

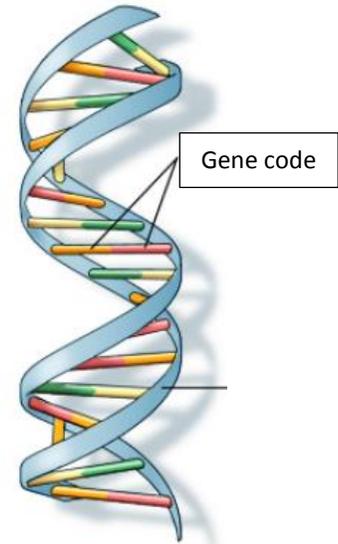


Genetics Glossary

Gene – a code for how our body grows and develops, like a recipe. This includes information about our appearance, like hair color or eye color, and traits, like blood type.

We inherit one gene from each of our parents, and pass them down to our children. In general, we all have two copies of each gene, one from our father and one from our mother.

Everyone has changes in their genetic code that make them a unique individual. Changes may be called mutations or **variants**. Terms like changes, mutations, and variants have been used interchangeably over time. Sometimes changes can prevent a gene from working properly. These types of changes may cause health problems or disease.



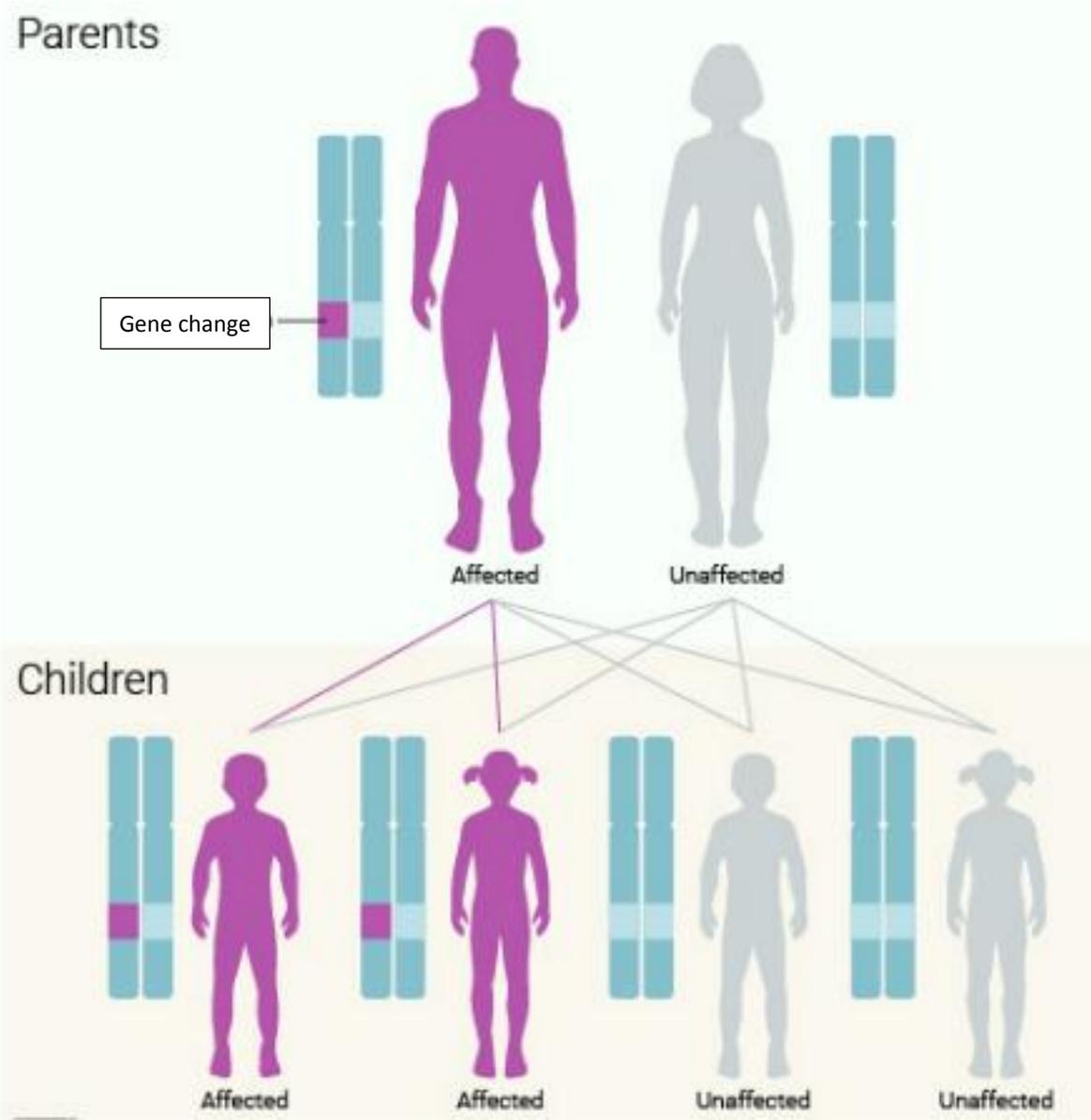
U.S. National Library of Medicine

Gene sequencing – laboratory testing used to find changes in the genetic code. It is similar to “spell check” for any “typos” in the genetic code. In KidsCanSeq, testing may be done on a sample of tumor, blood or saliva.

Genetic condition – health problems or disease caused by changes to a gene. Genetic conditions may be passed down in families, or someone may be the first person in their family to have a genetic condition.

Dominant genetic condition – a genetic condition caused by having only one change in a specific gene. An individual who is affected by a dominant genetic condition has a 50% chance to pass on the change to each of his or her children.

Dominant Genetic Condition



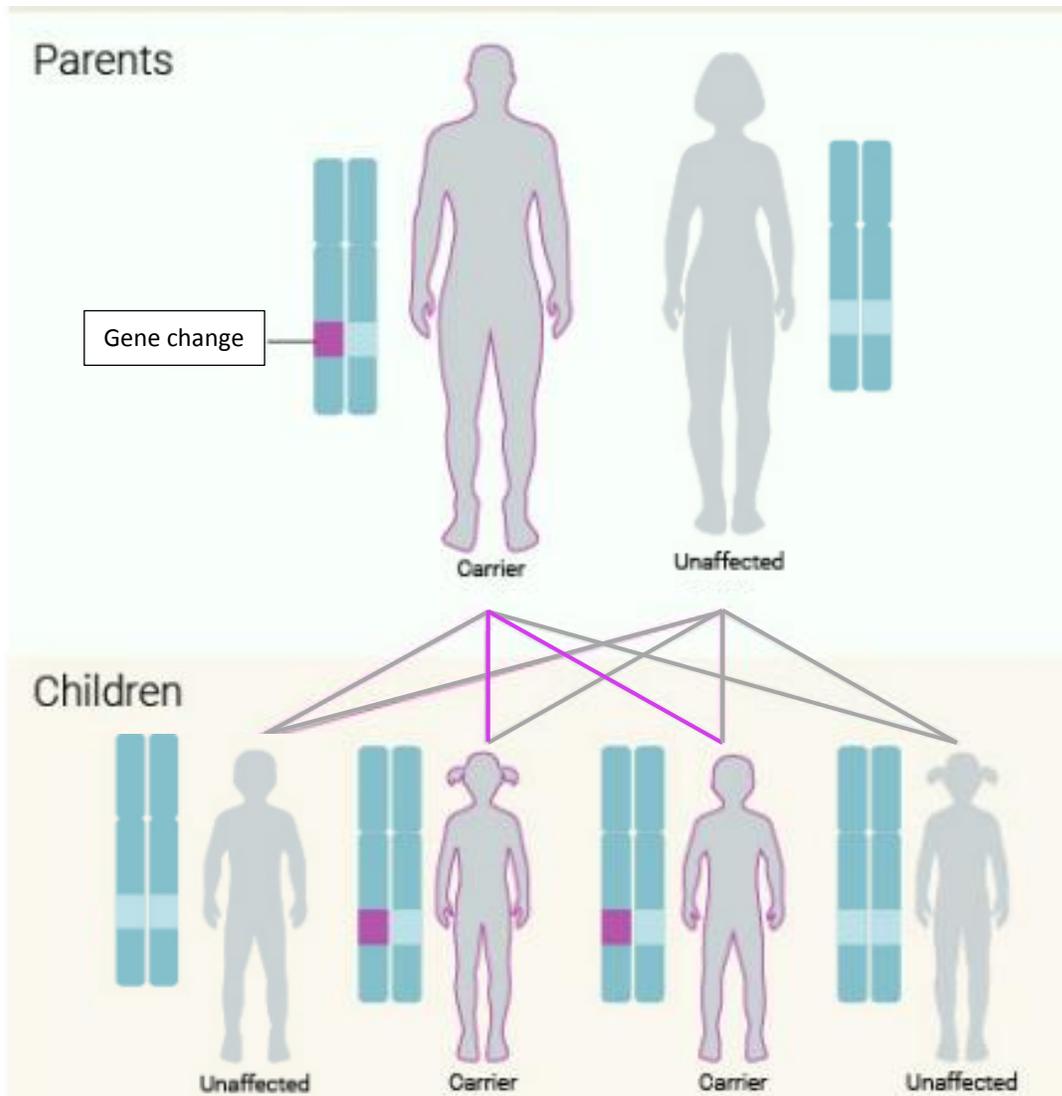
NIH Genetics Home Reference: <https://ghr.nlm.nih.gov/>

Affected – an individual who has a genetic condition
Unaffected – an individual who does not have a genetic condition

Recessive genetic condition – a genetic condition caused by having two changes in a specific gene. Someone with one recessive gene change typically does not develop the condition and is sometimes referred to as a “carrier”.

If only one member of a couple has a recessive gene change, there is a 50% chance with each pregnancy to have a child who also carries the gene change, and a 50% chance to have a child without the change.

Recessive Genetic Condition – One Parent is a Carrier



Adapted from NIH Genetics Home Reference: <https://ghr.nlm.nih.gov/>

Affected – an individual who has a genetic condition

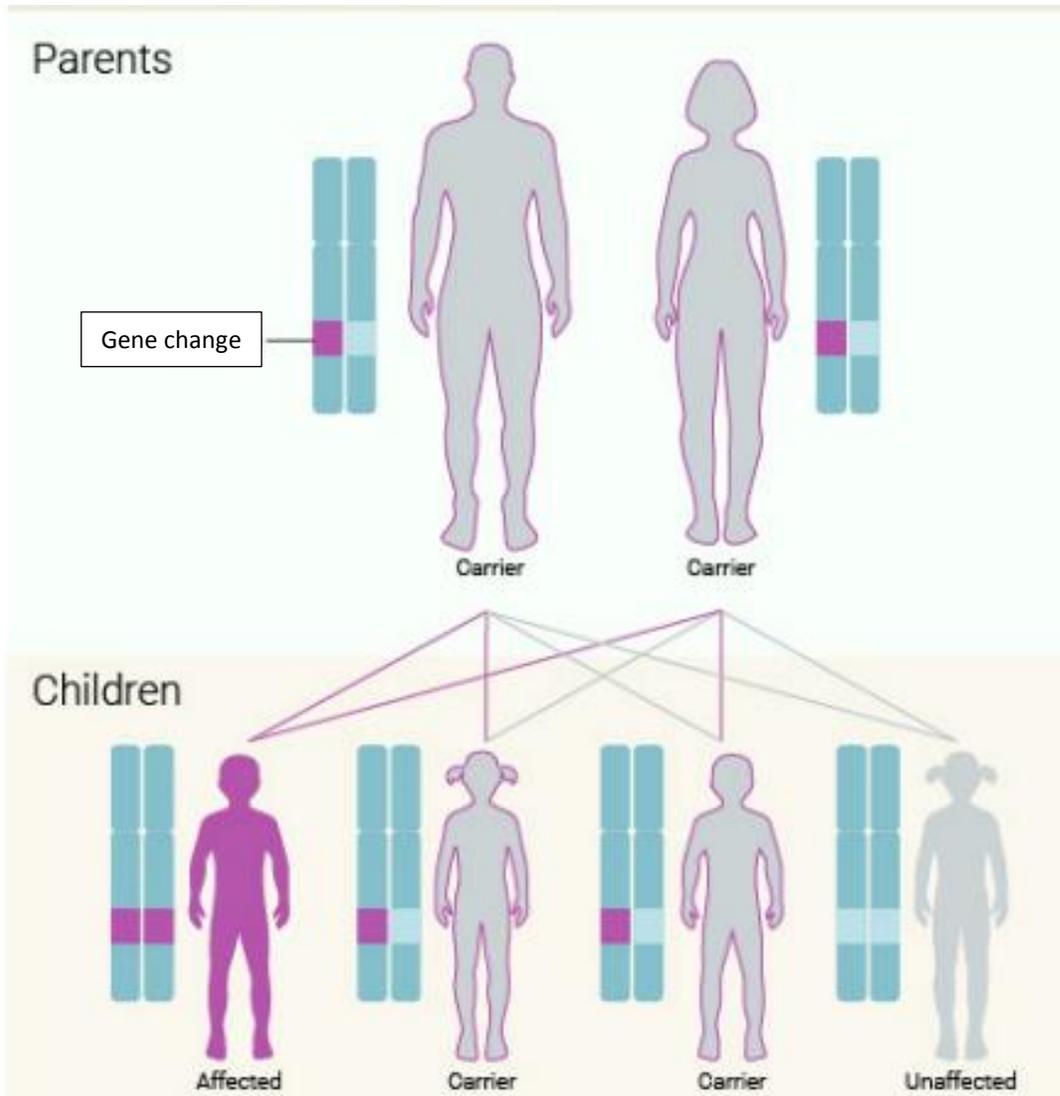
Unaffected – an individual who does not have a genetic condition

Carrier – an individual with one gene change that typically does not develop the genetic

For a couple who each have one recessive gene change, neither has the genetic condition, but they have a 25% chance of having a child with the genetic condition, a 50% chance of having a child who has one gene change, and a 25% chance of having a child who does not have the condition and does not have the gene changes.

Remember that someone with one recessive gene change typically does not develop the condition and is sometimes referred to as a “carrier”.

Recessive Genetic Condition – Both Parents are a Carrier



NIH Genetics Home Reference: <https://ghr.nlm.nih.gov/>

Affected – an individual who has a genetic condition

Unaffected – an individual who does not have a genetic condition

Carrier – an individual with one gene change that typically does not develop the genetic