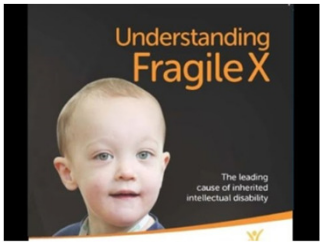
What is Fragile X?

Denise Devine, Western Mass Chapter

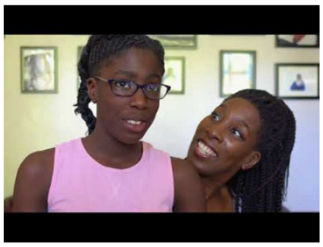


Features in Males with Fragile X

- Behavioral characteristics can include ADD, ADHD, autism and autistic behaviors, social anxiety, hand-biting and/or flapping, poor eye contact, sensory disorders and increased risk for aggression.
- The majority of males with Fragile X syndrome demonstrate significant intellectual disability. Disabilities in FXS include a range from moderate learning disabilities to more severe intellectual disabilities.

Features in Males with Fragile X

- Physical features may include large ears, long face, soft skin and large testicles (called "macroorchidism") in post-pubertal males. Connective tissue problems may include ear infections, flat feet, high arched palate, double-jointed fingers and hyper-flexible joints.
- No one individual will have all the features of FXS, and some features, such as a long face and macroorchidism, are more common after puberty.
- They are also very social and friendly, have excellent imitation skills, have a strong visual memory/long term memory, like to help others, are nice, thoughtful people and have a wonderful sense of humor.



Features in Females with Fragile X

- The 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.
- About one-third of females with FXS have a significant intellectual disability.
- Others may have moderate or mild learning disabilities, emotional/mental health issues, general anxiety and/or social anxiety.
- A small percentage of females who have the full mutation of the FMR1 Gene that causes FXS will have no apparent signs of the condition—intellectual, behavioral or physical.
- These females are often identified only after another family member has been diagnosed.

What is FXPOI

- FXPOI, or Fragile X-associated primary ovarian insufficiency, is one of three known Fragile X-associated Disorders (FXD). All the FXD are caused by changes in the FMR1 Gene.
- FXPOI is a condition in which the ovaries are not functioning at full capacity in an FMR1 premutation carrier. Common symptoms of FXPOI include absent or irregular periods, symptoms of menopause such as hot flashes, etc, early menopause and infertility.
- FXPOI occurs in about 20-25 percent of adult female FMR1 premutation carriers. It has also been reported in teenagers who are carriers, though it is less common in that population

Autism Spectrum Disorder and Fragile X Syndrome

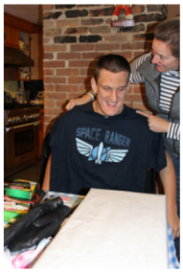
- Fragile X Syndrome is the most common known single gene cause of ASD
- Autism spectrum disorder (ASD) is a behavioral diagnosis.
- The range of symptoms in ASD vary and are generally characterized by an impaired ability to communicate and interact socially with other people
- Sometimes children will not meet the diagnostic criteria for ASD but will have autistic-like features.

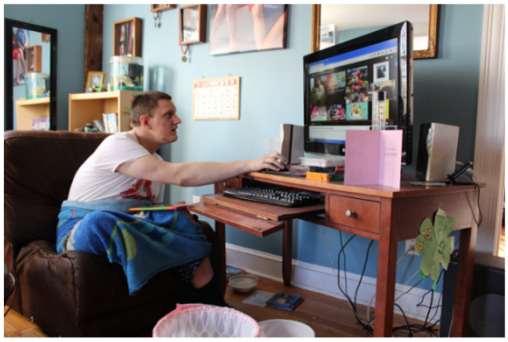
What is FXTAS

- Fragile X–associated tremor/ataxia syndrome (FXTAS) is an "adult onset" neurodegenerative disorder, 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 affects the neurologic system and progresses at varying rates in different individuals.

Treatment and Intervention

- While there is currently no cure for Fragile X syndrome (FXS), there are many areas of treatment and intervention that can improve the lives of affected individuals and their families.
- Given the proper education, therapy, and support, all persons with FXS can make progress.
- Most children with FXS qualify for special education services. Education can be complemented by a variety of therapies that will help your child become more independent in the transitions from childhood through adolescence and into adulthood.

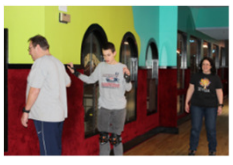




The National Fragile X Foundation Western Mass Chapter started in 1997

The group was started to support parents in the Western part of the state and raise awareness in the area. We raise awareness about Fragile X through autism conferences, fundraising, speaking to college students/medical students and many other events.

We support Western Mass families by offering events like roller skating, information about new research/drug studies, scholarships to Advocacy Day and the International Conference, local scholarships to conferences also.






Who should have Fragile X Testing?

There are three general circumstances in which Fragile X testing should be considered:

1. Clinical symptoms that suggest Fragile X Syndrome, FXTAS or infertility/FXPOI.
2. A family history of Fragile X syndrome, FXTAS, intellectual or learning disabilities or autism of unknown cause, or infertility.
3. Family or personal history of a Fragile X Genetics and Inheritance (carrier).

Specific indications for testing include:

- Any male or female with intellectual disabilities, developmental delay, speech and language delay, autism or learning disabilities of unknown cause.
- Any female with infertility, elevated FSH levels, premature ovarian failure, primary ovarian insufficiency or irregular menses.
- Any adult over 50 with features of FXTAS, including intention tremors, ataxia, memory loss, cognitive decline, personality change, especially in combination with a positive family history of Fragile X.
- Any preconception or pregnant woman who expresses interest in or requests Fragile X carrier testing.

Fragile X Clinics

Many Fragile X Clinics are opening across the United States and around the world. Clinics can guide parents to medication options that may address the symptoms of FXS, while new medications are coming on the market that aim to treat the underlying condition.Parents must address a new set of issues as their child with Fragile X syndrome grows into adulthood. The NFXF provides a variety of Resources to assist in this transition, from print and multi-media presentations to web-based materials.

Fragile X Clinics Part 2

A Fragile X clinic provides individuals and families affected by one or more of the Fragile X conditions with a comprehensive evaluation and treatment recommendations. These are supported by the latest medical, educational, and research knowledge available.

All of the clinics provide medical services (including medication evaluation and consultation) supervised by a Medical Doctor. Multidisciplinary services and/or referrals, such as occupational therapy, speech and language therapy, behavioral therapy and genetic counseling, are available within the institutions or through referral.

Many of the clinics also participate in collaborative research efforts with other Fragile X clinics and professionals.
All of the clinics have an emphasis on Fragile X syndrome (FXS) while also providing services and/or referrals for Fragile X-associated tremor/ataxia syndrome (FXTAS) and Fragile X-associated primary ovarian insufficiency (FXPOI).

Besides serving families closer to where they live, these clinics also benefit the Fragile X community by sharing knowledge, research, and clinical experience with one another. This in turn helps the patients whom they serve.

What is next for Fragile X..?

- Research
- BioBank
- FORWARD

fragile x syndrome (noun)
also,
- phenomenon that brings families closer together, creating powerful change and hope for the future.

Questions?