# Alpha thalassaemia genetic testing

Last updated: 8 June 2022

* From 1 July 2022, four new Medicare Benefits Schedule (MBS) items for genetic testing will be introduced which will allow individuals to determine their carrier status for alpha thalassaemia.
* New MBS items 73410, 73411, 73412, and 73413 will support the management of individuals of reproductive age and their reproductive partners who are at risk of being genetic carriers of alpha thalassaemia.
* These changes are relevant for all medical practitioners who manage pregnancies and provide antenatal care, and Approved Pathology Providers (APP) who provide these referred services.

## What are the changes?

Alpha thalassaemia is a genetic disorder which affects red blood cells. The severity of the disease can vary depending on the genetic variant that is inherited. The most severe type, called Hb Bart’s, is almost uniformly lethal for fetuses and is associated with high maternal morbidity.

From 1 July 2022, at-risk individuals of reproductive age and their reproductive partners will be able to access genetic tests under MBS items 73410, 73411, 73412, and 73413 to determine their carrier status for alpha thalassaemia. Services supported under these items will inform reproductive couples of their risk of having a fetus with alpha thalassaemia, and guide subsequent reproductive decisions.

## Why are the changes being made?

In March 2019, the Medical Services Advisory Committee (MSAC) considered the proposal for public funding for genetic testing for alpha thalassaemia and deferred its decision to seek further information on contemporary testing pathways and methodologies. In January 2021, following targeted consultation with peak bodies, the MSAC Executive supported the public funding of genetic testing for alpha thalassaemia.

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

All medical practitioners who manage pregnancies and provide antenatal care will be able to order genetic tests for the determination of alpha thalassaemia carrier status, following screening with a full blood count and an inconclusive thalassaemia study (supported under existing MBS item 65081).

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?

New MBS items 73410, 73411, 73412, and 73413 will inform reproductive couples of the risk of having a fetus with alpha thalassaemia and guide subsequent reproductive decisions. Services supported under these items, which are currently supported under funding arrangements that differ across states and territories, may better ensure equity of access and minimise out-of-pocket payments for genetic testing for alpha thalassaemia.

## Who was consulted on the changes?

Consultation has been undertaken with key stakeholders, clinical experts and providers as part of the MSAC process, including the Royal College of Pathologists of Australasia, the Haematological Society of Australia and New Zealand, and Monash Health.

## 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](http://www.mbsonline.gov.au/). You can also subscribe to future MBS updates by visiting [MBS Online](http://www.mbsonline.gov.au/) 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](https://www.servicesaustralia.gov.au/organisations/health-professionals/news/all)’ 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](http://www.mbsonline.gov.au/internet/mbsonline/publishing.nsf/Content/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.