

Fragile X At a Glance

Fragile X syndrome is a condition characterized by intellectual disability and physical features, that is more common in boys than in girls although it affects both. It is caused by a change in a gene (FMR1) on the X chromosome. The condition is characterized by mild to moderate intellectual disability in males and normal to mild intellectual disability in females. There is variability in the level of intellectual disability and other behavioral and physical features.



Meet Jack on page 12.

Fragile X is the result of one of two types of gene mutations. It can either be a full mutation (FMR1) or a loss of function mutation. Females have *some* of the characteristics and physical features as males who have Fragile X syndrome. There is variability of characteristics associated with this condition.

About 1 in every 3,600-4,000 baby boys is born with Fragile X, and 1 in 4,000-6,000 baby girls.

The diagnosis is made based on genetic testing. The testing identifies a difference in the FMR1 gene. The FMR1 gene produces a protein important for normal brain development. Fragile X syndrome is caused by an increase in the number of repeats (CGG repeats) of a portion of the FMR1 gene that causes the gene to not be able to make the protein.

The increase in the number of repeats can be small and results in the person being a carrier, or having a "pre-mutation". Fragile X **carriers** or FMR1 pre-mutation carriers may not experience any symptoms. A pre-mutation carrier may "carry" the potential to pass the pre-mutation on to their offspring.

MALES - The clinical findings in males with a full mutation may include the following:

- Intellectual disability is present. Males with full mutation have a significant intellectual disability (IQ may range from 22-65).
- Behavioral differences may occur. Other diagnoses/characteristics **may** include:
 - Autism spectrum disorder
 - Hyperactivity
 - Shyness
 - Gaze aversion
 - Hand flapping

- Hand biting
- Temper tantrums
- Specific facial appearance and physical findings. Not every individual will have all of the findings.

Facial features may include:

- Large head
- Long face
- Large forehead and chin
- Ears that stick out
- High arched palate or sometimes cleft palate
- These features become more noticeable with age.

Macro-orchidism (large testes) typically happens after puberty.

Loose joints mostly in fingers, thumbs, wrists. Also, flat feet occur.

Cardiac findings include mitral valve prolapse and aortic root dilatation.

Strabismus (crossed eyes) is present.

Skin is extra soft and smooth.

- Other findings may include: low muscle tone, reflux, repeated otitis media, high blood pressure, possible seizures.

FEMALES - Clinical finding in females who have a full mutation:

- Females have some of the same physical features as males.
- Intellectual disability may be found in up to 50% of females. Females with a full mutation have a mild intellectual disability (IQ may range from 74-91).
 - Girls who have a full mutation and a normal IQ may have learning difficulties or emotional problems. These may include: social anxiety, selective mutism, shyness, poor eye contact, hyperactivity, and/or impulsive behavior.
 - Girls may have subtle cognitive features, such as difficulty with math or excessive shyness without other traits.

Things to Think About

1. Medical / Dietary Needs

What you need to know

Medical home providers can help families access educational supports. Psychological and/or psychiatric evaluation, behavioral supports, and/or medications may be helpful with Attention Deficit Disorder (ADD).

No special diet is required for individuals with Fragile X syndrome. A well-balanced diet and exercise is recommended.

What you can do

- Refer as needed to early intervention, special education, and other supports.
- Medical Home can keep an eye on vision, hearing. School staff can do vision and hearing screenings.
- Early intervention for delayed milestones
- Special education
 - Small class size
 - Individual attention
 - Avoid sudden change or too much stimulation
- Vocational training
- Individual drug treatment of behaviors that affect social interactions
- Routine treatment of medical problems
- Behavioral and psychological testing as needed

Milestone averages:

- Sit alone ~10 months
- Walk ~20.6 months
- First clear words ~20 months

2. Education Supports

What you need to know

It is important to have HIGH EXPECTATIONS for learning for children who have Fragile X.

People who have Fragile X syndrome vary in their learning needs and cognitive abilities. Challenges vary between boys and girls.

Children who have Fragile X can be included in their neighborhood schools. They may do well when extra supports are provided. Being with their peers is good for developing social and communication skills. Their team can make decisions about the need for specific teaching and pace.

Complete developmental and educational assessments (including speech and language evaluation and occupational/physical therapy evaluation) are recommended for planning. A multidisciplinary team and approach is helpful.

Helpful Ideas

- Integrated teaching approach
- Support for developing friends
- Occupational therapist and speech therapists work together to design programs such as social-motor groups.
- Special education and anticipatory management. For example, avoidance of too much stimulation may prevent behaviors.
- Goals that emphasize the child's learning style and strengths
- Concrete, high interest examples and materials
- Recognize need for individual attention, small class size, and avoidance of sudden change
- Modeling and imitation for both behavioral and communication goals
- Highly structured, predictable routines with visual cues for changes in expected events
- Inclusion of functional skills, along with the academic skills
- Self-help goals integrated with language and motor skills (i.e. eating, dressing, toileting).
- Occupational therapists ensure appropriate settings and equipment for a calm, structured learning environment. Use of special chairs, postural supports, etc., may be helpful.

Intellectual Development

- Cognitive abilities range from typical, to learning disabilities, to intellectual disability. Males with a full mutation may have more pronounced cognitive issues than females with a full mutation.

Communication

- Fragile X Syndrome had been associated with delays in various aspects of language including: phonology (speech sounds), semantics (word meaning and vocabulary), syntax (word structure and grammar), and pragmatics (the functional use of language). (Roseberry-McKibbin & Hedge).
 - They may have difficulty with social language skills including phrases, sentences, and staying on topic.
 - Speech production may be fast or have a disordered rhythm. Verbal dyspraxia can result in unintelligible speech.
 - Language rich environments are important. They provide good role models for conversational goals.
 - Communication and sensory motor goals may be addressed together. Speech-language pathologists and occupational therapists have ideas for oral-motor stimulation and may work well together

- They may have difficulty with motor planning (praxis) and sequencing oral motor, gross motor, fine motor, and visual motor skills. Verbal apraxia can result in unintelligible speech.
- Children with Fragile X may have difficulty with gestures, eye contact, and symbolic play skills. These aspects, along with difficulties in making transitions, topic maintenance, and turn taking during conversation impact social pragmatic abilities. Intense speech and language therapy to remediate and focus on these skills is recommended.
- Speech may be difficult to understand due to errors in making the sound of speech and rapid rate of speech. Children have also been noted to have unusual rhythm and volume while speaking. Speech may sound “cluttered” as the child experiences difficulty planning the sequence of intricate movements needed to produce sounds and words.
 - Language rich environments are important. They provide good role models for conversational goals.

In boys who have fragile X syndrome full mutation, there may be:

- A strength in verbal and memory for meaningful information in context (i.e. pictures)
- Difficulty with the pragmatics of conversation. This is affected by physical, oral motor attention and behavioral issues.

Girls with fragile X syndrome full mutation:

- Have many good verbal skills. However, they may have difficulty in pragmatic speech with anxiety and shyness affecting social interaction.
- May need to work with speech pathologist for conversational goals.
- May be beneficial to work in small groups on social language.

Executive function difficulties

- People who have fragile X syndrome have executive function difficulties.
- Executive function difficulties affect planning, thinking flexibly, and understanding abstract ideas.
- This causes a struggle to remember, process, and organize information efficiently.
- This can lead to problems in mathematics and reading.

Attention, memory, reading and writing

- Children who have fragile X syndrome often struggle to hold information in their short term memory.
- Executive function difficulties affect planning, thinking flexibly, and understanding abstract ideas.
- This causes a struggle to remember, process, and organize information efficiently.

Math learning difficulties

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- People who have Fragile X syndrome have significant visuospatial dysfunction, poor math attainment, and executive dysfunction.
- Difficulties can be seen in areas of nonverbal processing, visual-spatial skills, complex verbal memory, attention, working memory, visual-spatial memory, and math.
- Math learning difficulties include difficulty understanding and representing quantities, and getting the numerical meaning from symbolic digits.

Learn more:

- Individuals show adequate fact retrieval. Development of procedural strategies is delayed.
- Word problems are a weakness. They are challenging due to their procedural nature and difficulty in reading comprehension.
- Important to emphasize: math skills for achieving independence.

In males who have fragile X syndrome full mutation:

- Processing sequential information is a weakness and may affect math performance (i.e. counting).
- They have difficulty with one to one correspondence when counting, sequential processing, inhibition, selective and sustained attention.

Females with fragile X syndrome full mutation:

- May require special education supports in developing math skills.
- May have challenges with visual perceptual skills, such as recalling location.
- May have executive function difficulties including: working memory, inhibition, and/or sustained attention.

What you can do

General Instruction Tips

- Keep distraction to minimum
- Use visual aids
- Teach using several short periods of time
- Use music, singing, and movement in instruction.
- Help with organization.
- Present information in concrete manner.

- Repetition will help children remember basic facts.
- Whole language based approach with high interest material has been successful for reading and spelling.
- Individuals may learn better when material is presented in a holistic manner rather than sequential one.
- Visual cues, including logos, pictures, and sign language are helpful.
- Use visual and tactile examples.
- Break down word problems to help with reading.
- Help children apply information they have learned to new situations.
- Use concrete examples.

Other helpful hints

- Help the child to chew on gum, fruit snacks, and hard, crunchy foods, rather than his hands, clothes, or backpack straps.
- Social-motor groups can be designed to incorporate movement into role-playing.
- Board games and computers might be used for turn taking, communication, and fine motor skills.
- Assistive technology
 - For children with fragile X syndrome who are not yet speaking, the assistive technology might be picture cards, a language board, or a computerized, talking device.
 - For a child with low muscle tone, the assistive technology could be a special chair to help with positioning and posture.
- Using a computer may help decrease frustration when writing.
- Foster association between numbers and quantities. This could include playing number board games.

Communication

- Communication and sensory motor goals may be addressed together. Speech-language pathologists and occupational therapists have ideas for oral-motor stimulation and may work well together.
- Otitis media may be common and ear infections should be monitored.
- Ensure the child is followed by a speech-language pathologist to assist with communication.
 - Help increase clarity of message
 - Help increase rate of speech, rhythm, volume, and articulation
- Early intervention is very important.

3. Behavior & Sensory Support

What you need to know

Behavioral and psychological assessments are important for individuals with fragile X. They can help determine the presence of concentration/attention problems, anxiety, obsessive-compulsive disorder, aggression, or depression.

Females with fragile X syndrome full mutation may have:

- Social anxiety
- Shyness
- Gaze avoidance

Males with fragile X syndrome full mutation may have:

- Autism (15-25%)

Learn more:

- An additional 50-90% of individuals with fragile X syndrome have symptoms of autism including poor eye contact, hand flapping, hand biting, perseveration in speech and tactile defensiveness.
- Autism spectrum disorder and/or attention deficit hyperactivity disorder may be seen in permutation carriers.

- Management of behavior using drugs

Learn more:

- No particular treatment has been found to be uniquely beneficial. Therapy must be individual and closely monitored.
- Medication may be necessary for ADHD and/or anxiety or depression.

- Often tactile defensiveness
- Poor eye contact
- Perseverative speech
- Problems in impulse control
- Easily distracted
- Often fidgety or overactive
- Postural control challenges
- Difficulty with self-control

- Sensory defensiveness
- Task avoidance
- Poor understanding of expectations
- Individuals with fragile X have difficulty with self-regulatory function. This causes difficulty with
 - Attention
 - Anxiety
 - Modulation of activity level
 - Easily aroused in situations with excessive auditory, visual and tactile stimuli.
 - This may lead to tantrums or increased hyperactivity
 - Situations like the cafeteria, theater, and/or gym may be challenging
 - Arousal states
 - Impulsivity

What can you do

Helpful hints that may work for children with Fragile X

- Quiet spaces when needed
- Small group instruction
- Calming activities
- Clear concrete plans and visual cues (i.e. visual sign for quiet)
- Proactive behavioral plans that include goals, rewards, and consequences for appropriate behavior
- Seating in back section of room and/or allow seating near exit.
- Structure and predictability
- Reduced level of environmental noise/sound, natural lightening, and avoidance of crowded areas.
- Predictable transitions and signal with visual cues
- Non-verbal cues and feed back
- Role play behavioral consequences
- Physical activities
- Alternative to stressful events
- Breaks and downtime if needed
- Work on conversational skills and friendships

4. Physical Activity, Trips, Events

What you need to know

Children who have Fragile X syndrome may have a hard time with change and transition. They can be easily aroused in situations with excessive stimuli. This may lead to tantrums or increased hyperactivity.

If you live in New England (USA) and qualify, Northeast Passage offers Therapeutic Recreation and Adaptive Sports programming (www.nepassage.org) .

What you can do

Be proactive and think about the potential for stress before going into the cafeteria, theater, and/or gymnasiums. If these are difficult settings, use some of the supports in the educational intervention section of this site to help prevent difficulties.

5. School Absences & Fatigue

What you need to know

Children who have Fragile X syndrome should not have increased absences due to their condition. They should not require any accommodations for fatigability.

What you can do

Notify school nurse or parent if you see anything out of the ordinary.

6. Emergency Planning

What you need to know

Emergency plans should be made on an individual child, based on child's behaviors and needs.

If any change is noticed in an individual who has Fragile X syndrome, it is important to contact the parents.

7. Resources

Fragile X Syndrome: Diagnosis, Treatment and Research

This book published in 2002 by John Hopkins University Press is a good resource. It was edited by Randi Jessen Hagerman, M.D. and Paul J. Hagerman, M.D., Ph.D.

Here is a link to Amazon.com for details:

<http://www.amazon.com/Fragile-Syndrome-Diagnosis-Treatment-Contemporary/dp/0801868440>

National Fragile X Foundation – Lesson

<http://www.fragilex.org/treatment-intervention/education/lesson-planning-guide/>

This Guide is intended for classroom teachers who may have little or no exposure to fragile X syndrome (FXS). Our goal is to help make the teaching and learning environment from preschool through high school more effective, more efficient, and more rewarding for teachers and the student(s) with FXS who come under their care.

FRAXA Research Foundation

<http://www.fraxa.org/>

“Finding a Cure for Fragile X”

GeneReviews

<http://www.ncbi.nlm.nih.gov/books/NBK22189/>

Learn more about the genetics of Fragile X.

American Academy of Pediatrics (AAP) – Health Supervision Guidelines

<http://pediatrics.aappublications.org/content/127/5/994.full.pdf%20html>

The AAP endorses the Health Supervision Guidelines. Families may find these helpful when talking to their pediatricians or family physicians.

Classroom Accommodations for Students with Visual Issues

Classroom Accommodations for Students with Visual Issues is a resource from Boulder Valley Vision Therapy, P.C. (<https://www.bouldervt.com/>), in Boulder, Colorado. It describes a variety of variety of issues and offers strategies.

<https://www.bouldervt.com/wp-content/uploads/sites/478/2015/12/227-Course-Handout-CCIRA-2016.pdf>

Genetics Home Reference

<https://ghr.nlm.nih.gov/condition/fragile-x-syndrome>

Consumer-friendly information about human genetics from the U.S. National Library of Medicine

Fragile X Foundation – Vermont Chapter

Vermont Chapter of the National Fragile X Foundation is a resource for all affected by Fragile X in the state of Vermont. They help families, spread awareness, find improved treatments, support research, act as advocates, and provide educational opportunities for all affected by a Fragile X-associated disorders. On Facebook, search for @fragilexvt.

8. Meet a Child with Fragile X

Meet Jack, “The Mayor”!

GEMSS would like to thank Jack and his mother for their generosity in sharing this story with us. You have made the site come to life with the addition of your thoughts and feelings. Thank you so much!

“Jack is one of the happiest children I have ever met,” says his mother. He has a smile that can light up a room and an endless supply of hugs. He is never too busy to say hello to a friend or to tell a stranger to “have a great day.” Being such a friendly little guy has earned him the nickname “the mayor” at his elementary school.



Jack loves tractors, trains, cows, and music of all types. “He is the only seven year old I know that enjoys listening to jazz while eating his after school snack or singing Tom Petty classics while getting dressed in the morning,” quips his mother. He prefers British cartoons to American and has a whole repertoire of funny little voices and accents that he uses on a regular basis. It is difficult to be around Jack and not smile! In fact, when the family goes out into the community, it is not unusual for him to leave a trail of smiling strangers behind him.

Ever since Jack was a tiny baby he has loved books. Snuggling and reading a favorite book has always been the one thing that makes him feel better when he is sad or anxious. It wasn't until he started talking at age four that his family realized that “he could read really, really well.”

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They also found out that his anxiety is much more severe than they ever imagined. “Over the years he would spontaneously cry when we were outside. It was after he started talking that he was able to tell us that he was afraid of car horns,” his mother remarks.

Jack has made huge gains since he was diagnosed with Fragile X Syndrome at sixteen months. “He has most of the symptoms typically found with Fragile X including: dyspraxia, sensory processing disorder, hypotonia, difficulty with communication and anxiety,” according to his mother. But, with the help of incredible private therapists, dedicated and experienced school staff, and, most of all, lots of peer support he is able to overcome the challenges that the symptoms cause. His mother affirms, “We believe that Jack's progress is a direct result of the support he receives in a variety of environments.”

The staff at his school communicates with his family and his private therapists on a daily basis to provide consistency. His classmates include him in school activities and social engagements outside of school. “They are a great group of kids who, despite being very young, are patient and supportive,” his mother says. Jack has a nine year old brother named Michael who reminds Jack on a daily basis that he is “the best brother in the world.” Jack has inspired Michael to get to know other children with disabilities. With permission from Michael's school special education director, he now spends time everyday reading to children with disabilities.

His mother feels that “Jack will face many more obstacles as he grows but I am confident that he will continue to conquer them as long as he has the support he needs.”