**FRAXA Research Foundation**

FRAXA's mission is to find effective treatments and a cure for all children and adults with Fragile X. Founded by three parents in 1994 and still run by parents, FRAXA is a national non-profit, tax-exempt organization.

FRAXA has funded over $17,000,000 in research, yielding discoveries which may soon improve the lives of all families struggling with Fragile X -- and possibly autism as well. FRAXA is one of the most efficient charities in the world, with management and general expenses under 7%.

*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

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all directors are parents of children with Fragile X

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**We need your help!**

We have arrived at the “translational” stage: developing and testing actual drugs. Yet we have not yet found the cure. The need for more funding is urgent.

To learn more about FRAXA and Fragile X, visit fraxa.org or call us at (978) 462-1866. You can donate at fraxa.org or send a check payable to:

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45 Pleasant Street  
Newburyport, MA 01950

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What is Fragile X?

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

Symptoms can include:

- intellectual disabilities
- autism
- severe anxiety
- seizures, in 20-25% of boys
- attention deficit and
- hyperactivity

Boys are often more severely affected than girls. Most boys have mental retardation, while two-thirds of girls have learning disabilities or normal IQ. 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

The brain pathways affected in Fragile X are also affected in other common disorders:

- Autism
- Down Syndrome
- Alzheimer’s Disease

What Causes Fragile X?

One gene in the brain shuts down.

In 1991, scientists discovered the gene on the X chromosome - FMR1 - that causes Fragile X. In affected individuals, this one gene is shut 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

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

How is Fragile X Diagnosed?

A simple and reliable DNA test can identify affected individuals. It can also test whether a person is a Fragile X carrier. This test is widely available - yet too rarely used. Most people who have Fragile X are currently undiagnosed.

FRAXA: Finding a Cure

We are on the verge of breakthrough treatment.

FRAXA-funded research has achieved a breakthrough: scientists have found a brain pathway (mGluR) which is defective in Fragile X and implicated in autism. Drugs now being developed can target this pathway and the first clinical trials have been successful.

“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

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

It is likely that treatments being investigated now will benefit all people with Fragile X, regardless of age.

“In the coming decades, an array of important discoveries will emerge from this research and ... help develop new therapeutic and diagnostic methods for not only Fragile X but also other developmental brain disorders such as autism.”

– Susumu Tonegawa, PhD
Nobel Laureate
FRAXA Scientific Advisor