The Fragile X Treatment Research Program
At Vanderbilt University Medical Center

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What is Fragile X Syndrome?

Fragile X Syndrome is the most common inherited cause of intellectual disability. Fragile X is caused by a defect in the *FMR1* gene that can cause symptoms such as intellectual disability, anxiety, unstable mood, behavioral problems and hyperactivity. Boys are typically more affected than girls and a diagnosis is made through DNA testing.

mGluR5 Theory of Fragile X Syndrome

Recent research has focused on the “mGluR5 Theory of Fragile X Syndrome.” Research in animal models shows too much mGluR5 receptor signaling in the brain. Improvement in brain function and behavior is seen when this signaling is decreased, either by manipulating the gene or by using medication to block the receptor. Medications that block the mGluR5 receptor are now being tested in individuals with Fragile X Syndrome in hopes that the same benefits will be seen.

GABA-B Theory of Fragile X Syndrome

Other research shows abnormalities in the GABA system in animal models of Fragile X Syndrome. Improvements are seen when a particular receptor, GABA-B, is stimulated. Medications that stimulate the GABA-B receptor have been used to treat individuals with nerve and muscle problems and are now being evaluated to treat symptoms of Fragile X Syndrome.

Reference:

There is no cure for Fragile X Syndrome but current research is looking to improve treatment options for affected individuals and their families.

Contact the Fragile X Treatment Research Program for more information regarding current research in Fragile X Syndrome.

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