Syndrome Specific Medical health check guide - Fragile X Syndrome (FXS)

Introduction
Fragile X Syndrome (FXS) is caused by an expansion of a segment of DNA (trinucleotide sequence CGG triplet repeat) in the FMR1 gene. Normally, this DNA segment is repeated 5 to 40 times. In people with Fragile X syndrome, however, this segment is repeated over 200 times. This inactivates the FMR1 gene, which prevents it from producing the Fragile X Mental Retardation protein, which in turn leads to the signs and symptoms of Fragile X syndrome.

History
As with all people with LD focus on an assessment of:

- eyesight and hearing
- feeding, bowel and bladder function
- behavioural problems.

As the most common cause of inherited learning difficulty, they have a normal life expectancy and generally less severe medical complications. It affects males more than females and has a characteristic physical appearance:

- long face
- large jaw
- prominent ears
- enlarged testicles (post puberty).

About one in four people with Fragile X have epilepsy, which can be generalised or focal (tonic-clonic seizures, complex partial seizures or absences). Seizures usually begin in childhood or adolescence and are not frequent, often being outgrown before adulthood.

Ask about anxiety (often highly anxious and overwhelmed), hyperactivity, autistic type features (such as hand flapping, biting, poor eye contact and shyness), ataxia, seizures and any joint dislocations (particularly patella and shoulder). About 30% of boys with FXS meet the criteria for autism.

In women ask about hot flushes as premature ovarian failure can occur before 30 years.
Examination

1. Sensory
Eye problems can include squint (strabismus), long-sightedness and visual perceptual problems. Eyelids tend to puffiness.

☐ Full assessment by optician/optometrist every 2 years.

Children with fragile X are prone to recurrent Otitis media.

☐ Assessment including Otoscopy, whisper test and refer if concerns.

2. Abdominal
Hernias are more common due connective tissue disorder. Men may develop enlarged testicles (macro-orchidism) after puberty, but this does not seem to pose any medical problems.

☐ Examine the abdomen and inguinal areas (when seen as new patient and then periodically).

3. Cardiovascular
Examine for adult onset mitral valve prolapse and aortic regurgitation.

☐ Auscultation of heart annually.

4. Musculoskeletal
Musculoskeletal Problems with connective tissue can lead to flat feet and low muscle tone. The joints are often extremely flexible and may dislocate.

☐ Assessment spine for scoliosis