Introduction to Fragile X Syndrome

Fragile X syndrome (FXS) is a genetic disorder. A genetic disorder means that there are changes to the person’s genes. FXS is caused by changes in a gene that is on the X chromosome. This gene is called the fragile X mental retardation 1 (FMR1) gene. Everyone has this gene on their X chromosome – women have two Xs and thus two copies of this gene; men have one X (and one Y) and thus have one copy of the gene.

Note: The term “mental retardation” in literature has been replaced with “intellectual disability.”

The FMR1 gene usually makes a protein called fragile X mental retardation protein (FMRP). FMRP is needed for normal brain development. People who have FXS do not make this protein. FXS affects both males and females. However, females often have milder symptoms than males because they have two Xs, and while one of the Xs may not be producing the FMR1 protein, the other one is.

The exact number of people who have FXS is unknown, but it has been estimated that approximately 1 in 3,600 to 4,000 males and approximately 1 in 4,000 to 6,000 are born with the disorder.

Some of the Issues You Might See

A child with FXS might have developmental delays – such as in walking and talking, learning disabilities – such as in school, and characteristics such as anxiety, hyperactivity and a short attention span. Some children with FXS also have autism.

They may also have an elongated face, large ears and loose connective tissue – this may result in chronic ear infections, hernias and loose joints.

Why Should You Consider Testing?

There is no cure for FXS, however, if your child has the condition, you can begin early intervention services – services to help with learning to walk and talk and services to help you learn about and address some of the characteristics of FXS, such as sensory processing difficulties. There are doctors who work specifically with people who have FXS and they can help develop the best treatment plan.

Early Intervention Services

Each state offers early intervention services for children from birth to 3 years old who qualify. Parents can ask for an evaluation, and the results of the evaluation drive the services that are provided.

Your Other Family Members

Also, if you have a child who is diagnosed with FXS, it means that it “runs” in your family. If you have other children, they could have the condition or they could “carry it.” You may want to take this into account for future children. It could also be in your extended family.

For Additional Information and Support:

- The National Fragile X Foundation -- Their website has a lot of information and they have parent groups – known as the Community Support Network – in most every state.
- There is a Fragile X clinic at Duke!
Introduction to Fragile X Disorders

Fragile X (FX) is often referred to as a group of genetic conditions known as Fragile X Disorders (FXDs).

Let’s look at the different FXDs:

- **Fragile X syndrome (FXS):** The most common *inherited* cause of intellectual disabilities, FXS occurs in both genders. Girls generally have less severe symptoms. FXS can cause developmental and language delays, learning difficulties, and behavioral and mental health issues. Individuals with FXS have a form of the Fragile X gene called a “full mutation.”

- **Fragile X-associated primary ovarian insufficiency (FXPOI):** A cause of infertility, early menopause, and other ovarian problems in women of reproductive age, FXPOI is seen in females who have a form of the Fragile X gene called a “premutation.”

- **Fragile X-associated tremor/ataxia syndrome (FXTAS):** This adult-onset (over age 50) neurological condition causes balance and memory problems, tremors and other neurological and psychiatric symptoms in people who have Fragile X premutations. It is more common in males than females.

**Other potential health issues for people who have Fragile X premutations:** Although people with Fragile X premutations are generally healthy, they may experience some medical issues more commonly than the general population. These problems include: high blood pressure, migraine headaches, depression, anxiety, and hypothyroidism. Researchers continue to study possible associations with these and other health issues in people with Fragile X premutations.

**If your child has been diagnosed as a FX premutation carrier:**

- He or she may be at risk for one of the above premutation carrier conditions, when they get older.
- He or she may be at risk to have a child with a full mutation of FX – called Fragile X syndrome.

**Your Other Family Members**

Also, if you have a child who is diagnosed as a FX premutation carrier, it means that it “runs” in your family. If you have other children, they could also be premutation carriers or there is a chance they could have the full mutation, OR they may not have either condition. Either way, you may want to take this into account for future children. It could also be in your extended family.

**For Additional Information and Support:**

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