Targeted gene sequencing and newborn bloodspot screening

Frequently asked questions – Consumers and families

This frequently asked question (FAQ) document has been developed in consultation with consumers and families. Not every question will be included, but we have tried to include answers to common topics.

If there are other topics or questions you would like to see included, please email them to SST@health.qld.gov.au

Please note that information provided in this document is not a substitute for professional medical advice. Always consult a qualified health professional for advice relevant to you and your family's circumstances.

What is targeted gene sequencing?

Genes are instructions that tell our bodies how to grow and develop.

Everyone has small differences in their genes. These differences are called variants. Most variants are harmless and don't affect how the body works. However, some people have variants that do affect how a gene works. These variants may lead to a genetic condition that can cause serious health problems if not treated early.

Most of the conditions we look for in newborn bloodspot screening are genetic conditions. These conditions have been carefully chosen by experts and patient advocate panels.

In Queensland, the lab may use a test called targeted gene sequencing (TGS) to look for these genetic conditions. This test only checks specific genes linked to the health conditions included in newborn bloodspot screening.

Our test doesn't look at or store information about any of your baby's other genes. We do not use your baby's sample or information for any other purposes.

For the full list of conditions that are included in the newborn bloodspot screening program, please access the Australian Government's website at www.health.gov.au/our-work/newborn-bloodspot-screening/.

How will my baby's genetic information be used if we consent to newborn bloodspot screening?

Your baby's blood sample will only be used to look for specific conditions included in the newborn bloodspot screening program.

There are strict rules in place to protect your baby's information, and we do not use your baby's sample or information for any other purposes.

We keep your baby's blood sample and screening information for up to 28 years.

For the first two years, we are required to keep your baby's blood sample so we can check the result again if it's ever needed. After that, you can ask us to destroy it at any time if you'd like. If you would like us to destroy your baby's blood sample after two years, please submit a request in writing to nnsu.pathology@health.qld.gov.au.

If we don't hear from you, we will destroy your baby's sample automatically once 28 years have passed.

Can I change my mind about testing once the sample has been collected?

Consent can be withdrawn at any time by telling your healthcare team. If the testing is already complete, the report will be stored in your baby's medical records, but you can ask not to be told the results.

Do you need to take additional samples from my newborn to perform targeted gene sequencing?

No. With your permission, your doctor, nurse or midwife will collect a few drops of blood in the first few days after birth. This sample is sent to our testing lab in Brisbane where we look for the rare, but serious health conditions included in newborn bloodspot screening.

Sometimes we might call you if we need to collect another blood sample from your baby. This might happen if the first sample wasn't able to be tested properly or we need to complete additional tests.

How is the genetic information stored and what steps are taken to keep genetic information secure?

After we finish testing, we only share your baby's results with your healthcare team such as your doctor, nurse or midwife. We cannot release this information further without your permission, unless required by law.

There are strict rules in place to protect your baby's blood sample and screening information. This means:

- We store the sample securely, and only authorised staff can access it.
- We keep your baby's information on secure systems, and we take several steps to protect you and your baby's privacy. This includes keeping the data from your baby's sample separately from their name, so your baby's identity stays protected.
- Your baby's results are only shared with your healthcare team. These results don't include any information about your baby's genes, unless the screening finds a possible condition and your baby needs some extra tests.
- We always follow Australia's privacy laws and our staff are trained to protect your baby's information.
- We don't share your baby's information unless you give permission or the law requires it.

Under no circumstances will Queensland Health sell your newborn's blood sample or genetic information.

Will the results impact life insurance for my baby or my family?

No. In Australia, national legislation has been passed to protect you and your family from discrimination or differential treatment as a result of genetic testing. Further information on this legislation is available at www.legislation.gov.au/C2026A00035/asmade/text, or by searching *Treasury Laws Amendment (Genetic Testing Protections in Life Insurance and Other Measures) Act 2026* into any search engine.

In Queensland, targeted gene sequencing only checks for changes to specific genes that can cause genetic conditions included in newborn bloodspot screening. This means we don't keep any other information about your baby's genes.

What do I do if a test result comes back positive?

Positive results do not mean that your baby has a condition. It may mean that your baby has an increased risk of a condition. If the result is positive, you will be contacted by a specialist healthcare provider who will discuss arranging more in-depth testing with you. These rare conditions can only be diagnosed and treated by specialised treatment teams.

If your baby's test comes back with a positive result, your healthcare team will contact you to support you and your family through treatment options and further care.

Will a positive result impact other members of my family or future children?

Sometimes these types of tests can show that a genetic condition may occur again in future pregnancies or be present in other members of your family who have not been tested.

The chance of this happening depends on how the condition is passed on. You can discuss this with your healthcare team if your baby is found to have a genetic condition.

Will targeted gene sequencing detect if my baby is a carrier of one of these conditions?

A carrier is someone who has inherited a condition but doesn't have symptoms (or only has mild symptoms). Carriers may be able to pass these conditions on to their future children.

Our tests may pick up that your baby is a carrier of one of the newborn bloodspot conditions.

If we identify your baby may be a carrier for one of these conditions, we may ask you to come in for some extra tests. Your healthcare team will also give your family extra information on what this might mean for your baby and answer any questions you may have.

What if screening finds a gene change, but it's unclear whether it will affect my baby?

There are some conditions that are picked up in the newborn bloodspot screening program that are considered "late-onset." This means symptoms may not develop for babies with these conditions until much later in life, sometimes as late as twenty or sixty years old.

If your baby is found to have one of these conditions, your healthcare team will give your family extra information on what this means for your baby or answer any questions you may have.

If you're a public patient who had your baby at a Queensland Health hospital, we'll work with you and your family to decide what happens next. Your healthcare team can also connect your family with other support services or share helpful information.

If you are private patient who has given birth through a private hospital, we encourage you to discuss what this pathway looks like with your healthcare team.

Will my baby's samples be used for research?

Not without your consent. Clinical research has strict ethical requirements. Queensland Health does not allow the use of newborn bloodspot screening samples without the explicit, informed consent of families.

If a clinical research team would like to use your baby's sample for research, you will be asked to agree and sign a separate consent form.

Where can I get more information on genetic and genomic services?

In Queensland, Genetic Health Queensland provide a statewide clinical genetics service. You can find further information on their website at www.metronorth.health.qld.gov.au/rbwh/genetic-health-queensland.

Further general information on genomics and genomic testing is also available at Genomics Australia at www.genomicsaustralia.gov.au/individuals/what-genomics-might-mean-you.

There's lots of helpful resources available on these websites. However, please be aware that they may also include information for different types of tests. This means that some of the information may not be relevant to targeted gene sequencing, which only looks at changes to specific genes.