TEST INFORMATION

Medicare Rebatable Carrier Screening

SA PATHOLOGY

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)n 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 *CFTR* 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 *CFTR* variant.
FXS	MBS 73300 / 73305 -The patient exhibits intellectual disabilities, ataxia, neurodegeneration, or premature ovarian failure consistent with a *FMR1* mutation; or -The patient has a relative with a *FMR1* 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

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