

Privacy Issues Forum
30th March 2006
Te Papa Tongarewa, Wellington

Health and Privacy Stream, 11.45am – 1pm
“The Field of Genetics – New Challenges For Health Privacy”
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E nga mana, Tena kotou
E nga reo, Tena kotou
E nga iwi o te motu, Tena kotou

Good morning to all

I understand the expectation from the Privacy Commissioner’s Office is that I help generate some discussion about some ethical, legal and practical issues in genetics during the course of proceedings in this stream.

In a nutshell:

At the outset and in general, for this morning, I would like to suggest (and I would encourage you to question this later) that the challenges in genetics are threefold, involving the following considerations:

- (1) enabling the sharing of samples or information for beneficial health goals,
- (2) attempting to anonymise – to the fullest extent possible – such samples or information without hampering (1) above, and
- (3) giving weight to (informational) privacy rights and addressing (wider) privacy concerns and interests while taking into account – and often, trying to reconcile – other competing rights or interests.

Ruminations and Preliminary Comments

I must confess being grateful to the Privacy Commissioner’s Office for helping set the title for this presentation because, sometimes, I find myself tongue-tied, not quite knowing where to start when general questions about new developments in genetics privacy are posed.

And even with the proposed title for this morning, “The Field of Genetics – New Challenges for Health Privacy”, I still hesitate a bit for a few reasons.

First, I am not always certain if what seems like a novel issue that suggests a new challenge of some sort is really new *in itself*, or if it would be regarded as new by the person with whom I am speaking or the audience to whom I am addressing.

Second, I am never quite sure whether a particular issue is *that* new as I may already be desensitised by the literature that I have been tracking over a period of time.

Third, it seems, at least to me quite often, that when questions or issues come up in the area of genetics, there is an expectant sense that they are unique and special, and by implication, a presumption created that they are new, with the onus on the person who disagrees to displace that presumption.

So how, with all that in my mind and after some agonising, did I finally decide on approaching this presentation? I thought I would try to carry this morning's audience with me by going through an exercise I frequently undertake – I thought I would, after reviewing relevant articles, news and summaries of recent weeks, select three that would convey a sense of:

- (1) something that is more of an immediate and practical issue, something that should demand some attention from policymakers and those involved or interested in the ethical and legal implications. I am referring to the issues raised in the BMJ piece noted,
- (2) something that has been underway for a few years and that has generated much interest but that has just, in the past fortnight, reached a milestone – by this, I am referring to the latest development in the ongoing effort to establish the UK Biobank (I will talk about this later),
- (3) something that seems to lie quite far ahead in the future and that sounds quite remarkable (although if one were to step back and contemplate, would not be totally unsurprising given the typical progression of how technology tends to develop and unfold and the associated effect or consequence of the lowering of costs involved) – this is also something that has been the subject of speculation in the ethics or policy literature but looks increasingly like there will not be much time left for more speculation as we will be faced with a barrage of questions and issues to consider which may require some decisions to be made and acted on. I am referring to news about genome scientists having recently completed a draft of the second nonhuman primate – the rhesus macaque – for US\$22m, and the prediction that researchers are closing in on a new generation of technology they hope will slash the cost of a genome sequence to US\$1000.

While thinking about this third item, together with the first two, it occurred to me that I should share a couple of things that I have personally found useful as 'reference points'

over the years when I try to sift through various issues that come up in this area. These two things involve:

- the different meanings or senses of ‘privacy’, and
- the definition of ‘genetic testing’.

With regard to the latter, I have found it more helpful to think about asking the broader question: what is involved in diagnosing a genetic disorder? Before I get ahead of myself, let me continue with the three recent items of interest I have chosen.

The BMJ piece: Anneke Lucassen, Michael Parker, Robert Wheeler, Implications of data protection legislation for family history, 2006; 332(7536): 299–301

I would like to make three quick comments, some of which repeat the points the authors make in this piece and elsewhere. First, family medical history can provide predictive genetic information similar to that obtained from genetic (laboratory) testing. The gathering of information about family members helps with investigation into the patients’/consumers’ risks of developing genetic disorders.

Second, constructing the family tree involves information or knowledge about the genetic risks of the patient’s/consumer’s relatives. The question here is – what is the extent of confidentiality owed to the patient/consumer, or rather, in what circumstances would the breaching of confidentiality be justified, for instance, by the health provider making disclosures to or informing one or more ‘at risk’ relatives about their genetic risks.*

Conversely, that is on the other side of the coin, the question is how can and to what extent should a person’s (moral or legal) right, or at least, wishes, *not* to know about her or his genetic risks be respected and given effect?

Perhaps, some may reason by analogy with or appealing to cases where disclosures to third parties would be justified, for example, in the mental health or communicable diseases contexts, where the gravity/magnitude and likelihood of harm justifies disclosure. Very briefly, it must be noted, however, that harms in the context of genetics are diverse and may not be comparable. Additionally, the availability or effectiveness of any possible intervention (or lack of) seems to assume a more significant dimension of importance in the genetics context.

Third, clinical geneticists/genetics providers invest a lot in relationships with their patients/consumers and, which may be frequently the case, a few members of their immediate or extended family. The decision whether or not to disclose information to ‘at risk’ relatives is – as a matter of standard practice – left to the clinician to ascertain what

* Here, I am drawing on Anneke Lucassen’s and Michael Parker’s article: Confidentiality and serious harm in genetics – preserving the confidentiality of one patient and preventing harm to relatives, *European Journal of Human Genetics* 2004; 12: 93-97

is the best course of action (or, perhaps, the least worst action). It may be of interest to note here that a French study showed “nearly 10% of women attending a breast cancer genetic clinic would inform *no one* of any genetic testing result, and a third would inform one relative but not necessarily all”.**

The UK Biobank

The UK Biobank was an idea that, going by my fallible memory, was first mooted in 2000, sometime around the excitement and anticipation over the near completion of the mapping of the human genome.

As the announcement about two weeks ago revealed, the UK Biobank is starting to recruit volunteers with 3000 people being sent letters in the South Manchester area.

The UK Biobank, a large-scale population database, will involve 500,000 volunteers, aged between 40 and 69, who will provide samples and their information that include clinical records, lifestyle information, and family history. The goals of the UK Biobank include directly improving the health of the wider community by integrating findings into the health care system, and encouraging and advancing health research. More specifically, the UK Biobank is expected to help locate susceptibility genes for common diseases like cancer, heart disease, stroke, diabetes and dementia. The £61.5m initiative is funded by the Medical Research Council, the Wellcome Trust, and the Department of Health.

Commercial Access: Information and samples from participants will be available only to medical researchers (public or private) for bona fide research that meets the relevant scientific and ethics approval and is in the public interest. No one will be given exclusive or preferential access. Results from any tests made on participants or their samples will be put in the database so that they are available to all other approved researchers. Pharmaceutical and other health-based companies can access the study data for approved research. Insurers and employers will not be given any individual’s information, samples or test results. Police, security services, relatives or lawyers cannot access the information or samples unless compelled to do so by the courts.

Access will only be granted to anonymous information to all ‘users’. In a sense, an approach similar to that adopted in the context of the US health information privacy rules is taken where anonymisation (of samples/information originally collected in identifiable form) makes it acceptable for use in health research. It is interesting to note here that, sometime ago, Genewatch and Liberty argued that genetic information is *itself* a personal identifier and so can never be kept truly confidential.

** Julian-Reynier *et al*, Disclosure to the family of breast/ovarian cancer genetic test results: patient’s willingness associated factors, *Am J Med Genet* 94:13, cited in Lucasen & Parker, Confidentiality and serious harm in genetics – preserving the confidentiality of one patient and preventing harm to relatives (*supra*)

The Race for the US\$1000 Genome

As mentioned briefly before, genome scientists have recently completed a draft of the second nonhuman primate – the rhesus macaque – for US\$22m. This piece of news in the 17th March 2006 issue of *Science* was also accompanied with the statement that, “by the end of [this] year, at least one company expects to turn out a full mammalian genome sequence for about \$100,000, a 3000-fold cost reduction in just 6 years [since the final draft of the human genome sequence] and the prediction that “[r]esearchers are closing in on a new generation of technology they hope will slash the cost of a genome sequence to (US)\$1000”.

This piece of news brings to my mind a report put out in March last year by a joint working group of the UK Human Genetics Commission and the UK National Screening Committee. The joint working group conducted an analysis of the ethical, social, scientific, economic and practical considerations of genetic profiling (that is, the analysis of a person’s entire genome to reveal their personal genetic information) at birth. Four things should be noted about the findings of the joint working group’s report:

- that there are important “ethical, legal and social barriers to the introduction of genetic profiling of babies at birth as a public health service”,
- that, however, it is “unlikely to be publicly affordable within the next 20 years, even though commercial services are likely to be offered in this timeframe, potentially raising issues of regulation”,
- that “it is important research continues in order to establish how far profiling could be clinically useful, and it is critical developments are kept under review”, and
- that the group recommends to government “the entire topic should be revisited in five years when technologies will have moved on and the prospect of this becoming a reality is closer”.

Given the news in the *Science* magazine, it may be the reality is drawing much closer than expected and that, if and when, any proposal for the genetic profiling of newborns gets mooted, issues surrounding – for example, appropriate consent or authorisation for the collection of samples or information, and more specifically as far as privacy is concerned, how samples or information will be stored, the uses of such samples or information (eg subsequent/future/secondary uses), and under what circumstances the samples or information will be accessed, who may access the samples or information, and what safeguards will have to be in place – will have to be considered in detail.

Privacy – in what sense?

The next part of what I would like to raise relates to the different senses or the range of meanings of ‘privacy’.^{***} One of the meanings of privacy relates to the constitutional right of the kind recognised by, for example, the US Supreme Court in cases like *Griswold v Connecticut*. *Griswold* involved a Connecticut law that prohibited the use of contraceptives and the court, by a vote of 7-2, invalidated the law on the grounds that it violated the “right to marital privacy”. The majority ruled that the right was to be found in the "penumbras" of other constitutional protections.

This kind of privacy right is of a decisional nature and may sometimes be heard invoked when arguments for ‘reproductive autonomy’ or ‘procreative liberty’ are made. In the genetics context, decisions about whether to undergo pre-natal screening and the continuation or termination of pregnancy, and choices and decisions to select against genetic disorders before implanting embryos or to select for sex on non-medical grounds, fall within the scope of this meaning of privacy.

There is another sense in which privacy is used and that is of being left alone or ‘in private’. This can be characterised as a kind of non-molestation right. The right or freedom (or maybe just the wish) to *not* want to know one’s genetic risks may be regarded as coming within this sense of privacy.

More commonly, privacy is understood and discussed in the ‘informational’ sense, i.e. the expectations, rules and safeguards governing the flow of personal information about the individual citizen – that includes the collection, use, storage and access of information about the individual, for example under the Health Information Privacy Code.

The fourth sense of privacy is proprietary – claims for intellectual property arising from findings made from discoveries in genetic knowledge fall within this sense of privacy.

Often in the genetics context, at least two of the meanings of privacy are involved, and potentially, all four senses can be raised.

Genetic Testing – definition?

I had, a few minutes ago, said that one of my ‘reference points’ in navigating through ethical and legal issues in genetics is the frequent reminder to myself to ask and consider the definition of ‘genetic testing’ and – more usefully – to ask and think about the broader question of what is involved in diagnosing a genetic disorder. There may sometimes be the tendency to think of genetic testing mainly or solely in terms of the analysis of DNA (whether directly, or indirectly by the analysis of RNA, chromosomes, proteins or certain metabolites) in the lab.

^{***} Here, I would like to acknowledge Maurice Ormsby, PhD, a member of the Health Research Council Ethics Committee for an e-mail correspondence sometime a few years ago when we both discussed the first three senses of privacy.

It should, however, be kept in the back of one's mind – or rather – at the forefront of one's mind that genetic diagnosis can be made from simple clinical observation and examination (ie by the health professional *seeing* the patient/consumer) or by the taking of personal or medical family history (ie by *asking* the patient/consumer and getting a response). The gathering of such information and subsequent uses and disclosures of that information attract privacy expectations, rules and safeguards (in the informational sense of 'privacy').

Closing words

Finally, the title of this presentation makes reference to new 'challenges'. However, there is another side to that as the title could well make reference to new 'opportunities' for the improvement of health for all and advancement of research in the public interest.

Thank you all for your attention.

Thanks to the Commissioner for the invitation to speak, and thanks to the staff of the Office of the Privacy Commissioner.

Thanks to the Chair for this stream.

Lastly, I would like to acknowledge the Law Foundation – the funder for the Human Genome Research Project, Otago University, which I am managing, that has provided the opportunity for the group of us working on the project to read, discuss together, and reflect on the issues.