What's new in Neonatology?

Vitamin D recommendations and the Newborn Bloodspot Screening Test

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Specialist Neonatologist
Royal Women's Hospital

Date: 24/10/2025

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Vitamin D: unique physiology

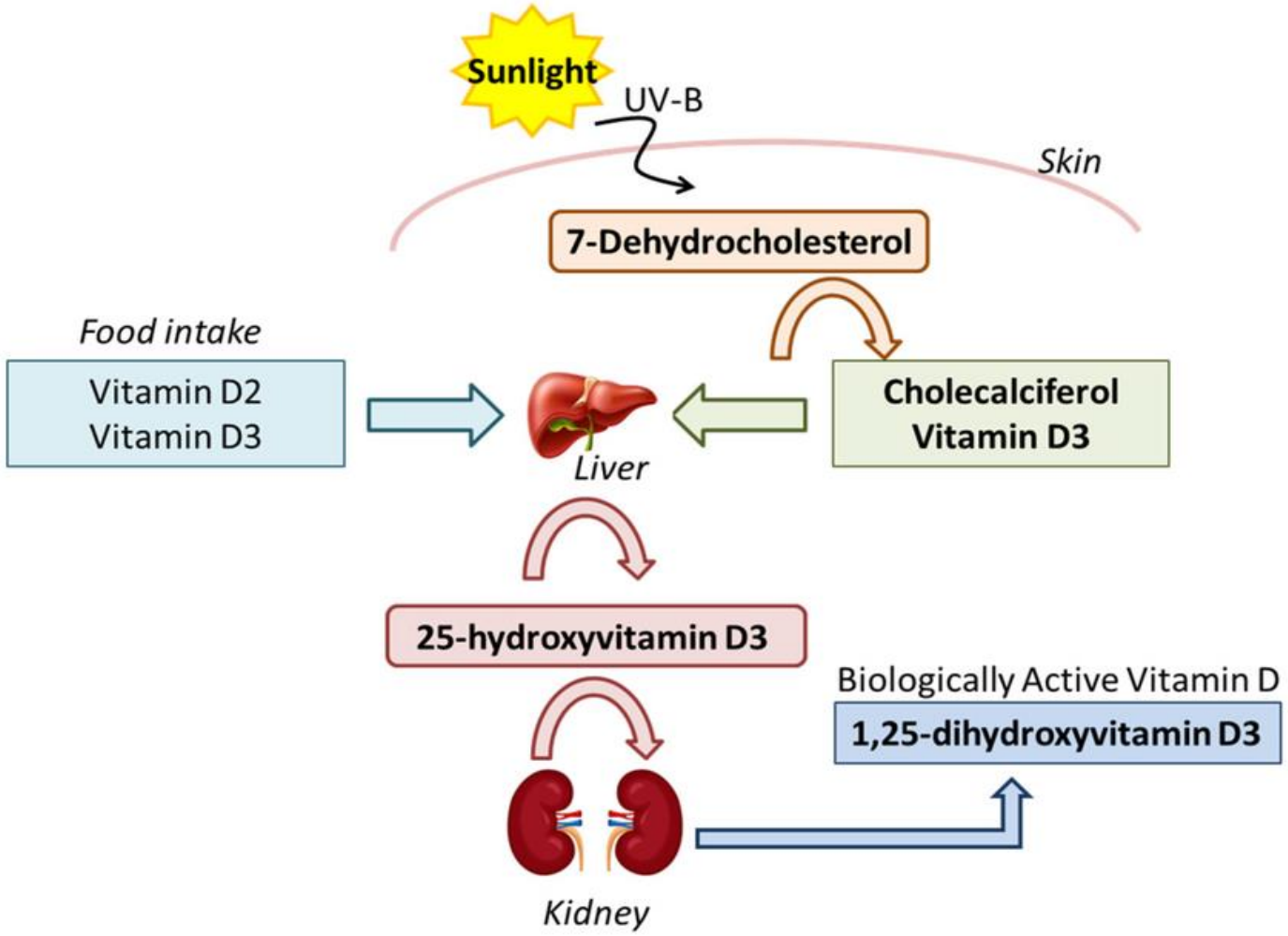

Figure from Aiello, Lombardo, Badelli. *Exploring Vitamin D synthesis in Cardiovascular Health: A Narrative Review.* Applied Sciences 2024

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Vitamin D in pregnancy

Vit D testing funded for high-risk groups only

- Deeply pigmented skin
- Chronic lack of sun exposure
- Fat malabsorption

Ensure adequate calcium intake (pregnancy RDI 1000mg/day)

All women should be supplementing 400-600IU Vit D per day until cessation of breastfeeding

Increase sun exposure
Summer: 6-7 minute walk with arms exposed mid-morning/afternoon
Winter: Up to 40 minutes at noon, exposed as much as possible

Increase food intake of Vitamin D

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Vitamin D in the Infant and Newborn

- Fetal calcium and phosphate accrual peaks at 36-38 weeks of gestation
    - Preterm born babies at risk of osteopaenia of prematurity
- Infant vitamin D is ~50-75% of maternal 25(OH)D concentration at birth

- Neither breast milk or formula milk reliably deliver 400IU vit D per day
    - Delivery of 400 – 1000IU per day to prevent rickets

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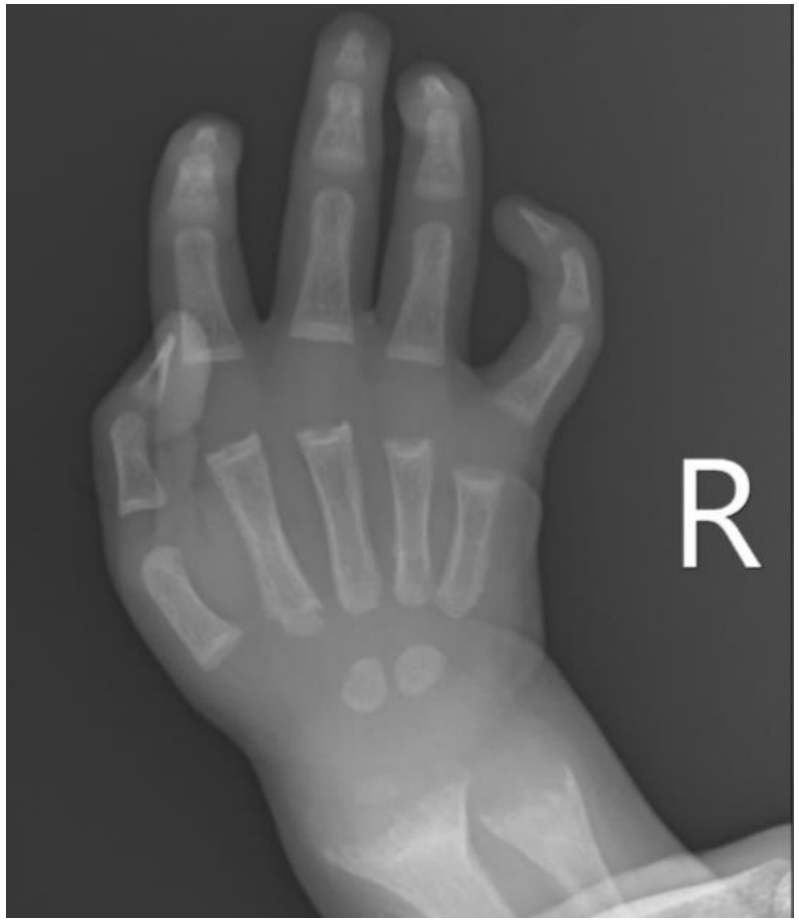
Nutritional Rickets



- Bony features
  - Swollen ankles, wrists, chostro-chondral joints
  - Frontal bossing
  - Delayed tooth eruption
  - Delayed fontanelle closure
  - Bone pain
- Other signs
  - Hypocalcaemic seizures
  - Failure to thrive
  - Muscle weakness and gross motor delay

Case courtesy of Zeeshan Ghias Khan, Radiopaedia.org, rID: 61376

Other vitamin D effects

Research

JAMA | Original Investigation

Effect of Vitamin D Supplementation on Recurrent Wheezing in Black Infants Who Were Born Preterm

The D-Wheeze Randomized Clinical Trial

Anna Maria Hibbs, MD, MSCE; Kristie Ross, MD, MS; Leigh Ann Kerns, MD; Carol Wagner, MD; Mamta Fuloria, MD; Sharon Groh-Wargo, PhD, RD; Teresa Zimmerman, MD; Nori Minich; Curtis Tatsuoka, PhD

Table 2. Primary and Secondary Outcomes by Randomization Group

Outcome	No./Total No. (%)		Risk Difference, % (95% CI)[a]
	Sustained Vitamin D Supplementation	Diet-Limited Supplementation	
Primary			
Recurrent wheezing	42/135 (31.1)	56/134 (41.8)	−10.7 (−27.4 to −2.9)

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Recommendations for Vit D supplementation

- **All babies** should be supplemented with 400 IU Vitamin D per day from birth to 12 months of age
  - Regardless of mode of feeding
    - Global consensus recommendations on prevention and management of nutritional rickets

Some infant and children's Vitamin D products available at community pharmacy:

Ostelin® Vitamin D3 1000IU Liquid	Ostelin® Infant Vitamin D3 Drops	OsteVit-D® Vitamin D3 Kids Drops
		
Strength: 0.5 mL = 1000 IU Dose: **0.2 mL daily** = 400 IU daily	Strength: 1 drop = 400 IU Dose: **1 drop daily** = 400 IU daily	Strength: 1 drop = 200 IU Dose: **2 drops daily** = 400 IU daily

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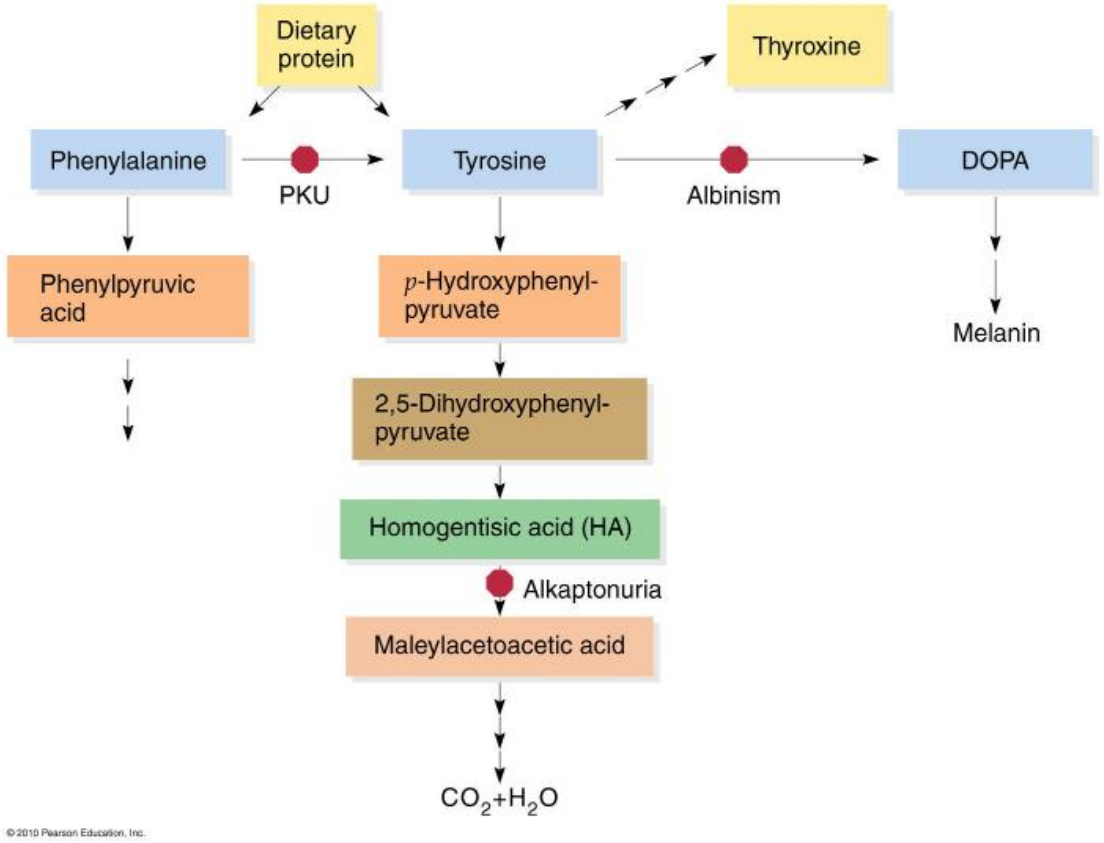
Newborn Bloodspot Screening Tests

Introduced to Australia in the 1960s- Phenylketonuria screening



Phenylalanine metabolic pathway

- Autosomal recessive, single gene mutation
- Phenylalanine Hydroxylase (PAH) Deficiency prevents conversion of phenylalanine to tyrosine
- Phenylalanine builds up in the brain with resultant toxicity
  - Loss of IQ
  - Microcephaly
  - Seizures

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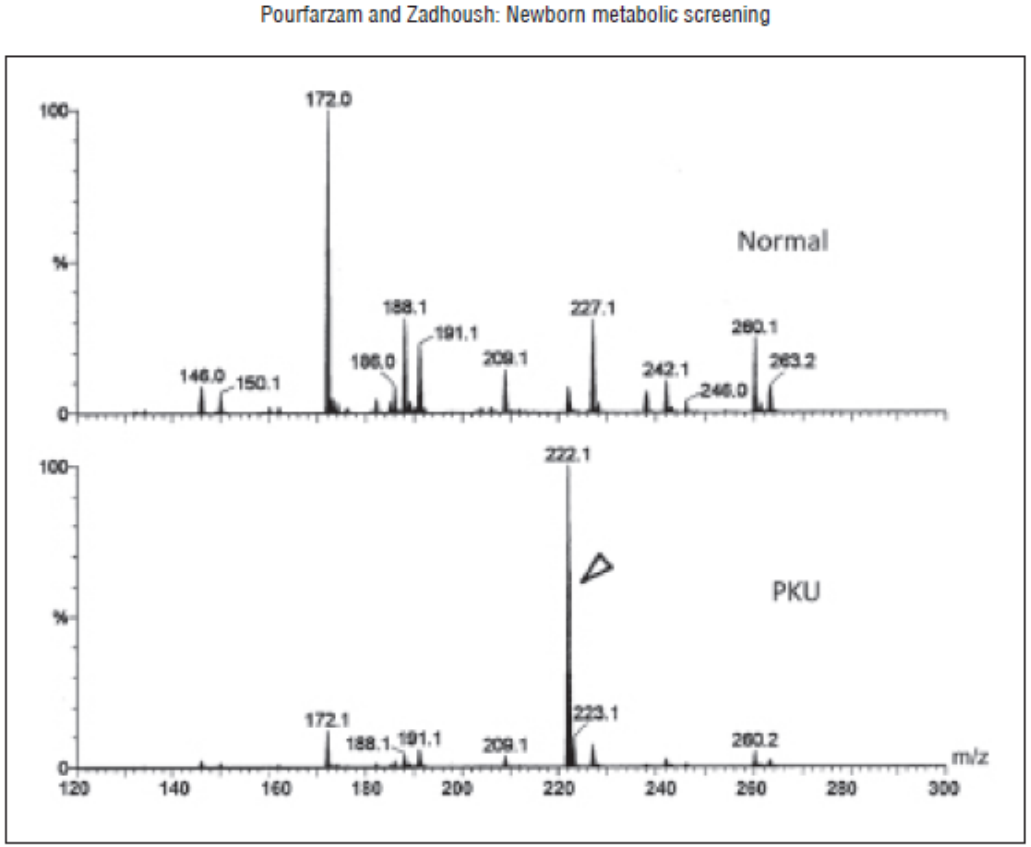
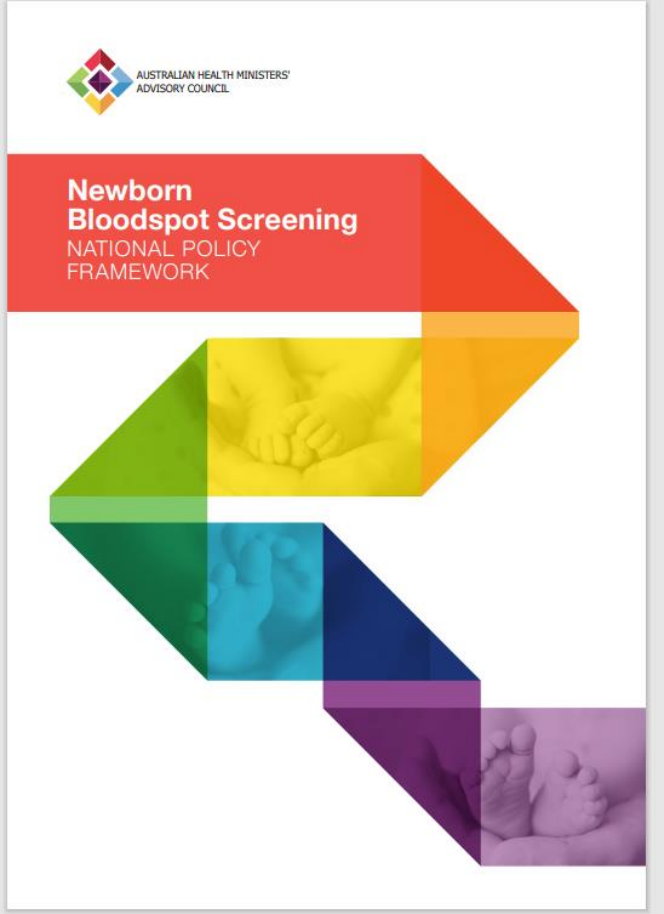
A brief history of Australian NBS

- 1970s
  - Congenital hypothyroid screening
- 1980s
  - Cystic Fibrosis
  - Galactosaemia EXCEPT IN VICTORIA
- 1990s
  - Introduction of Tandem Mass Spectrometry



2017- endorsement of a national policy framework



- Increasing knowledge and technology
- Previous ad-hoc approach duplicates work and risks inequity

Current Target conditions

- 2 endocrine disorders
- 7 amino acid disorders
- 8 fatty acid oxidation disorders
- 10 organic acid disorders
- Cystic fibrosis
- Spinal muscular atrophy (SMA)
- Severe Combined Immunodeficiency (SCID)

News

21 January, 2025

Newborn screening for Galactosaemia: Providing an earlier diagnostic pathway for Victorian babies

Home > News > Newborn screening for Galactosaemia: Providing an earlier diagnostic pathway for Victorian babies

Swenisha Victoria
Communications Officer

On Monday 20 January, Galactosaemia was officially added to Victoria's Newborn Bloodspot Screening (NBS) program.

This addition will help babies receive treatment quickly and improve their health outcomes in the long term.

Non-target disorders

- NBST not specifically designed to test for these disorders
- May be picked up due to abnormal NBST result
- Further testing usually required

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How are target disorders chosen?

- Decision to include a disorder is based on the WHO screening principles
  1. The condition should be a serious health problem
  2. There is a benefit to the baby in early detection
  3. The natural history of the condition should be known
  4. The condition can be suitably tested for
  5. The test is socially and ethically acceptable
  6. There is capacity in the health care system to deal with the result
  7. There is an intervention available

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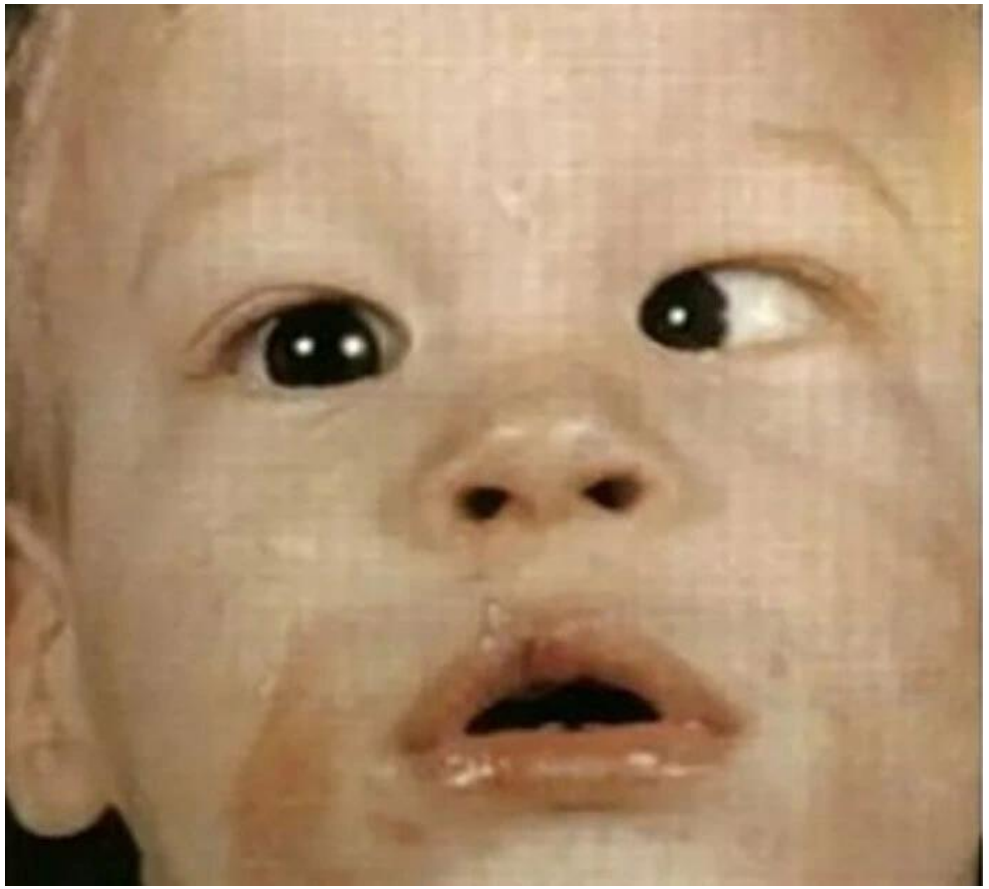
Phenylketonuria



- The condition should be a serious health problem
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Type II glycogen storage disease (Pompe's disease)

- The condition should be a serious health problem
- There is a benefit to the baby in early detection
- The natural history of the condition should be known
- The condition can be suitably tested for
- The test is socially and ethically acceptable
- There is capacity in the health care system to deal with the result
- There is an intervention available

Medical Services Advisory Committee (MSAC) Public Summary Document

Application No. 1774 – Newborn bloodspot screening for glycogen storage disease, Type II (Pompe disease)

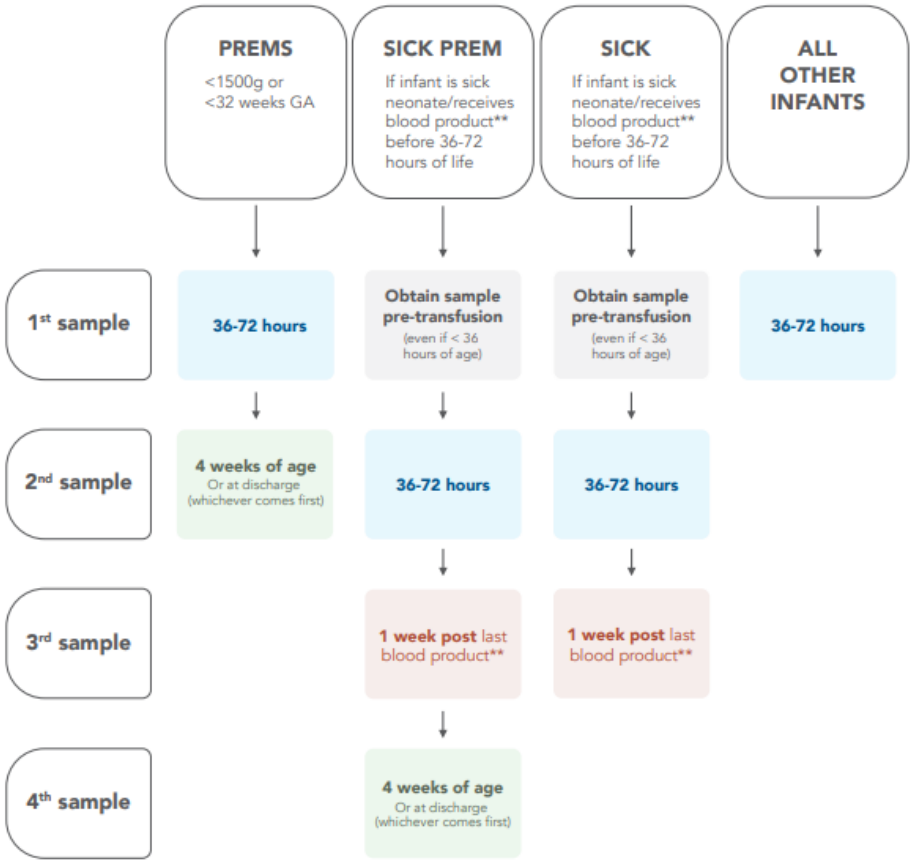
Applicant: **Department of Health and Aged Care - Newborn bloodspot screening section**

Date of MSAC consideration: **3-4 April 2025**

MSAC's advice to the Commonwealth Minister for Health and Aged Care

MSAC did not support newborn screening of GSD II because it may cause greater harms than health benefits. This was because the current screening tests and diagnostic tests (including genetic tests) will mostly identify babies who may develop the adult-onset (late-onset) form of GSD II. These children will not benefit from an early diagnosis because there is no effective early treatment. However, they may have psychological harms from being diagnosed with a potentially serious disease they might or might not develop in the future. MSAC advised that newborn screening for infantile-onset GSD II could be re-considered when a test is available that is able to accurately predict the type of GSD II.

NBS prem/sick neonate protocol



**Blood product means packed red cells or fresh whole blood transfusion or ECMO (this procedure requires exposure to RBC and should be a transfusion of blood products)

VCGS leaders in genetic health

Victorian Clinical Genetics Services
The Royal Children's Hospital
P +61 1300 118 247 W vcgs.org.au
BG-W-305 v1 9/03/2023

NBST Testing Protocol

Available to all babies regardless of place of birth

Declined tests- demographic details should be filled and cards returned to VCGS

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Declining NBS

"You did or did not participate in the heel prick test. Indicate how important the following reasons were for you in making a decision about participation"	Respondents who participated in NBS	"You did or did not participate in the heel prick test. Indicate how important the following reasons were for you in making a decision about participation"	Respondents who declined NBS
Most important reasons first:	n = 804 Mean (SD)	Most important reasons first:	n = 48 Mean (SD)
Because I can prevent my child from getting health complaints from a disorder	4.62 (0.65)	Because of my view of life (e.g. anthroposophical or naturopathic)	3.66 (1.24)
Because I am confident that the results of the heel prick test are reliable	4.16 (0.74)	Because I think the heel prick test is painful for my child	3.40 (1.44)
Because the heel prick test reassures me	4.06 (0.82)	How my data and my child's data are handled	3.38 (1.41)
How my data and my child's data are handled	2.85 (1.17)	Because I can prevent my child from getting health complaints from a disorder	2.74 (1.26)
Because the government arranges and pays for the heel prick test	2.83 (1.20)	Because I am confident that the results of the heel prick test are reliable	2.64 (1.13)
Because I think the heel prick test is painful for my child	2.13 (0.91)	Because the government arranges and pays for the heel prick test	2.57 (1.12)
Because of my view of life (e.g. anthroposophical or naturopathic)	1.65 (0.98)	Because of my faith or religion	2.54 (1.61)
Because of my faith or religion	1.36 (0.74)	Because the heel prick test reassures me	1.93 (1.02)
Because of Corona virus[b]	n.a.	Because of Corona virus[b]	1.74 (1.06)

n.a. = not applicable.

[a] 1 = not at all important—5 = very important. Maximum of 2 missing values per reason.

[b] Only assessed among non-participants in 2020.

https://doi.org/10.1371/journal.pone.0272585.t002

Van der Pal et al. *Parent's views on accepting, declining and expanding newborn bloodspot screening.* PLOS One; 2022

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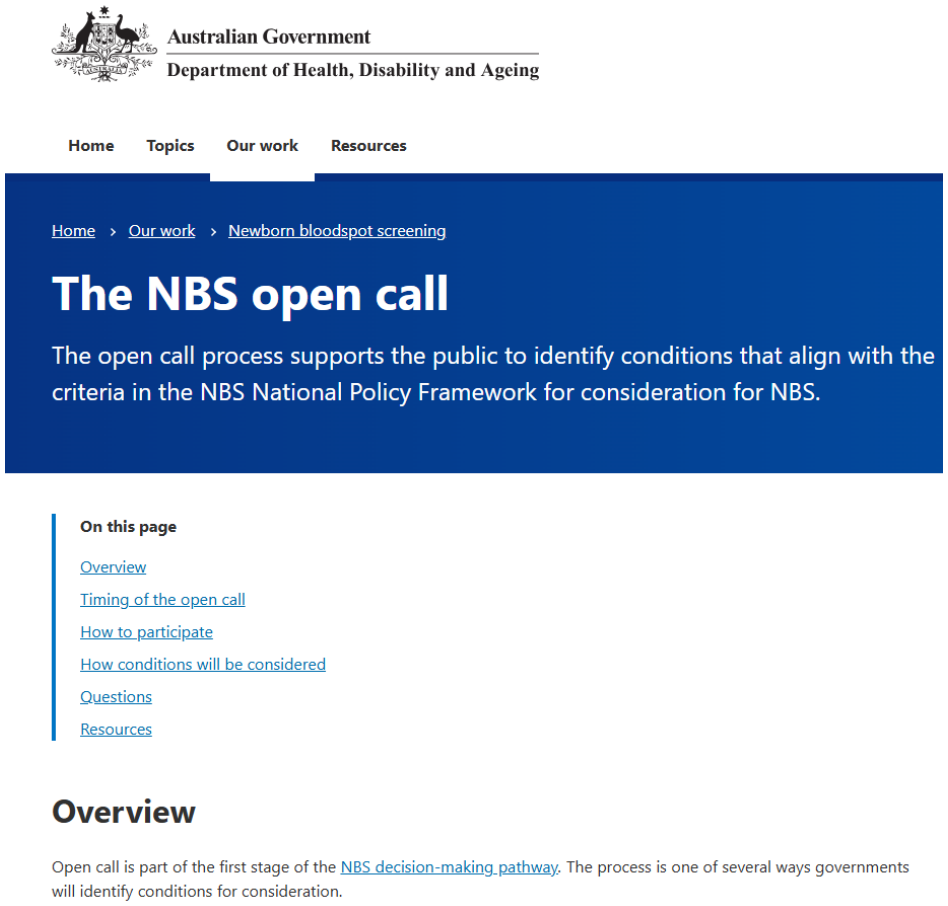
The future of NBST



Australian Government
Department of Health, Disability and Ageing

Home Topics Our work Resources

Home › Our work › Newborn bloodspot screening

The NBS open call

The open call process supports the public to identify conditions that align with the criteria in the NBS National Policy Framework for consideration for NBS.

On this page

Overview
Timing of the open call
How to participate
How conditions will be considered
Questions
Resources

Overview

Open call is part of the first stage of the NBS decision-making pathway. The process is one of several ways governments will identify conditions for consideration.

ELSEVIER

The Lancet
Volume 402, Issue 10398, 22–28 July 2023, Page 265

Editorial

Genomic newborn screening: current concerns and challenges

The Lancet

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Any questions?

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