

This provides important information regarding requests for genetic carrier testing of *CFTR* and *FMR1* genes.

Genetic carrier screening provides patients with information about their chance of being a carrier of common inherited conditions: cystic fibrosis (CF) and fragile X syndrome (FXS).

SA Pathology screen for carrier status in the predominant pathogenic variants, which include:

- Over 90% of pathogenic *CFTR* variants
- Repeat expansion of (CCG)<sub>n</sub> within the *FMR1* gene (full repeat expansion results in FXS)

### How to request carrier screening?

SA Pathology request form should include:

1. The test name (i.e., carrier screening for CF)
2. Relevant clinical information (i.e., chronic cough, azoospermia, family history)
3. Requesting clinician details and signature
4. Patient details and signature

### What are the costs associated with carrier screening?

The following patients are eligible for Medicare subsidised testing for CF and FXS under the Medicare Benefits Schedule (MBS).

#### Medicare Rebatable Tests:

Test	MBS Item
CF	MBS 73345 Clinical or laboratory findings suggesting a probability of CF or CF -related disorder: - Respiratory (e.g., chronic cough) - Male infertility (e.g., azoospermia, oligospermia)
	MBS 73348 Family history of CF: - laboratory-established family history of pathogenic <i>CFTR</i> variants - must be within a certain degree of relatedness
	MBS 73349 Patient's partner is affected by CF or is a carrier of a known pathogenic <i>CFTR</i> variant.
FXS	MBS 73300 / 73305 -The patient exhibits intellectual disabilities, ataxia, neurodegeneration, or premature ovarian failure consistent with a <i>FMR1</i> mutation; or -The patient has a relative with a <i>FMR1</i> mutation.

*NB: Although Medicare funding for genetic carrier testing for CF, FXS and SMA has been announced, this rebate is not projected to be available for patients until November 2023.*

#### More Information

Please call SA Pathology on 08 8222 3000 or visit [www.sapathology.sa.gov.au](http://www.sapathology.sa.gov.au)