What is Alkaptonuria?
Alkaptonuria, also known as AKU or Black Bone Disease, is a rare inherited condition that causes the cartilage on joints to become black and brittle.

It’s caused by a problem with a gene and leads to a build-up of a substance called homogentisic acid (HGA).

HGA builds up in connective tissue, especially cartilage, and over time causes it to darken and become brittle. This discolouration is known as ochronosis. This leads to early onset osteoarthritis and means multiple joint replacements are often needed.

HGA is secreted in large quantities in the urine, making the urine turn dark when left exposed to air. Because of this AKU can also be called Black Urine Disease.

One of the earliest signs of the condition is dark-stained nappies caused by the HGA in the urine. If this sign is missed, the disorder may go unnoticed until adulthood, as there are usually no other noticeable symptoms until the person reaches their early 30s.

As a result of the ochronosis a blue/black tinge to the ears can appear. There may be dark spots on the whites of the eyes. Ear wax will be darker and dark sweat can stain clothing.

The deposits of HGA can make people more prone to kidney stones, bladder stones, gall stones and prostate stones.

Deposits around heart valves can cause them to turn brittle and black. Blood vessels, veins and arteries may also become stiff and weaken. This can lead to heart disease and may require heart valve replacements.

People with AKU have a normal life expectancy. They may, however, experience symptoms, such as pain and reduced movement in the joints, which considerably impact on quality of life.
Kidney and prostate stones
Black spots in the eyes
Blue/grey tinge to the ears
Early onset osteoarthritis
Hardening of blood vessels in the heart
Kidney and prostate stones
Black Urine
AKU is a lifelong condition and there is currently no cure or specific treatment, although painkillers and joint replacement surgery can relieve symptoms.

There is a medicine called nitisinone which is showing promising results and research into this drug is ongoing. Nitisinone is offered ‘off-label’ at the National Alkaptonuria Centre at the Royal Liverpool University Hospital. This means it isn’t licensed for use in AKU but doctors believe it can treat AKU effectively. Some lifestyle changes may help you manage your AKU and cope with the symptoms.

**Diet**

It is important to follow a healthy balanced diet. A low-protein diet can also be useful in reducing the risk of potential side effects of nitisinone during adulthood. Your doctor or dietitian can advise you about this.

**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 as these can put stress on your back and joints. Regular gentle exercise can actually help by building muscle and strengthening your joints. Exercise is also good for relieving stress, losing weight and improving your posture, all of which can ease your symptoms.

**Pain relief**

Pain management strategies can be discussed with your doctor. There are a variety of medications and complementary therapies as well as support groups that can help manage symptoms.

**Emotional support**

A diagnosis of AKU can be confusing and overwhelming at first. Like many people with a long-term health condition, those who find out they have AKU may feel anxious or depressed. You can talk to your GP and the AKU Society can offer support and information to patients, carers and their families.