

Department of Health

health

Newborn screening
policy and guidelines
2011

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Contacts

Detailed newborn screening program information including brochures and the e-learning tool are available at: www.vcgspathology.com.au/nbs

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If you would like to receive this publication in an accessible format, please phone 03 9096 8975 using the National Relay Service 13 3677 if required, or email: newbornscreening@health.vic.gov.au.

This document is also available in PDF format on the internet at:
<http://www.health.vic.gov.au/nbs>

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Policy statement

Newborn screening is an important public health program that facilitates the early identification and management of babies at risk of having rare but serious medical conditions that can affect normal development.

- Newborn screening must be offered to parents¹ of all babies born in Victoria.
- To support informed decision making, parents are to be given written and verbal information prior to screening, ideally in the third trimester of pregnancy.
- Written consent for screening must be obtained before a blood sample is collected from the baby.
- Hospitals and service providers² are responsible for ensuring that a completed screening card is submitted to the Newborn Screening Laboratory for all births, including those babies who are not tested.
- Hospitals and service 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 screening and are competent to undertake the task.

¹ Or legal guardians.

² Including independent midwives

Rationale

A policy of written consent is being introduced to improve the quality of the screening program and to promote informed choice for parents.

Newborn screening in Australia is a voluntary program. In Victoria, parents are currently required to give verbal consent for a blood sample to be collected for screening.

Research³ has shown parents do not receive adequate information prior to screening and are not aware of the choices available to them. Also, the existing process of verbal consent has been found to be ad hoc and inconsistent.

In response, the Department of Health is implementing a new written consent process to support parents, to strengthen program quality and safety and ensure legal obligations are met.

3 Suriadi, C, Jovanovska, M & Quinlivan, J 2004, 'Factors affecting mothers' knowledge of genetic screening', *Australian and New Zealand Journal of Obstetrics and Gynaecology*, vol. 44, pp. 30-34; Hider, K & Naksook, C 2007, 'Identifying Fundamental Problems with the Victorian Newborn Screening Program', *Health Issues*, no. 90, pp. 36-39.

Newborn screening in Victoria

Newborn screening (NBS) 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 Service (VCGS) to operate the Newborn Screening Laboratory based at The Royal Children's Hospital in Melbourne.

Screening is a quick, safe and effective way to identify newborns at risk of having a rare but serious medical condition. Early identification allows for early intervention (usually with diet and/or medication) and can lead to a significant reduction in morbidity and mortality for affected infants.

Currently, conditions that can be identified through newborn screening include phenylketonuria (PKU), congenital hypothyroidism, cystic fibrosis (CF) and approximately 22 other metabolic conditions that affect fat or protein metabolism (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 an absorbent paper card and is processed at the Newborn Screening Laboratory.

Storage and access to screening cards

Storage and access to the screening cards are governed by a number of pieces of state legislation (Appendix 2). All screening cards collected in public and private institutions in Victoria are considered public records under the *Public Records Act 1973*.

After screening, the cards are stored in the laboratory for a period of two years in line with National Pathology Accreditation Advisory Council (NPAAC) guidelines. This allows further clinical testing if needed and is also a requirement for laboratory quality control. Currently, after the two-year period of laboratory storage all cards are stored indefinitely in a secure, off-site facility.

- Parents and individuals 18 years or older have the right to request transfer of the card to them after it has been stored for a minimum of two years.

In line with state legislation, stored cards may be accessed by Victoria Police with a court order and by the coroner. Cards may also be used for ethics approved, de-identified research. Identified 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.

Roles and responsibilities for newborn screening

Department of Health

- program funding
- program monitoring
- program policy development

Hospital/maternity service provider

- development of hospital policies to support newborn screening program quality and safety
- provision of information to parents
- ensure all parents are offered screening
- appropriate record keeping – screening result or decline for all births
- support continuing education for midwives about newborn screening
- assign responsibility for newborn screening to an individual who will:
 - be the first point of contact for the laboratory
 - ensure timely delivery of screening cards to the laboratory
 - ensure every birth has a screening result or a decline form on file

Midwife

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

Newborn Screening Laboratory, The Royal Children's Hospital

- timely screening of all samples received
- timely reporting of results to all hospitals/providers
- timely requests for repeat samples made to hospital/provider
- timely contact with hospital/clinical specialist to arrange diagnostic testing if required
- provision of regular feedback to hospitals/providers in terms of consent compliance and sample quality

Newborn screening guidelines for service providers

Informing parents about screening

Before sample collection, staff must ensure parents are properly informed about screening and its importance.

The information brochure *Newborn screening: for the health of your baby* should be provided to parents during the last trimester. The information must be discussed with parents and is available in a number of community languages at: www.vcgspathology.com.au/nbs

- Hospitals/providers must ensure parents⁴ of all newborns are offered 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 card. This section also allows parents to indicate their preference with regard to 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 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 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 screening counsellor 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.

⁴ Or legal guardians.

Sample collection

Newborn 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 to the screening laboratory, after air-drying.

Guidelines for sample collection are available here: www.vcgspathology.com.au/nbs/

More information and a video of collection are available in the newborn screening e-learning tool at: www.vcgspathology.com.au/nbs/etool/

Screening results and follow up

The screening laboratory will issue a report of results on a weekly basis (electronic or paper) to all hospitals/providers.⁵ Parents will not be contacted when screening results are normal. Positive screens will be followed up immediately with parents and the associated hospital/paediatrician by clinical staff from VCGS.

A repeat collection will be requested by the laboratory for inadequate/contaminated samples or samples giving borderline abnormal results. The request will be mailed to the relevant hospital/provider, along with an attached screening card. While concerning for some parents, reassurance should be given that repeat samples usually return a normal result.

Each hospital must identify a newborn screening liaison person⁶. This individual will be the first point of contact for the laboratory. In particular, this person 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.

⁵ including independent midwives

⁶ Independent midwives will work directly with laboratory staff

Appendix 1

#	Disorder	Other names
1	Hypothyroidism	
2	Cystic fibrosis	
Disorders detected by tandem mass spectrometry:		
3	3-hydroxy-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 deficiency, carnitine transporter defect, OCTN2 defect
12	Carnitine-acyl carnitine translocase	CACT
13	Cobalamin disorders	cb1C, cb1D, cb1F 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 deficiency, MCD
17	Isovaleryl CoA dehydrogenase	Isovaleric acidaemia, IVA
18	Medium-chain acyl CoA dehydrogenase	MCAD
19	Methylmalonic acidaemia	Methylmalonyl CoA mutase, MMA, cb1A, cb1B disease
20	Mitochondrial 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 hyperglycinaemia
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

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 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.

Information Privacy Act 2000 (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 (Fifth Edition 2009)

Human Genetics Society of Australia (HGSA) Newborn Bloodspot Screening Policy 2011

