

History and Prevalence of Fragile X Syndrome

Prevalence

The prevalence of fragile X syndrome differs between males and females, with estimates of 1 in 4000 for males and 1 in 8000 for females.

History

James Purdon Martin and Julia Bell first described fragile X syndrome in 1943. They reported a family case study in which intellectual disability appeared to be inherited and linked to the X chromosome.

In 1969, Herbert Lubs made the observation of a characteristic fragile site on the lower end of the X chromosome.

The FMR1 gene on the X chromosome that is associated with fragile X syndrome was identified and sequenced in 1991. To read more about the genetics of fragile X syndrome, [click here](#).