

What is FXTAS? Your Questions Answered

What is FXTAS?

Fragile X–associated tremor/ataxia syndrome (FXTAS) is an adult-onset neurodegenerative disorder, more common in males than females over 50 years of age with the Fragile X premutation. FXTAS is associated with tremors, balance problems, and other neurological signs. FXTAS progresses at varying rates in different individuals. The risk of developing FXTAS in any given individual is influenced by their CGG repeat size, sex, and age.

Fragile X-associated tremor/ataxia syndrome is more commonly known as FXTAS and is pronounced *faks-tas*.

All individuals with FXTAS have the *FMR1* premutation (CGG repeats 55–200). The job of the *FMR1* Fragile X gene is to make protein (FMRP) that is important in brain development. Researchers believe that (for unknown reasons) having the premutation leads to the overproduction of *FMR1* mRNA (which contains the expanded CGG repeats). They believe that the high levels of mRNA are what cause the signs and symptoms of FXTAS, but more research is needed.

Note: Not all individuals with the *FMR1* premutation will develop FXTAS, but all individuals with FXTAS have an *FMR1* premutation. Researchers are investigating what other factors might contribute to FXTAS in individuals with the *FMR1* premutation.

FXTAS Prevalence

FXTAS may be one of the most common adult-onset, single-gene neurological diseases, similar in prevalence to other neurodegenerative diseases such as ALS (Lou Gehrig's disease); however, more studies within the general population are necessary before the true incidence is known.[3,4,5,6] See published studies:

- Intention Tremor, Parkinsonism, and Generalized Brain Atrophy in Male Carriers of Fragile X
- Fragile X Premutation Tremor/Ataxia Syndrome: Molecular, Clinical, and Neuroimaging Correlates
- Fragile X-Associated Tremor/Ataxia Syndrome: Clinical Features, Genetics, and Testing Guidelines
- CGG Repeat Length Correlates with Age of Onset of Motor Signs of the Fragile X-Associated Tremor/Ataxia Syndrome (FXTAS)

Among individuals with the *FMR1* premutation, about 40% of males older than 50 years and 8%-16% of women older than 40 years develop FXTAS.[1]

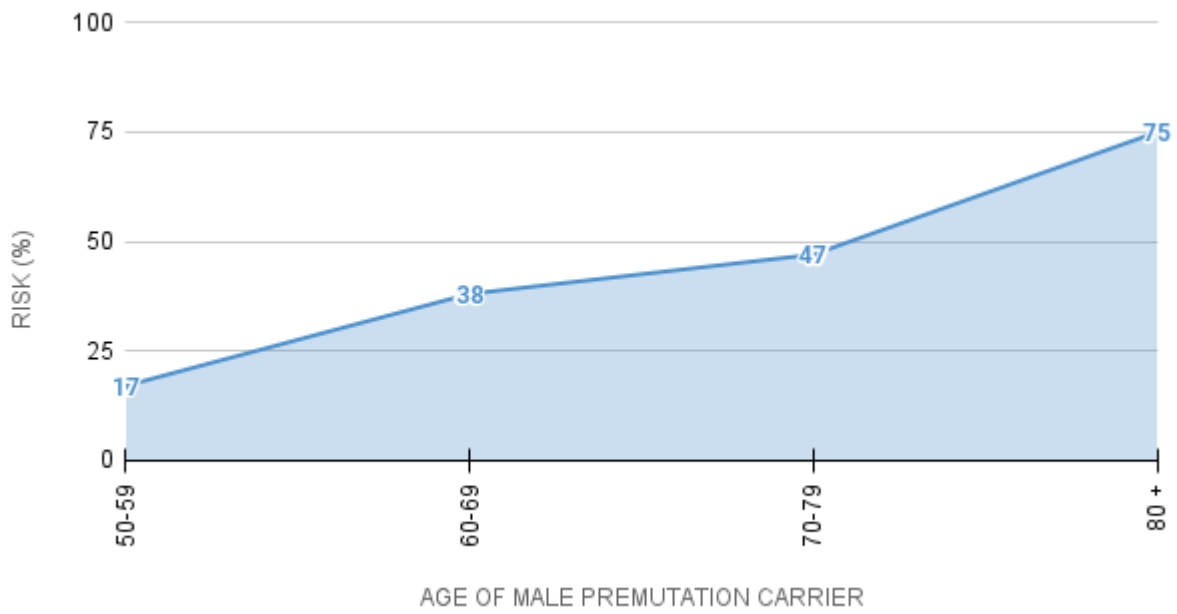
However, the risk of FXTAS in any given individual is influenced by their:

- CGG repeat size — A larger number of repeats increases the risk.
- **Sex** — Males are at greater risk
- **Age** — Symptoms are more common at older ages

Considering all of these factors and literature to date, it is estimated that the lifetime prevalence of FXTAS in the general population is approximately 1 in 8,000.[2] This indicates that FXTAS is significantly less common than essential tremor or Parkinson's disease in older adults.

For men who have the *FMR1* premutation, the chance of developing core symptoms of FXTAS (tremor, problems with walking or balance) increases with age, as shown below.

% Chance of Male Premutation Carriers Developing Core Symptoms of FXTAS



For males, the chance of developing symptoms of FXTAS increases with age.

For males who are premutation carriers, the chance of developing core symptoms of FXTAS (tremor, problems with walking/balance) increases with age:

- Ages 50-59 the chance is about 17%.
- Ages 60-69 about 38%.
- Ages 70-79 about 47%.
- Over 80 years old, about 75% will develop symptoms of FXTAS.

Studies of females with the *FMR1* premutation (within families already known to have someone with a Fragile X condition) have found that about 8%-16% develop some FXTAS symptoms. The symptoms in females tend to be milder.

Back to top

FXTAS Signs & Symptoms

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

There are also comorbid symptoms of FXTAS, which means they there often occur but aren't used to confirm the diagnosis. For example, a sore throat is a symptom of strep throat, but cultures, fever, and other observations and tests are also used to make the diagnosis.

Major Symptoms

- **Intention Tremor**: A tremor of the hand when using utensils, writing instruments, reaching for or pouring something. The tremor is not as apparent at rest.
- **Gait Ataxias**: Balance problems may include falling, needed support when walking or going up or down stairs, trouble stepping on or off curbs, generalized instability, or display of a wide-based gait.
- **MRI Findings**: Signs strongly associated with (but not unique to) FXTAS include white matter lesions involving middle cerebellar peduncles (MCP).
- **Neuropathology Findings**: "FXTAS inclusions" may be found within brain cells.

Minor FXTAS Symptoms

- **Parkinsonism**: Also known as resting tremors.
- **Short-Term Memory Problems**: This 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 or Decision-Making Problems**: Executive function includes the ability to initiate and complete an activity, to adapt and change behavior as needed, and to anticipate and plan for new tasks and situations. Executive function allows us to anticipate outcomes, solve problems, and generalize from one situation to the next.

Watch the webinar: FMR1 Carriers Report Executive Function Changes Prior to FXTAS: A Longitudinal Study, with Dr. David Hessl.

- **MRI Findings**: In addition to those listed above, are findings referred to as “lesions of cerebral white matter,” and other findings indicating “moderate to severe generalized brain atrophy.”

Other FXTAS Symptoms

These are not considered to be official diagnostic criteria.

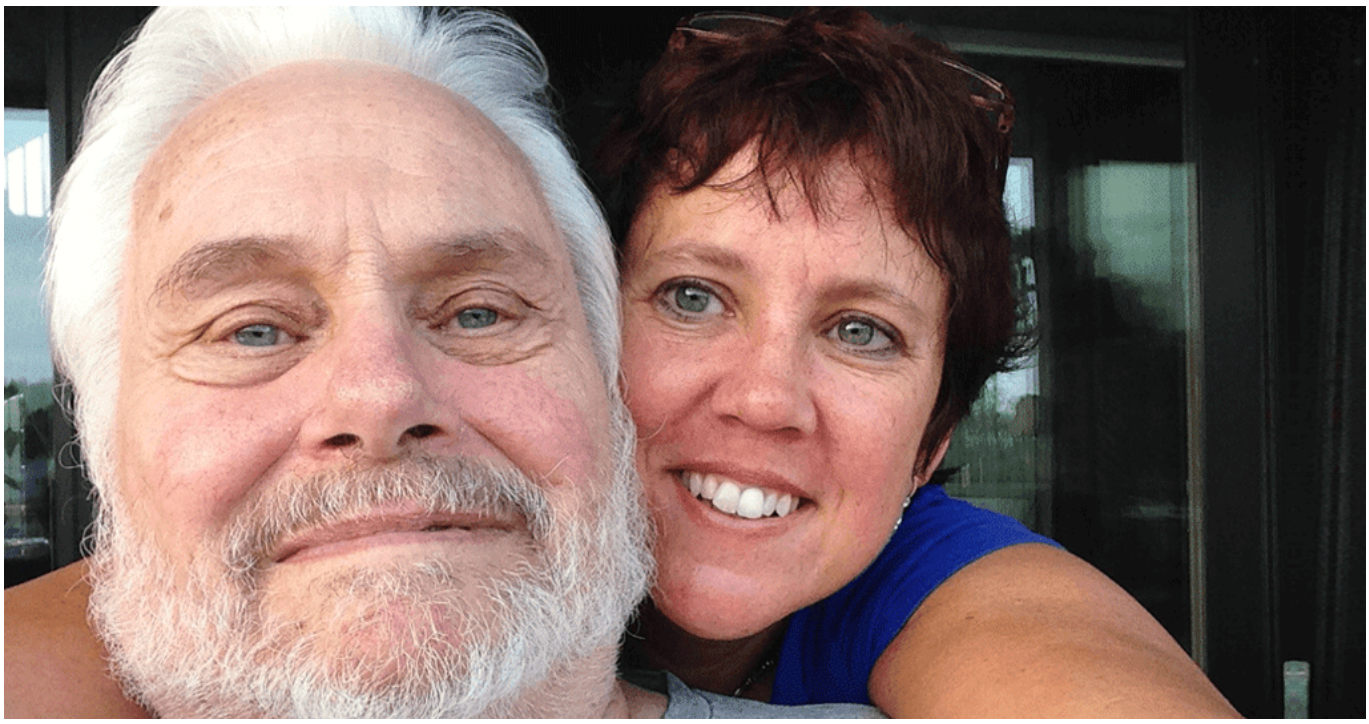
- **Neuropathy**: Numbness or tingling of the extremities.
- **Mood Issues**: Including instability, irritability, explosive outbursts, personality changes.
- **Cognitive Decline**: Including loss of skills such as math and reading.
- **Autonomic Functioning**: Including problems such as impotence and loss of bladder or bowel functions.
- **Co-occuring Conditions**: Including high blood pressure, thyroid disorders, and fibromyalgia (more common in females and very common in the general population).
- **Seizures**

TIP Speak with your healthcare provider about any concerns with any anesthesia. You could also consider sharing this article: General Anesthetic Use in Fragile X Spectrum Disorders from the NIH.

Back to top

Testing for FXTAS

FXTAS only occurs in individuals with a Fragile X (*FMR1*) premutation. Therefore, anyone considered for this diagnosis must be tested for and confirmed as having the Fragile X premutation. This involves DNA testing of the individual's *FMR1* gene. Please also see Diagnosis & Recognition of FXTAS.



Related Treatment Recommendation

Genetic Counseling and Family Support

Genetic counseling includes family and medical histories to assess the chance of carriers (premutations) throughout a family's lineage.

Go to the treatment recommendation
Back to top

FXTAS Treatment & Intervention

Each person with FXTAS is unique, with their own medical history, biological makeup, and personality, so each will have their own set of problems. While most will have handshaking and balance problems, these will vary in severity and the need for treatment.

While most of the FXTAS symptoms are neurological, many other body systems are also affected. To receive optimal care, it is best to have a medical care team that is knowledgeable about FXTAS. The team should include a neurologist and primary care physician at a minimum. Other specialists in the areas of psychiatry, psychology, rehabilitation, urology, cardiology, and movement disorders neurology, may also be needed.

Typically, treatments and interventions aim to reduce symptoms, slow disease progression, and even provide a cure. While there is no known cure or treatment to slow the progression of FXTAS, several treatments have been found helpful in reducing symptoms. Research is currently underway to identify potential treatments specific to FXTAS. In the meantime, clinicians may treat the symptoms of FXTAS with treatments already available for those symptoms.

Useful treatments for FXTAS include:

- Medications.
- Psychological and genetic counseling.
- Rehabilitative treatments such as speech, occupational, and physical therapy.
- Gait training.
- Surgery.
- Family supportive services and counseling are also important.

Related Treatment Recommendation

Fragile X-Associated Tremor/Ataxia Syndrome Treatment Recommendations

Therapy for FXTAS aims to reduce symptoms and eventually slow the progression of the disease.

Go to the treatment recommendation
Back to top

FXTAS Clinics

While there is currently no cure for FXTAS, specialists may be able to reduce symptom severity or slow symptom progression with various treatment options.

FXTAS clinics are members of the NFXF's International FXTAS Consortium (IFC) and specialize in FXTAS. They are comprised of doctors (primarily neurologists and movement disorder specialists), therapists, researchers, and other specialists who treat FXTAS using the best available scientific knowledge of its known causes.

Also listed in our clinic finder are National Ataxia Foundation clinics specializing in ataxia and ataxia-related disorders. While they do not specialize in FXTAS, they have indicated experience treating FXTAS patients, and we have provided them with FXTAS and general Fragile X-related materials.

Find a FXTAS Clinic

FXTAS clinics are led by doctors and other specialists who use the best available scientific knowledge about FXTAS. While there is currently no cure, specialists may be able to reduce symptom severity and slow symptom progression.

Locate a FXTAS Clinic
Back to top

More FXTAS Resources

FXTAS Webinars

View all FXTAS webinars



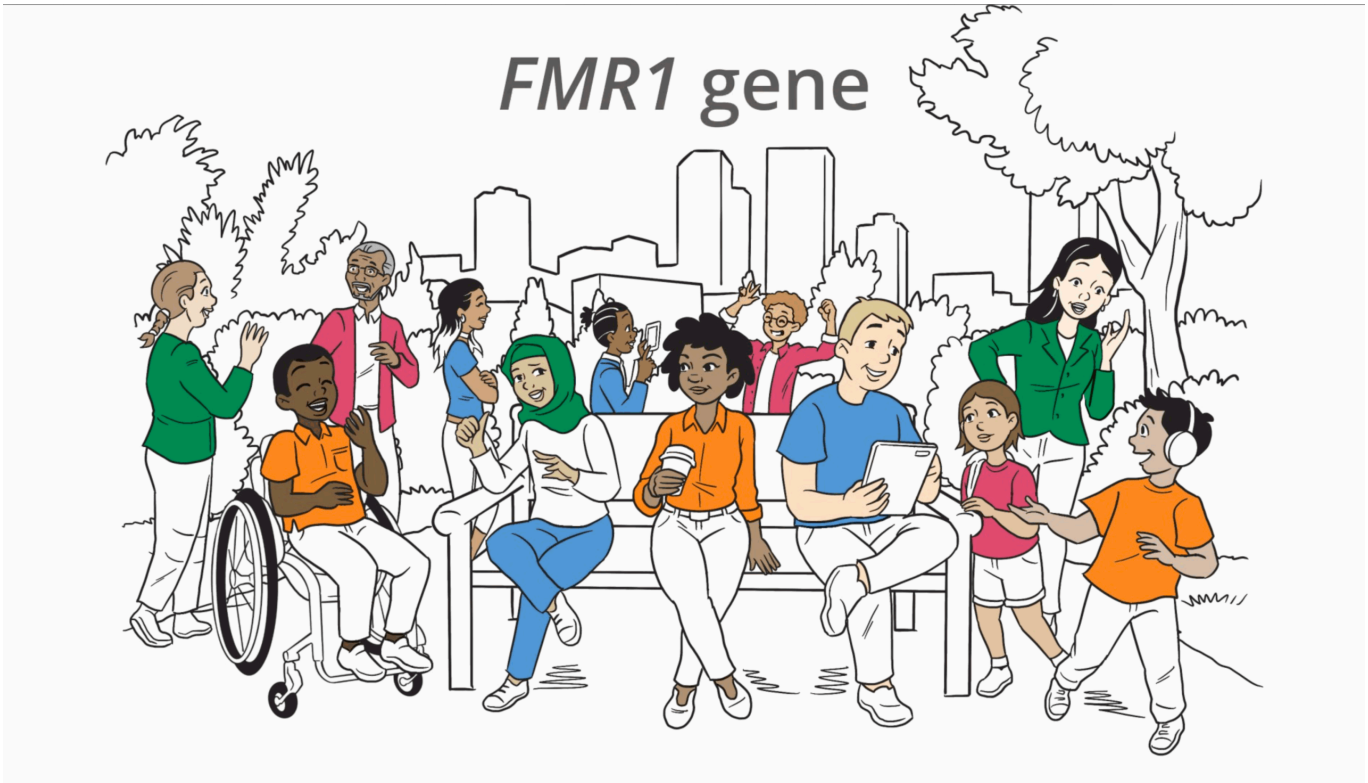
- FXTAS Issues
- Research

Advancing Clinical Trials for FXTAS

February 9, 2026

00 h 19 m

Watch Now



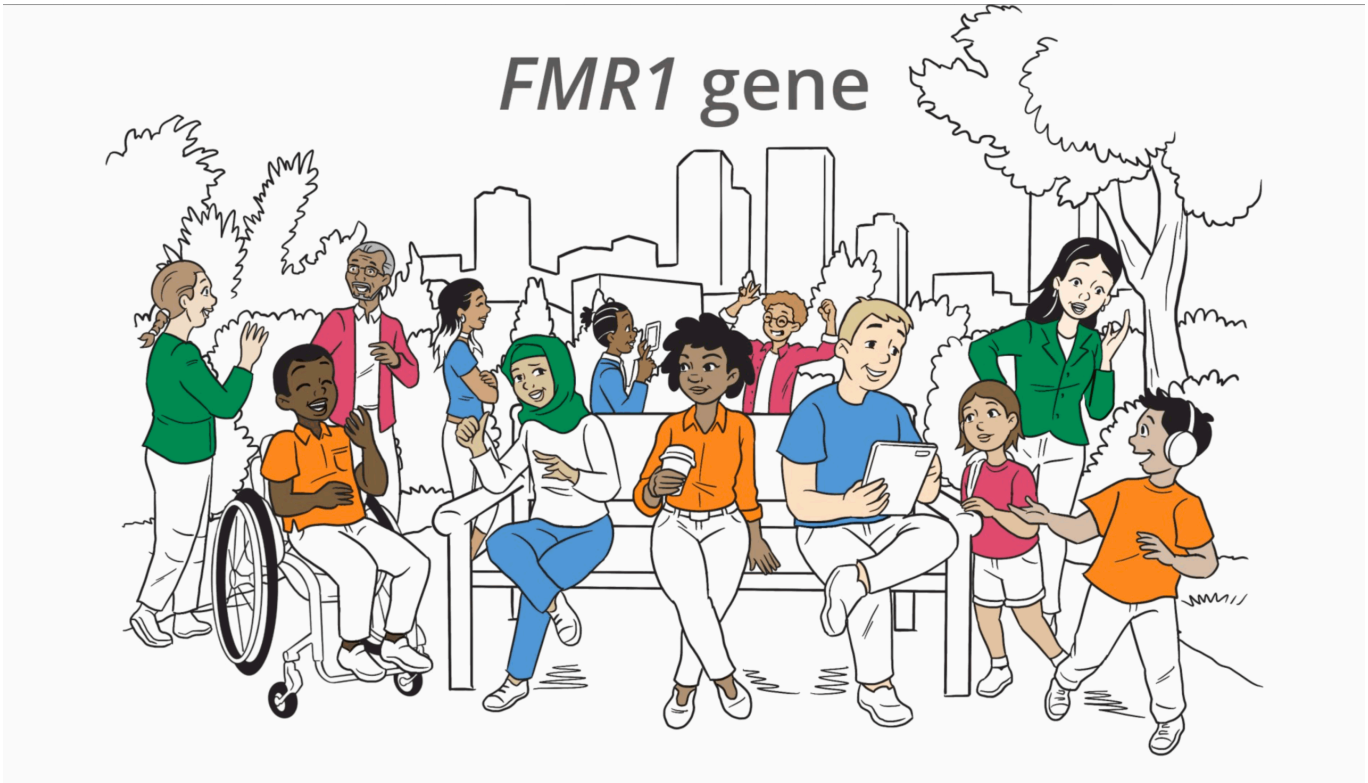
- Community
- FXPOI

¿Qué es la premutación del cromosoma X frágil?

January 8, 2026

00 h 01 m

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What is the Fragile X Premutation? A 90-Second Video

July 29, 2025

00 h 02 m

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Back to top

FXTAS Research Results

view all

View all FXTAS research results



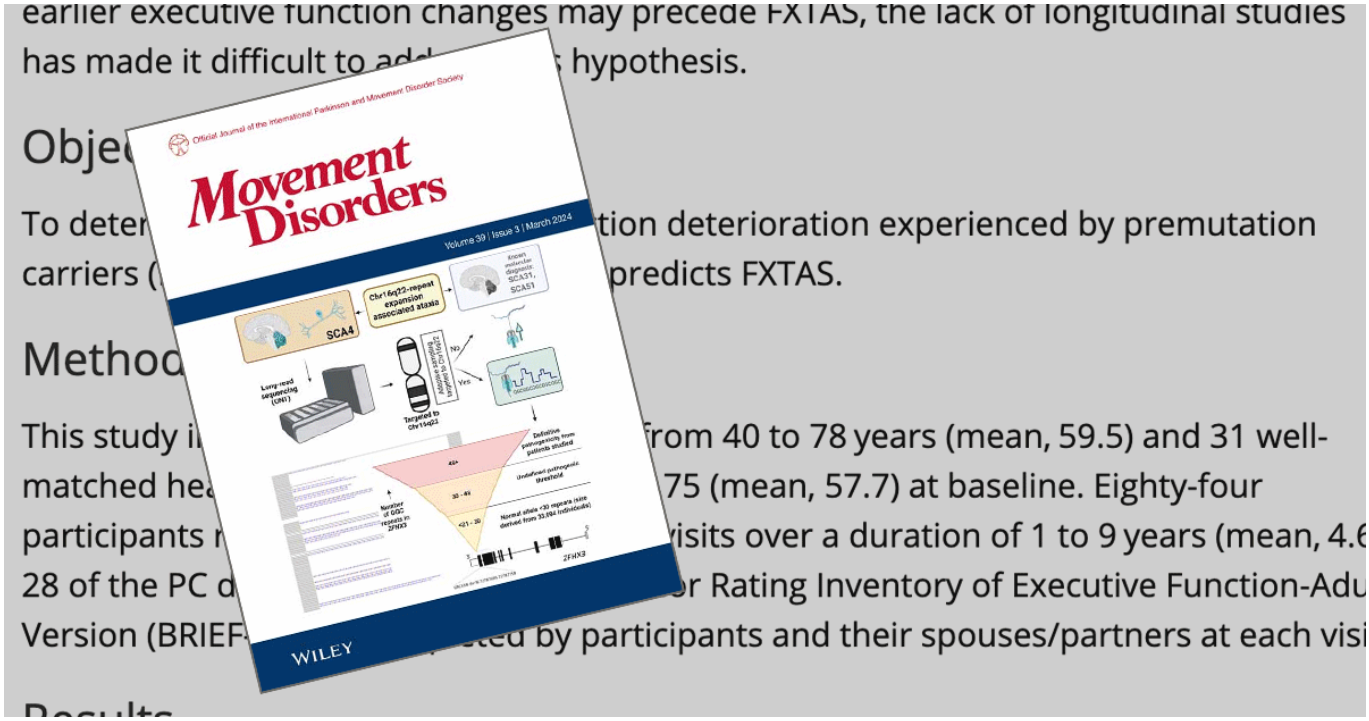
- FXTAS Issues
- NFXF & Programs

Symposium Summary: Prenatal and Newborn Screening in Fragile X Syndrome
September 16, 2026
05 min
Read Now



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- NFXF & Programs

FXTAS Neuromotor Signs in Women with the FMR1 Premutation: Elevated Risk at Mid-Range CGGs and Attenuation by College Degree Attainment
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- [FXTAS Issues]
- [Physical & Mental Health]

[FMR1 Carriers Report Executive Function Changes Prior to Fragile X-Associated Tremor/Ataxia Syndrome: A Longitudinal Study]
April 25, 2024
06 min
[Read Now]
[Back to top]

Additional FXTAS Resources

Support

[NFXF FXTAS Facebook Support Group]
For everyone who would like to connect with others dealing with FXTAS.

Organizations

International Parkinson and Movement Disorder Society

Helpful Tools

Find a Movement Disorders Specialist (*International Parkinson and Movement Disorder Society*)

Journal Articles

Treatment of Fragile X-Associated Tremor Ataxia Syndrome (FXTAS) and Related Neurological Problems (*Clinical Interventions in Aging*)

Fragile X-Associated Tremor/Ataxia Syndrome: Clinical Features, Genetics, and Testing Guidelines (*Movement Disorders*)

Back to top