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 how 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 doesn't work the other one takes over. However, this is not true for all genes. There are many genetic conditions where if one gene of the pair is not working, the other one cannot make up for it and it can cause a genetic disease. This illustrates a common pattern of heredity called autosomal dominant inheritance.

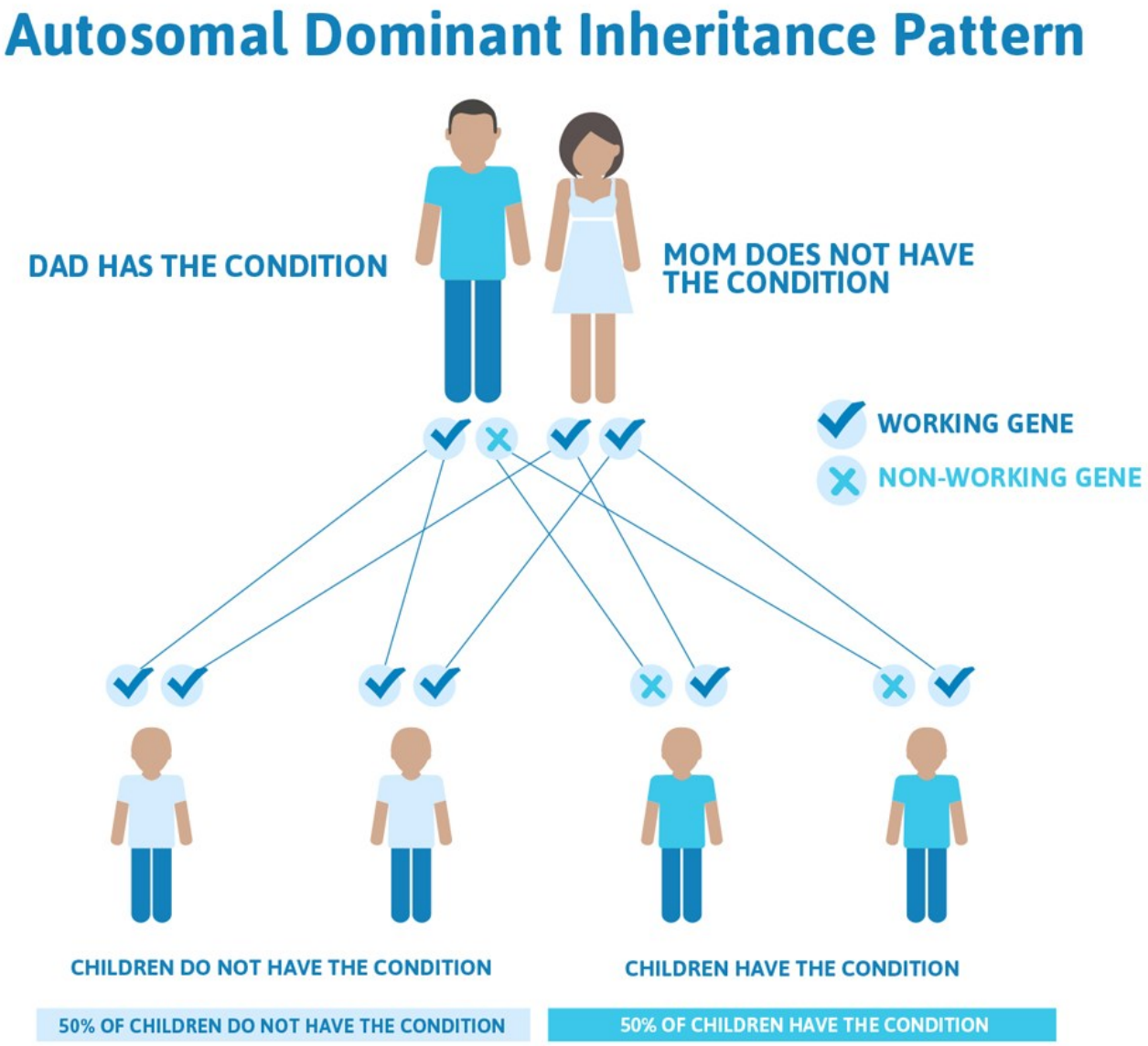
Autosomal means the gene that is involved is found on one of the first 22 chromosomes (called the autosomes) and not on the X or Y chromosome (the sex chromosomes).

Dominant refers to the above explanation that we have two copies of each gene, one from mom and one from dad, and in order to have an autosomal dominant condition, a person only has to have one copy of the gene not working. They can inherit this copy from mom or dad, who may also have the condition.

So how does an autosomal dominant condition look in a family?

Often autosomal dominant conditions can be seen in multiple generations within the family. If one looks back through their family history they notice their mother, grandfather, aunt/uncle, etc., all had the same condition. In cases where the autosomal dominant condition does run in the family, the chance for an affected person to have a child with the same condition is 50% regardless of whether it is a boy or a girl. It is the same 50% chance for each pregnancy.

There are cases of autosomal dominant gene changes, called pathogenic variants, where no one in the family has it before and it appears to be a new thing in the family. This is called a de novo pathogenic variant. For the individual with the condition, the chance of their children inheriting it will be 50%. However, other family members are generally not likely to be at increased risk.



Click the chart to view in full-screen.

This diagram illustrates autosomal dominant inheritance. The example below shows what happens when dad has the condition, but the chances of having a child with the condition would be the same if mom had the condition.

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