Early Check
Expanded health screening for your baby

Fragile X Syndrome Fact Sheet for Families

Fragile X syndrome (FXS) is the most common cause of inherited intellectual disability. The exact number of people who have FXS is not known, but scientists think that 1 out of every 3,600 to 4,000 boys and 1 out of every 4,000 to 6,000 girls have fragile X syndrome.

What problems does FXS cause?

- Children with FXS might learn new skills--like walking and talking--later than children who don't have FXS.
- They often have learning disabilities, anxiety, hyperactivity and a short attention span. Some also have autism and may have seizures.
- Children with FXS may also develop physical differences, such as a longer appearing face, large ears and loose connective tissue. Loose connective tissue can lead to ear infections, hernias, and less stable joints.
- Although there is no cure for FXS, getting treatments through early intervention can help.

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How to get early intervention

- Each state offers early intervention services for children from birth to 3 years old who qualify. In North Carolina, the CDSA (Children's Developmental Services Agency https://beearly.nc.gov/) evaluates children and provides therapies.

- Parents or healthcare providers can ask for an evaluation, and the results of the evaluation determine what services are provided (https://beearly.nc.gov/index.php/providers/eligibility-referral)

Why should you consider testing your baby?

- Once a child is diagnosed with FXS, parents can begin early intervention services to help with things like learning to walk and talk. These services can also help parents learn more about FXS so that they can understand and help their child.
- Although there is not a cure, doctors who work specifically with people who have FXS can develop the best treatment plan.

Who should be tested?

Fragile X can be tested for on a small sample of blood or saliva. Testing before birth (prenatal diagnosis) is available and accurate.

- Mothers of children with fragile X syndrome should be offered testing.
- Testing should be offered for children, brothers, sisters, and more distant family members of someone with FXPM or FXS. Testing grandparents can guide testing for distant family members.
- Testing should be offered for blood relatives with problems with development, learning, or behavior.
- Grandfathers of children with FXS should consider testing, especially if they have trembling hands, difficulty walking, or changes in mood, or difficulty thinking.

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- Blood relatives who are pregnant or considering pregnancy should speak with their doctor or a genetic counselor to learn more about fragile X, testing, and options for planning a family.

Why should parents and other blood relatives consider testing?

Fragile X runs in families and is passed down from one generation to the next.

- If your child is diagnosed with FXS, then fragile X runs in your family even if nobody else in the family has been diagnosed with FXS or FXPM. Usually FXS is passed from mothers who are healthy "carriers" of fragile X premutation (FXPM).
- For parents who have a child with FXS, there is a 50% chance (50:50) for FXS or FXPM in each pregnancy. Boys who inherit FSX will have fragile X syndrome. Girls who inherit FXS can have no features of fragile X syndrome, milder features, or all the features of fragile X syndrome.
- Knowing about these possibilities can be important for parents and other family members when deciding to have baby.
- Share information about fragile X with your blood relatives by printing the Letter for Family Members available when you log into your Early Check page.

What about genetics?

- To learn more about the gene involved in fragile X, see the Fragile X Genetics Fact Sheet when you log into Early Check. Also visit the National Fragile X Foundation genetics page: https://fragilex.org/understanding-fragile-x/fragile-x-101/genetics-inheritance/

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Reliable Information and Support

- You can learn more about fragile X syndrome and fragile X premutation at the **National Fragile X Foundation** website: https://fragilex.org/. The Foundation has parent groups – known as the Community Support Network – in most every state.
- For more information about fragile X premutation, **see the Early Check FXPM Fact Sheet for Families** when you log into Early Check.
- For general information about fragile X, visit the **Center for Disease Control** Fragile X webpage: https://www.cdc.gov/ncbddd/fxs/index.html
- Fragile X clinics specialize in caring for people with fragile X syndrome:
    - **Duke University Medical Center** in Durham North Carolina: https://fragilex.org/duke-university-medical-center/
    - **Greenwood Genetics** in various South Carolina locations: https://www.ggc.org/fragilex

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