

Fragile X Syndrome

What is fragile X syndrome?

Fragile X syndrome is an inherited form of intellectual disability that is primarily seen in males. It is the most common form of inherited intellectual disability. Fragile X syndrome is caused by the *FMR1* gene on the X chromosome. Females tend to be less affected because they have 2 X chromosomes, and one can compensate for a mutation on the other X chromosome. Males have only 1 X chromosome, so mutational changes are more likely to be expressed.

How common is it?

About 1 in 5,000 males born in the United States has fragile X syndrome. Females are less severely and less commonly affected (about 1 in 7,000).

What are some common characteristics of children with fragile X syndrome or of fragile X syndrome as children present with it?

- Facial features such as a long face, large ears, and a prominent jaw.
- Moderate to severe intellectual disability.
- Developmental delay such as gross-motor skill delay, fine-motor skill delay, or speech delay. Infants may have low tone.
- Behavioral abnormalities. Children with fragile X syndrome may have features of autism spectrum disorder (ASD) (including ASD comorbidities), which include hand flapping, poor social skills, poor eye contact, and speech delay. Anxiety and attention deficits are also common.
- Seizures may occur in a small portion of children with fragile X syndrome (about 15% of them).
- Some evidence supports that children with fragile X syndrome may be more prone to ear infections and eye problems.
- Sleep disturbances are common.



Child with fragile X syndrome

Fragile X Syndrome *(continued)*

Who might be on the treatment team?

- Pediatrician/primary care provider in the medical home
- Geneticist
- Developmental-behavioral pediatrician
- Psychologist
- Special education and vocational training teachers
- Physical and occupational therapists
- Speech-language therapist
- Social worker

What adaptations may be needed?

Dietary Considerations

Children with fragile X syndrome may have gastroesophageal reflux disease (see Gastroesophageal Reflux Disease [GERD] Quick Reference Sheet [page 129]) and may need medication or special feeding techniques.

Physical Environment and Other Considerations

- Behavioral interventions may need to be in place.
- Early intervention should be involved early on.
- Avoid excess stimulation in infancy. Quiet, low-light, soothing environments may be best.

What should be considered an emergency?

No emergencies are directly related to fragile X syndrome, but seizures may occur.

What types of training or policies are advised?

- Teachers and caregivers may need training about how to adapt daily lessons and schedules to accommodate these children.
- Medication administration training may be needed.
- Specific training about the individual child's special needs may be necessary.

- The child's schedule may need to be adapted if individual therapy (ie, physical, occupational, speech-language) will take place at the program. Providing therapy in the classroom helps teachers incorporate it into the child's ongoing daily activities, many of which may be suitable for the other children to share in.

What are some resources?

- American Academy of Pediatrics: www.aap.org
 - Kidd SA, Lachiewicz A, Barbouth D, et al. Fragile X syndrome: a review of associated medical problems. *Pediatrics*. 2014;134(5):995–1005
 - Raspa M, Wheeler AC, Riley C. Public health literature review of fragile X syndrome. *Pediatrics*. 2017;139(suppl 3):S153–S171
- Behavioral Data Collection Sheet (page 215) in Chapter 12
- Centers for Disease Control and Prevention: “Fragile X Syndrome (FXS)” (Web page), www.cdc.gov/ncbddd/fxs/index.html
- National Fragile X Foundation: <https://fragilex.org>, 1-800-688-8765
- National Institutes of Health, US National Library of Medicine, Genetics Home Reference: “Fragile X Syndrome” (Web page), <https://ghr.nlm.nih.gov/condition/fragile-x-syndrome>

