Newborn screening cards: a legal quagmire

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ABSTRACT

- Newborn screening (NBS) programs are a well established and cost-effective method for early identification of genetic disorders. However, a raft of legal questions surrounds the collection, storage, ownership and secondary use of NBS cards.
- The absence of clear legal rules governing NBS programs in Australia means that there are few straightforward answers to these questions. A series of controversial incidents have exposed this uncertainty in Australia, and remarkably similar controversies have occurred in the United States and European Union.
- We review the situation, using Victoria as a case study. We also make the case for a dedicated regulatory regime for NBS programs, arguing that the lack of such a regime threatens public trust and the robust operation of NBS programs in Australia.
- New rules would likely introduce stricter requirements for informed consent at the point of blood collection than has been the norm to date. However, the scope for use of cards in research could expand rather than contract, and it may be possible to reduce the risk that vast card archives will need to be destroyed in response to future public outcries.

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Newborn screening (or "Guthrie") cards have been a fixture in Australian maternity wards for 40 years.[1] Within 2–3 days of birth, the heel of nearly every baby born in Australia is pricked to collect several drops of blood on filter paper. Laboratories test the specimens for a variety of rare conditions, including phenylketonuria, congenital hypothyroidism and cystic fibrosis. The hospital or testing service then stores the card and a record of the test results.

The public health benefits of newborn screening (NBS) programs are well established.[2-4] These programs trigger timely treatment and advice to parents and are a highly cost-effective strategy for early identification of genetic disorders.[5-7] But despite their ubiquity and effectiveness, a raft of legal uncertainties surrounds NBS programs. Who owns the cards and the blood spots? Who may access them and under what conditions? What uses are permissible beyond the initial battery of genetic tests? In recent years, controversial incidents in Australia and other countries have brought these questions to the fore. There are few straightforward answers. The absence of specific legal regimes governing NBS cards and programs in Australia means that rules regarding ownership, storage and use must be drawn from a complicated mix of directly and indirectly related laws.

In this article, we review the situation, identify the main legal uncertainties and present a case for greater clarity. For illustrative purposes, we focus on the situation in Victoria where, despite several attempts to forge clearer rules, the legal substructure of the state's NBS program remains inchoate.

Global controversy

In Australia, a series of widely publicised incidents over the past 15 years has spotlighted legal uncertainties over access to and use of NBS cards. As part of a criminal investigation into an alleged incest in 1997, Western Australia Police obtained a court order allowing them to access and test DNA from blood spots on several cards held at Princess Margaret Hospital, Perth.[8] The incident attracted national media attention and prompted public outcry that eventually led to destruction of all cards older than 2 years in the state's archive.[9]

In Victoria, a mother who became concerned about storage of her children's cards requested their return and received those for children who were not hers, sparking public debate about oversight of the state's NBS program.[10] Further controversy with Victoria's program occurred in 2003–2004 when researchers conducting a study of cystic fibrosis[11] accessed about 500 cards from the state's collection without consulting the families. The ensuing debate centred on secondary uses of the specimens, whether de-identification obviated the need to obtain specific consent for research uses, and fundamental questions of ownership.[12]

Other countries with NBS programs have experienced remarkably similar incidents. In New Zealand, use of NBS cards for forensic purposes, such as paternity testing, has stirred community concerns.[13-15] In the United States, the Texas Civil Rights Project recently sued the state on behalf of five parents over storage and continuing use of NBS cards held by the Texas Newborn Screening Program.[16] The state settled the case, agreeing to destroy 5.3 million cards that had been collected before 27 May 2009 (the day that a new governance structure for the program became law).[17,18] Ireland's Data Protection Commissioner is currently investigating whether the practice at the Children's University Hospital, Temple Street, Dublin, of indefinitely storing NBS cards and blood samples breaches Irish law and the European Union (EU) data protection legislation (Directive 95/46/EC).[19] A finding that EU law is breached could have consequences for NBS programs in other EU member states.

A common element in these controversies is the public apprehension that they provoke. Critics charge that the retention of cards, followed by the secondary use of blood spots without the consent of the child or parents, effectively creates a national biobank. In the absence of explicit legislation establishing such a repository, it is decried as a kind of "biobank by stealth". The types of legal protections that regulators would normally point to in assuaging such concerns — clear restrictions on access and use, well defined property rights, and preservation of patient autonomy through informed consent — are often missing, or lack clear application to NBS programs.

Legal frameworks

Attempts by the Australian Health Ministers' Advisory Council Working Group on Human Gene Patents and Genetic Testing to develop a national framework for administering NBS programs have not borne fruit. Idiosyncratic programs continue to operate in each state and territory, with substantial variation across programs in the number and type of disorders screened, card storage

Legal instruments relevant to the Victorian Newborn Screening Program

Commonwealth	*Privacy Act 1988* (including the National Privacy Principles)
	National Pathology Accreditation Advisory Council guidelines
Victoria	*Health Records Act 2001* (including the Health Privacy Principles)
	Human Tissue Act 1982
	Public Records Act 1973
	Charter of Human Rights and Responsibilities Act 2006
	Public Record Office Standard 99/04
	Information Privacy Act 2000

periods, and the nature of entities responsible for storing and controlling access and use. No jurisdiction in Australia has enacted legislation specifically designed to govern operation of its NBS program, which means regulatory oversight is left to a jumble of state and federal statutes. In Victoria alone, we identified six state and two federal laws that are applicable or potentially applicable to the state's NBS program (Box). Common law doctrine (ie, judge-made law) adds to the mix.

A review of the particular legal issues raised in each jurisdiction is impossible within the scope of this article. However, the situation in Victoria sheds light on the types of uncertainties that vex NBS programs nationwide.

Victoria: a case study

Ownership

Understanding ownership of NBS products is an important first step in determining who may lawfully access, use and dispose of them, and in what circumstances. The ownership issues are quite complex. One source of the complexity is that the screening produces, or may lead to, multiple products. There is the physical card, the blood spot itself, and the written pathology report of the screening tests. Laboratory work on the blood may also give rise to new biospecimens, information and knowledge, and possibly even to commercially exploitable discoveries.[20] Different rules and regulations determine property rights in these various products, and those regimes do not always provide clear-cut answers.

For example, at common law, courts have traditionally allowed only limited property rights in human tissue specimens, including blood.[21] The approach has been to treat tissue as a *res nullius* (a thing belonging to no one) until it was transformed by labour into a product (like an anatomical display specimen or a human hair wig). The property rights would go to whoever provided the skill and labour in creating the product, not to the human source of the tissue. However, the Court of Appeal of England and Wales recently held that there are situations in which the *res nullius* presumption will not apply.[22] One view of this decision is that it dealt with an unusual situation; another view is that the courts are laying the groundwork for stronger proprietary rights in human tissue. For current purposes, the salient point is that, without legislative direction, it is unclear who Australian courts might favour today in a contest over blood removed from an NBS card.

Public records

Each state and territory has statutes that govern public records. Although these statutes generally do not resolve ownership questions, they establish protections over use, and rules for storage, handling and disposal of the information. The controversy in Victoria in 2003–2004 exposed uncertainty about the applicability of the state's public records statute to the NBS archive.[23] The statute covers information collected within public entities (eg, hospitals) by public service providers; information collected in the private sector falls outside the statute. Victoria's collection of NBS cards is held by a private non-profit company, Genetic Health Services Victoria (GHSV), a wholly owned subsidiary of the Murdoch Childrens Research Institute. This corporate structure raised fundamental questions about ownership and whether the cards constituted public records.

To resolve the problem, the government invoked a special provision in the state public records statute and "deemed" all NBS cards, past and future, to be public records and thus subject to the statute's storage and disposal rules.[24] This meant that the Victorian Department of Human Services became the legal custodian of the cards and GHSV, the entity in possession of the cards, the guardian. It was an expedient solution, but an ad hoc and incomplete one. The larger background question about who, if anyone, actually "owns" the physical cards was left unanswered because, like other medical records, they are not owned by patients.

Health records legislation

Ordinarily, a tissue sample would not be considered a health record, but NBS cards may present a special case because health information often resides on the same piece of paper as the blood spot. As health records, they would be regulated by privacy principles in the *Health Records Act 2001* (Vic), which set down rules regarding collection, use, disclosure, security and retention of health records. In addition, health records held by public hospitals in Victoria are subject to rules in the *Freedom of Information Act 1982* (Vic) regarding access and correction.

Informed consent

NBS programs in all Australian jurisdictions are bound by informed consent doctrine, which emanates chiefly from the common law. Of particular relevance is the rule that consent to medical treatment (including testing) given by parents and guardians on behalf of minors is valid only when those decisionmakers are properly informed and the intervention is in the best interests of the child.[25] The human tissue statutes in all jurisdictions, including Victoria,[26] codify a version of this requirement for tissue donation. It is also a core principle in research involving human participants.[27]

NBS guidelines in Victoria stipulate that before the heel-prick test is performed on a baby, the parents must be given written information about the program and provide their consent. Clinicians are supposed to document the process in the medical record. Whether the process adheres to informed consent standards must be determined on a case-by-case basis. Historically, informed consent processes in NBS programs provided minimal information about use, access and storage, raising serious questions about the legality of any uses of the blood specimens beyond the narrow screening purpose.[28]

Concerns in this area led to the formation of the Victorian Newborn Screening Review Committee, a body convened in 2004

with representatives from relevant government agencies. One of the committee's recommendations was that families should be provided with more information during the antenatal period and that consent to the heel-prick test be obtained in writing. The committee also recommended written consent for the long-term retention and subsequent use of the card for research purposes.[7] A pilot study trialling a revamped informed consent process is underway in Victoria.[29]

Secondary uses

The most controversial aspect of the NBS debates in Australia and elsewhere has been access to and use of the cards by courts, the police, researchers, estranged family members and others for purposes not directly connected with post-natal laboratory screening. The Victorian review committee noted that: "Most parents were not aware that blood samples are retained and may be used for secondary purposes under some circumstances".[7] The key determinant of the legality of secondary uses is the validity and scope of the original consent.

Human research ethics committees have some latitude in approving the use of personal information without specific consent. However, their authority is confined to research uses. One prerequisite for securing this waiver is that obtaining the consent would be "impracticable".[28] Whether this is true in the context of NBS card data is debatable. Retrospectively obtaining consent from many thousands of families may well be infeasible. On the other hand, it is difficult to ignore the fact that there was a clear opportunity to have done this at the time the blood was taken. Recognising the central importance of informed consent to the issue of secondary uses, the Victorian review committee recommended the introduction of a two-tiered consent process under which consent to screening would be separated out from consent to long-term storage and secondary uses, such as research.

Where to from here?

The public health value of NBS programs is well established and impressive. However, they operate in Australia in what one expert has called a "spaghetti junction" of legal rules (Professor Neil Rees, Chairperson, Victorian Law Reform Commission, personal communication). One view of the absence of a dedicated and coherent regulatory regime to govern these programs is that it ensures that their operation can continue in a manner that best promotes population health, without being overly burdened by lawyers and law.

We take the opposite view. The absence of a carefully considered and well articulated set of rules threatens the robust operation of NBS programs in Australia. When controversial incidents arise in such a legal environment, they are difficult to diffuse. Without the backdrop of a clear and rational set of laws, there is greater potential for public unrest and distrust, which could ultimately threaten the viability of the entire NBS enterprise.

If the status quo is a fair indication of policymakers' preferences, we hold a minority view. In Victoria, where the review committee exhaustively considered the issues, the state government was presented with three options for addressing the legal ambiguities underpinning the state's NBS program:

- referral to the Victorian Law Reform Commission for advice on a suitable regulatory regime
- enactment of a statute specifically tailored to govern the NBS program
- modifications to current legislative instruments and practices, especially in relation to consent and collection procedures.[7]

The state government appears to have chosen the third option.

Tweaking existing laws is not enough. In particular, we believe that heavy reliance on the informed consent process to resolve future uncertainties about storage, ownership and secondary uses is problematic. Informed consent is a necessary determinant of the proper scope of these practices, but it is not a sufficient determinant. What society may regard as legitimate and ethical in any given clinical encounter cannot necessarily be generalised to the population level. In other words, even with an exemplary informed consent process in place, long-term storage and secondary use of biospecimens routinely collected from millions of neonates raise broader questions for society about accountability, transparency, agency and privacy. A fragmented regulatory approach, in which discrete aspects of NBS programs are dealt with by discrete laws, risks losing sight of the forest through a focus on the trees.

A dedicated legal regime will almost certainly lead to stricter requirements for informed consent at the blood collection stage than has been the norm to date. It does not follow, however, that it would necessarily mean tighter restrictions on all secondary uses. Indeed, with respect to research uses, it may have the opposite effect: there is strong public support for health and medical research in Australia; NBS biospecimens are amenable to analysis in de-identified form; and they have the potential to make valuable contributions to population health research. (In our view, these research considerations are one reason why the destruction of card archives in response to public concerns is probably an overcorrection.) On the other hand, use of identifiable data for forensic or private purposes without consent, or over the objections of the family, raises deeper ethical concerns. Thus, in defining rules for secondary uses, it is important not to consider secondary uses in a monolithic manner.

We urge governments in Australia to develop a coherent legal framework for NBS programs. That framework should reflect a considered balance between the rights of parents and children, community attitudes to the storage and use of biospecimens, and the interests of all Australians in high-quality prevention and treatment services at birth.

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Competing interests

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