


INFORMED PARENTAL CONSENT  
FOR NEWBORN SCREENING IN  
VICTORIA

FINAL REPORT

*December 2005*



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<b>ACKNOWLEDGEMENTS</b> .....	<b>1</b>
<b>EXECUTIVE SUMMARY</b> .....	<b>2</b>
<b>BACKGROUND</b> .....	<b>5</b>
<b>INFORMED CONSENT: DEFINING THE MEANING</b> .....	<b>6</b>
<b>PROJECT DESIGN</b> .....	<b>8</b>
<b>THE FINDINGS</b> .....	<b>11</b>
<b>DEVELOPMENT OF ISSUES PAPER AND RECOMMENDED MODELS</b> .....	<b>26</b>
<b>VALIDATION PROCESS</b> .....	<b>27</b>
<b>FACTORS THAT IMPEDE INFORMED CONSENT</b> .....	<b>31</b>
<b>DISCUSSION</b> .....	<b>32</b>
<b>RECOMMENDED MODELS OF INFORMED PARENTAL CONSENT</b> .....	<b>34</b>
<b>RECOMMENDATIONS</b> .....	<b>37</b>
<b>APPENDIX A: Project Advisory Group Membership</b>	<b>44</b>
<b>APPENDIX B: Overview of documents related to Newborn Screening &amp; Informed Consent</b>	<b>48</b>
<b>APPENDIX C: Background Information about the Current Newborn Screening Program</b>	<b>55</b>
<b>APPENDIX D: Issues Paper Response Template</b>	<b>57</b>

## ACKNOWLEDGEMENTS

This report has been produced for the Department of Human Services by Kim Hider, Charin Naksook and Tony McBride with assistance from Souzi Markos, Mary Caruana and Dell Horey.

Health Issues Centre would like to thank the Department of Human Services Victoria for commissioning and funding this project. We would also like to express our gratitude to a number of people who have made a valuable contribution to the project. They include the members of the Project Advisory Group for their advice and guidance throughout the project's implementation. (See Appendix A)

We would also like to thank the specialists and experts in privacy, informed consent, consumer advocacy, maternity services and genetic services for participating in the key informant interviews; and the numerous health professionals (midwives, maternal and child health co-ordinators, shared care general practitioners, obstetricians, paediatricians and genetic and metabolic disease specialists) who participated in the consultations and validation process.

Our special thanks go to the parents from Meruka Childcare Centre, Dousta Galla Community Health, Frankston Youth Resource Centre, Warragul Playgroup, Collingwood Neighbourhood House Playgroup, the African Women's Group, Vietnamese Women's Group and the antenatal class at Northpark Private Hospital for their contribution to the consumer consultations and validation of the recommended informed consent model for newborn screening in Victoria.

## EXECUTIVE SUMMARY

Newborn screening tests have been available to all babies in Victoria for the last 30 years. The primary purpose of the newborn screening program is the early detection of a range of metabolic conditions; to reduce the risk of severe illness and to allow for earlier monitoring and treatment, which can result in better outcomes for most for these babies.

Both anecdotal and published evidence suggests that many parents in Victoria are unaware that their child has participated in a statewide newborn screening program. Such evidence also suggests they are possibly also unaware of the issues surrounding the rationale for the test, storage of the blood samples, secondary use of the samples and knowledge of who can later access the samples.

As part of a review process of these issues, in April 2005 the Department of Human Services (DHS) commissioned Health Issues Centre to develop effective strategies for providing information to parents about the newborn screening test, to determine the most effective method to obtain informed parental consent for the screening, and to identify the best method of recording parental consent.

Community consultation was undertaken with 155 participants including 69 consumers/parents, 71 health professionals and 15 experts in privacy, informed consent, consumer advocacy, maternity services and genetic services. The consultations identified numerous limitations and gaps in the current newborn screening consent practice and the factors that impede current practices in Victoria in obtaining informed parental consent.

The key findings were that no consistent approach existed among health professionals and health services for the dissemination of (verbal or written) information about the newborn screening program to parents. Significant differences in parent information was also evident between public and private practitioners, with parents using public maternity services often being better informed than those using private maternity services.

The dissemination of the newborn screening parent information pamphlet produced by Genetic Health Services Victoria also varied. The consultations identified that old versions of the parent information pamphlet (pre-2002) were still being used in some hospitals. Where the updated pamphlets were used, parents and health professionals alike were not routinely reading all the information, and therefore continued to be unaware of the additional information about storage, access and secondary uses of the cards.

Whilst all health professionals had a clear understanding of the importance of the newborn screening test for the three main conditions, only a few knew about the additional 20-plus conditions now screened for using tandem mass spectrometry (TMS).

Almost all parents consulted were unaware about the ongoing storage, access and secondary use of the blood samples. When informed of these issues through the consultation, many parents expressed significant concern at not having been adequately informed about this aspect of the newborn screening program. Several parents were also under the assumption that the newborn screening test was compulsory, and felt that not consenting to their child participating in the screening was not a choice they had.

Although many parents questioned the motive for and necessity of indefinite storage of the cards, the consultations still indicated strong parental support for the newborn screening program. After discussing in detail all the available information about the screening program (including storage, access and secondary use issues) all parents stated they would still consent to the collection of the blood samples, because they were aware that the screening has significant benefit to the health and development of their babies. They did, however, want to have options regarding storage, access and secondary uses of their baby's blood samples.

The newborn screening program and the later storage and access of the cards for population health and identification purposes are quite different endeavours, linked currently by an artefact of Victorian legislation and administrative arrangements. Separating the two components will eliminate the complexities for parents and health professionals caused by the current intertwining. It will remove much of the difficulties in designing and implementing a satisfactory and reliable informed consent process for the all-important screening program, and will remove a potential barrier to full uptake of the program by parents.

Stakeholders throughout the consultations expressed strong arguments for both a written and verbal consent model; however, the clear majority of support is for the introduction of a written consent model. Project Advisory Group (PAG) members could not reach consensus on one method of obtaining consent; therefore both verbal and written consent model options are presented in the final recommendations. Other key components of the recommended model of consent which were supported by most PAG members included:

- The development of a two-staged consent process (i.e., screening consented separately from storage, access and secondary use).
- A layered approach to providing parent information.
- The introduction of a checklist for health professionals.
- The development of a result notification system for the newborn screening program.

These will enable the separation of the screening test from the controversial issues surrounding the storage, access and secondary use of the screening cards. As noted above, this will reduce the confusion and possible barriers regarding the primary purpose of the screening program.

It is recommended that expressed consent for the screening alone be sought post-natally by midwives and/or other maternity care providers, prior to the blood sample being taken; and consent for the storage, access and secondary use be obtained at the time of result notification (around 8–10 weeks after birth) by Genetic Health Services Victoria.

The proposed model will also provide better mechanisms to ensure the provision of high quality consumer-friendly information for parents. These include the dissemination of a new parent information pamphlet, the provision of more information on-line, and mechanisms to ensure that both consistent information is provided to all parents and that a discussion takes place between parents and health providers about the newborn screening program. The model also introduces a formal process to provide all parents with information about their baby's screening results. At this later stage, the model includes a mechanism to give parents information about the storage, access and secondary use of their baby's blood samples, and to seek informed consent for some or all of these.

Consumers and health professionals universally supported recording of parental consent to the screening in the mother's files and baby's child health record. The

checklist provides additional and consistent documentation regarding parental consent including date, time, who provided consent, when written information was given to parents, and when a discussion with the health professional about the screening program occurred.

Fifteen recommendations (short- and medium-term) have been developed based on the consultation findings and are presented in detail in this report. Three essential recommendations include that a government decision be made as soon as possible regarding: future storage and access to the Victorian collection of newborn screening blood spots; the piloting and evaluation of both a written and verbal consent model for the newborn screening program prior to any statewide implementation of any new model of consent; and greater consumer input into the ongoing management and future direction of Victorian Newborn Screening Program.

## BACKGROUND

Newborn screening tests have been available to all babies in Victoria for the last 30 years. These screening tests ensure the early detection and treatment of a number of rare but important conditions.

The primary purpose of the newborn screening program is the early detection of a range of metabolic conditions; to reduce the risk of severe illness and to allow for earlier monitoring and treatment, which can result in better outcomes for most for these babies.

Both anecdotal and published evidence suggests that many parents in Victoria are unaware that their child has participated in a statewide newborn screening program. They are possibly also unaware of issues surrounding the rationale for the test, storage of the baby's blood sample, secondary use of the samples and knowledge of who can later access the samples.

As part of a review process of these issues, in April 2005, the Department of Human Services (DHS) commissioned Health Issues Centre to develop effective strategies for providing information to parents about the newborn screening test, to determine the most effective method to obtain informed parental consent for the screening, and the best method of recording parental consent.

### Project Aims

The aim of this project was to explore factors that facilitate informed consent about both newborn screening and its related issues such as secondary uses of the newborn screening card, and to determine those factors that obstruct/hinder/impede informed parental consent. Expected outcomes were effective strategies for health professionals to gain informed parental consent for the newborn screening program.

### Summary of Relevant Documents

A summary of documents and literature relevant to informed consent and newborn screening has been developed. This summary is an analysis of relevant documents including media reports, current research, medico-legal issues, guidelines and policies, and parent information around newborn screening and informed consent. The findings and references are presented in Appendix B.

### Abbreviations and Terminology Used

DHS	Department of Human Services
GHSV	Genetic Health Services Victoria
HIC	Health Issues Centre
PAG	Project Advisory Group
C	Consumer
HP	Health Professional
KI	Key Informant
TMS	Tandem Mass Spectrometry

Throughout this report the term consumers and parents have been used interchangeably. The use of the term parents represents both mother and father (unless stated otherwise), or the legal guardian/s of the child.

## INFORMED CONSENT: DEFINING THE MEANING

The High Court of Australia decision in *Rogers vs. Whittaker* (1992) is commonly cited when examining what constitutes consent. The common law recognises that a patient who is an adult and who is of sound mind has a right to make decisions which affect his or her life and welfare, and to decide what risks he or she is willing to take in receiving medical treatment. Before a patient can properly exercise this right and decide to either consent to or refuse particular medical treatment, he or she needs to have a reasonable understanding of what that treatment involves. As that information can only come from the treating health professional, it is part of their duty to provide a proper explanation of the medical treatment and the risks involved.

Various definitions of informed consent used in law, health and insurance industries have been sourced for the purpose of understanding the context of this research. Some of the definitions found include the following:

- A process by which a subject voluntarily confirms his or her willingness to participate in a particular trial, after having been informed of all aspects of the trial that are relevant to the subject's decision to participate. Informed consent is documented by means of a written, signed, and dated informed consent form.  
([www.brentwoodresearch.com/glossary.cfm](http://www.brentwoodresearch.com/glossary.cfm))
- Appropriate information is provided in order to enable patients to make decisions that reflect their own values. Health professionals subscribe to the values of non-directive counselling and informed consent, in order that patients can make decisions which are best for them, on the basis of full and balanced information.  
([www.antenataltesting.info/glossary.html](http://www.antenataltesting.info/glossary.html))
- Free and informed agreement with what is being done or proposed. Consent can be either expressed or implied (General Insurance Information Privacy Code, on the Insurance Council Australia Ltd website).  
(<http://app01.ica.com.au/PrivacyPrinciples/definitions.jsp>)
- Written consent that is freely given after information has been received and understood.  
([www.hc-sc.gc.ca/english/media/releases/2002/2002\\_34bk8.htm](http://www.hc-sc.gc.ca/english/media/releases/2002/2002_34bk8.htm))
- The Courts require four elements to be present to constitute adequate consent. These include explanation of the procedure and explanation of the risks and benefits, time for inquiries to be answered, and patient cooperation during the procedure (Knapp. R.M. Legal View of Informed Consent for Anesthesia during Labor, *Anesthesiology* 1990; 72:211)
- Informed consent is a legal condition whereby a person can be said to have given consent based upon a full appreciation and understanding of the facts and implications of any actions, with the individual being in possession of all his faculties (not mentally retarded or mentally ill), and his judgment not being impaired at the time of consenting (by sleepiness, intoxication by alcohol or drugs, other health problems, etc).  
([www.en.wikipedia.org/wiki/Informed\\_consent](http://www.en.wikipedia.org/wiki/Informed_consent))

- Informed consent is an ongoing process inherent in patient care. The underlying principle of informed consent is that patients have the right to be told what to expect and to determine what will be done with their bodies.  
([www.rmfm.harvard.edu/risklibrary/rmissues/i\\_infconsent\\_incP.asp](http://www.rmfm.harvard.edu/risklibrary/rmissues/i_infconsent_incP.asp))
- General Insurance Information Privacy Code explains that consent can be either express or implied. Express(ed) consent is given explicitly, either orally or in writing. Express consent is unequivocal and does not require any inference on the part of the organisation seeking consent. Implied consent arises where consent may reasonably be inferred from the action or inaction of the individual.  
(<http://app01.ica.com.au/PrivacyPrinciples/definitions.jsp>)
- Among legal professions, implied consent is “consent when surrounding circumstances exist which would lead a reasonable person to believe that this consent had been given, although no direct, express(ed) or explicit words of agreement had been uttered”.  
([www.massabruening.com/index.php](http://www.massabruening.com/index.php))

Interestingly none of these definitions specify conditions for parents consenting for their children, although the elements identified in the definitions still appear relevant in most cases.

It is clear though that there is no common agreement on exactly what constitutes informed consent. Information provision is key to many but not all of them. Further, the issue of how much information is enough is not really tackled. Health Issues Centre’s perception is that the quantum of information required is person-specific. That is how much is enough for someone to feel they are ‘informed’ will vary from person to person. Some people will want more than others.

There is also no reference to any agreed standards of adequate information provision. Several definitions do point to a baseline of information on: what is proposed; the risks; and the benefits.

Other elements common to several of the definitions below include:

- sufficient time to consider and understand the information;
- expressed consent (i.e. either verbal or written but unequivocal in intent, as opposed to implied consent)
- people making decisions based on their own values.

The following stages were implemented to undertake this project:

### **Stage 1: Establishment of Project Advisory Group**

A project advisory group was established with representatives from the Department of Human Services, the offices of both the Victorian Privacy Commissioner and the Health Service Commissioner, Victorian maternity services, Genetic Health Services Victoria, and consumers. The role of the Project Advisory Group was to comment on the direction and design of the project and the selected methodology, and to provide input into the recommendations and draft final report.

Appendix A provides membership details and terms of reference for the Project Advisory Group.

### **Stage 2: Community Consultations**

#### *Key Informants*

A combination of telephone and face-to-face interviews were conducted with 15 participants, with expertise in privacy, informed consent, consumer advocacy, maternity services and genetic services, to obtain background information and to identify relevant issues that could be used to guide the consultation questions with health professionals and consumers.

#### *Health Professionals*

Focus groups, individual interviews (telephone and face-to-face) were conducted, and email feedback received, from a total of 71 health professionals including midwives, maternal and child health coordinators, shared care general practitioners, obstetricians and paediatricians who work in a variety of different health settings across Victoria.

#### *Consumers (parents)*

Nine focus group interviews were held with a total of 69 consumers (included 61 mothers and 8 fathers) across different regional locations. These included two groups with mothers from culturally and linguistically diverse backgrounds (Vietnamese and Horn of Africa) using language interpreters.

A variety of recruitment strategies were implemented to obtain a wide and diverse statewide consultation. Most parents were recruited through existing networks and groups, not recruited specifically based on their level of interest in the research topic.

The participation of Aboriginal mothers or Aboriginal health workers, and of parents of children diagnosed with existing metabolic conditions, proved quite difficult, even with multiple and varied recruitment strategies. On the other hand, the consultation reached over 30 more participants than initially proposed (see Table 1). However, the broadening of the consultation and the diversity of parents involved was aimed at obtaining a valid representation of stakeholders to inform the recommended model of consent and implementation strategies.

Table 1: Newborn Screening Consultations Reach			
Consultations		Target Groups	Participation Reach
KI	Key Informants	Experts in privacy, informed consent, consumer advocacy, maternity services and genetic services	15
<b>Consumers</b>			
C1	Meruka Childcare Centre	Melbourne Metropolitan areas	8
C2	Doutta Galla Community Health	Western Region	3
C3	Frankston Youth Resource Centre	Southern Region - Young mothers	6
C4	Warragul Playgroup	Rural mothers	9
C5	Collingwood Neighbourhood House Playgroup	Lower socio-economic location	14
C6	African Women's Group	CALD group (recent migrants)	12
C7	Vietnamese Women's Group	CALD group (established migrants)	8
C8	Northpark Antenatal Group	Antenatal parents	9
<b>Health Professionals</b>			
HP1	Mercy Hospital	Public hospital midwives working in various settings (e.g., postnatal, domiciliary, midwifery model)	7
HP2	Maternal & Child Health Co-ordinators	Maternal & child health nurses	25
HP3	Kilmore Hospital Midwives	Rural hospital midwives	5
HP4	Northpark Hospital Midwives	Private hospital midwives	8
HP5	Midwives in Private practice	Independently practicing midwives	3
HP6	Australian College of Midwives	Rural and metropolitan maternity service providers	13
HP7	Shared Care GPs	Specialty medical practitioners	6
HP8	Obstetricians		
HP9	Paediatricians		
HP10	Community Midwives	Midwives working with aboriginal families	1
HP11	Endocrinologists/Metabolic Specialists	Consultants working in the genetics field	3
<b>Total</b>			<b>155</b>

### *Inclusion/Exclusion criteria*

The inclusion criteria for the consumer consultation were women and their partners who had had a baby in the Victorian health system between January 2003 and December 2004. The newborn screening program was changed in September 2002 with the introduction of a parent information handout about the screening test and changes to the timing of the test. Parents prior to this may have had different experiences with the newborn screening program than those who gave birth after 2003. Therefore the latter group had most relevance to this project and a greater chance of recalling their experiences. Parents were invited to participate and consent was totally voluntary. It was made clear that non-participation would have no impact on continuing care with their maternity service providers.

### *Consultation Materials*

Specific background information about the current Victorian newborn screening program was developed for use in the consultations (Appendix C). The information was extracted from the newborn screening parent information pamphlet and from the Department of Human Services and Genetic Health Services Victoria websites. Participants were also sent a copy of the participant information and consent form, as per Department of Human Services Ethics Committee requirements.

Results of the community consultations are presented in the Findings section of this report, and have been used to inform the development of consent models and recommendations.

### **Stage 3: Production of Issues Paper**

An Issues Paper was developed to report the key themes and issues arising from the feedback from health professionals and consumers. After presenting the initial findings to the PAG members, a new model of informed consent was designed along with potential strategies on how best to gain informed parental consent for newborn screening, record information and store blood samples, and address the constraints identified in the consultation process. It also included a summary of documents and literature relevant to informed consent and newborn screening. These were all incorporated into the Issues Paper and used in the validation process.

The Issues Paper is available at [www.healthissuescentre.org.au](http://www.healthissuescentre.org.au)

### **Stage 4: Validation Process**

This stage involved validation of the recommended model of expressed consent, as described in the Issues Paper. Written feedback on the Issues Paper was sought from interested consumers and health professionals who had been involved in the initial consultation process, and who had expressed an interest in participating further.

Twenty-five copies of the Issues Paper were sent by post or e-mail to key participants, including 16 consumers and nine health professionals. They were asked to assess the recommended consent model presented in the Issues Paper and respond using a response template provided. In addition, responses from members of the Project Advisory Group were sought.

The response template used in the validation process is presented in Appendix D.

### **Stage 5: Final Report**

This final report for the Department of Human Services documents all phases and achievements of the project. This was completed within nine months of the project being started.

The findings below are results from consultations with 15 experts in privacy, consent, consumer advocacy, genetics and maternity services, 71 health professionals, and 69 consumers across Victoria. The consultations illustrate the experiences and views of key informants, health professionals and consumers about the current practice of the newborn screening, and their opinions on how the parental informed consent should be obtained.

### 1. Issues arising from key informant interviews

The main issues identified during the key informant interviews that provided background and content knowledge for the project included the following:

- Newborn screening is a standard practice in every developed country; it is done for the benefit of the baby.
- Public confidence in the newborn screening testing process is critical.
- The accelerated pace of genetic testing will require that any consent model needs to take into consideration past, present and future testing options and possibilities.
- Identifying genetic conditions where there are no cures or limited management options can create significant harm to parents and families in the early postnatal period.
- The more specific description of the benefits of keeping the newborn screening cards, the greater justification for doing it.
- The Victorian maternity services hand-held record is currently being piloted and is based on the principles of informed decision-making for women. There is a possibility that information about the newborn screening program could be incorporated into the maternity record before it is disseminated statewide. An antenatal booklet around screening tests has also been developed and could be a good model for the newborn screening test.
- In 2002, the parent information brochure was reviewed and coincided with a statewide education program for health professionals informing them of the changes to the collection time (from 3–5 days to 48–72hrs), and of the additional tests now available using tandem mass spectrometry.
- Changes in technology and public perceptions have occurred over the past few years.
- When considering the consent model, the project will need to consider if there are any disadvantages to individuals from access, storage and secondary use of the newborn screening cards.
- The existence of a DNA database is an urban myth. There is a collection of newborn screening cards with blood spots and a database of results, but the two are stored separately and not linked into one large DNA database, as perceived by some.
- The two issues surrounding consent with the newborn screening program are the screening test performed to provide benefit to the child and family, and the subsequent collection of blood samples kept for public benefit.
- It is difficult to estimate the number of newborns screened compared to the number of babies born each year in Victoria, as this information is not routinely collected and analysed.
- The Australian Health Ministers Advisory Group is currently reviewing newborn screening programs at a national level.

- The issue of administering access to the cards is currently under review in Victoria by the Newborn Screening Review Committee.

## 2. Providing parent information: current practice in Victoria

The Victorian Newborn Screening Program Guidelines state that before the newborn screening test is performed, staff must ensure that parents or guardians are properly informed about the test and the importance of the screening. This information is summarised in the pamphlet produced by Genetic Health Services Victoria (GHSV) titled *Newborn Screening Program*; the Guidelines state that staff should discuss the information outlined in the pamphlet with parents before the test is performed. The guidelines also state that the pamphlets should be given to all mothers prior to delivery and needs to be available for review after their baby's birth. Pamphlets are supplied to maternity units by the newborn screening laboratory and are available in community languages.

- **Who is responsible for providing the information**

Midwives in public maternity services are responsible for providing information about the newborn screening to parents, and appear to do so. In contrast, private obstetricians and shared-care general practitioners (GPs) reported they did not routinely provide written or verbal information about the newborn screening program to families. Several stated that newborn screening was only discussed if parents asked about it during their antenatal visits.

- **What is provided (written/verbal)**

The consultations identified that no consistent approach existed among health professionals on the dissemination of (verbal or written) information about the newborn screening program to parents.

In most instances midwives stated that both written and verbal information about the newborn screening is given to parents. Health professionals in public maternity services stated that the newborn screening parent information pamphlet (published by Genetic Health Services Victoria) is given to all women.

Midwives in private practice stated they did routinely have a detailed discussion with parents about newborn screening and most provided parents with the newborn screening parent information pamphlet. One community midwife stated that they do not provide any written information to their Aboriginal women, as "many wouldn't read the information due to poor literacy" (HP10).

In contrast to the practices described, only a few parents recalled seeing, or being given the newborn screening parent information pamphlet, either antenatally or post-natally. Several stated they may have been given information about the test, but couldn't remember. Many stated that a lot of information was given to them just after their baby was born when they were still recovering from their labour. They found it overwhelming and difficult to take it all in. One parent commented that:

*They did briefly explain to me what it was about, but I guess with everything going on, with everything else being so overwhelming, this was just really down the bottom of the ladder of things. And if they say "oh they need to get it done", you go "oh alright then". (C2)*

Several health professionals also concurred with this stating that:

*Parents often forget that they received information about the newborn screening, as it is just one of the many pieces of information they are given. (HP6)*

- **Timing of information**

Most of the health professionals consulted in public maternity services provided the newborn screening information to parents antenatally. The discussion took place antenatally either at antenatal classes or during antenatal clinic visits. Post-natally, the health professionals discussed the information with parents again before they asked them to consent to the blood sample being collected on their baby. One health professional felt that:

*It would be best if parents are given information during pregnancy (when they register for delivery in a hospital, or with a midwife) and when they come to deliver (or thereafter). This will ensure that parents have had quality time to read and understand the process. (HP11)*

However, midwives in the private maternity hospital consulted did not routinely provide written information about the newborn screening test to consumers until after the birth, when the newborn screening parent information pamphlet was given out along with a variety of other pieces of information in a post-natal pack.

Most parents recalled hearing about the newborn screening test verbally during their antenatal classes or being told about it by a midwife post-natally before the blood sample was collected. Most of the antenatal couples consulted between 35–37 week gestation who had a private obstetrician and were booked into a private maternity hospital stated the newborn screening program was mentioned in their antenatal classes, but no detailed information was provided (written or verbal).

- **Level of information provided**

The level of verbal information provided to parents differed between individual midwives and hospital practices. Some health professionals justified the simplification of information to parents about the newborn screening program to “avoid them being overloaded with information”. A few health professionals also believed that parents would not want to know a detailed level of information about the newborn screening program, and that being given too much information might cause them not to consent to the screening test.

The level of written information was also inconsistent between health services and practitioners. The current parent information pamphlet, published in 2002 by GHSV, provides details about newborn screening, which included: why the test is recommended; details of conditions screened for; storage of the newborn screening card; and secondary uses of the newborn screening cards. However, it became apparent that these pamphlets have only been used by some maternity services. The consultations with health professionals found that older versions of the parent information pamphlet were still being used in some hospitals. These older pamphlets do not have information about storage, access and secondary uses of the blood sample cards.

Where the updated pamphlets were being used, parents and health professionals alike were not routinely reading all the information, and therefore continued to be unaware of the additional information on the pamphlet about storage and access of the cards:

- **Addressing language barriers**

Health professionals in some public maternity services reported that translated materials about the newborn screening information were available in major languages such as Vietnamese. The information was in an information sheet format, produced internally by the hospital and not as a translated version of the Genetic Health Services Victoria parent information brochure. Translated material about the newborn screening was not evident in private hospitals, or used by midwives, obstetricians and shared care general practitioners in private practice that were consulted.

Only a few women from culturally and linguistically diverse (CALD) backgrounds recalled being given the pamphlet in their language. However, some said they were provided with interpreters, and some did not have an interpreter but had relatives who could speak English with them in hospital when the information was discussed.

### **3. Existing knowledge about the newborn screening program**

The consultations identified that a large deficit in knowledge exists about the newborn screening program among both consumers and health professionals.

- **What health professionals know**

All health professionals had a clear understanding of the importance of the newborn screening test, and all health professionals knew about the main conditions (phenylketonuria, congenital hypothyroidism and cystic fibrosis) that were being screened. However, only a few knew about the additional 20-plus conditions screened for using tandem mass spectrometry (TMS).

Very few health professionals were aware that the blood samples are currently stored indefinitely in Victoria. Midwives stated that: "one presumes it is stored for a short period of time for the testing, then thrown out" (HP7). Only a few health professionals knew the cards could be accessed for secondary uses. Some had a small amount of information about these issues from previous media coverage. None of the health professionals interviewed knew that parents or children (18 years old and over) could apply for custodianship of the cards.

One consultant specialist who was aware of Victoria's indefinite storage of the newborn screening cards expressed the view that: "there is enormous community benefit to being able to use the cards for population screening". (HP11)

- **What parents know**

#### *Understanding why newborn screening is recommended*

Most parents had some understanding of why the test is recommended. Parents stated that the newborn screening test was being done to detect a few rare conditions that, if left untreated, could have serious consequences to their baby's health. Most knew at least one or two of the major conditions screened for.

A small number of consumers, especially from lower socio-economic locations and CALD backgrounds, did not understand why the screening was recommended. Comments included:

*The doctor said my baby would be pricked on the heel. She said every baby when born must have it. I don't know why. (C5)*

*They told me after my baby was born. I don't really know what it was about. She cried heaps. (C5)*

Only a few were aware that there were additional conditions tested for. Several parents also questioned why the blood sample needed to be taken so soon after their baby's birth. This created a lot of stress, particularly to mothers from CALD backgrounds (C6, C7). One mother recalled:

*I was scared. I didn't know if it was going to be painful for my newborn twins. I was so worried. (C7)*

Although many felt they were not given a detailed explanation of the reason for the test, the importance of having the screening test was clearly expressed by midwives, which contributed to parents agreeing to have the blood sample collected.

As noted, most mothers from CALD backgrounds did have had interpreters offered to them when discussing the newborn screening test and other postnatal issues. However, the consultations revealed that many of the Vietnamese and Horn of Africa mothers did not have a clear understanding about what they had actually consented for, and confused the newborn screening program with the Hepatitis B immunisation and/or Vitamin K administration.

#### *Knowledge about storage, access and secondary uses*

Nearly all parents were unaware of the information in the pamphlet about the storage, access and secondary use of the blood samples. Those who did have some knowledge of these issues had generally been informed through exposure to previous media coverage.

When parents were told about storage, access and secondary use of the blood samples through the consultation material (Appendix C), many felt they had not been adequately informed about the newborn screening program by the health professionals, and were concerned about why parents were not being informed about the use of the cards and the motives for the indefinite storage. The awareness that blood samples were being kept by a privately owned company and being kept indefinitely raised concerns for a small number of consumers. One mother commented that:

*Well, they've got our children's DNA, so what does that mean for the future? I don't know how it can be used. It just means someone else has got your data...I feel strongly against about them holding that information. (C2)*

Many parents were concerned about who could access their child's newborn screening card without their knowledge. Access for medical research and forensic use was generally well accepted, but there was greater concern regarding police access to the cards. A small number of parents asked if they could have their baby's cards back. Others said:

*The blood samples of our babies shouldn't be used for secondary uses without consent. If they are used for medical reasons, it may still be OK, but about the police check, I am not sure. There are potentials for misuse. (C1)*

*It is unforgivable if they said they take blood samples for the good of your baby, and then use it for something else. (C1)*

Younger mothers (C3) and mothers from CALD backgrounds (C6, C7) were less concerned about storage, access and secondary uses, but believed that parents should be told that this occurred. One young mother stated that:

*It's reassuring that my child's DNA is being stored.*

And

*If it was going to be destroyed, I would prefer to have the option of keeping it. (C3)*

The antenatal parents consulted were very concerned when told about storage, access and secondary uses of their unborn child's blood sample. These parents strongly vocalised the need for parents to have some choice about the storage and secondary uses of the newborn screening cards (C8).

Most health professionals supported parents being informed about storage, access and secondary uses of the screening cards, and supported parents having options about these issues, but they did not want to carry the responsibility of providing these options to parents. A rural obstetrician stated that:

*Parents should have options to have the card back, or destroyed after two years, or kept indefinitely for future research. It gets down to consumers. It's their samples. They are to be informed about secondary use in every case. They should consent every time someone accesses it. (HP8)*

#### **4. What information should parents know**

Parents were asked how important it was to be informed about various components of the newborn screening program, before they were asked to consent to their child's participation in the screening program. Table 2 demonstrates that all parents felt it was essential to have the rationale for why the screening test is recommended clearly explained to them. A majority of parents wanted more details about the conditions screened for in the newborn screening program, or at least information about where to access additional information about all the other conditions tested.

All parents felt it was important to be told about storage, access and secondary uses of the newborn screening cards. Of these, 83% or more of parents felt it was essential to be informed about these issues, whilst 17% or less saw it as important but not vital.

Table 2: Parents' responses to information provision

	Essential to Know	Important to know but not vital	Not important to know
Why the screening test is recommended	69	-	-
Details of the conditions screened for by the test	64	5	-
Storage of the newborn screening blood cards	60	9	-
Access to the newborn screening cards	58	11	-
Secondary uses of the newborn screening cards	57	12	-

While most health professionals felt it was important that parents were informed about storage, access and secondary uses of the screening cards, a few did not feel it needed to be part of the routine information provided. One stated that:

*Parents don't need to know this level of information routinely... but health professionals should have information in case they are asked about storage, access and secondary uses. (HP7)*

One of the consultant specialists felt that information about newborn screening:

*Needs to be written, simple and not overwhelming, and not to be too detailed about the extended metabolic screening. (HP11)*

Others stated that:

*Parents should be provided with written information. Verbal information is inadequate. Many midwives do not know exactly what this program is about. Written information should be comprehensive and include all issues surrounding the screening itself. Information must be comprehensive and include the fact that cards are stored and will not be given to parents for the two-year quality assurance period. (HP11)*

*Parents need to know how the samples are stored, who is responsible for the cards, what the procedures for accessing them are and what constitutes a proper secondary use of the cards. It must be remembered that the primary consent for screening is "only" for screening. (HP11)*

## 5. Obtaining parental consent: current practice in Victoria

The current Victorian Newborn Screening Guidelines state that staff need to obtain verbal consent from parents or guardians before performing the test. There should be documentation on the mother's/baby's file stating that there has been discussion about the newborn screening test, and the file should also show a record of completion of the test.

The guidelines also state that if parents wish to refuse the test on behalf of their baby, they should be referred to a newborn screening counsellor at GHSV for urgent discussion. Any parents refusing the test will need to sign a written statement showing that they understand the potential risk to the healthy

development of their baby. They note that the expanded program will diagnose babies for whom urgent treatment may be life-saving. Refusals must be documented and signed in the mother's/baby's file. A newborn screening sample card has to be completed with refusal written on it, and sent to the laboratory. The consultations told us the following:

- **Parental consent given verbally**

All midwives stated that they always got consent from parents to obtain the newborn screening blood sample, but some questioned whether it could actually be considered informed consent given the limited amount and type of information that is presented to parents.

Parents stated that consent was always provided verbally at the time when the midwife was about to take the blood sample. Fathers were rarely asked to consent to the newborn screening test, nor routinely provided about the newborn screening program. The Newborn Screening Program Guidelines make no reference to the inclusion of fathers in the information to parents about the screening test, only to mothers. However, they do refer more broadly to parents or guardians being asked to give consent.

While the consultation clearly identified most parents were unaware of the number of tests screened for, or the indefinite storage, access and secondary uses of the screening cards, all parents stated they would still have consented to their baby participating in the newborn screening program if presented with such information.

- **Compulsory or voluntary participation**

Several parents were under the assumption that the newborn screening test was compulsory. They felt not consenting was not a choice they had. Some women from the Horn of Africa, especially those with language difficulties, felt they did not consent to the newborn screening test, and that it was something they were told just needed to happen to make sure their baby was healthy. This limited information appeared to contribute to many women from CALD backgrounds feeling very concerned and anxious about the pain of the heel prick to their babies, and about what problems the test might reveal.

- **Timing of consent**

Most parents and health professionals supported consent being obtained post-natally. One consultant believed that: "Parents will not remember a thing beforehand, as the birth is a big barrier" (HP11).

Several parents also did not know and questioned why the test needed to be done so early after the baby was born. These parents felt that immediately post-natally was not a good time to be first told about the newborn screening program and be asked to give consent, as they were often exhausted and still recovering from their labour and birth experience.

- **Confusion over consent**

Many parents had problems comprehending what informed consent was. Some parents, including several from CALD backgrounds, were confused about what they had been asked to consent to. Consent for the newborn screening program was occasionally confused with consent for the Hepatitis B vaccination and Vitamin K administration. This was not helped by the timing and variability of each consent process by the different health providers.

- **Recording consent and documentation**

Midwives stated that the time and date when the newborn screening blood sample is collected is routinely recorded in the mother's notes and in the baby's child health record.

However, it was not common practice to note if a discussion had taken place with parents and that verbal consent was obtained. Staff at some hospitals also documented the baby's individual care plan when the newborn screening blood sample was taken. However, it appears there is no mention of consent, parental discussions or information given out to parents.

A few maternal and child health nurses commented that on some occasions this information is not recorded in the baby's child health record, and they then have to follow up with the birth hospital to ascertain the baby did actually have the screening test or if the parents did not give consent to the test. A few parents were aware that information about the newborn screening test is recorded in their baby's child health record.

- **Responding to parents who decline the screening test**

A few health professionals recalled one or two parents who declined to have the newborn screening test. Most of them obtained a written non-consent from the parents; one midwife recalled a family who "also refused to sign the non-consent form". (HP1)

Another midwife stated that: "There is a culture in independent midwifery to put decision-making into parents' hands, which may or may not lead to non-consent". (HP5).

Although they stated they were highly recommending all babies have the screening test, they also stated that they needed to "honour the parents' decision" (HP5).

Some parents questioned why a written process was required for parents not consenting, but not required for parents who were giving consent for the screening test. One mother's comments were:

*I find it interesting that if someone refuses, they are more worried about them. They're more concerned about that than actually informing people who will consent. (C1)*

It is understood that documentation of non-consent is currently required as there may be grave health consequences for a child whose condition is not detected as a result of the refusal of a parent to consent to screening; hospitals are therefore keen to ensure there is written evidence of the parents' refusal in the event of a medical negligence claim.

## **6. Limitations of the current consent practice**

The consultation findings indicated numerous limitations of the current consent practice in Victoria. The comments from health professionals and consumers re-affirms the anecdotal information that informed parental consent for the newborn screening program is not adequately being obtained in Victoria on a routine basis. The consultations identified several gaps and limitations in the current consent practice including:

- No consistent approach by health professionals regarding the dissemination of (verbal or written) information to parents. Parents who do not attend antenatal classes have a greater chance of not being informed about the newborn screening program until after their baby's birth. It also appears that parents in private maternity services may not receive the same level of information about the newborn screening program antenatally as those giving birth in public maternity services.
- Inadequate levels of parent information are common. Simplification of information occurs by some health professionals to avoid parents being overloaded with information, both antenatally and post-natally.
- Many health professionals have a misconception about parents' need for information. Deliberate censorship of detailed information was adopted by several health professionals, making the assumption that parents will not want to know all the information about the newborn screening test, or believing that if parents are given too much information they will not consent to the test.
- Inconsistency in the use of written information. Old versions of the newborn screening parent information pamphlet are still used by most maternity service providers interviewed. This does not contain information on storage and access.
- Health professionals are not aware of storage, access and secondary uses of the newborn screening cards, despite their roles in giving parent information and obtaining parental informed consent.
- Parents are not routinely given information about storage, access and secondary uses of the newborn screening cards.
- Parents are not informed they can nominate on the blood card that there should be no secondary access to the cards without their explicit permission.
- Recording of details about when the blood sample has been collected is recorded in the mother's hospital records and on care plans, but few state if any discussion- has occurred (as outlined in the newborn screening guidelines).

## 7. Re-designing a model of informed parental consent

The consultations indicated strong parent support for the newborn screening program. After discussing in detail all available information about the screening, parents were asked whether they would still participate in the program. All said they would still consent to the collection of blood samples, because they were aware that the screening has significant benefit to the health and development of their babies. They did, however, want to have options regarding storage, access and secondary uses of their baby's blood samples. One mother stating:

*My first preference would be to have the test done... I'd definitely have the test done no matter what, even knowing that they are kept and used for other things. (C8)*

Parents and health professionals were also asked how the current model of verbal consent could be improved, including when consent should be obtained and how best this could be done. The findings were as follows:

- **When to obtain parental consent?**

Both health professionals and parents supported informed consent being sought post-natally, before blood samples were collected from their babies. More importantly, the consent should be given after discussion about the newborn screening has occurred. This should include information on the program, the storage of the blood sample cards, people who have access to the cards and secondary uses of the blood samples on the cards.

However, some parents were concerned that their consent should not be given too soon after a baby's birth. One parent stated that: "I would have preferred it after 72 hours, as I was not able to rationally make a decision before that" (C5).

A small number of parents felt that consent could be given prior to their baby's birth. Their views were that during the postnatal period, especially between 48 to 72 hours after birth, mothers would often not be able to take in enough information, nor ask questions before giving consent.

One PAG member commented that:

*the suggestion from health professionals to link the newborn screening consent process with the Hepatitis B and Vitamin K consent may assist in streamlining the issues and minimising any suggested administrative burden in seeking written consent for the newborn screening program, so long as the significant differences in the programs is communicated to parents.*

They expressed caution with this method stating that: "In other contexts, bundled consents have been problematic" (PAG member).

- **Written or verbal consent**

Both consumers and health professionals were asked if parental consent for the newborn screening test should be obtained verbally (as recommended in current guidelines) or through the introduction of a written and signed consent form.

Stakeholders throughout the consultations expressed strong arguments for both options. However, the clear majority of support was for the introduction of a written consent model. Table 3 provides comments from the consultations for and against both consent models. Twelve of the 15 respondents (80%) in the validation process also supported a written consent model.

There appear to be four significant reasons for the strong support for a written consent model. These include:

- The consultation highlighted to consumers the limitations of the current verbal consent model in ensuring that parents are adequately informed about the newborn screening program.
- Health professionals are concerned about possible medico-legal ramifications if parents feel they have not been adequately informed prior to giving consent for the screening test.

- A belief from consumers that if written consent was a pre-requisite, the newborn screening program would be viewed more importantly by parents, and explained more thoroughly to them by health professionals.
- Both consumers and health professionals support the idea that parents should be given options regarding the storage, access and secondary uses of their baby's newborn screening cards, and this could not be obtained through a verbal consent process.

It is important to note that a key reason for the written consent model is the complexity of the issues. Were the screening cards *not* to be stored, or for them to be stored via a separate mechanism and consent process, then the level of support for written consent for the screening test alone might be lower.

A majority of health professionals supported a written consent process similar to the way in which many hospitals implement the Hepatitis B consent model, but for the test only. They also felt that options about storage and access should involve parents and Genetic Health Services Victoria. Some midwives stated that hospitals "need to be kept out of the loop", *as* obtaining consent for all the options was seen as too burdensome on midwives (HP1).

Table 3 provides comments from the consultations regarding people's views and preferences about a written or verbal consent model for the newborn screening program.

Table 3: Comments for and against written and verbal consent

	Written consent model	Verbal consent model
<b>For</b>	<p><i>"You're told more about Hep B and vitamin K because we have to sign for it. It makes people more aware of it". "Because the midwives have to hand you the paper to be signed, they probably talk more about it, whereas in the other way (verbal consent) they don't. "If there was a tick box option midwives would have to run through it with you" (C4)</i></p> <p><i>"Writing and signing is better". "I feel more secure having signed it, like evidence to confirm the decision". "Because a lot of Vietnamese women are not very good in English and we might say, yes, yes, to everything we hear, by signing something we will look at it more seriously" (C7)</i></p> <p><i>"When you sign something, you see it as more important". "I would like to be able to have the option of it being destroyed after 2 years". "Stored indefinitely is a big thing to consent to". "We should have some control over our child's samples" (C5)</i></p> <p>People can say they didn't give consent, but if it's written down "it's there in black and white". <i>"I think up to now verbal consent has worked quite well; however, I can see that in the future there is going to be the need for written consent, the same as the Hep B" (HP3)</i></p> <p>Written consent <i>"gives you medico-legal backup". "Given the longevity of the sample, written consent is preferable" (HP7)</i></p> <p><i>"Verbal is probably adequate, but in these litigious days, a simple written consent makes sense" (HP11), supported by all three GPs (HP7)</i></p> <p><i>"The relative seriousness of the process infers that consent should be more written than verbal" (KI)</i></p>	<p><i>"Verbal consent is fine... one general consent is fine... however, parents should be told if it used for research purposes" (C2)</i></p> <p><i>"If the information is explained clearly using an interpreter, verbal consent is OK" (C6)</i></p> <p><i>"Verbal consent is adequate, if explained well and the verbal consent is recorded. Written consent in this case is not necessary" (HP8)</i></p> <p>A majority of staff in (HP4) supported a verbal consent model following the dissemination and explanation of written information. They preferred that the responsibility for gaining consent for storage, access and secondary use should be put back on Genetic Health Service Victoria (as they are responsible for collecting and storing the samples)</p> <p><i>"A verbal consent is easy to obtain but might be problematic at times, if the information has not been provided properly" (HP11)</i></p>
<b>Against</b>	<p><i>"As soon as you start introducing a consent form parents start to think and ask questions. It changes people's perceptions" (HP8)</i></p> <p><i>"Obtaining written consent antenatally is a bit messy, and would still need a post-natal follow up process" (HP8)</i></p> <p><i>"A written consent is completely unrealistic. It is NOT practical and will eventually cause the 'death of the program' " (HP11)</i></p>	<p><i>"Verbal consent cannot be proved" (C1)</i></p> <p><i>"When it's verbal you're put on the spot and you feel like you have to make a decision right now, and you go 'okay' without having time to digest it... or even have the time to read the fine print" ... With verbal consent "it feels like an assumption that you always say yes" (C2)</i></p> <p><i>"I don't like being told I have no options". "Verbal agreement is not credible". "What happens 10 years from now with my babies blood sample worries me" (C8)</i></p> <p><i>"Verbal consent doesn't cut it 20 years down the track" (HP1)</i></p>

In order to improve informed parental consent, other areas of the newborn screening program were identified as needing to be improved or introduced to the program. These included:

- **Additional information to parents**

Findings from the consultations identified the need for a new parent information pamphlet, and the provision of additional parent information about the newborn screening program on established websites (e.g., Genetic Health Services Victoria, Department of Human Services). Parents want to have access to information about **all** the conditions tested for in the newborn screening program. Parents also want information about why the blood collection needs to be done 48–72 hours after birth, and who owns their baby’s blood samples. One PAG member response was that: “The findings that parents may want different degrees of information would support a layered approach to providing information” (PAG member).

Several health professionals also stated they would like to have information about each of the conditions tested, available electronically for them to read and print off for parents if requested.

- **Overcoming the barriers for health professionals**

The consultations clearly showed a significant deficit in health professional knowledge about the newborn screening program, including the variety of conditions tested for, and storage, access and secondary uses of the screening cards. For parents to give informed consent they need an appropriate level of information on which to base their decisions. Health professionals need to be aware of the issues so they can discuss the newborn screening program and any issues relevant to the program with parents. Maternity service providers stated that: “An education program for midwives is required, about how to talk to parents about storage, access and secondary uses of the cards” (HP6).

This was supported by one of the consultant specialists who stated that: “Midwives doing the test will need education on how to give a simple message to parents” (HP11).

- **Notification of screening results**

A majority of consumers stated they would like to have some confirmation of the results sent to them. Several commented that the current practice of ‘if you don’t hear anything, it’s OK’, was not adequate.

Parents from CALD backgrounds were worried about their baby’s wellbeing, as they did not know the results of the test. One recalled: “It was a big worry, I was worried if there were going to be any complications following the test” (C7).

And another stated: “I thought something was wrong with my baby. I was so worried” (C7).

Other parents said:

*The problem is you don't get notified so that where we're not being informed whether our baby had it in the first place. You don't get a letter saying that your baby is okay. .... I just think that it's something they should think about. (C1)*

*We should be told about all conditions that are screened for, and we should get the feedback about the results of the screening. (C5)*

General practitioners (GPs) and obstetricians also stated they would like to have newborn screening results provided to them so that they could retain a copy in their patient's files. It would be particularly valuable if GPs were notified when screening results were positive for any of the conditions, or a repeat test was required (HP7).

- **Options for parents**

Many consumers indicated they would like to have choices in making decisions about the screening and what happens to the blood samples after the screening. The consultation results clearly indicate that consumers understand the importance and benefit of the newborn screening program for their baby's health and wellbeing, and that they strongly support the program. However, they were very concerned about the storage of blood sample cards, the people who have access to the cards and how the card might be used:

*I think if there's a way for them to be separated it should be.... It's not okay to agree for one thing, but then add on all these other things, saying well if you agree to it for the health of your child, but then it comes with these attachments of things like research and secondary people that can access them. ... It's not an even playing field there. You're consenting to the screening but there're numerous things you are consenting to there. So it really should be broken down. (C1)*

*I think to be given the choice, and to be given the information that we have the choice would be good. And if I was given that opportunity then I might give my consent to one but not to the other. (C2)*

Two health professionals also made comments that there needed to be some way of identifying to researchers if the baby dies soon after birth. This will ensure researchers do not approach grieving parents unless they have given permission to have their baby's card used for research. It will also enable researchers to be made aware of the baby's death prior to contacting parents (HP2, HP6).

## DEVELOPMENT OF ISSUES PAPER AND RECOMMENDED MODELS

Following the analysis of the findings, three options for informed consent practice were developed. These were an enhanced verbal consent model; a written consent model; and a combined verbal and written consent model. However, major features recommended in all model options were:

- Separation of screening, from the storage, access and secondary use issues with a two-staged consent process.
- Expressed consent for the screening alone, to be sought post-natally – prior to the blood sample being taken.
- Results of the screening to be sent to all parents.
- At notification of results, parents should be given information and options about the storage, access and secondary uses of blood samples, and asked to give informed consent for these uses to GHSV.
- Better mechanisms to ensure provision of high quality consumer-friendly information for parents.

The three options were presented to the PAG members. Extensive discussion and comments were made on each option. It was decided that only the enhanced verbal consent option would be used at this stage for seeking feedback from key health professionals and consultants.

An Issues Paper was then developed, outlining the key findings of the consultations and identifying factors that impede as well as facilitate the parental informed consent process. The recommended enhanced verbal consent model for informed consent with the principles supporting its development was presented for comments. The Issues Paper also included summary of documents and literature related to the newborn screening program.

The Issues Paper is available at [www.healthissuescentre.org.au](http://www.healthissuescentre.org.au)

## VALIDATION PROCESS

Twenty-five copies of the Issues Paper were sent by post or e-mail to key participants, including 16 consumers and nine health professionals. They were asked to assess the recommended consent model and respond to the questions provided. In addition, responses from members of the PAG were sought.

The response template used to obtain feedback had three open-ended questions about: strength and weakness of the recommended model with suggested criteria for assessment; opinions on splitting the consent process into two steps — consent for the screening and options for the storage, access and secondary use of blood samples; and preference for verbal or written consent to the screening (Appendix D).

### Results

Fifteen out of 25 key participants, including eight consumers and seven health professionals, responded to the request. Three key consumers incorporated views of their partners into the comment. At least one key health professional discussed the recommended model in a meeting with their peers and hence responded as collective views. Four Project Advisory Group members also provided comments on the model.

The validation process was effective in highlighting the strengths and weaknesses of the new recommended model and supporting the findings from the initial consultations. The findings outlined below have been used to inform the final recommended models of consent and implementation strategies.

- **Strengths and weaknesses of the recommended model**

All key consumers and health professionals supported the recommended consent model. They agree that the consent model ensures that parents are fully informed about the newborn screening and gives parents choices to consent or not consent to the screening as well as to the storage, access and secondary use of the blood samples.

The key consumers and health professionals agree that the recommended model supports the primary purpose of the screening, which is to identify individuals at high risk of a condition so they can seek appropriate treatment or advice. One PAG member argues that the recommended consent model would not serve this purpose if the blood sample cards were kept for longer than the two-year quality assurance period.

The key consumers and key health professionals think that the strategies are practical for health professionals. Some indicate further education and training is required, and that a consistent process across both the public and private health systems is needed:

*Unfortunately a checklist will mean more work for the midwife taking blood samples from babies, but they are best to do the checklist, as they are the ones doing the hands-on. (Key health professional)*

One PAG member identified that under this model of consent, health professional training does not necessarily need to be extensive:

*Only being responsible for obtaining consent for the screening test will mean that health professionals will not need to acquire detailed knowledge about the storage, access and secondary use issues. They will only need to flag the issues with the parents and inform them that they will be sent detailed information about these issues with the test result notification. (PAG member)*

Most respondents think that the model reduces parents' concerns about privacy issues. Two key consumers and a key health professional are unsure, stating that privacy will remain an important issue between parents and GHSV. Another key health professional argues that informing parents is more important:

*Privacy is an issue but the most important issue is that the parents are informed correctly about the NS Test and all staff that are involved with their care know the test has been undertaken or declined. (Key health professional)*

One PAG member commented that:

*It is reassuring that the researchers found that given the extent of information discussed, parents would still consent to the newborn screening... this suggests that more information about testing and future uses etc. would not act as a barrier to testing. (PAG member)*

- **Support for a two-staged consent model**

Almost all key consumers, key health professionals and PAG members supported the model's strategy for a two-staged consent process. Comments included:

*You don't want people's decision about consenting to the test to be overshadowed by a fear of what will happen to the sample afterwards. Having two consents will alleviate this. (Key consumer)*

*We like the idea of the split model as it ensures that DNA research projects do not have a negative impact on the primary purpose of the newborn screening being identification of diseases. (Key consumer)*

*The fact that people will be more informed about the storage, access and secondary use will reduce confusion and the consent form will allow people to make an informed decision. (Key consumer)*

*I think that split consent is a great way to prevent parents' confusion. I like the idea of sending results to parents. (Key health professionals)*

*It ensures parents will not be inundated with all the information at the one time. Parents only have to be given as much information as necessary to enable them to make an informed choice about that particular stage of the screening program. (PAG member)*

*This is a good solution, as it is not feasible for health practitioners to give accurate information to patients about this – as such they should not be seeking consent to storage. (PAG member)*

One PAG member disagreed with the two-staged consent model, believing that the extra consent for storage of cards, although required at a later stage, would take the focus away from the primary purpose of the screening. Another stated that:

*seeking consent for the collection of blood should not be separated from the process of seeking consent for later storage/access/use... consent to testing must be informed by what will be done with the cards after they are created. (PAG member)*

A key consumer and a key health professional who did not support the two-staged consent model simply stated it was not necessary and that one consent would be easier to manage.

- **Verbal or written consent**

Consultation results showed that most health professionals and consumers preferred written to verbal consent, and in the context of the consultation, their consent would apply to both the screening and what happened to blood samples after the screening. As the model splits the consent in two steps and recommends enhanced and express verbal consent to screening, it is appropriate to ask whether the key participants still support this option.

It was found that all eight key consumers, six key health professionals and two PAG members strongly support obtaining consent for the taking blood samples in writing. Their reasons are:

*They're accountable professionals. If it's written, it's clear. People's memories are poor after having a baby generally. It's a record. (Key consumer)*

*I think that in writing is the most fail-safe way of gaining consent. It makes people read what they are signing, or in ESL (English as a second language) area, it will have to be verbally explained. (Key consumer)*

*If parents do not need to sign, I would not trust that all information is provided. (Key consumer)*

*Parental consent should be in writing – that is the only legal standard that would pass the test of proof particularly if there was any challenge by the parents at a later date. (Key health professional)*

*I support written consent because verbal consent could be mistakenly given. (Key health professional)*

*Individuals whose blood it is, need to be able to see what was consented to on their behalf years ago, and perhaps be able to amend the scope of that consent or possibly revoke it. (PAG member)*

One key health professional and two PAG members stress the importance of parents having information and choices rather than the mechanism of giving consent:

*Written does not necessarily mean that the consent is informed one; however, it's the signature that does suggest people have been given options. (Key health professional)*

*Verbally or in writing as long as there is a formal discussion with parents. (PAG member)*

*Verbal consent is OK as long as it is explicit and adequate information is provided before hand. (PAG member)*

- **Additional comments**

The additional comments received indicate the concerns that the key participants felt needed to be addressed. These include parents' frustration at not being informed about the current system of storing blood samples, a PAG member's concern regarding complexity of notifying results to parents, and a suggestion for piloting the recommended consent model. Another key consumer would like to be assured that changes to the current practice would take place and a key health professional suggests a strategy for public education campaign on the newborn screening. Comments included:

*What has been occurring seems improper and dishonest. People give their baby's (blood) samples for the good of their baby, not for other uses. I believe the samples that have been taken and that are only being kept for secondary purposes should now be returned. (Key consumer)*

*Sending out test results is problematic; there is no such thing as a "normal" screening result. Parents would have to be informed that their child was at low risk of all the conditions screened for, as this is a screening program, not diagnostic. This may be a confusing message without appropriate education. (PAG member)*

*If a new system is proposed that is quite different to the existing system, it must be trailed and evaluated (on a small scale) before implementation. (PAG member)*

*What follow-up system will be in place to ensure that this process is followed? If it doesn't happen, no one will be any wiser. (Key consumer)*

*Literature informing parents about the NS test should be produced in English as well as other CALD community languages and be made available to Maternity Services, Maternal & Child Health Services, medical practitioners for display in their offices/rooms. (Key health professional)*

Finally, a key health professional who sent in collective responses suggests the default option in the consent model be the present model; for example, ongoing storage and access:

*Split consent model is a good idea but I think parents (should be) required to decline the storage, access and secondary use... the default option should be the present model; for example, ongoing storage and access. (Key health professional)*

## FACTORS THAT IMPEDE INFORMED CONSENT

Analysis of the community consultations identified the following key factors that impede or hinder the current newborn screening consent practices from obtaining consistent informed parental consent. They include:

- Differing levels of knowledge by health professionals regarding the newborn screening program, particularly about storage, access and secondary use issues.
- Confusion regarding health professional responsibility, given the diversity of maternity service providers (particularly in the private health sector).
- Varying protocols in practice for the dissemination of the newborn screening parent information pamphlet across maternity services.
- Differing consent models in practice for other screening and immunisation programs (e.g., Victorian Infant Hearing Screening Program, Hepatitis B Immunisation Program, Vitamin K administration).
- Time constraint issues for health professionals providing maternity care that reduces staff's ability to adequately inform and discuss the newborn screening program with parents.
- Inappropriate timing of information to parents about the newborn screening program, and obtaining of consent.
- Attitudinal differences between health professionals about the level of information required to adequately inform parents about the newborn screening program.
- Strong concerns from parents that they are not adequately informed about storage, access and secondary uses of the newborn screening cards.

## DISCUSSION

The consultations have been very effective in identifying gaps and limitations in the current Victorian Newborn Screening Program, and in recognising a range of key factors that impede current practices in obtaining informed parental consent.

The findings support previous anecdotal information that parents were not being adequately informed about the screening program and associated storage, access and secondary use issues, nor being appropriately consented.

While the consultations from both consumers and health professionals showed overwhelming support for a written model of consent, there were mixed responses from PAG members, with consensus unable to be reached.

Support for the written consent model was based on views that it would:

- Constitute clear evidence that informed consent had in fact been obtained and on what basis.
- Provide an opportunity for discussion to occur where parents could ask questions or have any concerns addressed.
- Allow parents to express their preferences about future uses of the newborn screening cards (such as for research) separately from consent for screening.
- Allow individuals whose blood it is to determine (upon reaching maturity) whether consent was in fact given, and what the scope of the consent was, thereby facilitating their right to make decisions about the ongoing use of their blood sample.
- Improve parents' recall of information.
- Assist with compliance of Privacy Laws; for example, the obligation to take reasonable steps to ensure that individuals are aware of a range of matters, including their right of access, and any usual disclosure of information to other individuals or organisations.

Opposition to written consent within the PAG was based on its potential to create concern and a barrier for parents, increasing the risk of non-consent to screening (and hence potential harm to babies not screened). Although many health professionals believed that written consent would give them medico-legal cover, opposing views to a written consent model consider a signed form is not sound evidence that parents were actually fully informed before signing a consent form.

Irrespective of potential conflicting viewpoints around the written versus verbal consent models, many potential improvements to the current Newborn Screening Program have been identified through the consultations, and further developed from discussions with PAG members.

The analysis also highlighted the fact that the collection of newborn screening cards is **not** a required part of the Newborn Screening Program. It has evolved as a consequence of the screening over the past 30 years, for which few parents would have been informed about or been asked to consent to. This is a highly unsatisfactory situation and raises a host of questions, many of which are being

looked at in the broader Review of the Screening Program in Victoria. Not only is the existence of this large collection of blood samples problematic in its own right, more importantly it risks reducing the scope of effectiveness of the Newborn Screening Program. This is a serious issue and has led to Health Issues Centre to the recommendation that the screening program and practices associated with storage be separated.

## RECOMMENDED MODELS OF INFORMED PARENTAL CONSENT

Based on the community consultation findings, the results of the validation process, and the advice of the PAG members, Health Issues Centre presents two final models of informed consent, with its definitive preference for the written consent option.

### Key principles for both models

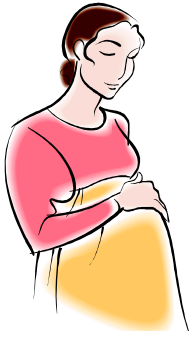
1. Parents are entitled to choose whether their baby participates in the newborn screening program.
2. Parents should be provided with high quality consumer-friendly written information about the Newborn Screening Program to inform their choice. They should also be given enough time to make an informed choice.
3. Parents should be informed about the conditions screened for, as well as the storage, access and secondary uses of the blood samples.
4. Health professionals should discuss the Newborn Screening Program with parents and gain expressed parental (not implied) consent prior to taking the baby's blood sample.
5. Parents have a right to information about their baby's screening results.
6. The consent process is developed taking into account possible unintended consequences of harm or risk of psychosocial distress arising from the screening program.

The models outlined below recommend the separation of consent for the screening test from the decisions concerning longer-term storage, access and secondary use. Whilst a few stakeholders regarded the two-staged consent process as unnecessary (HP7), the majority supported the concept and believed it would reduce confusion about, and possible barriers to, screening (the primary purpose of the screening program).

The consultations identified large majority support from both health professionals and consumers for a written consent model. Health Issues Centre also strongly supports this model on the basis that it formalises the choice being given to consumers. It clearly documents consumers' decisions, and supports research findings that written consent facilitates better quality decision-making by consumers. However, given that consensus could not be reached by the PAG members on one recommended method of obtaining consent, both verbal and written consent model options are presented in this report, with a recommendation that they be trialled.

### The proposed models aim to:

- Reduce some possible barriers to screening
- Reduce the burden of information and anxiety for parents
- Enable parents to have adequate time to read, discuss and choose preferences for screening storage access and secondary uses
- Notify parents of their baby's newborn screening test results
- Improve compliance with the Privacy and Health Records Acts.



# Newborn Screening

## Enhanced Informed Consent Model

(Written Consent Option)

### Parents Information

- The GHSV newborn screening parent information pamphlet should be given to all parents **antenatally** (preferably in early third trimester).
- The pamphlet should also be made available to parents after the baby's birth.
- The pamphlet should explain that the cards are provided to GHSV who conduct the test, and who may re-use the cards in the first two years for quality assurance purposes. Summary information about storage, access and secondary use and parent options should also be included in the pamphlet.
- Health professionals should discuss the information summarised in parent information pamphlet with parents.
- A newborn screening parent information checklist should be completed by a health professional before the blood sample is taken.
- Parents will be asked to provide **written consent** to the newborn screening test 48 – 72 hours after their baby's birth.
- Parents should also be made aware that they will be sent information and provided with options for storage, access and secondary uses of their baby's newborn screening card with the test result notification (approx. 8 – 10 weeks after their baby's birth).

### Obtaining Informed Parental Consent

- Health Professionals should inform parents that they have an option of participating in the newborn screening program (The test is offered to all parents, and is highly recommended for the wellbeing of their baby).
- Health Professionals need to discuss the newborn screening parent information pamphlet with parents prior to obtaining their written consent, and before taking the blood sample.
- Parents should be informed about storage, access and secondary uses of their baby's newborn screening card, and be advised that they will be sent information, and their preferences will be sought by Genetic Health Services Victoria when they receive the test result notification by mail (8–10 weeks after birth).

### Test Results

- All parents are notified by mail of their baby's newborn screening results (if normal), or by telephone (if re-testing required).
- With the test results notification, parents are also given the options regarding the ongoing storage, access and secondary uses of their baby's newborn screening card.

### Storage, Access & Secondary Uses

- All samples and information are securely stored in accordance with privacy laws and public records legislation and access to the card is restricted according to agreed protocols.
- A minimum period of two years of storage at Genetic Health Services Victoria is recommended for quality assurance purposes. After this time the cards are stored indefinitely in Victoria, in a secure off-site facility.
- Sometimes the cards may be used for approved research, once the identifying information has been removed. Testing on identified samples is only performed with the permission of the parents or otherwise authorised by law.
- Parents will be informed (with the test result notification) that after the two-year quality assurance period they can consent to having their baby's card:
  - a) Destroyed \*
  - b) Custody of the cards transferred to them \*, or
  - c) Stored by GHSV with no secondary access to the cards without their explicit permission, or
  - d) Stored by GHSV and made accessible for other diagnostic, research or forensic identification purposes (without additional parents notification) or as otherwise authorised by law

\* The options of the card being destroyed or custody of the card transferred to parents will be dependent on the relevant privacy and health records acts.

- If parents do not notify GHSV of their preference regarding storage and access, a default option should apply.

### Documentation

- The completed newborn screening parent information checklist (which could be attached to the screening card) should be detached once completed and kept in the mother's file confirming when:
  - a) The newborn screening parent information pamphlet was given to the parents
  - b) Key issues were discussed with the parents about the newborn screening program
  - c) When **written consent** was obtained and who consented to the test, and when
  - d) The newborn screening sample was collected.
- Details of when the newborn screening sample was taken should also be recorded in the baby's child health record book.
- The signed written consent form will be kept in the mother's file.
- Parents who choose not to participate in the newborn screening program will be asked to sign a non-consent statement, which will be included on the consent form. This should be filed in the mother's hospital file.

# Newborn Screening

## Enhanced Informed Consent Model

(Verbal Consent Option)



### Parents Information

- The GHSV newborn screening parent information pamphlet should be given to all parents **antenatally** (preferably in early third trimester).
- The pamphlet should also be made available to parents after the baby's birth.
- The pamphlet should explain that the cards are provided to GHSV who conduct the test, and who may reuse the cards in the first two years for quality assurance purposes. Summary information about storage, access and secondary use and parent options should also be included in the pamphlet.
- Health professionals should discuss the information summarised in parent's information pamphlet with parents.
- Parents will be asked to provide express consent (**informed verbal consent**) to the newborn screening test 48 – 72 hrs after their baby's birth.
- Parents should also be made aware that they will be sent information and provided with options for storage, access and secondary uses of their baby's newborn screening card with the test result notification (approx. 8 - 10 weeks after their baby's birth).

### Obtaining Informed Parental Consent

- Health Professionals should inform parents that they have an option of participating in the newborn screening program (The test is offered to all parents, and is highly recommended for the wellbeing of their baby).
- Health Professionals need to discuss the newborn screening parent's information pamphlet with parents prior to obtaining their express verbal consent, and before taking the blood sample.
- Parents should be informed about storage, access and secondary uses of their baby's newborn screening card, and be explained that they will be sent information and their preferences sought by Genetic Health Services Victoria when they receive the test result notification by mail (8-10 weeks after birth).

### Test Results

- All parents are notified by mail of their baby's newborn screening results (if normal), or by telephone (if retesting required).
- With the test results notification, parents are also given the options regarding the ongoing storage, access and secondary uses of their baby's newborn screening card.

### Storage, Access & Secondary Uses

- All samples and information are securely stored in accordance with privacy laws and public records legislation and access to the card is restricted according to agreed protocols.
- A minimum period of two years of storage at Genetic Health Services Victoria is recommended for quality assurance purposes. After this time the cards are stored indefinitely in Victoria, in a secure off-site facility.
- Sometimes the cards may be used for approved research, once the identifying information has been removed. Testing on identified samples is only performed with the permission of the parents or otherwise authorised by law.
- Parents will be informed (with the test result notification) that after the two-year quality assurance period they can consent to having their baby's card:
  - a) Destroyed \*
  - b) Custody of the card transferred to them \*, or
  - c) Stored by GHSV with no secondary access to the cards without their explicit permission, or
  - d) Stored by GHSV and made accessible for other diagnostic, research or forensic identification purposes (without additional parents notification) or as otherwise authorised by law.

\* The options of the card being destroyed or custody of the card transferred to parents will be dependent on the relevant privacy and health records acts.

- If parents do not notify GHSV of their preference regarding storage and access, a default option should apply.

### Documentation

- The completed newborn screening parents information checklist (which could be attached to the screening card) should be detached once completed and kept in the mother's file confirming when:
  - e) The newborn screening parent information pamphlet was given to the parents
  - f) Key issues were discussed with the parents about the newborn screening program
  - g) When express consent (**verbal**) was obtained and who consented to the test, and when
  - h) The newborn screening sample was collected.
- Details of when the newborn screening sample was taken should also be recorded in the baby's child health record book.
- Parents who choose not to participate in the newborn screening program will be asked to sign a non-consent statement, which should be filed in the mother's hospital file.

## RECOMMENDATIONS

The following recommendations have been developed to introduce a new and more effective informed parental consent model for the Victorian newborn screening program. The 14 recommendations and rationale behind them are provided below.

### Recommendation 1 (medium-term)

*That the Department of Human Services instigates changes to separate the operation of the newborn screening program (including its quality assurance processes) from any subsequent storage, access and secondary uses for research and identification purposes.*

#### Rationale

- The newborn screening program, and the later storage and access of the cards for population health and identification purposes, are quite different endeavours, linked currently by an artefact of Victorian legislation and administrative arrangements. A newborn screening program does not need a subsequent blood spot database, and a population health database does not need a newborn screening program as a pre-requisite for its existence. Separating the two programs will eliminate the complexities for parents and health professionals caused by the current intertwining. It will remove much of the difficulties in designing and implementing a satisfactory and reliable informed consent process for the all-important screening program. It will also remove a potential barrier to full uptake of the program by parents.

### Recommendation 2 (medium-term)

*That, given a separation of the programs, the consent process for the screening test be sought as per current arrangements (with some modifications to make it more effective), whilst information and options about future potential uses will be sought at a later date.*

#### Rationale

- This separation will therefore create two programs focused on quite different purposes and administered separately. Each program could focus on its primary purpose. Each program would need to seek informed consent but they would be separated in purpose, in time, and perhaps by the requesting organisation.
- Seeking consent for the screening test would become considerably more straightforward.
- This model will enable a more layered approach to parent information about the newborn screening program and associated issues of storage, access and secondary uses.
- This was supported by most of the PAG members, who saw it as a new way of clearly separating the screening test from the controversial issues surrounding the storage, access and secondary use of the screening cards.

### Recommendation 3a

*That informed **written consent** for the newborn screening test be obtained post-natally before the blood sample is collected.*

**OR**

### Recommendation 3b

*That informed **verbal consent** for the newborn screening test be obtained post-natally before the blood sample is collected.*

### Rationale

- The consultations identified a large majority of support from both health professional and consumer for a written consent model (2a). However, the Newborn Screening Project Advisory Group members could not reach consensus on the recommended method of obtaining consent, and therefore both options are outlined above. Piloting of both models was therefore recommended (see below).

### Recommendation 4 (medium-term)

*That a new statewide parent information pamphlet be developed and disseminated to services and hence parents. This should be provided by the relevant health professional to all parents antenatally (early in the third trimester, and again post-natally before the blood sample is collected). This should be available in multiple formats (e.g., pamphlet, websites, audio-visual) and multiple languages.*

### Rationale

- The existing newborn screening program parent information pamphlet needs to be reviewed and re-designed, to incorporate the feedback from the consultations. These include providing links to further information about all the genetic conditions tested and where additional information about storage, access and secondary uses can be obtained. The pamphlet should also make reference to how parents will be asked to record their preferences about storage, access and secondary uses of their baby's blood sample, and inform parents that screening results are not always 100% accurate.
- A dissemination strategy for the new parents information pamphlets is required to ensure that they are distributed to all maternity providers, and old versions are recalled.
- This was supported by all PAG members.

### **Recommendation 5 (medium-term)**

*That a checklist for health professionals be developed which provides confirmation and standardised documentation of:*

- a) When the parent information pamphlet was given to parents*
- b) That a discussion has taken place with the parent about the newborn screening program*
- c) Whether informed consent was obtained, when, and who consented to the test*
- d) When the newborn screening sample was collected.*

### **Rationale**

- The checklist will provide health professionals with a guide to what information should be provided to parents about the newborn screening program and what needs to be recorded.
- The checklist will be attached to the blood spot card, and have a perforated edge so that it can be completed and filed post-natally with the mother's history.
- The checklist should have a provision for the health professional taking the blood sample to sign it to acknowledge that all the recommended practices have been offered to parents before they consented to the newborn screening test being performed.
- This had mixed responses from PAG members. Some concern was expressed that health professionals in maternity services may not welcome this, given that it could be seen as another form of unnecessary documentation with which they have to comply. Others supported the checklist as a tool for assisting health professionals to provide consistent information to parents about the newborn screening program.

### **Recommendation 6 (medium-term)**

*That a newborn screening test result notification system be established to provide written feedback to all parents about their baby's screening results, and to provide options to parents regarding storage, access and secondary uses of their baby's blood samples.*

### **Rationale**

- To inform parents about their baby's screening results, a pamphlet (similar to the Victorian Infant Hearing Screening Program) or letter should be sent by Genetic Health Services Victoria to all parents (approximately eight to 10 weeks after birth) detailing the:
  - ◆ Conditions screened for
  - ◆ Baby's screening test results,
  - ◆ Information about options for parents regarding storage, access and secondary use of their baby's blood sample; and reply options.
- While most PAG members supported the introduction of this practice, a few felt that it was an unnecessary and expensive process to establish and maintain.

### **Recommendation 7 (medium-term)**

*That the newborn screening blood spot card be re-designed.*

#### **Rationale**

- Postal addresses will need to be recorded on the newborn screening cards so that test results can be mailed to parents with the additional information about their options regarding storage, access and secondary use of their baby's blood samples.
- The blood spot card will need to be re-designed to incorporate the health professional checklist to the blood spot cards, with a perforated edge for subsequent separation and storage.

### **Recommendation 8 (short- and medium-term)**

*That guidelines be developed for health professionals about discussing the newborn screening program with parents.*

#### **Rationale**

- The development of guidelines for health professionals (similar to the United Kingdom) will help guide health professionals about what constitutes an appropriate level of information about the conditions screened for by the current newborn screening program, and how best to explain what options parents have regarding storage, access and secondary use of their baby's blood samples. This needs to occur in the short-term to cover the current situation, and be revised if and when the programs are separated as above.

### **Recommendation 9 (short- and medium-term)**

*That a statewide education program be conducted targeting all maternity service providers. This should include hospitals, independent and community midwives, obstetric medical practitioners and maternal and child health nurses.*

#### **Rationale**

- The consultations showed that health professionals are not well informed about storage, access and secondary uses of the newborn screening cards, or the number of conditions now screened. There is a need for this to happen in the short-term.
- Health professionals will also need to be updated about any new consent model introduced, including use of the checklist and the guidelines about how to discuss newborn screening with parents in order to obtain informed consent.
- All PAG members supported the need for an educational update for maternity services providers.

#### **Recommendation 10 (short-term)**

*That information be made publicly available on the Genetic Health Services Victoria and the Department of Human Services websites, listing and providing a brief outline about **all** the conditions tested for in the current Victorian Newborn Screening Program.*

#### **Rationale**

- The consultations identified that both parents and health professionals wanted access to additional information through credible websites about all the conditions tested for in the newborn screening program.
- Additional information about storage, access and secondary uses of blood samples could also be available on-line for those parents or health professionals who would like more detailed information than that outlined on the newborn screening parent information pamphlets.

#### **Recommendation 11 (short-term)**

*That both a written and verbal consent model for the newborn screening program be piloted and evaluated in different-sized maternity services across Victoria, to obtain consumer, health professional and key stakeholder feedback, and assess impact on screening rates, before any statewide implementation is considered.*

#### **Rationale**

- Piloting of the proposed consent model with both the written and verbal consent options will help identify which model is practicable, workable and has least impact on the parental consent to the newborn screening test.
- Piloting of the proposed model will also enable modifications to be made before a final model is implemented statewide.
- PAG members were supportive of piloting and evaluating any new models of consent (both written and verbal options) prior to any statewide implementation.

#### **Recommendation 12 (short-term)**

*That a small quantitative study be undertaken to determine:*

- a) The uptake of newborn screening per birth in Victoria (with individual hospital sub-analysis)*
- b) The number of notified non-consents per year, and the number and consequence of subsequent genetic counselling advice*
- c) The number of transfer of custody requests per year, with outcome data.*

#### **Rationale**

- This level of information will be required to form baseline data, prior to the piloting of any new consent model.

### **Recommendation 13 (medium-term)**

*That a public awareness campaign be considered, targeting parents and health professionals about all public health programs recommended for newborns. This should incorporate the latest information about the newborn screening program, Hepatitis B vaccination, Vitamin K administration and Victorian infant hearing program.*

#### **Rationale**

- Parents are confused about the standard investigations, tests and procedures that are performed on their babies within hours and days after their birth. Consistent and clear information about what and why these procedures are recommended is required.

### **Recommendation 14 (short-term)**

*That a decision about future storage and access to the collection of newborn screening blood spots be made as soon as possible.*

#### **Rationale**

- If storage of the newborn screening blood samples is recommended by the Australian Health Ministers Advisory Committee (AHMAC) then guidelines and public awareness regarding storage and access to the blood samples needs to be improved.
- Based on the AHMAC recommendations, the Victorian Government will need to decide what to do with the existing including stored newborn screening cards (e.g., some form of notification process to parents, with destruction, transfer of custody, or ongoing storage options).
- If the proposed consent model is accepted and parents are given options regarding storage, access and secondary use of their baby's newborn screening cards, a default option for parents who do not reply to the notification process will also need to be determined based on the relevant Privacy and Health Records Acts.

### **Recommendation 15 (short- and medium-term)**

*That there be better consumer representation in the management, monitoring and future direction of the Victorian newborn screening program.*

#### **Rationale**

- The involvement of parents more directly in the management, monitoring and future direction of the screening program, including in the implementation of these recommendations for an improved informed consent model, may improve the transparency of the program to parents and encourage a high participation in the newborn screening program.

- A process to involve parents in the development and implementation of the new parent information pamphlet (recommendation 4), and the test result notification system (recommendation 6) should be endorsed to ensure the information is written in an appropriate, consumer-friendly style and language.
- Health Issues Centre recommends that ongoing monitoring of the newborn screening program be considered, incorporating consumer feedback, to ensure informed parental consent is maintained.



## APPENDIX A

### PROJECT ADVISORY GROUP - MEMBERSHIP

#### *Screening In Victoria Report On Informed Consent For Newborn*

Dr Rosemary Lester  
(initially Dr Christine Selvey) and  
Michael Batchelor  
Dianne Scott  
Wendy Dawson +/-or Joan O'Neil

Beth Wilson +/-or Jesinder Bhullar  
Paul Chadwick +/-or Michelle Fisher  
Dr Della Forster  
Associate Professor Martin Delatycki  
MaryAnne Aitken  
Leslie Arnott  
Kelley Stewart  
Tony McBride, Kim Hider and  
Charin Naksook

Department of Human Services  
 Preventions and Perinatal Health, Public Health  
 Strategy and Performance Reporting, Metropolitan Health and Aged Care  
 Maternity Services Programs, Metropolitan Health and Aged Care

Office of the Victorian Health Service Commissioner  
Office of the Victorian Privacy Commissioner  
Research Midwife (Mercy Hospital)  
Genetic Health Services Victoria  
Murdoch Childrens Research Institute  
Consumer representatives

Health Issues Centre

### Project Advisory Group - TERMS OF REFERENCE

#### 1. Background/Context

A Project Advisory Group will be established to provide direction and sector knowledge to Health Issues Centre, who is responsible for compiling a report on informed consent for newborn screening in Victoria.

The overall project objectives are to explore the factors that facilitate informed consent about newborn screening and related issues such as secondary uses of the card, and to determine those factors that obstruct/hinder/impede informed parental consent.

Terms of Reference for the Project Advisory Group have been developed by the project staff, and presented at the first Project Advisory Group meeting for endorsement.

#### 2. Role of the Project Advisory Group

The Role of the Project Advisory Group is to:

- Offer relevant knowledge and background information to project staff, to assist them to undertake the project;
- Provide advice to the project team on implementation and recruitment strategies;
- Make suggestions for effective and comprehensive stakeholder consultation;
- Provide comment on draft interview questions for stakeholder interviews and focus groups;
- Identify and raise any relevant or emerging issues that may have implication for the project;
- Advise on a recommended model of consent for newborn screening; and to
- Provide feedback on the draft final report.

#### 3. Membership

The Project Advisory Group will comprise representatives from the Department of Human Services, consumer advocacy groups, relevant professional bodies (midwifery and obstetrics), Office of the Victorian Privacy Commissioner, Office of Health Services Commissioner, and Genetic Health Services Victoria.

#### **4. Convener/Chair**

Dr Christine Selvey, A/Manager, Prevention and Perinatal Health, Department of Human Services will chair the Project Advisory Group. If the designated chair is not available, then Michael Batchelor, Manager, Genetics and Perinatal Programs will be responsible for convening that meeting.

#### **5. Support**

Health Issues Centre will provide executive support for the advisory group, including establishing meeting dates, setting agendas, taking and distribution of minutes and liaison with advisory group members.

#### **6. Frequency of Meetings**

The project will be conducted over eight to ten months, and it is anticipated that the Project Advisory Group will meet four or five times at key stages during the project. The initial meeting will be held on Friday 27<sup>th</sup> May 2005 (12:30 to 2.00 pm), with subsequent meeting dates to be determined at this meeting.



## APPENDIX B

### OVERVIEW OF DOCUMENTS RELATED TO NEWBORN SCREENING & INFORMED CONSENT

(For Project Advisory Group Members)

*The documents fall into six broad categories:*

- (a) Media reports related to newborn screening issues (4 items)
- (b) Medico-legal issues (7 items)
- (c) Guidelines and policies for newborn screening (3 countries)
- (d) Verbal vs. written consent (3 items)
- (e) Health research related to communication issues around newborn screening (4 items)
- (f) Information for parents about newborn screening (15 sources including 5 from USA, one UK, and 9 Australia)

#### **MEDIA REPORTS RELATED TO NEWBORN SCREENING ISSUES**

All media items relate to Genetic Health Services Victoria (GHSV) and the ownership of infant blood samples. The initial news item published in *The Age* in July 2004 raised concerns about the retention of the blood samples by a private not-for-profit company (Noble 2004). This followed the report by the Australian Law Commission (ALRC) on protecting human genetic information in March 2003. The news article contained a brief outline of the history of newborn screening in Victoria and the role of GHSV. In response, a press release from the Murdoch Children's Institute issued on the same day, aimed to reassure parents that State and federal guidelines cover the storage of screening cards, recasting the primary issue as the regulation of these samples not their ownership (Anon 2004). The GHSV are described as the custodians of the screening cards. A few weeks later the online newsletter of Monash University published another defence of the GHSV in regards to the privacy and security of its records following legal concerns about ownership and protection of the blood spot raised by law academics after the previous media coverage (Anns 2004). A further article on the topic, written by the same journalist appeared in *The Age* in July 2005, again raising the issues of ownership and use of the samples in research or by the police to confirm identity. Particular concerns arise from the potential use of newborn screening samples for commercial purposes such as confirming paternity (Noble 2005).

#### **MEDICO-LEGAL ISSUES**

Chapter 21 in the Australian Law Reform Commission (ALRC) report on the Protection of Human Genetic Information in Australia addresses population genetic screening and discusses privacy, consent to testing and the circumstances in place when consent is obtained, provision of counselling, costs to the health system, reliability of results, implications for insurance, the 'right not to know' (particularly for incurable conditions), and the use of genetic samples and information for research. The discussion paper points out that currently it is only the information obtained from genetic screening is subject to legislation and the retention of samples in effect creates a genetic database raising privacy concerns. The chapter concludes that there is a need for consistency in newborn screening policies and practices nationally and that testing must be reliable with an acceptable level of sensitivity (ALRC 2003).

An overview of the ALRC's discussion paper published in the newsletter of the Victorian Privacy Commissioner discusses the collection and storage of DNA in newborn screening samples highlighting concerns about the quality of the consent that parents give for their storage and secondary use (Anon 2003).

Several Information Privacy Principles appear to be relevant to the issues around newborn screening. These include: the manner and purpose of collection of personal information for a lawful purpose; the storage and security of personal information including protection against unauthorized access; entitlement of access to records containing personal information; limits on use of personal information including not using personal information for other purposes unless consent has been given, although this particular principle asserts that personal information can be used for another purpose if it is 'directly related to the purpose for which the information was obtained' (OFPC 1988).

Skene (2004) adds information about the variation in the practice of retaining newborn screening cards across Australia. Samples are kept for two years in Western Australia, 18 years in New South Wales, 25 years in Queensland, and indefinitely in South Australia and Victoria. Skene *et al* conclude that clarification is needed about the legal ownership of the cards and whether the transfer of newborn screening samples to parents is contrary to the requirement that cards be securely retained. Editorial comment to Skene's article raises doubt that current procedures for gaining parental consent would meet the test of law for informed consent and also points out that parents do not have absolute rights over their child and that decisions must be made in the child's best interests.

The test for informed consent is summarised by McPhee (2002) in a legal perspective of perceptions of risk in Australia. McPhee refers to the concept of material risk and the significance that a reasonable person would attach to it which was raised in the 1992 Rogers vs. Whitaker decision and affirmed in a 2001 judgement and cites Justice Kirby's observation that the burden of risk is ultimately borne by the patient.

An alternate view to the need for informed consent prior to newborn screening is offered in a 2004 Canadian article, which outlines international practices around screening sample storage and consent. Laberge *et al* (2004) agree that the paramount concern for all policies around newborn screening and dried blood spot storage should be the best interest of the newborn. However, unlike the situation in Australia and the United Kingdom, this leads these authors to recommend presumed consent for screening of treatable diseases with explicit consent required for additional testing for new disorders and for storage. On the other hand parents should give written consent for screening of untreatable disorders, for future use of samples if there is planned research at the time of collection or if the samples are to be used in research where they will not to be anonymised. Research in this latter case should only proceed only with approval by an ethics process (Laberge 2004).

Other issues raised by Laberge and colleagues are the need for education for both health professionals and the public, and that parents should be informed about screening and storage prior to the sample collection.

### **Guidelines and policies for newborn screening**

The documentations include policies and guidelines from three countries. Those from Australia and the United Kingdom support a voluntary model of informed consent for newborn screening whereas the Public Health Laboratories says that explicit parental consent is unnecessary for mandated screening but this applies only for those conditions that can be treated and with parents and provider education program (APHL 2002).

The joint policies of the Human Genetics Society of Australasia (HGSA) and the Royal Australasian College of Physicians (RACP) for newborn screening and the retention and

storage of samples form the basis of the policies in Victoria and New South Wales. Specifically the HGSA-RACP policy recommends universal screening for three disorders (PKU, CH and CF), voluntary participation, public funding and appropriate follow-up (HGSA undated). Additional reasons for the use of retained samples include the need for samples to maintain quality assurance in existing testing procedures and for the modification of existing tests, as well as for the development of new tests and the investigation of missed cases (HGSA 2004). In the event of refusal HGSA-RACP recommend that parents be required to sign a written statement confirming they have been informed and understand the consequences of not testing. The HGSA policy statements do not include guidance on how and when the procedure of heel prick screening should be conducted.

In contrast the policies of the State departments responsible for health services contain guidelines for the procedure. This includes how the test should be conducted and when. Both Victoria and New South Wales recommend that the test be performed when the baby is between 48 and 72 hours old. Both States offer similar advice about giving information to parents about newborn screening and how to manage refusals. Health services are advised to provide parents with a specific pamphlet prior to the test, outlining newborn screening prior to testing. Both policies also provide additional information about retesting and the role of people nominated for newborn screening liaison in hospitals (DHS 2001, Public Health Genetics 2005, NSW Health 2005).

There are a couple of minor differences between the policies of the two states. Parents who refuse newborn screening in Victoria are referred to genetic Health counsellor for 'urgent discussion', whereas the New South Wales policy advises that parents who refuse the test speak to a paediatrician and be offered the option of speaking to the Director of the Newborn Screening Program. Both states require documentation of any refusal to be recorded on file and the New South Wales policy stipulates that consent, provision of appropriate pamphlet, discussion and completion of test to be also documented. The NSW pamphlets are not to be distributed without relevant discussion.

Other differences between the policies are the inclusion of information related to warming the baby's heel prior to the test (this is deemed unnecessary in the UK material) and the need to note the occurrence of twin-to-twin transfusion on the sample card in New South Wales and additional detail about the test results for Cystic Fibrosis in Victoria.

New South Wales also provides information to health professionals about the storage of newborn screening samples including reasons for storage (laboratory audit, to develop new tests, for family use), an assurance that no DNA tests are done on 'the vast majority of samples' and no data about DNA is stored, and advice that there is a memorandum of understanding between the health department and the police for the use of samples to identify human remains.

Both New South Wales and Victoria screen for PKU, congenital hypothyroidism, and Cystic Fibrosis. NSW also identifies galactosaemia and 'some fatty acid, organic acid and other amino acid defects' in their newborn screening while Victoria identifies 'over 20 additional metabolic conditions' specifically identifying Medium Chain Acyl Coenzyme A (MCAD) deficiency, homocystinuria and maple urine disease.

Information downloaded from the website of GHSV in February this year contains more detail but is no longer available on-line.

The policy approach in the United Kingdom to newborn screening has been thorough. It includes the development of policies and standards for all aspects of newborn screening. The UK documents include the consultation paper, the approved policies and standards, communication guidelines for discussion with parents, a health professional handbook and an information pamphlet for parents (UK NSPC a & b 2004, UK NSPC a & b 2005).

The standards are to be used to monitor quality and performance of the screening program and cover the timely collection and dispatch of samples, universal offering of testing, capacity to track samples and identify babies who do not provide samples or have positive screening results.

The most significant difference between procedures in Australia and the UK is the timing of when the test is conducted. The UK stipulates that the heel prick test be performed between Days 5 and 8 (more than 96 hours after delivery) whereas in Australia the sample is taken between 48 and 72 hours after birth.

Another major difference between the two countries is the provision of comprehensive evidence-based information to health professionals to enable them to answer parents' questions. For example it differentiates between different forms of PKU, and outlines the difficulties around screening for secondary hypothyroidism. Overall the information describes the complex issues underlying newborn screening and provides a useful resource to address the questions that parents may have. Explanations are given about common disorders that are screened (PKU, CH, CF and sickle cell disorders), their incidence, the available treatment and benefits of screening, and the difficulties that screening raises. The accompanying guidelines to offering screening would be helpful in ensuring that parents are properly informed before consent, that will be told about the limits of screening, and that a repeat test or further diagnostic tests may be required. The final major difference between the information given to parents in the UK and Australia is the notification of the carrier status of the baby.

#### **Verbal v written consent**

Though most newborn screening programs screen for treatable disorders without explicit consent, there is general agreement amongst guidelines that parents should be adequately informed about newborn screening. Those who support a choice approach for screening argue that obtaining consent will not compromise uptake for screening (Laberge et al). Written consent is recommended for tests which are investigational or where the value has yet to be determined, and when it is anticipated that the sample may be used for other purposes. Detailed safeguards for the safe-keeping of stored cards are outlined (HGSA 2004). Supporters of a written consent model state that the written form can become an educational document, provides medico-legal evidence of the conversation, and there is evidence that verbal and written consent together improves recall of the information given (Grice, 1988; cited in SOAP. 1998)). Whilst some supporters of a verbal consent model feel that pre-printed written consent forms may detract from the verbal consent process, and have the potential to be misused by institutions (obtaining implied consent not necessarily informed consent). Laberge et al. also support an informed choice model that separates consent for the screening of treatable conditions from conditions that are not treatable and the ongoing storage and secondary use issues.

#### **Health research related to communication issues around newborn screening**

The only primary research included in the documents is a report of a descriptive cross-sectional survey of women's knowledge of genetic screening within 24 hours of their babies undergoing a heel prick test in Melbourne, Victoria (Suraidi 2004). The study involved 200 interviews with ethnically diverse women using interpreters where necessary. Two hundred and thirty-two women who delivered live born babies consecutively at a tertiary hospital and who had been offered prenatal screening for Down Syndrome were approached to take part in the study. Participants were comparable with other women giving birth in Victoria in terms of age, marital status, parity and whether they held religious beliefs, although Arabic and Turkish women were over-represented, as were women of Islamic faith. The authors do not report the year that the study was undertaken. Women had limited knowledge about the terms used in counselling for Down Syndrome and newborn genetic disease. The median score was 4 points (out of maximum knowledge score was 15) (IQR=2-8). While 72 percent of

women indicated that they had heard of the term 'genetic disease' only 43.5 percent were able to accurately explain the term. Sixty-two percent had heard the term 'genetic screening' but it could be explained adequately by only 30 percent and 37 percent had heard of 'Guthrie', 'heel prick test' or 'newborn screening' with only 3.5 percent able to describe its purpose. Only 26.5 percent of women said that they knew that baby had undergone the heel-prick test whereas it had not been done for only four babies (2 percent). The mothers of these babies had specifically refused the test. Two women refused because of the distress of a previous false positive result, one because of the distress it caused her older child, and one because it resulted in a heel infection in an older child.

The other research reports relate to systematic reviews. A comprehensive review assessed the impact of disclosing to parents their baby's carrier status following newborn screening (Oliver, Lempert *et al* 2004). There were no controlled trials found (reported separately as a Cochrane review (Oliver, Dezateux *et al* 2004). Five of six studies addressing parents' views were assessed to be of sufficient quality to be reliable. Parents whose babies were found to be carriers of cystic fibrosis said they preferred to know their child's status and anticipated informing them at a later time. Most parents did not change their reproductive plans and found discussing carrier status with the wider family necessary but difficult. Parents preferred familiar non-specialists to give them positive test results. Resources and support are needed for health professionals to perform this role (Oliver, Lempert *et al* 2004).

A 1999 review of controlled trials to assess interventions to affect informed consent concluded that there was a lack of good quality studies that were theory driven and more primary research in this area is needed (Bekker 1999).

#### **Information for parents about newborn screening**

Most information for parents is provided in the form of a pamphlet or fact sheet. There is information for parents from four Australian states: Western Australia, South Australia, New South Wales, and Victoria. The information for WA and SA parents explains some limits of a screening test, and that further tests may be required for diagnosis. Only the WA information advises that newborn screening is voluntary but highly recommended and includes advice for parents who have homebirths. All states report how long the cards will be kept (from 2 to 50 years), and state that samples may be used in research after the removal of personal information. All four states test for PKU, CH, and CF but there is some variation in other disorders that are screened.

The fact sheet prepared by the Centre for Genetic Education appears to be based on the HGSA policy statement. It includes mention of homebirth and unlike the information from the states says that no further tests will be done on samples without written consent. The information from the Better Health Channel website explains that a positive test result does not mean that a baby is affected by the disorder. The parent's information pamphlet from the UK covers all issues raised in other documents as well as advising that carrier status will be detected.

The documents include information for parents from five states of the USA (California, Washington, Minnesota, Oregon and Michigan). Screening is mandatory but there is provision for refusal in some states for religious reasons (Washington and Oregon). There is no public funding. A sixth pamphlet offers a commercial screening service to parents for the full range of disorders that can be detected (SBTSF 2004). In all but Michigan a second test is required in second week after birth but it is recommended in that state if the first sample was taken in the first 24 hours after birth. The range of disorders screened varies in each state but all test for PKU CH and CF. Washington and Michigan retain samples for over 20 years and Washington, Minnesota, Oregon allow for samples to be used for research with identification removed. Samples can be destroyed following a written request from parents.

## SUMMARY

The research undertaken by Suraidi *et al* demonstrates that there is a low level of knowledge among women who have recently given birth about genetic screening. It also reveals that women will refuse newborn screening for issues that are not addressed in the information given to parents (false positive results and the distress and harm that may result from the procedure itself). These findings support concerns about whether informed consent processes around newborn screening would meet the test of law. The review of studies of parents' views of the communication of positive test results by Oliver *et al* (2004) supports familiar non-specialists in this role. This requires health professionals to be educated about the complex issues around newborn screening. Other issues to be resolved are: disclosure of carrier status, reconciliation of differences in international practices about when it is best to conduct the test, and the need for written consent.

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### **Background Information about the current Newborn Screening Program**

The following background information was developed and used in the consultations to provide consistent and up-to-date information about the newborn screening program to the participants.

#### **1. Background**

The newborn screening program has been available to all babies in Victoria for the last 30 years. The newborn screening tests enables the early detection and treatment of a number of rare conditions that may cause serious complications. If these conditions are detected immediately after birth, early treatment can prevent or delay possible complications, and in most cases the child will grow and develop normally. Conditions screened for include:

- **Phenylketonuria (PKU)**

This is a condition that affects about five babies in Victoria each year. If the condition is not treated immediately severe, progressive intellectual disability results.

- **Congenital hypothyroidism**

This condition occurs in about 20 babies in Victoria each year. A lack of thyroid hormone can cause severe intellectual disability and growth problems.

- **Cystic fibrosis (CF)**

This is a severe genetic condition that affects about 20 babies in Victoria each year. CF affects the lungs and digestive system.

- **Other conditions**

More recently (from February 2002) because of advanced technology (tandem mass spectrometry) at least 20 other rare metabolic disorders can be detected. These affect about nine babies each year in Victoria. Early detection and treatment before they become sick results in a better outcome for these babies.

#### **2. Parental consent**

The Department of Human Services (DHS) guidelines for newborn screening (November 2001) state that before the screening test is performed, hospital staff must ensure that parents or guardians are properly informed about the test and its importance. Information for parents about the newborn screening program was developed in September 2002. The guidelines advise that these pamphlets should be given to pregnant women prior to delivery and the pamphlets should also be available for review after their baby's birth. The hospital where the birth occurs is responsible for ensuring all babies are offered the newborn screening test. This includes babies who are transferred to other hospitals or domiciliary midwifery programs.

Hospital staff should obtain verbal consent from parents or guardians before performing the newborn screening test. There should be documentation on the mother's/baby's file stating that there has been discussion about the test. The file should also show a record of completion of the test. Any parents refusing the test will need to sign a written statement showing that they understand the

potential risk to the healthy development of their baby. This statement must be documented, signed and placed in the mother's/baby's file.

### **3. Collection of blood sample**

The newborn screening test is carried out on a blood sample obtained by a heel prick, placed on special pre-printed filter paper, air-dried and processed at the Newborn Screening Laboratory of Genetic Health Services Victoria (GHSV). GHSV is funded by the Victorian Government to do the newborn screening tests. It is a not-for-profit organisation that is privately owned and operated by the Murdoch Childrens Research Institute and is located at The Royal Children's Hospital.

It is recommended that the newborn screening test be performed when the baby is between 48 and 72 hours of age. False positives and negatives can sometimes occur when the screening tests are done before 48 hours.

### **4. Testing and Storage**

Initial test results are normally available 24 hours after the sample is received in the laboratory. Newborn screening for cystic fibrosis involves several different tests and takes between four to six weeks to get the initial results.

When a test clearly shows the presence of one of the disorders, the parents of the baby is notified by telephone, and confirmatory testing organised.

The blood samples are securely stored for two years at GHSV in order to provide quality assurance for the testing. After this time they are stored indefinitely in a secure off-site facility.

### **5. Access and secondary use of blood samples**

The newborn screening card is regarded as a health record and as such the Victorian Health Records Act, State and Federal Privacy Act, and the Public Records Act govern its storage and access. In some instances Genetic Health Services Victoria on request has provided transfer of the card to parents. Access to the cards by the coroner, law enforcement agencies and researchers is available. However, specific protocols have been developed by Genetic Health Services Victoria and agreed to by the Department of Human Services regarding access.

Secondary use of the card includes:

- Quality assurance of testing - access to the blood sample in case of false negative results, cases of misdiagnosis, or where new tests become available.
- Diagnosis of other conditions in individual infants, diagnosis of retrospective infections, and genetic mutation information for current or future siblings - requires parental consent.
- Access by law enforcement agencies – police access to newborn screening cards by court order was introduced in 2003.
- Forensic identification – access to the newborn screening cards can be sought for identification of a body or body parts at the request of the Coroner's Court.
- Research – approved by a Human Research Ethics Committee with access to identified samples (with parental consent) and de-identified samples (not requiring parents consent)



INFORMED PARENTAL CONSENT FOR NEWBORN SCREENING

Validation of recommended model

Issues Paper Response Template

Please provide comment on the following questions then post this form to Health Issues Centre in the envelope provided or fax back to 94795977.

Q 1. What are the strength and weaknesses of the recommended model? We suggested some criteria for your assessment. Spaces are provided for your additional criteria/comments.

Table with 3 columns: Criteria, Yes/No/Unsure, Comments. Rows include questions about parental information, choices, screening purpose, practicality, and privacy.

Q2. Do you support the split consent model (one consent for the screening test and a second consent for storage, access and secondary use)?

.....

Q3. Do you support parental consent for the screening test being obtained verbally or in writing?

.....

Any additional comments

.....

Your name (optional) .....

Thank you for your time.