



New pathology items for targeted carrier testing for severe monogenic conditions

Last updated: 22 November 2023

- From 1 November 2023, three new Medicare Benefits (MBS) items were introduced for reproductive carrier testing for genetic variants known to cause nine severely disabling and/or life-threatening conditions, for which the Ashkenazi Jewish community is at significantly higher risk compared to the general population, in addition to Cystic Fibrosis (CF), Spinal muscular atrophy (SMA) and Fragile-X syndrome (FXS).
- Testing is available for people who identify as being of Ashkenazi Jewish descent and their reproductive partners.

What are the changes?

Effective 1 November 2023, three new items (73453, 73454 and 73455) for gene panel testing were introduced for variants in 12 genes relating to 12 conditions. The conditions are Tay-Sachs disease, Canavan disease, Familial dysautonomia, Mucopolysaccharidosis type IV, Glycogen storage disease type 1, Fanconi anaemia type C, Gaucher disease type 1, Niemann Pick disease type A and Bloom syndrome, CF, SMA and FXS.

People with Ashkenazi Jewish ancestry have one in five chance (20%) of being a genetic carrier for at least one of these conditions. Testing is available to people who identify as being of Ashkenazi Jewish descent (and who are of reproductive age) and their reproductive partners. **Attachment A** to this factsheet lists the new items.

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

- Clinical category: Support List (pathology)
- Procedure type: Type C

Why are the changes being made?

The changes allow equitable access to carrier testing for a high risk group. Previously, carrier testing in the Ashkenazi Jewish population was undertaken on a user-pays basis or through small-scale programs funded by private organisations, which was not accessible to all eligible patients.

The listing of this service was recommended by the Medical Services Advisory Committee (MSAC) in July 2022 under [application 1671 – Targeted carrier testing for severe monogenic](#)

[conditions](#). Further details about MSAC applications can be found under [MSAC Applications](#) on the MSAC website ([Medical Services Advisory Committee](#)). MSAC noted the high community demand in the Ashkenazi Jewish population to access such testing given the known severity of the conditions.

What does this mean for requestors and providers?

Treating practitioners are now able to request carrier testing for patients of Ashkenazi Jewish descent and their reproductive partners to determine the risk of having an affected child.

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 changes provide greater access to testing for patients of Ashkenazi Jewish descent (and their reproductive partners) who are planning families, leading to more informed reproductive-decision making.

Who was consulted on the changes?

Consultation was undertaken with the Royal College of Pathologists of Australasia, the Victorian Clinical Genetics Service, Private Pathology Australia, Australian Genomics Health Alliance, Australian Pathology, Human Genetics Society of Australasia, Royal Australian College of General Practitioners, Spinal Muscular Atrophy Australia (SMA Australia) and one individual.

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 P2 - Genetics

73453

Characterisation of germline pathogenic or likely pathogenic gene variants:

(a) in at least the following genes:

- (i) ASPA;
- (ii) BLM;
- (iii) CFTR;
- (iv) ELP1;
- (v) FANCA;
- (vi) FANCC;
- (vii) FANCG;
- (viii) FMR1;
- (ix) G6PC1;
- (x) GBA1;
- (xi) HEXA;
- (xii) MCOLN1;
- (xiii) SLC37A4;
- (xiv) SMN1;
- (xv) SMPD1; and

(b) in a patient of reproductive age who is of Ashkenazi Jewish descent, for the purpose of ascertaining the patient's carrier status for the following:

- (i) Bloom syndrome;
- (ii) Canavan disease;
- (iii) Cystic fibrosis;
- (iv) Familial dysautonomia;
- (v) Fanconi anaemia type C;
- (vi) Fragile-X syndrome;
- (vii) Gaucher disease;
- (viii) Glycogen storage disease type I;
- (ix) Mucopolysaccharidosis type IV;

Category 6 pathology Services

- (x) Niemann-Pick disease type A 7;
- (xi) Spinal muscular atrophy;
- (xii) Tay-Sachs disease

Applicable once per lifetime.

MBS fee: \$425.00

Benefit: 75% = \$318.75 85% = \$361.25

PN 7.18

Where the couple is already pregnant and both patients are of Ashkenazi Jewish descent, concurrent testing of any partner(s) not already tested is recommended.

73454

Whole gene sequencing of a gene or genes described in item 73453, in a patient who is the reproductive partner of an individual who is affected by or a known genetic carrier of one or more conditions described in item 73453 (other than cystic fibrosis, fragile-X syndrome or spinal muscular atrophy), for the purpose of determining the couple's combined reproductive risk of the conditions, if:

- (a) the patient is not eligible for a service to which item 73453 applies; and
- (b) the patient has not received a service to which item 73453 applies; and
- (c) the patient has not received a service to which this item applies for the purpose of determining their reproductive risk with their current reproductive partner.

Applicable once per couple per lifetime.

MBS fee: \$1,200.00

Benefit: 75% = \$900.00 85% = \$1,101.30

73455

Testing of a pregnant patient, where at least one prospective parent is known to be affected by or is a genetic carrier of one or more conditions described in item 73453, for the purpose of determining whether familial variant or variants are present in the fetus, if:

- (a) the detection is requested by a specialist or consultant physician; and
- (b) the fetus is at 25% or more risk of inheriting a condition described in item 73453.

MBS fee: \$1,600.00

Benefit: 75% = \$1,200.00 85% = \$1,501.30

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.