Shortfalls of a new Medicare-funded genetic screening program

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THE 2023 announcement of funding for reproductive genetic carrier screening (RGCS) for cystic fibrosis, spinal muscular atrophy and fragile X syndrome comes because of important advocacy work[1,2] as a part of a greater than $80 million pledge from the Australian Government tagged for supporting access to genetic screening in the population. Screening for these conditions corresponds to Medicare Benefit Schedule (MBS) items 73451 and 73452 and represents a combined screening initiative. However, a lack of corresponding health promotion and education for providers threatens to undermine this important initiative. If properly supported, general practitioners (GPs) are confident and capable of delivering genetic counselling;[3] however, in current practice this is not the case. This viewpoint asserts that this flaw (in not offering support to GPs) will lead to poor service utilisation, increased burden on GPs and potential healthcare anxiety.

There are two options when referring for reproductive carrier screening: sequential or couple testing. Medicare stated that the funding scheme would 'support informed reproduction decision-making' and improve access to RGCS (a test that previously cost more than $450).[1] This change came following the pilot study completed for Mackenzie's Mission in 2019.[2]

Mackenzie's Mission offered screening to 9107 couples across Australia via key providers such as GPs, obstetricians and midwives for 750 severe childhood-onset genetic conditions.[2] The three-year study aimed to determine the evidence for making free RGCS available to all couples in Australia. The offer of screening was provided to couples, who were then given information resources and a decision-making aid to supplement their participation in screening. The results were then provided to couples, who were supported with counselling services and genetics education to enable them to implement the results in a way that best suited their individual values.[2]

The evidence from Mackenzie's Mission was overwhelmingly positive from a patient perspective,[3] but there were concerns raised by healthcare providers, particularly GPs. For example, one paper out of Mackenzie's Mission that examined practitioner beliefs around RGCS stated that GPs reported concern about their ability to discuss high-risk results, especially the possibility of pregnancy termination in women in early pregnancy.[4] There was also concern about the 'concept of risk assessment, lack of confidence in offering prenatal genetic advice [and] apprehension around interpreting and explaining the screening results'.[4] Throughout the GPs included in the study, there was a call for education and support for practitioners around genetics[4] to support the delivery of post-test counselling services. This provided clear evidence for the barriers to implementation in general practice to be considered should a program like this be funded.

Instead of following these proposals, the Australian Government's funding program includes only three-condition screening (aforementioned),[1] in comparison to Mackenzie's Mission's extended carrier screening program. In addition to the structure of testing being minimally applicable to Australian MBS item funding, it also does not include allocation of funds to health promotion or education material. Nor does it include supports for either participants (eg in the form of genetic counselling) or providers. Although this is not the responsibility of the MBS, it is reasonable to suggest that at least part of the $400 subsidised to pathology services when RGCS is ordered could be distributed to assist colleges such as The Royal Australian College of General Practitioners (RACGP) to develop centralised practitioner and patient support. For example, Mackenzie's Mission went so far as to develop a decision aid designed for this Australian government-funded screening program to supplement conversations between GPs and their patients, and it too was not included within resources for practitioners.[5]

The Royal Australian and New Zealand College of Obstetricians and Gynaecologists (RANZCOG) official statement on genetic carrier screening (current 2019) states:

> Recommendation 3: Information on carrier screening for other genetic conditions should be offered to all women planning a pregnancy or in the first trimester of pregnancy.[6]

This is regardless of family history or geographic origin.[7] The Australian government-funded RGCS program recommends GPs (and the RACGP) upskill their Fellows further in genetics without allocating funding to do so and adds yet another service to their already demanding expected repertoire unsupported. GPs, as the forefront providers of RGCS, were not consulted in the development of these policies or their implementation. GPs, without the provision of support, seldom have the time and expertise to provide detailed genetic education to families who access screening services.[4,8] Thus, providing some of the pledged budget for GP education and the development of resources through colleges such as the RACGP is warranted.

This aside, the implementation of the screening program was minimally promoted to the public, and both the screening program itself and its utility are poorly understood by most community members.[9] Studies conducted in the field suggest that knowledge of RGCS is lacking in the general population, and ongoing education is necessary.[9,10] There have been no standardised patient resources provided to general practices as part of the pledged funding to reflect this area of deficit. It has been up to pathology providers to design such resources and have them available.[11] Compounding this problem, GPs reported that a couple's knowledge of RGCS was 'reflected in the time required at consultation'.[4] This not only affects patient understanding of their own healthcare experiences, but it also slows the day's appointment books and decreases earning capacity within general practice, another likely contributor to RGCS hesitancy among GPs.

Furthermore, in an Australian context, the evidence used to inform the usefulness of the RGCS program in Mackenzie's Mission does not translate to the program that eventuated in the MBS because of substantial differences in what is offered to participants of each screening program.

If international experience tells us anything, it suggests that appropriate use of RGCS requires significant consumer health literacy regarding prenatal genetics, which is frequently lacking in the general population.[9,10] Although international data on RGCS exist within very different subsets of populations and vary between paid versus subsidised services,[12] little is known about RGCS in Australia, even with Mackenzie's Mission in mind.

If you are a GP and wish to upskill in these matters yourself, some resources are available through the RACGP, including the *Genomics in general practice* online guide.[13,14] The Victorian Clinical Genetics Services also deliver online learning modules aimed at educating GPs.[15]

In summary, the new MBS program funding through items 73451 and 73452 is at grave risk of being underutilised or, worse yet, misunderstood despite its best intentions. Its lack of correlating education programs for doctors or patients has disrupted the smooth implementation and equitable use the program intended. The current state of practice risks creating another prenatal tool that generates health anxiety instead of decision-making freedom if it does not support GPs to discuss results with their patients. A review of the funding structure and allocated rebates is needed to ensure that taxpayer dollars are used in a way that generates the most health benefit for the lowest dollar value and supports the capacity of Australian GPs, with time and money allocated to detailed education for all those involved.

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