# Preparing for the genomics era

**Jon Emery**

*The Human Genome Project was one of the great feats of exploration in history – an inward voyage of discovery rather than an outward exploration of the planet or the cosmos.*

– NIH National Human Genome Project Research Institute

The Human Genome Project and its inward voyage of discovery was completed 15 years ago with considerable hype and expectation of benefits to human health and a revolution in healthcare.[1] The research translational pipeline is long, but we are now beginning to see how advances in genomic epidemiology and sequencing technology will change the way GPs practice medicine in the 21st century.

In this issue of the *Australian Journal of General Practice* (*AJGP*), four papers provide examples of this. Delatycki and colleagues discuss genetic carrier screening to identify couples who are at increased risk of having a child with a rare, recessively inherited, genetic condition.[2] Mackenzie's Mission, a Medical Research Future Fund project, aims to screen 10,000 Australian couples for carrier status of roughly 500 genes, with the expectation of significant involvement by GPs. Polasek et al describe how common genetic variants that affect response to medications could be used to individualise prescribing.[3] They estimate that, in 2017, roughly 1.7 million Australians received a medication for which pharmacogenomic testing could be relevant, and 40% of these patients would have at least one genetic variant affecting medication response. Isbister and Semsarian explain how genomics is increasingly used to identify specific causes of sudden cardiac death in the young, with important implications for families and prevention in relatives.[4] This reflects relatively rare genetic causes of cardiovascular disease, but soon we are likely to see tests for common variants that provide polygenic risk scores for multiple common chronic conditions.[5]

These advances raise many important considerations, not least who will pay for such tests and the implications of genomic testing for patients and insurance policies. Otlowski et al discuss current legislation regarding life insurance and genetic tests, and calls in Australia for a moratorium by the life insurance industry to reduce potential genetic discrimination.[6]

The National Health Genomics Policy Framework highlights the importance of a workforce that is genomically literate if patients are to benefit from genomic advances.[7] This is a significant challenge for general practice. The Royal Australian College of General Practitioners has developed the *Genomics in general practice* resource, which provides short, practical summaries about common genetic conditions and genetic tests.[8] The genomics era is finally upon general practice, and we need to be prepared to meet the challenges it raises and reap its benefits.

**Author**

Jon Emery MBBCh, MA, MRCGP, FRACGP, DPhil, Herman Professor of Primary Care Cancer Research, Department of General Practice and Centre for Cancer Research, University of Melbourne, Parkville, Vic

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correspondence **ajgp@racgp.org.au**