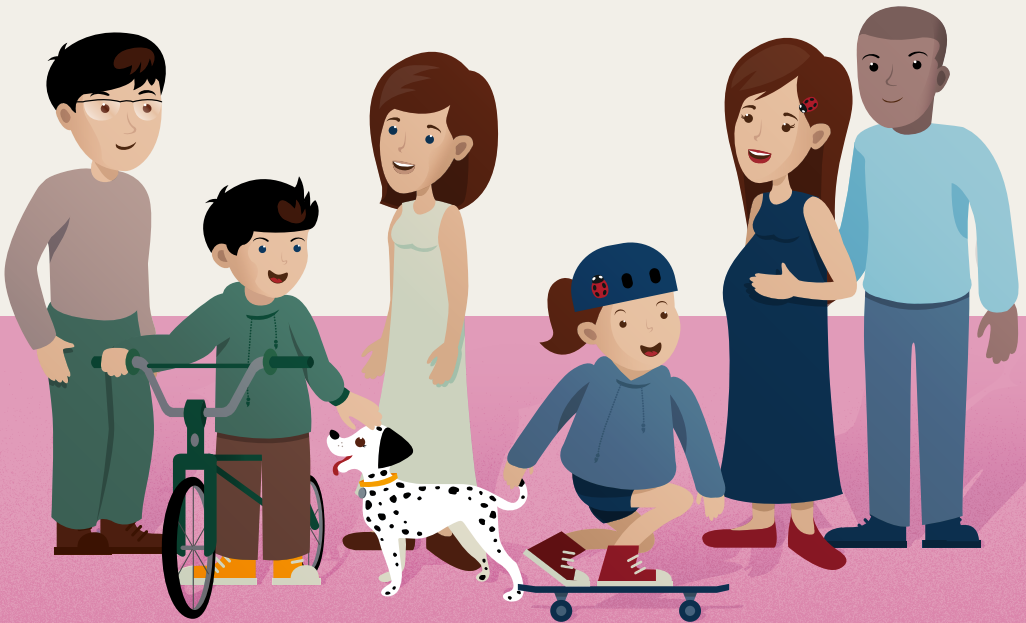


# AKU

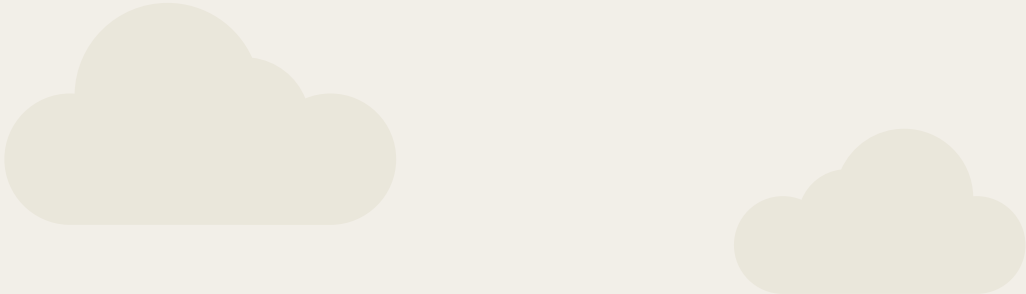
+ Alkaptonuria Society →

## → An introduction to Alkaptonuria (AKU)




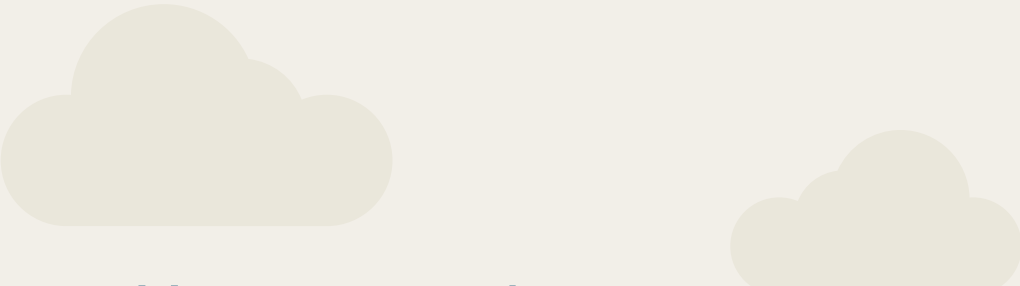


**Alkaptonuria**  
(sounds like al-kap-ton-u-re-ah)  
or **AKU** for short




# **AKU is a manageable condition which affects the way the body breaks down protein**

- 
- Children with AKU have a defect in the process which breaks down part of a protein called Tyrosine (TYR).
  - This leads to the build up of an acid called Homogentisic acid (HGA).
  - As you grow older HGA will affect your bones and joints.
  - With management, people with AKU attend university, build successful careers and enjoy a happy family life the same as everyone else.



You may notice some of these symptoms:

- **dark urine**
- **dark earwax**
- **dark sweat**



They are not painful and nothing to worry about.

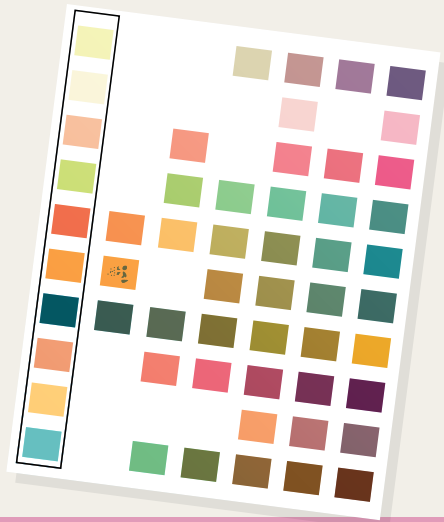
# When you get older you may notice more symptoms




Dark spots on your eyes.  
Pain in your lower back and joints.



# AKU is diagnosed by a simple urine test



If the urine test shows high levels of HGA, you will be referred to an expert doctor.

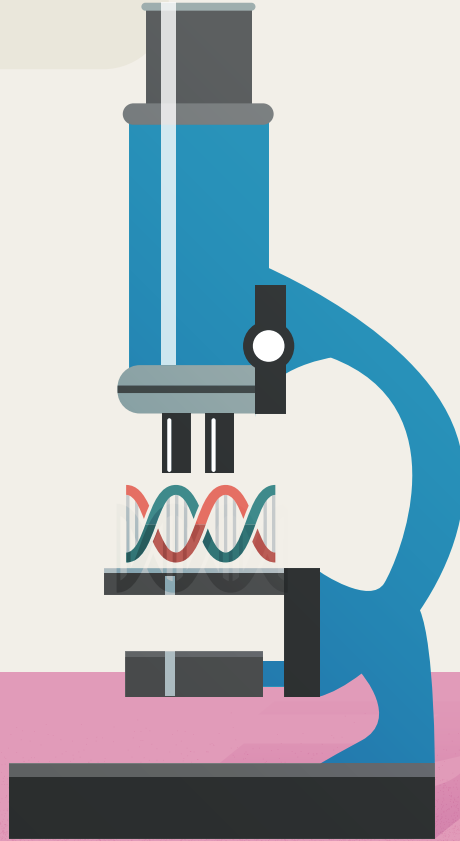
A cartoon illustration of a young girl with long brown hair, wearing a blue shirt and red boots, holding a large white sign. The sign has the text "Why do I have AKU?" written on it in blue. The background is a simple landscape with a pink ground, a white sky, and some stylized trees and clouds.

**Why do  
I have  
AKU?**

You can not give AKU to your friends.



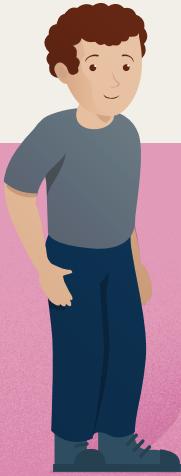
# What are genes?



Inside every cell of our body are sets of instructions called genes. These genes provide instructions on how we look and what features we inherit from our parents. These genes provide information on how to break down protein, including tyrosine. The body contains over 25000 genes all working together. They can only be seen under a microscope.

# AKU is an inherited condition

It's nobody's fault and there is nothing you could have done to prevent it.



FATHER  
Carrier



Normal Gene



AKU Gene



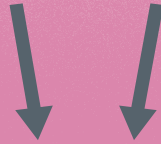
AKU Gene



Normal Gene



MOTHER  
Carrier



AKU Gene  
AKU Gene



Child with AKU

Your mum and dad will have one AKU gene and one normal gene. This is known as being a carrier.

Having AKU means you will have inherited 2 AKU genes, one from mum and one from dad.

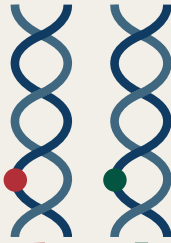
# Around 1 in 500 people are carriers of the AKU gene



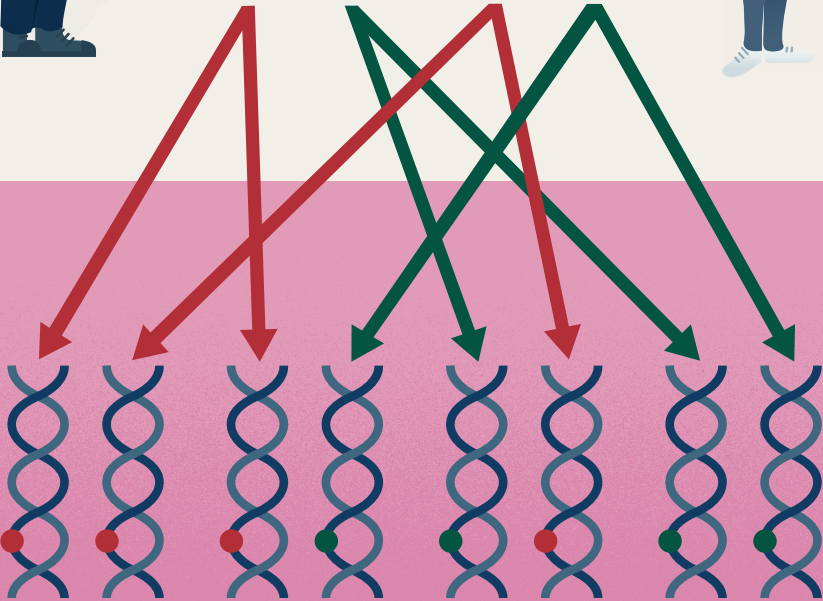
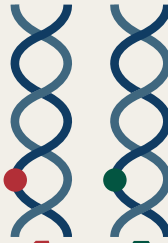
People who are carriers for AKU do not have AKU themselves and the faulty gene does not cause a problem to them.



Father



Mother



Child with  
AKU



Carrier  
Child  
without  
AKU



Carrier  
Child  
without  
AKU



Child  
without  
AKU and is  
not a carrier

When 2 people who are carriers of the AKU gene have a child there is a 1 in 4 chance of that child having AKU.

A cartoon illustration of a young boy with black hair, blue eyes, and a slight smile. He is wearing brown shorts and brown boots with orange laces. He is holding a large white rectangular sign in front of his chest. The background is a solid pink color, with stylized grey trees and light green clouds in the upper portion.

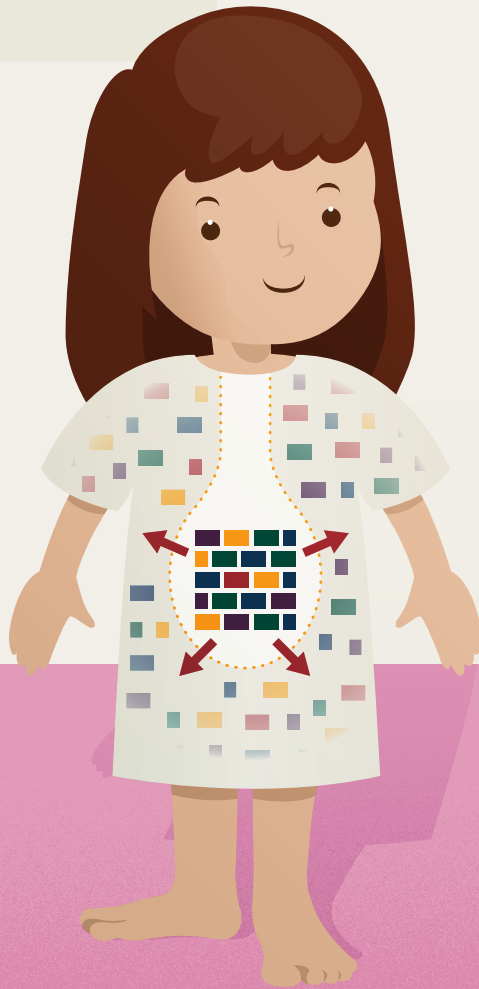
# The science bit... Protein explained

# Why do we need protein?



Protein is a nutrient needed by the body.

It helps to build, repair and maintain body cells and tissues, like your skin, muscles, organs, blood and even bones.



When protein is eaten, it is broken down in the body (during digestion) into smaller pieces (like building blocks).

These smaller pieces are called amino acids.

# Protein is made up of many building blocks called amino acids



There are 20 amino acids (building blocks) that make up protein. Some of these are ESSENTIAL and cannot be made in the body - so they must come from the food we eat.

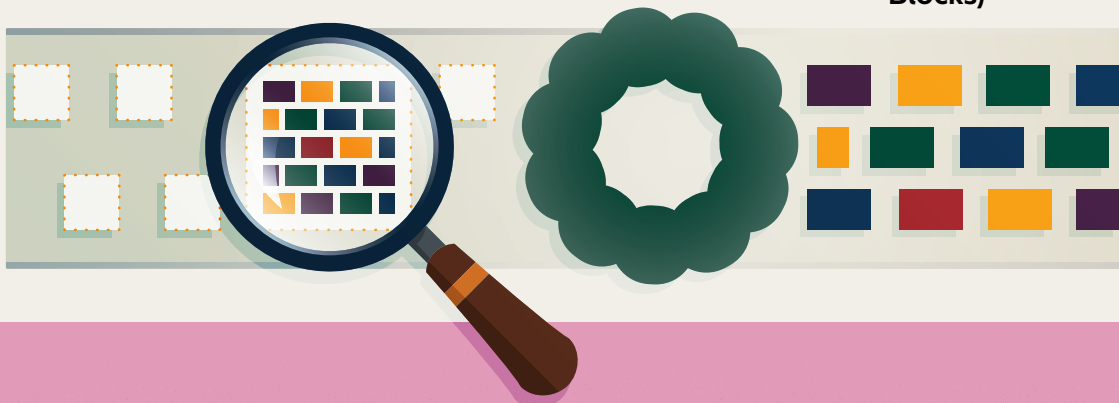
Tyrosine (Tyr) is one of these 20 amino acids. It is this particular amino acid that a person with AKU cannot breakdown correctly.




# What normally happens

The conversion of protein into amino acids

Protein → Enzymes → Amino acids (Building Blocks)

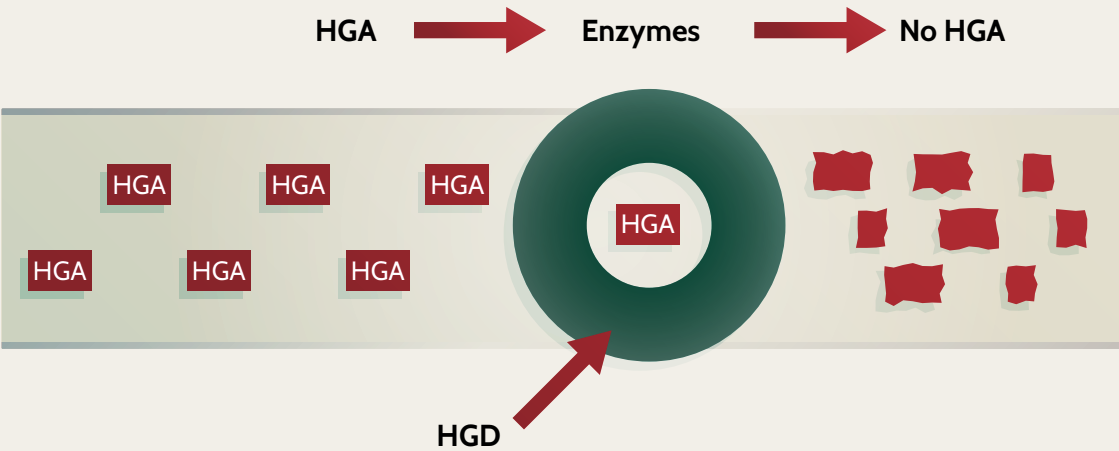


The body uses these amino acids for growing, building up muscle and helping the body stay healthy.

A cartoon illustration of a girl with long brown hair, wearing a blue shirt and red boots, holding a large white sign. The sign contains the text: "Why can someone with AKU not break down Tyr correctly?". The background features a pink ground, stylized trees, and clouds.

**Why can someone  
with AKU not  
break down  
Tyr correctly?**

# In a person **without** AKU, Tyrosine (Tyr) is broken down and removed from the body

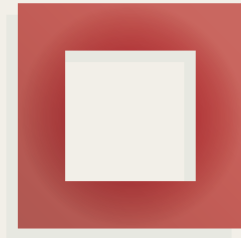


Tyrosine (Tyr) is converted into HGA.

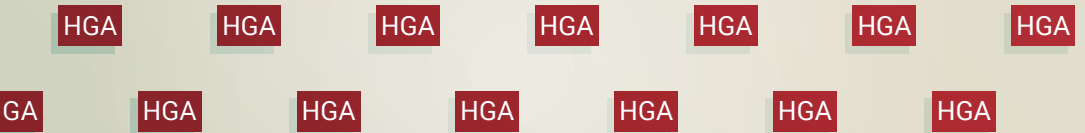
An enzyme called HGD breaks down HGA  
and removes it from the body.

# In a person **with** AKU, the HGA can not be broken down as it should be.

The HGD enzyme is missing, this means people have too much HGA. HGA causes the symptoms of AKU.



HGA → Faulty/No Enzyme → Too much HGA



The HGD enzyme is made in the wrong shape and so can not do its job of breaking down HGA.

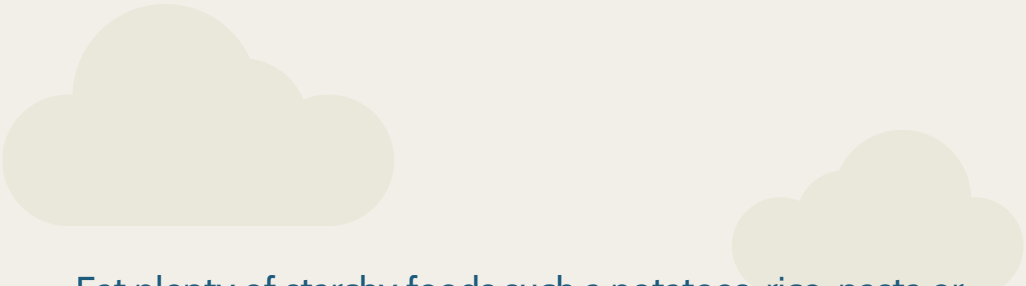

Since HGA comes from protein, it may be helpful to understand about protein in your diet. At this time there is no need to change what you eat. A healthy balanced diet is more important.



# A healthy diet for children



Choose a variety of different foods from each food group, to help the body get everything it needs to stay healthy and keep you fit.

- 
- Eat plenty of starchy foods such as potatoes, rice, pasta or bread, choosing wholegrain where possible. These foods give you energy.
  - Eat at least 5 portions of a variety of fruit and vegetables every day. A portion is the size of your hand.
  - Have 3 dairy foods every day for your bones and teeth.
  - Eat some beans, pulses, fish, eggs, meat and other proteins at each meal.
  - Choose olive or rapeseed in cooking oils.
- 

- Choose water, lower fat milk, sugar-free drinks. Limit fruit juice and/or smoothies to a total of 150ml a day which counts as 1 of your 5 a day.



# Physical activity and exercise

It is important to keep your joints moving without putting too much strain on them. Swimming is a good way to do this.

Try to avoid what is called 'high impact' exercise such as rugby or karate. These can put stress on your back and joints.

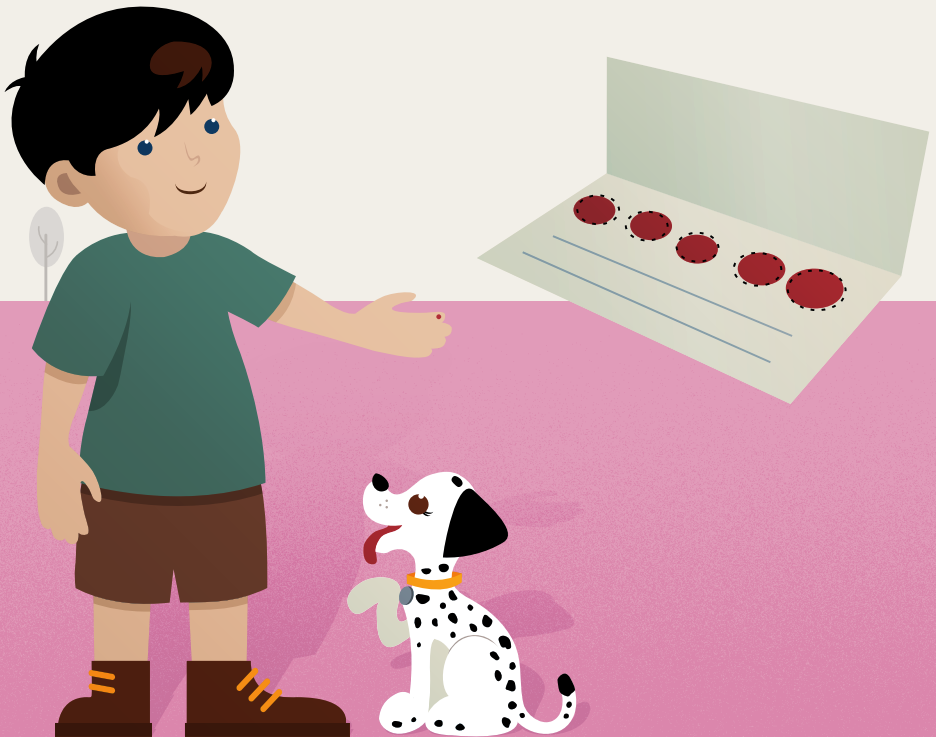
Being physically active will help you.








Up to the age of 16 you will be looked after by your GP and a doctor at a local hospital. You may have blood tests, x-rays and special scans.



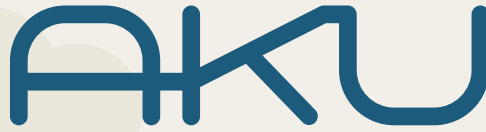
At 16, your doctor can refer you to the National Alkaptonuria Centre in Liverpool.

You will have an annual visit to the centre and meet a team of AKU experts who will look after you. You will be at the centre for up to 4 days and your mum & dad can go with you.



A cartoon illustration of a young boy with black hair, blue eyes, and a slight smile. He is wearing brown shorts and brown boots with orange laces. He is holding a large, light-colored rectangular sign in front of his chest with both hands. The sign contains the text "Where else can you find help" in a bold, blue, sans-serif font. The background is a simple landscape with a pink ground, three small grey trees on the left, and two light green clouds in the sky.

**Where else  
can you  
find help**

The logo for AKU, consisting of the letters 'A', 'K', and 'U' in a stylized, rounded, blue font. The 'A' and 'K' are connected at the top, and the 'U' is separate. The background features light green clouds.

+ Alkaptonuria Society →

AKU Society - a charity that helps and supports people with AKU.

They organise events where you can meet others with AKU and make new friends.

When you are 16, they will help you attend the National AKU Centre.

A member of the AKU Society team will support you at the centre and can visit you at home to help prepare you for your visit.



# Further information



AKU Society  
[www.akusociety.org](http://www.akusociety.org)



Climb  
Children Living with Inherited Metabolic Diseases  
[www.climb.org.uk](http://www.climb.org.uk)



Genetic Alliance UK  
[www.geneticalliance.org.uk](http://www.geneticalliance.org.uk)

The Robert Gregory   
National AKU Centre

Secretary of Clinical Director: 0151 706 4197

Managerial Lead: 0151 706 4289

Mobile: 07785447421

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The production of this booklet has been sponsored by  
Swedish Orphan Biovitrum Ltd.



# How to get in touch

The logo for the Alkaptonuria Society (AKU) features the letters 'AKU' in a bold, blue, sans-serif font. The 'A' and 'K' are connected at the top, and the 'U' is a simple, rounded shape. The logo is centered between two light green, stylized cloud shapes.

+ Alkaptonuria Society →

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