Newborn bloodspot screening

Policy and guideline

OFFICIAL

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Contacts and Resources

Victorian Clinical Genetics Services

Newborn Bloodspot Screening Laboratory
PO Box 1100, Parkville 3052

Laboratory operation hours:
Monday to Friday 0730 and 1730 (excluding public holidays)

Result and sample enquiries:

- phone: (03) 8341 6272
- fax: (03) 8341 6339
- email: screeninglab@vcgs.org.au

Parent counselling enquiries: phone: (03) 8341 6200

A video about the Victorian newborn bloodspot screening laboratory is available at https://www.youtube.com/watch?v=lT9SDde9b80

Detailed newborn bloodspot screening program information including brochures and the e-learning tool for maternity providers are available at: https://www.vcgs.org.au/tests/newborn-bloodspot-screening

Department of Health

Prevention and Population Health Services

- phone: (03) 8633 4596

Resources

This document is also available online at:

https://www2.health.vic.gov.au/public-health/population-screening/newborn-bloodspot-screening

The full National Policy Framework for Newborn Bloodspot Screening can be found on the Commonwealth Department of Health website at:

https://www.health.gov.au/health-topics/pregnancy-birth-and-baby/newborn-bloodspot-screening

Purpose

The purpose of this document is to provide a high-level summary of the policy positions in the National Policy Framework for Newborn Bloodspot Screening (the framework). It is recommended that information required for program management and decision making should be obtained from the full framework document, or by contacting the Victorian Department of Health.

Key Summary Points

- Newborn bloodspot screening must be offered to families of all newborn babies born in Victoria.
- Parents are to be given information to support decision making prior to screening, ideally in the third trimester of pregnancy, after birth (before the bloodspot sample is collected) and if recalled for further testing.
- Written consent for screening must be obtained before a blood sample is collected from the baby.
- Health care providers[1] are responsible for ensuring that a screening card is submitted to the newborn bloodspot screening laboratory for all births, including for those babies whose parents did not provide consent.
- Health care providers are responsible for ensuring that all births have either a screening result recorded or a completed decline form in the mother's medical record.
- Employers are responsible for ensuring that staff are aware of their role and responsibilities in relation to newborn bloodspot screening and are competent to undertake the task.
- Successful operation of the program requires that all stakeholders are aware of their role and responsibilities in delivery of newborns bloodspot screening in Victoria.

Introduction

Newborn bloodspot screening (NBS) is an important public health program that facilitates the early identification of rare but serious medical conditions that can affect normal development in newborn babies. Screening is a quick, safe and effective way to identify these conditions early and provide the appropriate intervention (usually with diet and/or medication). Early detection can lead to a significant reduction in morbidity and mortality for affected infants.

This program has been available to all babies born in Victoria since the late 1960s. The program is funded by the Victorian Department of Health, which contracts the Victorian Clinical Genetics Services (VCGS) to operate the Newborn Bloodspot Screening Laboratory. The laboratory is located at The Royal Children's Hospital in Melbourne.

Currently, conditions that can be identified through newborn bloodspot screening include phenylketonuria (PKU), congenital hypothyroidism, cystic fibrosis (CF) and approximately 22 other metabolic conditions that affect fat or protein metabolism (see full list at Appendix 1).

Screening is conducted using a small blood sample obtained by pricking the baby's heel, 48–72 hours after birth. This sample is collected onto a special filter paper card and is processed at the Newborn Bloodspot Screening Laboratory in Melbourne.

Parents are required to provide consent for newborn bloodspot screening prior to the blood sample being taken.

National Policy Framework

In December 2017, the Australian Health Ministers' Advisory Council endorsed the Nation Policy Framework for Newborn Bloodspot Screening (the Framework). The framework provides an overview of the elements that are needed to successfully delivery newborn blood screening. It outlines the high-level policies and recommended steps that support high-quality and family-focused newborn bloodspot screening. The framework is intended for clinicians, families, midwives, nurses, policy makers, program managers and scientists.

The framework provides information across the six policy areas:

- Program overview
- Program implementation
- Quality and safety
- Monitoring, evaluation and review
- Decision-making process

While this Victorian Policy and Guideline document provides a summary of the key points from the framework, it is important that information required for program management and decision making is obtained from the full version of the framework.

Roles and responsibilities for newborn bloodspot screening

The following section outlines the key responsibilities of stakeholders involved in newborn bloodspot screening. Further information is available in the Newborn Bloodspot Screening National Policy Framework 2018.

Department of Health

The Victorian Department of Health has overall responsibility for the delivery of newborn bloodspot screening in Victoria. This includes:

- overseeing state-wide policy development and program management
- provision of program funding
- monitoring program performance, quality improvement, evaluation and review

Hospital/Maternity Service Provider

The service provider has responsibility for delivery of the program. This includes:

- development of operational policies and procedures to support newborn bloodspot screening program safety and quality
- provision of information to families
- ensure all families are offered screening
- appropriate record keeping – screening result or refusal, for all births
- transport of samples to the laboratory
- support continuing education and ongoing competency for midwives, nurses and relevant staff about newborn bloodspot screening
- assign responsibility for newborn bloodspot screening to two individuals who will:
  - be available from Monday to Friday via email
  - be the first point of contact for the laboratory
  - ensure timely delivery of screening cards to the laboratory

- ensure timely follow-up of babies with abnormal results or who require further samples to be collected
- ensure that low birth weight babies having a follow-up screening test
- ensure that still birth babies are recorded and notification sent to the laboratory
- ensure every birth has a screening result or a refusal form on file
- ensure that the secure electronic recollection letters are followed up daily
- ensure appropriate stock of screening cards
- support program-wide quality improvement and safety

Midwife

The midwife has responsibility for:

- provision of information and discussion in relation to bloodspot screening with parents
- offer of screening
- obtain written consent
- appropriate sample collection
- record refusal – sign hospital decline form and generate a screening card for the laboratory

Newborn Bloodspot Screening Laboratory

The laboratory has responsibility for management of newborn bloodspot screen testing and providing program support. This includes:

- provision of regular feedback to hospitals/providers in terms of pre-analytical specimen quality issues, such as consent compliance, timeliness issues and sample quality
- ensuring that both the sample and information contained on the screening cards are processed in a timely manner
- timely reporting of results to all hospitals/providers, particularly abnormal results
- timely requests for repeat samples made to hospital/provider
- timely referral to hospital/clinical specialist in order for follow-up diagnostic testing to be arranged, if required
- supplying screening cards to service providers
- developing information for families, in collaboration with other stakeholders where required, and making it available to service providers
- reporting program data to governance and oversight agencies for monitoring

In Victoria, VCGS also provides education to midwives and health care providers on the appropriate collection of samples.

Information for service providers

Informing parents about screening

Health care providers must ensure families[1] of all newborns are offered screening. Before sample collection, staff must ensure parents are properly informed about screening and its importance.

[1] Or legal guardians

To support informed decision making, the information brochure called Newborn bloodspot screening: for the health of your baby should be provided to parents during the last trimester of pregnancy. The information must be discussed with parents and is available in a number of languages at: https://www.vcgs.org.au/tests/newborn-bloodspot-screening.

Written consent for screening

Implied consent for screening is inadequate. Staff must obtain written consent from a parent prior to sample collection. Written consent is provided by reading and signing a section of the screening card.

After reading the brochure and discussing the test with their midwife, one parent is to complete the consent section on the screening card. This section also allows parents to indicate their preference regarding the secondary use of the screening card in de-identified health research.

Making screening cards available for research use is a personal choice and should not deter parents from having their baby screened. Parents have the right to request that the screening card is not available for research use.

In addition, parents should be informed of their ability to request transfer of the screening card after a period of two years.

Decline of screening

While newborn bloodspot screening is strongly recommended for all babies, it is a voluntary program in Australia. If parents wish to decline screening, it is important to discuss their reasons and ensure they are aware of the risks (with referral to a paediatrician or newborn bloodspot screening counsellor at the Victorian laboratory if necessary).

If parents choose to decline, a signed screening card indicating that the test was declined must be provided to the laboratory. A hospital record of decline must also be signed by the parent and filed in the medical record.

- A screening card must be sent to the laboratory – this is a record that parents were offered and declined screening. The laboratory is not aware of a birth until they receive a screening card. This is an important record for the laboratory.
- A decline of screening form must be signed and kept in the mother's record – this is the equivalent hospital record that screening was declined.

Sample collection

Newborn bloodspot screening is carried out using a blood sample obtained by pricking the baby's heel, 48–72 hours after birth. Inaccurate results can occur when the sample is collected outside these times. Screening cards must be sent daily via pathology courier / courier or Express Post to the screening laboratory, after air-drying.

It is important that all still births are recorded by sending a completed screening card to the laboratory (with or without blood on it).

A guideline for sample collection is available at www.vcgs.org.au/nbs/ (under Resources>Downloads).

More information is available in the newborn bloodspot screening e-learning tool for midwives and administrative staff at: https://www.vcgs.org.au/newborn-screening.

A video about the Victorian newborn bloodspot screening laboratory is available at

https://www.youtube.com/watch?v=lT9SDde9b80

Screening results and follow-up

The Newborn Bloodspot Screening Laboratory will issue a report of results on a weekly basis (electronic) to all hospitals/providers.[2] Parents will not be contacted when screening results are normal, but this may take a few weeks. Positive screens will be followed up immediately with parents and the associated hospital/paediatrician by clinical staff from VCGS/The Royal Children's Hospital or Monash Medical Centre once contact details for parents are obtained from the maternity provider.

A repeat collection will be requested by the laboratory for inadequate/contaminated samples/missing data or samples giving borderline abnormal results. The request will be sent by secure electronic mail to the two hospital/ provider nominated with the NBS lab. While concerning for some parents, reassurance should be given that repeat samples usually return a normal result.

Each hospital must identify two newborn bloodspot screening liaison people. These individuals will be the first point of contact for the laboratory. In particular, these persons will be responsible for handling requests for repeat samples and will be required to check that the weekly report of screening results from the laboratory matches the hospital birth record.

- It is the responsibility of all hospitals to make certain every birth is accounted for.
- There may be legal implications for hospitals if appropriate records are not maintained.

Sample storage and secondary uses

Storage and access to the screening cards are governed by state legislation (Appendix 2). All screening cards collected in Victoria, whether in the public private or home setting, are considered public records under the *Public Records Act 1973*.

The newborn bloodspot screening cards must be stored by the laboratory for a minimum of two years in line with National Pathology Accreditation Advisory Council (NPAAC) Requirements for the Retention of Laboratory records and Diagnostic Material. Retention of the screening cards allows for further clinical testing if needed and is also a requirement for laboratory quality control, all of which are primary approved uses of the screening card.

After a two-year period of laboratory storage all screening cards are stored indefinitely in a secure, off-site facility. At this point, parents and individuals 18 years or older have the right to request transfer of the screening card to them after it has been stored for a minimum of two years by approaching the Victorian Bloodspot Screening Laboratory directly.

In line with state legislation, stored screening cards may be accessed by the coroner.

The screening cards may also be used for approved secondary purposes such as ethics approved, de-identified research, subject to the parents' consent. Identified screening cards may be accessed for research use only with the consent of the parents. At the time of sample collection, parents have the right to specify that the sample is not available for de-identified research use.

[2] Independent midwives will work directly with laboratory staff

Appendix 1: List of conditions screened in Victoria

Number	Disorder	Other Names
1.	Hypothyroidism	
2.	Cystic fibrosis	
	Disorders detected by tandem mass spectrometry:	
3.	3-hyd roxy-3-methylglutaryl CoA lyase	HMG CoA lyase
4.	3-methylglutaryl CoA hydratase	3-methylglutaconic aciduria type 1
5.	Argininosuccinic aciduria	Argininosuccinate lyase
6.	Citrullinaemia type 1	Argininosuccinate synthetase
7.	Beta ketothiolase	T2 deficiency, 3-oxothiolase
8.	Maple syrup urine disease	MSUD, branched chain keto acid dehydrogenase (mild/ intermittent forms may not be detected)
9.	Carnitine palmitoyl transferase 1	CPT1
10.	Carnitine palmitoyl transferase 2	CPT2
11.	Carnitine uptake defect	CUD, systemic carnitine defi ciency, carnitine transporter defect, OCTN2 defect
12.	Carnitine-acyl carnitine translocase	CACT
13.	Cobalamin disorders	cblC, cblD, cblF disease
14.	Homocystinuria	Cystathionine betasynthase, CBS (vitamin responsive forms may not be detected)
15.	Glutaric aciduria type 1	Glutaryl CoA dehydrogenase, GA1
16.	Holocarboxylase synthase	HCS, multiple carboxylase defi ciency, MCD
17.	Isovaleryl CoA dehydrogenase	Isovaleric acidaemia, IVA
18.	Medium-chain acyl CoA dehydrogenase	MCAD
19.	Methylmalonic acidaemia	Methylmalonyl CoA mutase, MMA, cblA, cblB disease
20.	Mitochodrial trifunctional protein	Long-chain hydroxy acyl carnitine dehydrogenase, LCHAD, MTP
21.	Multiple acyl CoA dehydrogenase	MADD, glutaric aciduria type 2, GA2, ETF deficiency
22.	Phenylketonuria	PKU, phenylalanine hydroxylase, including tetrahydrobiopterin defects
23.	Propionic acidaemia	Propionyl CoA carboxylase, PA, ketotic hype rg lyc inaem ia
24.	Tyrosinaemia 2	Tyrosine aminotransferase
25.	Very long chain acyl CoA dehydrogenase	VLCAD

NB: other disorders are occasionally detected as part of the newborn screening testing, including disorders affecting the mother, which will be followed-up and referred as defined by the relevant NBS standard operating protocols.

Appendix 2: Relevant legislation and guidelines

In various ways, the following pieces of legislation apply to the newborn screening cards and the blood samples and data derived from them. This legislation governs storage of the screening cards, access during storage and also their appropriate disposal.

Health Records Act 2001 (Vic) – screening cards are 'health information' for the purposes of the health records act. Privacy and Data Protection Act 2014 (Vic) Privacy Act 1988 (Cth)

Public Records Act 1973 (Vic) – all screening cards collected in public and private institutions are classified as public records.

Human Tissue Act 1982 (Vic)

National Pathology Accreditation Advisory Council (NPAAC). Requirements for the Retention of Laboratory Records and Diagnostic Material (Seventh Edition 2018).

Human Genetics Society of Australia (HGSA). Newborn Bloodspot Screening. 2017. https://www.hgsa.org.au/documents/item/8693

Newborn Bloodspot Screening National Policy Framework. 2018. Newborn Bloodspot Screening National Policy Framework. .