



OVERVIEW OF DOCUMENTS RELATED TO NEWBORN SCREENING & INFORMED CONSENT

The documents fall into six broad categories:

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

MEDIA REPORTS RELATED TO NEWBORN SCREENING ISSUES

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

MEDICO-LEGAL ISSUES

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

An overview of the ALRC's discussion paper published in the newsletter of the Victorian Privacy Commissioner discusses the collection and storage of DNA in newborn screening

samples highlighting concerns about the quality of the consent that parents give for their storage and secondary use (Anon 2003).

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

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

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

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

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

Guidelines and policies for newborn screening

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

The joint policies of the Human Genetics Society of Australasia (HGSA) and the Royal Australasian College of Physicians (RACP) for newborn screening and the retention and storage of samples form the basis of the policies in Victoria and New South Wales. Specifically the HGSA-RACP policy recommends universal screening for three disorders (PKU, CH and CF), voluntary participation, public funding and appropriate follow-up

(HGSA undated). Additional reasons for the use of retained samples include the need for samples to maintain quality assurance in existing testing procedures and for the modification of existing tests, as well as for the development of new tests and the investigation of missed cases (HGSA 2004). In the event of refusal HGSA-RACP recommend that parents be required to sign a written statement confirming they have been informed and understand the consequences of not testing. The HGSA policy statements do not include guidance on how and when the procedure of heel prick screening should be conducted.

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

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

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

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

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

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

The policy approach in the United Kingdom to newborn screening has been thorough. It includes the development of policies and standards for all aspects of newborn screening. The UK documents include the consultation paper, the approved policies and standards, communication guidelines for discussion with parents, a health professional handbook and an information pamphlet for parents (UK NSPC a & b 2004, UK NSPC a & b 2005). The standards are to be used to monitor quality and performance of the screening program and cover the timely collection and dispatch of samples, universal offering of testing, capacity to track samples and identify babies who do not provide samples or have positive screening results.

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

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

Verbal v written consent

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

Health research related to communication issues around newborn screening

The only primary research included in the documents is a report of a descriptive cross-sectional survey of women's knowledge of genetic screening within 24 hours of their babies undergoing a heel prick test in Melbourne, Victoria (Suraidi 2004). The study involved 200 interviews with ethnically diverse women using interpreters where necessary. Two hundred and thirty-two women who delivered liveborn babies consecutively at a tertiary hospital and who had been offered prenatal screening for Down Syndrome were approached to take part in the study. Participants were comparable with other women giving birth in Victoria in terms of age, marital status, parity and whether they held religious beliefs, although Arabic and Turkish women were over-represented, as were women of Islamic faith. The authors do not report the year that the study was undertaken. Women had limited knowledge about the terms used in counselling for Down Syndrome and newborn genetic disease. The median score was 4 points (out of maximum knowledge score was 15) (IQR=2-8). While 72 percent of women indicated that they had heard of the term 'genetic disease' only 43.5 percent were able to accurately explain the term. Sixty-two percent had heard the term 'genetic screening' but it could be explained adequately by only 30 percent and 37 percent had heard of 'Guthrie', 'heel prick test' or 'newborn screening' with only 3.5 percent able to describe its purpose. Only 26.5 percent of women said that they knew that baby had undergone the heel-prick test whereas it had not been done for only four babies (2

percent). The mothers of these babies had specifically refused the test. Two women refused because of the distress of a previous false positive result, one because of the distress it caused her older child, and one because it resulted in a heel infection in an older child.

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

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

Information for parents about newborn screening

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

The fact sheet prepared by the Centre for Genetic Education appears to be based on the HGSA policy statement. It includes mention of homebirth and unlike the information from the states says that no further tests will be done on samples without written consent. The information from the Better Health Channel website explains that a positive test result does not mean that a baby is affected by the disorder.

The parent information pamphlet from the UK covers all issues raised in other documents as well as advising that carrier status will be detected.

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

SUMMARY

The research undertaken by Suraidi *et al* demonstrates that there is a low level of knowledge among women who have recently given birth about genetic screening. It also reveals that women will refuse newborn screening for issues that are not addressed in

the information given to parents (false positive results and the distress and harm that may result from the procedure itself). These findings support concerns about whether informed consent processes around newborn screening would meet the test of law. The review of studies of parents' views of the communication of positive test results by Oliver *et al* (2004) supports familiar non-specialists in this role. This requires health professionals to be educated about the complex issues around newborn screening. Other issues to be resolved are: disclosure of carrier status, reconciliation of differences in international practices about when it is best to conduct the test, and the need for written consent.

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