Early Check
Expanded health screening for your baby

Fact Sheet: Genetics of Fragile X

To understand more about fragile X, it can help to review information about genes, chromosomes, and how we inherit traits from our parents.

What are chromosomes?

- Chromosomes are tiny bundles of DNA.
- The chromosomes all have names. One is called the **X** chromosome.
- The chromosomes are packed with thousands of even smaller structures called **genes**.

What are genes?

- On the chromosomes are thousands of **genes**. They are even smaller than chromosomes.
- Genes are like recipes for important chemicals in the body. They give the instructions that tell our bodies how to grow and develop.
- There are many different forms of each gene. Different forms give slightly different instructions. That's what makes us each unique!

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How are traits passed on to children?

- At the time a pregnancy is first made, half of the genes come from the mother and half from the father.
- This is how family traits are passed down from one generation to the next.
- It is also how some health conditions run in families.

How do health problems run in families?

- Some forms of genes can affect health or development. Forms of genes that cause health problems are called *mutations*.
- Mutations are passed down from parents to children just like other traits.
- There is nothing parents do to make this happen.

An important gene:

- Everyone has at least one copy of a important gene called FMR1.
- Girls have two. Boys have one.
- This gene makes a chemical that is very important for brain development.
- The name of the chemical is FMRP.

What causes fragile X syndrome?

- Mutations (also called "full" mutations) in the FMR1 gene stop it from making the needed brain chemical FMRP.
- Not having enough FMRP causes the features of fragile X syndrome.
- People often call the FMR1 gene "the fragile X gene."

Why is fragile X syndrome usually milder in girls?

- About half of girls with the full mutation have features of fragile X syndrome.
- Unlike males with just one fragile X gene, females have two copies.
- If one of a girl's fragile X genes has the mutation, her other fragile X gene makes some of the needed brain chemical, FMRP.
- Most females have fewer and or milder features of fragile X syndrome.

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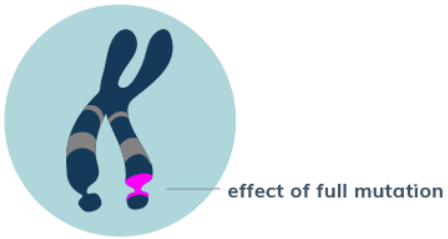
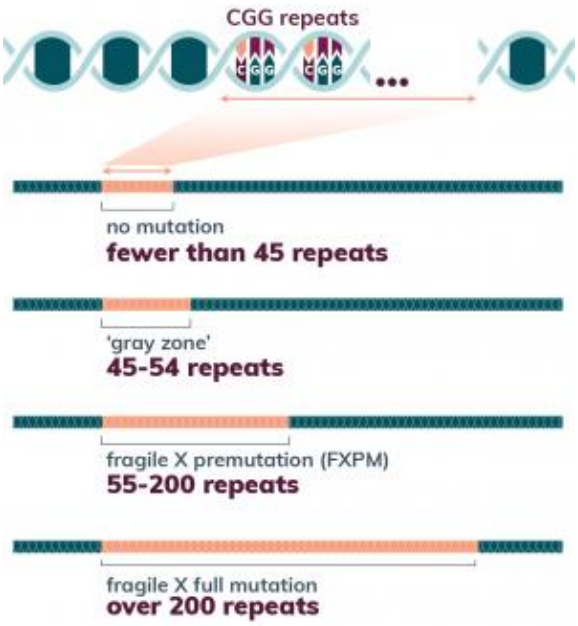
Why is it called fragile X?

- The fragile **X** gene is located on the end of the **X** chromosome. That's where the "**X**" in the name comes from**.**
- The full mutation in the gene makes the end of the **X** chromosome longer and look narrow or "fragile". This is only seen with a microscope.
- People with fragile X syndrome full mutation and carriers of the fragile X premutation are **not** physically weak or fragile.



What is a CGG repeat?



- The gene is measured by counting a repeated pattern in the gene called a *CGG repeat*.
- More CGG repeats means a longer mutation and a larger form of the gene.
- Most people have less than 45 repeats.
- People with the premutation have 55-200 repeats but they do not have fragile X syndrome.
- Large mutations (over 200 CGG repeats) stop the fragile X gene from working. This decreases the amount of the FMRP in the body and brain, which causes of fragile X syndrome.

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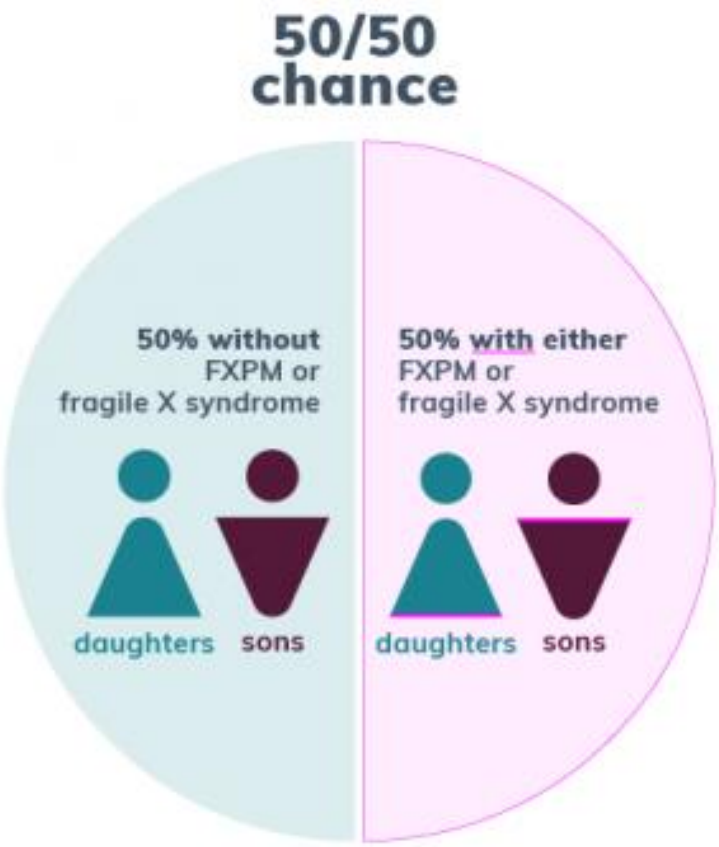
Where does the full mutation come from that causes fragile X syndrome?

- The number of CGG repeats often increases (expands) when the fragile X gene is passed down from a mother with the premutation or gray-zone allele to her child.

- When the mother has the premutation, baby girls and boys can inherit the full mutation that causes fragile X syndrome, fragile X premutation, or no mutation at all.



Where can I learn more about the genetics of fragile X?

- National Fragile X Foundation's Genetics and Inheritance webpage.
- The Center for Disease Control (CDC): https://www.cdc.gov/ncbddd/fxs/index.html
- Early Check webpage: https://portal.earlycheck.org/en/first-stop-for-parents-fxpm