Targeted gene sequencing and newborn bloodspot screening

Frequently asked questions – Healthcare providers

This frequently asked question (FAQ) document has been developed in close consultation with healthcare providers. Not every question will be included, but we have tried to include responses to common topics.

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

Please note we have also developed a consumer-focused FAQ which is available on at www.qld.gov.au/health/children/babies/newborn-bloodspot-screening.

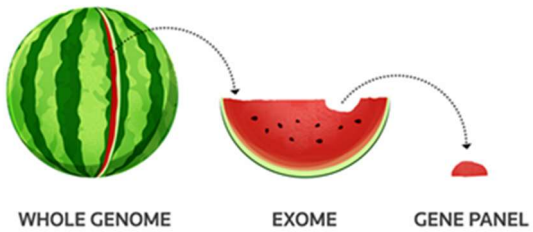
What is targeted gene sequencing and how is it being used in newborn bloodspot screening?

Genes are instructions that tell our bodies how to grow and develop. Each person has many differences (or variants) in their genes. Most variants are harmless and do not impact how the gene 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.

In newborn bloodspot screening, most of the conditions we test for are genetic conditions.

In Queensland, the lab uses a **gene panel** that looks for specific genes in the blood sample and checks for changes that can cause health issues. This testing method is called targeted gene sequencing (sometimes shortened to TGS) because it only "targets" specific genes.



Types of genomic tests
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The testing lab in Brisbane only uses the baby's blood sample to look for the specific genes that lead to genetic conditions included in newborn bloodspot screening. Our lab equipment does not look at or store information on any of the baby's other genes.

For conditions that are not inherited or where gene sequencing is not appropriate, Queensland Health will continue using alternative testing methods to detect conditions included in the newborn screening program. For example, we may continue our current pathology tests to detect abnormal levels of Thyroid-Stimulating Hormone (TSH) to identify newborns with primary congenital hypothyroidism. This is because not all cases of hypothyroidism are inherited and would not be detected through targeted gene sequencing.

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/.

Do we test for all target and non-target conditions?

Queensland Health tests for all target conditions included in Australia's national newborn bloodspot screening program.

Some non-target conditions may also be detected as a result of the types of pathology testing we perform. If a non-target condition is detected as part of the newborn bloodspot screen, results will be provided to the treating team and family following the normal clinical pathway.

What information should I include in my explanation to families?

We recommend avoiding technical language or jargon as part of these discussions and that healthcare providers follow principles and requirements set out in Queensland Health's Clinical Guidelines.

We also recommend that healthcare providers advise families that targeted gene sequencing may be used, but assurance is provided that:

- Only conditions included in the newborn bloodspot screening program will be tested for, and samples cannot be used for any other testing without their permission

- Queensland Health does not collect or keep records of genetic information outside of specific gene variations that are linked to NBS conditions.
- No additional blood samples are required.
- Queensland Health has several measures to protect the security of genetic information and ensure that only the necessary healthcare professionals will have access to records.
- There will be no impact to their baby's or family's insurance as a result of participating in the program.
- Samples and genetic information will **never** be sold.
- Samples, genetic information and results will never be distributed further without permission.

This might look something like the example script below, but providers should seek to understand what matters to their patient and what effective methods would support shared decision-making as part of best practice and person-centred care. This includes patient needs, preferences, values and specific cultural considerations.

We have included references throughout if you'd like further information to support your conversations with families.

EXAMPLE SCRIPT ONLY

Introduction

Hi [parent/carer], I'd like to talk to you about newborn bloodspot screening. This is a quick and simple test that is recommended for all babies in the first two to three days after birth. It's sometimes called the "heel prick test."

The purpose of this test is to check for over 35 rare but serious conditions that aren't always obvious at birth. These include conditions related to how the body processes food, makes hormones, or functions at a genetic level. [Refer to section 1.4 of the Clinical Guidelines]

We've put some of the more common conditions like Cystic Fibrosis and Hypothyroidism on the Parent Information Sheet, including what it is, what can happen if your baby has it and what treatment looks like if we pick it up early. [Refer to Parent Information Sheet or Australian Government website for full list of conditions]

If these conditions are identified early, we can start treatment to reduce the risk of complications such as developmental delays, permanent disability, or life-threatening illnesses. Early detection can make a big difference because starting treatment early can often prevent serious health problems.

We'll also provide you with written information about the test, and I'll make sure you have time to ask any questions you might have.

How the Test Works

We will collect a few drops of blood from your baby's heel in the first few days of life (usually 48-72 hours). This sample is sent to a specialised lab, where we check for specific conditions. This can include things like looking at certain hormone levels or checking if certain genes are working differently from usual. [Refer to section 2 of the Clinical Guidelines]

Here are a few important points to know:

- *We only test for the conditions included in the newborn screening program.*
- *We won't do any other testing on your baby's sample without your permission.*

Benefits of the Test

The test can identify conditions that might not show any signs at birth but could affect your baby's health later.

Early diagnosis means we can start treatment sooner, which can make a huge difference to your baby's growth and development.

Knowing these results could also help your family with future reproductive planning.

Are There Any Risks?
The risks of this test are very low, but it's important to understand them:

- ***Discomfort:*** *Your baby may feel some pain and discomfort when we take the blood sample. We can take steps to minimise any discomfort, like using soothing techniques during the collection. [Refer to section 2.6 of the Clinical Guidelines]*
- ***False Results:*** *While the test is very accurate, no test is perfect. Sometimes it might suggest a condition is present when it's not (a false positive), or it might miss a condition entirely (a false negative). If we do get a positive result or if the results are unclear, we'll always do further tests to confirm.*
- ***Emotional Impact:*** *Waiting for results or being asked to do follow-up tests can sometimes cause worry, stress and anxiety. If that happens, your healthcare team can support you through the process.*

Privacy and Security
We understand that you may be concerned about what happens to your baby's sample and information. Here's what you need to know:

- *Queensland Health doesn't collect or keep any genetic information beyond the specific genes linked to the conditions we're testing for. Genes are instructions that tell our bodies how to grow and develop. If some of your baby's genes are working differently from usual, this can lead to a genetic condition that can cause serious health complications for your baby. Our testing lab has special equipment that essentially pulls these genes out of the blood sample so we can test them without needing to collect any information about your baby's other genes. This is called targeted gene sequencing.*
- *Your baby's blood sample will be securely stored and only used by authorised healthcare professionals.*
- *We also store information from your baby's sample on a secure system which doesn't include information like your baby's name or date of birth. This keeps you and your baby's information safe.*
- *Under no circumstances will your baby's sample or information be shared with others, sold, or used for any other tests without your permission.*
- *There is no impact on your baby's or your family's health or life insurance as a result of the test, even if we do pick up a genetic condition.*

For the first two years, the lab 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 them to destroy it at any time if you'd like.

If they don't hear from you, the lab will destroy your baby's sample automatically once 28 years have passed. You don't need to do anything. [Refer to section 1.3 of the Clinical Guidelines]

What Happens If You Choose Not to Test?
You are able to decide not to test. However, it's important to know that:

- *The conditions we test for are rare but very serious. If they aren't treated early, they can cause permanent health problems, and in some cases, they can be life-threatening.*
- *If your baby doesn't have the test and becomes unwell, you should let your doctor know that your baby wasn't screened. This will help them know what to look for in case it's one of the conditions we usually test for.*

[Refer to section 2.1 of the Clinical Guidelines]

Alternatives to Newborn Screening
There is no other test that finds all the conditions in the newborn bloodspot screen.

Some other tests can look for genes that might lead to genetic conditions, such as reproductive carrier screening. This is done before or during pregnancy and involves testing the blood or saliva of parents to see if they carry certain genetic conditions. However, it doesn't test for all the conditions included in the newborn screening program, and only some conditions are funded by the Australian Government. This means there may be costs if you'd like to complete a more detailed test.

Important Things to Know / Remember

- *We will not test your baby without your permission.*
- *If your baby's results are negative, we won't need to contact you.*
- *We may need to contact you to collect another sample if we couldn't properly complete the tests.*
- *If we find something that needs follow-up, we'll contact you to explain the next steps and provide support. This doesn't necessarily mean your baby has one of these conditions, but we may want to complete some more in-depth tests to make sure.*

What Happens Next?
If you're still pregnant, you don't have to decide right now. You can take your time to think about it, talk to your GP or other members of your healthcare team, and read the Parent Information Sheet or visit our website for more details.

If you've already had your baby, we'll need to take the sample within the first 48 to 72 hours after birth. This can happen in the hospital, or it may need to be arranged once you have been discharged back home.

[Refer to sections 2.2 and 2.3 for initial and repeat samples of the Clinical Guidelines]

Your Decision
I strongly recommend all newborns have the test completed. It is your choice, and I'm here to answer any questions you have.

If you decide to go ahead, I'll ask you to sign a consent form before collecting the sample. If you decide not to participate, I'll record your decision and, with your permission, let your GP or other healthcare providers know so they can provide the best care if your baby ever becomes unwell.

Do you have any questions about the test or the process?

Further information, including points for discussion with families, are available in the newborn bloodspot screening clinical guideline available at www.health.qld.gov.au/__data/assets/pdf_file/0012/1321230/g-newborn-bloodspot-screening.pdf.

What should I say if a family asks me about full / whole genome sequencing or believes that we are sequencing their baby's entire genetic makeup?

Family's may have reasonable concerns related to how their child's genetic information is being used and want to understand the scope of what we collect, analyse, store and report.

We recommend that assurance is provided to families that the sample will not be used to detect conditions or store information outside of the scope of the newborn bloodspot screening program. The sequencing technology we have implemented only looks at specific genes or genomic regions that are related to conditions included in the newborn bloodspot screening program.

How will a child's genetic information be used if the family consent to newborn bloodspot screening?

Because Queensland Health has implemented targeted gene sequencing, genetic information will only be collected relevant to specific conditions included in the newborn bloodspot screening program. Queensland Health will not collect, store or retain any genetic information that is not relevant to the specific conditions included in newborn bloodspot screening.

All genetic information held in Queensland Health records complies with strict legislative, clinical governance and accreditation requirements. Queensland Health has measures in place to protect data security and prevent unauthorised access or breaches to information communication technology systems. This includes storing genetic information without identifying information to help protect family privacy.

All samples and information held comply with Queensland Government and Australian Government legislation.

Are there any changes to newborn bloodspot screening processes?

To support engagement with families, we have made some changes to the Guthrie Card for sample collection. This includes updates to the front sheet (which now provides a link to further information for families), small adjustments to the clinical information aligning to the Maternity and Neonatal Clinical Guidelines, and rewording of the consent statement based on input from multiple clinical and consumer stakeholders.

You can see the revised Guthrie Card layout on our website at http://www.qld.gov.au/health/children/babies/newborn-bloodspot-screening.

There are no changes to the type of sample or method of sample collection. If a family agrees to participate in the newborn bloodspot screening program, Queensland Health will continue to use the bloodspot sample that is collected from their baby in the first few days after birth.

Should we discuss targeted gene sequencing with families during the ante-natal and / or post-natal period?

Yes. In line with the Maternity and Neonatal Clinical Guidelines, we strongly encourage treating teams (including General Practitioners, midwives and nurses, and any other healthcare professional involved in the maternity pathway) to discuss newborn bloodspot screening with families antenatally and also post-partum.

Antenatal discussions will be particularly important and should include general information about NBS and discussions regarding consent. If you would like further information regarding the NBS Program, we encourage you to access the Clinical Skills Development Service free online training available at www.central.csds.qld.edu.au/central/courses/491. This course is available to both public and private healthcare staff at no cost.

Further information that can be provided to families is also available on our webpage at www.qld.gov.au/health/children/babies/newborn-bloodspot-screening. This includes a consumer-focused FAQ.

What advice or support can I provide to families who decline to participate?

There has been no change to recording participation consent as a result of introducing targeted gene sequencing. Treating teams should continue to document declines on the Guthrie Card and in the family's red (Qld) / blue (NSW) / yellow (NT) book and let other healthcare professionals relevant for the family know the screen has been declined. All Guthrie Cards (even from declining families) should be sent to our pathology laboratory.

If you would like further information on engaging with families who may decline or who have declined, resources are available for consumers and healthcare providers at *Partnering with the woman who declines recommended maternity care* webpage, available at www.health.qld.gov.au/consent/clinician-resources/pwdrmc.

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

All results are confidential and cannot be released without the family's permission, unless required by law.

Queensland Health takes significant steps to prevent data breaches and protect all health information that is collected. Additionally, because we are only testing for specific conditions, we will not store genetic information that is not relevant to the conditions included in the newborn bloodspot screening program. This means in the unlikely event of a data breach, information on a child's genetic information outside of these conditions is not at risk.

Under no circumstances will Queensland Health sell a newborn's genetic information.

Who will have access to the child's genetic information?

Queensland Health has several measures in place to make sure that only the family's treating team and staff who are directly involved in performing the testing in our pathology laboratory have access to newborn bloodspot screening information.

Once our pathology laboratory has finished testing the sample, results will only be provided to the treating team as required to support care.

Newborn bloodspot screening results may be included in a child's medical record, including their My Health Record if it is available. My Health Record is operated by the Australian Government and families can manage important information, control who has access to it, and see what has been accessed via their website at: https://www.digitalhealth.gov.au/initiatives-and-programs/my-health-record.

Will the results impact insurance for families?

No. In Australia, national legislation has been passed to protect families 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, our targeted gene sequencing testing only looks for specific genes that aren't working properly. This means we don't store any other information from baby's blood samples except for specific clinical information we need to identify genetic conditions included in the newborn bloodspot screen.

Will the introduction of targeted gene sequencing have an impact on result notification pathways?

No. Healthcare teams in public and private hospitals will continue to receive results through existing notification pathways.

Will the introduction of targeted gene sequencing impact clinical treatment pathways?

No. Healthcare teams in public and private hospitals will continue to lead treatment decisions in partnership with families where conditions are identified.

Where can I get more information on genomics or genetic 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 available through Genomics Australia at www.genomicsaustralia.gov.au/individuals/what-genomics-might-mean-you.

Please be aware that information may include information on genomic testing approaches and services which are broader than targeted gene sequencing. Not all information online or through these sources is necessarily applicable to TGS.