TEST INFORMATION

Reproductive Carrier Screening

This provides important information regarding requests for genetic carrier testing of *CFTR, FMR1* and *SMN1* genes.

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

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)
- The deletion of exon 7 in the *SMN1* gene (two exon 7 deletions account for ~ 95% of SMA cases)

Requesting reproductive carrier screening

The SA Pathology request form should include:

1. The test name: reproductive carrier screening for CFTR, FMR1 and SMN1
2. Requesting clinician details and signature
3. Patient details and signature
4. Financial responsibility

We recommend that testing of both partners be performed prior to conception. The tests can also be done in early pregnancy. Genetic counselling is recommended.

What are the costs associated with reproductive carrier screening?

Currently, Medicare does not cover reproductive carrier screening for CF, FXS and SMA.

All non-MBS patients must be given an SA Pathology Test Request & Agreement to Pay (ATP) form - Genetic Carrier Testing (PUB-0806).

These forms are to be completed and presented at the time of blood collection.

Non-Medicare Rebatable Tests

Test	Cost
CFTR only	$222 ATP required
SMN1 only	$378 ATP required
CFTR + FMR1 + SMN1	$410 ATP required

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

PUB-0804 v2 Published 08/06/22