



Fragile X Syndrome: A Public Health Challenge

Information on living with fragile X syndrome (FXS) is expanding, but important work must still be done to understand how well care and services are working, how different people experience the condition, and how to improve early diagnosis. Answering these questions will help make sure that every person with FXS can reach their full potential.



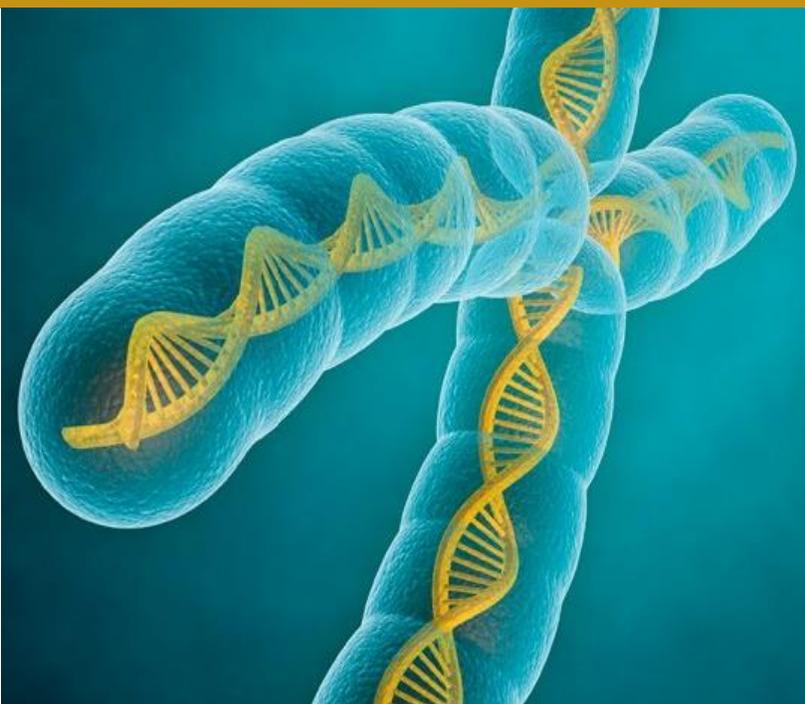
The more healthcare professionals learn about FXS, the more they can tailor their care for people living with FXS.

For families, having more information about FXS means having the tools to find and provide the best care for their loved ones.



What You Should Know About Fragile X Syndrome

- FXS is an inherited condition caused by a change in the genetic material found in every 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 brain function. A shortage of this protein often leads to intellectual disability and behavioral challenges.
- People with FXS may experience a range of behavioral and health challenges. These can include anxiety, depression, autism spectrum disorder, and difficulty with sensory issues. Difficulty with sensory issues means trouble receiving and responding to what a person sees, hears, smells, tastes, and touches. Ear infections, difficulty sleeping, seizures, and stomach or intestinal problems may also occur.
- Both males and females can have FXS. Males with FXS usually have some level of intellectual disability, ranging from mild to severe. Females can have mild to moderate intellectual disability or normal intellectual ability.
- The exact number is not known, but a review of research studies estimated that about 1 in 7,000 males and about 1 in 11,000 females may have FXS.



Public Health Needs

- **Improving the health and quality of life for people living with FXS.** Information on the medical, social, and behavioral challenges faced by people with FXS is increasing. The next step is to find out what care and services they are receiving, and which ones work best. We also need to know if different groups of people are more likely to have FXS, or whether they experience the condition in different ways.
- **Early diagnosis.** Children and adults who show signs of intellectual disability often visit many doctors before getting a diagnosis of FXS. The sooner a person is diagnosed with FXS, the more they can benefit from appropriate care and services. Since FXS is an inherited condition, other family members may also benefit from an early diagnosis.

CDC in Action

CDC's Work on FXS Includes

- **Finding out how often certain children at higher risk of having FXS are getting tested for it.** Children diagnosed with autism spectrum disorder (ASD) are at higher risk of having FXS. Experts recommend that children with ASD receive genetic testing for FXS so that if they have it, they can be diagnosed earlier. CDC researchers are examining health insurance data to find out how often children diagnosed with ASD are getting tested for FXS as recommended.
- **Collecting key data for public health research.** CDC funds the collection of data from people receiving care at FXS specialty clinics across the United States. These data are combined to form one of the largest and most detailed research databases on FXS available, known as FORWARD (Fragile X Online Registry With Accessible Research Database). Examples of information in FORWARD include:
 - Development and function over time, including language, toilet training, social interaction, intellectual disability, and mental health and behavioral challenges.
 - Medical problems and medications used by people with FXS to help with anxiety and behavior problems.
 - Care and services received by people with FXS.
- **Using the data to help people with FXS and their families.** CDC scientists and research partners analyze the FORWARD database and publish reports to help people with FXS and their families, for example by:
 - Describing the symptoms and challenges of FXS across the lifespan so that families and health care providers know what to prepare for.
 - Identifying the care and services provided to people with FXS and measuring how well that care is working for them.
 - Learning how to best measure factors such as quality of life, intellectual function, and mental health in people with FXS and their families. This helps researchers design better studies to find medications and other treatments for people with FXS.
- **Educating healthcare professionals about FXS.** CDC collaborates with the American Academy of Pediatrics to provide education on FXS for healthcare professionals to raise awareness of FXS and to encourage early diagnosis and appropriate care.

For more information, visit <http://www.cdc.gov/fragileX> or call 1-800-CDC-INFO