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Guthrie Cards: Legal and Ethical Issues

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Introduction

Newborn screening has been a routine part of neonatal care in most of the developed world since the late 1960s. In New Zealand, it began nationally in 1969. Screening is not mandated by law, unlike in many other countries, but coverage is wide with 95% of all infants being tested (National Advisory Committee on Core Health and Disability Services). Ensuring an infant is tested is usually the responsibility of the Lead Maternity Caregiver (LMC) involved: a midwife or doctor. At two days of age the infant's heel is pricked and the emerging drops of blood are blotted onto filter paper attached to a test card called a 'Guthrie Card'. This is then dried and sent to the National Testing Centre (NTC) for analysis and indefinite storage.[1]

Last year, New Zealand Courts sanctioned the evidentiary use of stored Guthrie Cards in two cases. In the Watson Murder Trial, those of victims Olivia Hope and Ben Smart were used to identify spots of blood found at the alleged crime scene, while in *H v G* (Unreported judgement, High Court, Auckland, May 1999, Salmon J) the Court ordered release of the stored card for paternity testing of a child who had previously died. On appeal to the High Court this decision was upheld (*H v G* (2000) 18 FRNZ 572). These recent precedents make it timely to address the legal and ethical implications of the storage and use of Guthrie Cards. As blood samples, they contain DNA, which can be extracted, amplified and examined (Redmayne, 1998),[2] making them potential sources of considerable genetic information. For this reason and because the disorders tested for are generally indicative of underlying genetic abnormalities, newborn screening, although mainly metabolic in nature, should be approached within the context of genetic screening (Skene, 1998).

Current Guidelines in New Zealand

In New Zealand there is no specific legislation dealing with either newborn screening or genetic privacy. However, both the Health Information Privacy Code 1994 (HIP Code) and the Code of Health and Disability Services Consumers' Rights 1996 (Code of Rights) have bearings on the situation. The information that is derived from a blood spot is 'health information' under the HIP Code and is, therefore, subject to the privacy principles it contains. Whether the blood spot itself can also be considered health information is more ambiguous. As a source of DNA, it may be seen as encoded health information. In *H v G* (Unreported judgement, High Court, Auckland, May 1999, Salmon J) it was submitted that a Guthrie Card is a "document" under Rule 3 of the High Court Rules, supporting the idea that the blood spots should be treated as health information for the purposes of the HIP Code. As 'bodily substances removed or obtained in the course of a health care procedure' Guthrie Cards are also covered by Right 7(9) and (10) of the Code of Rights.

Efficacy of Screening

To justify a national screening programme of this kind the benefits must be shown to outweigh the costs. All of the diseases tested for in New Zealand (biotinidase deficiency, congenital adrenal hyperplasia, cystic fibrosis, galactosaemia, hypothyroidism, maple syrup urine disease, and phenylketonuria (PKU)) are very rare, with 30-35 babies affected each year (National Testing Centre, 1997). The procedure of obtaining the blood spot (heel lancing) is relatively harmless, however, no screening programme is completely accurate, with false negative and false positive results.

In general, the Human Genetics Society of Australasia (HGSA) recommends newborn screening only when there is benefit for the individual from early diagnosis; the benefit is reasonably balanced against financial and other costs; there is a reliable test for screening; and there is a satisfactory system in operation to deal with diagnosis, testing,

counselling, treatment and the follow up of patients identified in the test (HGSA, 1999). Based on these criteria the HGSA recommends testing for PKU, cystic fibrosis and congenital hypothroidism throughout Australasia.

The Place of Informed Consent

Whatever the legal and ethical requirement in New Zealand for informed consent to be obtained with respect to medical procedures, newborn screening is concerned with individuals who cannot, by reason of age, consent for themselves. There is a significant change when one shifts from first person consent to parental consent, since it then becomes an issue of beneficence. Section 25(3) of the Guardianship Act 1968 gives guardians the right to consent to 'any medical, surgical...procedure' on behalf of a child, where consent by another person is necessary and sufficient. However, this is subject to s23 of that Act which makes the best interests of the child paramount. This provision has been invoked on many occasions to override the authority of parents with regard to medical treatment (For example *Re Norma Mati-Leifi* (High Court, Auckland, 20 December 1991 (M2796/91) Tompkins J) and *Re CL* [1994] NZFLR 352.).

Should parental choice be overridden in this way in the context of newborn screening? Different answers are given in different countries. For instance, in Maryland, in the United States, specific legislation has been passed making written informed consent a compulsory part of newborn screening. However, there are many places[3] where newborn screening is mandatory, particularly with respect to PKU. One argument for this is that we cannot allow some parents to consign their children to irreversible disorders for which there is treatment available. However, for PKU it has been reported that the chance of missing an infant due to parental refusal is 100 times less than the chance of missing one because of a false negative result (Faden *et al*, 1982). The idea that this low level of risk is unacceptable is inconsistent with the way our society operates, since we do not live in a zero risk society. In terms of this, it seems difficult to justify mandatory screening. Alternatively, it can be argued that as the screening procedure carries minimal risk it is justified even in light of the low chance of a true positive test result.

Instead of focusing on the few couples who withhold consent, attention should be directed to improving the consent process (Nicholas and Broadbent, 1996). Currently, the procedure for informed consent centres around a Fact Sheet produced by Genetic Services entitled *Your Newborn Baby's Blood Test.* In the instructions for blood collection printed on the reverse of the Guthrie Card itself, it is a requirement that the collector ensures that the mother or other caregiver has seen the Fact Sheet. There is no requirement that this be accompanied by further explanation or, indeed, that actual consent, oral or written, is necessary. The degree to which this happens varies depending on the LMC.[4]

An important issue here is the timing of the information delivery and whether it allows the guardian to become fully informed. It has been found that when women were asked for consent just before the test they tended to feel 'psychologically committed' to the test and so were less receptive to disclosed information (Holtzman *et al*, 1983). The Fact Sheet does not present screening as a choice but rather as something that will happen. As screening is not mandatory the choice of whether or not to screen needs to be clarified. Informed consent procedures must not only recognise the parents' right to consent but must also give them an opportunity to use their powers of refusal (ibid). Neither is there any indication that consent to the taking of the blood sample does not indicate consent to all of the proposed tests. The Fact Sheet needs modification to take account of these points. The case of *H v G*, mentioned earlier, went on to become the subject of a complaint to the Health and Disability Commissioner (Case 99HDC09011). In his opinion the Commissioner found that there had been a breach of Right 7(1) of the Code of Rights in the failure of the LMC to obtain consent for the taking of the sample.

Storage

At present, around 1.8 million cards are in storage, in an identifiable form, with data computerised since 1994.[5] In the Fact Sheet the reason given for the storage of the cards is to monitor false negative results and establish why they happen. It is also indicated that 'leftover scraps' of blood may be used in setting up new screening tests but only in an anonymous form. Assuming the samples are health information under the HIP Code it is questionable whether the samples should be stored at all. Rule 9 provides limits

on the retention of health information – under subrule (1) a health agency must not hold health information for longer than is required 'for the purposes for which the information may lawfully be used'. According to Rule 10 the only lawful uses appropriate here are use for the purpose for which the information was collected (here, the blood tests), use authorised by the individual or their representative and the use of the information in an anonymous form.

Therefore, any storage of Guthrie Cards, in an identifiable form, after testing is completed requires specific informed consent. If the guardian would like the child's Guthrie Card returned to them after testing they can write and ask for this, sending the letter with the card. This is in line with the Code as Right 7(10) reads that 'any body parts or bodily substances removed or obtained in the course of a health care procedure may be stored, preserved, or utilised only with the informed consent of the consumer'. Right 7(9) of the Code provides that 'every consumer has the right to make a decision about the return or disposal of any body parts or bodily substances removed or obtained in the course of a health care procedure'. However, the current practice of assuming consent in the absence of opting-out is open to the criticism that guardians may overlook the opportunity to refuse testing or storage (Holtzman et al, 1983). There are also suggestions that information is poorer when consent is obtained by a process of opting-out than by a process of opting-in (Stratham, Green and Snowden, 1983). This may be due to the fact that people end up consenting through failure to make a decision rather than through actually making an informed decision. For this reason, it is suggested that an opting-in system, which requires guardians to actively agree to testing, would be truer to the spirit of informed consent. It would also avoid the problems associated with routine treatments and encourage a more careful approach to gaining informed consent (Holtzman *et al*, 1983). The Health and Disability Commissioner found that it is indeed a breach of Right 7(9) of the Code of Rights to retain a baby's blood sample without consent (Report on Opinion – Case 99HDC09011, 4 August 2000).

Access and Use

In New Zealand, guardians are told that if they have not heard anything after a month they can be sure the tests are all negative for their baby. This approach is out of line with the requirements of the Code as Right 6(1)(f) and (g) gives consumers the right to the results of tests and procedures, implying that all test results are to be given to the consumer. Therefore, current practice should be amended and guardians notified of the results no matter what they are.

Access to residual tissues may be helpful in understanding disease and in the development of appropriate tests. In many countries Guthrie cards are used in an anonymous form for these purposes (for example USA, McEwen and Reilly, 1994). Neither the Privacy Act 1993 nor the HIP Code deals with this issue as they apply only to identifiable information. However, the Code of Rights (Right 7[10]) requires informed consent of the consumer as a prerequisite to any utilisation of the sample, anonymous or identifiable. Consent also has to be in writing for research procedures (Right 7[6]). Therefore, any use of stored samples without informed consent is in breach of the Code of Rights.

In terms of release to third parties, the test used by the NTC in deciding whether to release information, or the Card itself, is to ask the question 'would the individual agree to the release, if asked?'[6] The HGSA's recommendation and the general principle under Rule 11 of the HIP Code is that health information shall not be provided to third parties without the written permission of the individual concerned, unless the case falls within one of the statutory exceptions. In this context, a guardian's consent will suffice. We wish to recommend that a policy of release, only on the written permission of the individual, should be adopted.

As previously indicated, there are two recent New Zealand cases which illustrate the use of Guthrie Cards for purposes other than those for which they were collected and stored. Both were allowed due to the Courts' discretion to make such orders as preserved by clause 5 of the Code of Rights which provides that nothing in the Code 'prevents a provider doing any act authorised by any enactment'. A High Court order is made pursuant to s16 of the Judicature Act 1908 and is therefore not subject to Code of Rights' provisions. In New Zealand, there are several other instances when use is made of identifiable stored samples, including clinical use when trying to explain unexpected infant deaths. For example, the child may have had an undiagnosed metabolic disorder. Use for forensic purposes includes missing person and other no-body cases. In none of these cases could the release of information benefit the individual concerned.

Although these cases may not result in harm they are still uses of the Cards other than those for which they were stored. If stored Guthrie Cards are to continue to be used for these alternative purposes, this possibility needs to be made clear at the time of sample collection. It is also necessary that consent be obtained for the storage and use of the Card with respect to specific possibilities. It may also be desirable that consent to storage and use be obtained from the individuals themselves when they reach the age of 16 years.[7] In the absence of adequate legislation, decisions will continue to be made on a case-by-case basis, leaving the population uncertain as to when and for what purpose these Cards might be used.

The use of blood spots by third parties is uncommon in New Zealand. There have been only about one hundred requests from third parties since the inception of the storage programme.[8] However, a recent American study (Stratham, Green and Snowden, 1993) showed that several screening facilities would release the cards to an insurance company, an employer, a law enforcement agency (without subpoena), and other state agencies. The numbers that were prepared to release them for anonymous research or to a family member's physician were much higher. Many more were unsure of their policy on the issue. In New Zealand, third parties may also be interested in the information, such as insurance companies and employers. While the Code of Rights goes some way towards protecting individuals from this, specific legislation, requiring informed consent before the release of any information, is needed to regulate this area.

Guthrie Cards and Ownership

It is important not to blur the distinction between the tissue itself and the information contained in it or derived from it (Skene and Chalmers, 1997), since there may be different standards regarding what can be done with each and how each can be treated in terms of ownership. While the common law does not recognise property in, or ownership of, a dead body (Beattie J [1972] NZLJ 36,37; *Awa v Independent News Auckland Ltd.* [1995] 3 NZLR 701,709.), it does not follow that there cannot be property in human tissue derived from living bodies. In fact, the language used in the Code of Rights points to an interpretation consistent with the notion of ownership rights in human tissue. In the case of *H v G* (National Advisory Committee on Core Health and Disability Services, 1995) Salmon J found that neonatal blood spots come within the definition of property found in Rule 322 of the High Court Rules. This shift is a natural one as the usefulness and potential of body parts has changed radically. The very pressures that used to point away from property in human tissue now point towards it (Matthews, 1995). This means that any unauthorised interference would not only be a breach of the Code of Rights, but also theft or conversion. In the context of the information derived from the Guthrie Cards the concept of ownership is less helpful but it is noted that the HIP Code gives control of health information[9] to the individual to whom it relates.

Conclusion

Voluntary screening is more appropriate than mandatory screening as it is consistent with both public interest and parental liberty. Nevertheless, there are several changes that need to be made to the current policy to make it a reality. Parents need to be presented with the choice of whether to consent to screening for their children, rather than merely be given information about it. There is a need for streamlined policy for gaining informed consent for the collection of blood spots, commission of the tests, storage of the Guthrie Cards, and use of both them and information derived from them. Parents must opt-into these things rather than opt-out. It is also necessary that all test results, negative or positive, be reported to the parents. Owing to the Code of Rights, Guthrie Card blood spots are the property of the individual and may be used, in any form, only with consent.

Although both the Code of Rights and the HIP Code go some way towards protecting the privacy interests of individuals in this area, there is a need for more specific legislation, particularly with respect to the current and potential secondary uses of Guthrie Cards. Secondary use already occurs in New Zealand, as seen recently in the Watson Murder Trial and in the case of *H v G* (National Advisory Committee on Core Health and Disability Services, 1995). Specific legislation, requiring consent to the various possible implications of storage and use, would create more certainty for individuals whose Guthrie Cards are stored. This suggestion is in keeping with the recent recommendation of the Health and Disability Commissioner who recommended that a policy be developed to 'ensure that informed consent is obtained from parents or legal

guardians for neonatal blood tests' (Report on Opinion – Case 99HDC09011, 4 August 2000, p.15.) It would also provide more effective regulation of the practices and policies of the newborn screening programme.

Notes

1. The policy of indefinite storage is due for review at the next meeting of the Newborn Screening Advisory Committee.

2. Note that Redmayne points out that "in practice this would be an expensive and difficult way to gain information about the donor".

3. For example in Poland and in almost all states in the United States.

4. Information provided by the Midwifery Team Leader, Queen Mary Hospital, Dunedin.

5. Information provided by the director of the NTC.

6. Information provided by a member of the Advisory Committee to the NTC, February 2000.

7. The age at which s25 of the Guardianship Act 1968 recognises the right of individuals to make their own medical decisions.

8. Information provided by the Director of the NTC, February 2000.

9. The information derived from the blood spots is health information by virtue of s4(1)(d) of the HIPC as it is identifiable and is information derived from the testing of a bodily substance.

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