Genetic testing for heritable kidney disease

Last updated: 8 June 2022

- From 1 July 2022, six new Medicare Benefits Schedule (MBS) items will be introduced for genetic testing for heritable kidney disease other than Alport syndrome.
- The new items will support testing to determine whether the cause of a suspected or known heritable kidney disease is genetic, determine future risk and inform clinical management in asymptomatic biological relatives of affected patients, and enable informed reproductive decision making where appropriate.
- These changes are relevant for clinical geneticists, specialist nephrologists and specialist paediatricians who manage patients with suspected or known heritable kidney disease, specialist or consultant physicians who manage the first-degree relatives of patients known to carry a genetic variant(s) associated with heritable kidney disease, and Approved Pathology Providers (APP) who provide these referred services.

What are the changes?

From 1 July 2022, six new MBS items for genetic testing for heritable kidney disease other than Alport syndrome will be introduced to the MBS.

The new MBS items are:

- MBS item 73401, for genetic testing of variants associated with heritable cystic kidney disease in a patient suspected of having a monogenic condition
- MBS item 73402, for genetic testing of variants associated with heritable kidney disease in a patient with chronic kidney disease suspected of having a monogenic condition
- MBS item 73403, for re-analysis of genetic data obtained in performing a service to which item 73401 or 73402 applies, for testing of previously unreported genetic variants in a patient suspected of having a monogenic condition
- MBS item 73404, for genetic testing of first-degree relatives of an individual who is known to carry a genetic variant(s) associated with heritable kidney disease
- MBS item 73405, for genetic testing of a reproductive partner of an individual who is known to carry a genetic variant(s) associated with heritable kidney disease
- MBS item 73406, for genetic testing of a pregnant patient who is known to carry (or whose reproductive partner is known to carry) a genetic variant(s) associated with heritable kidney disease, to determine whether monogenic variants are present in the fetus

Why are the changes being made?

The listing of the services for genetic testing for heritable kidney disease other than Alport syndrome was recommended by the Medical Services Advisory Committee (MSAC) at its March-April 2021 and July 2021 meetings.

Further details about MSAC applications can be found under MSAC Applications on the MSAC website.

What does this mean for referrers?

Services to which new MBS items 73401, 73402, 73405 and 73406 apply may be referred by a clinical geneticist or specialist nephrologist.

Services to which new MBS item 73403 apply may be referred by a clinical geneticist or specialist paediatrician.

Services to which new MBS item 73404 apply may be referred by a specialist or consultant physician providing professional genetic counselling services.

Services for genetic testing in patients for whom clinical and relevant family history criteria are strongly suggestive of Alport syndrome to which existing MBS items 73298 and 73299 apply may continue to be referred by specialist or consultant physicians.

Appropriate genetic counselling should be provided to patients either by the treating practitioner who acted as the referrer for the service(s), a genetic counselling service or a clinical geneticist on referral. A patient with a diagnosis made under services to which new MBS items 73401, 73402 or 73403 apply should be referred for post-test genetic counselling as there may be implications for other family members who can access testing supported under new MBS item 73404.

What does this mean for providers?

To be eligible for Medicare rebates, a laboratory 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?

The services supported under new MBS items 73401 and 73402 will support patients who have signs or symptoms of a renal abnormality or heritable kidney disease (other than Alport syndrome) to undergo genetic testing to confirm if their suspected or known condition has a genetic cause. This testing may provide certainty of diagnosis and may lead to changes in treatment and clinical management options. It may also reduce the need for more invasive tests.

As the field of genetic and genomic medicine rapidly expands, additional genetic variants may be found to be associated with heritable kidney diseases. Patients who previously received a test to which new MBS items 73401 or 73402 apply and whose condition is still suspected to have a genetic cause may subsequently have their genomic data re-analysed. Medicare benefits may be claimed for these re-analysis services under new MBS item 73403.

Where a genetic cause is found in a patient (as an outcome of testing supported under MBS items 73401 or 73402, or reanalysis under 73403), asymptomatic first-degree relatives may be referred for testing supported under new MBS item 73404. This testing may support early diagnosis and treatment, which may in turn delay disease progression. As first-degree relatives are often considered for living donor kidney transplants, this testing may also reduce the risk of a first-degree relative with undiagnosed kidney disease donating a kidney to their relative. Alternatively, where first-

degree relatives are found not to have a genetic variant, a medical practitioner may reconsider the need for ongoing, regular monitoring for certain kidney diseases.

Reproductive partners of any patients found to have heritable kidney disease with a genetic cause (as an outcome of testing supported under MBS items 73401, 73402, or 73404, or reanalysis under 73403) which has a recessive mode of inheritance may be referred for genetic testing supported under new MBS item 73405. This testing may enable prospective parents to consider, in consultation with a medical practitioner, their combined reproductive risk of passing on heritable kidney disease and make informed reproductive decisions.

Where a pregnant patient and/or their reproductive partner are known to carry or have a heritable kidney disease with a genetic cause which has a recessive mode mode of inheritance, they may undergo genetic testing to determine whether the fetus is at-risk of inheriting the condition supported under new MBS item 73406.

Patients for whom clinical and relevant family history criteria are strongly suggestive of Alport syndrome and first-degree relatives of patients found to have a genetic variant linked to Alport syndrome may continue to undergo testing supported under existing MBS items 73298 and 73299 respectively.

Who was consulted on the changes?

Consultation has been undertaken with key stakeholders, clinical experts, providers, and consumer health representatives as part of the MSAC process.

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.

The utilisation of the new MBS items will be reviewed by MSAC approximately 24 months post-implementation.

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 when available can be accessed via 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.