Everything that is done in the world is done by hope.
-Martin Luther

Fragile X Association of Washington State serves as a resource for families, physicians, educators and therapists. Our mission is to increase awareness about fragile X in our community.

We provide:
* information packets, a resource library and regular meetings for newly and previously diagnosed families
* educational materials to the community, schools and social service agencies
* support to further research a cure for fragile X

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Other Resources:

National Fragile X Foundation
www. nfxf.org
800-688-8765

The ARC of the United States
www.thearc.org
301-565-3842

FRAXA Research Foundation
www.fraxa.org
978-462-1866

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He's still not talking at 18 months.

My daughter isn't sitting up on her own.

I'm so frustrated. I don't know what to do.

Fragile X Syndrome

The leading cause of inherited developmental and mental impairment

FRAGILE X
ASSOCIATION OF
WASHINGTON STATE

What Is Fragile X?

Most children with fragile X appear completely typical at birth, but gradually, developmental characteristics of the condition become evident.

Fragile X syndrome is a genetic condition caused by a change in the genetic code of a single gene on the X chromosome. This defect inhibits the body's ability to produce a protein called FMRP.

Messages that must be sent and received for proper brain development and functioning are disrupted when this protein is missing.When the gene is altered, it can cause developmental delays and mild to severe learning disabilities including mental retardation.

What Causes Fragile X?
The FMR1 gene was discovered in 1991. Fragile X is passed on by a carrier - a person who may show no signs of impairment, but whose gene changes as it is passed on to a child.

When a father passes on FMR1 it remains relatively stable, but he will pass on this gene to all of his daughters and none of his sons. With each succeeding generation the expanded area can become less stable and increases the likelihood that one's child may be fully affected by fragile X. The gene expands to the affected range only when passing from mother to child. This is why fragile X can travel silently through generations, undetected, and then appear.

When a carrier mother passes on the FMR1 gene there is a 50 percent chance that each child will inherit the mutated gene. The size of the expansion does not correlate to the degree of affectedness.

Anyone with undiagnosed mental retardation in his or her family should be tested.

How Frequently?
Even today, many people with fragile X are not correctly diagnosed. Worldwide, it is estimated that one in every 2,000 boys and one in every 4,000 girls is affected.

One in 260 women and one in 700 men carry the Fragile X gene.

Fragile X Characteristics
Children may have some or none of the common physical or behavioral fragile X features. The variability of symptoms contributes to the difficulty of diagnosing it.

Most boys have several physical characteristics and impairments and tend be more severely affected than girls. Because girls have two X chromosomes, the unaffected chromosome may compensate for the altered fragile X gene and girls tend to exhibit milder and fewer characteristics.

Learning disabilities can be slight or severe and range from math or reading difficulties to mental retardation.

Some Of The Most Common Fragile X Characteristics Include:

Physical	Behavioral
• Eye & vision impairments	• Anxiety & shyness
• Elongated face	• Autism & autistic-like behavior
• Flat feet	• Hand biting & hand-flapping
• High arched palate	• Hyperactivity & short attention spans
• Hyper-extensible joints (double jointed)	• Language delays
• Large testicles (evident after puberty)	• Perseveration - repetition of the same actions or words
• Low muscle tone	• Poor eye contact
• Prominent ears	

Is There A Test For Fragile X?

A simple DNA blood test is now available to determine if a child is affected by the condition or if a person carries the fragile X gene. The same test can be used for prenatal diagnosis.

What About Treatment?

Diagnosis is essential to early intervention programs involving occupational, physical, speech and sensory integration therapies. Individualized intervention plans, including medications if indicated, can greatly improve a child's ability to reach his or her full potential.

Many medications are also available which can help with behavior issues. Currently, only the symptoms of Fragile X can be treated. Research on treating the "root cause(s)" of the condition is moving forward at an increasing pace.

Is There A Cure?

Not yet. But recent progress has been made. With continued funding for research there is great hope for future generations.

"I got very excited when we first discovered the fragile X gene," said Dr. James D. Watson, Nobel Laureate and co-discoverer of the structure of DNA. *"I think it was the first triumph of the Human Genome Project. With fragile X we've got just one protein missing, so it's a simple problem. So, you know, if I were going to work on something with the thought that I were going to solve it, oh boy, I'd work on fragile X."*