While most screening tests we talk about during pregnancy are designed to give us more information about the baby, genetic carrier screening is a test that gives us information about mom and dad.

As you may recall from Genetics 101, we have two copies of most genes: one we get from our mom and one we get from our dad. When we say that someone is a 'carrier' for a genetic condition, it means that there is a harmful change (called a pathogenic variant) in one of the copies of the gene that make it not work how it should. Because there is still one gene that is working properly, carriers generally do not show signs or symptoms of any health issues related to being a carrier.

Genetic carrier screening usually looks for conditions that are recessive, meaning that for someone to have the genetic condition both copies of the gene need to be not working. In most instances, if someone is a carrier for a recessive condition, they don't have any medical or health problems, and may not have a family history of any genetic conditions.

Because being a carrier for a genetic condition does not cause any health problems in most cases, then what's the point of testing for it?

We are all probably carrier for five or more recessive genetic conditions. If two people who are carriers for the same genetic condition have a child, there is a 25% chance that they will have a child that has that genetic condition. This means there is a 1 in 4 chance that each parent will pass down their non-working copy of the gene to the pregnancy, and then the baby will have no working copies of that gene.

Carrier screening may also offer testing for X-linked recessive conditions. X-linked means that the gene is located on the X chromosome. To review from Genetics 101, women have two X chromosomes and men have one X and one Y chromosome (and thus only one copy of all of the genes on the X chromosome). If a male inherits a nonworking gene on his X chromosome, he usually will have that genetic condition because he does not have a second working copy of that gene.

If a woman, on the other hand, inherits a nonworking gene on one of her X chromosomes, she usually has a working copy of that gene on her other X chromosome. If a woman is a carrier for an X-linked recessive condition, she may have very mild or no symptoms at all. A woman who is a carrier for an X-linked recessive genetic condition would have a chance to pass that condition down to her children.

Click below to read more about prenatal carrier screening:

- How it works
- Commonly tested for conditions
- Expanded carrier screening
- Results
- How to decide

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