



Fragile X Syndrome

Fragile X Syndrome is the most common form of inherited mental retardation and accounts for approximately 40% of cases with X-linked mental retardation. The gene responsible for Fragile X Syndrome is located on the X chromosome and is called FMR1. If a full mutation occurs in this gene, women may have learning disabilities and hyperactivity, but normal fertility. If a partial mutation, called a premutation, occurs, infertility and premature ovarian failure (early menopause) may result in 20% of women. Approximately 1 in 250 women are carriers of the fragile X premutation. Many carriers of the fragile X premutation have no known family history of fragile X.

It is recommended that any person with unexplained mental retardation, developmental delay or autism be tested. The American College of Medical Genetics also recommends carrier testing on the basis of family history of unexplained mental retardation. It is not currently recommended to test all women who are pregnant. For more information see: <http://www.fragilex.org>, or http://www.cdec.gov/genomics/hugenet/facesheets/FS_FragileX.htm