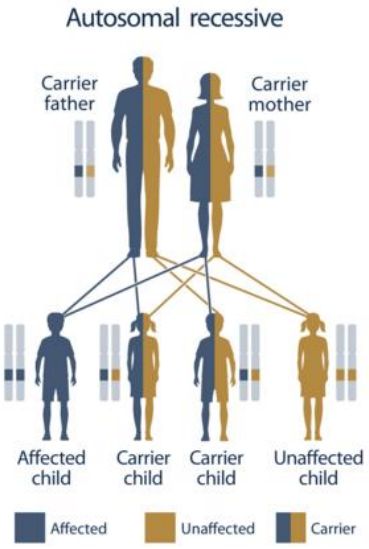
PA Genetics, Part 1

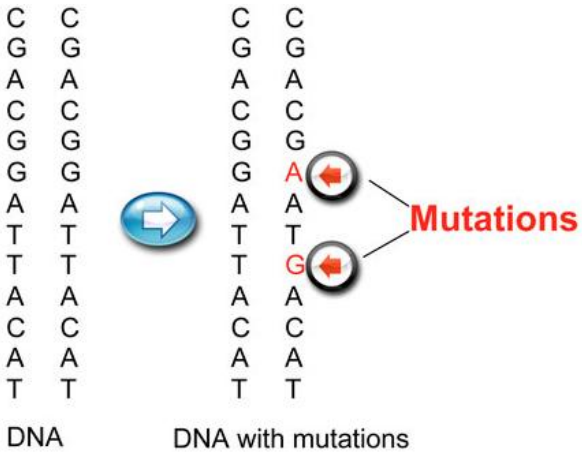
Propionic Acidemia (PA) is a condition caused by changes in the genes that make the propionyl-coenzyme A (CoA) carboxylase enzyme. **Genes** are made of **DNA** which is our hereditary material. Genes have the instructions that tell our bodies how to grow and function. Each gene provides specific instructions for various biological processes in the body.

The genes that make the propionyl CoA carboxylase enzyme are called PCCA and PCCB. The enzyme helps break down certain proteins and fats from food to make into chemical energy and other products the body needs. When there is a change in the gene called a **mutation**, the genes cannot perform their normal function. If these genes do not work and the body cannot break down fats and proteins, there is a buildup of organic acids in the body which can cause the symptoms associated with PA such as vomiting, weak muscle tone, and developmental delays.

If someone has a mutation, it is something he or she was born with. These mutations happen randomly and they are not caused by something the person did. We have two copies of each gene. We inherit one copy from each parent. If someone has one gene with a mutation and one gene that works properly, they are called a **carrier**. Carriers do not have symptoms of propionic acidemia because having one working gene copy means the body is still able to break down fats and proteins.



If both parents are carriers of propionic acidemia, there is a 1 in 4 or 25% chance of having a child with propionic acidemia. This is called **autosomal recessive** inheritance. The condition can affect males and females and an individual has to inherit two mutated genes to be affected with PA. Therefore, in order to be affected by PA, the child has to inherit a gene mutation from both parents. If a child inherits one working gene and one mutated gene, they will also be a carrier of PA and will not have symptoms. If a child inherits both normal copies of the gene, they will not be a carrier and not have the condition.



To find out if you are a carrier of PA, you can have **genetic testing**. Our DNA is written in a four-letter code. Genetic testing works by reading through the code like a spellchecker looking for a change, also called a mutation.

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