

# Policy Statement on the Retention, Storage and Use of Sample Cards from Newborn Screening Programs

The following Policy Statement has been developed by a joint subcommittee of the Human Genetics Society of Australasia and the Division of Paediatrics of the Royal Australasian College of Physicians. The policy provides guidance to Newborn Screening Programs on issues that relate to sample cards after screening tests are completed. The policy includes the following:

- Background
- General recommendations
- Ownership and retention issues
- Storage and release issues
- Quality assurance issues
- Privacy, legal and ethical issues
- Other dried blood samples
- References

The policy will be reviewed regularly to consider scientific information and debate on the value of retaining the sample cards and their role in public health research and genetic testing. General operating guidelines for newborn screening programs are contained in a separate HGSA policy<sup>1</sup>.

## 1 Background

Newborn screening to detect congenital metabolic disorders is an accepted part of neonatal care in all developed countries and has been established in New Zealand and Australia since the late 1960's. The sample cards (Guthrie cards) are special absorbent paper on which blood samples are collected from all newborns. The provenance of the samples cannot be guaranteed by the screening program. Along with the dried blood samples the cards and/or associated forms also contain information about the baby (and often mother) for identification (name of baby and mother, baby's sex, date of birth) as well as sample details, hospital of birth, the referring health care giver. There is also variable other information, such as birth weight and mode of feeding, specifically related to interpretation of the test results. The dried blood samples are tested for a number of congenital biochemical abnormalities. Following completion of testing the

screening programs retain the sample cards, for varying times, primarily for screening program audit. When cases are missed by the screening program, retention of sample cards enables a check for proof of the existence of a sample and reconfirmation of initial test results. In addition, sample cards may also be used for:

- confirmation of laboratory normal ranges (using anonymised samples)
- modification of existing screening tests (using either anonymised or identified samples)
- development of new screening tests (using either anonymised or identified samples)
- epidemiological or public health research (using anonymised samples)
- testing of deceased members of a family if a specific disorder is suspected or known (using identified samples)
- assisting in coronial and forensic investigations (using identified samples).

When anonymised samples are used, the sample identification does not contain the baby name or original sample number. The samples may be tested without any identification, or a new sample identification may be created and the code linking the new and old identifications destroyed. However, other information necessary for interpretation of test results may be retained linked to the new sample identity.

Newborn screening cards are seldom mentioned in policies relating to retention of laboratory specimens. The National Pathology Accreditation Advisory Council (NPAAC) recommended that the sample cards be stored for 50 years although no justification or explanation was given for the recommendation.<sup>1</sup> The Royal College of Pathologists recommended storage for 20 years advising due care that no deterioration should occur and that records should be retained to prove the existence of a sample.<sup>2</sup> In Australia, various State Public Health Acts cover retention of laboratory samples and information, eg NSW legislation states records for minors should be retained for a minimum of 15 years after the child reaches the age of 18<sup>3</sup>.

Retention practices vary internationally. Some countries, for example Denmark, store cards indefinitely for screening program audit and for future research projects.<sup>4</sup> Cards are destroyed soon after completion of testing in France<sup>5</sup> and by most programs in the United States of America.

Recently, the Council of Regional Networks for Genetic Services (CORN) in the USA produced comprehensive guidelines for those laboratories developing policies to retain their sample cards.<sup>6</sup> Currently, the six screening programs in New Zealand and Australia store their sample cards for times varying from two years to indefinitely.

In Australia, it has been suggested that sample cards are owned by the hospital or laboratory that prepared them, like other hospital records.<sup>7</sup> They may be owned by the laboratory which analyses and stores them. In New Zealand, in practice, samples belong to the person from whom they were collected but in the case of a deceased person, ownership is with the testing laboratory.

In both Australia and New Zealand, a principle of "informed refusal or dissent" has applied to sample card collection rather than a requirement for informed consent.<sup>7</sup> Testing for purposes outside those originally specified requires further informed consent, testing of unlinked (anonymous) samples (in some approved cases) or an intermediate course agreed by local ethical and regulatory bodies.

The possibility of misuse of the stored cards by insurers, employers and other third parties gives rise to issues which threaten privacy.<sup>8</sup> The major public health value of the screening programs must not be jeopardised by concerns over misuse of the samples.

## **2 General recommendations**

- 2.1 Newborn Screening Programs should have a policy regarding the retention, storage and use of their sample cards. The policy should address each of the issues outlined in this document.
- 2.2 Development of this policy should consider local customs and laws.
- 2.3 The public should be informed about the policy in general terms, and that further information on screening program storage policies and procedures is available to them on request.
- 2.4 The policy governing retention, storage and use should be reviewed regularly.

### **3 Ownership and retention issues**

- 3.1 Ownership of residual sample cards. The screening program should document, as far as possible, who is considered to own the cards. Policies regarding further use should consider ownership issues.
  
- 3.2 Retention of sample cards. The screening program should document how long it will retain their cards, including the purposes for retention. The information to parents should state how long the cards will be retained and what additional uses may be made of the cards.

### **4 Storage and release issues**

- 4.1 Storage of sample cards. Following completion of the newborn screening analysis, the sample cards should be stored in a manner appropriate to the intended future uses of the cards.
- 4.1.1 In general, this would involve security of access, indexed storage to allow retrieval, and consideration of the storage conditions to prevent undue deterioration of the sample.
  - 4.1.2 Appropriate control cards, such as those taken from the HGSA Newborn Screening Quality Assurance Program, should be included with the sample cards.
  - 4.1.3 Only authorised representatives of the Newborn Screening Program should have access to the storage facility and sample cards.
  - 4.1.4 It may be appropriate to record the names of persons accessing the storage, the date and time at which access occurs and the reasons for access.
- 4.2 Release of sample cards. All releases of residual samples should be documented. Such records should include the purpose for release, what material was released, by whom it was used, and the authority for use.
- 4.3 Use of sample cards. Specific uses of residual samples include, but are not limited to, the following. Each program policy should state what permission and documentation are required in each situation-:
- 4.3.1 Investigation of cases missed by the screening program. This is the primary purpose of retention of screening samples. Confirmation of test results and any related testing may be done in the program laboratory, or may be sent to

another laboratory with suitable methodology. This must be done on any reported missed case.

4.3.2 Screening program development, method development, and establishing normal ranges for new or existing tests. Within the screening program these uses do not require consent when the samples are unlinked (anonymised). Institutional ethics committee approval is required if samples are to be used outside the screening laboratory and/or remain identified and if new information about the health status of the person from whom the sample was collected will be obtained. Such approval should be specific about who will be informed of any abnormal test results, what they will be told, and what follow-up will be necessary.

4.3.3 Individual requests. Programs must develop policies to deal with requests for cards to be returned to the family. Information about test results should be treated in the same way as requests for other medical information within the institution. Release should not be made to persons other than the person whose blood is on the card, or if that person is a minor, the child's parents. Release should follow a written request, and should be discouraged if there is a living person from whom an alternative system can obtain a sample. If cards are returned to the family, the family should be encouraged to retain the card/s and to store it/them in an appropriate manner (taking cognisance of the safety requirements of pathology sample storage). An alternative to returning cards to families is an agreement

with the family to destroy the card after an agreed period of time.

- 4.3.4 Requests from Health Professionals. Any request from a health professional to release a card from a deceased person to determine the cause of death or to gain genetic information for family reasons must be accompanied by a request from a custodial parent or next-of-kin.
- 4.3.5 Research Studies. Requests to use the sample cards in research studies are permissible if the researcher has appropriate approval from a local ethics committee and the screening program advisory committee (where such exists). The research should be performed under conditions established by the screening program advisory committee and the ethics committee and conform to NH&MRC guidelines.<sup>9</sup>
- 4.3.6 Coronial and forensic. Material should be released from dead or missing persons for coronial or forensic investigations with parental or next-of-kin permission. In exceptional circumstances, if legal authorities have a reason for not seeking parental permission an appropriate legal permission (search warrant, court order) should be obtained. Ethical and legal issues surrounding stored human material must be considered in approval of such uses and may include notification to the parents that the sample has been provided. Sample cards should not be released from a person who is neither dead nor missing as alternative systems exist for obtaining new samples.

## **5 Quality assurance issues**

- 5.1 Quality assurance systems must be designed to ensure that stored samples can be retrieved, and that on retrieval, samples will be suitable for their intended purposes. This requires clearly defined pathways for collection, retention, storage, security and retrieval.
- 5.2 Samples with known concentrations of the analytes tested should be stored with the sample cards. Storage of the HGSA Newborn Screening Quality Assurance Program samples with the sample cards meets this requirement.

## **6 Privacy, legal and ethical issues**

- 6.1 The screening program must ensure that the sample cards and associated information are stored in a secure manner so that unauthorised access or use of the cards is prevented.
- 6.2 Laws, customs and systems governing ethical and privacy principles will vary between jurisdictions and screening programs. The program should be cognisant of these and acknowledge them in policy development. Programs should be aware of the Privacy Acts (Australia 1988, New Zealand 1993) and other specific policies<sup>9-12</sup>.

## **7 Other dried blood samples**

Stored dried whole blood on filter paper has also been considered by researchers or public authorities for the creation of DNA banks.<sup>6</sup> Such samples may be desirable for economic reasons and permit safe storage over long periods, for example in the case of late onset hereditary diseases.

The principles and issues outlined in this policy apply equally in such situations. Reference may also be made to the NH&MRC Guidelines for Genetic Registers and Associated Genetic Material.<sup>13</sup>

## 8 References

1. HGSA. Newborn Screening Policy. Sydney: Human Genetics Society of Australasia, 1999.
2. RCPATH. The Retention and Storage of Pathological Records and Archives. Report of the Working Party of the Royal College of Pathologists and the Institute of Biomedical Science. London: Royal College of Pathologists, 1999.
3. NSW State Records Act. NSW Department of Health General Disposal Authority 5: Patient Client Records. Sydney: NSW State Government, 1998.
4. Almind G, Neilsen L, Strandberg Petersen N, Riis P. Health Science Information Banks. Bio Banks. Vol. ISBN 87-90343-06-9. Copenhagen: The Danish Council of Ethics, 1996.
5. Association française pour le dépistage et la prévention des handicaps de l'enfant. Comité d'éthique. Les prélèvements de sang sur papier pour le dépistage neonatal. Recommandations pour leur collecte, leur traitement et leur conservation. Arch Pediatr 1995; 2:3-7.
6. Therrell BL, Hannon WH, Pass KA, et al. Guidelines for the Retention, Storage, and Use of Residual Dried Blood Spot Samples after Newborn Screening Analysis: Statement of the Council of Regional Networks of Genetic Services. Biochem Molec Med 1996; 57:116-124.
7. Skene L. Access to and Ownership of Blood Samples for Genetic Tests: Guthrie Spots. Journal of Law and Medicine 1997; 5:137-139.
8. Section 63. Health Services Act. Canberra: Australian Federal Government, 1991.
9. NH&MRC. National Statement on Ethical Conduct in Research Involving Humans. Canberra: National Health and Medical Research Council, 1999.
10. NH&MRC. Draft Guidance on Ethical Aspects of Human Genetic Testing. Canberra: National Health and Medical Research Council, 1999.
11. NH&MRC. Draft Guidelines for the protection of privacy in the conduct of medical research. Canberra: National Health and Medical Research Council, 1999.
12. Personal privacy protection in health care information systems. Australian Standard AS4400. Canberra, 1995.
13. NH&MRC. Guidelines for Genetic Registers and Associated Genetic material. Canberra: National

Health and Medical Research Council, 1999.