Reproductive carrier testing for cystic fibrosis, spinal muscular atrophy and fragile X syndrome

Last updated: 22 November 2023

- From 1 November 2023, two new Medicare Benefits Schedule (MBS) items were introduced for genetic reproductive carrier testing for cystic fibrosis (CF), spinal muscular atrophy (SMA) and fragile X syndrome (FXS).
- The new items support testing in all Medicare eligible individuals who are pregnant or are planning a pregnancy, and their reproductive partners, to determine their reproductive risk of having a child affected by one of the three conditions.
- These changes are relevant for medical practitioners and geneticists who manage patients who are pregnant or are planning a pregnancy.

What are the changes?

New MBS items 73451 and 73452

From 1 November 2023, new MBS items 73451 and 73452 were introduced to support genetic reproductive carrier testing for CF, SMA and FXS for people who are pregnant or are planning a pregnancy, and their reproductive partners. **Attachment A** to this factsheet lists the new items.

Reproductive carrier testing identifies whether an individual is a genetic carrier for a particular genetic condition, meaning they have specific changes in their genetic sequence that can cause a condition, despite showing no symptoms of the condition.

New MBS items 73451 and 73452 allow people to determine the reproductive risk of having a child affected by CF, SMA, or FXS and allow them to make informed decisions regarding current or future pregnancies on the basis of this risk.

Contrary to most other genetic tests currently funded on the MBS, which require patients to satisfy certain clinical or family history criteria, the new MBS items are available to any Medicare eligible individual who is pregnant, or planning pregnancy in the absence of family history of CF, SMA or FXS,.

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

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

Carrier testing for fragile X syndrome

FXS differs from CF and SMA, because it is inherited in an 'X-linked dominant' fashion, therefore the likelihood of a child being born with FXS is dictated by the carrier status of the patient who is already pregnant or planning pregnancy.

For this reason, the intent of the new MBS items is to:

- first test the patient who is pregnant or planning pregnancy for CF, SMA **and** FXS under MBS item 73451; then
- subsequently, test the reproductive partner of the patient who is pregnant or planning pregnancy for CF and SMA **only** under MBS item 73452**.**

That is, the intent is to **not** test the reproductive partner of the patient who is pregnant or planning pregnancy for FXS carrier status.

This is because the FXS carrier status of the reproductive partner is not directly relevant in determining whether a child will be born with FXS. For the purposes of determining the risk of having a child with FXS, it would therefore not be informative to conduct carrier testing of the reproductive partner for all three conditions under MBS item 73451.

Why are the changes being made?

CF, SMA and FXS are the three most common inheritable monogenic (involving a single gene) conditions that substantially reduce the life expectancy of those affected.

Reproductive carrier testing for these conditions supports informed reproduction decision-making. These changes also improve equity of access for those who cannot afford to pay privately for the test.

At its meeting in July 2020, the Medical Services Advisory Committee (MSAC) supported the creation of new MBS items for reproductive carrier testing under MSAC application 1573 - Reproductive carrier screening for fragile X syndrome, spinal muscular atrophy and cystic fibrosis. 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?

Medical practitioners who manage patients who are pregnant or planning pregnancy are now able to request reproductive carrier testing for CF, SMA, and FXS for their patients and their reproductive partners.

This testing allows medical practitioners to give advice to their patients about their individual or combined risk of passing on CF, SMA, and FXS to their children. This testing also helps medical practitioners to support their patients in making informed decisions about current or future pregnancies.

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 provides people with the option to be informed of their carrier status for CF, SMA, and FXS.

This allows people to make an informed decision about how to plan a pregnancy, or what to do if they are already pregnant. Reproductive couples identified as high risk may be referred to a clinical genetics service or obstetrician to discuss reproductive options.

Who was consulted on the changes?

The introduction of new MBS items 73451 and 73452 was informed by consultation feedback from the Royal College of Pathologists of Australasia, the Royal Australian and New Zealand College of Obstetricians, Rural Doctors' Association of Australia, Australian Pathology, the Australian Medical Association, Fragile X Association of Australia, Cystic Fibrosis Australia and Spinal Muscular Atrophy 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 and explanatory note (commenced 1 November 2023)

Category 6 – Pathology Services

Group P7 - Genetics

73451

Testing of a patient who is pregnant, or planning pregnancy, to identify carrier status for pathogenic or likely pathogenic variants in the following genes, for the purpose of determining reproductive risk of cystic fibrosis, spinal muscular atrophy or fragile X syndrome:

a. CFTR;
b. SMN1;
c. FMR1

One test per lifetime.

MBS fee: $400.00

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

73452

Testing of the reproductive partner of a patient who has been found to be a carrier of a pathogenic or likely pathogenic variant in the CFTR or SMN1 gene identified by testing under item 73451, for the purpose of determining the couple's reproductive risk of cystic fibrosis or spinal muscular atrophy.

One test per condition per lifetime.

MBS fee: $400.00

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

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MBS items 73451 and 73452

Fragile X syndrome (FXS) is inherited in an X-linked dominant fashion.

The sex chromosomal pattern of a patient determines the likelihood of being a genetic carrier of FXS and the risk of FXS carriers passing on a variant(s) in the FMR1 gene that would cause their child to be born with the condition. Patients with certain sex chromosomal patterns have no risk of influencing whether their child is born with FXS, regardless of whether they are a carrier of the condition.

Category 6 – Pathology Services

The intent of MBS item 73451 is to test a patient who:

(a) is either planning a pregnancy or is already pregnant; and

(b) if found to be a genetic carrier of fragile X syndrome, is at risk of passing on a variant(s) in the FMR1 gene that would cause their child to be born with the condition

The intent of MBS item 73452 is to test a patient who:

(a) is the reproductive partner of the patient planning pregnancy or already pregnant tested under item 73451.

(b) is not at risk of passing on a variant(s) in the FMR1 gene that would cause their child to be born with fragile X syndrome, regardless of whether they are a genetic carrier of the condition

The patient who is planning pregnancy or already pregnant must be tested first under MBS item 73451 prior to testing the reproductive partner patient under MBS item 73452, to ensure an informative and clinically relevant test result is obtained in the FMR1 gene.

MBS item 73451

The laboratory used to undertake reproductive carrier testing under item 73451 should use a methodology appropriate to the clinical setting with:

(a) sufficient diagnostic range and sensitivity to detect at least 95% of pathogenic variants likely to be present in the patient; and

(b) at least 50 of the most frequently encountered cystic fibrosis transmembrane conductance regulator variants in the Australian population.

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.