

# Getting the Diagnosis of Fragile X Syndrome

## Findings from the National Fragile X Survey

August 2009

This report summarizes information about how families found out their child has fragile X syndrome. The findings show that when families first express concerns about their child, they are often told to “wait and see” if development improves . It often takes between 3 and 5 visits to a specialist before a test for fragile X syndrome is ordered. As a result, the average age of diagnosis has remained the same over the past 7 years.



# How do families find out their child has fragile X syndrome?

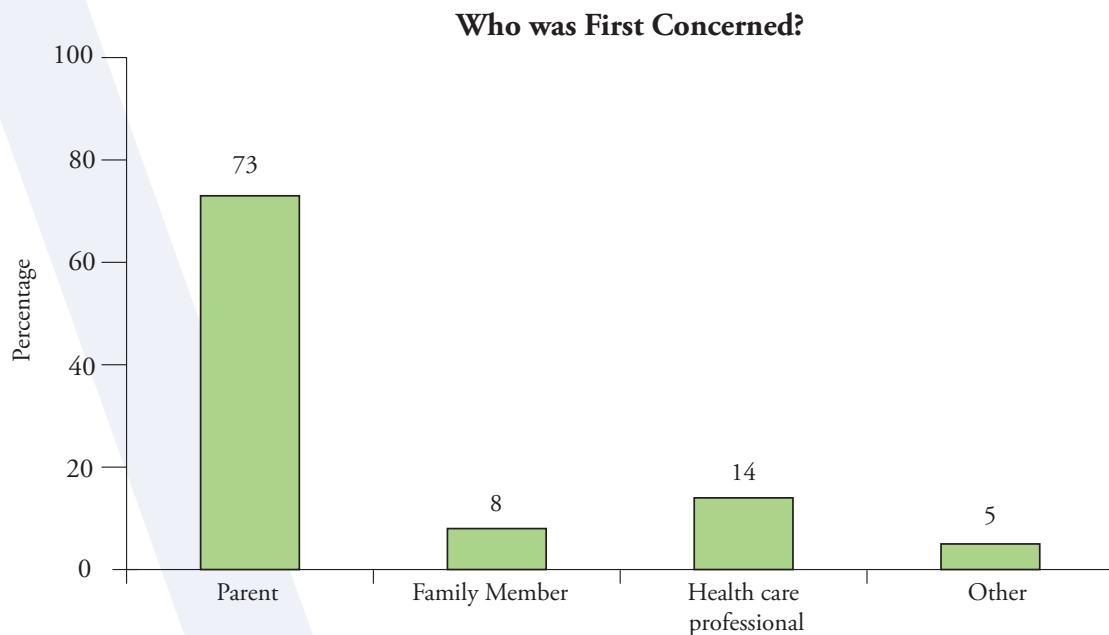
## Overview

This report provides information about how families find out their child has fragile X syndrome. Previous research has highlighted the need for earlier diagnosis. Recent efforts by the national fragile X organizations as well as medical associations have encouraged doctors to screen for developmental delays during well child visits and test for fragile X if there are any unexplained delays. The results in this report will help to highlight the status of these efforts.

These data are based on 249 families who had their first child diagnosed in 2001 or later and answered questions about:

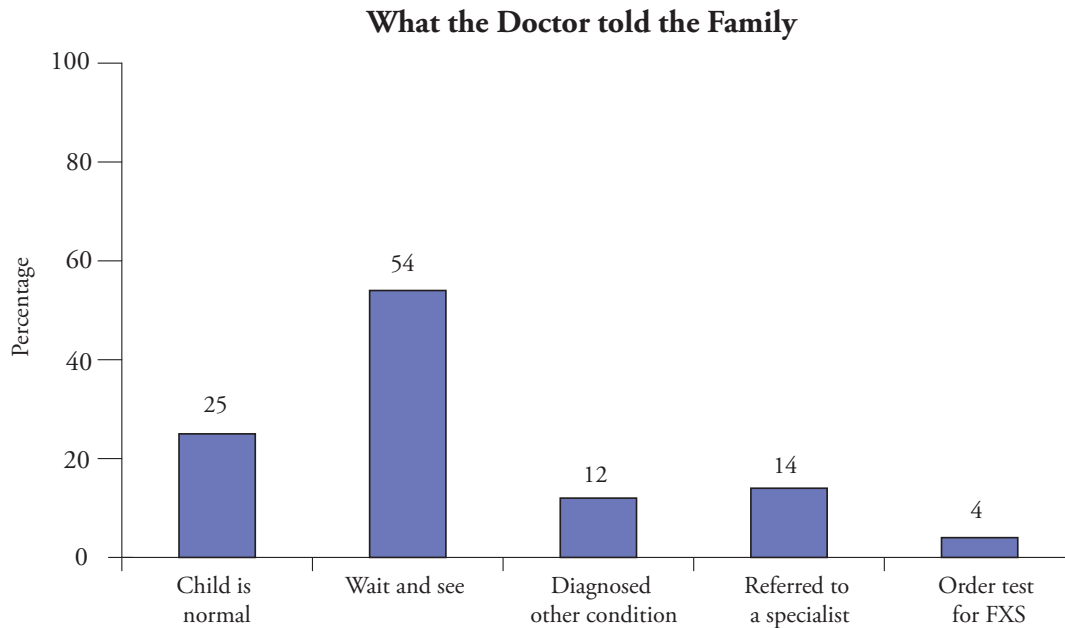
- Who was first concerned about their child's development or behavior?
- What the doctor told them when they were concerned?
- How many visits they made to a doctor before the test for fragile X was ordered?
- Who recommended the test for fragile X?
- Whether the diagnosis affected their decision to have additional children?

The first chart shows information about who was first concerned about the child's development or behavior.

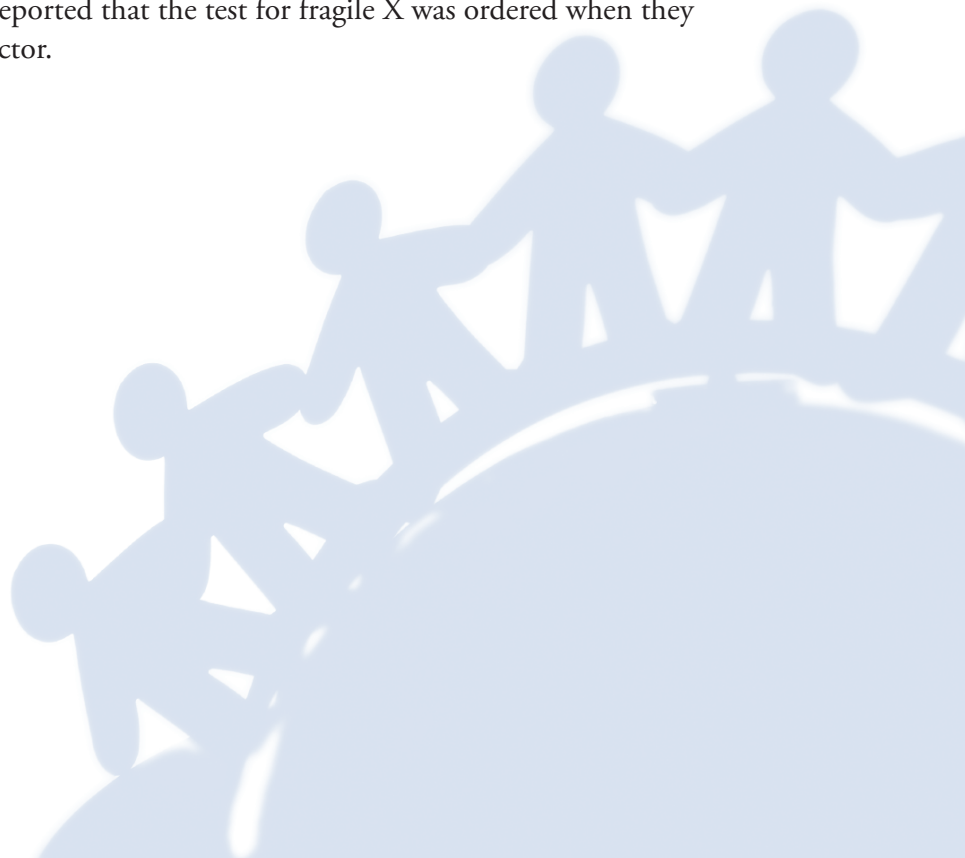


- Most of the time parents were the first to express concerns about the child's development of behavior. Most families (81%) reported they were concerned because the child was behind in development. A small number reported behavior problems (3%) or lack of eye contact and difficulty interacting with others (5%).

The next chart provides information about what the doctor or health care professional told the family when they first expressed concern about their child's development or behavior.

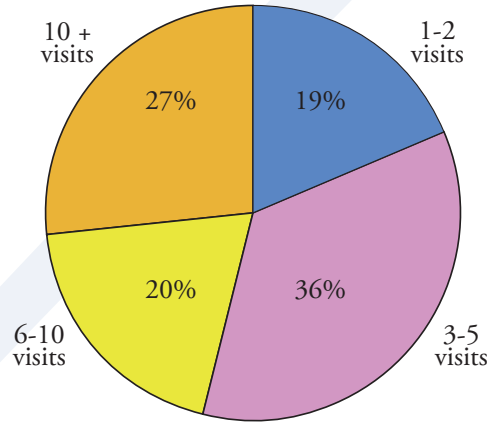


- Most often, a doctor or health care professional told the family to either “wait and see” if the child’s development improves or that the child’s development was normal and there was no need for concern.
- Only a small number of families reported that the test for fragile X was ordered when they first expressed concern to their doctor.



The chart below presents information on how many visits families had to make to a doctor before a test for fragile X was ordered.

**Number of Visits to Doctor**

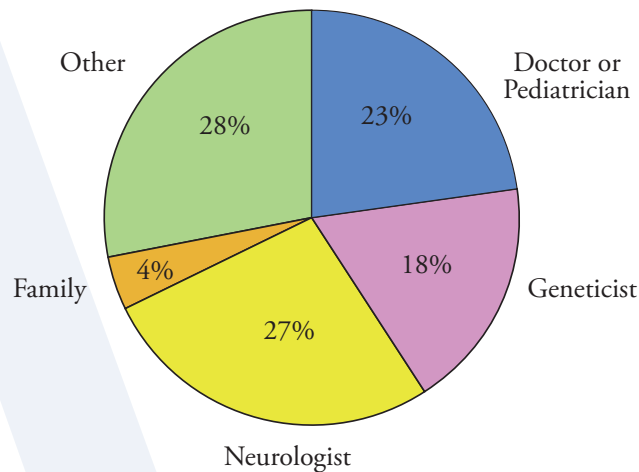


- Most families reported that it took between 3 and 5 visits to a doctor before a test for fragile X was ordered. However, a large number of families required more than 10 visits.

The chart below shows who first recommended testing for fragile X.

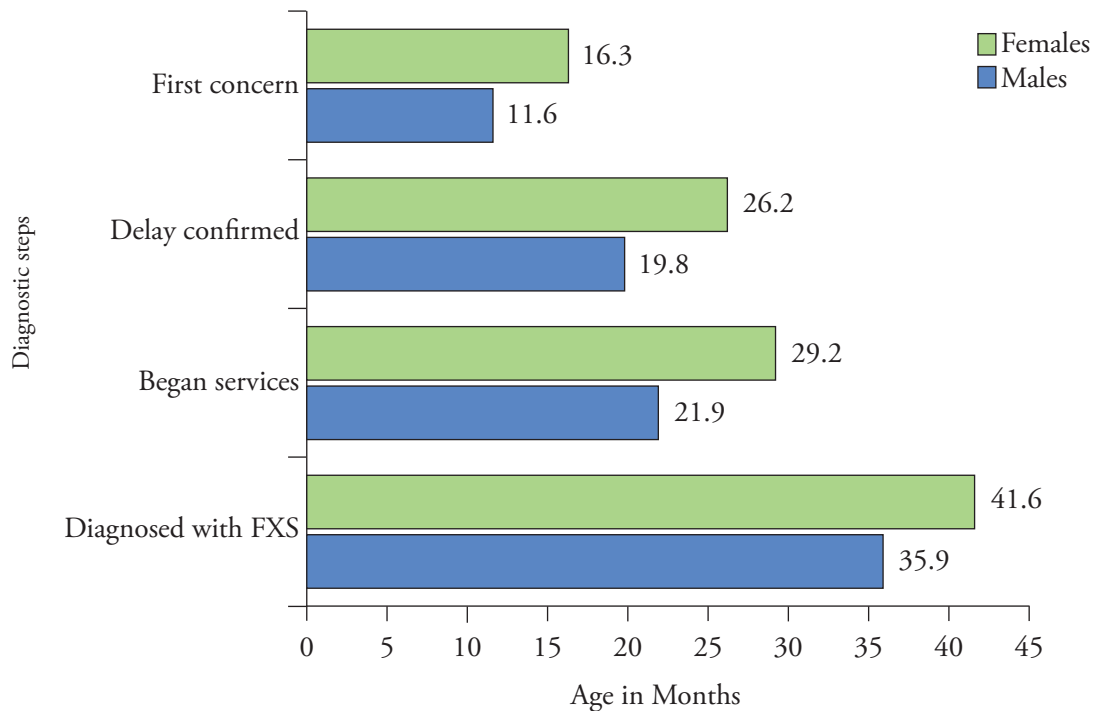
- Testing was most often ordered by a neurologist, family doctor or pediatrician, or geneticist. A small number of families requested the test themselves.

**Who First Recommended the Test for Fragile X**

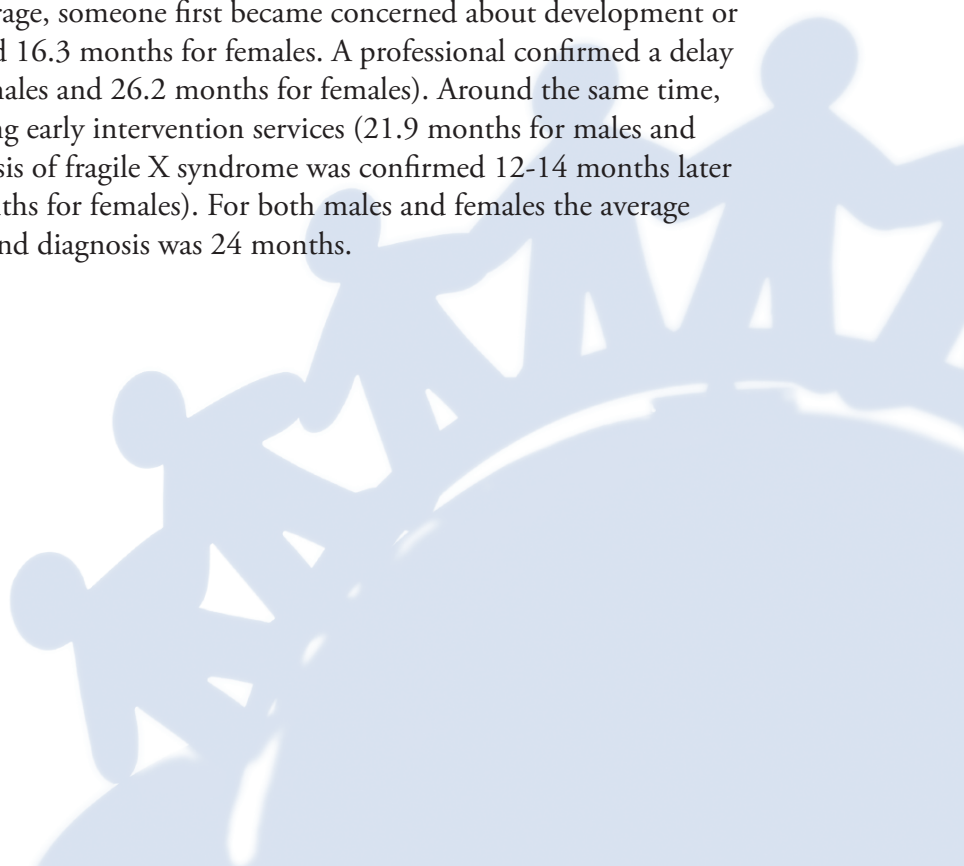


The next chart shows the average age of each step in the diagnostic process for males and females.

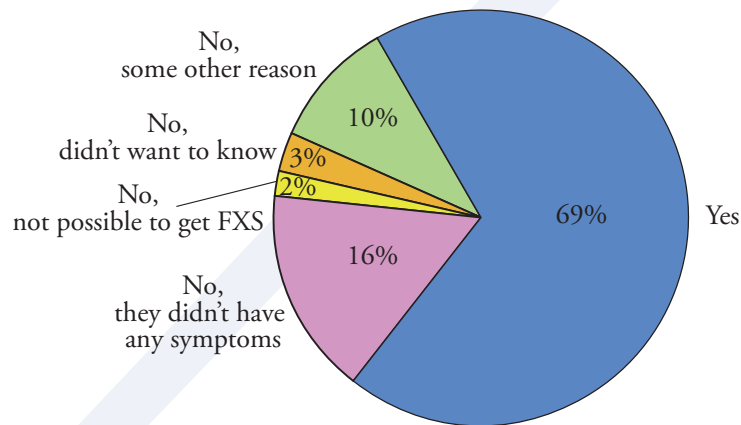
### Average Ages of Steps in Diagnosis Process for Males and Females



Overall, the diagnostic process for males with the full mutation of fragile X syndrome started earlier than it did for females. On average, someone first became concerned about development or behavior at 11.6 months for males and 16.3 months for females. A professional confirmed a delay 8-10 months later (19.8 months for males and 26.2 months for females). Around the same time, children were eligible to begin receiving early intervention services (21.9 months for males and 29.2 months for females). The diagnosis of fragile X syndrome was confirmed 12-14 months later (35.9 months for males and 41.6 months for females). For both males and females the average length of time between first concern and diagnosis was 24 months.



The last chart shows whether families decided to test other children in their family after the initial diagnosis of fragile X syndrome.



- Most families (69%) had tested all of their children for FXS. About half the families who did not have all their children tested said they decided not to because they did not have any symptoms. A handful of families indicated that they could not genetically inherit FXS or said they did not want to know the status yet. The remainder gave a wide range of other reasons.
- The majority of respondents (76%) said the diagnosis affected reproductive decisions. Almost all parents who said the diagnosis did not affect reproductive decisions reported that by the time they had received the diagnosis, they had already decided not to have additional children.
- Approximately 25% of the families of male children had a second child with the full mutation prior to the diagnosis of the first child; 39% of the families of female children had a second child with the full mutation prior to the diagnosis.

## Summary of Results

In most cases, a parent or family member first expressed concern about their child's development. Doctor's often told families to either "wait and see" if development improves or that there was no need for concern. Most frequently, a test for fragile X was ordered by a neurologist after 3 to 5 visits. When compared to earlier reports on the diagnostic process, children with fragile X syndrome are identified earlier and enter intervention at younger ages when compared to data from 2001. However, there were no changes in the average age of diagnosis despite recent efforts by professional organizations and advocacy groups. Delayed identification has important implications for families, many of whom have additional children before finding out about fragile X syndrome.

## Acknowledgement

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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](http://www.fragilex.org/html/fx_survey.htm)

## Suggested citation

Raspa, M., Bailey, D. B., & Bishop, E. (2008, August). Getting the diagnosis of fragile X syndrome. Research Triangle Park, NC: RTI International. Available at <https://fragileX.rti.org/USresults>

These results also have been published in *Pediatrics*, available at: <http://www.pediatrics.org>

Reference: D. B. Bailey, M. Raspa, E. Bishop, & D. Holiday (2009). No change in the age of diagnosis for fragile X syndrome: Findings from a national parent survey. *Pediatrics*, 124, 527-533.

## Questions?

Please contact Melissa Raspa at RTI International ([mraspa@rti.org](mailto:mraspa@rti.org)).

