

SPOT FRAGILE X SYNDROME



Look at the face

Long face, prominent jawline, protruding ears, strabismus, recurrent ear infections.

Muscle tone

Low muscle tone leads to hyperflexible joints. Heart murmur. Gastro reflux issues.



Autism/autistic traits

Fragile X is one of the leading causes of inherited Intellectual Disability and Autism. Children with Autism should be tested for Fragile X Syndrome

Mother's reproductive health

Women carriers can have ovarian insufficiency which often leads to fertility problems. Another important factor is that being an inherited condition they need to be careful for the next child.



Cognitive

Intellectual disability/autism.
Repetitive/echolalic speech.
ADHD, hyperactivity and impulsivity.

Fragile X is caused by alterations on the FMR1 gene and as it is passed through generations, it may cause a woman to be a carrier for Fragile X. This can result in her having fertility problems or having children affected by fragile X syndrome. Fragile X can be diagnosed by a blood test called the "Fragile X" or "FMR1" DNA test.