The need for genetic studies of Indigenous Australians

To the Editor: The continuing integration of genetic technologies into clinical medicine is providing opportunities for health care improvement. This has the potential to reduce health disparities between Indigenous and non-Indigenous Australians in several ways: improving our understanding of disease pathogenesis, obtaining a perspective from which to view environmental risks, improving prediction of disease risk, identifying new diagnostic techniques and drug targets, and making advances in pharmacogenomics.

Unfortunately, the dearth of genetic research involving Indigenous Australians may exclude them from such benefits of biomedical innovation.[1] This is beginning to affect the clinical utility of genetic tests. For instance, in selected circumstances, single nucleotide polymorphism chromosomal microarrays (CMAs), which examine one's chromosomal constitution at high resolution, are being employed to investigate a variety of phenotypes. To interpret the results of genetic investigations such as CMAs, it is a critical to understand the range of normal genetic variation, which is partly population specific. In addition, technologies such as next-generation sequencing, which can be used to analyse an individual's exome or genome, will soon be used in clinical care. A survey conducted by *Nature* suggests that the current bias against studies of non-European people is likely to pervade these techniques.[2] If so, a biased picture of which variants are important will emerge and, globally, genomic medicine will largely benefit a privileged (European) few.[3] Notably, rare genetic variants (ie, those occurring in less than 5% of the world's population) are disproportionately important in terms of complex disease risk and pharmacogenomics,[4] and some are the cause of monogenic disease. Rare variations tend to be population specific. Accordingly, reference data from historically marginalised populations are needed to separate real from spurious findings.[3] In addition, to help determine the significance of genotypic variation, there is a parallel need for accurate phenotypic information. Therefore, genetic studies will need to be coupled with detailed phenotyping, for which high-throughput phenomic assessments may be required.

> "the dearth of genetic research involving Indigenous Australians may exclude them from such benefits of biomedical innovation"
>
> Baynam

Advances in genetic testing that will be used for clinical care therefore require engagement of Indigenous communities, so that normal genetic variation in these populations can be ascertained. To maximise benefit and minimise harm, this will need to be managed in an inclusive and culturally sensitive manner, with open discussion among all relevant parties.[1] Failure to prioritise Indigenous genetic studies may perpetuate health inequity.

Gareth S Baynam Clinical Geneticist

Genetic Services of Western Australia, King Edward Memorial Hospital, Perth, WA.
gareth.baynam@health.wa.gov.au

Competing interests: No relevant disclosures.

doi: 10.5694/mja11.11459

1 Kowal E, Rouhani L, Anderson I. Genetic research in Aboriginal and Torres Strait Islander communities: beginning the conversation. Melbourne: The Lowitja Institute, 2011.
2 Genomes by the thousand. *Nature* 2010; 467: 1026-1027.
3 Bustamante CD, Burchard EG, De la Vega FM. Genomics for the world. *Nature* 2011; 475: 163-165.
4 Manolio TA, Collins FS, Cox NJ, et al. Finding the missing heritability of complex diseases. *Nature* 2009; 461: 747-753.