

Genetic carrier screening can generally be done by either a blood or a saliva test. The blood or saliva is sent to the lab, where they can then pull out your DNA from either your blood cells or your cheek cells that are in your saliva. This DNA is what makes up your genes. Your genes are basically a string of letters that are an instruction for how to make a protein. Each protein has a very specific job to do in the body. Some proteins tell our body how tall we're going to be, while others tell our eyes what color they will be.

Some proteins perform such important jobs in the body, that when we're born without them, they can cause health problems, known as genetic conditions. If there is a harmful spelling error in the DNA, known as a pathogenic variant, it can change the instructions for that gene. If the instructions are changed, then that gene can make either an abnormal protein or no protein at all. Not having these proteins can lead to different health concerns.

Each lab offers screening for various different genetic conditions. Most of the conditions that are offered for prenatal or preconception carrier screening are passed down in a family in a pattern we call [autosomal recessive](#), although there are some [X-linked](#) conditions that can be screened for.

What is a pathogenic variant?

Remember that your genes are a string of letters that are basically a set of instructions to build a protein. A pathogenic variant is any change in those instructions that affects how the gene works. In other words, a pathogenic variant is a spelling error, or a missing or extra piece, in the instructions that affects how the protein is made.

A similar example is self-assemble furniture. Let's say you buy a kit at the store to build a jungle gym for your kids in the back yard. You lay all of the pieces out, start going through the instructions and putting things together. When you get done, the jungle gym doesn't look like it's supposed to and you have a bunch of extra pieces left over. You go back through the instructions and realize that there is a page that is missing. Without that missing page, you don't have proper instructions on how to put together your jungle gym. The instructions for how a gene builds a protein is very similar.

To learn about the different kinds of pathogenic variants, see our pages on [sequencing](#) and [deletion/duplication](#) testing.

Click [here](#) to learn more about scheduling a genetic counseling appointment for pregnancy-related questions.

Click [here](#) to learn more about scheduling a genetic counseling appointment for infertility or preconception questions.

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