



# CFTR gene testing

## Medicare update for Doctors

The CFTR gene is responsible for cystic fibrosis (CF) and related disorders. Testing of the CFTR gene is now rebated for certain patients when testing is requested by a specialist or consultant physician.

### Key points

- > Mutations in the CFTR gene are responsible for CF and other clinical disorders (male infertility, bronchiectasis and pancreatitis) which are collectively referred to as CFTR-related disorders (CFTR-RD).
- > From July 1, 2018, rebated testing of the CFTR gene is available in a number of settings:
  - Testing of an individual with clinical or laboratory findings of CF or CFTR-RD.
  - Testing of a fetus to confirm or exclude a diagnosis of CF or CFTR-RD.
  - Testing of an unaffected individual to determine if they are a genetic carrier of CF or CFTR-RD when they have a family history of these disorders, or their reproductive partner is a carrier, or their fetus has ultrasonic findings of echogenic gut.
- > Specific requirements apply for Medicare eligibility. Full item descriptors are available at [www.mbsonline.gov.au](http://www.mbsonline.gov.au).
- > Medicare stipulates that requests for CFTR gene testing can only be made by a medical specialist or consultant physician. General practitioners cannot order rebated CFTR gene testing.
- > Sonic Genetics will continue to provide testing for patients who do not meet the Medicare requirements, including test requests from general practitioners. These patients will be privately billed \$310\*.

**The information in this leaflet may change as new decisions are made by Medicare regarding testing and therapies. Please contact the laboratory for updated information or refer to the Medicare Benefits Schedule (MBS).**

### Testing an individual with clinical or laboratory findings of CF or CFTR-RD

#### MBS Item 73345

Inherited mutations of the CFTR gene cause CF or CFTR-RD. The detection of two pathogenic mutations in the CFTR gene (one on each chromosome) confirms a diagnosis, and may also provide eligibility for access to genotype-specific therapies available on the PBS.

This item covers diagnostic testing for pathogenic CFTR variants for the purpose of investigating, making or excluding a genetic diagnosis of CF or CFTR-RD. The patient must have clinical or laboratory findings highly suggestive of CFTR-associated disease, as determined by the requesting doctor.

### Testing when prenatal ultrasound identifies echogenic gut

#### MBS Items 73346 and 73347

An echogenic or hyperechoic bowel pattern on prenatal ultrasound may suggest intestinal obstruction by abnormal meconium. About one in 10 fetuses with this pattern will have CF, however echogenic gut can also be associated with a number of other disorders, or be a normal variant that resolves by the third trimester.

These MBS items cover testing when a fetus has ultrasonic findings of echogenic gut. Testing is eligible for rebate under the following circumstances:

- Testing of a pregnant woman to determine whether her fetus has pathogenic variants, in order to make or exclude a genetic diagnosis of CF or CFTR-RD in the fetus.
- Testing for pathogenic variants in a prospective parent in order to determine the risk of the fetus having CF or CFTR-RD.

## Testing to determine genetic carrier status

### MBS Items 73348 and 73349

Genetic carriers of pathogenic CFTR gene variants are themselves unaffected by CF or CFTR-RD but are at increased risk of having affected children. An individual has a higher pre-test risk of being a genetic carrier if they have a family history of CF or CFTR-RD. A couple is at greater risk of having an affected child if it is already known that one partner is a genetic carrier.

These items cover CFTR mutation carrier testing of unaffected individuals to determine genetic carrier status. Testing is eligible for rebate under the following circumstances:

- Testing to determine if a patient is a genetic carrier of pathogenic variants previously identified in the family. The patient must have a personal risk of being a carrier of at least 6% (this includes family relatedness of parents, children, full-siblings, half-siblings, grand-parents, grandchildren, aunts, uncles, first cousins and first cousins once-removed, but excludes relatedness of second cousins or more distant relationships).
- Testing for pathogenic variants to determine the reproductive risk of a patient because their reproductive partner is already known to have pathogenic CFTR variants.

While the items do not stipulate that the individual being tested must be an adult, we do not recommend genetic carrier testing in minors unless the family has received genetic counselling from an appropriately qualified professional.

## Testing a fetus for CF or CFTR-RD

### MBS Item 73350

If both partners of a couple are carriers of a pathogenic CFTR variant, there is a 25% risk with each pregnancy of their having a child affected with CF or CFTR-RD. As part of prenatal care, couples may choose testing by CVS or amniocentesis to detect those variants in their fetus.

This item covers testing of a pregnant woman to determine whether her fetus has pathogenic CFTR gene variants, in order to make or exclude a genetic diagnosis of CF or CFTR-RD in the fetus. Testing is eligible for rebate if at least one parent is known to be a genetic carrier, and the fetus must be at 25% risk or greater of having CFTR-related disease due to familial pathogenic gene variants.

## A note on genetic carrier screening

The introduction of Medicare items for CFTR gene testing for only certain patients does not negate the utility of preconception genetic carrier screening in the absence of family history or evidence of an affected fetus.

Requests for genetic carrier screening will still be accepted from GPs and will be privately billed.

## Implications for relatives

The primary purpose of testing the CFTR gene is to confirm a genetic diagnosis of CF or CFTR-RD in, or determine the genetic carrier status of, the individual or fetus being tested. However, the identification of pathogenic variants in the CFTR gene in the patient can have implications for male and female relatives, who may also be affected or be genetic carriers. For this reason, we recommend genetic counselling for patients in whom pathogenic CFTR variants are identified.

## Test sensitivity

Testing for CFTR gene variants can be performed by a number of methods. These methods may test for a single variant, a set or panel of commonly encountered variants, or scan the entire CFTR gene for all variants by full gene sequencing. The ability of each of these methods to detect all CFTR gene variants is referred to as the test sensitivity.

It is important to understand the difference in sensitivity between methods, so as to request the most appropriate test for your patient's circumstances. Factors that may affect choice of test in this regard include the purpose of testing (diagnostic or carrier assessment), whether there is a family history of a previously identified CFTR gene variant and the ethnicity of the patient.

The Medicare items described in this leaflet do not dictate the sensitivity of the test used to conduct testing. By default, requests for CFTR gene testing received by Sonic Genetics will be tested by a 50-mutation panel. This panel will identify approximately 85% of people in Australia who are carriers of CF. If full gene sequencing is required, please contact Sonic Genetics on 1800 010 447 for this to be arranged.

\*Price correct at the time of printing. Please refer to [www.sonicgenetics.com.au](http://www.sonicgenetics.com.au) for current price.