

CDC's Work: Fragile X Syndrome



Our Goal

Over the past two decades, scientists have made advancements in understanding the genetics of fragile X syndrome (FXS). However, we are just beginning to understand how this complex condition affects individuals and their families.

Some parents spend years taking their child to different doctors before getting a diagnosis of FXS. Others struggle with the behavioral and communication challenges that come with FXS. These are just two examples of the broad impact of FXS. The more we know, the better we can design services and care that will improve lives.

The Centers for Disease Control and Prevention (CDC) partners with physicians, university researchers and state health departments to better understand FXS.

Building a National Fragile X Registry

CDC supports the Fragile X Clinical and Research Consortium. The consortium is a group of more than 25 fragile X clinics across the United States. These clinics collect information in a registry to paint a picture of the full scope of FXS. The registry allows CDC scientists and research partners to gather information on a large enough group of people with FXS to make the research scientifically valid. The registry also serves as a centralized database for individuals who want to be contacted for other FXS research projects.

Some examples of information in the registry are age at diagnosis, treatment and services provided, and the range of behavioral and intellectual challenges faced by individuals with FXS. Researchers, therapists, doctors, and nurses can use information from the registry to develop better treatments and educational programs for individuals with FXS.

National Fragile X Family Survey

CDC works with researchers from the University of North Carolina at Chapel Hill and Research Triangle Institute International on a national survey of families affected by FXS. This survey asks families questions about support and medical services that they receive, issues surrounding transition of their child to adulthood, and access to health care. Researchers have analyzed these survey results and have published papers on medication use, therapy services, daily functional skills, and how families adapt to FXS. Results from this survey will be used to find better ways to support families affected by FXS.

One scientific paper published from this study looked at the employment impact and financial burden felt by families of children with FXS. The study found that almost half of families affected by FXS reported that they had experienced an increased financial burden. More than half the families said they had to change their work hours or stop working to care for a family member with FXS.¹

South Carolina Study for Adolescents and Young Adults with Rare Conditions

The state of South Carolina has a robust records system which lets CDC researchers study 15-24 year-olds with FXS. Records tell us what health care services are being used, other medical conditions that a person with FXS has, their progress in high school, vocational school or college, and whether they are participating in state employment programs. Studying 15-24 year-olds is important because this timeframe is when teenagers switch from pediatric to adult medical care. This is also the timeframe that shows how people with FXS perform in school and progress towards their employment goals.

CDC is studying the data from South Carolina because this level of detailed and connected formal records is not usually available. This approach may become a new way to more thoroughly research complicated medical conditions like FXS, using existing state records along with interviews and surveys.

Reference

¹Employment Impact and Financial Burden for Families of Children with Fragile X Syndrome: Findings from the National Fragile X Survey. Ouyang L, Grosse S, Raspa M, Bailey D. J Intellect Disabil Res; October 2010;54(10):918-28. Epub ahead of print August 26, 2010.

For further information about Fragile X Syndrome, visit <http://www.cdc.gov/ncbddd/fxs>.