virtus DIAGNOSTICS

Virtus Diagnostics offers a complete range of general pathology services as well as specialist fertility and genetic testing.

Virtus Diagnostics conducts over a million tests each year across the following disciplines: general pathology, fertility pathology and advanced genetic testing.

We continually innovate and expand this range and with over 200 scientists, many who are internationally renowned in their field, you can be assured you are accessing world class diagnostic services.

Virtus Diagnostics is committed to supporting patients' health throughout their lifetime, and in particular, during their reproductive life. For women this includes general gynaecology and for both men and women it includes general health, fertility planning and conception, through to infertility treatment.

Our service offers:

- Specialist pathologists
- A comprehensive range of tests
- Fast turnaround of results
- Highly skilled and experienced scientists, with access to the latest technology
- Convenient collection centres, and
- Personalised service

Virtus Diagnostics Collection Centres

The network of Virtus Diagnostics collection centres are across NSW, QLD, VIC & TAS. Please refer to virtusdiagnostics.com.au for the complete list of our collection centres.

Virtus Diagnostics General enquiries
1800 837 284

Virtus Diagnostics Accounts enquiries
1800 090 325

virtusdiagnostics.com.au

6 JUNE 2018

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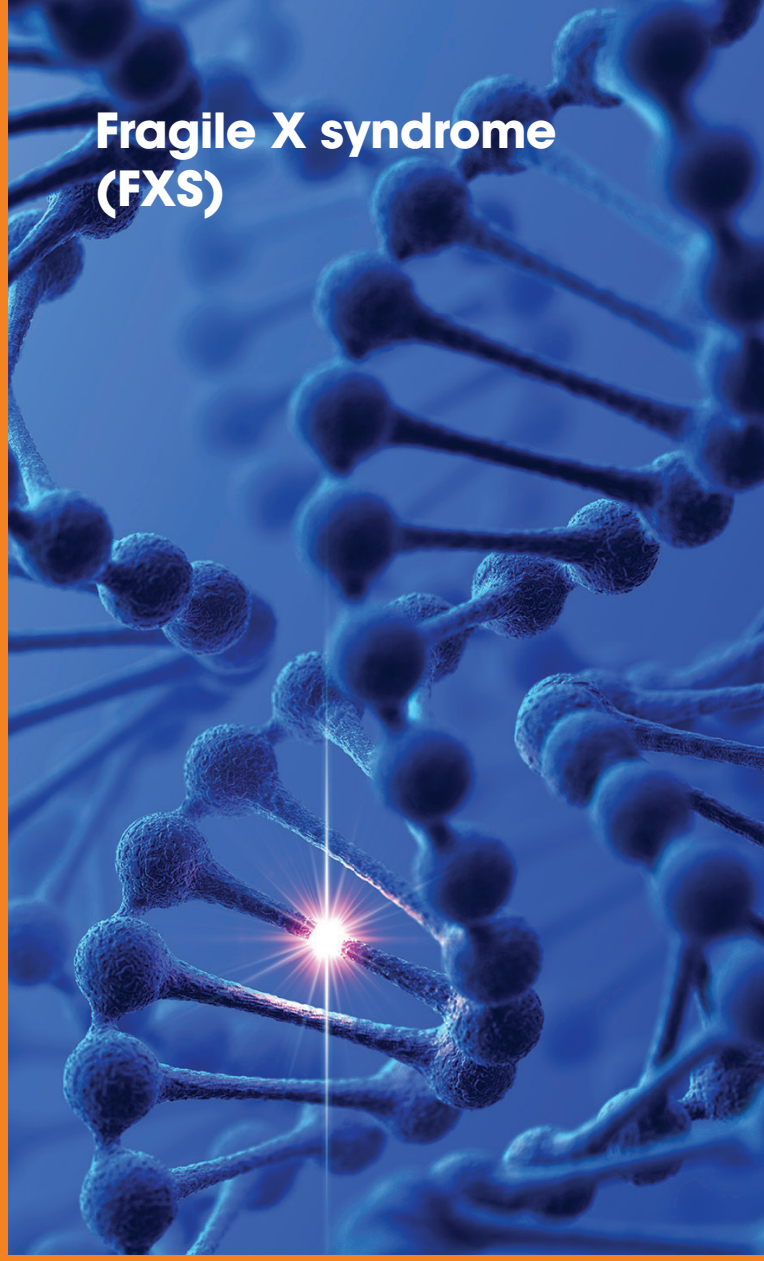
Fragile X syndrome (FXS)



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Fragile X syndrome (FXS)

The facts:

- FXS is a major cause of inherited intellectual disability
- features range from mild to severe
- this is an X-linked disorder and males are more likely to be severely affected than females
- the disorder is inherited from pre-mutation carrier mothers
- presenting features in children include developmental delay, learning difficulties, anxiety, autism and hyperactive behaviour
- some individuals have seizures
- FX and primary ovarian insufficiency (FXPOI) – one in 50 women with an unknown cause of POI are Fragile X carriers
- FX and tremor/ataxia (FXTAS) is a late-onset disorder, usually after the age of 50 years and males who carry the pre-mutation repeat are more likely to be affected than females (40% vs 16%)

Fragile X and intellectual disability:

Fragile X Syndrome (FXS) occurs in approximately 1 in 4,000 males and 1 in 8,000 females. This X-linked genetic condition causes a range of developmental problems that encompass learning disabilities and cognitive impairment. Males are usually more severely affected than females. The gene causing FXS (FMR1) is divided into three categories: normal, pre-mutation and full mutation depending on the repeat region length. The full mutation causes FXS. There is no cure for the disorder - educational and behavioural interventions may improve the outcome.

Inheritance: The syndrome is inherited from a carrier mother who has one X chromosome with the FMR1 gene repeat region in the pre-mutation range. The disorder is caused by an increase of the repeat region length to full mutation of the FMR1 gene. As males only have one X chromosome, when they inherit the full mutation they develop the disorder. Females have two X chromosomes and thus tend to have milder symptoms when they inherit the full mutation.

With each pregnancy a carrier mother has a 50% chance of passing on the X chromosome with the pre-mutation repeat region. The chance it will expand to the full mutation increases with increasing size of the pre-mutation.

Fragile X and primary ovarian insufficiency (FXPOI) – pre-mutation carriers

Ovarian insufficiency occurs when the ovaries stop functioning normally in a woman younger than 40 years of age. Some women with POI can get pregnant, but once they have completed menopause they can no longer fall pregnant.

Studies of female pre-mutation carriers show that 25% experience ovarian insufficiency and 25% experience early menopause.

Fragile X and tremor/ataxia (FXTAS) – pre-mutation carriers

This condition is characterized by problems with movement and thinking ability. It is a late-onset disorder (>50years) and the signs and symptoms worsen with age.

Symptoms include an intention tremor ie trembling or shaking of a limbs when trying to perform a voluntary movement and there are problems with coordination and balance. The tremors develops first and loss of balance may follow, but not all affected individuals exhibit both signs. There can be cognitive problems with loss of short term memory, the ability to plan and problem solve.

Male carriers of the pre-mutation occur with a frequency of one in 450 but only 40% of these will develop the condition. One in 200 females have the pre-mutation but only 16% develop the condition.

Cascade screening:

If you are shown to be a pre-mutation carrier and you have siblings and other close relatives, they may also be at risk of being carriers. Testing for these individuals is also available should they wish to determine their carrier status.

Accuracy of testing:

The test determines the number of triplet repeats in the FRX gene and determines if an individual falls into the normal, intermediate, pre-mutation of full mutation range. A small percentage of cases may require further testing.

How much does it cost?

The test has a Medicare rebate for individuals with an intellectual disability or women with ovarian insufficiency (this must be indicated by your doctor on the blood collection form). There is no rebate if the test is to determine carrier status in a normal individual. Please see Virtus Diagnostics for costings.

This test can be ordered separately or bundled as part of the Virtus Genetic Screen (for Cystic fibrosis, Spinal Muscular Atrophy and Fragile X).