



Genetic testing for major fetal structural abnormalities

Last updated: 27 October 2021

- From 1 November 2021, a new MBS item will be available for genetic testing for major fetal structural abnormalities, in pregnancies where abnormalities are detected by ultrasound.
- These changes are relevant for all medical practitioners who manage pregnancies and provide antenatal care.

What are the changes?

Major fetal structural abnormalities are typically caused by a genetic condition. From 1 November 2021, pregnant individuals whose fetus has a known major structural abnormality will be able to undergo genetic testing for the fetus via genome-wide microarray (GWMA) testing. The major structural abnormalities must be determined by ultrasound.

Why are the changes being made?

In November 2019, the Medical Services Advisory Committee (MSAC) supported public funding for fetal GWMA testing in pregnancies with major fetal structural abnormalities. In their recommendations, MSAC noted the superiority of GWMA compared to current options for genetic testing in this group, namely karyotyping. Following the recommendation, the Australian Government agreed to public funding of GWMA for major fetal structural abnormalities as part of the 2021-22 Budget.

The item is to be listed in the *Health Insurance (Pathology Service Table) 2020*, Group 7 – Genetics.

What does this mean for providers/referrers/other stakeholders?

Medical practitioners who manage pregnancies and provide antenatal care will be able to request GWMA to determine if an affected fetus has a genetic condition, should the fetus be identified with major structural abnormalities on ultrasound.

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?

This service will be able to inform expectant parents of the likelihood of their fetus having a genetic condition. This in turn may guide decisions surrounding the management of the pregnancy.



Who was consulted on the changes?

Consultation has been undertaken with key stakeholders, clinical experts and providers, and consumer health representatives as part of the MSAC process, including the Royal College of Pathology Australasia and the Royal Australian and New Zealand College of Obstetrics and Gynaecology.

How will the changes be monitored and reviewed?

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

Significant variation from forecasted expenditure may warrant review and amendment of the items and fees, and incorrect use of MBS items can result in penalties including the health professional being asked to repay monies that have been incorrectly received.

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 provides an email advice service for providers seeking advice on interpretation of the MBS items and rules and the Health Insurance Act and associated regulations. If you have a query relating exclusively to interpretation of the Schedule, you should email askMBS@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 was released on 22 September 2021 and can be accessed 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.