



# Genetic Research in Aboriginal and Torres Strait Islander Communities: Beginning the Conversation

## *Discussion Paper*

Emma Kowal, Lobna Rouhani and Ian Anderson





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THE UNIVERSITY OF  
MELBOURNE



the  
**Lowitja**  
INSTITUTE

Australia's National Institute  
for Aboriginal and Torres Strait  
Islander Health Research

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**The Lowitja Institute**

PO Box 650, Carlton South  
Vic. 3053 AUSTRALIA

**T:** +61 3 8341 5555

**F:** +61 3 8341 5599

**E:** admin@lowitja.org.au

**W:** www.lowitja.org.au

**Authors:** Emma Kowal, Lobna Rouhani and Ian Anderson

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**Definition:** Within this guide the term ‘Indigenous’, when used in an Australian context, refers both to Aboriginal people and to Torres Strait Islanders.

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## Abbreviations

DNA	deoxyribonucleic acid
GENA	Genetic Education for Native American
GERIS	<i>Guidelines for Ethical Research in Indigenous Studies</i>
GLAI	Genetics Literacy Assessment Instrument
GWAS	Genome Wide Association Study
HRCNZ	Health Research Council of New Zealand
ICIP	Indigenous Cultural and Intellectual Property
MJD	Machado Joseph disease
MOA	monoamine oxidase
mtDNA	mitochondrial deoxyribonucleic acid
NHMRC	National Health and Medical Research Council
REAL-G	Rapid Estimate of Adult Literacy in Genetics
REALM	Rapid Estimate of Adult Literacy in Medicine
SNPs	nucleotide polymorphisms
WAAHIEC	Western Australian Aboriginal Health Information and Ethics Committee



## Preface

Genetics is at the forefront of medical research, but it is rarely used in Indigenous health research projects. In the past, proposals to conduct genetic studies in Aboriginal and Torres Strait Islander communities in Australia have been highly criticised and rarely funded. However, genetic researchers worldwide argue that genetics has the potential to reduce health disparities (including Indigenous health disparities) in multiple ways: through understanding disease pathogenesis, using genetics to probe environmental risk, predicting disease risk, finding novel diagnostics and drug targets, and pharmacogenomics.

Understandably, many Indigenous people interpret genetic research in the context of their experiences of colonisation. Multiple fears constitute barriers to effective research partnerships between Indigenous communities and genetic researchers. These concerns include genetic theft or 'biopiracy', that genetics will be used to determine Aboriginality and may fuel racism, of poor access to potential health care innovations, of bad experiences of the Human Genome Diversity Project (known by some as the 'Vampire' project) and of struggles over access to deoxyribonucleic acid (DNA) extracted from human remains.

Clearly, there are serious risks from genetic research for Aboriginal and Torres Strait Islander people, as well as potential benefits. The only way that these risks and benefits can be managed effectively is through open discussion among all of those parties with an interest in the conduct of genetic research in Indigenous communities.

On 2 July 2010 the first Roundtable on genetic research in Aboriginal and Torres Strait Islander communities was convened by The Lowitja Institute, Australia's National Institute for Aboriginal and Torres Strait Islander Health Research. The Roundtable was attended by twenty-four Indigenous and non-Indigenous attendees, including experts in genetics, Indigenous health research, Indigenous research ethics, genetic ethics and genetic literacy. This discussion was especially significant because it was the first national discussion of genetics and Indigenous health to have taken place in Australia. Although comparable nations such as Canada, the United States of America and New Zealand have a long history of discussing these issues, in Australia these conversations had not yet taken place.

The Lowitja Institute is ideally positioned to manage 'difficult conversations' such as those about genetic research. The aims of this conversation were to create a community of interest around Indigenous genetic research; to foster collaborative genetic research in Indigenous communities that is ethical, productive and of high quality; and to begin to consider the tools and models that are needed to make genetic research a culturally safe option for Indigenous people.

The Lowitja Institute is well aware of the potential difficulty of this conversation, but remains committed to bringing stakeholders together in a safe space. Broaching topics like these, which have long been put in the 'too-hard' basket, can end up in unproductive conversations. But we know that it is important to have those conversations. Indeed, as the summary of the discussions presented in this discussion paper

plainly shows, discussions were honest and constructive and considered many difficult issues. These included the sensitivities inherent to the method of genetics (such as how the diversity of ancestry within Indigenous populations is dealt with); the risks involved when research findings are generalised to the entire Indigenous population; the existence of highly publicised examples of 'bad' Indigenous genetic research in other countries; and the need to provide short-term benefits to participating Indigenous communities, given the long-term nature of the potential benefits of genetic research.

This discussion paper presents background material prepared for the Roundtable participants on ethical issues associated with genetic research in Indigenous populations; international approaches to guidelines for genetic research; and genetic literacy in Indigenous populations. It also presents a summary of roundtable discussions. Roundtable participants were offered the opportunity to amend or comment on this summary and approved the final version.

We hope that this discussion paper generates many more conversations about the potential of genetic research to contribute to Aboriginal and Torres Strait Islander health research efforts, and, more importantly, the mechanisms that need to be in place to ensure that Indigenous communities are empowered to make informed decisions about participating in genetic research.

**Professor Ian Anderson**

April 2011







## Background Papers

Four papers were sent to the participants who were invited to attend the Indigenous Genetics Roundtable. The papers were:

- *Genetic Research and Aboriginal and Torres Strait Islander Australians*
- *Summary of International Guidelines for Indigenous Genetic Research*
- Review of *Summary of International Guidelines for Indigenous Genetic Research* by E. Kowal and L. Rouhani
- *Genetic Literacy Discussion Paper*.

### *Genetic Research and Aboriginal and Torres Strait Islander Australians*

*Emma Kowal, Glenn Pearson, Lobna Rouhani, Chris S. Peacock, Sarra E. Jamieson and Jenefer M. Blackwell*

#### **Abstract**

Human genetic research is a rapidly developing field that promises to deliver a range of health benefits to the population in general. However, genetic research that takes place with minority groups raises many sensitive issues and has generated much debate in scientific journals and the media. Attempts at genetic research in Indigenous communities have proven particularly controversial. Indigenous peoples have raised concerns about a lack of benefit to their communities, a diversion of attention and resources from non-genetic causes of health disparities and racism in health care, a reinforcement of ‘victim-blaming’ approaches to health inequalities, and possible misuse of blood and tissue samples. The philosophical and cultural implications of genetic research can fundamentally change the ways that we think about our bodies, disease, human variation and what it means to be human. For Indigenous peoples, these can also challenge one’s sense of identity and cultural beliefs.

In Australia, these issues have acted as a barrier to conducting genetic research in Indigenous communities, and, as a result, genetic research in Australian Indigenous populations is in its infancy. This is in sharp contrast with comparable Indigenous groups in Canada, the United States and Aotearoa/New Zealand, which have been considering the use of genetic technologies and participating in genetic research for some time.

Drawing on international literature and local contexts, this paper reviews the ethical issues relevant to genetic research in an Australian Indigenous context, including informed consent, storage of samples, privacy and confidentiality, ownership and use of samples, commercialisation, benefits to the community, effects on collective identity and risks to community harmony. Finally, it considers how Australian researchers and Indigenous communities can constructively respond to the challenges of genetic research such that Indigenous Australians can equally benefit from genomic advances.

## ***Introduction***

Human genetic research is a rapidly developing field that promises to deliver a range of health benefits to the population in general. However, genetic research that takes place with minority groups raises many sensitive issues and has generated much debate in scientific journals and the media (Juengst 1994; Wade 2005; Hausman 2008). Attempts to carry out genetic research in Indigenous communities have proven particularly controversial (Foster & Sharp 2000; Scott et al. 2005). There is a growing international literature showing that genetic research can have a detrimental effect on minority groups, including Indigenous people (Lone Dog 1999; Reardon 2005; Marks 2006). Indigenous peoples have raised concerns about a lack of benefit to their communities, a diversion of attention and resources from non-genetic causes of health disparities and racism in health care, a reinforcement of 'victim-blaming' approaches to health inequalities, and possible misuse of blood and tissue samples (Dodson & Williamson 1999; Pearce et al. 2004). The philosophical and cultural implications of genetic research can fundamentally change the ways that we think about our bodies, disease, human variation and what it means to be human (Rose 2001). For Indigenous peoples, these can also challenge one's sense of identity and cultural beliefs (Greely 1998).

Issues such as these have been found to affect the participation of members of minority groups in genetic research (Sanner & Frazier 2007; Bowen & Penchaszadeh 2008). In Australia these issues have acted as a barrier to conducting genetic research in Indigenous communities. A recent review of public health genomics in Australia commented that 'very little is known about the specific genetic issues relevant to Indigenous Australians' (Metcalf et al. 2009:121–8). This is in sharp contrast with other comparable Indigenous groups in Canada, the United States and Aotearoa/New Zealand, which have been considering the use of genetic technologies for some time (Dukepoo 1998; Glass & Kaufert 2002; Du Plessis et al. 2004).

There is a large gap between Indigenous and non-Indigenous health outcomes. In Australia the life expectancy at birth for Aboriginal and Torres Strait Islander people is 17 years less than non-Indigenous Australians, largely due to higher rates of chronic disease and injury (AIHW & ABS 2008). Indigenous health disparities in comparable nations exist to a lesser degree. In Canada and the United States, for example, the life expectancy at birth for Indigenous people is six years less than non-Indigenous people, while in New Zealand there is an eight-year difference (Cooke et al. 2007:7)

The question of whether genetic research has anything to offer Indigenous populations has been debated. Given the large socioeconomic inequalities that exist between Indigenous and non-Indigenous populations, some argue that social factors rather than genetic factors are solely responsible for Indigenous ill health (Pearce et al. 2004; Paradies, Montoya & Fullerton 2007). However, others argue that although the poor health of Indigenous people is largely due to social factors, it is likely that the interaction of genetic factors with the environment also plays a role (Durie 2003).

This paper does not argue whether or not genetic research should take place in Indigenous communities, although we consider this an important issue that will continue to provoke debate. Rather, we recognise that it is likely that Indigenous Australians will increasingly be invited to participate in genetic research. Given this, both researchers and research participants should be aware of the issues that Indigenous people elsewhere have raised, and the solutions that have been developed to manage these issues. This paper reviews the issues that Indigenous people have raised

with reference to genetic research projects, DNA banks and other population genetic initiatives, and points to a way forward for resolving these issues. Although the paper focuses on Aboriginal and Torres Strait Islander populations in the Australian context, it also considers other Indigenous populations within settler states, particularly Canada and New Zealand. The authors have an interest in this topic from a variety of positions, including as non-Indigenous genetic health researchers working in Indigenous communities (JB, SJ), as an Indigenous researcher engaged in health research projects (GP), and as a non-Indigenous cultural anthropologist of genomics and indigeneity (EK).

### ***Indigenous responses to genetic research***

Indigenous and minority communities around the world have expressed concern about research that takes place in their communities, and about health research in particular (Smith 1999; Freimuth et al. 2001; Humphery 2001). The history and ongoing incidences of exploitative relationships between Indigenous peoples and the societies that colonised them have severely eroded Indigenous peoples' trust in non-Indigenous societies. This lack of trust affects all activities that are initiated by non-Indigenous people, including research that aims to improve Indigenous health. Genetic research is among the most controversial types of health research, and many Indigenous people interpret genetic research in the context of their experiences of colonisation. Research in New Zealand reported that some Māori perceived genetic research in the context of past research practices that had exploited Māori communities. They worried that genetic researchers did not have their best interests at heart, and that poor access to health care would mean they might not benefit from any breakthroughs that resulted from genetic research (Du Plessis et al. 2004). The Indigenous Peoples Council on Biocolonialism in the United States argues that much genetic research on Indigenous people is 'genetic theft' or 'biopiracy' (IPCB 2000). In contrast, prominent Māori health researcher Mason Durie argues that genetic vulnerability is a key cause of Indigenous ill health, along with socioeconomic disadvantage, resource alienation and political oppression (Durie 2003). In Australia recent consultations with Indigenous people to identify health research priorities, facilitated by Australia's National Health and Medical Research Council, have identified genetic research for the first time as an area requiring more attention (Dwyer & Silburn 2009).

Many different kinds of genetic research projects have been conducted in Indigenous communities, only some of which have been contentious. For the purpose of this discussion, genetic research should be considered in three categories: research on rare genetic disorders; population genetics; and the study of common, complex diseases.

Generally, genetic research into rare diseases affecting some families within Indigenous communities has not been controversial. For example, Canadian researchers began researching primary biliary cirrhosis (an autoimmune disease that causes liver failure) in a First Nations community in northern Canada after local doctors noted the high rates of this disease (Arbour et al. 2004). Research into a rare neurological disease on Groote Eylandt in the Northern Territory of Australia was similarly welcomed by the community (Burt et al. 1996).

The bulk of Indigenous opposition to genetic research relates to population genetics. Some Indigenous groups believe that genetic research into human population history threatens their cultural beliefs. For example, when evolutionary accounts of history stated that Native Americans migrated to North America through the Bering Strait 15,000 to 45,000 years ago, an upheaval

was caused because many people in the Native American community believe their ancestors had always been in North America and did not ‘arrive’ from somewhere else (Foster & Sharp 2000). Native Americans have also resisted DNA research on ancestral remains found on their lands (TallBear 2003).<sup>1</sup>

The Human Genome Diversity Project, the largest project of its kind, generated the greatest opposition to date. It sought to collect DNA from Indigenous groups in order to understand the diversity of human species. It created much controversy in Australia and internationally by calling Indigenous groups ‘Isolates of Historical Interest’ that had to be sampled before they ‘vanished’ (Dodson & Williamson 1999; Reardon 2005; Marks 2006). Indigenous advocates have stressed the links between researchers’ desire for Indigenous DNA and the rush to exploit Indigenous lands in the colonial era (Mead 1996; IPCB 2000; Marks et al. 2005). The current Genographic project has also been resisted by Indigenous groups (UNESCO 2006). However, some Native American groups in the United States have used ancestry testing to determine tribal membership (Lock 1999; TallBear 2007). All the existing Australian literature that comments on Indigenous genetic research concerns population genetics (Dodson 1996; Dodson & Williamson 1999; van Holst Pellekaan 2000; Rimmer 2007).

A third type of genetic research focuses on common diseases such as diabetes, heart disease and infectious diseases that occur at higher rates in Indigenous populations than in the general community. Researchers hope that by looking at the DNA of these groups they may be more likely to find genes that are relevant for understanding these diseases. Finding these genes may lead to the development of new treatments or vaccines that will help the whole population. It may also allow people at higher risk of these diseases to be identified as targets for prevention and health promotion (Davey-Smith 2007; Khoury 2009).

Although this type of research is less controversial than research into population genetics, it still attracts criticism. As mentioned previously, Indigenous people have argued that Indigenous health inequalities are not due to genetics, but to social and environmental factors, including colonisation and racism (Pearce et al. 2004; Paradies et al. 2007). Seen from this perspective, genetic research diverts attention from other potentially more important influences on health. By encouraging the idea that health inequalities are due to genetics, not social factors—an effect called genetic determinism—some argue that genetic research could even worsen health inequalities (CRG 2006).

Those arguing that genomics can contribute to the elimination of health disparities emphasise that genetic studies ‘have turned from a science du jour to a necessary and integral part of how we identify disease susceptibility among populations with ancestry from different parts of the world’ (Ramos & Rotimi 2009:5). Contemporary genetic research generates much new information about disease development and progression, and possible new ways to combat diseases. The dearth of genetic research in Indigenous communities means that Indigenous people may be excluded from biomedical innovations that result from genetic research, such as new pharmaceuticals and advances in pharmacogenomics. More generally, researchers have tried to counter genetic determinism by stressing that genes interact with the environment to cause disease, and only rarely cause disease on their own. Therefore, using genetic research to explore a health problem should not be interpreted as saying that a disease is ‘genetic’ and that social and environmental

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1 Note that the recent announcement that researchers are planning to sequence the genome of the legendary Native American Chief Sitting Bull is yet to be criticised by Native American scholars (Witze 2010).

interventions are not relevant. Indeed, because we sample our environment through our genes, genetic research can often help us to identify environmental variables (e.g. micro-nutrients or vitamins) that contribute to disease susceptibility. This has led to the suggestion that, because functional genetic variants can act as a proxy for environmental exposure, genetic epidemiology may help to 'redress the failures of observational epidemiology' (Ebrahim & Davey-Smith 2008:15). The next section reviews the major ethical issues relevant to the conduct of genetic health research in Indigenous communities.

### ***Ethical issues in genetic research with Indigenous communities***

In Australia there are now a number of excellent resources to help Aboriginal and Torres Strait Islanders communities and researchers ensure their research is valued by the community, is conducted ethically and provides meaningful benefits to participants (NHMRC 2003, 2006; AHMAC 2004). Although these provide excellent ethical guidance for all health researchers, issues specific to genetic research are not mentioned.

In contrast, Canadian and Aotearoa/New Zealand Indigenous health research guidelines do provide guidance for genetic health researchers. As stipulated in the Canadian Institutes of Health Research guidelines for research involving Aboriginal people, ethics in research is not just a 'review moment' (CIHR 2007). Rather, ethics should comprehensively encompass all issues, including the need to balance collective and individual interest; authentic opportunities to participate in the dissemination of results and the interpretation of data, affording due credit to those actively involved in the research project; and explicit communication of commercial potential. Moreover, the establishment of ethics should be undertaken within a framework of mutual trust and co-operation, whereby both the researcher and those participating in the research project will benefit from the study, as well as fully understanding their rights and responsibilities. Canadian guidelines also reinforce the notion of biosamples being 'on loan' to researchers, with researchers acting as 'stewards' rather than owners, and research participants retaining the right to access data about themselves (CIHR 2007). The New Zealand guidelines share this concept of ownership and state that genetic research must take place within the 'paradigms of a Maori world view' (HRCNZ 2008:19).

An overarching ethical principle of Indigenous research of any type must be Indigenous participation. In particular, given the social dangers of genetic research, it is imperative that Indigenous people are in control of any genetic research that takes place in their communities. Ideally, Indigenous geneticists and genetic epidemiologists would lead genetic research initiatives (Dukepoo 1998). As this is not yet feasible, at least in the Australian context, non-Indigenous researchers must collaborate with Indigenous researchers on every part of the research process, from planning to data collection to publication and research dissemination (NHMRC 2003).

A formal agreement with the community, such as a memorandum of understanding, is an important way for issues of participation and control to be formalised. Arbour and Cook (2006) identify five issues that should be addressed in a 'research contract' between the researchers and a community representative body: the provision of culturally appropriate counselling when and if genetic results are fed back to individuals; the possibility of stigma or discrimination from within or outside the community as a result of the research; the use, storage and disposal of genetic samples; feedback of research findings; and potential commercialisation of results, data ownership and sharing, review of publications and authorship (Arbour & Cook 2006).

The remainder of this section briefly discusses the important issues that Indigenous communities, and the genetic researchers working with them, need to consider. As well as Arbour and Cook's work, it draws particularly on a New Zealand study examining Indigenous peoples' attitudes to genetics (Du Plessis et al. 2004).

### ***Consent to genetic research***

Like any form of research, it is important that research participants understand a genetic research project and freely give their consent to participate. Depending on the pre-existing knowledge of genetics and DNA within the community, informed consent may require the use of culturally appropriate tools for promoting genetic literacy. Where English is not the first language of Indigenous people, an interpreter should be used. Even where English is a first language, an Indigenous researcher or cultural consultant will often need to be involved in the consent process to ensure that there is clear communication between the researcher and the research participant (NHMRC 2003). Consent for genetic research must be free and informed, and should cover all the issues that the community thinks are important for individuals to know including storage of genetic samples, access and ownership of DNA, and commercialisation. In an Indigenous context, group-level consent must also be obtained from a collective organisation such as a community council (in the case of a discrete Indigenous community) or a community-controlled Indigenous health service.

### ***Privacy, confidentiality and storage of genetic samples***

Our genetic code contains a huge amount of information about us and our kin. Fears have been raised that information about the diseases someone is at risk of could be used by insurance companies and employers to discriminate against people (and potentially their family members) with certain DNA markers (ALRC 2003). Concerns have also been expressed about the potential use of DNA to exclude Aboriginal and Torres Strait Islander people from Native Title claims or from government assistance (ALRC 2003; Sutton 2005). Moreover, there are concerns that the police could access DNA for forensic purposes, or that agencies dealing with child maintenance could access DNA for paternity testing. For these reasons, many Indigenous people are particularly concerned about the maintenance of privacy and the storage of and access to DNA samples. Further, issues of confidentiality relate to the community as a whole, as well as individuals. For instance, Indigenous community representatives may decide they do not want the community to be named in research publications in order to prevent any harm arising from potentially negative portrayals of the community (see below).

The governance of the research project is important for ensuring that privacy and confidentiality of research participants is maintained. Indigenous communities may not have trust in the oversight provided by mainstream ethics committees (Humphery 2001; Castellano 2004). Many Indigenous health projects will have their own oversight committees, such as a steering committee or reference group, depending on the size and nature of the project. Where these groups exist, it is important that members of the community are part of the group.

### ***Ownership, use and commercialisation***

Many Indigenous communities consider that their natural and cultural resources are collectively owned by Elders or the whole community. Some Indigenous groups extend this belief to DNA, and



consider that the DNA of an Indigenous person is the property of the Indigenous nation, as well as the property of the individual (Mead 1996). This is particularly the case where the DNA sample is wanted because it is 'representative' of the DNA of the group, as is the case in population genetic studies (IPCB 2000).

Research with Māori people found that their concerns about ownership relate to the potential for third parties to access their DNA for commercial purposes such as drug development (Du Plessis et al. 2004). Native American communities have similarly expressed concern about their DNA being used for research to which they did not actively consent (Foster & Sharp 2000). Indigenous communities and genetic researchers need to negotiate both the ownership and nature of the research conducted on the DNA samples. As mentioned above, Canadian ethical guidelines suggest that samples should be considered 'on loan' to researchers, a concept developed by genetic researcher Laura Arbour and Indigenous researcher Doris Cook (2006).

Some genetic research has led to the discovery of new drugs that have generated profits for drug companies, raising concerns about the sharing of benefits between the researchers and the community that participated in the research (Knoppers 2000). Profit deriving from the genetic modification of organisms and from the patenting of DNA has created particular controversy among Indigenous people (Cunningham 1998; IPCB 2000; Du Plessis et al. 2004). Therefore, inclusion of the issue of commercialisation in any research agreement between Indigenous communities and genetic researchers is desirable.

### ***Benefits and risks to the community***

All health research in Indigenous communities must benefit those communities in a meaningful way. As with many types of health research, there is likely to be a significant delay before the research findings translate into any health benefits. In many cases, these benefits will be provided to the whole population, and not Indigenous people in particular. Māori people have expressed concern that they may even miss out on any health benefits of research because of their poorer access to health care, leading to a 'double oppression' (Du Plessis et al. 2004). Researchers and community members should agree on the benefits the research will aim to offer the community, both short term and long term.

The corollary of group benefits is group risks. Commentators have argued that Indigenous (and other socially identifiable) groups face culturally specific risks of genetic research as a group, in addition to their individual risks (Foster & Sharp 2000; TallBear 2001; Brodwin 2002). For example, the portrayal of an Indigenous group as 'genetically susceptible' to particular diseases can affect the way Indigenous people are treated in society, and even the way they experience their indigeneity. This is of particular concern to young people whose cultural identity and self-esteem are inter-related and vulnerable to external influences (American Academy of Pediatrics 2004). This is another aspect of the problem of 'genetic determinism', where problems that have complex causes are attributed solely to genetics, and thus to the individual, rather than their social context. Accordingly, New Zealand's ethical guidelines stipulate that findings of genetic research projects with particular Māori groups should not be generalised to the Māori population at large (HRCNZ 2008).

There is also a risk that the conduct of research could disrupt a community, creating conflict and disturbing the balance of power between different community groups. These risks are not unique

to genetic research, but because issues of genetics and DNA can be particularly controversial (Darby 2002; Denholm 2007), genetic research may be more likely than other kinds of health research to disrupt community harmony. This risk could be alleviated with widespread community consultation, including community meetings where any concerns could be raised openly.

### ***Conclusion: Model for Indigenous genetic research or cautionary tale?***

Despite criticism, genetic research continues in Indigenous communities around the world and, in fact, many research projects are conducted successfully with the consent and participation of the Indigenous community concerned. This final section discusses a further example where genetic researchers and Indigenous communities appear to be finding solutions to the challenges posed by genetic research, but not without controversy. This example illustrates both the potential for culturally sensitive Indigenous genetic research and the dangers inherent in even the most sensitive Indigenous research efforts.

Māori and non-Māori health researchers at New Zealand's Environmental Science Research Institute have collaborated with Te Iwi o Rakaipaaka (the organisation representing members of the Rakaipaaka community) on the Rakaipaaka Health and Ancestry Study, based predominantly in Nuhaka (Hawke's Bay). It aims to recruit 3000 Māori to participate in a longitudinal 'envirogenomics' project that will investigate common diseases that affect families such as diabetes, gout, heart disease and cancer. The research team is composed of Māori and Pakeha (non-Māori) researchers led by Rod Lea and Marino Lea, the latter of whom is a member of the Rakaipaaka iwi (clan).

The research team has taken many of the ethical issues raised in this paper into account. An incorporated community organisation has control of the project, and retains ownership of the genetic information. It has also formed a 'Māori *kaitiaki* [guardianship] group' to oversee research that uses Māori genetic information and to develop policy regarding secondary use of samples. One example of such an arrangement for culturally appropriate procedures for the storage of blood and other samples comes from the New Zealand cancer tissue bank, which, since 2004, has offered all donors the option of having their samples disposed of with a Māori blessing or *karakia* (Morrin et al. 2005). The research team also expresses concern about possible stereotyping in the media if its study findings are interpreted as proof of 'Māori genetic susceptibility' to certain diseases. This political context could lead to further discrimination and 'blaming the victim'. To prevent this, researchers will be careful not to extrapolate their findings beyond the community they are studying (Hudson et al. 2007).

However, the lead researcher, Rod Lea, generated worldwide controversy in 2006 when he presented findings from a separate research project that found that Māori were twice as likely as non-Māori to carry a gene associated with alcohol and tobacco use (Lea 2006). The particular polymorphism of the gene that encodes the enzyme monoamine oxidase (MOA) has also been associated with risk taking and aggression, and is consequently known as the 'warrior genes' (Stoke 2006). This episode was widely reported in the international media as proving that Māori were genetically predetermined to be violent, a depiction that the researchers argued was a misrepresentation of their research (Lea & Chambers 2007). A recent critique by a Māori academic argued that linking the MOA allele with high levels of violence among Māori is scientifically unsound, effectively makes



being Māori a ‘disease’, and may lead to genetic and racial discrimination by insurance companies. Further, ‘contributions to racial stereotyping by trained scientists are unethical and scandalous’ (Hook 2009:6).

This leaves us with a dilemma: are we to perceive Rod Lea as the culturally appropriate lead co-researcher of the Rakaipaaka Health and Ancestry Study or as the proponent of the potentially damaging ‘warrior gene’ hypothesis? Rather than a split personality, this apparent contradiction may reflect the dangers of conducting Indigenous genetic research (particularly in areas such as behavioural traits) in an environment in which both long-held racial stereotypes and health inequalities are widespread.

Consequently, the greater levels of controversy surrounding Indigenous genetic health research in New Zealand and North America may simply reflect the fact that more genetic research has taken place in those Indigenous populations relative to Aboriginal and Torres Strait Islander communities. This paper has considered the lessons that international experience in Indigenous genetic research offers in the hope that such research in Australia can reap the benefits while avoiding the pitfalls. What is clear is that developing relationships of trust between Indigenous people and non-Indigenous researchers, negotiating shared control and understanding of research projects, and articulating the potential risks and benefits the research poses to both the community and the researchers are the building blocks of partnerships that have the capacity to conduct culturally sensitive genomics research.

## *Summary of International Guidelines for Indigenous Genetic Research*

*Emma Kowal and Lobna Rouhani*

### **Introduction**

Genetic research in Indigenous communities must follow established guidelines for health research in general. Countries including New Zealand/Aotearoa, Canada and Australia have also produced detailed guidelines for the conduct of health research in Indigenous communities (NHMRC 2003; CIHR 2007; HRCNZ 2008). They include measures such as the need to consult with the community in question on the aims and design of the research; to involve community members in the research process as much as possible (for example, as co-investigators, reference group members or employees); to gain formal permission from a valid community representative body, as well as individual informed consent; to feed results back into the community; to ensure the community benefits from its participation; and to consult with community representatives on the appropriate manner of reporting and disseminating research results.

Genetic research poses specific concerns to Indigenous communities as well as other ‘socially identifiable’ groups (Foster & Sharp 2000; Arbour & Cook 2006; Crampton 2007; Tsosie & McGregor 2007). This review considers issues specific to genetic research in Indigenous communities that are covered in national guidelines referred to above. North America has no established guidelines

for this kind of research, so this paper discusses a few key journal articles that provide advice about the conduct of genetic research in Native American communities (Sharp & Foster 2002; Burhansstipanov, Bemis & Dignan 2002).

### **Health Research Council of New Zealand**

New Zealand/Aotearoa has been the site of a substantial body of research on Māori responses to a range of genetic issues, including genetic engineering, genetically modified foods and crops, genetic testing and genetic research (Gardiner 1997; Cram, Pihama & Barbara 2000; Mead & Mead 2003; Du Plessis et al. 2004; Roberts et al. 2004; Gillett & McKergow 2007; Hudson et al. 2007). The Health Research Council of New Zealand (HRCNZ) prepared the *Guidelines for Researchers on Health Research Involving Māori* (HRCNZ 2008), which includes a section on genetic studies involving Māori participants. The guidelines consider:

- **Research within paradigms of ‘Māori world view’:** genetic research is an extremely contentious issue among many Māori. Attitudes range from acceptance to total rejection. Intending researchers should familiarise themselves with the issues outlined in the 1995 HRCNZ Consensus Development Conference report *Whose Genes Are They Anyway?* (Baird et al. 1995). Māori at this conference expressed ‘support for genetic research that enhances quality of life for Māori as defined by Māori... [if that research occurs] within the paradigms of a Māori world view’.
- **Local support and approval:** genetic studies require access to both *whakapapa* (genealogical) knowledge and blood or tissue samples. The development of such studies requires close co-operation between the research team and the *whanau* (family) concerned, as well as thorough consultation with the *iwi* (clan) to which the *whanau* belong. This process can be lengthy and occasionally difficult, but if undertaken in good faith such consultation will ensure the commitment of all parties to the project and minimise the potential difficulties in such a study. Genetic research projects involving Māori require the approval of the *iwi* or *hapu* (sub-clan) organisation representing the *whanau* involved. This approval could be obtained in a *hui* (meeting) of the representative body following discussion of the project and should be documented in writing.
- **Consent and collectivity:** it is extremely important to be aware that informed consent for research should, in many cases, be gained from more than just an individual. It is also important for researchers to be aware that even though they may have received consent to gather *whakapapa* knowledge and/or blood, hair, saliva, tissue or other human samples from a *whanau* member, *whanau* or even *hapu*, the findings that result from such research should not be generalised to the Māori population at large (adapted from HRCNZ 2008:19).

### **Canadian Institutes of Health Research**

In 2007 the Canadian Institutes of Health Research published a set of comprehensive *Guidelines for Health Research Involving Aboriginal People* (CIHR 2007).

Genetic research is not specifically mentioned in the guidelines. However, sections 2.12 (containing Articles 12.1 to 12.5 inclusive) and 2.13 (Article 13), reproduced below, concern the use and storage of biological samples. These sections are relevant to genetic research and draw on the work of medical geneticist Laura Arbour (who was on the Aboriginal Ethics Working Group, which authored

the guidelines). Section 2.13, in particular, is based on the concept of biological samples being ‘on loan’ to researchers (Arbour & Cook 2006). This concept stipulates that researchers should act as ‘stewards’ rather than owners of any DNA samples, and that research participants must inherently retain the right to access data obtained from their samples.

### ***2.12 Initial and secondary use, proprietary interest, and storage and transfer of data and biological samples***

*Article 12.1* – A researcher should recognize and respect the rights and proprietary interests of individuals and the community in data and biological samples generated or taken in the course of the research.

*Article 12.2* – Transfer of data and biological samples from one of the original parties to a research agreement, to a third party, requires consent of the other original party(ies).

*Article 12.3* – Secondary use of data or biological samples requires specific consent from the individual donor and, where appropriate, the community. However, if the research data or biological samples cannot be traced back to the individual donor, then consent for secondary use need not be obtained from the individual. Similarly, if research data or biological samples cannot be traced back to the community, then its consent for secondary use is not required.

*Article 12.4* – Where the data or biological samples are known to have originated with Aboriginal people, the researcher should consult with the appropriate Aboriginal organizations before initiating secondary use.

*Article 12.5* – Secondary use requires REB [Research Ethics Board] review.

Much of the criticism directed towards research involving Aboriginal populations stems from the loss of control of data or of biological samples collected from Aboriginal people. Additionally, serious concern has been raised over the inappropriate use of stored biological samples, including DNA and cell lines, for unauthorized research. Ownership of traditional and sacred knowledge should always remain with the community.

These guidelines set out basic principles for the collection, disclosure, use and transfer of data and biological samples. The details of safeguards protecting the privacy and confidentiality of data and biological samples should be negotiated as part of the research process and specified in a research agreement. Subject to the community’s views on traditional or sacred knowledge, co-ownership of data and samples between researchers and communities is recommended.

If there is to be transfer of the data or biological samples to a third party, this should be done only with the consent of the researchers, the individual participants and the community. If the third party is to engage in secondary use of the transferred data or samples, then a further consent to that use must be obtained. Consent should address how confidentiality and privacy will be respected.

In any case, secondary use of the data or biological samples requires new consent unless such use is specifically agreed to in the research agreement.

Notwithstanding the above, individuals retain the right to access data about themselves (CIHR 2007:24–5).

### **2.13 Biological samples on loan**

*Article 13* – Biological samples should be considered ‘on loan’ to the researcher unless otherwise specified in the research agreement.

This Article reflects Aboriginal philosophies regarding ‘full embodiment’, in which it is held that every part and product of the body is sacred, and constitutes an essential part of the person.

Most Canadian Aboriginal communities advocate a participatory approach in the collection, use, storage and potential future use of human biological samples. These should be negotiated as part of the research agreement. In keeping with this model, the researcher needs to understand that his or her beliefs may not be reflective of the community’s and should be respectful of the latter.

Therefore, the researcher should be considered the steward, rather than the owner, of the samples.

The research agreement and consent process should address the conditions of collection, place of storage, research lab/researcher involvement, industry roles, plans for governance and potential future use.

Unless samples have been destroyed or anonymized, requests by an individual to withdraw, return or dispose of samples should be accommodated, in accordance with the terms of the research agreement and any applicable law.

In the case of existing tissue banks, consultations with the community and the individuals should be held to determine under what circumstances the samples can be used for future research (CIHR 2007:25–6).

### **Genetic research in Native American contexts**

As far as we are aware, no official guidelines exist for the conduct of health research in Native American communities. However, Linda Burhansstipanov and colleagues present a useful list of issues relating to the conduct of genetic research based on a series of consultations held between 1995 and 1999 with Native Americans (Burhansstipanov, Bemis & Dignan et al. 2002). They highlight a number of contextual issues, such as the need for better science education for Native students and greater numbers of Native geneticists. They also discuss a number of issues that are common to all Indigenous research projects, including the need to ensure that the research topic is a priority for the community concerned; the need to consult with community elders on the cultural appropriateness of research methods, such as the collection of blood or hair; the need for community representatives to approve publications to ensure the community is portrayed in an acceptable manner; and the need for cultural awareness training for geneticists. They also raise some less discussed issues such as the pressures faced by Native American geneticists to conduct research that some consider insensitive, and the need to mentor Native scientists and limit their administrative responsibilities (Burhansstipanov, Bemis & Dignan et al. 2002:154-156).

Sharp and Foster (2002:165) argue that guidelines for the conduct of genetic research in Native communities must strike a balance between ‘minimizing harm, treating sample contributors with respect, and promoting intellectual freedom to pursue a range of research questions’. Their review of existing guidelines for human research in the United States notes that guidelines that focus on human research tend to take minority groups into account but do not consider the implications for collecting large numbers of samples for potential multiple uses (including biobanks). Conversely, guidelines that focus on large collections of biological material for multiple purposes focus on the

individual donor and ignore the potential risks for identifiable communities. On the basis of their review, Sharp and Foster (2002:174–180) make five recommendations:

- community representatives should be consulted about study design, recruitment strategies, sample collection procedures and repository policies for distributing biological materials
- sample collection should proceed in a culturally sensitive manner; anticipated risks to identifiable communities should be discussed with prospective contributors during the informed consent process
- potential uses of stored biological materials should be clearly defined before sample collections are initiated and secondary sample uses should be discussed with contributors; mechanisms should be established to permit subsequent withdrawal of samples
- assessments of potential research applications should assign a higher priority to those uses that may potentially benefit contributing communities; clear policies on commercial applications should be defined prior to sample collection
- communication between sample-contributing communities and those who maintain the biological repository should not end after the samples are collected; where appropriate, researchers should allow for community review of findings prior to release.

### ***Australian guidelines***

Genetic research in Australia must comply with the *National Statement on Ethical Conduct in Human Research* produced by the National Health and Medical Research Council (NHMRC 2007). The National Statement includes a section specifically relating to human genetic research (Section 3.5; note that other sections specifically relevant to genetic research are the sections on databanks (3.2) and human tissue samples (3.4); also see Section 4.7 on Aboriginal and Torres Strait Islander research). The National Statement considers that genetic research requires particular ethical attention because:

- many of an individual's genes are shared with close genetic relatives (commonly called 'blood relatives') and with unrelated people in the population; and
- genetic research can reveal information about predispositions to disease. Although people with such a predisposition may not develop the disease, the information may have implications for their access to employment and education and to benefits or services, including financial services such as banking, insurance and superannuation. The information may also have similar implications for blood relatives (NHMRC 2007:41).

The National Statement advises various measures to manage these risks inherent to genetic information. Measures include constructing a plan for how the researchers will manage a situation where previously unknown paternity or maternity (or non-blood relationships to siblings) is detected in the course of the research; ensuring that data is held securely and that participants are aware of any statutory requirements to disclose information; and that only personnel qualified in clinical genetics and/or genetic counselling provide genetic information to research participants.

Two aspects of the statement are particularly relevant to Indigenous research. The first states that when:

*complex socially significant characteristics or the genetic characteristics of communities are being investigated, there is a risk that the research may be misrepresented or misused in ways*

*that lead to prejudice, disrespect or other harm to participants or communities* (NHMRC 2007:43).

Researchers are advised to plan to minimise this risk.

Second, the statement requires researchers to seek community, as well as individual, consent where:

- a. researchers propose to collect genetic material and information from individuals who are chosen because of their membership of a particular community;
- b. the research involves sensitivities for that community; and
- c. there is known to be a culturally relevant community structure involved in such matters (NHMRC 2007:44).<sup>2</sup>

The NHMRC provides guidance to Indigenous health researchers through *Values and Ethics: Guidelines for Conduct of Aboriginal and Torres Strait Islander Health Research* (NHMRC 2003). The guidelines do not discuss genetic research, but do highlight it as an issue that needs to be discussed. Further, the document does not provide guidance on the use and handling of biological samples, as the Canadian guidelines do (see above).

The Australian Institute of Aboriginal and Torres Strait Islander Studies produced *Guidelines for Ethical Research in Indigenous Studies* (GERIS) (AIATSIS 2000), which presents the ethical principles of research in Indigenous contexts. Although the AIATSIS guidelines are not intended to guide the conduct of health research, they are often considered useful for some health research contexts (in addition to the NHMRC guidelines).

The GERIS is currently being reviewed, and it is included here because the discussion paper associated with that review explicitly mentions human genetic material. The discussion paper (AIATSIS 2010) mentions human genetic material within a discussion of Indigenous Cultural and Intellectual Property:

*The term Indigenous Cultural and Intellectual Property (ICIP) refers to the whole range of Indigenous cultural heritage—especially its intangible components—for which Indigenous people seek protection. This has been described by Janke (1998) as ‘the intangible and tangible aspects of the whole body of cultural practices, resources and knowledge systems developed, nurtured and refined by Indigenous people and passed on by them as part of expressing their cultural identity’. It includes:*

- Literary, performing and artistic works (including music, dance, song, ceremonies, languages, symbols and designs, narratives and poetry);
- Scientific, agricultural, technical and ecological knowledge (including cultigens, medicines and sustainable use of flora and fauna);
- Spiritual knowledge;
- All items of moveable cultural property including burial artefacts;
- Indigenous ancestral remains;
- Indigenous human genetic material (including DNA and tissues);

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<sup>2</sup> Note that the recent Biobanks Information Paper (NHMRC 2010) also discusses the importance of cultural sensitivity in recruitment, consent and data management, but does not specifically address the needs of Aboriginal and Torres Strait Islanders.

- Cultural environment resources (including minerals and species); and
- Immovable cultural property (including Indigenous sites of significance, sacred sites and burials); and
- Documentation of Indigenous peoples' heritage in all forms of media. (including scientific, ethnographic research reports, papers and books, films, sound recordings) (AIATSIS 2010:12).

The discussion paper advises that ICIP is not the same as Western intellectual property and should be managed with:

*a 'knowledge trust agreement' which is an agreement between all parties to a research project about what elements of knowledge and intellectual property they hold, and bring to a project, and how that will be managed through the course of the project (AIATSIS 2009:13).*

This association of human genetic material with other forms of ICIP, such as Indigenous ecological knowledge and performance, is controversial. In considering whether the Convention for Biological Diversity should cover human genetic resources (it currently excludes it), Schroeder and Lasen-Diaz (2006) argue that there are essential differences between human and non-human genetic resources and that health research ethics are adequate and appropriate for human genetic research.

The view that human genetic material is cultural and intellectual property has been influenced by responses to the Human Genome Diversity Project conducted in the 1990s. The report cited in the discussion paper above—*Our Culture, Our Future: Report on Australian Indigenous Cultural and Intellectual Property Rights* (Janke 1998)—was commissioned by the Aboriginal and Torres Strait Islander Commission (which was disbanded in 2004). The report discusses the issue of human genetic material in section 3.8. Janke (1998:28) argues that:

*Indigenous people are concerned about their lack of control over the use of their genes and tissues in genetic testing and screening projects, such as the Human Genome Diversity Project... They feel they are being exploited, as their genes are used for research without their control or ownership and often without their knowledge or consent as a group. Under the existing framework of intellectual property rights, Indigenous peoples cannot control the use of the genetic material taken from them.*

Like other commentators, Janke highlights concerns over generating information about a family, clan or 'Indigenous people' in general when individual Indigenous people participate in genetic research without the permission of the collective; the potential for genetic racial discrimination if genetic research is used to argue that 'Indigenous people' are susceptible to alcoholism or cancer; and patenting genes and lack of benefit sharing (Janke 1998:28–9; Cunningham 1998; MacIntosh 2005; Mead & Ratuva 2007).

As this report illustrates, from one perspective issues relating to genetic health research are strongly tied to population genetic research and even archaeological research involving DNA from human remains.

## **Summary**



Genetic material and information is considered to be particularly sensitive in health research ethics. In Indigenous health research, these sensitivities are compounded by the historical context of colonisation and the contemporary power differential between Indigenous people and the broader society and cultural differences in attitudes towards inheritance and genetics. Contemporary examples of the perceived misuse of genetic research in Indigenous health contexts (primarily the Havasupai case in the United States of America<sup>3</sup> and the warrior gene controversy in New Zealand (McGregor 2007; Hook 2009) further compound any sensitivities that may exist.

Although there are important differences in the context and purpose of the different guidelines reviewed here, the following issues are prominent:

- genetic research must be conducted with awareness and respect for local Indigenous cultural and spiritual beliefs
- because genetic material can provide information about other people beyond the research participants, community consent for the research and control over how the research is reported is important
- researchers and communities should negotiate a model of ownership of genetic samples and/or the intellectual property arising from the samples that gives research participants control over the current and future use of samples, and ensures that a benefit-sharing arrangement is established where appropriate
- for some people there is an association between human and non-human genetic material (in that they are both seen as forms of cultural property), and subsequently an association between medical genetics, population genetics and archaeological genetics (in that they all seek to access Indigenous cultural property).

This review has highlighted the ways that guidelines for Indigenous genetic research have been considered internationally. Questions to consider further are whether the current *National Statement on Ethical Conduct in Human Research* adequately covers these issues and their implications, whether a revised version of *Values and Ethics: Guidelines for Conduct of Aboriginal and Torres Strait Islander Health Research* should include discussion of genetic research (or perhaps biological samples in general, as in the Canadian guidelines), or whether a separate document specific to genetic research in Indigenous communities would be helpful.

## ***Review of Summary of International Guidelines for Indigenous Genetic Research by E. Kowal and L. Rouhani***

*Don Chalmers*

1. This is a sound paper identifying key articles on Indigenous genetic research, particularly the paper by Tsosie and McGregor (2007) in the *Journal of Law, Medicine and Ethics*.
2. This paper asks the fundamental question whether the current *National Statement on Ethical*

<sup>3</sup> This case involved the Havasupai tribe of Arizona who live in the base of the Grand Canyon. They participated in genetic research into diabetes in the early 1990s, and their samples were used for a range of other research projects they did not specifically approve. Tribal members began legal action against Arizona State University and were awarded \$700,000 in a settlement in 2010 (see Mello & Wolf 2010).



*Conduct in Human Research* (NHMRC 2007), along with the *Values and Ethics: Guidelines for Conduct of Aboriginal and Torres Strait Health Research* (NHMRC 2003), provides adequate and proper guidance on genetic research among Indigenous communities in this country. The paper asks whether new or revised guidelines may be required for genetic research on Indigenous communities. In general terms, I hold the view that the current National Statement and Values and Ethics guidelines are both valuable general documents, but are not specific on Indigenous research. Some form of additional guidelines, perhaps by way of a guidance note to supplement the Values and Ethics guidelines, are desirable and worthy of discussion.

Briefly, separate guidelines or a guidance note require some justification. New guidelines would be a national reference point for ethical genetic research on Indigenous communities. Second, such guidelines could particularise the unique issues on genetic research in Indigenous communities, rather than relying on the more general guidelines on genetic research and databases, included currently in the National Statement. Third, the guidelines/guidance note could establish firm foundations for the ethical relationships between researchers and Indigenous communities, avoiding the generalisations implicit in the National Statement. Fourth, the issue of DNA and tissue sampling could be specifically contextualised to Indigenous communities. In this respect the Canadian guidelines are helpful. The current National Statement is too general to apply to some of the specific points about Indigenous communities raised during the Roundtable discussion of Friday 2 July. In this respect, the five salient points made in the Sharp and Foster *Jurimetrics* article in 2002, annotated in the paper, are not fully addressed in the current National Statement or Values and Ethics.

3. The paper could perhaps include reference to some key developing principles in relation to genetic research and biobanking of tissue and data. These principles have particular importance to Indigenous communities and are consistent with the points made in the paper. These principles are:

#### 3.1 Trust

In the 'Genetic research and Aboriginal and Torres Strait Islander Australians' paper (above) there is an excellent example of a breakdown in Indigenous trust in research in Victoria; similarly, revelations about the comments of Lea in New Zealand. Trust has become a key concept in genetic research in general and has been widely discussed in the development of biobanking guidelines. Trust is a key element in the *Governance Framework* for the UK Biobank (UK Biobank 2007). Similarly, the *OECD Guidelines on Human Biobanks and Genetic Research Databases* (OECD 2009) makes some reference to the concept.

#### 3.2 Benefit sharing

The paper mentions the need to establish proper relations between researchers and Indigenous communities and for benefits to flow from the research to the communities. The work of Bartha Knoppers is mentioned in relation to benefit sharing. There are now specific official reports and references to benefit sharing, particularly in the 2005 UNESCO Universal Declaration of Bioethics and Human Rights. This is an important concept that aims to reverse historical patterns of paternalistic and exploitative research on Indigenous communities. The concept of benefit sharing establishes a more equal and contractual base in the conduct of research.

4. Reference could be made to other guidelines. The *OECD Guidelines on Human Biobanks and Genetic Research Databases* has been mentioned. This was the product of wide international consultation. These guidelines establish the importance of engagement in consultation with communities. The guidelines expect researchers to take into consideration engagement methods that recognise the special attributes of the community, and its need for information, understanding and agreement to the research.

Second, the OECD guidelines are important when establishing storage (biobanks) of tissue. It is inevitable that genetic research will involve the collection of human biological materials and data that will be stored. Storage must be properly legally and ethically regulated and future use of the tissue also regulated. In this respect the concept of 'loan' in the Canadian guidelines is helpful. Importantly, governance arrangements generally must be established in a manner that accommodates the specific needs of the Indigenous communities.

5. These brief general comments can be developed and expanded should a decision be made to develop guidelines or a guidance note to supplement the Values and Ethics guidelines.

## ***Genetic Literacy in Indigenous Communities***

*Emma Kowal and Lobna Rouhani*

### ***Introduction***

There are many factors that are required for the successful conduct of genetic research in Aboriginal and Torres Strait Islander communities. One of these is the ability of Indigenous representatives and research participants to understand the genetic concepts that underlie both a particular genetic research project and the risks and benefits common to all genetic research. This discussion paper explores the concept of genetic literacy, the tools used to measure it and the tools used to enhance it. The paper aims to inform discussion on whether genetic literacy is a useful frame to guide the development of culturally safe models of genetic research.

### ***What is health literacy?***

Genetic literacy can be conceptualised as one type of health literacy, so it is useful to consider how health literacy is defined. The United States Institute of Medicine has defined health literacy as 'the degree to which individuals have the capacity to obtain, process and understand basic health information and services needed to make appropriate health decisions' (Institute of Medicine 2004). It sees health literacy as determined by individual capabilities within four separate domains: cultural and conceptual knowledge, speaking and listening skills, writing and reading skills, and numeracy. Health literacy is strongly related to overall literacy and socioeconomic status (Institute of Medicine 2004). The two most prominent tools for measuring levels of health literacy are the Rapid Estimate of Adult Literacy in Medicine (REALM) (Davis et al. 1999) and the Test of Functional Health Literacy in Adults (TOFHLA) (Parker et al. 1995). Higher levels of health literacy have been found to correlate with measures of health service utilisation and health outcomes, including 'effective management of chronic disease, compliance with medication and other health advice, and participation in health and screening programs' (Weiss 2005; Keleher & Hagger 2007; Nutbeam 2008:2073). Poor health literacy

can be addressed in two main ways: with educational interventions for health consumers and with changes to the organisation and delivery of health care.

In a recent review, Nutbeam (2008) classifies this conventional model of literacy as a 'risk' model because poor health literacy is considered a risk condition. He proposes an alternative 'asset' model of health literacy that is based on empowerment. Rather than seeing health literacy as skill based and determining appropriate use of health care services and compliance with medical advice (involving only 'functional' literacy), this model sees health literacy as the outcome of 'higher' levels of literacy ('interactive' and 'critical' literacy) that allow one to critically reflect on the social determinants of health and help motivate others to make healthy decisions through negotiation and advocacy. Importantly, Nutbeam argues that while the 'risk' model of health literacy is relevant only to clinical situations, the broader 'asset' model has potential applications beyond clinical care.

### ***What is genetic literacy?***

Genetic literacy has been discussed in two different contexts. First, it has been considered as a form of health literacy necessary for making decisions about genetic-related health care such as genetic testing. Second, it has been argued that the general population needs a high level of genetic literacy to understand and participate in debates about genetic modification of organisms, genetic screening and genetic discrimination (Knoppers 2002; Barash 2008). While this wider notion of genetic literacy is arguably related to Nutbeam's 'asset' model of health literacy, it is generally the health-related conception of genetic literacy that has been explored in the literature. Therefore, mainly the health-related form of genetic literacy is considered here.

Although over the past 15 years, issues such as the Human Genome Diversity Project, prenatal genetic diagnosis and human cloning have meant genetics have received an abundance of media attention, this does not mean that there is a great understanding of how genetics can impact health. In other words, despite increased exposure to genetics, many lay people struggle to gain a real understanding of how genetics can impact them and their families (Bowling et al. 2008).

Genetic literacy is usually taken to mean the ability to understand genetic concepts communicated in a clinical setting for the purposes of health care. The communication of complex genetic concepts is an increasingly common and daunting task for medical professionals, not least because genetic literacy now requires not just a reasonable level of scientific literacy but also statistical literacy (Erby 2007; Monahan 2008; Lea et al. 2010). Literature on the topic reveals that health workers are challenged by the task of discussing genetic concepts with people and that the task is even more difficult when dealing with minority or socially identifiable groups (Saleh et al. 2009).

Although there is no consensus on the definition of 'genetic literacy', many see it as beyond merely understanding genetic terms and concepts. Rather than knowledge outcomes, scholars focus on the ability to use genetic information. Jennings (2004:38), for example, defines genetic literacy as the 'ability to understand and act to protect one's needs and interests', while Molster (2009:85) states that it is the 'capacity or empowerment for democratic and informed decision-making and participation around genetic issues'. McNerney (2002:372) approaches a definition of genetic literacy by asking:

*what knowledge and skills does the average person require to manage uncertainty and to participate as a full partner in a prevention-based health care system that is increasingly informed by genetic perspectives?*

There is a paucity of literature that considers whether and how genetic literacy translates to the context of health research rather than health care. The process of informed consent is the most common place for genetic literacy to be mentioned in the context of genetic research, particularly where the research concerns non-English speaking and/or non-White groups (Gong 2008). The definitions of genetic literacy discussed above suggest that the concept can be translated in the research context within the broad scope of 'participation'. This would include the individual decision to participate in a genetic research project. Genetic literacy in this context may be defined as 'the state of having sufficient knowledge and understanding of genetic concepts to make an informed decision about participating in a genetic research project'.

However, where genetic research involves socially identifiable groups, genetic literacy would also be required when collective bodies make a decision to participate or decline to participate in research. This process of consultation and negotiation between genetic researchers and community representatives occurs at the start of a project, usually prior to receiving ethics approval and sometimes before funding is sought (NHMRC 2003, 2006). At this level of community representation, genetic literacy may be defined as 'having sufficient knowledge and understanding of genetic concepts to make an informed decision about the participation of the *group* I represent in a genetic research project'. This second level of genetic literacy, in particular, approaches Nutbeam's 'asset' model of health literacy because it requires consideration of the possible long-term implications of participation for the relevant community, as well as advocacy and negotiation.

### ***Why is genetic literacy important?***

Competence in genetic literacy is necessary for making good decisions that involve genetics, whether these decisions involve individual health care, individual or collective decisions about participating in genetic research, or wider public debates.

In the context of health care, the transformation of medicine has necessitated a transformation in genetics education with a focus on the development of genetic literacy that encourages collaboration between patients and medical professionals for the purpose of health promotion and disease prevention (McInerney 2002). Erby (2007:174) argues that low levels of genetic literacy 'constitute a significant barrier across the health services spectrum with consequences in virtually every public health and medical care domain'.

Many people lack the genetic literacy required to make good decisions. Although much written genetic health information (of both good and poor quality) is available, many people in the general population lack the general literacy, health literacy and/or genetic literacy to benefit from this information. In the United States, for example, 43 per cent of adults have only basic or below basic reading skills and 36 per cent have only basic or below basic health literacy levels (Kutner 2006). A recent study of genetic literacy among 1200 Americans of varied ethnic/racial origin found that all respondents lacked a good understanding of eight key genetic concepts, with the highest number (70 per cent) correctly understanding the genetic similarity of twins and siblings, and less than half of respondents correctly understanding the other seven concepts. Less than one-quarter of respondents correctly understood that 'races' are not genetically distinct and that behavioural traits are not determined by single genes (Christensen et al. 2010).

The Australian Bureau of Statistics reports that 59 per cent of adults in Australia have poor or very poor health literacy skills, and that in general people in this group are those who lack a formal

education or are unemployed, or where English is not their first language (ABS 2008). This reinforces the notion that health literacy is intimately connected with socioeconomic status. Moreover, a study conducted in Western Australia that aimed to determine the public's knowledge of genetics and health found that around half of the respondents (46 per cent) stated that they knew a 'small amount' about genes and health, while 29 per cent stated that they knew 'not very much' about this topic (Molster 2009). Those who were more comfortable with genetic content were likely to have a higher socioeconomic status.

Neither health literacy nor genetic literacy levels in the Indigenous population are available. However, the Survey of Aspects of Literacy conducted by the Australian Bureau of Statistics showed that approximately 44 per cent of Indigenous Australians had low literacy levels compared with 19 per cent of non-Indigenous Australians (ABS 1996). Therefore it is likely that health literacy and genetic literacy levels are also lower in Indigenous populations.

In addition to varying literacy rates, cultural difference is also a factor in poor communication about genetics in the clinical setting (Browner et al. 2003). Sivell et al. (2008) argue that cultural beliefs or assumptions, which may conflict with genetic knowledge, can influence the understanding of risk information. African-American cancer patients have expressed a desire for culturally appropriate education materials that are 'personalized and made relevant to the lives of the target population' (Baty, Kinney & Ellis 2003:146). Increasing genetic literacy through targeted genetic literacy tools and better systems (of both research and health care) will improve the effective level of genetic literacy in Indigenous and other minority populations.

In a research context, enhancing genetic literacy will enable more meaningful partnerships between communities and researchers at all stages of the research project, in addition to being the basis of a sound informed consent process. Taussig (2007:200) points out that the purpose of training Native American college students in cultural and genetic issues as part of the Genetic Education for Native American Project was to facilitate an 'active, not passive informed consent'.

From the perspective of researchers, genetic literacy is also important for ensuring that the project is accepted by the community, and that levels of participation are maximised. A study of White and African-American participants in genetic research found that African-Americans were less likely to give a blood sample and this was explained by lower levels of trust in researchers (Bussey-Jones et al. 2010). Furthermore, other research that aimed to compare African-American and White attitudes to genetic research revealed that African-Americans were more likely to perceive genetic research as harmful to society (Furr 2002), and were more likely to believe that clinical trials may be dangerous and that the federal government conducted unethical research (Achter, Parrott & Silk 2004).

Although it is possible that African-Americans and other minorities are making informed choices not to participate in genetic research, relatively lower levels of genetic literacy may also play a role. Given the low levels of trust in the Indigenous community in relation to health research (VKHRCU 2000),<sup>4</sup> it is important that efforts are made to ensure Indigenous people who are considering participation in genetic research have access to effective and culturally appropriate genetic literacy tools.

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4 This has been related to the history of Indigenous health research; see Thomas 2004. Note that in an African-American context, it has been found that knowledge of the Tuskegee research project is directly related to non-participation in medical research; see Shayers, Lynch and Burmeister 2000.



### ***What are the most common tools for measuring genetic literacy?***

In seeking to improve genetic literacy, it is important to have tools that measure it. The most popular existing tool for measuring genetic literacy is the Rapid Estimate of Adult Literacy in Genetics, or REAL-G (Erby 2007). This screening tool was developed for use in the clinical setting and is patterned on the widely used REAL-M. The tool is composed of sixty-three items in three groups of increasing complexity (Table 1). Patients are asked to pronounce the words aloud, or to say 'pass' if they are unable to read a word. The score is the number of correctly pronounced words. A shorter list of eight words can be used with slightly less specificity (that is, more people are erroneously judged to lack genetic literacy when the shorter tool is used as opposed to the longer tool). The short version is made up of the eight items that were most frequently missed and had at least a 0.5 item-scale correlation with the full version of the REAL-M. The tool was developed on a sample of 203 cancer and prenatal screening patients from several United States cities (consisting of 75 per cent African-American, 18 per cent White and 7 per cent 'other') (Erby 2007).

**Table 1: The REAL-G (note: the 8 items in the short version are marked with an asterisk (\*))**

List 1	List 2	List 3
Test	Detect	Syndrome
Egg	Lifetime	Condition
Male	Percent	Surgery
Risk	Onset	Ultrasound
Gene	Pattern	Disorder
Cell	Sibling	Carrier
Blood	Certain	Sporadic*
Sperm	Ethnic	Uterus
Change	Severe	Instruction
Chance	Needle	Insurance
Screen	Sample	Mutation*
Related	Fluid	Miscarriage
Trait	Protein	Development
Odds	Option	Variation*
Pap	Cystic	Chromosome*
Result	Negative	Generation
Working	Genetic*	Population
Normal	History	Retardation
Defect	Positive	Hereditary*
Likely	Infection	Abnormality*
Mistake	Affected	Susceptibility*

The REAL-G can be used to quickly identify patients with low genetic literacy in the clinical and research setting. The utility of the REAL-G is not only in its ability to identify patients with literacy deficits, but also in its ability to assist clinicians in more effectively meeting the informational needs of patients with such deficits. Ideally, health professionals (and researchers) who use the REAL-G and find that someone has low genetic literacy can respond by adjusting their communication practices to suit the patient's needs, paying particular attention to word choice and sentence structure (Erby 2007), providing clear and concise definitions, and simplifying concepts without losing their meaning (Molster 2009). However, further study is needed to explore ways in which providers can adjust their communication practices to meet these needs (Erby 2007). Further research would also need to establish whether this method of measuring genetic literacy is appropriate or useful in other settings, including Indigenous communities.

Another tool developed to measure genetic literacy is the Genetics Literacy Assessment Instrument (GLAI). Bowling and colleagues (2008) developed and evaluated an instrument to assess the genetic literacy of undergraduate students. It consists of a thirty-one item multiple choice test that tackles seventeen concepts considered to be critical to genetic literacy. The evidence for the validity and effectiveness of this tool, however, has only been shown for undergraduate students in the biological or genetic sciences (Bowling et al. 2008). There is currently no evidence that this tool can be extrapolated to the general population.



GLAI sample questions are as follows (the asterisk (\*) indicates the correct answer):

***Q5 What is the most likely way the genetic system (genetic material and the genetic code) of living organisms evolved?***

- a. The same genetic system repeatedly developed at different times in various organisms.
- \*b. One genetic system developed early in the evolution of life in all organisms and remained.
- c. One genetic system developed early but later changed into quite different genetic systems in different organisms.
- d. One genetic system developed but well after numerous different species existed.
- e. Different genetic systems evolved in different species.

***Q12 A woman has been told she carries a mutation associated with breast cancer. How does this influence her likelihood of developing breast cancer?***

- a. Her risk will be no different from any other healthy woman.
- b. She will likely not get breast cancer.
- \*c. She is at an increased risk for breast cancer.
- d. She will definitely get breast cancer.
- e. She already has breast cancer since she carries the mutated gene.

***Q15 Which of the following is a characteristic of mutations in DNA?***

- a. They are usually expressed and result in positive changes for the individual.
- b. They are usually expressed and cause significant problems for the individual.
- c. Those that occur in the body cells of a parent are usually passed on to their children.
- d. They usually occur at very high rates in most genes.
- \*e. They result in different versions of a gene within the population.

***Q22 Your muscle cells, nerve cells, and skin cells have different functions because each kind of cell:***

- a. contains different kinds of genes.
- b. is located in different areas of the body.
- \*c. activates different genes.
- d. contains different numbers of genes.
- e. has experienced different mutations. (Bowling et al. 2008:21)

While the REAL-G is more targeted to the general population than the GLAI, it is likely that even the REAL-G would need amendment for use in Indigenous populations. In particular, the short version is nearly all made up of the most difficult category of words and thus is less useful for measuring any changes in genetic literacy in a low-literacy population from a non-dominant cultural background.

### ***Tools for increasing genetic literacy in Indigenous populations***

A plethora of resources for mainstream genetic education exists. Some important hubs where



many resources can be found include websites for the Centre for Genetics Education ([www.genetics.edu.au/home.asp](http://www.genetics.edu.au/home.asp)), the United States National Human Genome Research Institute ([www.genome.gov/10000002](http://www.genome.gov/10000002)) and the National Health Service National Genetics Education and Development Centre ([www.geneticseducation.nhs.uk/](http://www.geneticseducation.nhs.uk/)). Although some of these resources focus on clinical genetics, there are many targeted at improving general genetics education.

The only known genetic literacy project specifically targeting Indigenous people is the Genetic Education for Native American (GENA) Project, a National Human Genome Research Institute project designed for Native American university students (Dignan, Burhansstipanov & Bemis 2005). The desire to educate Native communities was born from a reaction to requests from medical researchers for samples from Native Americans (Taussig 2007).

The goal of GENA was to provide ‘culturally sensitive genetics instruction to Native Americans to increase their awareness of genetics as a topic’, in addition to improving informed decision making about genetic issues including research and consent issues (Dignan, Burhansstipanov & Bemis 2005:517). Once a curriculum was developed, it was implemented in workshops in different areas throughout the United States in conjunction with scientific conferences that included a large number of Native American attendees. The original curriculum included twenty-four objectives illustrated in Table 2.

**Table 2: Original 24 GENA objectives, plus 5 new objectives (Dignan, Burhansstipanov & Bemis 2005:518)**

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**Original 24 GENA objectives, plus 5 new objectives**

- Objective 1 Examine selected Native American cultural/political issues (10 min)
- Objective 2 Review basic principles of cell biology and genetics; e.g., cell structure, location of DNA and RNA, protein expression, transcription, and translation (45 min)
- Objective 3 Examine examples of ethical, legal, and social implications that humanity faces in genomic research (50 min)
- Objective 4 Examine special concerns of Indigenous people (40 min)
- Objective 5 Identify the types of genetics research that are of interest/priority to Native communities (45 min)
- Objective 6 Identify current scientific, cultural, ethical, social, and legal resources to clarify the patenting process (45 min)
- Objective 7 Review genetics concepts (45 min)
- Objective 8 Understand classical patterns of inheritance and cultural traditions related to these patterns (60 min)
- Objective 9 Describe genetic testing (30 min)
- Objective 10 Examine selected Native American cultural and ethical issues related to genetic testing (60 min)

- Objective 11 Identify common misconceptions related to genetic testing (30 min)
- Objective 12 Analyse the benefits and risks of genetic testing (30 min)
- Objective 13 Determine factors that should be considered when deciding whether or not to take part in genetic testing (10 min)
- Objective 14 Examine current genetics-research-related issues and discoveries and their potential impact for Native communities (60 min)
- Objective 15 Understand the NIH Human Genome Project (50 min)
- Objective 16 Describe benefits and drawbacks to pharmacogenetics (60 min)
- Objective 17 Examine current botanical genomic research (30 min)
- Objective 18 Examine Native American cultural issues that are related to contemporary genetics research (30 min)
- Objective 19 Analyse the tribal research approval process relevant to genetics research (60 min)
- Objective 20 Describe guidelines and ways that future genetics research could be conducted to be culturally respectful and allow for informed decision making by Indian communities (30 min)
- Objective 21 Recognise the roles of the health care team involved with cancer genetic counseling (20 min)
- Objective 22 Describe culturally acceptable methods of collecting a family history (45 min)
- Objective 23 Examine selected ethical, legal, and cultural issues of genetic counseling (30 min)
- Objective 24 Describe how other Native and Latino/Chicano/Hispanic scientists have addressed challenges in their (1) education, (2) careers, and (3) family life (video)
- Objective 25a Identify advantages and limitations of selected models for human diseases (60 min)
- Objective 26a Describe parts of a cell (45 min)
- Objective 27a Describe components of the NHGRI HapMap Project (120 min)
- Objective 28a Describe potential benefits and drawbacks regarding participation of tribal nations in the HapMap Project (40 min)
- Objective 29a Distinguish between facts and myths of genetics issues of concern to Natives (60 min) a *New objectives*.

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The curriculum was developed by means of focus groups and extensive review of existing curricula and material. The development process reinforced the need to emphasise cultural perspectives related to genetic issues. The curriculum was then implemented either as sixteen-hour 'comprehensive' workshop over the course of two days, or as 'customised' three- to five-hour workshops. The comprehensive workshop covers the twenty-four GENA objectives, while the content of the customised workshop was tailored to suit the needs of the audience. Both of these workshops are presented by at least one individual with substantial genetics knowledge and another person with relevant training in cultural and scientific methods. In terms of knowledge assessment, both workshop formats were successful in increasing participants' knowledge

measured using pre- and post-test evaluations, where each knowledge item is an open-ended question designed to assess a specific workshop objective conducted across the same time intervals for both workshop formats; for example:

#### **Examples of pre- and post-workshop knowledge items**

What is a historical example of classical inheritance in everyday life?

Where would you find a centromere?

How would you refer to the short arm of chromosome 3?

What genetics research study is currently going on with the support/partnership of at least one Native American tribe?

What is one example of how a scientist should work with a local Native American community to demonstrate respect?

What type(s) of tribal approval are required for a research project to be conducted in a Native American community?

What is one of the issues concerning the use of 'cell lines' among Native American communities?

What is one concern among Native communities regarding patenting? (Dignan, Burhansstipanov & Bemis 2005:519).

For the customised workshops, the mean test score increased by 55 per cent, while there was a 38 per cent increase for the comprehensive workshops. More importantly, the cultural benefit of GENA has been recognised by students who have applauded the program for its effective integration of genetics, culture and ethics (Dignan, Burhansstipanov & Bemis 2005).

The content and format of GENA may be a useful approach for improving the genetic literacy of Indigenous leaders and community representatives involved in research. Health conferences where there are a significant number of Aboriginal and Torres Strait Islander people (such as the Northern Territory Chronic Disease Network conference and the Kulunga Research Network's National Aboriginal Health Worker Conference) may provide an opportunity to attract Indigenous leaders to a tailored genetics education workshop.

Within a research context, the major tool to improve genetic literacy is the Plain Language Statement (also known by other terms such as 'Information Sheet') provided by researchers to potential research participants. Although, in relevant research, these statements are essentially genetic literacy tools, details of the contents of such information sheets are rarely reported in the academic literature. One exception is Sheila van Holst Pellekaan's article (2000:68) on her experience of conducting ancestry related genetic research in western New South Wales. She reproduces part of her information sheet, which is in the form of a dialogue and is illustrated with simple pictures:

What have 'genes' got to do with it? What are they?

Sheila: Genes are made of chemicals called DNA. Each person has their own set of genes which control what we look like and how our bodies work.

All living things, plants and animals have DNA or 'deoxyribonucleic acid' — say 'dee-oxy-rye-bow-new-clay-ick asid'. 'DNA' is a short way of saying it.

Whole bodies have lots of tiny cells... like muscle cells or blood cells (van Holst Pellekaan 2000:68).

Another genetic literacy tool for Indigenous Australians has been developed by the Machado Joseph Disease Foundation. Machado Joseph disease (MJD) is a hereditary neurological disease in the same 'disease family' as Huntington's disease. It causes a progressive loss of motor abilities and sufferers are wheelchair bound and fully dependent within 10–15 years of the first symptoms occurring. It is autosomal dominant, meaning that children of affected people have a 50 per cent chance of inheriting the disease. It affects families living on the coast of Arnhem Land from Groote Eylandt to Oenpelli and has been present for at least four generations. The disease is thought to be Portuguese in origin and to have passed into Arnhem Land from Macassan sailors from Indonesia who visited the Arnhem Land coast to trade for hundreds of years until the early twentieth century.

The Machado Joseph Disease Foundation was set up in 2008 to provide support for MJD sufferers and the 400 people at risk of inheriting the disease. The foundation has produced a booklet that explains MJD and genetics more generally in English (MJDF n.d.) and in Anindilyakwa, the language spoken on Groote Eylandt. A sample of the text is shown here:

When a baby is born, most the way his body will grow and the way his body will move is in his flesh already worked out inside his tiny body.

The baby has a genetic 'story' stored inside its body. The 'words' in the story are passed on to the baby by both its mother and father.

There are some parts of the baby's story that we can see at the beginning. We can see what colour the baby's skin and hair are.

These parts of the baby's story are called '**genetic information**' (MJDF n.d.:2).

There are millions of 'cells' in our bodies.

Inside every cell is a plan that decides all the things that will happen inside the body. The plan is stored in the chromosomes...

The body's plan is stored in a special way.

There are always 23 pairs of chromosomes.

The first 22 chromosomes are the same in everyone. Chromosomes number 23 decides if you are a male or female (MJDF n.d.:7–8).

There is scope for drawing on existing tools for genetic education to develop basic genetic literacy tools for use in Indigenous health research projects. These could be in the form of sample Plain Language Statements and presentations for use at the negotiation phase of the research, as well as more intensive genetic education programs modelled on GENA.

## **Conclusion**

Genetic literacy may offer a useful way to think about how Indigenous people can be empowered to make informed decisions about both individual and collective participation in genetic research projects. A combination of tailored genetic education materials, effective models for delivering genetic education and culturally appropriate tools for measuring genetic literacy would provide a sound basis for the negotiation and conduct of genetic research projects in Indigenous communities.

# Discussions at the Indigenous Genetics Roundtable

*Graduate House, The University of Melbourne*

*2 July 2010*

*8.45 am–4.00 pm*

## Summary

Genetics is at the forefront of medical research, but it is rarely used in Indigenous health research projects. In the past, proposals to conduct genetic studies in Indigenous communities have been highly criticised and rarely funded (only twenty papers have been published on a variety of conditions since 1980). However, genetic research has the potential to improve Indigenous health in multiple ways: through understanding disease pathogenesis; using genetics to probe environmental risk; predicting disease risk; finding novel diagnostics and drug targets; and pharmacogenomics. The fact that genetics was mentioned by a number of participants in the Road Map consultations and is included in Road Map II indicates that this is an emerging research area of interest and concern.<sup>5</sup>

Understandably, many Indigenous people interpret genetic research in the context of their experiences of colonisation. Fears of genetic theft or ‘biopiracy’, fears that genetics will be used to determine Aboriginality and may fuel racism, poor access to the potential health care innovations that genetics may bring, bad experiences of the Human Genome Diversity Project (known by some as the ‘Vampire’ project) and struggles over access to DNA extracted from human remains all constitute barriers to effective research partnerships between Indigenous communities and genetic researchers.

On 2 July 2010 a Roundtable on Indigenous genetic research was organised by Emma Kowal (The University of Melbourne) and convened by Ian Anderson on behalf of the Lowitja Institute. There were twenty-four Indigenous and non-Indigenous attendees, including experts in genetics, Indigenous health research, Indigenous research ethics, genetic ethics and genetic literacy.

Discussions were honest and constructive and considered many difficult issues, including the sensitivities inherent to the reporting requirements of sample quality control (degree of heterozygosity or ‘inbreeding’ in the population, and corrections for population structure and level of ‘admixture’ with non-Indigenous populations); the risks involved when research findings are generalised to the entire Indigenous population; the existence of highly publicised examples of ‘bad’ Indigenous genetic research (including the Havasupai case in the United States and the ‘warrior gene’ controversy in New Zealand); and the need to provide short-term benefits to participating Indigenous communities given the long-term nature of the potential benefits of genetic research.

The risks involved in media reporting of results from genetic studies and the potential to elicit or aggravate existing racist attitudes in the community was discussed in particular. Opinions varied over whether these risks can be adequately managed through partnerships between researchers, Indigenous communities and Indigenous ethics committees.

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<sup>5</sup> The Roap Map is a document outlining the strategic priorities for Aboriginal and Torres Strait Islander health research and is the result of intensive community consultations with Indigenous communities across the country supported by the National Health and Medical Research Council (NHMRC 2010).

Various new strategies were discussed to address these issues, including the development of guidelines or a guiding note for Indigenous genetic research to complement the National Statement and Values and Ethics documents (advocated by Don Chalmers); genetic literacy tools for researchers working in Indigenous contexts; genetic literacy workshops (offered before or after relevant conferences) for Indigenous health workers and Indigenous leaders; media guidelines for reporting Indigenous genetic research findings; and cultural competence training for genetic researchers.

Realising the benefits of genetic research will require a program of Indigenous genomic research that combines scientific research (such as a high-powered Genome Wide Association Study (GWAS) enabled through collaboration between a number of research groups) with social science research into the ethical and cultural risks of genetics and the governance arrangements required to manage those risks. As a first step, the conversation between different stakeholders on Indigenous genetic research should be continued at another Roundtable or similar event.

### **Welcome, aims and overview**

**Moderator:** Ian Anderson, Director, Director of Research and Innovation, The Lowitja Institute and Director, *Onemda* VicHealth Koori Health Unit, The University of Melbourne.

Ian Anderson welcomed all attendees to the Roundtable and discussed the aims of the workshop:

- to create a community of interest around Indigenous genetic research
- to foster collaborative genetic research in Indigenous communities that is ethical, productive and of high quality
- to begin to consider the tools and models that are needed to make genetic research a culturally safe option for Indigenous people.

Anderson gave some background on the Lowitja Institute. One of the roles of the Lowitja Institute is to facilitate conversations on difficult topics:

*We have hosted conversations on lateral violence in Indigenous communities and on sexual violence. These are topics that are in the 'too-hard' basket that end up in unproductive conversations. But we know that it is important to have those conversations.*

### **Presentations**

Following Ian Anderson's introduction, presentations were given by:

- Jenefer Blackwell: Head of Genetics and Health, Telethon Institute for Child Health Research, The University of Western Australia
- Sheila van Holst Pellekaan: School of Biotechnology and Biomolecular Sciences, The University of New South Wales
- Emma Kowal: Research Fellow, Discipline of Anthropology, The University of Melbourne.

### ***Jenefer Blackwell***

Jenefer Blackwell began by acknowledging the Wurundjeri people, who are the traditional owners of the Melbourne area. She then discussed the many barriers to Indigenous genetic research. These include the fact that Indigenous people interpret genetic research in the context of their experiences of colonisation; fears of genetics being used to determine Aboriginality; fear of genetic

theft or biopiracy; concerns that poor access to health care will prevent Aboriginal people from benefiting from any medical breakthroughs resulting from genetic research; concerns generated by the conduct of the Human Genome Diversity Project in the 1990s, which called for research into the genetics of Indigenous groups before they ‘vanished’; and specific concerns about access to, and use of, DNA extracted from human remains of Indigenous ancestors.

Blackwell outlined the enormous advances in genomic research over the past two decades, partly as a result of the Human Genome Project (which is distinct from the Human Genome Diversity Project), which sequenced the human genome in 2000. She argued that genetic research has the potential to improve Indigenous health through improving our understanding of disease pathogenesis; using genetics to probe environmental risks; predicting disease risk; developing novel diagnostics and drug targets; and through pharmacogenomics (tailoring drugs to individuals).

She emphasised that genes and environment are intimately connected and can’t be separated from each other: ‘You sample your environment through your genes. Your genes determine your response to your environment (Genetics); your environment influences expression of your genes (Epigenetics).’

Blackwell then discussed the tool that is currently the basis of most genetic research, the GWAS. A Genome Wide Association Study typically examines more than 500,000 single nucleotide polymorphisms (SNPs) using a ‘SNP chip’. Either a case-control design or a family-based study is usually used with ideally a large number of participants (at least 2000). Since the year 2007, known by some as ‘the year of GWAS’, many articles presenting the results of GWAS have been published, and as of March 2010, 779 highly significant associations have been published on 148 traits.

Blackwell then discussed the various technical procedures that geneticists must perform on their data and their samples to ensure that the data are of high quality and that their study findings are true. She indicates the ‘some of these issues have implications that we might want to talk about’ during the Roundtable.

The first issue she raised was heterozygosity, which is the proportion of SNPs across an individual’s profile where they carry two different alleles. Geneticists need to know about this to make sure that the SNP chips will be effective in this population and to interpret their data correctly. Low or high levels of heterozygosity can occur for many reasons, including if there has been sample contamination in the laboratory. Blackwell presented some data comparing the heterozygosity of two different populations, one of which was lower than the other. She explained that the population with lower heterozygosity:

*can be explained by a founder effect, or it could be that we are using the wrong chips, or because of consanguinity [partnerships between people who are related in some way], or ‘inbreeding’, which is a nasty word that comes up.*

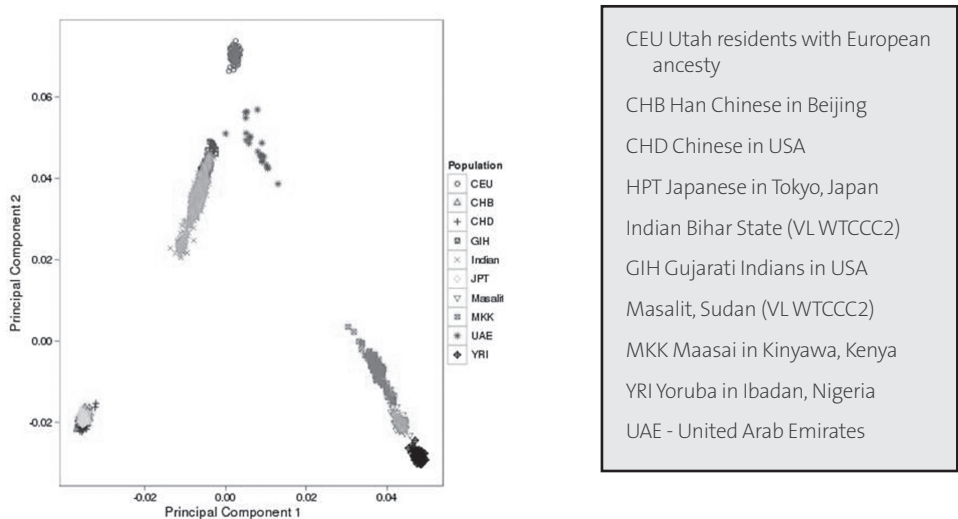
The second issue she raised was relatedness. Geneticists use analyses called ‘identity-by-descent sharing’ analysis to determine the level at which the study participants are related to each other. This may indicate a different (closer or more distant) relationship to that reported by the individuals themselves. This analysis may show that some people in the population are related in ways they did not realise. This information is not reported back to research participants but is needed to make sure that any results from the GWAS analysis are accurate.




The third issue is that geneticists need to work out what is called the ‘population structure’ using the method of Principal Component Analysis. Blackwell explained that ‘population structure can cause a problem’. Working out the population structure is required to ‘check matching of cases and controls, to control for mixed ethnicity in the association analysis, and to determine the origin of the disease associated allele/haplotype’. On the third point, she gave the example of the genetic neurological disease Machado Joseph disease, which is found in Arnhem Land and where genetic research has established that the origin of the disease gene is Portugal. Drawing on some of her work from India, she showed how Principal Component Analysis found that Hindu and Muslim people from one village she worked in were genetically distinct. If she had found a gene that was present in the Muslim population and not in the Hindu population, she might have mistakenly thought it was a disease gene when it really reflected a different population history.

Blackwell showed a graph presenting the results of Principal Component Analysis for a number of different populations. The graph is an upside-down ‘V’ with Asian populations on the bottom left-hand side, African populations on the right-hand side, and Caucasian populations at the top, with South Asian and Middle Eastern populations in between.

Figure 1: Principal components 1 and 2 for a range of ethnic populations







Blackwell then addressed the issue of whether disease genes that might be found in Indigenous Australian populations are likely to be different from disease genes found in any other populations. ‘Will we find anything novel by studying Indigenous Australians?’, she asked. Blackwell argued that out of fourteen diabetes genes found in Caucasian populations, only four have been found in Asian populations and only one has been found in the United Arab Emirates. The novel diabetes gene found in the United Arab Emirates is a gene that plays an important role in insulin secretion and may be a potential drug target. Based on experiences such as these, she argued that ‘there is a fair chance we would find novel genes in the Indigenous population’. This is one reason why it is important to do genetic research with the Indigenous population.

She then presented information from the database of genetic research projects that have been conducted in Indigenous Australian communities (prepared by Lobna Rouhani, Sarra Jamieson and Emma Kowal). Most genetic research reported in the literature has been conducted in Central Australia, the Kimberley, Arnhem Land, and north and central Queensland. There are a number of other studies where the location the samples were collected from is not specified beyond ‘Australia’.

One participant asked whether the communities in north Queensland would be aware of what was being done with their DNA. Blackwell deferred this question until later. (In fact, all the studies in the database of genetic research projects are different projects and issues relating to community consent would be different in each case.)

Blackwell also briefly mentioned other issues including data storage, sharing and linkage; the potential impact of ‘nextgen’ sequencing (the fact that within five years or so sequencing the whole genome will be economically viable on a large scale); and the impact of direct-to-consumer genetic testing.

Blackwell went on to share the experiences of data collection in a remote community in the mid-west of Western Australia. She outlined the 18-month process of consulting with the community, preparing a memorandum of understanding between the Telethon Institute for Child Health Research and the community, successfully applying for a research grant, and gaining ethics approval from the Western Australian Aboriginal Health Information and Ethics Committee (WAAHIEC). She discussed the experience of collecting ‘pedigrees’ (family trees) at the community. She described that while WAAHIEC was concerned that community members may be concerned about viewing family trees, especially where they include family members who have passed on, her team found that ‘they love looking at their family trees. WAAHIEC were very worried about their family trees but they love them.’ Finally, she outlined their current work on metabolic disease and otitis media.

An Indigenous participant was invited to respond to the presentation. He highlighted the challenges that genetic research presents to Indigenous people: ‘it’s a tough gig’. Regarding research more generally, he said that ‘one of [the] things I’ve noticed about research is that it can... disconnect us as human beings, especially data collectors’. He raised the question of how Indigenous people can build up their own capacity to understand these concepts, as the language is a challenge. The researchers can be thought of as ‘the great white centre’, and the Roundtable provided a space for Indigenous people to talk about these issues. He argued that it’s about power and control, and that it is critical to ask the researchers why they want to do their research—to what end?

*There are bells ringing for me when I hear Jennie [Blackwell] speak. It speaks to this idea that we've broken and we are busted and we need to disappear and vanish. And it connects with our lived experience around not trusting whitefellas.*

He then talked about the Principal Component Analysis graph: 'And when I see the graph and see how we are moving up towards the Caucasians again, I think, why can't we move down towards the Caucasians?' Another genetic researcher in the audience suggested that 'we can put it [the graph] upside down' so that the Caucasians are at the bottom. The Indigenous speaker finished by saying that if there are barriers to this kind of research, then they should be made into strengths.

### **Sheila van Holst Pellekaan**

Sheila van Holst Pellekaan discussed her research in western New South Wales, where she has worked since the early 1990s. Although she is currently pursuing a genetic health research project, for which some results from a paper under review were shown, research until now has focused mainly on mitochondrial DNA (mtDNA), which is DNA that is passed down the maternal line. Published results from that work have identified very ancient deep maternal lineages and indicate a long period of genetic isolation from near neighbours. She talked about her ongoing experiences of talking to Aboriginal people about genetics over 18 years. She stated that plain language documents were an essential tool when talking with individuals face to face and with local groups. This type of contact, she argued, is very powerful when appropriate information is shared.

van Holst Pellekaan raised the issue of relationships between community organisations that agree to the research but that want individuals to make their own decisions about their individual participation. She has also had comments from some individuals who feel insulted that over-riding 'distant' committees can impose conditions that seem to imply that participants and/or families are not capable of making decisions for themselves. Other critical points made in the presentation included the importance of explaining that mtDNA work on maternal lineages does not ignore men, that genetic research does not mean disrespect for religious beliefs or cosmology, does not threaten Aboriginality or native title, and that mixed parentage does not deny Aboriginality. The criteria by which people identify as Aboriginal does not require genetic evidence. Sheila and collaborator Joanne Lind want the research to continue because they believe it is important not to leave Aboriginal people out of the types of genetic epidemiological health studies being done throughout the world and because Sheila has built up a very good trusting, working relationship with families, communities and Maari Ma Health Aboriginal Corporation in western New South Wales. It should be possible for Maari Ma to have a high level of involvement if funding becomes available to expand the study.

One Indigenous Roundtable participant commented that although he finds the research to be very interesting, 'while I'm not an expert, I'm concerned about the impact of research'.

One member of an Aboriginal ethics committee commented that, of 160 projects that the committee had been asked to review in recent years, only three had raised serious issues, all of which were genetics based. The only project formally rejected was a genetic-based study in which an overseas researcher wanted to pick a hundred people at random and get published: 'he was going to pick up a hundred people off the streets of Sydney and get their DNA and get into a journal'. The committee member said that a lot of time was spent working with this individual

but it was felt that his publications would not reflect the actual research. The other two genetics projects were passed but also created controversy.


In particular, discussion of ‘admixture’ in terms of ‘percentages’ plays into the hands of conservative commentators: ‘It’s like offering Andrew Bolt<sup>6</sup> a gold-plated Christmas present. He will say that benefits should only be given to those above a certain percentage.’ The committee member also raised concern about the idea that it is effective to work directly with Indigenous families rather than with representative bodies: ‘The idea of small groups can be seen as cherry picking people who will agree with you.’ He suggested that it’s going to be very hard to get rid of risks—that you can manage risks and explain them—but at the end of the day people will still be worried about the risks. It is up to researchers to convince Indigenous people of the benefits because the risks are high. He argued that the researchers’ perspective is distinct from the Indigenous perspective, and said that the ethics committee wants to build the capacity of non-Indigenous researchers to work with Indigenous people. He also argued that although people obviously want to know about family trees, and their predisposition to disease, he is yet to read a study that doesn’t generalise about the Aboriginal population. He pointed out that the Māori guidelines specifically addressed this problem.

Another genetic researcher discussed research undertaken in a remote community in the Northern Territory, where the rates of renal disease are 30 times the rate in the general population. The larger project looking at renal disease gained funding in 1994, but the genetic component was not carried out because ethics approval was not given. He commented on the effect of the Human Genome Diversity Project and general negative perceptions of genetic research (curiosity-driven research in general) within the Indigenous and research communities: ‘Every time that Luca Cavalli-Sforza<sup>7</sup> opened his mouth our ethics approval died a death.’ Eventually, ethics approval for the genetic research was given in 2005 by both institutional and Aboriginal ethics committees. The research project initially had 250 samples that they had permission to conduct genetic research on, but there were significant technical issues. The DNA had not lasted very well and it took a few years to generate the technology to amplify it: ‘If you have any forensic samples you want to do health research on, we now know how to do it’, the researcher joked. Reinforcing the issues that Jenefer Blackwell raised, he discussed the level of heterozygosity of the sample and the fact that the known renal disease genes were not found in the community. He talked about some of the other technical challenges of Indigenous genetic research in this context. Because of the close family ties within the community, and the very long length of occupation of Indigenous people, there may be insufficient polymorphism (genetic variation between people) to find the disease genes, and it is possible that the disease genes have been selected for. Furthermore, because renal disease is so common, it is difficult to find a ‘control’ group that does not have the disease.

Another genetic researcher introduced research with a community in Central Australia. In this case, the samples were collected in the early 1980s. At that time, the research centred on population history. In more recent years the research has focused on genes that relate to inflammation, which may contribute to understanding how to treat and prevent infectious diseases. The research has suggested that Indigenous people in this community may have a lesser anti-inflammatory

6 Andrew Bolt is a popular conservative commentator who is known for his outspoken views on Indigenous affairs, including attacks on so-called ‘fair-skinned’ Indigenous people who he says should not take up Indigenous-identified positions.

7 Luca Cavalli-Sforza was the main proponent of the Human Genome Diversity Project in the 1990s. This project was dubbed the ‘vampire’ project by Indigenous people worldwide who resisted it (IPCB 2000).



response and a stronger pro-inflammatory response because of the effects of a hunter-gatherer lifestyle over tens of thousands of years. The researcher discussed another reason why genetic research could be useful in Indigenous communities, namely ‘ecological niches’:

*What people haven’t talked about today is that Aboriginal people have filled ecological niches in Australia for at least 50,000 years. The environment has affected genes which have created great genetic diversity. Nobody has determined what the diversity is. They were hunter-gatherers with no cities and no herding of animals. As a result, there was no exposure to those infections until we came and messed everything up.*

His research has also highlighted the links between genetics and environment: ‘Smoking targets the genes for interleukin-10 [a protein involved in inflammation] and further reduces the anti-inflammatory response and therefore reducing smoking will also improve infections in disease populations.’ In this case, genetic research has reinforced the need for health promotion and disease prevention measures.

Another researcher discussed her knowledge of a genetic testing program for a rare genetic disease in an Aboriginal community: ‘There was a lot of frustration about the length of time the results took to come back and the co-ordination of genetic counselling.’ This frustration has been somewhat alleviated by the formation of a foundation to help families affected by the disease. The other issue concerns shame: ‘families with the disease do experience shame, so the take up of any kind of genetic research is likely to be affected by how visible the disease is in the community’. She commented that for the research she is developing, the issues of waiting times and clarity about research results is important:

*For the kind of research we want to do, we would be looking for a genetic mutation, and this is something that potentially could take months or up to a year to identify (if we found something, but there is a good chance that we won’t find a mutation). So, there obviously needs to be a lot of education to address the expectations of the community, and we need to be upfront about the timeframe that we can provide results in.*

### **Emma Kowal**

Emma Kowal presented a review on current ethical international guidelines on genetic research in Indigenous contexts, including New Zealand, Canada and Australia: ‘While all Indigenous health research is ethically sensitive—hence the existence of Indigenous-specific research guidelines—Indigenous genetic research raises specific issues.’

In order to illustrate these potential ethical issues, she discussed two recent case studies of ethically sensitive genetic research projects: the long-running court case over allegedly unauthorised use of Indigenous DNA samples involving the Havasupai people and Arizona State University, and the ‘warrior gene’ controversy in New Zealand. Lessons to be learned from these experiences include the potential for additional and third-party use, sensitivity among Indigenous people to population genetics (which can contradict with their cultural or religious beliefs), sensitivity to the reporting of ‘low heterozygosity’ (which is often reported as ‘inbreeding’) and the potential for media reporting of results to increase racism.

Kowal discussed the overarching issues that researchers working in this context must continually

be mindful of, including widespread racist stereotypes and beliefs about Indigenous people's biological inferiority (which can potentially be exacerbated by media reports), lack of trust and suspicion felt by Indigenous people towards genetic researchers, and the inability of researchers to manage these factors (hence the need for dialogue and capacity building in this area). Kowal concluded with a list of critical questions that aimed to facilitate small group discussion:

1. How can (and can) genetic researchers manage the risks of Indigenous genetic research?
2. Do the potential benefits to Indigenous people outweigh the potential risks?
3. How should the ethical issues identified be dealt with in an Australian Indigenous context?
  - a. Existing or new ethical guidelines and role of Aboriginal ethics committees?
  - b. Research governance structures?
  - c. Other measures?
4. Are there additional issues that need to be considered? Existing issues include informed consent, privacy and confidentiality, storage of samples, ownership and use of samples, publication of results, benefits to community intellectual property (IP) and commercialisation versus free access to data.
5. Do we need separate guidelines for Indigenous genetic research?
6. Do the existing Values and Ethics document and/or the National Statement need to be revised to cover these issues?
7. Is it a matter of better relationships, rather than better guidelines?

## **Group discussions**

After the presentations by Blackwell, van Holst Pellekaan and Kowal, the Roundtable participants broke into four small groups for further discussions of the ethical issues surrounding Indigenous genetic research.

Ian Anderson followed the group discussions by raising the issue of whether there are genetic issues that are specific to Indigenous people or whether 'what we are really talking about are socially identifiable groups'. He also briefly mentioned how rapidly genetic technology is evolving and how this therefore makes it a unique type of research.

Afterwards, a spokesperson from each group reported on the discussions.

### **Group A**

A range of issues were discussed, including the ownership of samples and how this can be negotiated with each community in question. Group A discussed the danger that publication of Indigenous genetic research may be reported in the media in a way that plays into existing stereotypes about Indigenous people. Group members discussed whether publication is necessary, or whether there is another way to ensure that research results improve health outcomes. They discussed the ways that researchers can try and report results in ways that minimise the risk of harm. The non-geneticists in the small group were surprised to realise there was some flexibility to how results can be reported to allow for sensitivities, as this contradicted what they had previously believed: 'Is it about cold, hard science versus choosing what to say? There is a contradiction there

[between the proposition that scientists must report results in a standard way and the proposition that scientists have flexibility in how they report results].’ The notion of benefits was discussed. As the benefits of genetic research are likely to be long term, genetic researchers need to provide more immediate benefits to the community such as providing employment and training in research and/or health service provision. Issues of consent, language, media reporting and generalisation of data were discussed. The group discussed the ‘warrior gene’ controversy, and some group members asked why the sensitive issues of alcohol and violence were chosen as topics to research in that instance, rather than something else. Other issues discussed included the future use of data and concerns over use of genetic data for native title claims, as well as other concerns not specific to Indigenous people, including the potential for genetic discrimination in employment and obtaining insurance.

### **Group B**

This group discussed informed consent and issues surrounding publication. Group members discussed who owns genetic samples and data, and considered the idea of community power-of-veto to stop publication of results if the community felt the results were potentially damaging to the community. They discussed the level of involvement within the community in terms of skill building, employment opportunities and the potential to receive valuable insight from the community’s own historical knowledge. With regards to informed consent, they considered what a potential research participant really needs to be informed about and whether the researcher has a conflict of interest in obtaining consent. They considered whether there is a need for an independent advocate to get consent.

### **Group C**

This group discussed the need to demonstrate tangible benefits to Indigenous communities that participate in genetic research. Part of any genetic research proposal must be to help the community better understand and prevent the relevant disease and not just extract DNA from them. However, with complex diseases (e.g. cardiovascular conditions), we do not yet have sufficient information to do this, so some baseline genetic studies need to be done to this point. The researcher needs to have a team that can continue to work with the community.

The problem of low levels of genetic literacy in the community was also discussed.

Some people in the group felt that in their experiences of genetic research the level of understanding of genetics among Aboriginal communities was underestimated by researchers and by ethics committees: ‘They do know about genetics, they all watched CSI.’ Another researcher felt there was a good understanding of long-term benefits as well:

*They do understand that it may not benefit them tomorrow but it will in the future.  
Grandmothers understand that it may not benefit them but are okay [with the research  
because]... it will benefit future generations.*

Pharmacogenomics (tailoring drugs to individuals) was also discussed, particularly the first ‘ethnic’ drug, called BiDil, a blood pressure medication for African-Americans that has been available since 2005. One participant argued that having an ethnic-specific drug ‘is not racist because it is moving towards personalised targeted medicine’. Another participant argued that we do not know nearly enough about the genetics of Indigenous populations: ‘It is a matter of urgency to get a few



Aboriginal genomes sequenced so we can stop stabbing in the dark.'

The group then discussed the process of ethics approval for genetic research projects. Some genetic research projects in Indigenous communities have received ethics approval from one of the Indigenous ethics committees, while others have approval from their university ethics committee. One geneticist working in a remote community said:

*We got ethics [approval] from [the university ethics committee] in 1986 and that [approval] has continued on. In 2007 we thought we better go and see about ethics and we went to [the community] and asked them whether we should go to the [Aboriginal ethics] committee and they said, 'no, were not interested in them, don't go.'*

The researchers followed the wishes of the community leaders and continued working with the university ethics committee and did not submit an application to the relevant Aboriginal ethics committee. Another participant expressed concern that the Indigenous community leaders did not want the researchers to have their project approved through an Aboriginal ethics committee in addition to the university ethics committee: 'We need Indigenous people to be speaking with one voice.'

#### **Group D**

Group D discussed the possibility of future research conducted on public genetic databases, and more generally of the need to be cognisant of what can be done in the future, as the field of genetics is rapidly advancing. When we collect samples and data now, it is hard to know how others might be able to use that data in the future. The group couldn't resolve the tension between the benefits and the risks of genetic research. Some of the benefits are potentially shorter term, such as risk profiles of diseases. In terms of the risks, some group members thought that we all have responsibility to address inaccuracies in how data are used by the media and others. Some group members questioned whether samples need to be labelled as Aboriginal at all.

The group discussed how environmental issues (such as housing, smoking and nutrition) can be rectified, a problem that may prevent some of the potential knowledge generated by genetic research from being implemented. The group discussed how the links between genetics and environment—gene expression—need to be understood a lot better.

#### **Presentation**

##### ***Kristine Barlow-Stewart***

After the group discussions, a presentation was given by Kristine Barlow-Stewart, Director, Centre for Genetics Education, Royal North Shore Hospital and The University of Sydney.

Kristine Barlow-Stewart began her presentation by summarising the discussion paper on genetic literacy in Indigenous contexts (prepared by Emma Kowal and Lobna Rouhani). She discussed the lack of, and importance of, health and genetic literacy, particularly for socially identifiable and low socioeconomic groups. Barlow-Stewart defined health literacy and genetic literacy as the information that individuals need in order to access the health services appropriate to them. Barlow-Stewart argued that it is imperative that health literacy is interactive and engaging, and that it is critical to talk to people and gauge their understanding of issues affecting them, in

addition to including them as full partners in the process of improving their health. She discussed an extreme example of genetic literacy in the Personal Genome Project, a genetic research project that requires potential participants to study for and pass an online exam on gene transmission, expression, regulation, ethical issues and project literacy before they are permitted to participate in the research.

Barlow-Stewart then discussed tools for increasing genetic literacy in socially identifiable populations. She first discussed a genetic carrier screening program for high school students in Ashkenazi Jewish populations. Education materials for these screening programs are developed within the community and owned by the community. Sample access and genetic data is protected by a 'gene trustee'. A gene trustee is an independent person from the studied community who has the key to re-identify data. Knowledge about, and attitudes towards, genetics were significantly improved through the education process and knowledge was retained up to three to five years later.

To illustrate the importance of culturally appropriate genetic literacy, Barlow-Stewart told a story about a Chinese woman who initially refused to participate in a DNA test for a genetic disease because she did not believe that genes caused the hereditary disease that her family suffered from; she believed that it was caused by 'bad luck'. Barlow-Stewart responded by presenting genetic testing in a way that would be more applicable to the woman's beliefs: 'we say we are doing genetic test but when we are doing it we are looking for the first sign of bad luck'. After expressing it in this way, the woman agreed to have the test.

Barlow-Stewart then discussed the Latino Consumers Genetics Education Network in New York, which developed a 'train the trainer' genetic literacy program for community health workers. The curriculum was developed using local health workers, geneticists, adult literacy consultants, translators, Latin American-trained physicians, and a Board composed of community and religious leaders. Education materials were based on telling stories and the program was promoted at community events, beauty salons, social clubs, schools and in home gatherings. She also discussed the Genetic Education for Native American Project (covered in 'Background Papers'), which may serve as a model for developing genetic literacy workshops designed for conferences that significant numbers of Aboriginal Health Workers and Indigenous researchers attend. Finally, she discussed the genetic literacy tools developed by Sheila van Holst Pellekaan and the tool developed by the Machado Joseph Disease Foundation (also discussed in the 'Background Papers'). A final point made reiterating the importance of language selection was 'the need to explain scientific terms in order for people to engage and the need to walk alongside people in the process and not talk down to them'.

## **General discussion**

Kristine Barlow-Stewart's presentation led to a discussion about whether we need to use technical scientific language so that people understand concepts when they hear things in the media or from their local health professional, and whether using culturally translated concepts was thus not in the interests of people from diverse backgrounds because it underestimates their capacities to understand or culturally translate everything they hear: 'How can people consent to something that they don't understand?'

The issue of a gene trustee was pertinent to one genetic researcher who is concerned about



what will happen to personalised information, currently kept locked away, upon the researcher's retirement:

*We need some kind of mechanism for storing personalised information, particularly for ancestry-related research. Community reports and papers have depersonalised information only. What will happen to the records when I die? Why should it not be possible for a member of a participant's family, maybe several generations into the future, who knows that their granny/relative was in the study and would like to find out about the work. It is part of their story but they may not have heard about it through their family. I've been to AIATSIS [Australian Institute for Aboriginal and Torres Strait Islander Studies] and they won't hold the personal information there.*

This researcher discussed one idea of a local Aboriginal-controlled organisation or 'keeping place' to take responsibility for housing personalised information. The issue remains unresolved. Another researcher commented that database custodians need to exercise discretion over how to execute their data.

There was a general discussion on how literate you need to be to give informed consent in a research context versus a clinical setting. One participant argued that researchers should avoid using acronyms and jargon, which exclude people from the conversation. He also stated that:

*we don't need to know the science but we need to know the implications, you need to know the risks and benefits of the operation, not how the operation is done.*

Another participant pointed out that when obtaining informed consent in clinical contexts, the clinicians emphasise the risks so that they protect themselves from potential litigation, but in a research context the researcher emphasises the benefits of the research in order to maximise participation. Another participant asked, 'how literate do you need to be to give informed consent? Do they need to know what an SNP is, for example?' Another participant argued that 'people don't need to know the science, but what can go wrong, and the benefits etcetera'. Another participant stated that organ donation is a good example of this, because families don't need to know the details of how an organ is transplanted, but do need to understand the procedure. Ian Anderson commented that 'in order to participate ethically, you don't need to be a geneticist'.

Another participant made the point that scientific knowledge can threaten cultural beliefs and scientists can underestimate the effect of this. He says that the dichotomy between knowing about genetics and understanding the risks is tied together because genetic knowledge can threaten religious beliefs, so the act of knowing about genetics involves a risk itself: 'Indigenous people can't keep two sets of beliefs simultaneously like we can.' A second participant argued that some anthropological literature posits that Indigenous people can hold two opposing views better than the average Westerner because oppositions are at the heart of classical Aboriginal worldviews. The first participant also talked about the tension between the scientific idea of sharing knowledge and the fact that within Aboriginal society knowledge is private. He argued that there is a need to explain to scientists how Indigenous people view knowledge. He emphasised that genetic literacy argument is not just about what terminology means, but what it means to people's lives and cultural understandings.

One participant asked why Aboriginal people need to get involved in genetic research.

Another participant replied that high-quality evidence-based studies that use the very best science available are needed to improve Indigenous health.

An Indigenous participant stated that we need to look at the research that needs to be done specifically in Indigenous populations and only do the research that is necessary: 'If it can be done somewhere else [in another population], then do it elsewhere.'

An Indigenous participant discussed how in North American bioethics you must include minorities, otherwise you need to state why you are not including them. Another participant commented that while the American model is one of 'inclusion', the dominant framework in Indigenous research in Australia is one of 'protection'. Another Indigenous participant commented that 'whereas [the] mindset in Australia is that research is dangerous, in America the perception is that it is not dangerous'.

A genetic researcher asked about whether ethical approval is needed from an Aboriginal ethics committee if data are collected about the indigeneity of the participants. An Indigenous participant answered that if comparisons are made between the Indigenous and non-Indigenous data, then specific ethical approval is needed. Another participant commented that researchers need to think, before collection, about how their data will be used in future so that they can tell what the ethical implications will be.

Referring to the Background paper, another researcher asked, 'what does research in a Māori worldview mean in a practical sense?' An Indigenous participant responded by talking about *Kaupapa Māori*, the Māori system of knowledge that guides Māori research.

Ian Anderson posed the question of whether there is 'something in particular about genetic research or do existing ethical guidelines cover all the issues?' Another Indigenous participant argued that specific guidelines are needed because 'there is an intimacy to genetic information. It's very personal, it's your story and it becomes the community story.' This led to discussion of how both the risks and benefits of genetic research are trans-generational.

An Indigenous participant raised the issue that the sensitivity of genetics also affects the willingness of non-Indigenous people to be involved in this research because they are at risk of being called racist: 'when White people get called a racist, it numbs them'.

Another participant commented that 'we are constantly looking at the downside when we should refocus on looking at the upside' of genetic research.

However, another participant reiterated the potential for further stigmatisation (when population structure is reported in publications as 'percentages' of Aboriginality) and harm inflicted by the potential misuse of samples. He argued that this may make it difficult to convince Indigenous communities to be involved in genetic research: 'The potential benefits are given, I believe in them, but we need to convince Indigenous people of that.'

An Indigenous participant stated:

*researchers should talk to the people. Blackfellas are scientific [enough] to be able to survive in this country for 50,000 years. They are the best environmentalists on the planet, and they didn't*

*need science to tell them that. Don't think we know nothing.*

Summing up the feeling in the room, a researcher commented that ‘everyone in this room has good will but we need to get inside each other’s heads’.

### **Next steps**

Ian Anderson and Emma Kowal then talked about the ‘next steps’ in further developing this important discussion about genetic research in Indigenous communities. Although the Roundtable succeeded in identifying many challenging issues and generating constructive debate, much more discussion and research is needed to work through these issues. The ideas for further development and research include the possibility of media reporting guidelines for reporting genetic research through a science–media exchange. Such guidelines could be developed by a group of people who advocate for better reporting of Aboriginal genetic research in a way that minimises the chance of reinforcing stereotypes. Two other major ‘next steps’ are consideration of whether specific guidelines for genetic research are necessary, and whether Aboriginal Health Worker training programs on genetic literacy can be developed.

Ian Anderson stated that the Lowitja Institute can have a role in continuing this conversation about genetic research, because it is an example of the kind of ‘dangerous’ conversations that the Institute sees its role in facilitating. As an example of ‘dangerous’ conversations for the future, Kowal suggested that an Aboriginal biobank could be discussed as a mechanism for ensuring that access to samples was controlled by Indigenous people in the long term, and that sample sizes were large enough to generate useful research results. This was briefly discussed, including the process of using a ‘stepped’ consent process so that participants can consent to having their samples used only for a specific kind of research or for any research that is approved by an Indigenous governing board. Another participant suggested that, as a first step, researchers could explore collaborating on a project that is of interest to more than one community. One participant made a final comment that these issues are of great sensitivity, and if the day’s discussions were brought up in the media, they could create angst.

# Appendix: Indigenous Genetics Roundtable program and list of attendees

## Indigenous Genetics Roundtable program

Friday 2 July 2010

8.45 am–4.00 pm

Graduate House, The University of Melbourne

8.45am Registration, tea and coffee

9.00 Welcome, aims of workshop and overview (Ian Anderson)

9.15 Overview of genetic health research (Jenefer Blackwell):

- Brief discussion of inheritance, genetic variation and diversity, gene-environment interactions
- Aims and basic study designs of typical research projects
- The range of genetic research that has been conducted in ATSI communities (drawing on database)

9.45 Introductions:

Each group present to give a 5–7 minute overview of research/area of work in relation to Indigenous genetics (others to give 1–2 minute introduction):

- Simon Foote/Jim Stankovich/John Mathews, Menzies
- Jenefer Blackwell/Sarra Jamieson, TICHR
- Glenn Pearson, Kulunga
- Sheila Holst van Pellekaan/Joanne Lind, UNSW/UWS
- Danny Kelly/Bob Davidson, AHMRC
- Rodney Scott, U Newcastle

10.30 Overview of ethical issues and international guidelines for the conduct of genetic research in Indigenous communities (to guide small group work after morning tea)

11.00 Networking morning tea

11.30 Small group work: How should the ethical issues identified be dealt with in an Australian Indigenous context? Are there additional issues that need to be considered?

12.00 General discussion of how ethical issues impact specifically on ATSI communities participating in genetic research including:

- Informed consent
- Privacy and confidentiality

- Storage of samples
  - Ownership and use of samples
  - Publication of results
  - Benefits to community
  - IP and commercialisation vs free access to data
- 1.00      Networking lunch, pre-allocated seating at 4 tables.
- 1.45      Discussion of review of genetic literacy tools (pre-circulated)  
             Kristine Barlow-Stewart to give 10 minute introduction, then general discussion
- 2.30      Next steps: What do we need to do to move this discussion forward?  
             Discussion of potential project that aims to develop a culturally-appropriate model  
             of Indigenous genomic research including:
- (1) an assessment of the ethical issues associated with genetic research in  
 Indigenous communities;
- (2) an assessment of the governance arrangements required to manage the  
 potential risks and benefits of genetic research; and
- (3) culturally-appropriate genetic literacy tools to provide the basis for a sound  
 informed consent process.
- Emma Kowal to give 10 minute introduction, then general discussion
- 3.15      Networking afternoon tea
- 3.30      Closing comments (led by Ian Anderson)
- 4.00      Close

## List of attendees

**Ian Anderson**, The Lowitja Institute and *Onemda* VicHealth Koori Health Unit, The University of Melbourne.

**Debra Knoche**, *Onemda* VicHealth Koori Health Unit, The University of Melbourne.

**Yin Paradies**, *Onemda* VicHealth Koori Health Unit and McCaughey Centre: VicHealth Centre for the Promotion of Mental Health and Community Wellbeing, The University of Melbourne.

**John Mathews**, Centre for Molecular, Environmental, Genetic and Analytic Epidemiology, The University of Melbourne.

**Anna Forsythe**, Centre for Molecular, Environmental, Genetic and Analytic Epidemiology, The University of Melbourne.

**Simon Foote**, Menzies Research Institute, University of Tasmania.

**Jim Stankovich**, Menzies Research Institute, University of Tasmania.

**Emma Kowal**, School of Anthropology, The University of Melbourne.

**Lobna Rouhani**, School of Anthropology, The University of Melbourne.

**Jenefer Blackwell**, Telethon Institute for Child Health Research.

**Sarra Jamieson**, Telethon Institute for Child Health Research.

**Elizabeth Scaman**, Telethon Institute for Child Health Research.

**Glenn Pearson**, Kulunga Research Network and Telethon Institute for Child Health Research.

**Sheila van Holst Pellekaan**, School of Biotechnology and Biomolecular Sciences, University of New South Wales.

**Joanne Lind**, School of Medicine, University of Western Sydney.

**Kristine Barlow-Stewart**, Centre for Genetics Education.

**Rodney Scott**, School of Biomedical Sciences, University of Newcastle.

**Bob Davidson**, Aboriginal Health and Medical Research Council.

**Danny Kelly**, Aboriginal Health and Medical Research Council.

**Don Chalmers**, Faculty of Law, University of Tasmania.

**Doug Hilton**, Walter and Eliza Hall Institute.

**Steven Tong**, Menzies School of Health Research.

**Alwin Chong**, The Lowitja Institute and Aboriginal Health Council of South Australia.

**Barbara Beacham**, The Lowitja Institute.

## With Apologies

**Ted Wilkes**, Centre for Developmental Health, Curtin University of Technology.

**Ngiaire Brown**, Poche Centre for Indigenous Health.

**Maureen O'Donnell**, Maari Ma Health Aboriginal Corporation.

**Nola Whyman**, Maari Ma Health Aboriginal Corporation.

**Colin Thomson**, School of Medicine, University of Wollongong.

**David Weisbrot**, School of Law, Macquarie University.

**Kalinda Griffiths**, Aboriginal Ethics Sub-Committee, Human Research Ethics Committee of the Northern Territory Department of Health and Community Services and the Menzies School of Health Research, Charles Darwin University.

**John Condon**, Menzies School of Health Research, Charles Darwin University.

**Peter Visscher**, Queensland Institute of Medical Research.

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Australia's National Institute  
for Aboriginal and Torres Strait  
Islander Health Research

**The Lowitja Institute**

PO Box 650, Carlton South  
Vic. 3053 AUSTRALIA

**T:** +61 3 8341 5555

**F:** +61 3 8341 5599

**E:** [admin@lowitja.org.au](mailto:admin@lowitja.org.au)

**W:** [www.lowitja.org.au](http://www.lowitja.org.au)