Early Check
Expanded health screening for your baby

Fragile X Premutation Fact Sheet for Families

About Fragile X Premutation (FXPM)

- **FXPM is common in people around the world.**
- **Nearly 1 out of every 200 women and 1 out of every 400-500 men has FXPM.**
- **Most people with FXPM do not know they have it and do not notice any health effects.**

How does FXPM affect health?

Most people with fragile X premutation do not have related health problems, but FXPM can affect the health and well-being of some adults and even fewer children.

- **In Children:** Most children have no effects at all or mild effects of FXPM. Some children may have a shorter attention span, hyperactivity (ADHD), and a tendency to worry or to be shy in social situations, or have difficulties communicating.
    - Scientists and doctors do not yet fully understand the effects of FXPM on health and development in children, but Early Check researchers are looking for signs of these effects in babies and young children in order to know when early treatments or therapies could be most helpful.

- **In Men:** Later in life, some men with FXPM develop a nerve condition that effects movement, mood, and thinking. This is called FXTAS (*Fragile X-Associated Tremor/Ataxia Syndrome*). For more information about FXTAS, see the National Fragile X Foundation FXTAS

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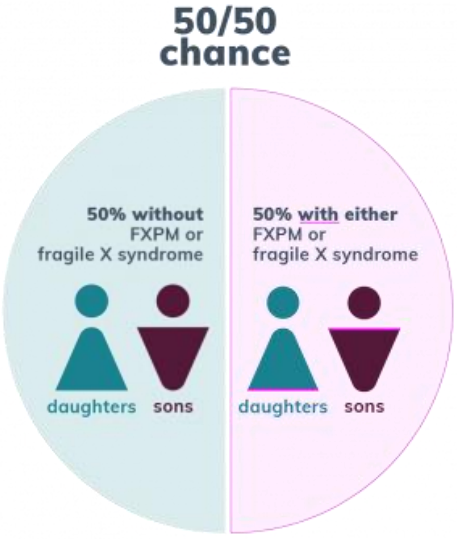
- **In Women:** Some women with fragile X premutation have irregular periods, difficulty getting pregnant and can have menopause earlier than usual. This condition is called FXPOI (*Fragile X Premature Ovarian Insufficiency*). For more information, see the National Fragile X Foundation FXPOI.
    - Later in life, some women also develop FXTAS, though it is usually milder for women than men.
    - Some women with FXPM may experience anxiety and depression, and there is treatment that can help.

Can it affect other family members?

- Fragile X runs in families, so parents, brothers, sisters, and aunts, uncles, cousins, and grandparents may also have FXPM or fragile X syndrome.
- Fragile X syndrome (FXS) is a serious condition that causes problems with thinking, learning, communication, and behavior.
- The premutation is often passed from one generation to the next without anyone knowing and can cause fragile X syndrome.
- Share information about fragile X with your blood relatives by printing this page and this letter for family members: https://portal.earlycheck.org/en/fragile-x/letter-for-family-members

How can people with FXPM have children with fragile X syndrome (FXS)?



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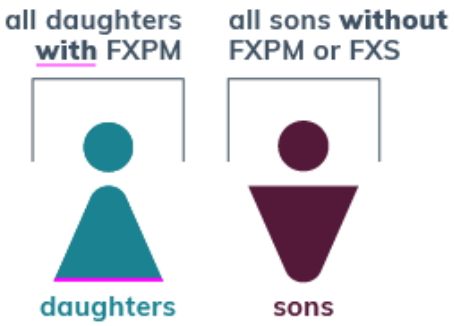
- Each time a woman with FXPM has a child, there is a 50/50 (50%) chance of passing on the fragile X premutation. When passed from a woman to her child, the premutation can change to become a larger "full" mutation that causes fragile X syndrome.

- Boys who inherit the full mutation will have fragile X syndrome.
- Girls who inherit the full mutation can have fragile X syndrome, mild features, or no features of fragile X syndrome.
- Every daughter of a man with FXPM will inherit FXPM. The sons of men with who have FXPM will not inherit it from their father.



- To learn more about how fragile X runs in families, see the Fragile X Genetics Fact Sheet at https://portal.earlycheck.org/en/genetics-of-fragile-x
- To learn about options for planning a family and testing for fragile X during pregnancy (prenatal diagnosis), see the **National Fragile X Foundation Premutation Carrier page** at https://fragilex.org/learn/premutation-carriers/ and talk with your doctor or a genetic counselor.

What is a fragile X premutation "*carrier*"?

- People with FXPM are sometimes referred to as "*fragile X carriers*," meaning they "carry"--or have--the premutation and can pass it on to their children.
- For *most* conditions that run in families, being a carrier does not cause any health problems.
- In fragile X, having the premutation can sometimes affect health and well-being.

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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.

- Parents of children with the fragile X premutation (FXPM) should consider testing first to see if one of them also has FXPM.
- Testing should be considered for children, brothers, sisters, and more distant family members of people with FXPM, especially if they have problems with development, learning, or behavior.
- Fathers of women who have the fragile X premutation should consider testing, especially if they have trembling hands, difficulty walking, or changes in mood, or difficulty thinking.
- Family members who are pregnant or considering pregnancy should speak with their doctor or a genetic counselor to learn more about testing and options for planning a family.
- Share information about fragile X with your blood relatives by printing the Letter for Family Members available when you log into Early Check.

What Causes Fragile X?

To learn more about the gene involved in fragile X, see the Fragile X Genetics Fact Sheet at https://portal.earlycheck.org/en/genetics-of-fragile-x.

What are good sources of reliable information?

Learn more about fragile X syndrome and what it means to have the fragile X premutation.

- **National Fragile X Foundation** website: Fragilex.org
- **Center for Disease Control Fragile X** webpage: https://www.cdc.gov/ncbddd/fxs/index.html

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