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Perspective

The Genetic Disorder: Fragile X Syndrome

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DESCRIPTION

The increase of CGG nucleotide repeats in the FMR1 gene near the end of the long arms of the X chromosome causes Fragile X Syndrome (FXS). Researchers culture cells in folic acid-deficient medium to find the mutation, which causes the ends of the X chromosome to look to be about to split off. This was the only way to see the mutation before molecular testing. Martin and Bell first characterised Fragile X Syndrome, also known as Martin-Bell Syndrome, as a kind of Intellectual Disability (ID) with an X-linked inheritance pattern. The Fragile X Mental Retardation Protein (FMRP) is encoded by the FMR1 gene and regulates gene expression and protein translation in the brain. FMRP modulates the expression of critical molecules involved in receptor signalling and spine formation by regulating mRNA metabolism in the brain.

FXS is caused by a mutation in the FMR1 gene on chromosome X. One of two types of sex chromosomes is the X chromosome. The Y chromosome is the other. Men have one X chromosome and one Y chromosome, while women have two X chromosomes. The FMR1 gene has a flaw, or mutation, that prevents it from producing the fragile X mental retardation 1 protein appropriately. This protein is involved in the nervous system's operation. The protein's function isn't completely understood. The symptoms of FXS are caused by a loss or shortage of this protein. Fragile X syndrome is diagnosed using DNA from blood, amniotic fluid, or other tissues. Your healthcare provider will send the sample to a lab to see if your child carries the FMR1 gene. If you're pregnant and think your child might have fragile X syndrome, you can visit a genetic counsellor and have the following prenatal testing done:

Amniocentesis is a procedure in which a healthcare provider extracts a sample of amniotic fluid for examination.

Chorionic villus sampling is a procedure where a sample of cells from the placenta is taken for testing by the healthcare provider.

In cells that mature into eggs, the FMR1 gene premutation on the X chromosome in women can grow to more than 200 CGG repeats. This means that women who have the premutation are more likely to have a kid who has fragile X syndrome. The premutation in men, on the other hand, does not extend to more than 200 repeats when it is passed down through the generations. Only their daughters inherit the premutation from their fathers. Their sons are born with a Y chromosome that lacks the FMR1 gene.

CONCLUSION

The goal of treatment is to help people with the disease gain important language and social skills. Extra assistance from teachers, therapists, family members, doctors, and coaches may be required. While there is no cure for FXS, knowledge of FMRP activity has cleared the way for reasonable treatment designs that could potentially reverse many of the neurobiological abnormalities seen in the disease. Epilepsy is a common comorbidity in FXS. FXS has a wide differential diagnosis that includes both syndromic and non-syndromic psychomotor delays/ID. Sotos Syndrome, Prader-Willi Syndrome, Klinefelter Syndrome, and FRAXE are among the possible diagnoses.

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