



New pathology MBS items for genetic testing for neuromuscular disorders (NMDs)

Last updated: 5 October 2022

What are the changes?

From 1 November 2022, seven new items (73422 – 73428) for genetic testing for the diagnosis of NMDs will be listed on the Medicare Benefits Schedule (MBS). **Attachment A** to this factsheet lists the new MBS items.

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), reproductive partners of patients who have been diagnosed with an inherited genetic variant linked to a NMD, and data re-analysis to support re-testing where new genetic variants of significance are identified.

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

- MBS items 73422 – 73424 and MBS items 73427 and 73428:
 - Clinical category: Support list (pathology)
 - Procedure type: Type C
- MBS items 74425 and 73426:
 - Clinical category: Support list (pathology)
 - Procedure type: Type B

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 July 2021, the Medical Services Advisory Committee (MSAC) supported the creation of new MBS items for the genetic testing of eligible patients for the purposes of diagnosis, providing prognostic information, enabling cascade testing of at-risk family members and enabling informed reproductive decision-making. [1585 - Genetic testing for the diagnosis of early-onset or familial neuromuscular disorders](#).

Prior to the listing of these MBS items, NMDs would be diagnosed by imaging studies, muscle and/or nerve biopsies, and nerve conduction studies. MSAC considered genetic testing to be more effective.

What does this mean for requesters and providers?

Specialists who manage patients with suspected NMDs will be able to request a confirmatory test to determine appropriate treatment. Relatives of patients and patients planning families will also be able to determine their risk of disease or having an affected child respectively.

To be eligible for Medicare rebates, 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 this service will lead to more rapid diagnosis and better targeted treatment, in order to improve patient outcomes.

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 descriptors and information on other changes to the MBS will be available on the MBS Online website at www.mbsonline.gov.au. You can also subscribe to future 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, please email askMBS@health.gov.au.

Subscribe to '[News for Health Professionals](#)' on the Services Australia website to 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 Services Australia on the Provider Enquiry Line – 13 21 50.

The data file for software vendors will be available via the MBS Online website under the [Downloads](#) page.

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

Attachment A: item descriptor details

New item 73422

Descriptor: Characterisation of a gene variant or gene variants using a gene panel, in a patient presenting with clinical signs and symptoms suggestive of a genetic neuromuscular disorder (other than signs and symptoms associated with variants that are not detected by massively parallel sequencing), if the service is requested:

- (a) by a specialist or consultant physician; and
- (b) after exclusion of non-genetic causes

Applicable once per lifetime

(See para PN.7.6 of explanatory notes to this Category)

MBS Fee:\$1,200.00

Benefit: 75% = \$900.00 85% = \$1,112.10*

New item 73423

Descriptor: Detection of a single identified gene variant, in a biological relative of a person with a germline gene variant for a neuromuscular disorder identified by a service described in item 73422, 73425 or 73426, if the service is requested by a specialist or consultant physician

Applicable once per variant

MBS Fee:\$500.00

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

New item 73424

Descriptor: Prenatal detection of an actionable pathogenic familial gene variant or gene variants (including maternal cell contamination assessment), requested by a specialist or consultant physician, for a genetic neuromuscular disorder previously identified in an index person in the patient's family as a result of a service described in item 73422

Applicable once per pregnancy

MBS Fee:\$1,600.00

Benefit: 75% = \$1,200.00 85% = \$1,512.10*

New item 73425

Descriptor: Prenatal detection of unknown gene variants (including maternal cell contamination assessment) using a gene panel, if:

- (a) the service is requested:
 - (i) by a specialist or consultant physician, for a suspected genetic neuromuscular disorder; and
 - (ii) after exclusion of non-genetic causes; and

(b) the service is performed using a sample from the fetus; and

(c) the service is not performed in conjunction with a service to which item 73426 applies

Applicable once per pregnancy

MBS Fee:\$1,800.00

Benefit: 75% = \$1,350.00 85% = \$1,712.10*

New item 73426

Descriptor: Prenatal detection of unknown gene variants (including maternal cell contamination assessment) using a gene panel, if:

- (a) the service is requested:
 - (i) by a specialist or consultant physician; and
 - (ii) for a suspected genetic neuromuscular disorder; and
 - (iii) after exclusion of non-genetic causes; and
- (b) the request states that singleton testing is inappropriate; and
- (c) the service is performed using a sample from the fetus and a sample from each of the fetus's biological parents; and
- (d) the service is not performed in conjunction with a service to which item 73425 applies

Applicable once per pregnancy

MBS Fee:\$2,400.00

Benefit: 75% = \$1,800.00 85% = \$2,312.10*

New item 73427

Descriptor: Single gene testing for the characterisation of a germline gene variant or germline gene variants, if requested by a specialist or consultant physician, 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 item 73422, 73425 or 73426

Applicable once per gene

MBS Fee:\$1,200.00

Benefit: 75% = \$900.00 85% = \$1,112.10*

New item 73428

Descriptor: Re-analysis of whole genome or exome data obtained in performing a service described in item 73422, 73425 or 73426, for characterisation of previously unreported gene variants related to the clinical phenotype, if the re-analysis is requested by:

- (a) a consultant physician practicing as a clinical geneticist; or
- (b) a consultant physician practising as a specialist paediatrician, following consultation with a clinical geneticist

Applicable twice per lifetime

(See para PN.7.7 of explanatory notes to this Category)

MBS Fee:\$500.00

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

New note PN.7.6

Genetic testing for the diagnosis of neuromuscular disorders (NMDs) (item 73422)

Single gene tests for variants that are not detectable using next generation sequencing (NGS) methods (such as in SMN1, DMPK1, DUX4 or DMD) should be conducted before panel testing, where one of these NMDs is clinically suspected.

New note PN.7.7

Genetic testing for the diagnosis of neuromuscular disorders (NMDs) - data reanalysis (item 73428)

Variants may be previously unreported because the relevant gene was not included in the original virtual panel, or because the pathogenicity of the variant has been re-classified in the interim.

- [MBS fee is subject to the Greatest Permissible Gap.](#)