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LETTER TO THE EDITOR

Genomic Testing Access for Hearing Loss Must Catch Up to the Evidence

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To the Editor,

Congenital hearing loss affects about 1–2 in every 1000 babies born in Australia [1]. Around 50% of childhood hearing loss has an underlying genetic cause [2]. To date, over 500 causative genetic variants are known to be responsible for both syndromic and non-syndromic hearing loss [3].

Genomic testing has revolutionised clinicians' ability to provide accurate and timely diagnosis for individuals with hearing loss and their families. Australia is at the forefront of demonstrating exome sequencing's cost-effective value in informing prognosis, guiding management and clarifying the risk of recurrence in family members [4]. This piece of evidence was successfully used to initiate Medicare funding for exome sequencing in childhood hearing loss, available since 2023 with Medicare Benefits Schedule (MBS) items 73,440–73,444.

Despite strong evidence of the benefit of genomic testing for these families, the combined uptake of these five MBS items—193 over 2 years—remains lower than the expected 180–200 per year and 3000 retrospectively eligible cases (Table 1). The available funding ($1200 for singleton test) is insufficient for laboratories to complete testing (as of December 2025: Victorian Clinical Genetics Services, $2195; Pathology Queensland, $2181), and testing processes are laboriously slow. In addition, there is limited access to genetic counselling, a vital service for families for adequate understanding and informed consent for testing. Genetics services often have long waitlists, inequitable and variable decisions about funded testing and a long wait for results. Consequently, most families are unable to realistically access genomic testing for hearing loss, and for those who can afford the test, the results may take several months. These delays mean that families might not receive results in time to inform reproductive decisions based on genetic risk or to plan early interventions for their infant, leaving them uninformed at a critical period. This situation reflects the broader challenge of MBS items in genomics (including paediatrics, renal, cardiac, neurology and cancer), whereby funding a test in isolation without considering staffing costs for laboratories, genetics services and patient perspectives has led to an implementation problem [7].

This frustrating gap and inequity in clinical care must be addressed.

We must ensure there is adequate funding, resources and training for laboratories to complete genomic testing for hearing loss and other conditions, and increase access to genetic counselling through training, funding and recognition of the profession's

TABLE 1 | Services Australia utilisation data for childhood hearing loss Medicare Benefits Schedule (MBS) items (November 2023–October 2025) [5, 6].

		Services						
		State						
MBS item	Rebate	NSW	VIC	QLD	SA	WA	TAS	Total
73,440 singleton	$1200	55	23	4	29	20	2	133
73,441 trio	$2100	0	16	3	0	0	0	19
73,442 reanalysis	$500	1	0	0	0	0	0	1
73,443 cascade	$400	34	0	0	5	0	0	39
73,444 reproductive partner	$1200	1	0	0	0	0	0	1
Total		91	39	7	34	20	2	193

invaluable work. Most of all, we owe affected families equitable access to the opportunity for genomic testing to provide better, personalised and informed care.

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Emma McGonigal: writing – original draft and writing – review and editing. Andrew White: conceptualisation, supervision and writing – review and editing. Valerie Sung: supervision and writing – review and editing. Karen Liddle, Lilian Downie and Emily Shepard: writing – review and editing.

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Conflicts of Interest

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