



# 10 things you should know about Fragile X syndrome.

1. It's genetic – caused by a defect in the X chromosome.
2. It is the most common inherited form of mental impairment. 1 in 4,000 males and 1 in 6,000 females are affected – from all ethnic and socio-economic backgrounds.
3. It's a spectrum disorder, with symptoms varying from mild learning disabilities (including shyness and social anxiety) to severe cognitive impairment (mental impairment) and autism.
4. The effect on boys tends to be more severe. Most boys have mental impairment and approximately 1/3 have autism. Girls generally exhibit a milder form of Fragile X syndrome because they have two X chromosomes – one that works properly and one that doesn't.
5. Fragile X syndrome does not shorten lifespans. People affected by it do not suffer from major medical problems because of the syndrome and thus are generally healthy – but often require special care for life.
6. If a man is a carrier (1 in 800 men are carriers), he will pass it only to his daughter(s), and they will always be carriers.
7. If a woman is a carrier (1 in 260 women are carriers), she has a 50/50 chance of passing the defective gene on to her son(s) or daughter(s); the child will either be a carrier or be affected by Fragile X syndrome.
8. Some male carriers (and rarely women) will be impaired by Fragile X-associated Tremor Ataxia Syndrome (FXTAS) later in life. This is a neurological disorder that can involve tremors, balance irregularities, difficulty walking and dementia – and may be misdiagnosed as Parkinson's disease and/or Alzheimer's disease.
9. Some female carriers (approximately 20-28%) experience Fragile X-associated primary ovarian insufficiency (FXPOI) – menopause occurring prior to the age of 40.
10. Fragile X syndrome can be detected through a simple blood test. Testing should be considered for any individual with otherwise unexplained developmental delay, mental impairment or autism. Testing is also recommended if there is any family history of developmental problems.