

Report prepared for
NSW Genetic Services Advisory Committee, NSW Health

Community Views and Perspectives of Newborn Screening

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Background

The NSW Newborn Screening (NBS) programme has been operating for almost 40 years and each year about 85,000 babies are screened through a blood test. The NBS programme detects about 90 babies each year with rare medical conditions. Current NBS protocols, procedures and rules are outlined in a NSW Health Policy Directiveⁱ and are underpinned by the understanding that informed parental consent must be obtained.

This research has explored the experiences, awareness, perceptions and attitudes towards NBS amongst a sample of health consumers. The study has used qualitative research methodologies to explore participants' attitudes towards the conditions for which NBS is offered, the policies and procedures governing the programme in NSW and the issues arising from the retention of the NBS Sample Cards.

Methodology

Forty health consumers were recruited from a range of backgrounds with a potential personal interest in NBS and were interviewed in 9 re-convened group discussions. The groups, comprising 24 women and 16 men from a range of socioeconomic levels, included both first time and older parents and also young people without children. The sample was not intended to be statistically representative of the NSW population but to explore a range of views likely to be held in the wider community.

Each group met on two occasions. The first meetings began by exploring participants' general knowledge and perceptions of health, screening and testing before moving on to specific knowledge, experiences and views of newborn screening. Carefully prepared stimulus and information materials were used to support some more focussed consideration of specific NBS related issues. During the second meeting of each group, participants considered these issues further and explored how their views had developed. Interviews were audio recorded, transcribed, coded and analysed to identify themes and relationships.

Results

Perceptions of Newborn Screening

- NBS was considered in the wider context of their experiences and views towards other health issues.
- The term Newborn Screening as specific to the programme offered was generally unfamiliar although mention of the heel prick blood test did prompt some recall.
- There was little specific knowledge of protocols, procedures and rules related to NBS but rather this was seen to be simply a part of all the testing and screening done in the newborn period.

i. *Newborn Screening Guidelines, Policy Directive PD2005_566*. New South Wales (AUS): New South Wales Health;2005. Available from http://www.health.nsw.gov.au/policies/pd/2005/pdf/PD2005_566.pdf (accessed 21 Feb 2006)

Support for Newborn Screening

- All participants were highly supportive of the principles of current testing performed as part of NBS. The value of testing for treatable conditions such as phenylketonuria (PKU) was widely accepted.
- Access to all available screening was often seen as a right by parents.
- Participants also saw potential benefits for inclusion of screening for conditions where there was no current medical treatment, many citing the benefits of possible social interventions such as how early information could help families plan their lives.
- It was also recognised that results of wider screening had the potential to increase psychological stresses for some families.

Storage of Samples

- The concept of storage and further use of blood, tissue and biological samples after initial medical testing had not previously been considered by participants.
- Information provided to the participants about the current practice of storage and further use of NBS Sample Cards was surprising and even shocking to them.
- Participants widely perceived that it was vital for parents to know about these aspects of the NBS programme.

Using NBS Sample Cards - 'Family Benefit'

- The concept of 'family benefit' from the storage and potential for future testing of the NBS Sample Cards was not clear and often interpreted as parents in general being able to access the NBS Sample Cards at a later date to access further health information.

Using NBS Sample Cards - Medical Research

- Use in medical research was generally viewed supportively. The concept of using anonymised samples in medical research was clearly grasped. It was widely agreed that parents should be informed and consulted if identifying information would be provided to researchers requesting the cards.
- Generally prepared to let decisions about appropriate use in this context lie with others representing the public good such as Ethics Committees. However, the prospect of their use for human cloning was most commonly put forward as an example of what would be an unacceptable use of the NBS Sample Cards. Research uses that lead to discrimination by insurers or employers were also of concern. Some participants had strong views that access by pharmaceutical and biotechnology companies was also unacceptable.

Access By Police and Courts

- Access to the samples for solving crimes and identifying missing persons was not controversial for many participants.
- Male participants did have some concerns in this area and some participants had a clear general lack of trust in the behaviour of the police.
- While outlining the current agreement between police and health authorities appeared reassuring, there was still some continuing concern expressed among many of the male participants.

Retaining NBS Sample Cards and Records

- The rationale for the 18 year storage period for the NBS Sample Cards perplexed several participants. The concept of different NBS Sample Card retention policies in different Australian States was unsettling for many participants.
- Some questioned why the NBS Sample Cards were not stored for longer if they had such value for further medical research.

- No participant suggested that currently stored cards should be destroyed prior to the current 18 year retention period.
- Generally ownership of the NBS Sample Cards was not claimed by participants although they did view that they had some rights over the samples and should have some say over how the sample was used.
- A distinction was made by several participants between the storage of the NBS Sample Card and the records containing the results derived from testing. They questioned whether the electronic records were also destroyed after 18 years.
- Several male participants suggested unprompted that the samples could be used to create a DNA database.

Informed Consent

- Almost all thought that consent was an important matter and that parents should be asked or informed in some way.
- Parents were often uncertain what information they had been offered about NBS and if any discussion had taken place with health professionals. Parents that did recall giving some sort of consent noted their various experiences of how the topic was raised by health professionals. Most seemed satisfied with the manner with which this had been raised but a few expressed some frustration and that they were offered little real choice.
- Many viewed that written consent had some advantages such as making people think more about NBS before consenting. A consistent reason proposed by the participants for the use of written consent was that such a process would give legal protection to the hospital.
- Most felt that even if they personally were very positive towards NBS others might have objections and parents had a right to make this choice. It was perceived that very few parents would refuse testing and their reasons would likely be based upon religious or cultural beliefs.
- Several felt that NBS should be compulsory or mandatory when considering PKU testing. This topic prompted considerable discussion and the views of many individuals appeared quite fluid and liable to change.
- Few parents directly recalled having seen information materials which the NSW Policy Directive requires should be given to all parents. Much of the detail mentioned in the pamphlet was entirely new information to them.

Delivering Information

- *Timing:* During pregnancy rather than after birth was repeatedly suggested as a more effective option.
- *Format:* Using human stories such as those presented to them as stimulus materials in the discussions was thought to be a powerful mechanism for conveying the importance of screening to parents. Inclusion of information about NBS in other printed materials and health resources available to parents was also recommended.

Conclusion

This study demonstrates that members of the wider community are clearly able to engage and consider some of complex issues and grasp some of the key scientific, social and ethical issues raised by developments in genetic health. This bodes well for a more inclusive strategy in the development of public policies in the developing field of genetic health.

Recommendations

- 1) ***Understanding Current Practice*** - The experiences and views of health professionals involved in directly delivering NBS testing needs to be actively sought and explored further.
- 2) ***Delivering Information*** - Further policy development regarding best practice in the timing and distribution of information about NBS is required.
- 3) ***Developing Materials*** - The timing and mechanism for future review and revision of the NBS pamphlet needs to be considered. Future printed information may benefit from a greater emphasis on the use of accessible and non-technical language, the use of narratives and personal stories. The provision of up to date printed information in languages other than English should be an integral part in the development of future materials.
- 4) ***Supporting Health Professionals*** - The level of current support and training for health professionals in this area needs to be considered. This includes those professionals directly involved with implementation of the NBS programme, those providing information and education as well as NBS programme staff.
- 5) ***Communication Strategy*** - The apparent high level of public confidence in the NBS programme may be vulnerable and needs to be actively maintained and supported by a communications strategy.
- 6) ***Public Confidence and Transparency*** - In order to maintain public confidence and meet future challenges it is likely that certain areas of policy and practice may require further public explanation or justification.
- 7) ***Community Involvement*** - There is a need to continue to incorporate community perspectives into further programme and policy developments.



1. Background

1.1 Introduction

The NSW Newborn Screening (NBS) programme has been operating since the late 1960's. The programme was initiated by the development of a biochemical assay by Guthrie in 1963 to detect the chemical imbalances found in the blood of babies affected by phenylketonuria (PKU).¹ PKU is a rare metabolic condition that, without early treatment, leads to brain damage. However early detection of PKU in the first few days of life and the use of a special diet can prevent the development of life threatening symptoms.

The blood sample from a newborn baby is taken by a heel-prick 48-72 hours after birth. The sample is dried onto cards as three blood 'spots'. The NBS Sample Cards are sent to the NSW NBS Laboratory located in the Children's Hospital at Westmead. The cards were originally known as Guthrie cards in reference to the PKU test but they are now preferably referred to as Newborn Screening Sample Cards.

Screening for several additional disordersⁱⁱ was introduced during the 1970s and 80s and testing for cystic fibrosis was introduced in 1981. The introduction in 1998 of testing using Tandem Mass Spectrometry (TMS) methods² enabled the inclusion of a much larger panel of conditions. Currently the collected newborn blood spots are screened for about 30 conditions where early detection and treatment can positively impact on the health of an affected child and family.

Each year about 85,000 babies born in NSW³ are screened under the programme. About 90 babies are diagnosed with one of the conditions for which screening is undertaken.

While NBS programmes have been operating in Australia for over 40 years, the experiences, awareness, perceptions and attitudes towards NBS amongst the wider community and health consumers have not been formally explored. Such community consultation is essential to guide the development of educational materials and strategies. Additionally, the importance of engaging the public in the development and application of new technologies is now generally accepted by scientists and policy makers. NSW Health has recognised the principles of community involvement and participation in health services. These policies encourage the health system to give the community *"a greater say in health decision-making"*.⁴

Moves towards the creation of a deeper public dialogue with Australians about genetic health and medical research will need to be underpinned by a better understanding of perceptions, knowledge and attitudes and what underlies these. They will also likely require some further development and testing of appropriate public involvement methodologies.

In 2005 the NSW Genetics Services' Centre for Genetics Education, a NSW Health funded statewide program, undertook a study using qualitative research methodologies to explore participants' attitudes towards the conditions for which screening is offered in

ii Hypothyroidism, Galactoseamia and Homocystinuria

NBS, the policies and procedures governing the programme in NSW and the issues arising from the retention of the NBS Sample Cards.

1.2 Procedures and Consent

Current NSW Health policy⁵ defines how NBS should be delivered to consumers. It is based on the understanding that informed parental consent must be obtained before the procedure is carried out. The Directive outlines the procedures for the collection of samples by health professionals, the forwarding of samples to the NBS laboratory and the keeping of appropriate records. Local hospitals are required to identify a responsible person for the coordination of screening and develop their own protocols to manage newborn screening.

Communications that should operate between hospitals, the NBS laboratory and clients are also described. For example where testing results require urgent follow-up, the responsible health professional is notified by telephone of the test results and should then ensure that the baby is promptly referred for appropriate investigations and treatments. If further testing or retesting is required, they must ensure that a second sample is obtained, the procedure recorded and the sample sent to the laboratory.

The Directive states that parents should be asked to give verbal consent to the test before the heel-prick is performed and this consent be noted in an agreed format in the medical record of the mother/child. In cases where the parents do not initially consent to NBS the hospital protocol should include further informing the parents about the importance of the test, providing an opportunity to talk to a paediatrician, listening to the concerns expressed by parents and offering parents the option of discussing the matter by telephone with the Director of the NBS Programme.

Information from the NBS programme indicates that following such discussions, most parents do consent to screening. In cases where parents still refuse NBS, the Directive requires that refusal is recorded in the mother's/baby's file and signed by a health professional.

1.3 Information Provision

The Directive states that “at some time during the pregnancy, it is recommended that parents be told about newborn screening, and that they be provided with the pamphlet “Tests To Protect Your Baby”.⁶ Discussion of the information is mandatory and group situations such as antenatal classes or one-on-one situations are suggested as appropriate settings for this discussion.

The current version of the pamphlet contains information covering:

- Reason for screening, and why early diagnosis is important
- Information about the conditions screened
- The possible detection of cystic fibrosis genetic carriers
- Notification of results – only informed if retest needed, or condition suspected
- Reasons for retest
- That there is no direct cost for screening
- How the sample is collected
- Where the sample is tested
- How it should be recorded that the NBS sample was taken

- Period of storage of NBS Sample Cards
- Potential uses of NBS Sample Cards
- Access to NBS Sample Cards for further uses
- Where to find further information
- That the pamphlet should be kept by parents for 3 months after baby born
- Privacy Notice
- Contact details.

1.4 Clarification and Notification of Results

Sometimes it is necessary to repeat the test or conduct further testing and this may require another blood sample to be sought. This may be necessary if the results of an initial test are inconclusive or if the initial sample is insufficient or contaminated. In approximately one percent of cases a DNA test is required to clarify the initial results.⁷ A DNA test on the sample is required when a diagnosis of cystic fibrosis is suspected.

Parents are not notified of NBS results if no condition is detected.

1.5 Storage and Destruction of Cards

Current retention policies differ significantly across Australian States and Territories with some States retaining NBS Sample Cards indefinitely and others holding cards for just two years.

In NSW the NBS Sample Cards are stored in a secure facility at the NBS Laboratory located in the Children's Hospital at Westmead. Currently, cards are destroyed by being shredded once they have been stored for 18 years.

The retention of NBS Sample Cards in Australia has been addressed by several professional bodies which have made their own policy recommendations.^{8,9,10} The issues were also considered by the Australian Law Reform Commission which recommended the development of nationally consistent standards.¹¹ Proposals for a uniform approach to newborn screening are being developed by an advisory group of the Australian Health Ministers' Advisory Council and put forward in an initial public consultation.¹²

1.6 Access to and Further Use of NBS Sample Cards

A key value in storing the NBS Sample Cards is for quality assurance and audit practices by the laboratory. The blood spots may also be used to develop and test protocols to further expand newborn screening.

The stored card may be used, with the consent of parents, for the medical benefit of affected families such as enabling prenatal testing in future pregnancies where information from an affected child is otherwise unavailable.

The blood spots, with some provisions, may also be used for medical research. Such research must adhere to the NHMRC National Statement requiring that all research proposals involving human participants are soundly designed and conducted according to high ethical standards.¹³ All the research conducted requires the approval of a Human Research Ethics Committee (HREC).¹³

Under the National Statement, HREC's may waive the requirement for individual or parental consent in certain circumstance. Where the research is undertaken on anonymised blood spots, parental consent may not be required. However, HREC approved research on identified blood samples requires parental consent. The NSW Health policy states that *"no further tests will be performed on any identified stored blood sample by the newborn screening laboratory without written consent from both parents/guardians, the child if old enough or other lawful authority."*¹³

Approximately 400 NBS Sample Cards have been used in the last 3 years for HREC approved research.⁷

There is also a Memorandum of Understanding between NSW Health and NSW Police that provides a protocol for police access to the NBS Sample Cards as part of their investigations.¹⁴ The limited circumstances cover "for the purpose of identifying human remains" and/or "where the NSW Police is in possession of forensic samples suspected to come from a victim of a crime taken from a suspected crime scene in relation to the commission of an offence involving harm to another person's life, safety or liberty and that victim cannot be located."¹⁴ The NSW Police must make reasonable efforts to secure consent.

Access under the MOU requires the approval of the NBS Programme Director. About ten NBS Sample Cards have been requested by the Police under the MOU protocol since its instigation in 2002.⁷



2. Methodology

2.1 Research Questions

The aim of the study was to consult the community about NBS specifically focussing on the:

- Exploration of participants' knowledge and views of what is currently included in NBS
- Investigation of participants' perspective and attitudes towards potential future expansion of NBS with a particular focus upon the inclusion of other conditions where there was no current treatment, later onset conditions and genetic susceptibility screening
- Exploration of participants' knowledge and perspectives of provision of information about NBS to parents.
- Exploration of recall of information by parents with direct experience of NBS and their views as to how communications might be improved
- Examination of participants' views towards the consent to NBS. The current process of verbal consent was explored alongside alternatives such as written consent and mandatory screening
- Investigation of the knowledge, awareness and views among the participants' about the storage and further uses of NBS samples. Consideration of the time period cards are stored
- Exploration of the perceived benefits and concerns raised with the further uses of the stored samples
- Exploration of the issue of trust among the participants' in the medical profession, researchers and other stakeholders such as police and courts, commercial organisations and government as it related to governance of NBS.

2.2 A Qualitative Approach

A qualitative approach was called for to explore the range and depth of views held by the community. A quantitative approach, such as developing a survey, would be of little value at this stage since we simply do not understand where people begin when they consider these issues. Whilst many people would probably give a response to a survey question, if they do not understand the question and the background, these responses would simply be 'non-opinions'.

The reconvened group discussion methodology that was used has previously been applied to explore community perspectives of new developments in medical and genetics research¹⁵ and the study built upon these experiences. In particular this methodology can be useful to explore beyond initial responses to relatively unfamiliar topics and to explore more considered views where there are opportunities for further information, deliberation and reflection.

The project was developed with guidance from an Expert Advisory Group (EAG) with experience of NBS, policy, health ethics and communications. The research protocol was approved by the local Human Research Ethics Committee.

2.3 Sample and Recruitment

The sample included people from a variety of backgrounds and was chosen to offer a range of views about the research topic. Groups were structured such that participants shared similar socioeconomic backgrounds and all groups were single sex. Potential recruits from fields related to market research and health sectors were excluded since it was felt their specialist knowledge would have a detrimental effect on the development of group dynamics. The sample was not intended to be statistically representative of the NSW population but covered a range of socioeconomic levels, and age groups and included:

- Young adults whose NBS Sample Cards had recently been destroyed recruited through youth agencies in regional NSW (Groups 1 and 2)
- Parents who had likely recently experienced NBS recruited through several Child Health Centres in suburban North and West Sydney (Groups 3-5)
- Parents with older children where the child's NBS blood spots were being stored recruited through a professional market research field recruitment agency in suburban North and West Sydney (Groups 6-9).

Nine group discussions were undertaken with 40 participants in total, comprising 24 women and 16 men. There were between three and eight participants in each group (Table 1).

Table 1. Sample Characteristics

Group	Gender	Group Size	Age Youngest Child	Area
1	F	8	-	Regional NSW
2	M	3	-	Northern Sydney
3	F	4	< 6months	Inner West Sydney
4	M	3	< 2 years	Northern Sydney
5	F	3	< 6 months	North West Sydney
6	F	4	5 -18 years	Northern Sydney
7	M	5	5 -18 years	North West Sydney
8	F	5	5 -18 years	Inner West Sydney
9	M	5	5 -18 years	Inner West Sydney

Participants were provided with refreshments, childcare support where required and a \$50 shopping voucher for attending both sessions.

2.4 Initial Discussions

Discussion groups were held in June 2005. Each group met initially for about 90 minutes. Interviews were conducted using an agreed topic guide (Appendix 6.1) and by presenting stimulus materials (Appendices 6.3 and 6.4) intended to support further in depth discussion. The discussions about NBS were introduced from a base of general health perceptions and information sources and then moved to understanding of the concepts of screening and testing. Whilst the research approach and use of the stimulus materials required some structure and order to the conversation, it was also possible to consider issues as they were naturally raised by participants.

The principle was to explore unprompted knowledge and understanding in several areas then to elicit further consideration of the stimulus material. Each initial group began by

exploring participants' knowledge and images of health, screening and testing in general before moving on to specific knowledge, experiences and views of newborn screening.

2.5 Reconvened Discussions

Each group was then reconvened between one and three weeks later for a further discussion session of about 75 minutes. These sessions also followed a general topic guide (Appendix 6.2) although this was moderated in a less structured manner and focused upon the issues that participants themselves raised after considering the issues.

The intention here was to examine how participants had considered the issues over time and to what extent their views had developed. Emphasis was placed upon discussions of the storage and further usage of the NBS Sample Cards.

2.6 Stimulus Materials

The materials (Appendices 6.3 and 6.4) were developed with advice from a medical journalist and the EAG with the aim to be concise, accurate, accessible and engaging. They addressed key information from the NSW Health NBS pamphlet which was also shown in the groups. The stimulus material covered information about the tests undertaken for specific conditions, storage of NBS Sample Cards and further uses and information about procedures and rules.

The materials included several showcards and four scenarios.

The showcards were introduced as outlined in the topic guide. They presented in simple bullet-point information about:

- Current testing and NSW NBS programme
- Consent procedures and policies
- Storage of the NBS Sample Cards
- Possible uses of the samples
- Rules governing further use of the NBS Sample Cards.

The groups were also asked in the first session to consider four fictional scenarios which were intended to simulate further discussion. The scenarios covered:

- A mother whose child is diagnosed with PKU
- An couple overseas whose child is diagnosed with Duchenne Muscular Dystrophy (DMD)
- A couple who have been asked to consent to the further use of a stored sample
- A woman who wishes to access a sample in a paternity case.


After the first session, participants were provided with a hard copy of these stimulus materials in the form of an eight page A5 booklet to take away. They were encouraged to further consider the issues that had been discussed in the first session and also to seek out the views of others with whom they had contact. A small notebook was provided to assist them in keeping track of their considerations and wider discussions.

2.7 Analysis

Group discussions were audio recorded and professionally transcribed. Transcripts were checked and corrected by all authors. Where possible the individual speaking within a

group was identified in the transcript such that comments could be followed through each session and used in the analysis.

Transcripts were analysed using a Framework methodology¹⁶ which identifies and codes common themes. Data was managed by entering into a series of Microsoft Excel spreadsheets which allowed cross tabulation of individual responses against these themes. Where found necessary, further development and editing of themes was undertaken during the coding process. Consistency in coding was assured by independently coding several transcripts with differences discussed and resolved by the researchers.



3. Results

The study first probed how individuals gathered health information, especially about child health matters, and how they viewed these different sources. In this context the moderators then explored if, and how, participants made distinctions between the role of “testing” and “screening” in health and these terms were presented as discussion prompts to the groups. It was only at this point that the issue of NBS was raised. The basic perceptions and associations held by participants with the term “newborn screening’ were explored before any specific information on the topic was provided.

3.1 Considerations of Health, Testing and Screening

Key factors commonly mentioned as having an influence upon health included lifestyle factors such as diet and exercise. Holistic factors such as mental wellbeing, stress and being in a loving relationship were also viewed as influencing physical health and the ability to fight disease.

Perceptions were influenced by their personal and wider experiences of health or ill-health as well as parenting. Participants offered specific personal narratives about the health experiences of themselves and that of their family and friends. Whilst their stories were often initiated or at first framed around family relationships it was rare for family history to be described as a major influence upon health. In one case however a participant did describe her direct experience of genetic testing for cancer and the wider impact this had had upon her family.

Participants described varying levels of interest and knowledge in health matters and different strategies for seeking health information. First time parents often described themselves as “*information hungry*” and had read extensively during their pregnancy. Sources of information included material given to them in prenatal classes and by health professionals. Amongst the younger fathers most had also researched several child health issues on the internet.

Many parents recalled receiving a copy of the Personal Health Record (commonly known as the “Blue Book”)¹⁷ which is given to parents after the birth of their baby. They noted that they are encouraged to keep this safely as the child grows up and this document was seen to be a key source of reliable information for future reference. Parents with older children seemed to also rely more upon experienced friends and family for information and specific advice.

Across all the groups the media was described as a source of health information, including television news and current affairs programmes. Talk back radio was a key source of information for some and they judged that if a health issue was of general public relevance they would expect to hear it discussed here. Participants also referred in the later discussions to popular television drama shows such as CSI. For example, almost all were very familiar with the potential these programs raised for DNA information to be used in forensic and criminal investigations.

Some participants viewed the terms “testing” and “screening” as almost interchangeable and it was often difficult for them to separately define the two terms. It was also suggested that screening might not be as accurate or definitive as testing but screening was also seen to be less invasive and safer than testing.

Testing was most widely perceived to be applicable as part of an ongoing medical investigation of an individual and where some form of medical problem had already been identified. In most cases testing was perceived as involving the removal of part of the body, most likely blood or tissue, and its further analysis. As one participant succinctly put it, testing was seen to involve *“cutting something off and sending it away”*.

Screening on the other hand was most commonly associated with cancer and specifically breast, cervical, bowel and prostate cancer. Common examples of such screening were mammograms and pap smears. In this context screening was seen to have particular relevance for older groups and populations. Some suggested that one difference between screening and testing could be that screening was likely to be a regularly repeated process rather than a one off procedure.

As an example, prenatal screening and testing were raised by several parents who referred to their recent personal experiences. In some of these experiences screening was viewed positively whilst in others participants talked about some of the difficulties this had raised for them.

Unprompted, just a few participants suggested that the heel prick test was an example of screening or testing. One younger father recalled how he had been present at the heel prick of his first child and even videotaped the procedure.

3.2 Exploring Newborn Screening

For most participants NBS was not a familiar term by itself but they associated it with a number of newborn interventions of which they had some experience. The NBS test was not perceived as different or special as compared to other tests conducted in the newborn period. The Vitamin K injection and Hepatitis B vaccination given to newborns were repeatedly mentioned and it was thought that these would likely fall under a heading such as newborn screening. Others suggested that the heel prick was part of testing for conditions such as autism, HIV, jaundice or blood clotting diseases.

Following prompting with the first showcard that described NBS and discussion between participants, the key trigger for most participants who recalled the test was mention of the heel prick blood test rather than the term Newborn Screening. Most women with young babies recalled the heel prick and mothers with older children also generally recalled the tests. Recall was also reported by all the younger fathers while a few of the fathers with older children had quite distinct recollections of the test being done. In particular parents recalled whether they had been present at the time of the test and whether the baby cried after the heel prick. Another common recollection was that this was the test where parents were only informed of the results if there was some problem.

Recollection of the heel prick was not generally linked to an understanding of what was being tested for but this did not seem a cause of concern. For example, a father who had videotaped his child having a heel prick stated that *“I don’t know what they test for. I know its something good. It’s not bad”*.

A few participants however seemed more confident in their knowledge and stated that the heel prick test was for cystic fibrosis. Two individuals recalled that the test was in some way linked to the *“artificial sweetener condition”*. In one case this information was gleaned from reading the label on the can of a diet soft drink. The other participant (a woman with several family members in the medical profession) specifically mentioned that one of the

conditions included was called PKU and that individual's affected needed to follow a special diet.

The screening was generally unfamiliar to the two groups of younger people even when prompted.

There was also no recall of any media coverage of NBS in any of the first discussion groups as a possible source of information. However, it did emerge in the reconvened groups that the partner of one participant had seen a SBS television program that included some discussion about the storage of NBS Sample Cards.

3.3 Views About The Conditions Included In NBS

All participants were highly supportive of the principles of NBS as outlined in a showcard and early detection of a condition was a key positive aspect for them. They knew little about the specific conditions detected and were surprised to hear that as many as 30 different conditions could be detected by NBS. A few participants were keen to have information about these other unfamiliar "*alphabet diseases*" currently included in NBS. Participants often did not initially realise that many of the conditions detected were inherited in families.

When presented with a short scenario around current PKU screening their responses were overwhelmingly positive. This was widely praised and the medical benefits of early PKU screening clearly outweighed any possible harms. None of the participants we spoke to raised personal or specific concerns with testing for PKU. Participants assumed that almost all parents would see the value in screening for such a treatable condition.

3.3.1 Treatable Versus Non-treatable Conditions

A scenario presented about screening for DMD raised the possibility of including additional conditions for which there is no medical treatment. Most participants immediately viewed this very positively and supported inclusion. Generally the view was initially expressed that if screening for a condition was available and could offer some information to parents they would want to know and even expected to know. Even if the information offered by a test was incomplete or in some way problematic, the information was still considered potentially valuable to parents.

That there was no treatment for a condition such as DMD was hard for many participants to accept and it was expressed that surely physiotherapy or even natural therapies would have some effect or a new treatment might be developed at anytime. But many participants also focused on the social value of having early information that would be critical for a family to plan and reshape their lives. Parents were seen to have a right to such information and to decide how to use it.

There was also however a general appreciation that some parents would not want to be offered information about a condition like DMD so soon after birth. The potential for psychological stress and for increased parental worry was raised. Also having an early diagnosis where there was no treatment could detrimentally change the relationship between parents and baby.

In the reconvened groups participants reported that most of the people they had spoken to outside the group felt parents should be offered all available screening.

3.3.2 False Positive Results

When considering the false positive rate for an initial screening test for DMD, some participants recognised that it may make parents unnecessarily worried. However most

viewed that this was an acceptable cost and expressed the view that parents would want to know if their child was affected. This situation was compared to prenatal screening that a number had personally experienced, where they had faced uncertainty and anxiety.

Participants were more likely to raise concerns about such false positive results at the reconvened groups. For some participants, further consideration of this issue had led them to shift their position such that they no longer supported the inclusion of DMD testing in NBS.

3.3.3 Options for Including Additional Tests

When considering the possibility of including screening for additional conditions a number of barriers were raised by participants. These included financial pressures upon the Australian health system. Several suggested that the fact that DMD screening was not available in NSW, yet available overseas, was an example of how medical developments in Australia were lagging behind other countries.

The economic value of NBS was queried by several participants, especially men, who requested further details when considering additional tests and indeed for the current screening. They felt that this was an important factor that needed to be carefully assessed alongside other medical and ethical questions.

One option suggested was that the initial screening for DMD should be included in NBS but then allowing parents to decide if they wished to have the further confirmatory test. Along similar lines it was suggested that the tests for additional conditions could be undertaken as part of the NBS package but the results only given to parents if and when they specifically requested them.

3.3.4 Late Onset, Susceptibility Testing And The Limits Of NBS

The possibilities of including additional tests within NBS for childhood cancer, diabetes, heart disease, multiple sclerosis, mental illness, depression and even hair loss were put forward by participants. There was a wide range of views as to the acceptability of such screening. Their personal limits on what they would support screening for were difficult to define. Most felt that this would be a matter of personal choice for the parent should such screening be available.

Screening for other conditions with childhood onset seemed natural and their views often followed from their support for including DMD as part of the NSW programme. Screening for susceptibility to conditions such as depression might initially be viewed in a positive and accepting vein. However concerns began to be raised that the information might be used to discriminate against individuals. Just a few personally supported screening their child for susceptibility to conditions such as depression. The concept that some potential tests might only show a greater risk of developing a condition did not always seem to be grasped thoroughly.

Overall, despite sometimes being personally supportive of having their child screened for a wider range of conditions than currently offered, many participants held back from

actively supporting the inclusion of screening for late onset conditions or susceptibility screening as part of the current NBS package. Some participants thought that it was important for NBS to remain focused on childhood conditions which could be treated.

3.4 Storage of Samples

3.4.1 Health Samples

The subject of storing NBS Sample Cards was approached by prompting some more general discussion about health samples, their experience, knowledge and views. Participants' awareness and knowledge of the use of blood, tissue and biological samples after testing was limited.

Almost all believed that such health samples would be destroyed after initial testing, citing reasons such as storage would serve no purpose; the sample is no longer needed; the patient can just give another sample if it is necessary and the results are probably kept but physical samples would be impractical to store. On probing and with further reflection a few suggested that there could be some value in storing certain samples, especially tissue for further testing and medical research.

3.4.2 NBS Sample Cards

All participants were initially unaware that the NBS Sample Cards, including those of their own children, were being stored. When presented with this information on a showcard reactions included surprise, curiosity and even shock. It immediately raised questions such as *"What's point of storing these blood spots?"*, *"Why the 18 years?"* and *"Wouldn't such widespread storage not be cumbersome?"*

Considering this new knowledge that NBS Sample Cards were stored led some to begin considering what had happened to other health samples that had been taken from them.

In the period between groups where participants were encouraged to engage their friends and family in discussion, only one participant found someone who was aware that the NBS Sample Cards were stored. Participants found that those with whom they spoke were equally unaware that the cards were kept and sometimes they were quite shocked.

Almost all participants felt that it was vital for parents to know that the NBS Sample Cards are stored for 18 years and potentially used for various purposes.

3.4.3 Reasons For Storage

When asked to further consider and speculate just why they thought the NBS Sample Cards were stored, unprompted responses included that they could be used for quality assurance, re-testing, and some participants felt they could be useful for the family to find a reason for a future illness in a child. Some considered it would be possible to extract DNA from the dried blood spots. Others had not previously made this link but as they considered what they knew about DNA forensics from television shows such as CSI it was clear to them this was indeed a possibility.

3.5 Using NBS Sample Cards

3.5.1 'Family Benefit'

The term 'family benefit', as suggested in the stimulus materials, was not immediately clear to participants although they did have several ideas as to how the NBS Sample

Cards might be used in family situations. These included using the sample in the circumstances where a child had died to help identify the cause.

However most participants also perceived wider direct family benefit to storing and using the NBS Sample Cards. For example, parents might request that the NBS Sample Card be used for further tests as their child grew up. They commonly suggested that being able to access this sample would avoid having to go back to the child to collect further blood samples. There was a generally strong perception that if their child's sample was stored, it must be able to be used to provide some direct benefit to that child or the family.

A number of young women were concerned that their samples were no longer in existence and could therefore not be used in identifying them if something was to occur. They were also concerned about 'missing out' on the perceived benefits to their family following the destruction.

Two fictional scenarios were presented to illustrate some of the issues around using the NBS Sample Cards.

3.5.2 Medical Research

Use in medical research was generally viewed supportively. It was widely agreed that parents should be informed and consulted if identifying information would be provided to researchers requesting the cards. The concept of research upon anonymised samples was clearly grasped and seeking parental consent in such cases only viewed as important by a few.

A couple of individuals in different groups came up with the suggestion that the samples could be permanently anonymised at 18 years by simply cutting off the identifying information from the card and this could extend the life of this apparently valuable research resource.

When exploring what would and would not be acceptable and under what conditions, the prospect of human cloning was most commonly put forward as one example of what would be unacceptable and for others that the research might lead to discrimination by insurers or employers.

Some participants had strong views that access by pharmaceutical and biotechnology companies was unacceptable, views that were underpinned by a belief that health research and developments should not be driven by a profit principle. It was also argued that commercial companies might bias or manipulate research findings. Such suggestions prompted others to counter that advances in health would not happen without such commercial motives. The idea was often more palatable if there could be some return of benefits to the community or public health system.

Whilst participants generally agreed that there would be some research areas and situations they would find problematic, they were generally prepared to let these

decisions lie with others representing the public good such as Ethics Committees. The concept of Human Research Ethics Committees was also easily grasped and much support was expressed for these bodies.

3.6 Access by Police And Courts

Popular television programs focussing on forensics were often mentioned as a source of knowledge about use of DNA from blood samples in identifying missing and solving crimes.

Access of the NBS Sample Cards by the Police was not controversial for many participants but was a particularly sensitive issue for a few.

3.6.1 Missing Persons, Identifying Suspects

Several saw considerable potential value in the stored samples to identify missing children. This connection appeared to be linked to media stories published during the fieldwork period which documented attempts to identify a missing child using DNA testing¹⁸. A concern amongst some young women was that the recent destruction of their NBS Sample Card would prevent later identification if this was ever required.

Several also viewed positively the potential to use the cards in some manner to identify a suspect in a crime and for many it was a case of *“if you have nothing to hide what would the problem be?”*. However generally the male participants had some concerns in this area. These participants had a clear general lack of trust in the behaviour of the police. One suggestion was that NBS Sample Cards could still be accessed and used by corrupt police officers to frame an individual.

3.6.2 Memorandum of Understanding

When the current MOU arrangements were outlined to participants, this appeared reassuring. Such rules, once outlined, were generally seen as appropriate and workable. However, there was still some concern expressed among many of the male participants that even if the sample was given out for apparently legitimate reasons it might later be used for other purposes.

3.6.3 Access in Legal Cases

A scenario based around access by families and the involvement of the courts in use of the sample for paternity testing, stimulated some further discussion. In general, the view was that this was a matter that should be dealt with by the justice system. Many commented that if the court demanded access to the blood spots for testing, little resistance could be made and this was the appropriate mechanism through which requests for access to the samples in cases such as this should proceed.

3.7 Retaining NBS Sample Cards, Ownership and Records

Participants views regarding the retention period for NBS Sample Cards were probed in the reconvened sessions once they had had time to consider what, if any, period they supported.

The rationale for the current 18 year storage period was unclear and perplexed several participants. It was suggested that the current cut off was likely explained in some way by

the child reaching maturity and therefore fresh consent might need to be sought. Based on what they had heard and learnt in the group discussions most were prepared to accept the existing position.

A common question posed was why the NBS Sample Cards were not stored for longer if they had such value for further medical research. Several participants suggested that the cards be stored for longer although it was raised that this might require further consent from the individual from whom the blood sample was originally taken.

No participant suggested that currently stored cards should be destroyed prior to the current 18 year retention period. The possibility of reducing the retention period by several years for future cards was considered seriously by individuals in a couple of groups.

In the reconvened groups moderators raised the fact that there were different card retention policies in different Australian States. The variation was surprising and unsettling for many participants. They had expected that the NBS policies and issues they were discussing would be similar across Australia. That retention periods differed so significantly led them to question if there was further information available which might provide a rational explanation.

Concerns were also expressed if an individual relocated to another State it may be difficult for them to access the NBS Sample Card if ever necessary. They questioned in such cases if it was possible for the cards to be transferred between States and also which State's retention policy would apply.

3.7.1 Practicalities and Security of Storage

A common initial response to learning about the storage of the NBS Sample Cards was to query how practical this was and the space requirements. Questions were raised as to how securely access was controlled and it was suggested that no system could ever be impenetrable to abuse. A number of participants, mostly men, also commented that media coverage of inappropriate use of information by police and other government agencies, identify theft and credit card fraud raised concerns for them that may not have been considered a few years ago.

3.7.2 Ownership

Also discussed was who might actually own these cards. Would it be the parents, individuals, the laboratory, the State? Generally participants did not claim ownership of the samples - although of course they had not previously been aware of their existence - but they did view that they had some rights and should have some say over how the sample was used.

3.7.3 Electronic Records

A distinction was made by several participants between the storage of the NBS Sample Card and the records containing the results derived from testing the blood spot. They questioned whether the electronic records were also destroyed after 18 years. Some concerns were raised about the management of electronic records and several men with an IT background were concerned that such electronic data was inherently insecure. They did not feel the information we had provided answered their questions.

Several male participants suggested unprompted that the samples could be used to create a DNA database or that an illicit one might exist already and the public was unaware.

3.8 Importance of Choice

A range of views was expressed within each group when they considered the importance of being informed about and consenting to NBS but most participants viewed these as important issues. Participants generally placed emphasis upon the importance of parents being aware that the heel prick was being done and, further, having a choice.

It was evident that some individuals' thoughts and specific attitudes towards this issue were still developing and that they were liable to change. Many participants had not previously considered in detail their views regarding consent to NBS but offered and justified a clear position when first asked. When further information was provided and there was opportunity to deliberate with others these initial standpoints could shift. In particular, individual views changed when considering if NBS consent should be collected in writing and also whether mandatory screening was acceptable. Changes in views did not appear to consistently operate in a single direction.

Almost all thought that consent was an important matter and that parents should be asked or informed in some way. The areas on which their arguments focussed included:

- Parents should always be given a choice
- They wanted to be informed about what was done to their child
- Ultimately a parent has responsibility for the child and for decisions.

3.8.1 Recalling Consent

Parents noted that the period immediately after the birth of the baby was an emotional and physically tiring time and they could not be certain what information they had been offered or if any discussion had taken place. It was difficult for many to separately recall giving consent for the heel prick from the other interventions they had agreed to at this time.

Of those younger parents that did recall giving some sort of consent there was a mixture of recollections as to what this had actually involved. Parents noted their various experiences of how the topic was raised by health professionals. Most seemed satisfied with the manner with which this had been raised but a few expressed some frustration.

In some cases participants were quite clear that health professionals had presented them with a choice and given them opportunity to consider. Some reported basic discussions with health professionals regarding logistical matters such as the timing of the heel prick. A few participants reported having quite detailed conversations with health professionals about the heel prick and the NBS tests where they had opportunities to clarify issues and ask questions. Several specifically recalled that, after the birth of their baby, they had been given information and offered a choice by midwives or other health professionals. Some believed that they had "*signed something*" after the birth or at the time of admission for labour and that this would likely have included consent to NBS. Several were quite adamant that they had specifically been asked for and they had given written consent to NBS.

Those parents who did not recall giving specific consent to the heel prick test did not seem to be overly concerned. However, several participants did express some disquiet

that they were not really presented with any choice or options by health professionals. They described situations where they perceived that it was assumed they would have no objections to NBS since this was simply a routine procedure or that they were *told* “*this is going to happen now, OK?*”. These parents questioned if they had *been* “*offered any real choice*”.

3.8.2 Value of Written Consent

Many participants viewed that expressing their choice in writing had some advantages. A handful maintained the view consistently through the project that in the case of NBS this really should be in writing. Written consent was also seen to have the added advantage of making people think about NBS more before consenting. Participants felt that when someone is asked to sign something, they consider it much more carefully. It was viewed that this would result in parents being more aware about the various aspects of NBS, including which conditions are tested for, that the NBS Sample Cards are stored for 18 years, and that the samples may be used for other purposes after testing is completed.

Conversely there were a few expressed concerns that seeking written consent would raise the awareness and inappropriate anxiety of some parents who might then refuse testing. Some participants also initially felt that changing the consent procedure to written would increase the workloads of already busy health professionals. Participants suggested several possibilities which might alleviate this burden such as parents might sign directly on the NBS Sample Card or on a sticker that was then transferred onto the card.

A consistent reason proposed by the participants for the use of written consent was that such a process would give legal protection to the hospital. Participants noted that we live in an increasingly litigious society. Some saw that medical errors are inevitable and they viewed that evidence of written consent would be important in future litigation. It was suggested that written consent would provide unequivocal evidence that the parent either did or did not consent to NBS. Participants expressed concern that reliance on only noting verbal consent could leave the hospital open to a claim that they had not undertaken the tests when a parent had consented. There might also be a risk that parents could claim not to have consented to NBS where the testing had in some way led to an adverse outcome.

3.8.3 Refusing Consent

Most felt that even if they personally were very positive towards NBS others might have objections and parents had a right to make this choice. It was perceived that very few parents would refuse screening and their reasons would likely be based upon religious or cultural beliefs. Most participants felt that the views of parents who still didn't want their child screened probably needed to be respected and they could not be forced to have their children tested. Nevertheless it was also viewed that although ultimately parents should have the choice about whether their child should be screened, it should be strongly suggested that they do.

Offering the opportunity to have additional conversations with experts in the field was seen as one way of achieving this. Some felt that the multistage process outlined in the stimulus materials would deter those with initial concerns and that they would likely acquiesce and agree to NBS.

Participants supported the idea that refusal for NBS should be made by the parents in writing.

3.8.4 Mandatory Screening

Several felt that NBS should be compulsory or mandatory. In such cases, which were most clearly expressed when considering PKU screening, they were likely to express the position that parents had a responsibility to ensure their child was screened. It was felt that perhaps parents would not fully understand the medical importance of early detection or objections may be trivial. A few participants felt that NBS should be compulsory despite even religious or cultural objections: they believed that the child had a right to have the screening regardless of what the parents felt. Their view was that NBS was for the benefit of the child, not the parents, and that was the most important thing.

The differences of opinion about whether NBS should be voluntary or compulsory prompted considerable discussion. Later in the groups and with further deliberation and information, the views of many individuals appeared quite fluid and liable to change.

3.8.5 Consenting For Different Elements Of NBS

It was evident that some participants considered that need for information, choice and consent would differ dependent on the condition for which screening was being offered. The idea was floated by participants in a number of groups that there could be conditions that parents could choose to test for in NBS although this was also seen to be difficult to implement.

The view that the consent for different elements in NBS such as the testing itself, the storage and further uses of the samples could also be separated was not explored in detail but hinted at by a couple of participants. These proposed that verbal consent was sufficient for the testing, but that written consent should be required for the storage and further uses of samples.

3.9 Delivering Information about NBS

The current version of the NSW Health pamphlet 'Tests to Protect Your Baby' was shown in the discussion groups at the point the moderator first raised it. While few parents directly recalled having seen the pamphlet before, mothers of young babies were most likely to remember. Quite when parents recalled receiving the pamphlet varied. Some believed it was included in prenatal materials, others received it after birth and others a few hours before the heel prick was done.

Those parents that recalled that they had been given information were not able to recall what this had covered. As the discussions developed it was evident that much of the detail mentioned in the pamphlet was entirely new information to them.

Those participants that did not recall seeing the pamphlet suggested that they may have been given a copy but they had likely overlooked it. Other parents in the groups were adamant that they had never previously seen the pamphlet.

3.9.1 Better Communication

Once it was explained that every parent should receive a copy of the pamphlet a number of suggestions were made as to how the material might be better communicated and

delivered. Parents commonly described how they received a large volume of baby health material in the form of booklets and pamphlets and that it was not feasible to absorb it all. They also described how the period of time in hospital after the birth of their child was a quite overwhelming experience and that it was unlikely they could have realistically absorbed much new medical or technical information at this time.

Provision of information during pregnancy rather than after birth was repeatedly suggested as a more effective option. Parents were seen to be more receptive to such information during a pregnancy and it was put forward that they would have time to absorb and consider the information more fully. Later in the discussions it was also suggested that this would result in people being more aware of the storage and further uses of NBS samples.

The scenarios used in the research were cited as very effective in communicating the importance of NBS. Several participants felt that stories such as the one included about PKU would be a powerful mechanism for conveying the importance of screening to parents. Such human stories might be influential where parents had doubts or were unsure of the value of screening. A video, possibly to be shown in antenatal classes, was also suggested as suiting some people better, particularly those with low literacy levels. Some form of group discussion, perhaps in an antenatal class, was also suggested since this offered opportunities to ask questions.

Participants also mentioned that information about NBS should be included in other printed materials available to parents such as the 'Blue Book' and other general child health resources.

3.9.2 Engaging Discussion

Participants reported that they had found the initial discussions accessible and engaging. Most participants had made some attempts to raise the matters discussed with others in the intervening period between groups. Women generally found it easier than men to engage others in discussion, especially friends and family who had children. Some younger people and older men reported that the topic was difficult to raise and did not seem that relevant to their everyday lives.

It did not appear that the provided information booklet was generally used to support further external discussion. Also few participants used the notebooks provided to record their thoughts or document discussions with family or friends. Rather participants explained the topic to their friends and family face to face and in their own terms. Topics that were easier to engage others in included giving consent to NBS, testing for conditions where there was no treatment and the storage of NBS Sample Cards.

Most participants reported that those they had spoken to had similar views to themselves. However there were several cases where the participants' partners had a somewhat different view.

4. Discussion

Participants did not come to these discussions with any preconceived ideas about the topic and this enabled an assessment of unprompted, existing knowledge and awareness of NBS and the extent to which parents recalled any direct experiences. The particular strength of this study however, compared to tools such as a simple survey, is that it offers a window into participants' deeper opinions. The opportunities for extended discussion, consideration of information and deliberation over several days support a much richer exploration and understanding of community views.

While there was some vague understanding of the procedures of NBS, participants knew very little about its specific purpose and implications. There was little awareness of the conditions for which NBS was undertaken and no participant knew that the blood spots from their children, or themselves in the case of the younger participants, were currently or had been stored for 18 years. The concept of use of these samples was therefore new to the groups but all quickly understood the issues and were able to participate in the discussions.

4.1 Communication and Information

It would seem that current systems do not effectively inform and communicate to many parents some key aspects of NBS including the conditions tested for, the storage of the card and its possible further uses. Yet, whilst there was a diversity of views within and across groups it was evident that a consistent view was that parents wanted to be informed about NBS. Indeed they saw it as their right to know what was being done to their child and why and what was happening with the samples.

Although recall of NBS experiences is unlikely be entirely reliable, the clear suggestion from this study is that there is considerable variation in how NBS is communicated to parents. There is a need for further engagement with health professionals to better understand current practices in the delivery and timing of information.

Participants were keen to suggest modifications to current practices for informing parents and frequently suggested that the appropriate time to raise the topic was during the pregnancy in antenatal classes. Providing information at the time of the heel prick was considered inappropriate if this was the first time that information was offered. The current NSW Policy Directive provides a recommendation that the pamphlet is offered to parents during pregnancy although it is not clear how this is implemented by hospitals.

Regardless of the version of the pamphlet given to parents over the years, it does not appear that much of the content is well absorbed or understood by parents. Effective communication is unlikely to be achieved through printed materials alone and the role of discussion will also need to be addressed further. It is perhaps noteworthy that in this study few participants appeared to refer back to the specific printed material we offered them at the end of the first groups but relied upon their recall of our discussions when talking to others in the community about these issues.

Printed materials need to be engaging and accessible whilst still providing a sufficient level of detail for parents to make an informed choice. The use of stories to illustrate aspects of NBS were viewed positively.

It is also noteworthy that the current pamphlet in English contains additional information about several aspects of the NBS programme compared to materials available in other languages.¹⁹

4.2 Expanding NBS

These consumers all viewed current NBS testing such as for PKU in a very supportive light. This view also went beyond existing tests in NBS and there was a clear belief that if a test was available parents should be able to make decisions whether or not to access this. Many participants were also very keen to see their child screened for all possible conditions as part of NBS and for further tests to be included in the NBS programme. Participants did not perceive the presented information about higher rates of false positives, the likely need for further testing of suspect cases and the lack of medically accepted treatments to be issues which greatly influenced their positions.

Perhaps it is not surprising that such enthusiasm was shown for expansion of tests. However, it is also possible that some participants had not fully considered the realities of including further tests as part of NBS. Although they had already discussed these topics at length in the groups, it is possible that their views would continue to develop and modify further over time beyond our discussions with them.

A consistent message from other social research shows there is general public interest and general support for the development of new medical biotechnologies.²⁰ This apparent strong desire to embrace further testing as technologies become available poses some challenges and it would be difficult to meet all public expectations. The decision whether or not to include a test for a particular condition in the screening programme is complex.

Policy decisions depend upon the medical and technical possibilities within a framework of public health, social, ethical and economic factors. Criteria originally developed by the World Health Organisation²¹ are usually cited as the framework for considering health screening programmes. As NBS programmes have expanded the strict application of such principles has been questioned by some scientists.²² In particular it may not always be possible to apply some of the WHO principles in regard to having information about the natural history and treatment of some very rare conditions and abnormalities. It has been suggested that the above criteria may need to be reconsidered in the face of technical developments such as TMS and others in future. However, many genetic health professionals and policy makers would challenge the expansion of NBS to include testing for conditions which are untreatable, of late onset and where the test would only indicate susceptibility.²³ This may illustrate a gap between medical and public viewpoints.

4.3 Consent Matters

Consumers have increasing expectations that they will be informed about medical interventions and expect to have a say in key health decisions and NBS is likely to be no different. The principle of gaining verbal rather than written consent for NBS was generally accepted as valid but seen to be problematic in practice. Whilst written consent was seen to have some advantages it was usually not seen as critical if parents were properly informed. There was however significant support for parents providing written refusal for NBS.

There was also significant support for mandatory screening for conditions such as PKU given its public health benefits. However it was also recognised that some people might

have justifiable objections to this. The challenge to the development of policy concerning consent to NBS must balance this diversity of views whilst also holding to high ethical standards.

There was also a diversity of views on the issue of consent for storage and further use of the samples but it was clear that the consent process must involve specific communication about these issues.

4.4 Storage and Further Use of Samples

There would seem to be a very low level of public awareness that NBS samples are stored and indeed many people do not seem to have previously considered what happens to other biological samples they have given in health settings. Yet, despite the surprising discovery that these cards were kept, there was no evident rush to demand the cards should be destroyed because they were unaware of their existence.

However it appeared that this high level of support for storage was underpinned in part by a misconception that the NBS Sample Cards could *commonly* have direct future benefit to an individual or family which occurs only infrequently. It was evident that when participants were discussing testing as part of NBS, they were also often considering that the blood spots might be used for additional tests at a later date in a child's development.

The support for the use of the cards in medical research may be reassuring. Participants grasped distinctions between identified and non identified research and seemed satisfied with the rules and regulations as they were presented to them. This study suggests that there are currently very high levels of trust in the medical profession and regulation. There was very little questioning of the mechanisms in place to oversee research upon samples and rather participants were accepting that if an ethics committee is in place it can be trusted to act in the public good. There was little evidence of the mistrust and cynicism with scientists and institutions that had been described in some European Countries.²⁴

Critical coverage of NBS in the media is likely to impact upon community confidence and trust in a NBS programme. In this context it is noteworthy that, to date, there has been little media coverage related to NBS in NSW. A search, using an online catalogue, for recent media articles about NBS revealed almost nothing in NSW yet several stories were found from other Australian states and also from New Zealand.^{25,26}

Some health professionals in other States have suggested that the concerned media and "conspiracy theorists" should be reassured by the strict controls that already exist to protect information.²⁷ However it is also clear that it is better to consider and address at the outset issues which are likely to be sensitive for the community. If initiated early, before it is sensationalised, such debate may be easier to manage.

Support amongst participants for use of NBS Sample Cards in identifying missing persons was particularly strong. Access by the Police in such situations is defined in the current MOU¹⁴ between police and health authorities. Linked to this view were the concerns raised by young people whose samples had recently been destroyed about the loss of utility for them in this context.

There were also aspects where some public views towards police access differed from current policy. There was some support in this study for the use of NBS Samples Cards by police to identify suspects in criminal cases, yet there is no provision for such access

in the MOU. Findings from international public surveys²⁸ also suggest that many members of the public, at least in the UK, would express support for this use.

While this general support for access by the police was largely reassuring, it was also very clear that there were a number of participants who strongly objected to access by such third parties. Trust, or lack thereof, again seemed to underpin these views as well as the experiences of some of the participants, particularly men, in the use of electronic data. Concerns were also expressed about the privacy and security of the samples, the possibility of identity fraud as well as inappropriate use of data and forensic samples by the police.

It is also possible and even likely that some sections of the community whom we have not interviewed may have strong views and concerns about access to the NBS Sample Cards. This study included few people from culturally diverse backgrounds or other groups whose views may be quite different from those reported here based on their experience and values. It is worth noting that several public surveys in other countries suggest that those from certain ethnic minority backgrounds are much more likely to have concerns with the storage of biological samples, access to genetic databases by third parties and lower level of trust in health providers.^{29,28}

4.5 Engaging and Involving the Community

Community consultation and participation in the development and delivery of health services is now a common feature across Australia and internationally. NSW Health has recognised the principles of community involvement and participation in health services and recently revised the structures to support these. At the State level a Health Care Advisory Council is now *“the peak clinical and community advisory group to set future directions”* for the health system whilst at a more regional level Area Health Advisory Councils have been established to promote greater clinical and community involvement in planning local health services.⁴

Studies of community attitudes in other Australian States are beginning to meet such needs in the specific case of NBS. A community study commissioned by the Victorian Government³⁰ is nearing completion and initial reported key findings include that there is *“limited parent and health professional knowledge of current storage, access and secondary uses of newborn screening cards”*.³¹

Further, a mail survey from Western Australia with recent mothers confirms the picture that many parents do not feel well informed about NBS.³² The survey findings also highlight some of the methodological difficulties in engaging consumers and assessing their views. When asked in the survey, almost one in three mothers supported the current 2 year retention for NBS sample cards. However these mothers also noted that they did not possess adequate knowledge of the issues to consider alternative options. It is in situations such as this that there is significant value in a more deliberative approach to exploring valid community views.

Moves towards the creation of a deeper public dialogue with Australians about genetic health and medical research will need to be underpinned by a better understanding of perceptions, knowledge and attitudes and what underlies these. They also likely require some further development and testing of appropriate public involvement methodologies.

4.6 Limitations of the study

The qualitative methodology implemented is designed to collect a range of views, explore these in depth and also explore the attitudes and beliefs which underlie these. Participants were not pushed to build a consensus, compromise or make trade offs and reach a unified public position. These approaches can however have particular value in the development and testing of more specific policy options which require community involvement.

In this study discussions about NBS and associated issues of storage and further use were framed in the context of health, testing and screening generally. Essentially NBS was presented to participants in a medical context which initially highlighted its direct application in detecting rare and treatable conditions. It was on this canvas that further discussion was initiated. Many participants seem to find it difficult to move beyond the immediate perceived benefits NBS testing could offer them.

Participants views and reactions may have been rather different if the approach had been to initiate discussions directly with the storage of the NBS Samples Cards or access to the cards. It is possible that the study's approach may have masked some concerns to these issues.



5. Conclusions and Recommendations

5.1 Conclusions

This study demonstrates that members of the wider community are clearly able to engage and consider complex issues and grasp some of the key scientific, social and ethical issues raised by developments in genetic health. These participants were not specialists in health, policy or ethics yet they were able to contribute unique perspectives as consumers of genetics services. Those we spoke to embraced the sometimes challenging and complex issues, considered these carefully and were willing to share and listen to views of others. They were also willing to problem solve and make positive suggestions as to how policies and practices could be further developed. This bodes well for a more inclusive strategy in the development of public policies in the developing field of genetic health.

5.2 Recommendations

1) Understanding Current Practice

The experiences and views of health professionals involved in directly delivering NBS testing, such as midwives, needs to be actively sought and explored further. Emphasis needs to be placed on improving what is generally communicated to parents about the storage of NBS Sample Cards, the further use of samples and how consent is obtained. This will be important to better understand the barriers to effectively communicating all aspects NBS to parents.

The NSW NBS Policy Directive recommends that a note is made on the mother's medical records of the dates that the pamphlet is given out, when discussion takes place and also when the NBS testing is undertaken. One possible approach for further research could involve the examination of such existing medical records to shed more light on the implementation of the existing Directive.

2) Delivering Information

Further policy development regarding best practice in the timing and distribution of information about NBS is required. If evidence is available it may be possible to give more specific advice than the current Directive which simply states the current pamphlet be offered "during pregnancy".

Better communication of NBS will in part be dependent upon parents having multiple opportunities to hear and absorb information. The pamphlet should not be relied upon as the sole source of information parents are likely to encounter. There are also likely to be further opportunities to disseminate information about NBS to parents and the Blue Book is one obvious example. Other existing health information materials targeted at parents may be appropriate for the inclusion of some information about NBS.

3) Developing Materials

The timing and mechanism for future review and revision of the NBS pamphlet, 'Tests to Protect Your Baby', needs to be considered. This will depend in part upon progress of the Australian Government's¹² review and any outcomes from its public consultation

since this may ultimately involve significant changes to policy and practice at a national level.

Future printed information may benefit from a greater emphasis on the use of accessible and non-technical language, the use of narratives and personal stories. A mechanism needs to be developed for consumer input into the development of information material and the pilot testing of draft versions. The value of evaluative tools for consumer health information materials such as DISCERN³³ should also be considered in the writing and testing stages.

The provision of up to date printed information in languages other than English should be an integral part in the development of future materials. It is important that this is developed in consultation with the relevant communities.

4) Supporting Health Professionals

The level of current support and training for health professionals in this area needs to be considered. This includes those professionals directly involved with implementation of the NBS programme, those providing information and education as well as NBS programme staff. It is possible, and even likely, that some further educational and support initiatives will be necessary and the additional resource implications will need to be considered and addressed.

The opportunities to discuss NBS with parents during pregnancy, especially in antenatal and parenting classes, should be explored further. Current models where this already occurs need to be identified and assessed so that further advice and best practice can be more widely disseminated. Enhanced involvement of parenting educators would also have training and resource implications.

5) Communication Strategy

The apparent high level of public confidence in the NBS programme may be vulnerable and needs to be actively maintained and supported by a communications strategy. The wider community may in the future hear more about NBS and retention of NBS Sample Cards through the media and in some cases it is possible that partial or inaccurate information may lead to concerns. It is important that the programme is able to respond rapidly and consistently to media queries. One option, if it does not already exist, would be to prepare some Questions and Answers which can be used as a reference for media responses. These should consider the NBS programme's responses to some of the concerns raised in this research as well as those that have been raised in other Australian States.

The communication strategy may also need to consider how the NBS programme would rapidly respond to other adverse events which although considered unlikely could seriously threaten public confidence in the programme.

Aside from being in responsive mode to the media there are opportunities to take a more active role which sets a positive agenda. This should attempt to also highlight the value of storing the NBS Sample Cards. Such coverage could increase community awareness of the importance of the programme.

6) Public Confidence and Transparency

In order to maintain public confidence and meet future challenges it is likely that certain areas of policy and practice may require further public explanation or justification.

One area which would benefit from a more precise but succinct explanation is the further use of stored NBS Sample Cards. In particular there is a need to put data in the public domain which outlines how the cards have been previously used in medical research. There may be some individual published research reports where NSW samples have been used and if so these could be summarised and used in wider communications. There are also likely to be cases where there have been no published findings from HREC approved research on NSW samples and, in some manner, these also need to be explained or summarised. One option would be to develop a review document which collated data illustrating the uses of NBS Sample Cards in medical research.

The existing MOU with NSW Police does require that details of requests are forwarded to the NSW Privacy Commissioner although to date this information has not been made public. It would be of benefit in maintaining public confidence for at least some information about MOU requests to be in the wider public domain.

7) Community Involvement

There is also a need to continue to incorporate community perspectives into further programme and policy developments. This can in part be achieved through community representation in on-going advisory and policy structures and through regular engagement with stakeholders and interested parties.

Community perspectives also have a role when considering specific policy issues such as expansion of the programme to include further tests. These consultations would benefit from a deliberative approach such as the one we have begun to develop in this study. However there remains a need to further develop community consultation models which promote interactions between policy makers, scientists and the wider community.



6. Appendices

6.1 Topic Guide - Initial Groups

Health (5 min)

Awareness, knowledge, attitudes

Key associations

Determinants of good health

What has changed in last 20 years in understanding health?

Any specific personal beliefs that underpin these viewpoints?

Testing, Screening and Health (5 min)

Lay out Showcards – “Testing” and “Screening”

Awareness, experiences, perceptions, purpose, uses

Any differences between testing and screening?

New Born Screening (10 min)

Perceptions, awareness, level of knowledge

Showcard - “Newborn Screening “

Awareness of “Guthrie card” – explore

Direct experiences of NBS

Perceived benefits of NBS, risks?

What questions does this raise?

What further information is sought?

Any recollection of NBS leaflet?

If direct experience explore circumstances booklet provided.

Any additional discussion with health professionals, partner, further information seeking?

Consent (5 min)

What consent, if any, should be sought?

Why? What is the perceived purpose of consent?

Awareness of current consent procedures

Showcard: “Consent”

Clarify current guidelines for informing parents and verbal consent

What other questions does consent raise?

Newborn Screening tests (30 min)

Present Jenny’s Story from booklet - PKU testing

Is this test acceptable, perceived benefits, any risks?

Any aspects new or different to present understandings?

Does this information change any earlier comments made?

Present Colin’s Story from booklet – DMD testing

Is this test acceptable, perceived benefits, any risks?

Any aspects new or different to present understandings?

Does this information change any earlier comments made?

Break (5 min)

What questions are in people’s minds?

Biological Samples and Health (5min)

Awareness, perceptions, uses

Add showcard “ biological sample”, “ blood sample” , “tissue sample”
Explore relationships between terms and to “testing” and “screening”
Perceived benefits of using biological samples
What happens to them after use – storage?
Experiences
Any concerns?

Storage (5 min)

Awareness that sample is stored (unlikely)
Showcard – “Storage”
Perceived purposes of storing samples

Further uses of samples (15 minutes)

Showcard “ possible uses of samples”
Awareness and perceptions of current medical research use – inc DNA research
prompting if necessary
Present Mike and Rebecca’s Story – medical research using NBS spots
Perceived benefits and risks raised
Clarify and explore linked vs anonymous research
Should parents be informed, consent for research sought ?
Present John’s Story – non medical uses of samples
Is this an appropriate use? Why?
Who should be able to access samples
Should samples be destroyed

Preparing for reconvened group (5 min)

Using the booklet
Use centrespread image in booklet to check understanding and any outstanding queries
Discussing issues with others – ideas they may have

6.2 Topic Guide - Reconvened Groups

Review

Review experiences, deliberations since last group

How views have developed , changed

Focus on stories 3 and 4

Identify and note key issues

Individually, participants note issues they see as most important , both +ve and –ve (keep these notes)

Attempt to group issues

Which are most important groups of issues? Why?

Any additional information still required?

Explore issues raised above noting examples below which are guide. Not all issues may be covered and also explore additional areas if raised.

e.g. Testing

What should be tested for?

Age of onset?

Susceptibility to cancer or depression?

What constitutes treatment?

e.g. Consent

How and when information should be provided?

Should consent be sought in writing?

Should tests ever be compulsory?

What is perceived impact of changing consent model?

One consent for testing, storage, other uses?

e.g. Storage

Acceptability of storage – should they be stored?

Security of storage

Period of storage

Destruction of samples

Consent for storage

e.g. Access to and use of samples

Attitudes towards use in medical research – what is more or less appropriate? What are the boundaries?

Comparing requirements for identified research, anonymised research

When consent for research is sought how should this be done?

What about commercial involvement in research?

Police – just what are the concerns, if any and what underlies these

Who should NOT have access to samples – prompt : Insurance companies, employers (noting that access prohibited already)

Regulation and Trust (all groups)

Need for regulation of health and genetic science

Difference between personal attitudes and what we would permit others to do?

Particular areas where regulation required?

Compare to regulation in other areas, esp privacy?

What type of regulation is appropriate e.g, Legislation, policies, codes of conduct?
Local, State, national, international?
Prompt rules and laws different in different parts of Australia? Response?
Disadvantages to regulation?
Trust in regulation
Who should be included, consulted?
Does it matter what the community thinks?

Explore any other topics as raised

6.3 Stimulus Materials – Showcards

Newborn Screening

- ◆ Newborn Screening is a public health funded system for testing newborn babies' blood for about 30 rare conditions that can cause physical and/or intellectual problems if not treated promptly.
- ◆ A few days after the baby is born, a blood sample is taken by a heel-prick and collected on a newborn screening card. The blood is dried on the card and sent to the laboratory.
- ◆ A series of tests is performed that aim to detect a number of rare genetic or metabolic conditions. For 1% of cards, a DNA test is required to clarify the result.
- ◆ Each year newborn screening finds about 90 NSW babies who have one of these conditions and most of these benefit from treatment.

Consent

- ◆ Parents need to give consent for newborn screening to be carried out.
- ◆ Parents should be given a leaflet and the matter discussed with a member of hospital staff.
- ◆ The parent's agreement to screening is noted on the medical records.
- ◆ Parents must confirm in writing if they refuse screening for their child.

Storage of Cards

- ◆ After testing the blood spot cards are securely stored at a hospital.
- ◆ The cards are kept for 18 years in New South Wales and then destroyed.

Possible Uses of Sample

- ◆ Quality control and development of new tests
- ◆ Family
- ◆ Police and courts
- ◆ Medical Research

Further use of cards

- ◆ Special laws and rules control who can access the stored samples and how they can be used.
- ◆ Medical research on the blood spots must be approved by an Ethics Committee.
- ◆ Parents must also be asked for their consent for any medical research if it could be linked to the identity of their child.

6.4 Stimulus Materials – Booklet and Scenarios

Note: includes the four scenarios presented in initial groups

The specific stories about families and individuals described in this booklet are fictional.

What's Next ?

- Think some more about the issues raised in your discussion group and in this booklet.
- You may want to discuss some of these issues with others in your family, partners, friends or colleagues at work.
- Try to keep some notes of the questions, thoughts and ideas you have in the journal provided.
- Come along for the second group meeting.

More information

We cannot cover all the questions you may have in a short booklet but we have tried to give you enough information to start thinking about some of them.

There are some further web links to other sources of information on the Centre for Genetics Education website which you may also find useful. www.genetics.com/testingquestions

We are looking forward to seeing you at the second discussion. If you need to contact us please phone Luke on (02) 9926 7307.

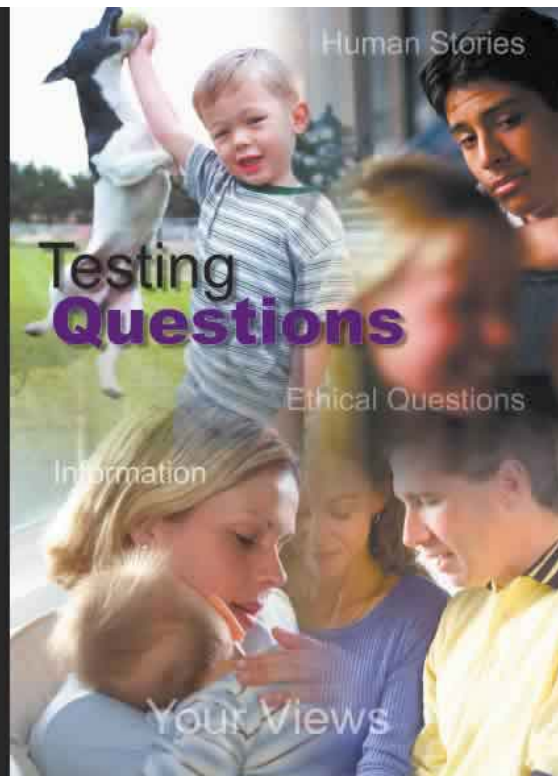
As a thank-you, you will be given a shopping voucher at the second meeting.

This material has been prepared by the Centre for Genetics Education with the assistance of a medical journalist and in consultation with experts in newborn screening, ethics and public health.

Your next group discussion is...

Date: _____ Time: _____

Location: _____



What is Newborn Screening?

- Newborn Screening is a public health funded system for testing newborn babies' blood for about 30 rare conditions that can cause physical and/or intellectual problems if not treated promptly.
- A few days after the baby is born, a blood sample is taken by a heel-prick and collected on a newborn screening card. The blood is dried on the card and sent to the laboratory.
- A series of tests is performed that aim to detect a number of rare genetic or metabolic conditions. For 1% of cards, a DNA test is required to clarify the result.
- Each year newborn screening finds about 90 NSW babies who have one of these conditions and most of these benefit from treatment.

Jenny's story

In NSW, about 10 babies each year are found to have PKU.

Baby Nicola looked healthy when she was born, so Jenny was shocked to hear her baby was at risk of growing up with brain damage if she didn't eat a special diet.

Jenny knew nothing about phenylketonuria (PKU) but it is one of the most important diseases that newborn screening detects because treatment makes such a difference to the lives of these babies as they grow up.

Now Jenny has seen the doctors and other specialists, she feels she can cope with the special diet Nicola will need to follow to protect her brain from permanent damage.

On the special PKU diet, Nicola's brain will develop normally and she will have the chance to do well at school and even university.

Because early treatment is so important, PKU testing is offered to all newborn babies in Australia as part of the newborn heel-prick tests. Over 99% of parents agree to such newborn screening tests.

PKU testing has been available since 1968. Since then new technologies have allowed additional conditions to be tested for from the blood spots.



John and Danni's Story

Last year John and his mother died in a car accident. John was 17 years old. Six months later, Danni, a girl he knew from his school has a baby. She says that John was the baby's father. John's remaining family do not believe that he is the father of Danni's baby.

Danni knows that John's blood spot is still stored and wants to use it for paternity tests to prove that he really was the father.

John's remaining family, including his own father, will not agree to the card being used for paternity testing. They say Danni has no right to have the sample and it should not be given out without their permission. John's family want the blood spot card destroyed.

If it is proved that John is the father, then the child could claim financial support from John's family and may stand to inherit money in the future. John came from a wealthy family.

The newborn screening laboratory will not give the card to Danni without a court order. They normally destroy cards collected for newborn screening after 18 years.



What do you think?

- Do you think the blood spots should be stored?
- How long should the cards be stored?
- Who should be able to access the cards?
- When should parents be asked for their permission?
- Should the cards be destroyed or returned if parents ask?

Mike and Rebecca's Story

Mike and Rebecca were heartbroken when their son died when he was 5 years old.

Medical researchers have now asked for their permission to use the blood spot taken just after their son was born. They had forgotten or not realised that the blood spot card was being stored.

The hospital said the blood spots are stored in a secure place. The blood spots are kept for 18 years in New South Wales and are then destroyed.

Special privacy laws and rules control the way newborn blood spots can be used. The rules say that if medical researchers want to use the stored blood spots where any identifying information is also given to them then they must get written permission from the parents.



In research where no information is given out that could identify the person then the rules do not require parents to be asked.

In all cases, researchers will also need to get the approval of a local Ethics Committee before they can go ahead.

Colin's Story

In NSW, 1 in 5000 boys is born with DMD.

Colin's parents live in Canada. They were devastated when their doctor said the newborn blood test showed Colin might have a muscle wasting disease called Duchenne Muscular Dystrophy (DMD).

The doctor said Colin had to have another test to be sure as nine times out of ten the baby will not have DMD. But, the second test showed that Colin really did have DMD.

There are no treatments for DMD and early detection is unlikely to directly affect Colin's health.

By their teenage years, boys with DMD are likely to need a wheelchair and most die by their 20s.

His parents have now joined a support group of families in similar situations. They hope this will help them know what to expect as Colin gets older.

If they want to have another baby they can have tests to check whether the next pregnancy is also affected.

Only a few countries like Canada and Wales test babies for DMD at birth. DMD is not currently tested for during newborn screening in New South



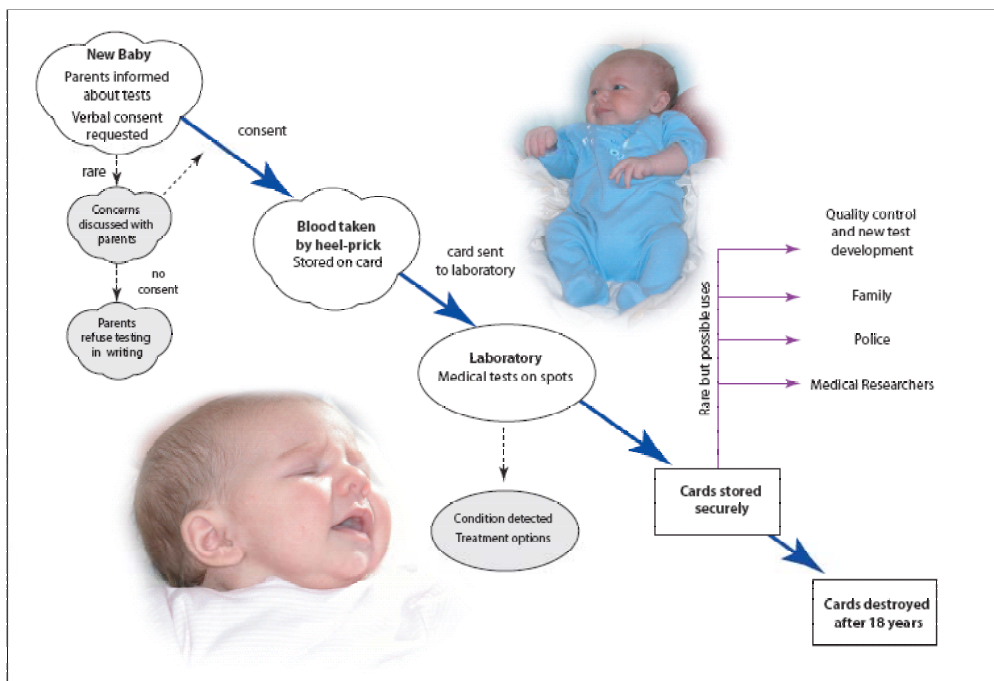
Wales. Children with the condition will usually be picked up when symptoms appear in the first five years.

Storage and Further Use of Cards

- ❖ After testing the blood spot cards are securely stored at a hospital.
- ❖ The cards are kept for 18 years in New South Wales and then destroyed.
- ❖ Special laws and rules control who can access the stored samples and how they can be used.
- ❖ Medical research on the blood spots must be approved by an Ethics Committee.
- ❖ Parents must also be asked for consent if the research could be linked to the identity of their child.

What do you think?

- ❖ What do you think are benefits of testing babies for such diseases?
- ❖ Do you see any risks or have concerns?
- ❖ How should parents be asked if they agree to testing?
- ❖ Should such testing ever be compulsory?



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