

Fragile X Signs & Symptoms

Fragile X Syndrome Symptoms

In Males

Behavioral Characteristics

Behavioral characteristics can include:

- ADHD
- Autism and autistic behaviors
- Social anxiety
- Hand-biting or flapping
- Poor eye contact
- Sensory disorders
- Increased risk for aggression

Physical Features

No one individual will have all the physical features of FXS, and some are more

common after puberty. Physical features may include:

- Large ears
- Long face
- Soft skin
- In post-pubertal males, a long face and large testicles (called “macroorchidism”)

Connective tissue problems may include:

- Ear infections
- Flat feet
- High arched palate
- Double-jointed fingers
- Hyper-flexible joints

Intellectual Disabilities

Intellectual disabilities in FXS include a range from moderate to more severe. The majority of males, as opposed to females, demonstrate significant intellectual disability.

Disposition

Individuals with FXS are very social and friendly, have excellent imitation skills, and have a strong visual and long-term memory. In general, they like to help others and are nice, thoughtful people with a wonderful sense of humor.

In Females

A small percentage of females who have FXS will have no apparent behavioral, physical, or intellectual signs of the condition. These females are often identified only after another family member has been diagnosed.

Behavioral Characteristics and Physical Features

Behavioral characteristics seen in males can also be seen in females, though females often have milder intellectual disability and a milder presentation of the syndrome’s behavioral and physical features.

Intellectual Disabilities

About one-third of females with FXS have a significant intellectual disability. Others may have moderate or mild learning disabilities, emotional and mental health issues, and general and social anxiety.

Also see our clinic finder for facilities specializing in FXS.

Learn more about Fragile X syndrome →

Back to top

FXPOI Symptoms

Fragile X-Associated Primary Ovarian Insufficiency

Common symptoms of FXPOI include absent or irregular cycles, “sub-fertility” or infertility, hot flashes, and premature ovarian failure (often referred to as POF), which is the complete cessation of menstrual periods before the age of 40. In the wide range of symptoms, premature ovarian failure is at the more severe end of the spectrum.

FX premutation carriers can have normal ovarian function, but can still go through early menopause, which is menopause occurring at age 40–45. Menopause normally occurs at age 45–55.

Even though women with FXPOI may develop symptoms similar to those of menopause, such as hot flashes and vaginal dryness, FXPOI differs from menopause in some important ways.

After menopause:

- Typical women cannot get pregnant because their ovaries no longer release eggs, but women with FXPOI can get pregnant *in some cases* because their ovaries can occasionally function to release viable eggs.
- Typical women will never have another menstrual period, but women with FXPOI may experience a return of menstrual periods.

Learn more about FXPOI →

Back to top

FXTAS Symptoms

Fragile X–Associated Tremor/Ataxia Syndrome

Both the type and severity of FXTAS symptoms vary among individuals. Some will have multiple symptoms that progress rapidly, others will have few symptoms that remain mild over many years.

The symptoms of FXTAS are divided into *minor* and *major* clinical and MRI findings. The diagnosis is then categorized into *definite*, *probable*, or *possible* FXTAS.

This criteria was developed primarily as a reflection of the presenting symptoms in males. As we learn more about FXTAS in females, the diagnostic criteria may become different for females, since they usually have milder symptoms.

There are also comorbid symptoms of FXTAS, which means they often occur in individuals with FXTAS but aren't used to confirm the diagnosis (just as a sore throat is a symptom of strep throat, but other symptoms are needed to make the diagnosis).

With FXTAS being a relatively new disorder, there may not appear to be as many services and professionals available as there are for other medical conditions. However, a number of support services and informational materials may be of value to those impacted by FXTAS, some of which address symptoms common to other medical conditions, such as Parkinson's. One important resource is our clinic finder, which lists both FXS and FXTAS treatment facilities.

Major FXTAS Symptoms

- **Intention Tremor —** Intention tremor is a tremor of the hand when using utensils or writing instruments, or reaching for or pouring something. The tremor is not as apparent at rest.
- **Gait Ataxias —** Gait ataxias include balance problems that may include falling, needing support when walking or going up and down stairs, trouble stepping on and off curbs, generalized instability, or a wide-based gait.
- **White Matter Lesions —** White matter lesions involving the middle

cerebellar peduncles, signs found via an MRI, are strongly associated with (but not unique to) FXTAS.
- **FXTAS Inclusions —** FXTAS inclusions are neuropathology findings within brain cells.

Minor FXTAS Symptoms

- **Parkinsonism —** Parkinsonism is also known as resting tremors.
- **Short-Term Memory Problems —** Short-term memory problems can be difficult to determine since it is natural for short-term memory to deteriorate as we age. However, in FXTAS, it can change more rapidly than normal or may be more dramatic, such as forgetting what one ate, said, or did shortly after the event.
- **Executive Function —** Executive function and decision-making problems include initiating and completing activities, adapting and changing behavior as needed, and anticipating and planning for new tasks and situations. Executive function allows us to anticipate outcomes, solve problems, and generalize from one situation to the next.
- **MRI Findings —** MRI findings that are more general than those listed above, such as cerebral white matter lesions and moderate to severe generalized brain atrophy.

Other FXTAS Symptoms

These are not considered to be official diagnostic criteria.

- **Neuropathy** or numbness and tingling of the extremities.
- **Mood instability**, including irritability, explosive outbursts, and personality changes.
- **Cognitive decline**, including loss of skills such as math and reading.
- **Autonomic functioning problems** such as impotence and loss of bladder or bowel functions.
- **High blood pressure, thyroid disorders, and fibromyalgia**, all of which are more common in females — and very common in the general population.

Learn more about FXTAS →

Back to top