Need to talk? Call us 0845 2412173 Monday to Friday 9am to 5pm

Climb™

Supporting those affected by Inherited Metabolic Disorders

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 discolouration (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

Telephone: **0845 241 2173** Freephone: **0800 652 3181**
Email: contact@climb.org.uk Website: www.climb.org.uk