

PRECISION MEDICINE: NOW IS THE TIME TO GET IT RIGHT

EMBARGOED UNTIL 12:01am Monday 28 November 2022

THERE have been promising clinical benefits arising from genomics, leading to publicly funded precision medicine available for some conditions. However, due to the rapid advances in this field, there remain barriers to full uptake of this emerging therapy, with authors of a Perspective, published today by the *Medical Journal of Australia*, calling for local, state and federal genomic medicine implementation research.

"Precision medicine is a tailored approach to health, incorporating an individual's genetic make-up, environment and lifestyle," wrote the authors, led by Dr Rosie O'Shea from the University of Sydney.

"Its recent rise has been largely driven by rapid advances in genomic medicine, with sequencing of an individual's genetic code identifying opportunities for precision health care, therapies and diagnostics. An ever-increasing proportion of families are receiving accurate genetic diagnoses, access to screening and counselling, and clinical management from publicly funded genomic technologies."

Medicare item numbers are now approved for genomic diagnostics in cancer, pre-implantation genetic diagnosis, and certain paediatric, renal and cardiac conditions.

However, say O'Shea and colleagues, barriers and challenges still remain.

"Many non-genetics professionals are not well prepared to use the newly funded genomic diagnostic tests," they wrote.

"Medical and training curricula covering genetics and genomics require updating, including guidance from professional bodies and colleges, both in primary care and specialty groups.

"There are reports from clinicians that they may rather refer to local genetics services or professionals to perform genomic testing, interpretation and clinical management of cases.

"However, this is not possible given the current clinical genetics workforce in Australia which has only an estimated 150 genetic physicians and 220 genetic counsellors.

"This means that current systems require new approaches to cope with the increased workload, which may include improved education and capability for local and disease-specific specialists to manage genomic care with increased capacity.

"An average of 17 years is required to integrate evidence-based practices into routine health care, and genomics has exploded from widespread sequencing availability to TGA-approved therapies requiring a precise genetic diagnosis in less than a decade."

What's needed, write O'Shea and colleagues, is translational research informing policy and practice, now known as "implementation science".

"An implementation science approach can address many of the already identified barriers and gaps in precision medicine," they wrote.

O'Shea and colleagues wrote that there was an urgent need for genomic medicine implementation research, "to ensure effective models of genomic care are created".

"Such research would allow evidence generation for optimal adoption, knowledge of factors affecting practice, and would inform policy about precision medicine program design.

"A focus on pre-implementation research commensurate with the introduction of new Medicare numbers for genomics will help define the best scalable models of care to implement genomics into routine practice," they concluded.

"This call to action will bring the benefits of precision medicine for all Australians."

All MJA media releases are open access and can be found at: <https://www.mja.com.au/journal/media>

Please remember to credit *The MJA*.

The *Medical Journal of Australia* is a publication of the Australian Medical Association.

The statements or opinions that are expressed in the MJA reflect the views of the authors and do not represent the official policy of the AMA or the MJA unless that is so stated.

CONTACTS:

Dr Rosie O'Shea
University of Sydney
Email: rosie.oshea@sydney.edu.au

Sarah Palmer
Media and Content Manager
Sydney Children's Hospitals Network
Ph: 02 9845 0512
Email: sarah.palmer@health.nsw.gov.au