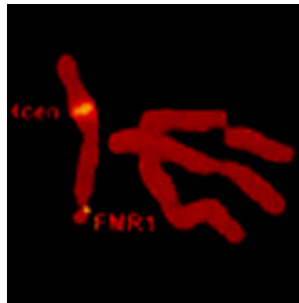




## Fragile X syndrome



An unstable nucleotide repeat is associated with the most common form of mental retardation known as Fragile X syndrome.  
[Image credit: Steve Warren, Emory University School of Medicine, Atlanta, GA, USA.]

Fragile X syndrome is the most common inherited form of mental retardation currently known. Fragile X syndrome is a defect in the X chromosome and its effects are seen more frequently, and with greater severity, in males than females.

In normal individuals, the FMR1 gene is transmitted stably from parent to child. However, in Fragile X individuals, there is a mutation in one end of the gene (the 5' untranslated region), consisting of an amplification of a CGG repeat. Patients with fragile X syndrome have 200 or more copies of the CGG motif. The huge expansion of this repeat means that the FMR1 gene is not expressed, so no FMR1 protein is made. Although the exact function of FMR1 protein in the cell is unclear, it is known that it binds RNA.

A similar nucleotide repeat expansion is seen in other diseases, such as Huntington disease. Research in mice has proven helpful in elucidating some of the mechanisms that cause the instability of this gene. Our methods for identifying carriers of Fragile X syndrome have also improved, and further research will help people carrying "premutations" to avoid having children who have a larger expansion (i.e. more CGG repeats) in FMR1, and therefore suffer from Fragile X syndrome.

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