



# IMPROVING THE HEALTH

of People with Disabilities

## Fragile X Syndrome: A Public Health Challenge

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



The more healthcare professionals learn about fragile 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.

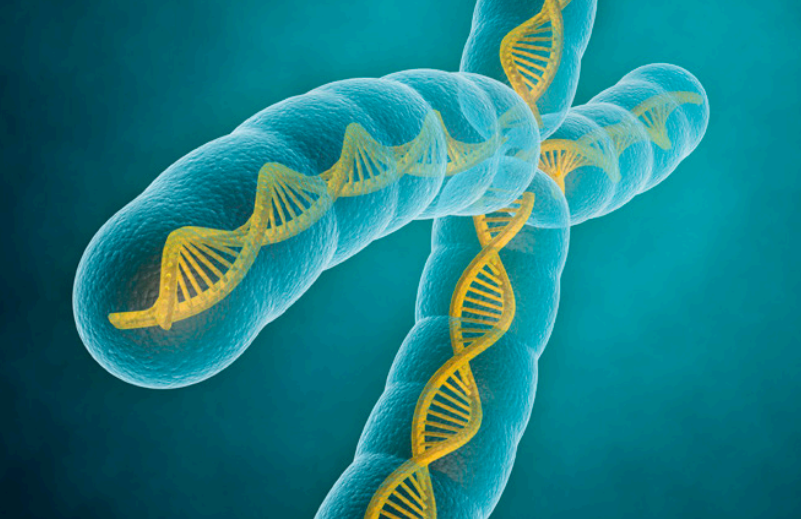


### What you should know about fragile X syndrome

- 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](#), 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 a range of symptoms from normal intellectual ability to moderate intellectual disability.
- The exact number of people who have FXS is not known, but it is estimated that about 1 in 5,000 males and about 1 in 6,000 to 1 in 8,000 females have FXS.

### Public Health Need

- **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.
- **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 improving. 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.



## What is CDC doing?

- **Building evidence through public health research.** CDC funds the collection of data for public health use from people receiving care at FXS specialty clinics across the United States.
- Examples of these data include age at diagnosis, the age when a child reaches milestones like walking and toilet training, level of intellectual ability, care and services received by each person, and each person's access to preventive services. CDC scientists and research partners can use this information to
  - » Describe the symptoms and challenges of FXS across the lifespan; and
  - » Identify the care and services provided to people with FXS and measure how well that care is working for them.
- **Using data to address the impact of FXS on people and their families.** CDC scientists and research partners are using existing data to
  - » Report on school services used by individuals with FXS, such as how many school-aged children with FXS are
    - Receiving instruction in a regular classroom setting;
    - Assisted by a personal aide at school; and
    - Receiving occupational therapy, speech therapy, and/or physical therapy.
  - » Report on the reasons why people with FXS go to the emergency room.
- **Collaborating with partners.** CDC is collaborating with the American Academy of Pediatrics to develop and distribute educational materials to healthcare professionals and families. These materials are designed to raise awareness of FXS and encourage early diagnosis so that people with FXS can receive appropriate care and services.

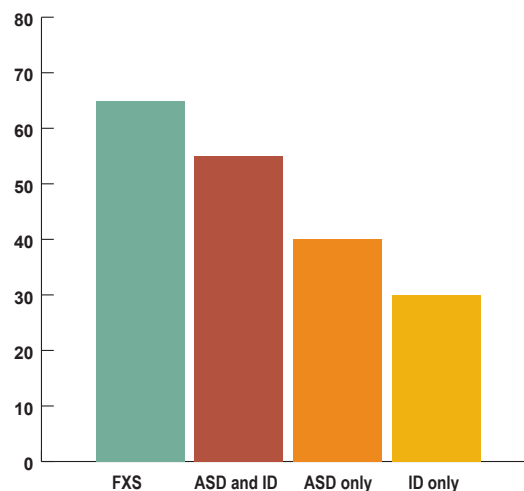
For more information visit <http://www.cdc.gov/fragileX> or call 1-800-CDC-INFO

## Future Opportunities

- There is limited information about whether the number of people affected by FXS varies by race or ethnicity. A large-scale study could provide a more specific estimate of how many people live with FXS. A large-scale study could also yield more information on the relationship between changes in genetic material and symptoms faced by people with FXS and [fragile X-associated disorders](#).
- Healthcare professionals, researchers, and families have shown an interest in screening newborns for FXS as a way to identify FXS as early as possible. To date there has not been a study comparing people who receive care and services for FXS shortly after birth with those who receive care and services for FXS after they develop symptoms of the condition. A study of this type could assess whether people with FXS do better when medications and therapies begin in the newborn phase or when these begin later.

## Percentage of families reporting financial expense

A comparison of family, financial, and employment impacts of fragile X syndrome, autism, and intellectual disability<sup>1</sup>



More families caring for a child with FXS reported excessive financial expense as compared to families caring for a child with autism spectrum disorder (ASD), with an intellectual disability (ID), or those caring for a child with both ASD and ID.<sup>1</sup>

### Reference

1. Ouyang L, Grosse SD, Riley C, Bolen J, Bishop E, Raspa M, Bailey DB. [A comparison of family financial and employment impacts of fragile X syndrome, autism spectrum disorders, and intellectual disability](#). Research in Developmental Disabilities. 2014 Jul;35(7):1518-27.