



Genetic testing for childhood syndromes

Last updated: 27 October 2021

What are the changes?

From 1 November 2021, amendments will be made to the descriptors of three items relating to genetic testing for childhood syndromes. These items are:

- amended item 73361, for genetic analysis of a single gene variant for detection purposes in a person who has a biological sibling with a known monogenic condition,
- amended item 73362, for genetic analysis of a single gene variant for detection purposes in a person who has a first-degree relative with a known monogenic condition, and
- amended item 73363, for genetic analysis of a single gene variant for segregation analysis in a person who is a biological parent or biological relative to an individual with a known monogenic condition.

Why are the changes being made?

The Medical Services Advisory Committee (MSAC) recommended the creation of new items for genetic testing for monogenic childhood syndromes in August 2019, which were listed on 1 May 2020. It has been identified that item descriptors for items 73361, 73362, and 73363 may be confusing to providers and consumers. The amendments will ensure that these items will be accessible by the intended population identified in the MSAC recommendations and approved by the Australian Government as part of the 2021-22 Budget.

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

Consultant physicians practising as a clinical geneticist or a specialist paediatrician will be able to request MBS-funded genetic tests for siblings or biological relatives of patients with a known monogenic childhood syndrome for cascade testing, as well as MBS-funded genetic segregation analysis for the biological parents or relatives of the affected patient.

To be eligible for Medicare rebates, laboratories providing these services 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?

Monogenic childhood syndromes are caused by genetic variants. When a genetic variant is identified in a patient with a suspected monogenic childhood syndrome, testing can be performed to identify whether this genetic variant can be found in the biological parents or relatives. If the genetic variant is absent from the parents' or relatives' genetic material, this confirms that the genetic variant cannot be inherited, also known as a *de novo* mutation. This is called a segregation analysis.



If a causative genetic variant is identified in a patient with a suspected monogenic childhood syndrome, testing can also be performed to screen individuals who have a biological relationship with the affected patient and identify family members who carry the genetic variant. This is called cascade testing.

The previous item descriptors for cascade testing (item 73361 and 73362) and segregation analysis (item 73363) may be interpreted in a manner that restricts their access due to confusing descriptors of eligibility. The changes will remove any ambiguity and provide access to family members of patients with monogenic childhood syndromes, for whom the items were intended.

Who was consulted on the changes?

Following the listing of these items, the Murdoch Children's Research Institute raised its concerns and was consulted in the drafting of the amendments. In addition, the following peak bodies and professional organisations were consulted: Royal College of Pathologists of Australasia, Victorian Clinical Genetics Service, Australian Mitochondrial Disease Foundation, Genetics and Rare Disease Network Rare Voices Australia, Syndromes Without A Name, Genetic Alliance Australia, and Australian Genomics Health Alliance.

How will the changes be monitored and reviewed?

MBS items 73361, 73362, and 73363 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.