

Prevalence of Fragile X Revisited and Revised

In the Winter 2000 Foundation Quarterly, Dr Randi Hagerman addressed the question of the prevalence of fragile X syndrome across the entire population. In our editor's note, we affirmed our commitment to review new data and adjust our published statistics as appropriate. Based on a recent report from the U.S. Center for Disease Control (CDC) we here revise our prevalence statement. Our year 2000 article cited prevalence of the fragile X, full mutation in boys as: 1:2000 and in girls as: 1:4000. These numbers were meant to include not only those with mental retardation, but also those with learning disabilities and emotional problems *without* mental retardation.

Based on the recent CDC report, which is itself a review of existing prevalence studies, the National Fragile X Foundation will henceforth publish a prevalence for males, with the full mutation, of 1:3600. The CDC did review *some* studies which did include some individuals with learning disabilities in addition to those with mental retardation.

Prevalence in females is less well understood and certain because of a lack of studies. However, it is likely that at least 1:4000 to 1:6000 females has the full mutation for fragile X - a figure higher (more prevalent) than the CDC estimate. We propose this adjustment because girls with fragile X often present with more mild involvement than boys, including learning disabilities and emotional problems, and thus may not be screened or tested.

Whatever the prevalence of fragile X, it remains the most common known cause of inherited mental impairment. Some of the uncertainties and differences of opinion stem from differing definitions of just who is "affected." Whether just those with the full mutation are affected, and to what degree those diagnosed with learning disabilities and/or emotional problems have fragile X, are two such questions which can impact prevalence. Differences in defining the cut-off point between pre-mutation carriers and those with a full mutation also cloud the issue. Most importantly, a lack of large-scale, comprehensive studies allows for wide-ranging differences of opinion. While a greater degree of certainty regarding prevalence would be desirable, and important in the quest for increased government-funded research, what is most important is that fragile X syndrome:

is prevalent,
has a significant impact on society,
is relatively unknown,
is under or largely undiagnosed,
is a key to understanding other neurodevelopmental disorders such as autism,
and, as such, needs our full attention - no matter how prevalent.

To view the CDC report, visit http://www.cdc.gov/genetics/hugenet/factsheets/FS_FragileX.htm.