

Overview of the National Fragile X Survey

December 2009

This report provides an overview of the National Fragile X Survey, including why and how the survey was conducted, a description of the families and children who participated, and what has been learned during the 3-year project.



Why Was the National Fragile X Survey Conducted?

Conducted between August 2008 and July 2009, the goal of the National Fragile X Survey was to better understand how having a child with fragile X syndrome affects family life. Although other studies have been conducted on children with fragile X syndrome and their families, these were limited to small samples. In addition, studies often focused on a specific topic, such as how fragile X syndrome affects language development. The National Fragile X Survey was designed to address these two issues.

The national survey format was an effective method to reach a much larger number of people than was previously possible. In addition, the survey was able to ask questions on a broad array of topics and items, which were selected because of their relevance to clinical practice and policy.

The survey asked specifically about families and their children with fragile X syndrome. Family-level topics included questions about the diagnostic process, knowledge of fragile X syndrome, types of support and needs, family history of fragile X associated tremor/ataxia and fragile X associated primary ovarian insufficiency, costs associated with raising a child with fragile X syndrome, and insurance information.

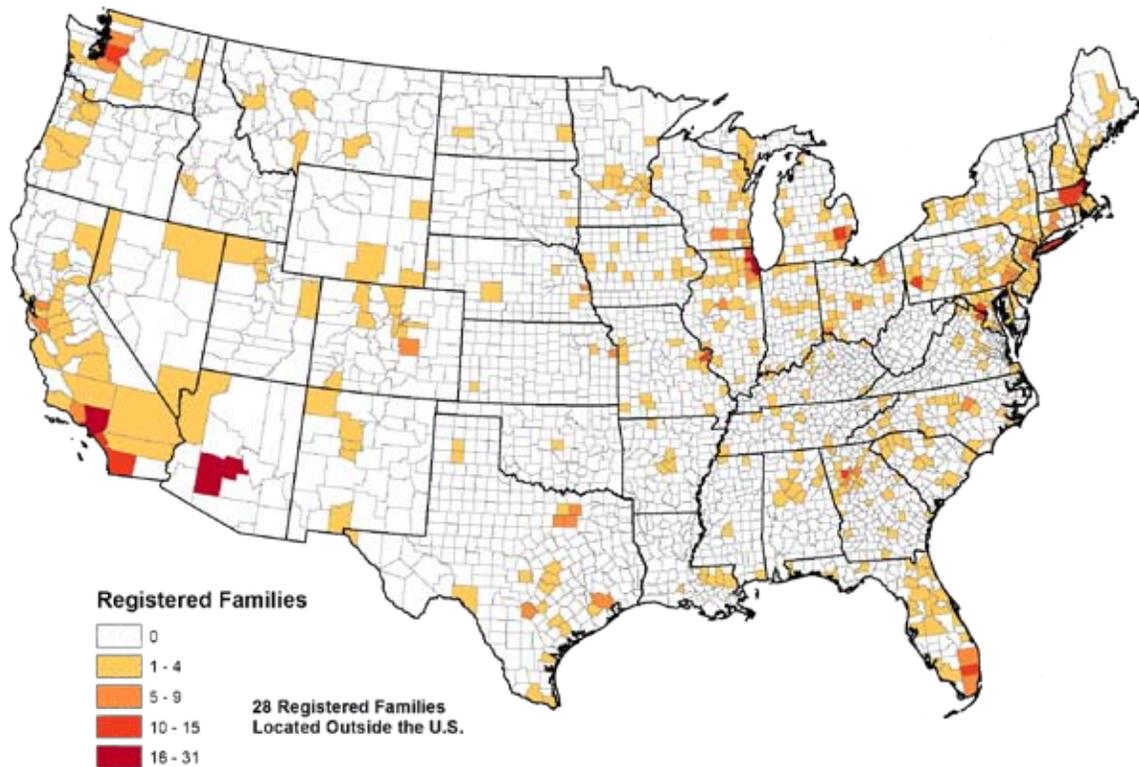
The survey also asked about the children with fragile X syndrome, both carriers and those with the full mutation. Families provided information on children's nutrition and physical activity, functional skills, sleep, seizures, self-injury, sensitivity to pain, medication use, education and schooling, services, and adult-specific topics, such as employment and living arrangement.

Who Participated in the National Fragile X Survey?

To maximize the number of families who could participate, the survey was designed to be completed on the Internet or over the telephone. The majority of families (80%) completed the survey online. This proved to be an effective method to reach a large number of families in a short period of time. A total of 1,124 families completed the National Fragile X Survey. This also allowed families across the United States to participate, including 23% of families who lived in the Northeast, 28% from the South, 30% from the Midwest, and 17% from the West Coast. *Figure 1* shows the geographical location and the number of families who registered for the survey. In addition, a small number of families (2%) from Canada, France, the United Kingdom, Switzerland, Puerto Rico, and Bermuda participated in the survey.

The majority of families that completed the survey are Caucasian (92%). Only a small number of families are African American (2%), Hispanic (5%), or from another racial or ethnic background (1%). Most participants indicated they were married (85%).

Figure 1. Geographical location of families that participated in the National Fragile X Survey



Overall, families reported a range of incomes. Just over half of the families reported an annual income over \$75,000, with 19% of families having incomes between \$75,000 and \$100,000 and 36% having incomes over \$100,000. Only a small number of families (6%) reported an annual income of less than \$25,000. Compared with the U.S. population, the median annual income was higher for families who participated in the survey.

Combined, more than half of the survey participants had a four-year college degree (32%) or a graduate or professional degree (25%). Another 18% had attended college but did not have a degree and 10% had a high school diploma or GED.

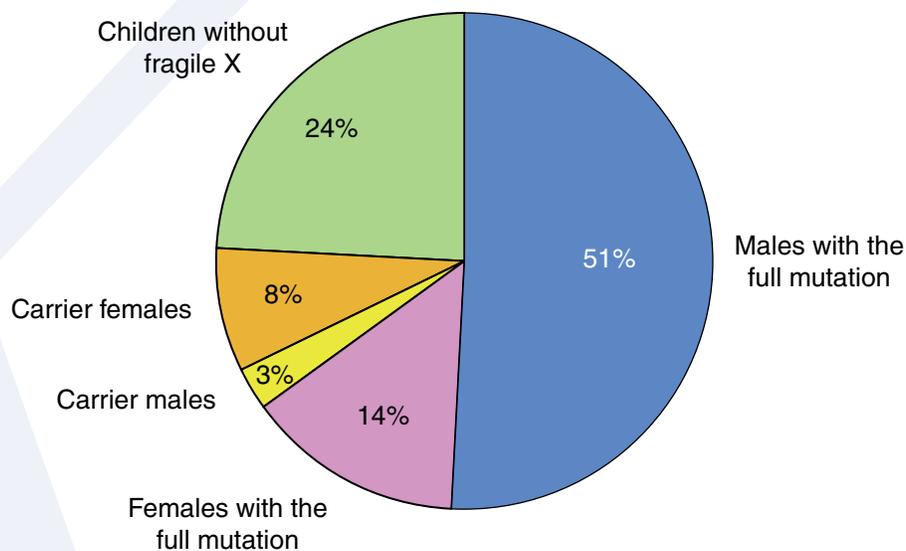
This group of families is not representative of U.S. families in terms of racial/ethnic distribution, income, or education. Thus, it was necessary to examine the effects of these variables in subsequent analyses.

What Do We Know about the Children?

The families that participated in the survey had a total of 2,672 children: 1,674 are male and 998 are female. Approximately 81% (2,154 children) had been tested for fragile X syndrome. **Figure 2** shows the genetic status of tested children. Some notable findings include the following:

- Just over half of the children are males with the full mutation and 14% are females with the full mutation.
- Only 3% of the children are male carriers, but 8% are female carriers.
- About a quarter of the children tested did not have fragile X syndrome.

Figure 2. Genetic status of tested children in the National Fragile X Survey (n = 2,154)



One noteworthy characteristic is the age range of the “children” in the study. The youngest child was less than a year old and the oldest was 65 years old.

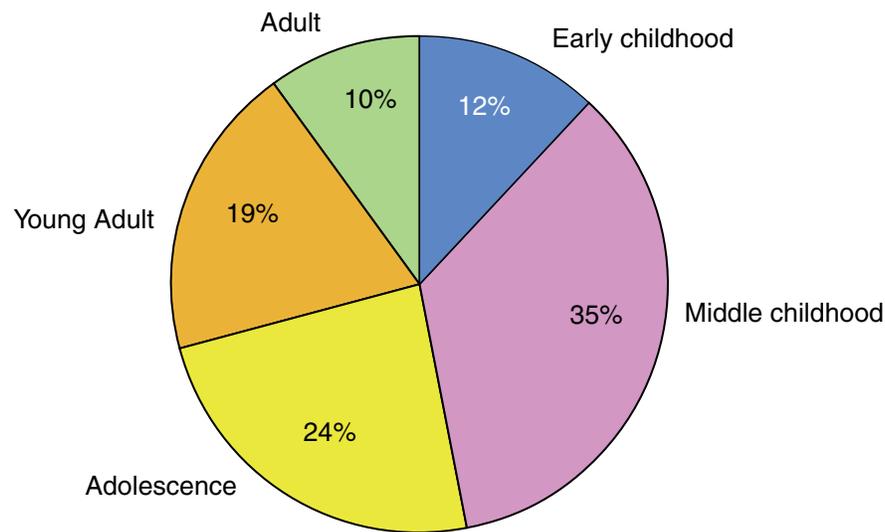
Among males, 1,090 have the full mutation. Males were divided into five age groups:

- Early childhood (birth to 4 years)
- Middle childhood (5 to 11 years)
- Adolescence (12 to 18 years)
- Young adult (19 to 30 years)
- Adult (over 30 years)

The percentage of males with the full mutation in each age group is shown in **Figure 3**. The largest group of males with the full mutation is in middle childhood. Almost one third of the children were either young adults or adults.

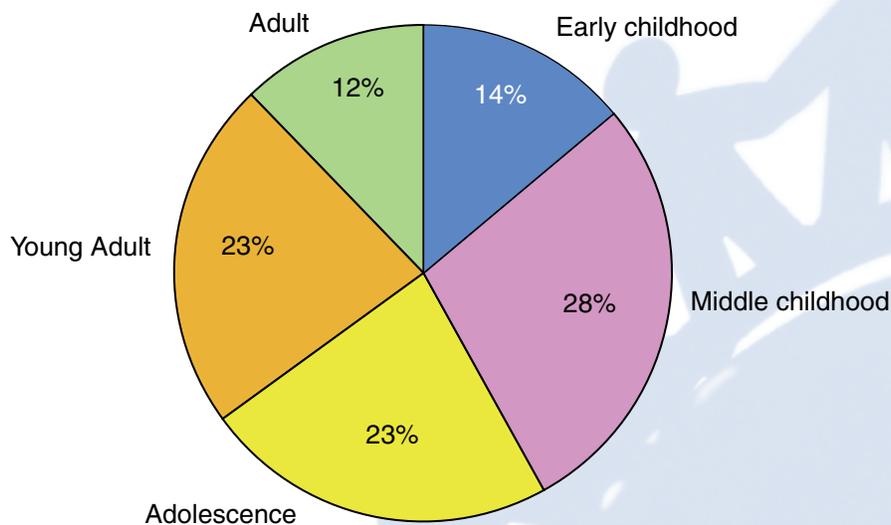
Figure 4 shows the percentage of females with the full mutation, 304 in total, in the corresponding age groups. Similar to males, the largest group of females with the full mutation is in middle childhood. Slightly more young adult or adult females than their male counterparts participated in the survey.

Figure 3. Age of males with the full mutation in the National Fragile X Survey



Note: Early childhood=birth to 4 years, Middle childhood=5 to 11 years, Adolescence=12 to 18 years, Young adult=19 to 30 years, Adult=over 30 years.

Figure 4. Age of females with the full mutation in the National Fragile X Survey



Note: Early childhood=birth to 4 years, Middle childhood=5 to 11 years, Adolescence=12 to 18 years, Young adult=19 to 30 years, Adult=over 30 years.

What Did We Learn from the National Fragile X Survey?

Overall, the National Fragile X Survey was very successful. First, it yielded the largest sample of families who have a child with fragile X syndrome to ever participate in a research study. Using both the Internet and a toll-free number allowed many people to participate in a short amount of time. Second, a great deal of rich information was collected about what life is like for both parents and children. This will help researchers, clinicians, and policy makers to better serve families who are affected by fragile X.

Despite this success, there were some challenges. One challenge involved enrollment in the survey. Once families started enrolling, it became clear that the requirements for participating in the survey needed to be described in more detail. For example, frequently both a grandmother and a mother wanted to enroll the same child with fragile X syndrome. Because many different types of families were encouraged to participate, it was decided that the grandmother should enroll her own daughter, who was a fragile X carrier, and the mother should enroll the child. Because of confidentiality requirements, it was not possible to link the two families together to look at fragile X across multiple generations in a single family. However, this would be interesting to do in a future study.

Another challenge was the complexity and amount of information collected from each family. In many parts of the survey, families were asked to report on each of their children with fragile X syndrome, causing them to loop through many items. This may have imposed a burden on some families who had multiple children who are affected by fragile X.

However, the survey yielded a rich data set that sheds light on many important questions about how fragile X affects family life. This information will be used to help inform future research, best clinical practices, and policy; and most importantly, to make a difference in the lives of families with children with fragile X syndrome.

Acknowledgments

The National Fragile X Survey project team would like to thank all of the families who participated in the survey. Because of you, researchers now have a better understanding of how fragile X affects individuals and their families.

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For more information

Please visit the National Fragile X Foundation Web site at http://www.fragilex.org/html/fx_survey.htm

Questions?

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* RTI International is a trade name of Research Triangle Institute.