

# Genetic research in Indigenous health: significant progress, substantial challenges

Genomics is a powerful research tool, but it must be handled with care

Of all the measurements and biological samples used in health research, it is the extraction and analysis of genetic material that has caused the most controversy in indigenous health research across the globe. No other kind of research has a specific international non-governmental organisation dedicated to opposing it — the Indigenous People's Council on Biocolonialism. This United States-based organisation was founded in 1999 in the wake of the Human Genome Diversity Project (HGDP). That project was a worldwide effort to collect DNA samples from indigenous populations for population genetic research that aimed to understand the ancient migration of *Homo sapiens* out of Africa beginning 100 000 years ago, and populating the rest of the world.

## Pitfalls of indigenous genetic research

The HGDP created controversy among indigenous leaders who dubbed it “the vampire project”.<sup>1</sup> Indigenous Australians were among those who expressed their opposition.<sup>2</sup> The Director of the Central Australian Aboriginal Congress at the time, John Liddle, accused the HGDP of seeking to take blood samples without proper consultation and use them for a range of purposes, including patenting Aboriginal DNA to produce new pharmaceuticals.<sup>3</sup> Although HGDP organisers denied such claims, no samples for the project were collected in Australia. Unsurprisingly, this episode affected Indigenous genetic research in Australia, with at least one project losing its ethics approval as a direct result.<sup>4</sup>

Since then, indigenous genetic research has continued to make international headlines. One example involving Maori occurred in 2006. A geneticist presented research findings that found Maori were twice as likely as non-Maori to carry a gene associated with alcohol and tobacco use.<sup>5</sup> The particular polymorphism of the gene that encodes the enzyme monoamine oxidase has also been associated with risk-taking and aggression, and is consequently known in some circles as the “warrior gene”. This episode was widely reported in the international media as proving that Maori were genetically predetermined to be violent, a depiction that the researchers argued was a misrepresentation of their findings.<sup>6</sup> Maori academics voiced their concern about this project, and genetic research in general. As one critic put it, “contributions to racial stereotyping by trained scientists are unethical and scandalous”.<sup>6</sup>

Ethical research in an indigenous context requires awareness of the broader social, cultural and political context in which the research takes place.<sup>7</sup> Aboriginal and Torres Strait Islander and Maori populations alike have

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experienced centuries of oppression and discrimination, once supported by scientific theories of racial inferiority. Genetics has the potential to reinforce these old stereotypes, portraying indigenous health problems as biologically foretold, rather than the complex interaction of genes and environment that determines the health of every human.<sup>8</sup>

## Indigenous genetic research in Australia

Given this history, it is understandable that little genetic research has taken place in Indigenous Australia. When I began looking at this issue in 2007, there were no genetic health research projects operating in Indigenous communities, with very few in the preceding decades. Genetic researchers attempting to work in indigenous health have shared with me tales of failed grant applications, denied or revoked ethics approvals, and requests by indigenous organisations to remove genetics components of research projects. The belief that “indigenous people don't like genetics” was widespread among both indigenous and non-indigenous people involved in health research.

In just the past 2 years, the situation has changed significantly. In 2010, the first of four Indigenous genetic research projects was funded by the National Health and Medical Research Council. This was a study of the genetic associations between metabolic disease and otitis media in a remote Aboriginal community in Western Australia. Results from that project have been fed back to the community by means of an animation made with children at the local school,<sup>9</sup> and are soon to be published. In 2011, another project was funded, this time on possible genetic associations with vulval cancer, which is very rare in the general population but occurs among women in Arnhem Land at 70 times the national rate.<sup>4</sup> Two further funded projects began this year, focused on genetic associations with rheumatic heart disease and renal disease in a number of remote communities in the Northern Territory.

Internationally, the profile of indigenous genetic research received a boost when the 2010 President of the American Society of Human Genetics gave his presidential address on the issue.<sup>10</sup> Perhaps the most dramatic recent event was the sequencing of the “first Aboriginal genome”, published in the journal *Science* in 2011.<sup>11</sup> The researchers sequenced the genome from a hair sample held in a Cambridge museum that was collected by British ethnologist Alfred Haddon, from a young Aboriginal man at a railway station near Kalgoorlie in 1923. As well as being a scientific first, it also took us into uncharted ethical territory regarding the use of previously collected biological samples for scientific or medical research. In this case, the researchers approached the Goldfields Land and Sea Council, the organisation representing traditional owners from the Kalgoorlie region, to ask for their consent to publish the research.

## The future of indigenous genetic research

What does all this mean for indigenous health research? There are two pertinent lessons to learn from recent events.

First, genomics is here to stay, with increasing numbers of researchers proposing genetic studies and obtaining funding. As a group, Australian researchers working in Indigenous health have embarked on a steep learning curve as they work with local communities, Aboriginal ethics committees and other stakeholders to conduct genetic research projects that are acceptable to, and beneficial for, participating communities.

As researchers work through the specific issues raised by genetic research, they will develop important expertise in this area. So too will Aboriginal and Torres Strait Islander communities who participate in genetic research. It is quite possible, however, that this period of significant learning will involve mistakes and missteps. A mistake on the scale of the "warrior gene" controversy could have significant negative implications, not only for a specific project, but for other communities and researchers involved in this field.

In response to these concerns, the Lowitja Institute has supported two national roundtable discussions on genetic research in Aboriginal and Torres Strait Islander communities in 2010 and 2011.<sup>4,12</sup> At these events, genetic researchers working in Indigenous communities have come together with Indigenous researchers and community members, Aboriginal ethics committee members, and experts in ethics and genetic literacy, to discuss issues specific to Indigenous genetic research. Discussions ranged from cultural beliefs surrounding DNA storage to how results should be presented in scientific papers to minimise the risk of misleading and damaging media coverage. Emerging from these roundtables, a multidisciplinary team of Indigenous and non-Indigenous Australian researchers, together with indigenous geneticists from the United States and New Zealand, are engaged in a project that aims to develop guidelines and literacy tools to enable culturally appropriate genetic research in Aboriginal and Torres Strait Islander communities.

Second, issues raised by genetic research are closely related to more general concerns about the use and long-term storage of biospecimens collected from Indigenous communities. One example of this issue has been debated in the pages of this Journal. Prominent Indigenous health researcher Wendy Hoy criticised the Australian Health Survey (an initiative of the Australian Bureau of Statistics [ABS]) for excluding the option of collecting blood and urine samples from Indigenous participants. She suggested this exclusion was based on "poorly articulated fears" and argued that Indigenous people would miss out on any health benefits as a result.<sup>13</sup> In reply, Jelfs explained that biospecimens may be collected in subsequent waves of the health survey, once trust had been established between the Australian Health Survey and Indigenous communities.<sup>14</sup> However, trust may not be the only reason that numerous representative and consultative bodies advised the ABS against collecting biospecimens. Without a concerted effort to outline the ethical and cultural issues associated with collecting and storing biospecimens in an Indigenous

context, it is quite possible that consultations for the next round of the Australian Health Survey may end with Indigenous people being excluded once again. Exploring the ethical and cultural implications of genetic research will therefore inform a wider range of research activities.

Canada provides a salutary example. Experiences with Indigenous genetic research there have resulted in guidelines for the ethical conduct of health research in First Nations communities that include specific guidance on how biospecimens should be collected, stored and used.<sup>15</sup> There is a clear need for similar guidelines in Australia, so that both Indigenous communities and researchers have a starting point from which to negotiate these issues. Without clear guidelines, declining to participate in genetic research may be the only "safe" option for Aboriginal ethics committees and local community representatives.

To move forward, we need to transcend both old stereotypes of indigenous people as biologically inferior and new stereotypes that declare "indigenous people don't like genetics". Equally, we need to transcend stereotypes of genetic research as focused on proving the inferiority of indigenous people or the solely "genetic" basis of disease. Only then will indigenous communities and researchers be in a position to realise the potential of genomics as a tool in indigenous health research.

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