An introduction to Alkaptonuria (AKU)
What is 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.
Diagnosis of AKU
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.
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.

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.
People who are carriers for AKU do not have AKU themselves and the faulty gene does not cause a problem to them.
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.
The science bit... Protein explained
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.
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.
The body uses these amino acids for growing, building up muscle and helping the body stay healthy.
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 cannot 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.

The HGD enzyme is made in the wrong shape and so cannot 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.
Management... a healthy diet in AKU
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.
Monitoring
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.
Where else can you find help
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

Climb
Children Living with Inherited Metabolic Diseases
www.climb.org.uk

Genetic Alliance UK
www.geneticalliance.org.uk

The Robert Gregory
National AKU Centre

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