FRAGILE X AWARENESS MONTH

NATIONAL **FRAGILE X** FOUNDATION

31 Shareable Facts about Fragile X and the Fragile X Premutation

#fragilex #fragilexsyndrome #fragilexawarenessmonth #thisisfragilex

1. July is Fragile X Awareness Month – Get Involved!

July is Fragile X Awareness Month! We understand the ups and downs that come with Fragile X; we're here to support you during challenging times and as well as celebrating the wins! Join us as we share more about Fragile X throughout the month of July. Each day we will present a new shareable fact for you to share on your social media, and with family and friends! Help raise awareness for #FragileX by sharing the facts along with your story.

Resource: https://fragilex.org/get-involved/national-fragilex-awareness-month/

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

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 Fragile X-associated tremor/ataxia syndrome, or Fragile X-associated primary ovarian insufficiency – or a "full-mutation" which causes Fragile X syndrome.

Resource: https://fragilex.org/understanding-fragile-x/fragile-x-101/

3. FXS is an inherited condition that occurs in both males and females.

Fragile X syndrome or FXS is an inherited condition affecting intellectual, behavioral, and social development. It occurs in both males and females who have a full mutation of the FMR1 gene.

Resource: https://fragilex.org/genetics/7-facts-did-not-learn/

4. Approximately 1 in 7,000 males and 1 in 11,000 females have FXS.

Approximately 1 in 7,000 males and 1 in 11,000 females have Fragile X syndrome (FXS).

Resource: https://fragilex.org/understanding-fragile-x/fragile-x-101/prevalence/

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 is not rare; it is estimated that up to 1 in 151 females and 1 in 468 males have the Fragile X premutation.

Resource: https://fragilex.org/understanding-fragile-x/fragile-x-101/

6. FXPOI is a condition in which the ovaries are not functioning at full capacity.

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

Resource: https://fragilex.org/understanding-fragile-x/fxpoi-primary-ovarian-insufficiency/

7. FXTAS is a neurodegenerative condition associated with the Fragile X premutation.

Fragile X–associated tremor/ataxia syndrome or FXTAS is an "adult-onset" neurodegenerative condition associated with the Fragile X premutation.

Resource: https://fragilex.org/understanding-fragile-x/tremor-ataxia-syndrome-fxtas/

8. Fragile X is both genetic and hereditary.

Fragile X is genetic, meaning it is caused by a change in the gene. Fragile X is also hereditary, meaning that 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.

Resource: https://fragilex.org/genetics/7-facts-did-not-learn/

9. Both males and females can the Fragile X premutation.

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.

Resource: https://fragilex.org/genetics/7-facts-did-not-learn/

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

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.

Resource: https://fragilex.org/genetics/7-facts-did-not-learn/

11. Individuals with the Fragile X premutation are at risk of developing Fragile X-associated conditions and disorders.

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).

Resource: https://fragilex.org/genetics/7-facts-did-not-learn/

12. Women with FXPOI may struggle to get pregnant or experience early menopause.

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, experience irregular menstrual cycles and early menopause.

Resource: https://fragilex.org/understanding-fragile-x/fxpoi-primary-ovarian-insufficiency/

13. There are various assisted reproductive options that individuals with the Fragile X premutation may want to consider.

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 your 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.

Resource: https://fragilex.org/understanding-fragile-x/fxpoi-primary-ovarian-insufficiency/

14. About 20% of women with the Fragile X premutation develop FXPOI.

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

Resource: https://fragilex.org/understanding-fragile-x/fxpoi-primary-ovarian-insufficiency/

15. FXTAS is associated with tremors, balance problems, and other neurological signs.

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. It is often mis-diagnosed as Parkinson's. 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, and is associated with tremors, balance problems, and other neurological signs.

Resource: https://fragilex.org/understanding-fragile-x/tremor-ataxia-syndrome-fxtas/

16. About 40% of males with the Fragile X premutation develop FXTAS.

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 Fragile X-associated tremor/ataxia syndrome (FXTAS).

Resource: https://fragilex.org/understanding-fragile-x/fragile-x-101/premutation-carriers/

17. FXS exists worldwide, in all populations and ethnic groups.

Fragile X syndrome (FXS) has been documented worldwide, in all populations and ethnic groups.

Resource: https://fragilex.org/understanding-fragile-x/fragile-x-101/prevalence/

18. Fragile X was first termed Martin-Bell syndrome in 1943.

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.

Resource: https://fragilex.org/understanding-fragile-x/fragile-x-101/

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

Fragile X is diagnosed by a simple yet highly accurate DNA test. Individuals find out their CGG repeat number(s), which determine their Fragile X status.

Resource: https://fragilex.org/understanding-fragile-x/fragile-x-101/testing-diagnosis/

20. The FMR1 gene makes an important protein called FMRP (Fragile X Protein).

The FMR1 gene makes a very important protein called FMRP (Fragile X Protein) that is found in all of our cells and performs very specific tasks. FMRP is especially important for brain development. Expansions in CGG repeats can impact our body's ability to make this important protein.

Resource: https://fragilex.org/understanding-fragile-x/fragile-x-101/genetics-inheritance/

21. Everyone has CCG repeats on the FMR1 Gene.

Everyone has the FMR1 gene. The FMR1 gene lives on each X chromosome. Everyone has CCG repeats on the FMR1 gene. Most people have CCG repeats below 45, which means they do not have Fragile X.

Resource: https://fragilex.org/understanding-fragile-x/fragile-x-101/genetics-inheritance/

22. FXS is diagnosed when CGG repeats on the FMR1 gene are over 200.

Individuals with CGG repeats on the FMR1 gene over 200 receive a Fragile X syndrome (FXS) 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 CCG repeat of 700 to be more affected than someone with a repeat number of 205.

Resource: https://fragilex.org/understanding-fragile-x/fragile-x-101/genetics-inheritance/

23. Some individuals with FXS also have an autism diagnosis.

Fragile X syndrome (FXS) 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.

Resource: https://fragilex.org/understanding-fragile-x/fragile-x-syndrome/autism/

24. FXS is the most common single gene linked to autism.

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

Resource: https://fragilex.org/understanding-fragile-x/fragile-x-syndrome/autism

25. Unlike autism, FXS is diagnosed by a DNA blood test.

Fragile X syndrome (FXS) is diagnosed by a DNA blood test, unlike autism, which is a behaviorally defined diagnosis.

Resource: https://fragilex.org/understanding-fragile-x/fragile-x-syndrome/autism

26. Individuals with FXS are "gestalt" learners.

Individuals with Fragile X syndrome (FXS) are "gestalt" learners who need to see and understand the "whole" and not the parts that add to a whole.

Resource: https://fragilex.org/family-resources/school-edu-fxs/

27. Individuals with FXS are visual learners.

Individuals with Fragile X syndrome (FXS) are visual learners. Visual schedules help ease their anxiety and can prevent over-stimulation.

Resource: https://fragilex.org/family-resources/school-edu-fxs/

28. Screening for genetic conditions, such as Fragile X is available.

Screening for genetic conditions, such as Fragile X is available. According to the American College of Obstetrics and Gynecology, "Carrier screening, whether targeted or expanded, allows individuals to consider their range of reproductive options. Ultimately, the goal of genetic screening is to provide individuals with meaningful information that they can use to guide pregnancy planning based on their personal values."

Resource: https://fragilex.org/understanding-fragile-x/fragile-x-101/testing-diagnosis/

29. Consult with your healthcare provider to discuss your options for testing and screening.

You can consult with your healthcare provider to discuss your options for testing and screening. Fragile X genetics, testing and diagnosis is complex. It is recommended to consult with your healthcare provider and a licensed genetic counselor to help identify the indications and most appropriate testing. Some genetic screening panels may include Fragile X, while others may not. It is important to discuss with your healthcare team if Fragile X testing has been included. Many labs now offer the ability to complete testing through saliva or buccal (cheek swab) in addition to blood samples.

30. Participating in Fragile X research is incredibly important!

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.

Resource: https://fragilex.org/our-research/myfxresearch-portal/

31. FMR1 stands for fragile X messenger ribonucleoprotein 1.

The FMR1 gene has officially been renamed! FMR1 now stands for fragile X messenger ribonucleoprotein 1, removing the reference to "mental retardation" which has long been outdated in 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!

Resource: https://fragilex.org/blog/research-results/the-use-of-retardation-in-fraxa-fmrp-fmr1-and-other-designations/