



Genetic Education Materials for School Success

# Fragile X Syndrome

## Table of Contents

At a Glance .....	2
Medical and Dietary Considerations .....	3
Education Supports .....	3
Behavior & Sensory Support .....	6
Physical Activity, Trips, Events .....	7
School Absences & Fatigue .....	8
Emergency Planning .....	8
Resources .....	9
Info for School Nurse and Primary Care Staff .....	10
Meet a Child with Fragile X: <i>Meet Jack, "The Mayor"!</i> .....	11

## Acknowledgment:

Genetic Education Materials for School Success (GEMSS) was created and maintained by an interdisciplinary team of genetic counselors, physicians, families, and students working with New England Regional Genetics Network (NERGN) and the Institute on Disability at the University of NH. Thank you to these amazing partners. This work was supported by HRSA grant UH7MC30778.

## Disclaimer:

This is not intended to be medical or clinical advice. Any recommendations here should be reviewed for the appropriateness for your individual child and their unique situation. Also, please note that each document should include the date that it was last reviewed or updated. Research and guidelines that have been released after that date may represent more up-to-date recommendations.

# Fragile X Syndrome

## At a Glance

Fragile X syndrome is a rare genetic condition that causes intellectual disability, behavioral issues, and specific physical features. There is a lot of variation in the seriousness of the physical and intellectual effects between affected children. It is more common and generally more severe in males.

Fragile X syndrome is caused by a change in a gene on the X chromosome. Females have two X chromosomes, but males have one X and one Y chromosome. If a female has a mutation in one of her X chromosomes, she most likely still has one X chromosome that does not have a mutation. That female will only show some of the symptoms of fragile X. However, if males have a mutation on their X chromosome, they do not have a second “healthy” X chromosome to moderate the effects. These males will have the disorder.



*Meet Jack on page 11*

## Common Features of Fragile X Syndrome:

*Note – Females can present with any of these characteristics though they tend to have less significant intellectual disability and less obvious facial characteristics than males.*

- Intellectual disability
- Learning difficulties
- Autism spectrum disorder
- Anxiety generally worsens after puberty
- Behavioral issues
  - ADHD - hyperactivity, poor impulse control, distractibility
  - Flapping their hands or biting their hands
  - Social anxiety, shyness, poor eye contact
  - Temper tantrums, irritability, aggression
- Sensory issues - sensitivity to crowds, sounds, touch, foods, textures
- Specific facial features - long face, prominent forehead, large ears, and prominent jaw
- Other medical issues:
  - Seizures
  - Ear infections
  - Crossed eyes or other vision problems
  - Low muscle tone

## In Depth

### Medical and Dietary Considerations

#### What You Need to Know

Fragile X syndrome affects every child differently. The healthcare team and family can provide specific information about the unique needs of their child. While aspects of the condition can be challenging, there are many positive features. For example, researchers have noted that people with fragile X are helpful, kind, thoughtful, friendly, good imitators, good at visual and long-term memory, and humorous.

There are no FDA approved drugs specifically for the treatment of fragile X. In many cases, medications are used to treat certain symptoms of fragile X syndrome that are found in other conditions such as ADHD (attention deficit hyperactivity disorder) or anxiety. Many individuals with fragile X syndrome experience seizures. Communicate with the family about that or any other medical condition that may require management.

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

#### What you can do

- Encourage at least annual comprehensive visits to child's medical home. Obtain any updated recommendations.
- Routine treatment of medical problems identified
- Refer as needed to early intervention, special education, and other supports
- School staff should do annual vision and hearing screenings
- Communicate any changes in child's behavior, mood, or activity with parent

### Education Supports

It is important to have high expectations for learning for children who have fragile X, utilizing their strengths and interests as a foundation. Encourage use of the core educational curriculum and modify it to meet the child's individual needs.

#### What you need to know

People who have fragile X syndrome vary in their learning needs and cognitive abilities. Individuals with fragile X who receive appropriate education, therapy services, and medications have the best chance of using all of their individual capabilities and skills. Even those with an intellectual or developmental disability can learn to master many self-help skills.

Because a young child's brain is still forming, early intervention gives children the best start possible and the greatest chance of developing a full range of skills. 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.

## **Intellectual Development**

Cognitive abilities range from typical, to learning disabilities, to significant intellectual disability. Males may have more pronounced cognitive issues than females.

## **Communication**

- Fragile X Syndrome has 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).
- There may be difficulty with social language skills including phrases, sentences, and staying on topic.
- 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.
- 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.

## **Executive Function**

- There may be difficulties affecting planning, thinking flexibly, and understanding abstract ideas.
- This causes a struggle to remember, process, and organize information efficiently leading 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.

## **Math**

- 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.
- Individuals may show adequate fact retrieval, while development of procedural strategies are delayed.
- Word problems are a weakness. They are challenging due to their procedural nature and

difficulty in reading comprehension.

- *In males who have fragile X syndrome full mutation* you may observe challenges with one-to-one correspondence when counting, sequential processing, inhibition, selective and sustained attention.

## What you can do

Complete developmental and educational assessments (including speech and language evaluation and occupational/physical therapy evaluation) are recommended for planning. An integrated, multidisciplinary team and approach is helpful. Develop goals that emphasize the child's learning style and strengths.

### General Recommendations

- Communicate regularly with parents
- Use early intervention services for delayed milestones
- Consider vocational training
- Prioritize small class size and individual attention
- Avoid sudden changes or too much stimulation
- Keep distraction to minimum
- Use visual aids such as logos, pictures, and sign language
- Teach in multiple shorter blocks of time
- Use music, singing, and movement in instruction
- Help with organization
- Present information in concrete manner and use concrete examples
- Repetition will help children remember basic facts
- Use whole language-based approach with high interest material for reading and spelling
- Present material in a holistic manner rather than a sequential one
- Use visual and tactile examples
- Break down word problems to help with reading
- Help children apply information they have learned to new situations

### Other helpful hints

- Board games and computers might be helpful for turn taking, communication, and fine motor skills.
- Foster association between numbers and quantities. This could include playing number board games.
- Occupational therapists ensure appropriate settings and equipment for a calm, structured learning environment.
- 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 children 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.
- Provide support for developing friends.
- Occupational therapist and speech therapists work together to design programs such as social-motor groups.
- Include functional skills, along with the academic skills
- Self-help goals can be integrated with language and motor skills (e.g., eating, dressing, toileting).

## **Communication**

- Language-rich environments are important; 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.
- Ensure that the child is seen by a speech-language pathologist to assist with communication.
  - Help increase clarity of message
  - Help increase rate of speech, rhythm, volume, and articulation
- *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.

## **Behavior & Sensory Support**

### **What you need to know**

Psychological and/or psychiatric evaluation, behavioral supports, and/or medications may be helpful with behavioral issues.

Behavioral and psychological assessments are important for individuals with fragile X. They can help identify and develop treatment plan for concentration/attention problems or mental health issues like anxiety, obsessive compulsive disorder, or depression. Social anxiety is common among females with fragile X syndrome. Autism spectrum disorder and/or attention deficit hyperactivity disorder are also frequently seen.

### **Common Behaviors**

- Tactile/sensory defensiveness
- Poor eye contact
- Hand flapping/hand biting
- Perseverative speech

- Problems in impulse control
- Easily distracted
- Often fidgety or overactive
- Postural control challenges
- Difficulty with self-control
- Task avoidance
- Poor understanding of expectations
- Difficulty with self-regulatory function can causes problems with
  - Attention
  - Anxiety
  - Modulation of activity level
    - Easily aroused in situations with excessive auditory, visual and tactile stimuli
- This may lead to meltdowns or increased hyperactivity
- Situations like the cafeteria, theater, and/or gym may be challenging

## What can you do

- Provide behavioral and psychological testing as needed
- Develop proactive behavioral plans that include goals, rewards, and consequences for appropriate behavior
- Utilize anticipatory management (e.g., avoiding too much stimulation may prevent behaviors.)
- Encourage highly structured, predictable routines with visual cues for changes in expected events
- Provide quiet spaces when needed
- Employ small group instruction
- Foster calming activities
- Encourage appropriate physical activities
- Provide alternatives to stressful events
- Allow breaks and downtime if needed
- Consider seating in back section of room and/or allow seating near exit
- Reduce the level of environmental noise/sound, natural lighting, and avoid crowded areas
- Consider non-verbal cues and feedback
- Provide modeling and imitation for both behavioral and communication goals
- Employ role-play behavioral consequences
- Work on conversational skills and friendships
- Design social-motor groups which incorporate movement into role-playing
- Offer the child gum, fruit snacks, and hard, crunchy foods to chew on, rather than his hands, clothes, or backpack straps.
- Allow prescribed medications

## 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 overstimulated in new situations. This may lead to meltdowns or increased hyperactivity.

## **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 behavioral intervention section of this site to help prevent difficulties.

## **School Absences & Fatigue**

Children who have fragile X syndrome should not have increased absences due to their condition. They should not require any accommodations for fatigue. Notify school nurse or parent if you see anything out of the ordinary.

## **Emergency Planning**

Emergency plans should be made for an individual child, based on the 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.



## Resources

### **Medline Plus**

MedlinePlus is an online health information resource for patients and their families and friends. It is a service of the National Library of Medicine. This link will provide a good medical overview of the condition without being too technical.

<https://medlineplus.gov/genetics/condition/fragile-x-syndrome/>

### **National Fragile X Foundation**

The National Fragile X Foundation has a very useful website. <https://fragilex.org/>

Check their Family Resources page for some great info. Below the banner you will see a list of resources by topic. Click on “School & Education” to find helpful documents for school settings.

<https://fragilex.org/family-resources/>

### **FRAXA Research Foundation**

Fraxa supports research to develop treatments and an eventual cure for fragile X syndrome. Go to their website for research updates and opportunities to participate. <http://www.fraxa.org/>

## Info for School Nurse and Primary Care Staff

Fragile X syndrome is a complex condition that will be best managed by good collaboration between the school nursing staff and primary care staff as part of the student's team. Work with the parents to get signed releases to share information at the school nurse's office and the primary care office.

*Gene Reviews* is a peer-reviewed point-of-care reference accessible via the National Library of Medicine. The information for fragile X syndrome is included in the entry for FMR1 disorders. It provides a comprehensive summary of the condition and describes some ongoing surveillance and management issues. It is a great resource for medical providers.

<https://www.ncbi.nlm.nih.gov/books/NBK1384/>

### HIPAA Concerns

Sometimes concerns by primary care staff about violating HIPAA regulations can hamper care by limiting access to necessary medical information. Please note that if care is intended to be delivered by school staff, clarifying medical orders or medication instructions represents continuation of medical care and is not a violation of HIPAA. To avoid any issues like this, remind the parents to sign a release of information form at the primary care office. The school nurse should keep a copy of that release in the student's file, if possible, to facilitate sharing of information in the event that there is difficulty obtaining needed information.

### Example letter for physician to provide to school

"My patient \_\_\_\_\_ has been diagnosed with fragile X syndrome, a rare genetic condition which can lead to intellectual and behavioral challenges. There is also an increased risk of certain medical complications. A thorough psychological assessment is warranted to identify frequently co-occurring conditions like ASD or anxiety and then develop a management plan. This condition necessitates an Individualized Education Program (IEP) or 504 plan.

## Meet a Child with Fragile X: *Meet Jack, “The Mayor”!*

“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.” 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.”

*Thank you so much to Jack and family for sharing their story! And thank you to the team at the Institute on Disability at UNH for assisting in collecting these stories.*