

We have over 20,000 genes in our bodies, and each gene performs a very specific job. Some genes determine our hair color or our height, while other genes control how our bones and organs form and function. Typically both copies from the pair are functional, but we all have genes that are not working as they should. In fact, scientists estimate that we each have approximately 20 non-working genes! This usually doesn't cause a problem because you have two, so there's a built-in backup system; if one gene doesn't work then the other one takes over. However, if both copies of a gene that a person inherited do not work, this can lead to a genetic condition that is caused by autosomal recessive inheritance.

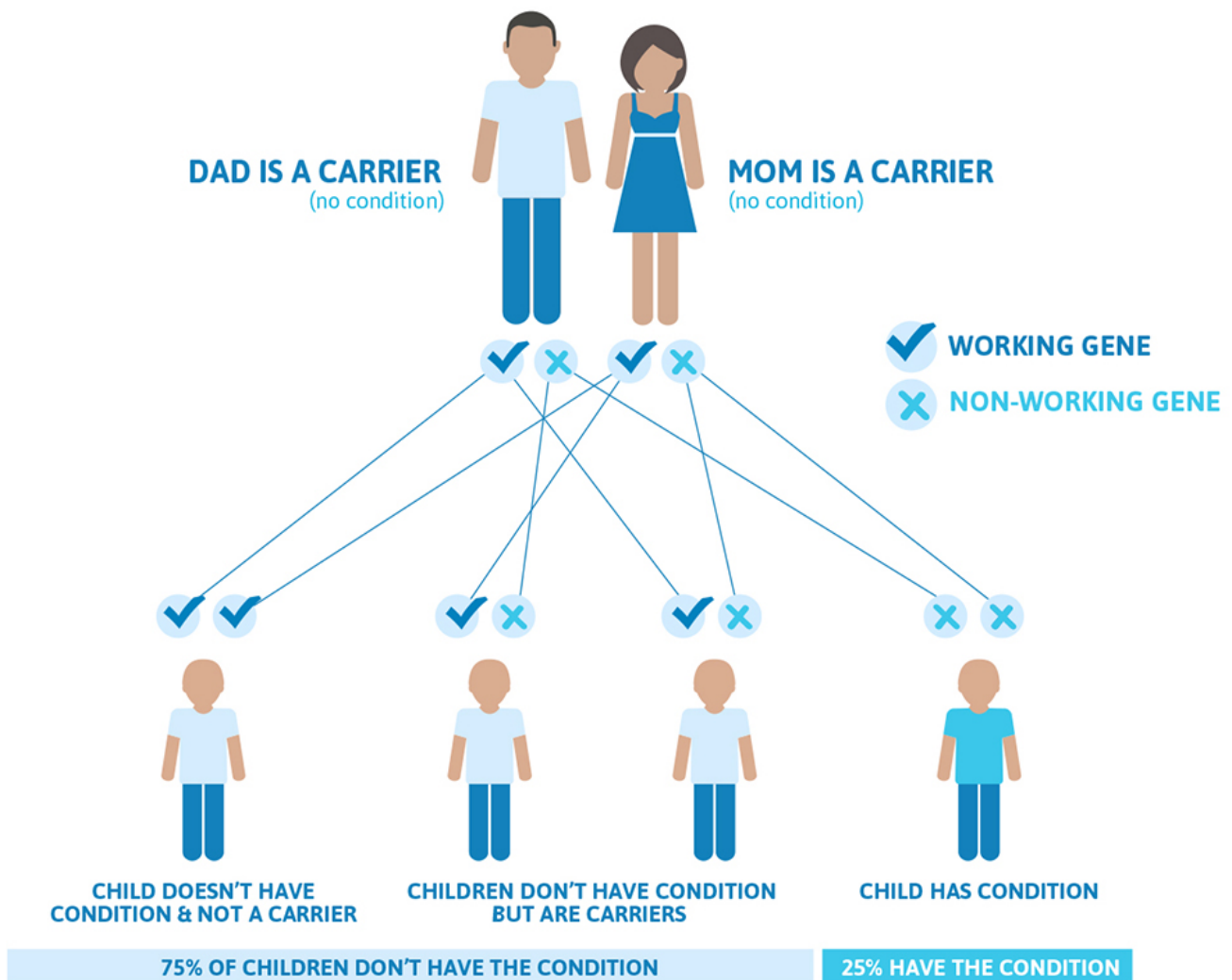
Autosomal means the gene that is involved is found on one of the first 22 chromosomes (called the autosomes) and not on the X or Y chromosome (the sex chromosomes).

In order to have a recessive condition a person has to have BOTH copies of a gene that are not working. The copy they inherit from mom is not working AND the copy they inherit from dad is not working, resulting in zero functioning copies of that gene. With recessive conditions, if you have one copy of the non-working gene (called a carrier), you do not have the condition and typically do not have any related symptoms.

How does an autosomal recessive condition look in a family?

It is rare to see any history of autosomal recessive conditions within a family because if someone is a carrier for one of these conditions, they would have to have a child with someone who is also a carrier for the same condition. Most autosomal recessive conditions are individually rare, so the chance that both people are carriers for the same recessive genetic condition are likely low. Even if both partners are a carrier for the same genetic condition, there is only a 25% chance that they will both pass down the non-working copy of the gene to the baby, which would lead to the baby having the genetic condition. This chance is the same with each pregnancy, no matter how many children they have with or without the condition.

Autosomal Recessive Inheritance Pattern



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That means also there's a 50% chance the baby would be a carrier just like it's parents, and as discussed above carriers generally do not have any symptoms. There's also a 25% chance that the baby would inherit a working copy of the gene from both parents and not be a carrier.

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