

Implementing Genomics in Primary Care

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INTRODUCTION

Genomic testing is becoming more widely available across all health care settings in Australia, including primary health. The evidence-base of clinical utility and cost effectiveness of many genomic applications is now well established and growing, and consumer attitudes are becoming more positive to testing.

Recent additions of genomic tests onto the Australian universal insurance scheme, Medicare, have made tests such as genetic carrier screening for couples planning a family, and testing for actionable genetic diseases such as familial hypercholesterolaemia, more feasible in primary practice. However, many general practitioners (GPs) [family physicians] have limited experience in genetic testing. There are also concerns over the capacity of specialist genomic services to handle any increases in referrals.

AIM

The aim of this study is to define the challenges of implementing genomics into primary care, and strategies and resources being used to address them.

METHODS

This study draws on findings from three studies conducted at different system levels.

1. Macro level ❖ Rich picture of implementation of genomics

Data on the implementation of genomics in Australia came from synthesised grey and peer-reviewed literature, and interviews with nine key informants from Australian Genomics. Key stakeholders were defined including clinicians across various settings, patients, industry, research institutions, government, regulatory bodies, and consumer advocates. Interactions between these stakeholders and the constraints and enablers influencing the system were mapped ¹.

2. Meso level ❖ Literature review on genomic workforce issues
❖ Policy brief

Grey and peer-reviewed literature reviews summarised genomic workforce issues across Australia, including shifting demands and capacity for genomics services, and education needs for non-genetically trained clinicians².

3. Micro level ❖ Interviewing 23 GPs from across Australia involved in recruiting couples for reproductive genetic carrier screening

Finally, interviews were conducted with 23 GPs from across Australia who were involved in recruiting couples for reproductive genetic carrier screening for the research project “Mackenzie’s Mission”³. The Theoretical Domains Framework was used to deductively analyse data to define barriers the GPs faced in offering the screening.

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RESULTS

Findings from the macro level view of genomics were the dynamic interplay of government, professional bodies (e.g., Royal College of Pathologists Australasia) and patient advocacy groups that provided the context in which GPs were working. Key challenges were around sustainable and equitable funding of tests, and coordination of fragmented endeavours across separate sectors.



Figure 2: Rich picture of the clinical genomics landscape in Australia; Source¹: Long et al., 2021

At the meso level, it was recognised that GPs along with the majority of the healthcare workforce were not prepared for genomics. Classroom education was shown to be inadequate to develop the skills needed for genomic practice, and uncoordinated resource development was confusing rather than assisting. Innovative models using genetic specialists as “shepherds” of non-genetic trained clinicians are showing promise, as are nation-wide projects developing resources such as a Directory of genomic tests.

At the micro level, individuals reported lack of time as a barrier to offering genomic testing, and lack of skills rather than knowledge per se. Flexible, high quality education modules, “shepherding” and consumer facing resources (including in other languages) by genetic counsellors, were seen as mitigating these issues.

CONCLUSIONS

As genomic testing and screening becomes mainstream, a systems approach to implementation allows a holistic approach to be taken. It can also avoid unintended consequences of solutions developed in isolation. Attention to all levels of the system is important to ensure the quality of care and the sustainability of health systems. This work is being used to inform further work with Primary care settings in which general practitioners are overseeing genomic testing and screening.

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