Diagnosis of Fragile X Syndrome

The diagnostic procedure may start if an infant is presenting signs of developmental delay, intellectual disability and/or signs of autism. The average age reported for diagnosis of fragile X syndrome in boys is 36 months of age and 42 months of age for girls.

A diagnosis of full mutation fragile X syndrome may be given to an individual if genetic testing shows that there is an excess of 200 CGG repeats in the FMR1 gene on the X chromosome alongside abnormal methylation. It is important to remember that the borderline number of repeats needed for a diagnosis are only estimates and a diagnosis will depend on a range of other factors. To read more about the genetics of fragile X syndrome, click here.

There are several different genetic testing methods used to test for FMR1 mutation but the most common and effective methods are targeted mutation analysis and methylation analysis.

In a few cases, diagnosis may occur through prenatal testing on women who have premutation FXS to screen for the mutated FMR1 gene in the foetus. Some of the prenatal tests include amniocentesis (test of the amniotic fluid) and chorionic villus sampling (testing of the cells from the placenta).

Further information:

For more information on the genetic tests used for fragile X diagnosis click here. Note: this is aimed at professionals

The Fragile X Society's website provides useful information for families on the different aspects of diagnosis. Click the relevant links below.

Testing

FXS carriers

Newly diagnosed