KAISER PERMANENTE®

INFORMATION SHEET

FRAGILE X CARRIER SCREENING

Intermediate (Gray-Zone) Result

Your carrier screening test showed that you have an intermediate result in the fragile X gene. This is also called a gray-zone result. **This is not expected to affect your health or the health of your offspring.**

What does it mean to have an intermediate result?

The fragile X gene varies in size from person to person. An intermediate result means that the fragile X gene is a little larger than usual, but the gene still works the way it should. No extra testing is needed. A person with an intermediate result is **not** considered a fragile X carrier. This is usually a family trait that has been passed down from generation to generation. About 2% to 3% of the population has this same change in the fragile X gene. There is a small chance for the gene to get slightly larger when it is passed on. If this happens, future generations (like your grandchildren or great-grandchildren) may be at risk for fragile X syndrome.

Should my child be tested?

There is no need to test your child (or a future child). This result will not lead to fragile X syndrome or other health conditions in your offspring. Fragile X testing could be offered to any child of yours when they are planning children of their own.

MORE ABOUT FRAGILE X SYNDROME

Fragile X syndrome (FXS) is an inherited condition that happens when a single gene is not working. A person with FXS often has intellectual disability and autistic behaviors.

The gene that causes fragile X syndrome is called *FMR1*. This gene makes a protein needed for normal brain development. The *FMR1* gene has an area where three letters of DNA, "**CGG"**, are repeated over and over. The total number of CGG repeats affects how the gene works.

The *FMR1* gene is found on the X-chromosome. Males have one X-chromosome and females have two X-chromosomes. That means males only have one copy of the *FMR1* gene and females have two copies. This is why FXS is more likely to affect males.

Fragile X Results

Normal: 5 to 44 CGG repeats
This is the typical number of repeats.

Intermediate: 45 to 54 CGG repeats
The gene still works in this range. The number of repeats can increase a little when passed from a parent to a child, but does not increase to a full mutation.

Premutation: 55 to 200 CGG repeats
The gene is unstable in this range. A person is a fragile X carrier, but does not have fragile X syndrome. Both males and females can have a premutation. The gene can get larger when passed from a mother to her child and may cause FXS in the child. It can also cause early menopause or a movement disorder in some people.

Full mutation: Over 200 CGG repeats
The gene stops working in this range and causes FXS in boys. It can also affect some girls.