

Myth Busters

For Providers

Fragile X Syndrome

Fragile X syndrome (FXS) is an identifiable genetic disorder that is one of the more common heritable forms of intellectual disability. FXS and other fragile X-associated disorders are caused by mutations in the Fragile X Mental Retardation 1 gene (FMR1). Any child with unexplained developmental delay, intellectual disability and/or autism spectrum disorder should receive genetic testing for FXS. Pediatric clinicians can support early identification and evaluation, which empowers families to make informed decisions about FXS-specific services for their child, as well as family planning. A diagnosis also helps healthcare providers assess comorbidities and associated conditions.

There are many myths associated with FXS that can contribute to a delayed or missed diagnosis. Here are some of the more common myths:

MYTH: There has to be a family history of FXS for a child to have the condition.

FACT: Because the FXS mutation is an expanding mutation that can become bigger when passed on to the next generation, diagnosed individuals may lack a recognized family history of FXS. However, a family history of other fragile X-associated disorders, such as tremors and early menopause, can be an indication that a child with unexplained developmental delay, intellectual disability or autism has these features because of unrecognized FXS in the family.

MYTH: FXS is diagnosed using a chromosome test or microarray.

FACT: FXS cannot be detected with a standard chromosome test or microarray. A specific test called the “FMR1 DNA Test for Fragile X” must be ordered.

MYTH: Girls cannot have FXS, and boys with FXS are always severely affected.

FACT: Both girls and boys can have FXS. Symptoms are usually more severe in boys, but both boys and girls can exhibit symptoms ranging from normal functioning to severe intellectual disability.

MYTH: All FXS patients can be recognized by their unusual physical features.

FACT: Individuals with FXS may have dysmorphic features like prominent ears, a long, narrow face, and large testes after puberty, but many children do not have any characteristic features. Some physical features may not start to develop until puberty, if at all, but can sometimes be seen in younger children too.

MYTH: There is no value in making a diagnosis of FXS if there is no cure.

FACT: Although currently there is no cure for FXS, there are important supportive treatments that can be applied once the diagnosis is known. These include educational and therapeutic approaches tailored to individual strengths and weaknesses, screening for and treatment of medical issues, and behavioral treatment methods. Having a diagnosis also enables families to receive counseling for family planning, and for them to connect with support groups of other families affected by FXS.

What you can do as a pediatric healthcare provider

- Ensure that all children with developmental delay, intellectual disability and/or autism spectrum disorder are considered for a genetic evaluation.
- Use a family history tool to evaluate for a family history of related problems that can be seen in fragile X-associated disorders:
 - A history of ataxia or “Parkinson-like” tremors in older males and some females on the maternal side.
 - A history of early menopause or fertility problems on the maternal side.

Resources for pediatric clinicians to support the diagnosis and evaluation of patients who have or may have FXS

American Academy of Pediatrics

www.aap.org

– *Health Supervision for Children with Fragile X Syndrome (Pediatrics 2011)*

– *Comprehensive Evaluation of the Child with Intellectual Disability or Global Developmental Delays (Pediatrics 2014)*

– *Genetic Literacy in Primary Care Colloquium (Pediatrics 2013)*

PediaGene application for mobile devices



AAP Genetics in Primary Care Institute

www.geneticsinprimarycare.org

National Fragile X Foundation

<https://fragilex.org/>

Centers for Disease Control and Prevention

www.cdc.gov/fragilex



This document was supported by the Cooperative Agreement Number 5 U38 OT000167, funded by the Centers for Disease Control and Prevention. Its contents are solely the responsibility of the authors and do not necessarily represent the official position of the Centers for Disease Control and Prevention or the Department of Health and Human Services.

American Academy
of Pediatrics



DEDICATED TO THE HEALTH OF ALL CHILDREN™