



New pathology MBS items for single gene testing for neuromuscular disorders (NMDs)

Last updated: 13 June 2023

- From 1 July 2023, two new items will be listed on the Medicare Benefits Schedule (MBS), and one existing item amended, for single gene testing for the diagnosis of NMDs to ensure patients suspected of having NMDs are better identified and more appropriately managed.
- These changes are relevant for medical practitioners that specialise in or consult on neuromuscular disorders, neurogenetics, and/or genetic pathology.

What are the changes?

From 1 July 2023, two new items (73434 and 73435) and one amended item (73427) for single gene testing for the diagnosis of NMDs will be listed on the MBS. **Attachment A** to this factsheet provides detail on the new items and item descriptors.

The new MBS items will be available for patients who are suspected of having NMDs, biological relatives of affected individuals for the purpose of identifying the causative variant(s), and for reproductive partners of patients who have been diagnosed with an inherited genetic variant linked to an NMD.

The Medical Services Advisory Committee (MSAC) previously supported public funding through the MBS for gene panel testing to diagnose NMDs. These items were listed on the MBS from 1 November 2022 (MBS items 73422 to 73428). However, there are some NMDs caused by specific types of genetic variants that the gene panel test **cannot** detect. The new MBS items for single gene testing of NMDs is intended to be undertaken **prior to** gene panel testing, where applicable.

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

- New items 73434, 73435 and amended item 73427:
 - Clinical category: Support List (pathology)
 - Procedure type: Type C

Why are the changes being made?

NMDs are a broad range of disorders affecting nerves and/or muscles and how they function. A great proportion of NMDs present antenatally or in early infancy and are associated with significant disability or life-threatening complications.

In May 2022, the Medical Services Advisory Committee (MSAC) supported the creation of new MBS items under [Supplement to Application No. 1585 – Addition of single gene testing for the diagnosis of heritable neuromuscular condition](#) for the purposes of single gene testing for patients with suspected NMDs, cascade testing and reproductive partner testing.

Prior to the listing of these items, NMDs would be diagnosed by gene panel testing, which cannot detect some types of genetic variants linked with NMDs.

What does this mean for requesters and providers?

Specialists who manage patients with suspected NMDs will be able to request single gene testing to rapidly detect gene variants linked to NMDs. Relatives and reproductive partners of patients and patients planning families will also be able to determine their risk of disease or having an affected child respectively. Single gene testing for NMDs should be undertaken **prior to** panel testing, where applicable.

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?

The listing of these services will allow diagnosis of certain NMDs without biopsy, as well as allow identification of causative genetic variants – which can improve patient management and better inform family planning.

Who was consulted on the changes?

Consultation was undertaken with the Royal College of Pathologists of Australasia (RCPA) and the Murdoch Children’s Research Institute (MCRI).

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 and amended item descriptors and explanatory notes (to take effect 1 July 2023)

New items and explanatory notes:

Category 6 – Pathology Services

Group P7 - Genetics

73434

Detection of pathogenic or likely pathogenic gene variants, requested by a specialist or consultant physician, for any of the following:

- (a) a patient with a suspected neuromuscular disorder;
- (b) a relative of a patient with a pathogenic or likely pathogenic germline gene variant associated with a neuromuscular disorder (confirmed by laboratory findings);
- (c) the reproductive partner of a patient with a recessive pathogenic or likely pathogenic germline gene variant associated with a neuromuscular disorder (confirmed by laboratory findings)

Applicable once per gene per lifetime

MBS Fee: \$392.00

Benefit: 75% = \$294.00 85% = \$333.20

PN.7.11

Single gene testing for the diagnosis of heritable neuromuscular disorders - item 73434

Where a recessive variant has been identified using item 73434, reproductive partners should first be tested using item 73434, prior to gene sequencing under item 73427 where no relevant variant was detected by item 73434 and if considered appropriate, after genetic counselling.

Item links: 73434

PN.7.12

Item 73434 minimum gene list

Testing for pathogenic or likely pathogenic gene variants associated with neuromuscular disorders (item 73434) should include, but is not limited to, any one of the following genes: DMPK, CNBP, HTT, PABPN1, C9orf72, AR, SMN1, PRNP, MTND1, MT-ND4, MT-ND4L, MT-ND6, MT-TK, MT-L1, MT-ATP6, FXN, ATN1; or all five of the following genes: ATXN1, ATXN2, ATXN3, CACNA1A, ATXN7.

Item links: 73434

Category 6 – Pathology Services

Group P7 - Genetics

73435

Detection of pathogenic or likely pathogenic DUX4 gene variants, requested by a specialist or consultant physician, for:

- (a) a patient with a suspected neuromuscular disorder; or
- (b) a relative of a patient with a pathogenic or likely pathogenic germline gene variant associated with a neuromuscular disorder (confirmed by laboratory findings)

Applicable once per gene per lifetime

MBS Fee: \$1,000

Benefit: 75% = \$750.00 85% = \$906.80*

Amended Items and explanatory notes (deletions in red strikethrough, additions in red text):

Category 6 – Pathology Services

Group P7 - Genetics

73427

Single gene testing for the characterisation of a germline gene variant or germline gene variants:

- a) if requested by a specialist or consultant physician; and
- b) within the same gene in which the patient's reproductive partner has a documented pathogenic germline recessive gene variant for a neuromuscular disorder identified by a service described in:
 - i) item 73422, 73425 or 73426; or
 - (ii) item 73434, if the patient has been provided a service described in item 73434 and that service has not identified a relevant variant

Applicable once per gene

MBS Fee: \$1,200.00

Benefit: 75% = \$900.00 85% = \$1106.80*

Item links: 73422, 73425, 73426

* [Greatest Permissible Gap](#) applies.

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.