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FROM PHENOTYPES TO FAMILIES: FINDINGS FROM THE NATIONAL (U.S.) FRAGILE X FAMILY SURVEY

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Most studies of individuals with fragile X (FX) necessarily rely on small samples, due to the cost associated with locating and assessing individuals who live in diverse locations. Large-scale studies are needed, however, to answer important questions about the nature and consequences of a condition, especially when the effects of a condition can be variable. This presentation summarizes the methodology and findings of national survey of parents of children with FX in the U.S. A total of 1,276 families completed study enrollment. The families had a total of 2,964 children. Of the children who had been tested for FX, parents reported that 1,167 males and 323 females had the full mutation; 65 males and 211 females were premutation carriers. Families were able to complete the survey on-line or through a telephone interview. The survey covered a wide range of topics, including phenotypic characteristics, life circumstances, and family adaptation. This presentation highlights major findings with regard to age of diagnosis, co-occurring conditions and other phenotypic consequences (e.g., obesity, sleep problems, self-injury, seizures), functional skills, work and living environments, quality of life, and family adaptation. Although all data are parent report, the approach was cost-effective and could be a model for collecting certain types of data for other X-linked conditions. The results provide insights into the nature and consequences of FX, and suggest hypotheses that need to be tested through more controlled studies.

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