

## FRAXA Research Foundation

FRAXA's mission is to find effective treatments and a cure for children and adults with Fragile X and related disorders such as autism.

Founded by three parents in 1994 and still run by parents, FRAXA is a national non-profit, tax exempt organization.

FRAXA has funded over \$24,000,000 in biomedical research, yielding discoveries which can change the lives of all families struggling with Fragile X. FRAXA is one of the most efficient charities in the world, with management and general expenses under 7%.

### *We need your help!*

We have made enormous progress: developing and testing actual drugs. But we have not yet found the cure. The need for funding is urgent.

Learn more about FRAXA and Fragile X at [fraxa.org](http://fraxa.org) or call us at (978) 462-1866. You can donate at [fraxa.org](http://fraxa.org) or send a check payable to:

**FRAXA Research Foundation**  
10 Prince Place, Suite 203  
Newburyport, MA 01950

“Studies of Fragile X have been galvanized by those most directly affected: the families and loved ones of sufferers...Experience shows that dedicated, resourceful, and, above all, motivated organizations like FRAXA sometimes do hold the key to cracking these diseases.”

– James Watson, PhD, Nobel Laureate  
FRAXA Scientific Advisor

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*All directors are parents of children with Fragile X*

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# Finding a Cure for Fragile X

The most common inherited cause of autism and intellectual disabilities worldwide



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## What is Fragile X?

*Fragile X is the most common inherited cause of intellectual disabilities and autism.*

Symptoms can include:

- learning problems
- autism
- severe anxiety
- seizures in 20-25% of boys
- attention deficit
- hyperactivity



Boys are often more severely affected than girls. Most boys have mental retardation, while two thirds of girls have normal IQ or learning disabilities. Emotional and behavioral problems are common in both sexes.

*Fragile X can occur in all races and ethnic groups.*

*It affects:*

- 1 in 4000 boys
- 1 in 6000 girls

*Brain pathways affected in Fragile X are also affected in other common disorders:*

- Autism
- Alzheimer's
- Down Syndrome



## What Causes Fragile X?

*One gene in the brain shuts down.*

In 1991 scientists discovered the gene on the X chromosome that causes Fragile X. This gene, called FMRI, shuts down and cannot manufacture the protein it normally makes—a protein vital for normal brain development.

*Fragile X can lurk in a family for generations before a child is born with a fully defective gene.*

Fragile X is carried by:

- 1 in 260 women
- 1 in 800 men

Carriers can also have symptoms: some older male carriers have Fragile X-associated Tremor Ataxia Syndrome (FXTAS) and women are at risk for early menopause.

*Fragile X testing is available.*

A simple, reliable DNA test can identify affected individuals. It can also test whether or not a person is a Fragile X carrier. This test is widely available yet most people who have Fragile X are still undiagnosed.

## FRAXA is Finding a Cure

*We are on the verge of breakthrough treatment.*

FRAXA-funded scientists have discovered a brain pathway (mGluR) which is defective in Fragile X and implicated in autism.

Investigational new drugs which target this pathway are being tested in clinics around the world.

FRAXA-funded studies have also discovered other compounds which can reverse symptoms in Fragile X animal models. We are working with university and pharmaceutical researchers to bring these compounds into clinical trials.

Treatments being investigated are likely to benefit all people with Fragile X, regardless of age.



“Fragile X is poised to become a triumph for translational research and the design of rational therapeutics for brain disease.”

– Justin Fallon, PhD  
FRAXA Scientific Advisor

“This is perhaps the most promising therapeutic discovery ever for a gene-based behavioral disease.”

– Edward M. Skolnick, MD  
Broad Institute, Harvard and MIT  
New York Times, 4/30/10

