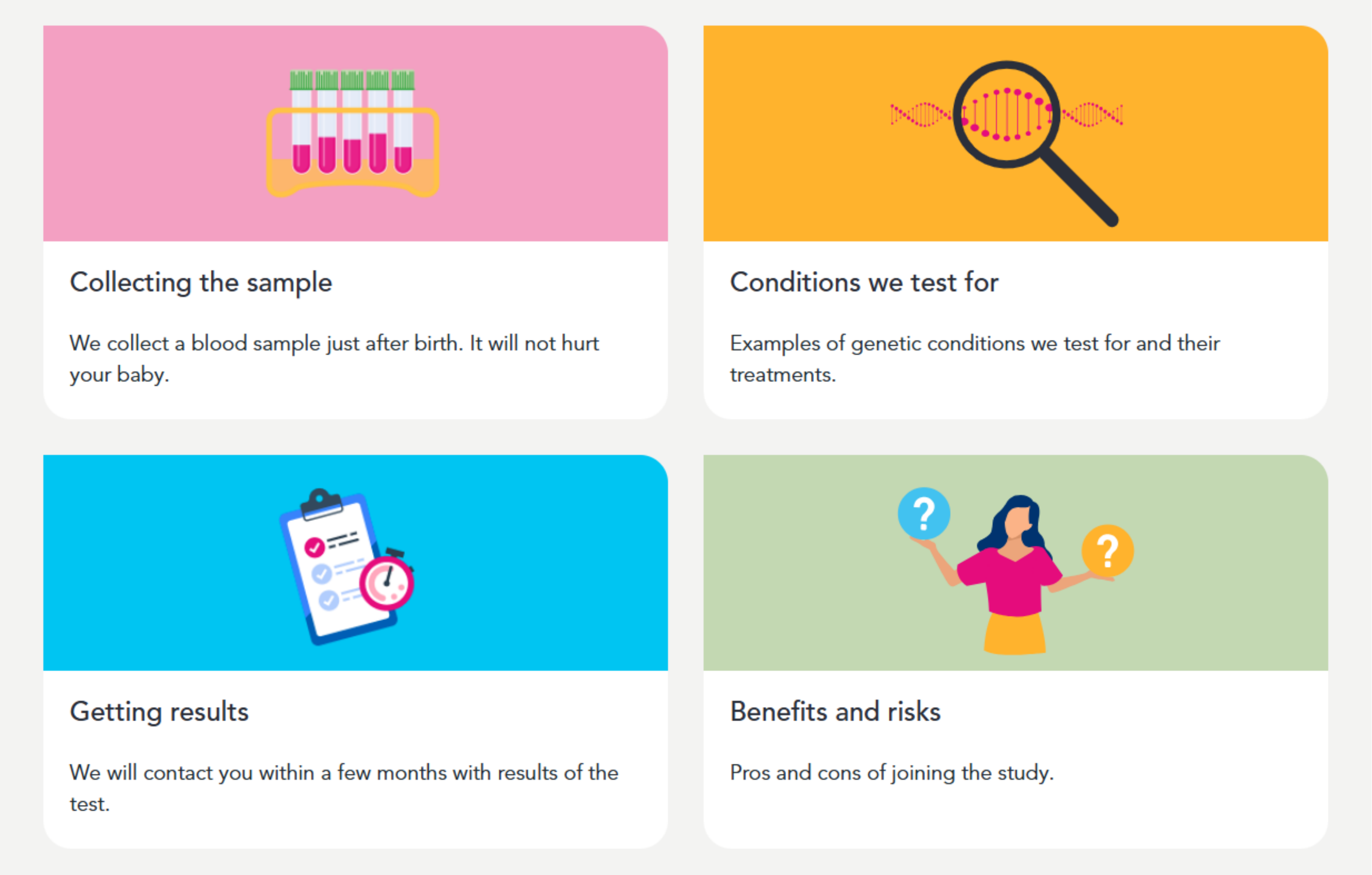
Test your baby for 200+ genetic conditions

Contribute to research that could lead to new treatments. Sign up when you're pregnant, and the test will happen shortly after birth. Any suspected conditions can be treated in the NHS.

Scan to visit our website or talk to your study team

Generation Study resources for health visitors

- Generation Study - Generation Study



Collecting the sample

We collect a blood sample just after birth. It will not hurt your baby.

Conditions we test for

Examples of genetic conditions we test for and their treatments.

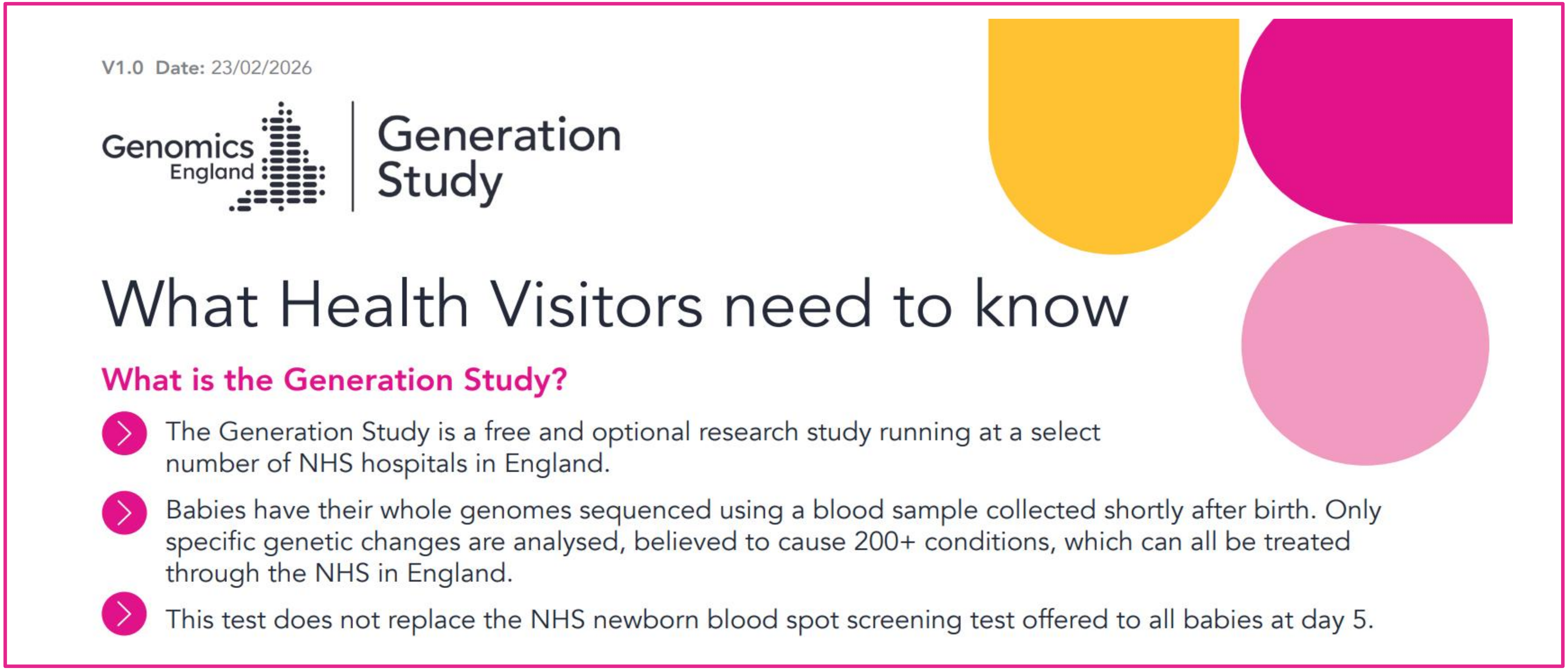
Getting results

We will contact you within a few months with results of the test.

Benefits and risks

Pros and cons of joining the study.

Infographic for health visitors and teams



V1.0 Date: 23/02/2026

Genomics England | Generation Study

What Health Visitors need to know

What is the Generation Study?

- The Generation Study is a free and optional research study running at a select number of NHS hospitals in England.
- Babies have their whole genomes sequenced using a blood sample collected shortly after birth. Only specific genetic changes are analysed, believed to cause 200+ conditions, which can all be treated through the NHS in England.
- This test does not replace the NHS newborn blood spot screening test offered to all babies at day 5.

Current Data and Forums

- Total number recruited: 44,858.
- Samples taken: 34,444.
- Condition suspected results: 117.
- No condition suspected results: 30, 737.
- 75 hospitals recruiting across 48 trusts.

- Data Genomics England 23/02/2026

Monthly forum 3
11/03/2026 12.30-13.30

Over to you!

Help us to tailor our events to meet your needs!

- Submit your anonymised responses to the poll
- What more can we do to support you in your practice?
- Email events@ihv.org.uk with any suggestions

Join us again!

Strengthening Safeguarding Practice: Professional Curiosity and Confident Challenge

26 March 2026 – 15:30-16:30

#iHVInsights

www.ihv.org.uk

Future iHV Member Benefit Event Dates

TOPIC	DATE
Retired Member Network	12 March 2026
Research Network	18 March 2026
Practice Education Network	15 April 2026
iHV Corporate Service Leads Network	16 April 2026

All future dates can be found at: bit.ly/4enHn2H

iHV Learning and Development

Institute of Health Visiting
Excellence in Practice

NEW Practice Assessor and Practice supervisor Development Programme

3/4/12 Mar

NEW Safeguarding Supervision Champions Programme

Safeguarding Supervision

18/19 Mar

Respiratory Health Ambassadors

Our Air, My Lungs

17 Mar

Genomics Ambassadors

Genomics Ambassador

14/21 Apr

PIMH Champions

22/23 Apr

Discounted rates for iHV members

Book today:

Accredited
cpd
Event
THE CPD STANDARDS OFFICE
CPD PROVIDER: 21481
2024-2026
www.cpdstandards.com

http://bit.ly/4mLxsrR

Contact us at:
training@ihv.org.uk

Wednesday 6 May 2026 | Bournemouth International Centre

iHV Evidence-based Practice Conference

From Evidence to Action: Getting it right from the start

iHV Institute of Health Visiting
Excellence in Practice

More information

Hybrid Conference

Thank you so much for joining us

CERTIFICATE OF ATTENDANCE

This is to certify that

has attended the following 1-hour activity:

iHV Insights:

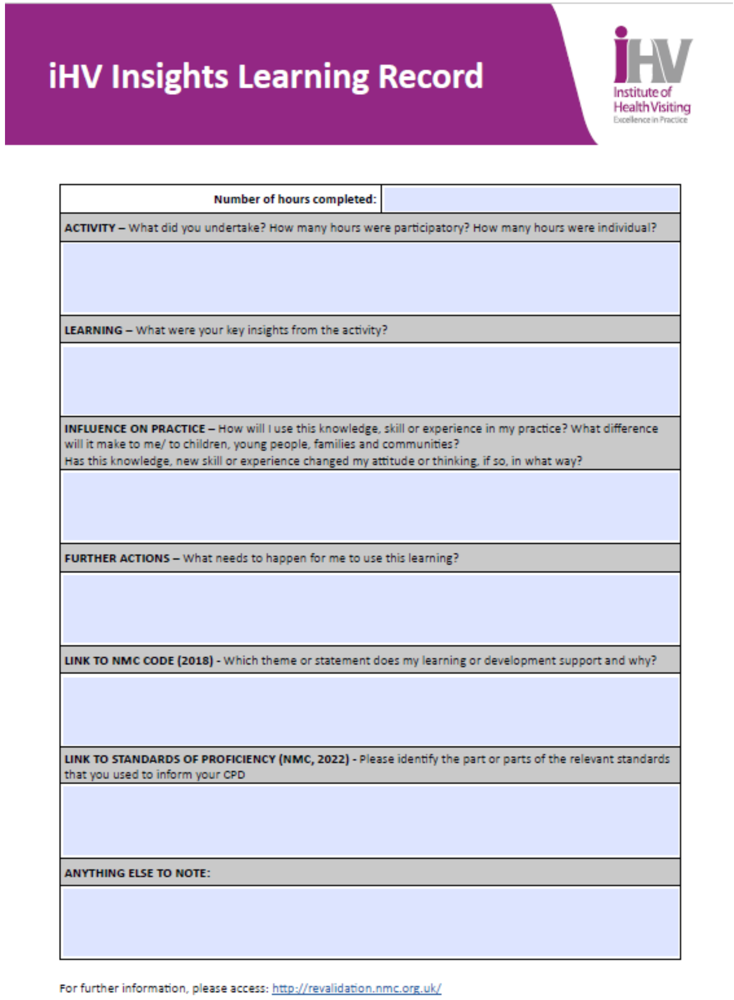
Delivered By:
INSTITUTE OF HEALTH VISITING

Date: :

This certificate should be kept within your CPD record and may be used as supporting evidence in revalidation with the Nursing and Midwifery Council.

Alison Morton
CEO, Institute of Health Visiting

Institute of Health Visiting
A Registered Charity Number 1149745
c/o Royal Society for Public Health, John Snow House, 59 Mansell Street, London E1 8AN
Telephone: +44 (0) 207 265 7352 | Email: info@ihv.org.uk | www.ihv.org.uk



iHV Insights Learning Record

Number of hours completed:	
ACTIVITY – What did you undertake? How many hours were participatory? How many hours were individual?	
LEARNING – What were your key insights from the activity?	
INFLUENCE ON PRACTICE – How will I use this knowledge, skill or experience in my practice? What difference will it make to me/ to children, young people, families and communities? Has this knowledge, new skill or experience changed my attitude or thinking, if so, in what way?	
FURTHER ACTIONS – What needs to happen for me to use this learning?	
LINK TO NMC CODE (2018) - Which theme or statement does my learning or development support and why?	
LINK TO STANDARDS OF PROFICIENCY (NMC, 2022) - Please identify the part or parts of the relevant standards that you used to inform your CPD	
ANYTHING ELSE TO NOTE:	

For further information, please access: http://revalidation.nmc.org.uk/

Continue your CPD!

After this event you will be able to access the following resources on the iHV website:

- A recording of this live session (no recording on other devices is allowed)
- This slide set
- Links and background reading
- A link to download a Record of Attendance & reflection template

Log in at www.ihv.org.uk and head to the Insights page