Alkaptonuria

In Alkaptonuria (AKU) there is a deficiency of an enzyme which prevents homogentisic acid (HGA), a naturally produced substance, from being broken down. This causes an accumulation of HGA in the body. It is deposited in the connective tissue causing a bluish-black discoloration (ochronosis) and brittle and weak bones and cartilage leading to severe arthritis. HGA is also excreted in large amounts in the urine causing the urine to turn dark when exposed to air. AKU is a rare metabolic disorder affecting 1 in 250,000 people worldwide.

Synonyms
Alternative names for this condition are:

- AKU
- Black Bone Disease
- Black Urine Disease

Further information about this condition is available from Climb.

Disclaimer
Please read our disclaimer and information on data protection.

Updated 12/05/17