To Whom It May Concern:

A member of your family has been identified as either having a condition called Fragile X syndrome (FXS) or being a carrier of a genetic change that could cause a Fragile X disorder (FXD). This means that other family members may also be carriers of this gene change and/or be at risk to have children with FXS. This letter has been sent to you to summarize information regarding the genetics of FXDs and Fragile X syndrome.

Fragile X syndrome is the most common form of inherited intellectual disabilities. It can occur in both males and females, though females often have milder features than males. Affected individuals can have intellectual disabilities, learning and behavioral disorders, and/or autism. Other characteristics include repetitive speech, hyperactivity, and poor eye contact. Physical characteristics include large ears, flexible joints, and a long face.

Fragile X is caused by a genetic change (mutation) in a gene called the FMR1 gene, which is on the X chromosome.

The Fragile X gene exists in several forms. An individual may carry a normal size gene, a “premutation” size gene, or a “full mutation” size gene. Both males and females can inherit this gene in either the premutation or full mutation form. Often individuals with a premutation are referred to as “premutation carriers.”

Individuals with FXS have a full mutation gene. Individuals with a premutation are usually not affected with FXS but are at risk to have children or grandchildren with a full mutation.

Females have two X chromosomes. Women who have inherited the Fragile X gene in either the full or premutation form are at risk to have a child with FXS.

Males have one X chromosome inherited from their mother and one Y chromosome inherited from their father. Males who have inherited a full mutation from their mother will have FXS. Males who inherit a premutation will pass it on to all their daughters and to none of their sons. In other words, all the daughters of male carriers will be carriers themselves.

Individuals who are premutation carriers are usually not affected, but they are susceptible to two other conditions:

* Fragile X-associated primary ovarian insufficiency (abbreviated FXPOI, pronounced “fax-poy”)—Seen in some women with the premutation, it results in early menopause and/or reduced fertility.
* Fragile X-associated tremor/ataxia syndrome (abbreviated FXTAS, pronounced “fax-tas”)—A late onset (over age 50) neurological condition, sometimes initially diagnosed as Parkinson’s disease, that is more common in males than females.

Therefore, even if you are not in your childbearing years, the possibility of being a Fragile X gene carrier has implications for your health, your children’s, or your grandchildren’s health.

A DNA test for FXDs, called the “FMR1 DNA test,” has been available since 1991. This test is extremely reliable in detecting the premutation or full mutation of Fragile X. Since a member of your family has been identified as carrying this gene change, you, and your children (if you have any), may also carry this changed gene. It is therefore recommended that you and/or your children contact a genetic counselor to discuss your family history and the advisability of genetic testing. Your physician or other health care provider can order this testing on your behalf.

You have several options should you wish to discuss the contents of this letter:

* You can locate a genetic counselor in your geographical area at https://findageneticcounselor.nsgc.org/.
* You can find additional information on the National Fragile X Foundation website by reviewing the section for “Newly Diagnosed” under the “Living with Fragile X” heading on the main menu at https://fragilex.org.

We hope this information is of help to you. Please feel free to use any or all of the resources described above, and should you have any further questions, you can contact the National Fragile X Foundation by phone at 800-688-8765 or by email: treatment@fragilex.org.

Sincerely,

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