

Fragile X Syndrome

Fragile X syndrome (FXS) is one of the more common known causes of intellectual disability that can run in families (inherited). FXS is caused by a change in the genetic material in each cell of the body. This change in genetic material makes it hard for cells to produce a protein that is necessary for normal brain development and normal brain function. As an inherited condition, FXS can be passed on to the next generation. People with FXS may have some or all of the following symptoms:

- Walking, talking, or toilet training later than other children of the same age
- Problems with learning
- Trouble making eye contact
- Frequent ear infections
- Trouble sleeping
- Seizures
- Autism
- Sensory difficulties (trouble with what a person sees, hears, smells, tastes, and touches)

Currently there is no cure for FXS, but an early diagnosis can help a family get treatment and services for their child sooner, and having a diagnosis may provide valuable information for other family members.

MYTH: I thought my child was tested for FXS when I was pregnant or after my child was born?

FACT: FXS requires a special blood test that is not usually included in the genetic tests that a pregnant woman gets or in the tests done right after a baby is born. The only way to diagnose FXS is with a special blood test called the “FMR1 DNA Test for Fragile X.”

MYTH: I thought girls couldn't have FXS, and boys always have severe symptoms.

FACT: Both boys and girls can have FXS. The symptoms are usually more severe in boys than in girls, but both boys and girls can have symptoms that range from mild to severe.

MYTH: Does everyone with FXS have large ears or a long, narrow face?

FACT: Many people with FXS do not have certain physical traits that textbooks attribute to FXS. However, some people with FXS do have some of these physical features. These features can be seen in younger children, but some may not show up until puberty.

MYTH: If there is no cure for FXS, why does my child need a diagnosis?

FACT: Even though there is no cure for FXS, there are educational, behavioral, and therapeutic services which can help. A diagnosis may also help families with family planning and connecting with support groups of other families affected by FXS.

MYTH: How can my child have FXS? We don't have a family history of FXS.

FACT: FXS is caused by a change in genetic material that ranges in size, and can become bigger from one generation to the next. Small size changes typically do not cause FXS, but large size changes often do cause FXS. Therefore, a person can have FXS without a family history if that person inherits a large change in the size of the genetic material while everyone else in the family has small size changes. Families who do not have FXS but have small size changes can have other signs that FXS could occur in future generations. These families have members that may have fragile X-associated disorder symptoms, such as tremors and early menopause, which could be identified through a more thorough evaluation of family history.

What to do if you think your child might have FXS

- Talk to your doctor about genetic testing if your child is not sitting, walking, or talking at the same time as other children the same age, has trouble learning new skills, or has social and behavioral problems like not making eye contact, anxiety, trouble paying attention, hand flapping, acting and speaking without thinking, and being very active.
- Talk to your family to see if anyone remembers a history of “Parkinson-like” tremors in older men on the mother’s side of the family, or a history of early menopause or fertility problems in women on the mother’s side of the family. These are symptoms of fragile X-associated disorders, which suggest that FXS could run in the family.

What to do if your child has been diagnosed with FXS

- Early intervention services in each state <http://www.parentcenterhub.org/repository/ei-overview/> help children from birth to 3 years old learn important skills. You can ask to have your child evaluated, and these services may improve your child’s development. Even if your child has not been diagnosed with FXS, he or she may still be eligible for services.
- Work with your child’s pediatrician to get care and services for your child.
- Contact the organizations who work with FXS families and become familiar with FXS resources.
- Consider joining a local group of FXS families to share information and support each other.

Resources for people and families living with FXS

National Fragile X Foundation
<https://fragilex.org/>

FRAXA Research Foundation
www.fraxa.org

Centers for Disease Control and Prevention
www.cdc.gov/fragileX



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