New MBS items for genetic testing for childhood hearing loss

Last updated: 22 November 2023

- From 1 November 2023, five new Medicare Benefits Schedule (MBS) items were introduced to support genetic testing for childhood hearing loss.
- The new items support testing to determine whether the cause of hearing loss in affected patients is genetic, determine future risk, inform clinical management in asymptomatic biological relatives of affected patients and enable informed reproductive decision making where appropriate.
- The change is relevant for medical practitioners, specialists and consultant physicians who manage patients with hearing loss.
- The change is also relevant for biological relatives and reproductive partners of patients known to carry a genetic variant(s) associated with hearing loss.

What are the changes?

From 1 November 2023, five new MBS items were introduced to support genetic testing for childhood hearing loss (HL). **Attachment A** to this factsheet lists the new items.

Childhood HL can be genetic (can be inherited) or non-genetic. The new items support singleton (affected individual only) and trio (affected individual and both biological parents) virtual gene panel-based analysis of whole exome sequencing (WES) or whole genome sequencing (WGS) data to determine whether the cause of hearing loss is genetic. The new items also support re-analysis of genetic data taken in the initial test.

Testing is supported in affected patients whose HL presented before 18 years of age. That is not restricted to those who are under 18 years of age at the time of testing. The new items support testing in affected patients regardless of whether their HL is ‘syndromic’ (one part of a syndrome that affects the patient in other ways) or non-syndromic (where the patient has no symptoms other than HL).

The new items also support testing in biological relatives and reproductive partners of patients known to carry a genetic variant(s) associated with hearing loss to determine future risk, inform clinical management, and enable informed reproductive decision making where appropriate.

For private health insurance purposes, the new items were listed under the following clinical category and procedure type:

- New items 73440, 73441, 73442, 73443, and 73444:
    - Clinical category: Support List (pathology)
    - Procedure type: Type C

Why are the changes being made?

Genetic testing by WES or WGS methods are more effective for affected patients than sequencing in the *GJB2* and *GJB6* genes only, which is the current standard of care. The new MBS items may allow affected patients to avoid further investigations such as magnetic resonance imaging (MRI) and also reduce the time to diagnosis, resulting in better patient health outcomes.

There may also be non-health benefits to this testing, such as supporting patients to access appropriate schooling, including bilingual education, and other accommodations they may require.

At its meeting in November 2022, the Medical Services Advisory Committee (MSAC) supported the creation of five new MBS items for genetic testing for childhood hearing loss under MSAC Application Application 1680. Further details about MSAC applications can be found under MSAC Applications on the MSAC website (Medical Services Advisory Committee).

What does this mean for requestors and providers?

Specialists or consultant physicians are now able to request genetic testing for people whose hearing loss first presented before 18 years of age, data re-analysis in these patients, and testing in the reproductive partners of patients known to carry a genetic variant(s) associated with hearing loss.

Medical practitioners are able to request genetic testing for biological relatives of patients known to carry a genetic variant(s) associated with hearing loss.

To be eligible for Medicare benefits, laboratories providing this service must be accredited according to the pathology accreditation standards specified in the *Health Insurance (Accredited Pathology Laboratories-Approval) Principles 2017.*

How will these changes affect patients?

Listing this service allows affected individuals to receive confirmation of whether the cause of hearing loss is genetic, which will inform clinical management and result in better health outcomes.

The new service also allows biological relatives and reproductive partners of patients known to carry a genetic variant(s) associated with hearing loss to access genetic testing to determine whether they also carry a relevant a genetic variant(s). This testing determines future risk, inform clinical management, and enable informed reproductive decision making where appropriate.

Who was consulted on the changes?

Consultation input was received from twelve organisations, all of whom were supportive of the application: Australian Genomics, Australasian Newborn Hearing Screening Committee,

Australian Pathology, Aurora School, Centre for Genetics Education NSW Health, Deafness Foundation, Genetic Undiagnosed and Rare Disease Collaborative Australia, the Human Genetics Society of Australasia including its Ethics and Social Issues Committee, Neurodevelopmental and Behavioural Paediatric Society of Australasia, Public Pathology Australia, the Royal College of Pathologists of Australasia, and UsherKids Australia.

How will the changes be monitored and reviewed?

All MBS items are subject to compliance processes and activities, including random and targeted audits which may require a provider to submit evidence about the services claimed.

Where can I find more information?

The full item descriptor(s) and information on other changes to the MBS can be found on the MBS Online website at www.mbsonline.gov.au. You can also subscribe to future MBS updates by visiting MBS Online and clicking 'Subscribe'.

The Department of Health and Aged Care provides an email advice service for providers seeking advice on interpretation of the MBS items and rules and the *Health Insurance Act 1973* and associated regulations. If you have a query relating exclusively to interpretation of the Schedule, you should email askMBS@health.gov.au.

Private health insurance information on the product tier arrangements is available at www.privatehealth.gov.au. Detailed information on the MBS item listing within clinical categories is available on the Department's website. Private health insurance minimum accommodation benefits information, including MBS item accommodation classification, is available in the latest version of the *Private Health Insurance (Benefit Requirements) Rules 2011* found on the Federal Register of Legislation. If you have a query in relation to private health insurance, you should email PHI@health.gov.au.

Subscribe to 'News for Health Professionals' on the Services Australia website and you will receive regular news highlights.

If you are seeking advice in relation to Medicare billing, claiming, payments, or obtaining a provider number, please go to the Health Professionals page on the Services Australia website or contact the Services Australia on the Provider Enquiry Line – 13 21 50.

The data file for software vendors when available can be accessed via the Downloads page.

Attachment A:

New item descriptors (commenced 1 November 2023)

Category 6 – Pathology Services

Group P7 - Genetics

73440

Genomic testing and copy number variant analysis of genes known to be causative or likely causative of childhood hearing loss in a patient, if:

(a) the testing and analysis is requested by a specialist or consultant physician; and

(b) the patient has congenital or childhood onset hearing loss that presented before the patient was 18 years of age and is permanent moderate, severe, or profound (>40 dB in the worst ear over 3 frequencies) and classified as sensorineural, auditory neuropathy or mixed; and

(c) the patient is not eligible for a service to which item 73358 or 73359 applies; and

(d) the testing and analysis is not associated with a service to which item 73441 applies

Applicable once per lifetime

MBS Fee: $1,200.00

Benefit: 75% = $900.00 85% = $1,101.30 (Greatest Permissible Gap (GPG) will apply)

73441

Genomic testing and copy number variant analysis of relevant genes known to be causative or likely causative of childhood hearing loss in a patient, if:

(a) the testing and analysis is requested by a specialist or consultant physician; and

(b) the patient has congenital or childhood onset hearing loss that presented before the patient was 18 years of age and is permanent bilateral moderate, severe, or profound (>40 dB in the worst ear over 3 frequencies) and classified as sensorineural, auditory neuropathy or mixed; and

(c) the testing and analysis is performed using a sample from the patient and a sample from each of the patient's biological parents; and

(d) the patient is not eligible for a service to which item 73358 or 73359 applies; and

(e) the testing and analysis is not associated with a service to which item 73440 applies

Applicable once per lifetime

MBS Fee: $2,100.00

Benefit: 75% = $1,575.00 85% = $2,001.30 (Greatest Permissible Gap (GPG) will apply)

Category 6 – Pathology Services

73442

Re-analysis of whole exome or genome data obtained under a service to which item 73440 or 73441 applies, for characterisation of previously unreported germline gene variants for childhood hearing loss in a patient, if:

(a) the re-analysis is requested by a specialist or consultant physician; and

(b) the re-analysis is performed at least 24 months after:
  (i) the service to which items 73440 or 73441 applies has been provided to the patient; or
  (ii) a service to which this item applies is performed for the patient

Applicable twice per lifetime

MBS Fee: $500.00

Benefit: 75% = $375.00 85% = $425.00

73443

Characterisation of one or more familial germline gene variants known to be causative or likely causative of childhood hearing loss in a person, if:

(a) the person tested is a biological relative of a patient with a germline gene variant known to be causative or likely causative of hearing loss confirmed by laboratory findings; and

(b) the result of a previous proband testing is made available to the laboratory undertaking the characterisation

MBS Fee: $400.00

Benefit: 75% = $300.00 85% = $340.00

73444

Characterisation of all germline variants in one or more genes known to cause hearing loss in a person, if:

(a) the characterisation is requested by a specialist or consultant physician; and

(b) the characterisation is for the reproductive partner of a patient with a pathogenic or likely pathogenic recessive germline gene variant known to cause hearing loss confirmed by laboratory findings; and

(c) the result of the patient's previous testing is made available to the laboratory undertaking the characterisation

MBS Fee: $1,200.00

Benefit: 75% = $900.00 85% = $1,101.30 (Greatest Permissible Gap (GPG) will apply)

Category 6 – Pathology Services

PN.0.23 - Informed consent and genetic counselling for genetic tests

Items 73297, 73300, 73305, 73334, 73339, 73340, 73393, 73394, 73417, 73418, 73440, 73441, 73442, 73443, and 73444

Prior to ordering these tests the ordering practitioner should ensure the patient (or approximate proxy) has given written informed consent. Testing should only be performed after genetic counselling. Appropriate genetic counselling should be provided to the patient either by the specialist treating practitioner, a genetic counselling service or a clinical geneticist on referral. Further counselling may be necessary upon receipt of the test results.

Items 73295, 73296, 73304, 73333, 73392, 73395, 73416 and 73419

Patients who are found to have any form of affected allele should be referred for post-test genetic counselling as there may be implications for other family members. Appropriate genetic counselling should be provided to the patient either by the specialist treating practitioner, a genetic counselling service or a clinical geneticist on referral.

PN.0.27 - Germline gene mutation tests (Items 73416 and 73392)

Items 73416 and 73392

The rapidly expanding field of genomic medicine has resulted in recognition of an increasing number of genetic causes of cardiac diseases. Use of genomic testing methods that permit reanalysis of existing data for variants in newly described clinically relevant genes are recommended/encouraged.

PN.7.13 - Genetic testing for childhood hearing loss - MBS items 73440, 73441, 73443, 73444

MBS Items 73440 and 73441

When determining the genes to be assessed on the virtual panel, the list of phenotypically driven genes should be based on a recognised test directory.

MBS Items 73443 and 73444

Prior to requesting or performing these tests, the requesting practitioner or pathologist should consider if the patient has previously received equivalent testing. Testing should not be required more than:

- once per variant per lifetime, for item 73443
- once per gene per lifetime, for item 73444

Additional testing should only be performed if it is clinically relevant.

Please note that the information provided is a general guide only. It is ultimately the responsibility of treating practitioners to use their professional judgment to determine the most clinically appropriate services to provide, and then to ensure that any services billed to Medicare fully meet the eligibility requirements outlined in the legislation.

This factsheet is current as of the Last updated date shown above and does not account for MBS changes since that date.