

**GUIDELINES FOR TESTING FOR FRAGILE X**

In Australia government funding for fragile X testing allows for the following clinical indications:

* Intellectual disability, ataxia, neurodegeneration, or premature ovarian failure consistent with an FMR1 mutation;
* The patient has a relative with a FMR1 mutation.

These terms are broad based and overlap clinically. Patients can present with subtle features of the above terms and should be tested for the following indications:

* Intelligence problems, ranging from learning disabilities to severe mental retardation.
* Social and emotional problems, such as aggression or shyness.
* Autism.
* Attention Deficit and Hyperactivity Disorder (ADHD)
* Developmental delay.
* Speech and language problems.
* Clinical indications suggestive of FXPOI: irregular periods, elevated levels of follicle stimulating hormone, fertility problems or premature ovarian failure (POF)
* Clinical indications suggestive of FXTAS: late onset tremor and ataxia, balance problems, or features similar to Parkinson’s disease.

These presentations can be applied to Medicare item numbers 73300 and 73305.  
  
Ref: Human Genetics Society of Australasia, April 2015.

**PLEASE NOTE:**Both the **American College of Obstetricians and Gynaecologists** **(ACOG)** and **Royal Australian and New Zealand College of Obstetricians and Gynaecologists (RANZCOG)** now support carrier screening including for FXS and in particular recommend this is best offered prior to pregnancy in order to allow more options for subsequent management. Links to the relevant position statements are below:

RANZCOG: <http://bit.ly/2pvkD9r>

ACOG: <http://bit.ly/2oGcoY0>