Ordering Medicare-funded genomic testing for childhood hearing loss

A guide for paediatricians and ENT surgeons

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Step 1.
Is my patient eligible?

Has the child or family already had genetic testing? Discuss with a genetics experts before proceeding with further testing.

Patients must meet ALL of the following criteria:[1]

- ☐ Has congenital or childhood onset hearing loss before 18 years of age
- ☐ Hearing loss (unilateral or bilateral) is moderate to profound in the worst ear [2]
- ☐ Hearing loss is classified as sensorineural, auditory neuropathy or mixed
- ☐ Child is not eligible for a childhood syndromes exome [3]

1. Criteria for MBS Item Number 73440 (singleton exome) and 73441 (trio exome)
2. Moderate to profound hearing loss is >40 dB over 3 frequencies
3. Children are eligible for the childhood syndromes exome (MBS Item 73358) if they have dysmorphic features, congenital anomalies, intellectual disability or developmental delay

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Step 2.
Pre-test counselling

- What is the family's lived experience of hearing loss?
- Reasons for genetic testing e.g. benefits of a diagnosis, access to community support, future pregnancy planning
- What are the family's values and goals?
- Potential outcomes of genetic testing - positive, negative, uncertain and incidental findings
- How would the family cope with various testing results?
- Practicalities of testing - type of sample, turnaround time etc.

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Step 3.
Arrange genomic testing

Discuss all aspects of the consent form with the patient's family.

Complete the consent form, and document under the patient file.

Order singleton or trio exome testing. The ordering process is usually site dependent.

Example of <u>consent form</u> from *Australian Genomics*

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Step 4.
Contact an expert

Contact a genetic counsellor or geneticist with any unanswered questions from you or the family

Do not refer to genetics (paediatricians can order genomic hearing loss tests independently)

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Step 2.
Post-test counselling

- Check in with the family - are they ready and expecting to receive the results?
- What do the results mean for the child? (e.g. management implications)
- What do the results mean for the family? (e.g. recurrence risk, inheritance patterns)
- For VUS results, how does the family process uncertainty?
- For incidental findings, what are the medical implications? Does the family recall the possibility of an unexpected result coming up?
- Noticing body language and asking questions to assess understanding of the result
- Referring to genetics when the family requires additional support for complex results e.g. discussing prenatal options or clinical actions based on VUS results and incidental findings

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Frequently Asked Questions

Q: Are there any pre-requisite genetic tests for the childhood hearing loss exome?

A: No. There is no need to order Connexins or microarray testing first.

Q: Are connexins included in the childhood hearing loss exome?

A: Yes, *GJB2* and *GJB6* are both included.

Q: Does the exome include mitochondrial testing for aminoglycoside ototoxicity?

A: No. This can be ordered separately if clinically indicated (e.g. family history or sudden onset hearing loss with administration of medication)

Q: What is the difference between a singleton and trio exome test?

A: A singleton exome only tests the child, whereas a trio exome tests the child and both parents. A trio exome can help clinicians determine the source of a child's genetic variant(s), and whether these variants are pathogenic.

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