

# Retention of Guthrie cards: reassuring parents

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The newborn screening program is a well-regarded standard of care for newborn infants. Although restricted later use of retained Guthrie cards, which form an inadvertent DNA sample bank, has the potential for enormous personal and community benefit, access is rarely granted and strict controls are in place to protect the information that may be derived from retesting the samples.

New parents who are approached to consent to a newborn screening test on their baby may be alarmed by recent publicity regarding privacy issues concerning Guthrie card blood spots. In Victoria, these blood spots have been collected since the commencement of the newborn screening program in the late 1960s and the Guthrie cards are stored indefinitely. In Western Australia, the State's Guthrie cards stored from 1990 to 1995 were destroyed in 1998 and are now only retained for two years. This decision was apparently made after the Western Australia Police Service sought access to stored newborn



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blood sample cards for one of its investigations. Although there is widespread community support for newborn screening, some people may now be concerned about the privacy implications, especially since more information may be revealed as potential genetic tests become increasingly sophisticated and cards are kept for later reference for the family. This article aims to clarify the facts and thereby assuage these concerns.

## Purpose of newborn screening

Newborn screening performed on blood obtained by a heel prick from infants soon after birth aims to detect conditions that can be treated before a child becomes sick or dies.<sup>1</sup> The conditions for which tests are conducted vary between jurisdictions. They are all serious and sometimes fatal, but can be treated or palliated by early intervention. Phenylketonuria (PKU), hypothyroidism and cystic fibrosis are commonly included. An expanded testing using the technique of tandem mass spectrometry (TMS) screens for many other metabolic conditions.

The advantage to the baby is obvious. The 'invasion' of the test is a jab to the heel with brief pain as a small quantity of blood is obtained. However, a baby found to have a serious condition that threatens his or her life or health can be given immediate treatment and avoid the worst consequences of the condition. When parents understand this, they almost always

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consent to the test. Thus, although the test is not compulsory, the informed refusal rate is very low.

### **Guthrie cards**

The information printed on Guthrie cards is limited. In Victoria, it consists of the mother's name, the name of the hospital in which the baby was born, the sex of the baby and the baby's date of birth. The baby's name is not on the card and there are no test results or medical information recorded on the card.

After the blood on each card is tested, the cards are stored at an offsite secure facility in sealed boxes. Cards are not stored alphabetically, but by month and year of birth.

Access to the Guthrie cards is restricted and closely self-regulated by Genetic Health Services Victoria. People who want to gain access to a stored Guthrie card cannot do so directly. Access to the facility where the cards are stored is limited to specially authorised personnel and then only under strict criteria of quality assurance and research approved by an independent hospital ethics committee. The authorised personnel must use a fingerpad code to enter the facility. As there is limited information on the cards, finding the card of a particular baby requires knowing when the baby was born, the name of the hospital and the mother's name – as mentioned before, the baby's name is not on the card. This information is stored at a physically separate site.

### **Why are Guthrie cards kept?**

The National Pathology Accreditation Advisory Council (NPAAC) recommendations and the Standards issued by the Keeper of Public Records require Guthrie cards to be retained for a minimum of 25 years. Currently the cards are retained indefinitely in Victoria and South Australia, 25 years in Queensland and 18 years in NSW. Initially the blood spots were retained for quality assurance purposes but with developments in genetic tests, there are greater potential uses that render the cards an invaluable asset to the community. Although cards are kept securely and contain very limited information, some people are worried about them being retained at all because of concerns for potential and future misuse of the blood spots. However, there are several important reasons for keeping the cards and any concerns would be best addressed by stringent regulation of their storage and access.

### **Quality assurance**

Having cards available for retesting is vital for quality assurance. If there is a suggestion later that a baby's condition was not diagnosed by newborn screening (for example, because the child develops the condition), the baby's card can be retrieved and retested, together with the cards of all other babies tested that day to ensure that other cases have not been overlooked. If

there was a fault in the diagnostic process, it will be identified and corrected, and the babies affected can be offered new tests.

Also, earlier samples can be retested for accuracy of diagnosis if a new test is later developed and believed to be more effective. Samples can also be retested with other equipment.

### **Retrospective testing**

Retrospective testing can be done after a baby has died. For example, a woman planning a later pregnancy may want to know whether her dead child had a genetic condition for which no test was then available or was not undertaken during the child's life. Indeed, she may want this information to help her blood relatives even if she is not planning another pregnancy herself. Such information is critical for clarifying the risk of recurrence and being able to provide the option of prenatal diagnosis to family members who choose to avoid having a child with the identified genetic condition.

### **Identification of remains**

Guthrie cards may be the only sure way to identify people when bodies are found in incinerated buildings or in other circumstances in which identity cannot be confirmed (many of the dead in the Bali bombings were identified in this way). Such identification is vital for grieving relatives and the 'closure' it brings.

Guthrie cards may also be important in identifying crime victims. Blood spots are not used to identify alleged offenders. If a suspected offender is alive, a sample can be obtained from that person directly by obtaining a court order, although a judge or magistrate would not make such an order unless satisfied that there was reason to believe that an offence has been committed. (The Crimes Act 1958 [Vic] ss 464R-464U sets out the circumstances in which nonconsensual taking of blood and other bodily substances may be taken for forensic tests.) Victoria Police has signed a Memorandum of Understanding with Genetic Health Services Victoria (GHSV), who hold all Guthrie cards in Victoria, that they will not seize any card without first obtaining a court order.

### **Research**

Researchers may seek access to samples of blood from Guthrie cards for use in research. This will only be permitted if there is prior specific approval from a hospital ethics committee. Samples will generally be anonymous: only the blood spot will be available to the researcher and not the entire Guthrie card, unless specific consent is obtained from the parents to use the samples in research in an identified manner.

In Victoria, to date, approval has been granted for only three projects. One of these was the testing of the hypothesis that it is only carriers of cystic fibrosis and not other children

who have a predisposition to intussusception when challenged with rotavirus vaccine.<sup>2</sup> Unfortunately no connection to cystic fibrosis was found, but had there been a connection, a rotavirus vaccination program for thousands of children in developing countries could have been reinstated, with huge potential benefits. The researchers needed anonymous samples from a random control group as well as from cystic fibrosis carriers to test their theory. As the researchers explain in a letter to the *Journal of Paediatric Child Health*, it was not possible – given the cost restraints of their study – for them to obtain consent from people whose samples were tested and this did not seem ethically necessary since the samples were provided without identifying details.<sup>3</sup> Another project was a study of the incidence of a gene causing deafness in a random anonymous group; the findings may have implications in planning services for deaf people.

### Who can access Guthrie cards?

Of the two million or so Guthrie cards held in Victoria, fewer than a thousand cards have been accessed. In line with the above, the people who seek access are:

- staff of the testing laboratory wanting to check the accuracy of the original test – related to a particular child or his or her family in light of later information or because a new test has been developed or as part of a wider blanket checking
- individuals seeking retrospective genetic diagnosis – Genetics Victoria has granted access to people wanting to retest their own sample
- the Coroner – to assist in identification of remains or determination of the cause of death
- researchers – after specific consent has been obtained from an ethics committee
- the police – if a specific court order has been obtained (to date in Victoria, police have obtained access only where the person has died)
- people whose sample is held, or their parents – the law on access to Guthrie cards or ownership of them is unclear:<sup>4</sup> Guthrie cards may be transferred to both parents on request in writing that their child's Guthrie card be transferred to them for safe-keeping, but this contravenes the requirement that the cards be retained; the matter needs clarification.

### Review of Guthrie card procedures

The Victorian Department of Human Services is currently undertaking a review of Guthrie card procedures. This includes an audit of hospital records to ensure that consent is being sought from parents before the test is done and an audit of storage procedures to ensure that they comply with standards

prescribed by the National Association of Testing Authorities.

There are moves to promote greater national consistency. The Australian Law Reform Commission in its report 'Essentially yours' has identified the need for clarifying issues relating to storage and access to Guthrie cards.<sup>5</sup> The National Public Health Partnership has embraced the Victorian recommendation for a national approach to Public Health Genetics and in particular to newborn screening practices.

### Conclusion

The newborn screening program is regarded internationally as a vital standard of care for newborn infants. There are good reasons for hospitals to retain Guthrie cards. Restricted later use has the potential for enormous personal and community benefit. Access is rarely granted and there are strict controls to protect the information that may be derived from retesting the samples. Various inquiries are currently under way to audit and strengthen these controls.

### Series Editor's comment

Newborn genetic screening is self-evidently of major benefit, both to the individuals tested and to the community. The test for phenylketonuria fits the definition of a perfect screening test. It is cheap to perform so that even though the condition is rare, the cost per positive detected is acceptably low. More importantly, the condition is now detected early enough for a very cheap intervention (dietary modification) to prevent what were previously the inevitable complications of the disease.

Since Dr Robert Guthrie developed, in 1963, a simple, cheap and accurate screening test for phenylketonuria, many new technologies have been developed enabling screening for many conditions using a single heel prick blood sample. Screening for 30 conditions is now possible on the one sample, using the technique of tandem mass spectrometry. These are currently biochemical tests. When 'gene chip' technology is available, it will be possible, once sufficient genetic markers are developed, to screen for hundreds of different conditions at the one time, on the one specimen of dried blood.

### Informed consent for the test

The first issue this raises is the issue of informed consent for the test itself. Despite the ready availability of information leaflets, it is doubtful that, in a busy maternity ward and with very short-stay patients, informed consent processes that would stand the test of law currently occur. Is anyone really going to sit down with an expectant mother and inform her (and the child's father) about every single one of those hundreds of conditions for which testing will be available in the future? Ensuring that parents truly understand the possible consequences of agreeing (informed consent) or not agreeing to

testing (informed refusal) is very difficult, but the law requires that an informed decision must be made about the test. A heel prick sample cannot be taken as part of routine postnatal care without an informed decision by the parent(s).

Another issue is whether parents should have a right of refusal when that refusal has the (remote) potential to cause their child preventable harm. Parents do not have an absolute right to give or refuse consent for procedures for their children. Decisions must be made in the best interests of the child. We have discussed previously in this column that it is unlawful to perform a nontherapeutic sterilisation procedure on a minor or an incompetent person without seeking court approval for the proposal, and we have also considered whether a court could authorise prenatal screening if parents refuse.<sup>6,7</sup> In each case, it is ultimately the court, not the parents or guardian, that must determine whether the proposed procedure is in the best interests of the patient.

### Informed consent for prolonged storage of cards

Although the current tests performed on Guthrie card blood spots are for metabolic conditions and cystic fibrosis, concerns have been raised regarding the potential for using Guthrie card samples for genetic testing in the future. Fortunately, once informed, almost all parents currently agree to their child having newborn testing. But look at what happened in the UK when a television program seized upon a study and reported a possible link between pertussis vaccine and encephalopathy: immunisation rates plummeted and deaths from pertussis rose. An article in *JAMA* in 1984 had extrapolated the results of a cautious English study and had suggested that pertussis vaccine might be causing 25 cases a year, in the USA, of vaccine encephalopathy.<sup>8</sup> A later study, published in *JAMA* in 1990, failed to replicate these results and an editorial in the same issue of *JAMA* said 'It is time for the myth of pertussis vaccine encephalopathy to end.'<sup>9,10</sup> How many children died of complications between 1984 and 1990 due to not being immunised? Public confidence is fragile and is easily shaken by alarmist reporting.

Anything to do with genetic testing brings the conspiracy theorists out of the woodwork, but prolonged storage of blood spots is a far cry from a population-wide DNA or genetic fingerprint data bank. Until now, only in a few very rare cases has the Guthrie card of a named individual been retrieved and the DNA extracted and compared with DNA extracted from unidentified human remains for the purpose of identifying those remains. That is a bit like what happened when fingerprinting was in its infancy. Then it was not possible to compare a fingerprint taken from a crime scene against a data bank of fingerprints, only against the fingerprint(s) of one or a small number of suspects. Now, however, fingerprints

can be electronically scanned and checked against very large databases of fingerprints. It is conceivable that over the next few years or decades similar advances in technology will allow DNA to be extracted from blood much more easily, quickly and cheaply than now, and be able to be compared against a database of genetic fingerprints. Conspiracy theorists are concerned that indefinite storage of Guthrie cards may provide the raw material from which such a genetic database could be derived.

The response to the conspiracy theorists' Big Brother concerns is the legal protections that already exist, and have been detailed in this article. If there is concern that these protections are inadequate then they should be strengthened; we should not react hastily by destroying a valuable resource.

### Conclusion

Great good has come already from newborn screening to prevent disease progression. The destruction of stored Guthrie cards in Western Australia seems a gross over-reaction to the conspiracy theorists' alarms. Doctors have an important role in reinforcing the very great benefits to the children of tomorrow of newborn screening and providing accurate information about the legal and other protections that surround these programs. MT

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