AKU

Alkaptonuria Society

How AKU is Inherited

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Have you ever asked yourself why members of the same family look similar?

Genes carry the information that determines the traits or characteristics that are passed on to us, or inherited from our parents. Blood relatives have a large number of the same genes in common. This is why family members may have many similar traits such as the same hair colour and eye colour.

AKU is what's known as an autosomal recessive condition. Autosomal recessive is one of several ways that a trait, disorder, or disease can be passed down through families.

An autosomal recessive disorder means two copies of an affected gene must be present for the disease or trait to develop.

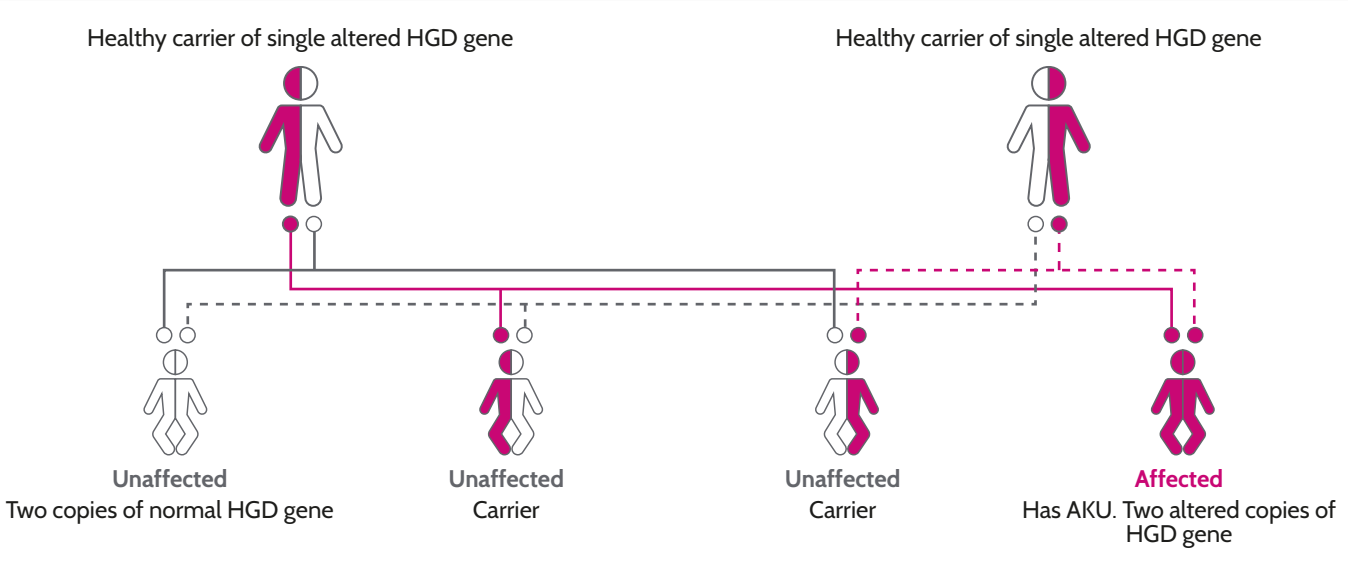
Genes are passed on from parents to children. We all carry two copies of each gene, one inherited from our mother and one from our father.

The gene involved in alkaptonuria is the HGD gene. You need to inherit two copies of the affected HGD gene (one from each parent) to develop alkaptonuria. The chances of this are slim, which is why the disease is very rare – affecting just 1 in 250,000 to 500,000 people worldwide.

The parents of a person with alkaptonuria will usually only carry one copy of the affected gene themselves. They are known as 'healthy carriers' and will not have any signs or symptoms of the condition.

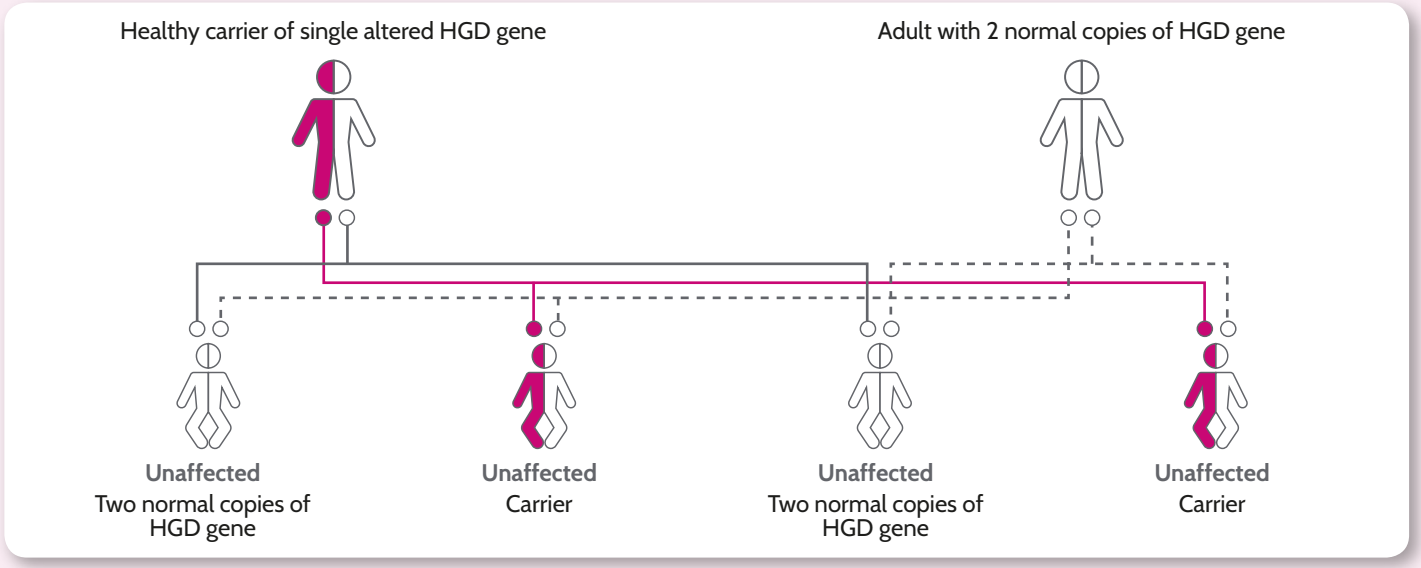
If two healthy carriers of AKU have children together each child they have has a:

- **1 in 4** (25%) chance of having AKU
- **1 in 4** (25%) chance of not having AKU
- **2 in 4** (50%) chance of being a carrier

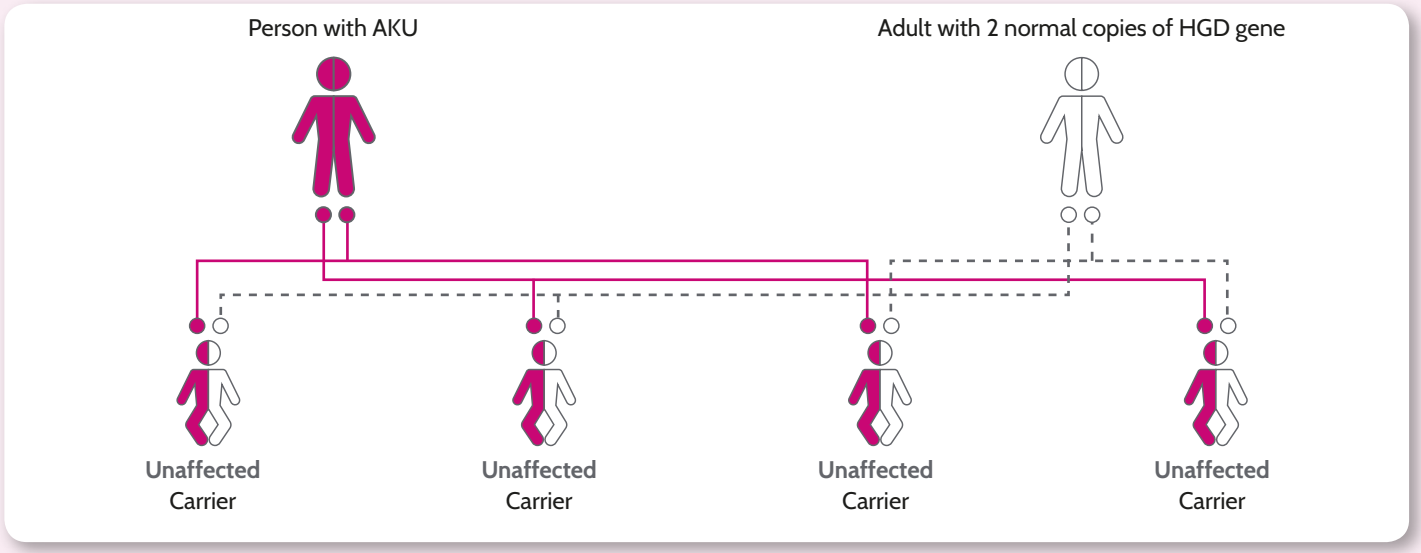


If a healthy carrier of AKU has children with someone who is not a carrier of the same condition each child they have has:

- 1 in 2 (50%) chance of not being a carrier
- 1 in 2 (50%) chance of being a carrier



If a person with AKU has children with someone who is not a carrier of the same condition each child they have will be a healthy carrier of AKU.



Support and Useful Addresses

Carrier Testing

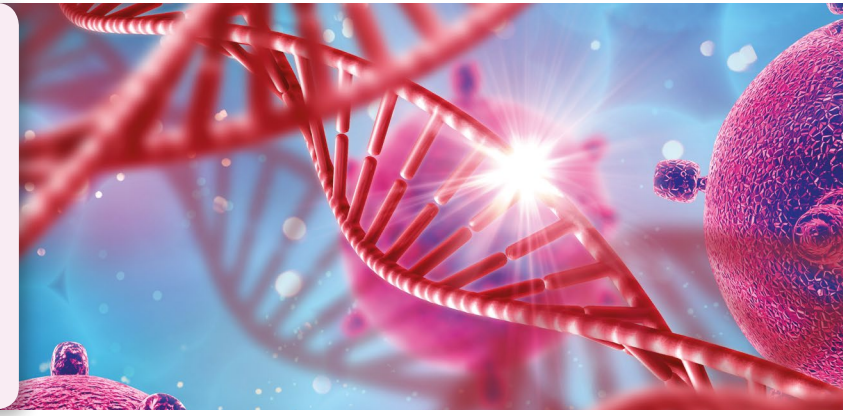
If one of your relatives is affected with AKU or is a carrier for AKU, you and your partner may be able to have carrier testing for the condition. Carrier testing can tell you one of two things:

1. You do not carry AKU. In this case, there is no chance of you having a child with AKU. This will also be the case whether your partner is a carrier of AKU or not.
2. You are a carrier for AKU. Most of the time, there is a low chance of having a child affected with the condition even when one of the partners is a carrier. This is because their child is only at risk of having the condition if both parents are carriers.

There are some situations where the chance may be higher if you and your partner share common relatives such as grandparents.

AKU is a rare genetic condition where you usually need to inherit the affected gene from both parents. However, there are a very small number of people who have AKU even though they have only one affected gene. This is ultra-rare but not unheard of.

If you are concerned about having AKU or being a carrier speak to your GP. Your GP can offer more information and support and refer you to an appropriate specialist service.



Genetic Alliance UK provides information and support to individuals and families affected by inherited disorders.

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AKU Society

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The Robert Gregory
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