

Newborn bloodspot screening

Clinical Guideline Presentation



45 minutes

Towards CPD Hours

References:

Queensland Clinical Guideline: Newborn bloodspot screening is the primary reference for this package.

Recommended citation:

Queensland Clinical Guidelines. Newborn bloodspot screening clinical guideline education presentation E25.53-1-V1-R30. Queensland Health. 2025.

Disclaimer:

This presentation is an implementation tool and should be used in conjunction with the published guideline. This information does not supersede or replace the guideline. Consult the guideline for further information and references.

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Objectives

Increase knowledge and awareness of:

- Common conditions included in the newborn bloodspot screening (NBS) program
- Indications for initial and repeat screening
- Correct sampling technique
- Care of baby having a NBS sample collected



Information for parents

Georgia is planning to birth at your facility. What points of discussion will you include when you talk with Georgia about NBS?

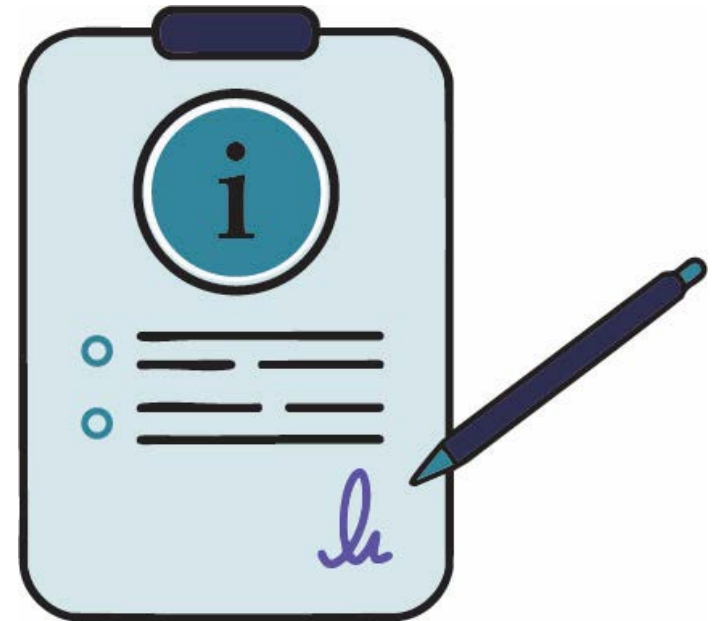
- Purpose of NBS
- Conditions screened
- Benefits and limitations of screening
- Consent
- Storage and retention of collection card
- Sample collection technique
- Timing of collection
- How results are reported
- Indications for repeat screen collection



Consent

Georgia asks you if the NBS test is compulsory. How will you respond?

- Advise NBS is not compulsory
- Parental consent is always required
- NBS is strongly recommended because of significant health benefits for baby if a condition is detected
- If NBS is declined, advise to notify baby's healthcare providers so they can be alert for signs of undiagnosed conditions



Common conditions screened

You talk with Georgia about the 30+ conditions included in the NBS screen and discuss the more common conditions.

Condition	What is it?	What can happen?	Treatment
Cystic fibrosis	Causes problems with the lungs and digestive system	<ul style="list-style-type: none"> problems with breathing, lung infections, digesting food, growth 	diet, medicine, other therapy
Phenylketonuria	A protein (phenylalanine) is not broken down properly	<ul style="list-style-type: none"> developmental delay intellectual delay 	special diet
Hypothyroidism	Not enough thyroid hormone is made	<ul style="list-style-type: none"> intellectual delay 	medicine
Galactosemia	A sugar (galactose) is not broken down properly	<ul style="list-style-type: none"> liver, kidney, eye problems intellectual delay 	special diet
Congenital adrenal hyperplasia	The hormones from the adrenal glands are not made in the right amounts	<ul style="list-style-type: none"> dehydration growth and developmental delay 	medicine
Spinal muscular atrophy	The nerves controlling muscle movement don't work properly	<ul style="list-style-type: none"> muscle wasting growth and developmental delay 	medicine, other therapy
Severe combined immunodeficiency	The body's defence to infection doesn't work properly	<ul style="list-style-type: none"> repeated infections, sometimes severe 	bone marrow transplant

Intellectual delay: slow to learn, think and reason | **Developmental delay:** doesn't reach milestones, difficulty with everyday activities

Sample collection timing

Georgia asks you when the NBS sample is collected. How will you advise Georgia?

- Collection recommended for all babies at 48–72 hours of age
- Repeat collection indicated if:
 - Insufficient milk feeds at time of collection
 - Birth weight < 1500 g
 - Baby has blood transfusion before 48 hours of age (collect before and after transfusion)

Indication and timing of sample	asap	48–72 hours of age	variable^	28 days of age
Early neonatal death or Stillbirth	✓ if possible			
Birth weight < 1500 g		✓		✓ or discharge (whichever is earlier)
Blood transfusion before initial NBS sample collected (48–72 hours of age)	✓ pre-transfusion	✓	✓ 14 days post transfusion	
Milk feed criteria not achieved*		✓	✓ milk feed criteria met	
At laboratory request	✓			

* Milk feed criteria

- Milk feeds contain protein and lactose
- 50% of total daily fluid volume is milk feed (minimum) AND this volume has been given for 24 hours or more

^ Variable

- Cluster sample collection with other blood test requirements
- If there are 2 or more indications for repeat sampling (not including the universal collection at 48–72 hours of age) consult a neonatologist about optimal timing of collection

Changes to repeat screening

Georgia says she has heard there have been recent changes to the indications for repeat sampling. What will you tell Georgia?

Yes in 2025 indications for repeat sample collection were revised with the aim of reducing the number of repeat samples collected while maintaining a high-quality newborn bloodspot screening program.

Unchanged indication

- Blood transfusion before 48 hours of age

Indication removed

- Parental nutrition
- 10% glucose infusion
- Steroids before and/or after birth
- Dobutamine and dopamine
- Babies of a multiple birth

Consolidated indications

- Birthweights < 1000 g and < 1500 g combined as < 1500 g

New indication—milk feed criteria

- Milk contains protein and lactose
- 50% of total daily fluid volume is milk feed AND this volume been given for 24 hours or more

Discomfort during procedure

Georgia says she is worried about baby having a painful test so early in life. How will you advise Georgia?

- Heel prick does cause discomfort
- Discomfort is brief and health benefits are substantial
- Comfort measures usually effective
 - Breastfeeding
 - Skin-to-skin
 - Holding, face to face
 - Swaddling
 - Non-nutritive sucking
 - Sucrose can be offered



Preparation

What will you discuss with Georgia about preparing baby for the NBS test?

Preparation

- Environmental warmth and light
- Warm foot (e.g. socks)
- Position leg lower than level of heart to enhance blood flow
- Collect immediately prior to feed
- Prepare comfort measures (pharmacological and non-pharmacological)

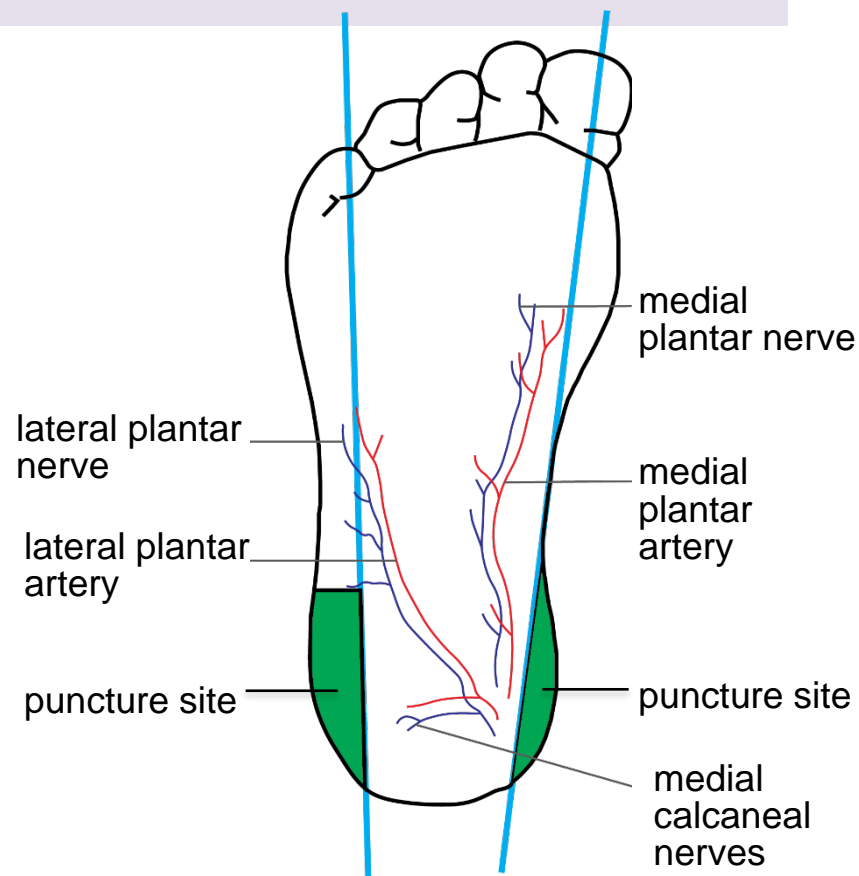


Sample collection technique

Georgia asks you exactly what will happen during the collection of the screening sample. What will you tell her?

Technique

- Clean heel with alcohol wipe and allow to air dry
- Puncture the medial or lateral plantar aspect of heel with lancet
- Wipe first drop of blood with sterile cotton wool
- Apply gentle intermittent pressure to heel



Sample collection management

What will you do/not do to support an optimal sample collection for Georgia's baby?

DO

- Record all clinical information on the card
- Completely fill each circle before progressing to the next circle
- Check blood has soaked through to the reverse side

DON'T

- Milk/squeeze puncture site
- Touch puncture site to the card
- Use petroleum jelly
- Layer successive drops
- Under or overfill the circles
- Mark the circles with pen
- Fill circles from both sides of card