# 31 Shareable Fragile X Facts

## 1. July is Fragile X Awareness Month!

In July 2000 the 106th Congress designated July 22 as National Fragile X Awareness Day. We took that a step further and consider the month of July as Fragile X Awareness Month.

Learn more about Fragile X Awareness Month

## 2. Fragile X is a group of conditions associated with alterations in the FMR1 gene on the X chromosome.

These changes result in a “premutation,” which can lead to Fragile X-associated conditions and disorders like FXTAS (Fragile X-associated tremor/ataxia syndrome), FSPOI (Fragile X-associated primary ovarian insufficiency), or a “full-mutation,”

which causes Fragile X syndrome.

Learn more about the FMR1 gene

## 3. Fragile X syndrome is an inherited condition affecting intellectual, behavioral, and social development.

Though Fragile X syndrome occurs in both genders, males are more frequently affected than females, and generally with greater severity. Some females may not experience any of the behavioral, cognitive, or physical features that appear more widely in males.

Learn more about Fragile X syndrome

## 4. Approximately 1 in 7,000 males and 1 in 11,000 females have Fragile X syndrome.

FXS has been detected in all populations and ethnic groups. As a result, efforts have been made to determine the overall prevalence of FXS and the difference in prevalence between males and females. Studies have been undertaken both in the "special needs" population and the general population.

Learn more about the prevalance of Fragile X syndrome

## 5. Fragile X syndrome is considered a rare disease.

Rare diseases are defined as less than 200,000 individuals in the United States. It is estimated that about 100,000 Americans have Fragile X syndrome. The Fragile X premutation, however, is not rare. It is estimated that up to 1 in 151 females and 1

in 468 males have the Fragile X premutation.

Learn more about prevalance

## 6. FXPOI (Fragile X-associated primary ovarian insufficiency) is a condition in which the ovaries are not functioning at full capacity in an individual with the FMR1 premutation.

About 20% of women who carry a Fragile X premutation over their reproductive life span develop POI, compared with only 1% in the general population.

Learn more about FXPOI

## 7. Fragile X–associated tremor/ataxia syndrome, or FXTAS, is an “adult-onset” neurodegenerative condition associated with the Fragile X premutation.

Fragile X–associated tremor/ataxia syndrome or 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.

Learn more about FXTAS

## 8. Fragile X is genetic, meaning it is caused by a

## change in the gene.

Fragile X is also hereditary, meaning this gene change can be passed from one generation to the next. Fragile X is unique among rare diseases because it is both genetic and hereditary.

Learn more about genetics and inheritance

## 9. Fragile X is an “X-linked” condition, which means the FMR1 gene is on the X chromosome.

Males have one X and one Y chromosome and females have two X chromosomes. Both males and females can have Fragile X syndrome or the Fragile X premutation.

Learn more about the *FMR1* gene

## 10. Fragile X needs only one parent to pass the gene along.

This is different from many other conditions where both parents need to have the “carrier” gene.

Learn more about inheritance

## 11. Traditionally, a “carrier” of a genetic mutation is defined as a person who inherits an altered form of a gene but shows no effects of that mutation.

Not the case with Fragile X, as “carriers” of the Fragile X premutation can be impacted even by the partial mutation. Individuals with the Fragile X premutation are at risk of developing Fragile X-associated conditions and disorders including Fragile X-associated tremor/ataxia syndrome (FXTAS) and Fragile X-associated primary ovarian insufficiency (FXPOI).

Learn more about the Fragile X premutation

## 12. Fragile X-associated primary ovarian insufficiency, or FXPOI, is a condition associated with females with the FRM1 premutation.

The ovaries in women with FXPOI do not function to full capacity, and women with FXPOI may struggle to get pregnant and experience irregular menstrual cycles and early menopause.

Learn more about FXPOI

## 13. Some women with the Fragile X premutation are initially identified because they have fertility problems and are considering fertility treatment.

There are various assisted reproductive options that individuals with the Fragile X premutation may consider, including IVF with their own eggs, IVF with eggs donated by a non-carrier, attempting to get pregnant naturally, or adoption. Some may consider prenatal genetic testing through amniocentesis or chorionic villus sampling (CVS) to evaluate the genetic status of your pregnancy.

Learn more about reproductive options

## 14. About 20% of women with the Fragile X premutation develop primary ovarian insufficiency over their reproductive life span, compared with only 1% in the general population.

Evidence shows women with a premutation, on average, experience natural menopause at an earlier age compared to those without a premutation. The mean age of natural menopause being reduced by about five years from the typical age of about 51 years.

Learn more about the prevalence of FXPOI

## 15. Fragile X–associated tremor/ataxia syndrome, or FXTAS, is associated with the FMR1 premutation.

FXTAS is an “adult-onset” neurodegenerative condition, usually affecting males over 50 years of age. Females comprise only a small part of the FXTAS population, and their symptoms tend to be less severe. FXTAS progresses at varying rates in different individuals.

Learn more about FXTAS

## 16. Among individuals with the Fragile X premutation, about 40% of males older than 50 years and 8%–16% of women older than 40 years will develop FXTAS.

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50 years of age. Females comprise only a small part of the FXTAS population, and their symptoms tend to be less severe. FXTAS progresses at varying rates in different individuals.

Learn more about the prevalence of FXTAS

## 17. Fragile X syndrome, or FXS, has been documented worldwide, in all populations and ethnic groups.

While researchers do not have an exact number for how many male and female Americans could have Fragile X syndrome, the ratios noted here suggest that the raw number could be between 38,000 and 87,000. Worldwide, the number could be between 777,000 and 1,400,000.

Learn more about the prevalence of Fragile X Sydrome

## 18. Fragile X is a fairly “new” condition.

Fragile X was first termed Martin-Bell syndrome in 1943. In the 1990s, genetic testing technologies improved, and the specific gene associated with Fragile X syndrome — *FMR1* — was discovered.

Learn more about genetic testing

## 19. Fragile X is diagnosed by a simple yet highly accurate DNA test.

Individuals find out their CGG repeat number, which determines their Fragile X status.

Learn more about CGG repeats

## 20. The FMR1 gene makes a very important protein called FMRP that is found in all of our cells and performs very specific tasks.

FMRP (Fragile X messenger ribonucleoprotein) is especially important for brain development. Expansions in CGG repeats can impact our body's ability to make this important protein.

Learn more about proteins and *FMR1*

## 21. Everyone has the FMR1 gene.

The *FMR1* (Fragile X messenger ribonucleoprotein 1) gene lives on each X chromosome. Everyone has CGG repeats on the *FMR1* gene. Most people have CGG repeats below 45, which means they do not have Fragile X.

Learn more about the FMR1 gene

## 22. Individuals with CGG repeats on the FMR1 gene over 200 receive a Fragile X syndrome diagnosis.

Fragile X syndrome is not known to be more severe with a higher repeat number. For example, we would not expect someone with a CGG repeat of 700 to be more affected than someone with a repeat number of 205.

Learn more about testing and diagnosis

## 23. Fragile X and autism are not the same, though there are similarities.

Autism is generally characterized by an impairment in social interaction and communication, and the presence of restricted and repetitive patterns of behavior, interests, or activities. Some individuals with Fragile X syndrome also have an autism diagnosis.

Learn more about the similarities and differences between autism and Fragile X syndrome

## 24. Fragile X syndrome, or FXS, is the most common single gene linked to autism, accounting for about 1%-6% of all cases of ASD.

About 10% of children with autism are identified as having another genetic and chromosomal disorder, such as Fragile X syndrome. Given the possibility of a link, it is recommended that all children with autism, both male and female, be referred for genetic evaluation and testing for Fragile X syndrome and any other genetic cause of autism.

Learn more about the prevalence of FXS with autism

## 25. Fragile X syndrome is diagnosed by a DNA blood test, unlike autism, which is a behaviorally defined diagnosis.

There are three general circumstances in which Fragile X testing should be considered, one of them is a family history of FXS, FXTAS, intellectual or learning disabilities, autism of unknown cause, or infertility.

Learn more about who should have Fragile X testing

## 26. Individuals with Fragile X syndrome are “gestalt” learners who need to see and understand the “whole” and not the parts that add to a whole.

Methodologies using spatial-visual memory are thought to be more effective than sequential-successive approaches.

Learn more about FXS learning styles

## 27. Individuals with Fragile X syndrome are visual learners.

Visual schedules help ease their anxiety and can prevent over-stimulation.

Learn more about the Fragile X learning style

## 28. Carrier screening for many conditions, including Fragile X, is available.

Some families may choose to do carrier screening prior to starting a family. Other families may choose to do carrier screening when they find out a member of their family has been diagnosed with a genetic condition that prompts carrier screening. Some individuals may never choose to be screened.

Learn more about genetic counseling

## 29. Primary care physicians or OB-GYNs may offer carrier screening services.

There are also several options for at-home or lab-based carrier screening. Many carrier screenings include screening for many conditions. If you are interested in being screened for Fragile X, you should confirm Fragile X is included in the screening panel.

Learn more about genetic counseling

## 30. There is a lot of Fragile X research going on — including research for individuals with Fragile X syndrome, the Fragile X premutation, FXPOI, and FXTAS.

Some of these studies even need participants without Fragile X to serve as a group to compare to — or a "control" group. You can review the active research opportunities by visiting the NFXF's MyFXResearch portal.

Find a research opportunity

## 31. The FMR1 gene has officially been renamed!

*FMR1* now stands for "Fragile X messenger ribonucleoprotein 1," removing the reference to "mental retardation" that has long been outdated in the common vernacular. At the time of the gene's discovery, mental retardation was an accepted term for what we now call "intellectual disability." *We know that individuals with Fragile X are more than an intellectual disability!*

Learn more about the renaming of the FMR1 gene