Liverpool Clinical Laboratories

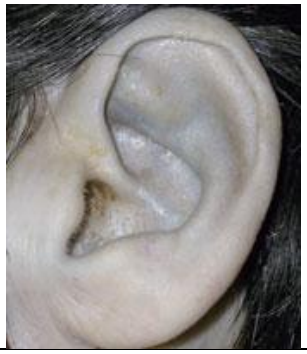
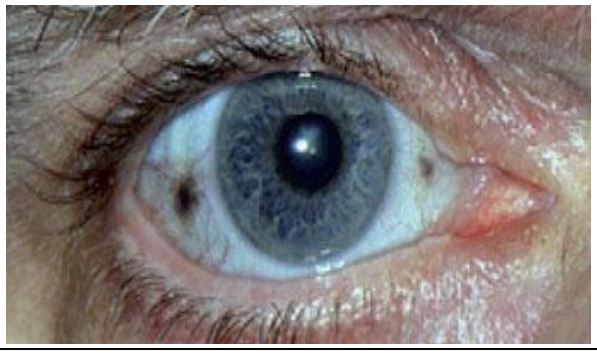
FACTSHEET

Alkaptonuria

pronounced: al-kapt-o-nuria

Also known as: black urine disease, black bone disease, AKU or alcaptonuria.

Affects: One in 250,000 people every year. There are currently 66 people in the UK living with the disease.

What is AKU?	AKU is very rare genetic condition caused by a mutation of the gene homogentisate dioxygenase (HGD). AKU prevents the body from fully breaking down two protein building blocks; tyrosine and phenylalanine, which results in the build-up of a chemical called homogentisic acid (HGA).
Symptoms:	Over time, HGA can cause a number of problems for patients, including: - change in skin colour - joint and spinal damage - heart problems and kidney stones. Black urine stains in a baby's nappy are an early indicator of the disease and blue-black discolouration of the eye and ear cartilage is common in adults.  
Treatment:	Alkaptonuria is a lifelong condition and there's currently no specific treatment or cure other than pain management medication and joint replacements. Research takes place at Liverpool Clinical Laboratories which is the international AKU centre, based at The Royal Liverpool Hospital. Patients visit the centre every year for advice and guidance on managing their condition and to monitor disease progression. Patients also receive the drug nitisinone, which is being researched as the first potential medication to stop the progression of the disease.
Research:	**ANNALS EXPRESS: The effect of nitisinone on homogentisic acid and tyrosine: A two-year survey of patients attending the National Alkaptonuria Centre, Liverpool** Anna Milan, Andrew T Hughes, Andrew S Davison, Jean M Devine, Jeannette L Usher, Sarah Curtis, Milad Khedr, James A Gallagher, Lakshminarayan Ranganath
Find out more:	For support living with AKU visit www.akusociety.org For more information on AKU research and clinical trials, please contact: anna.milan@rlbuht.nhs.uk or andrew.davison@rlbuht.nhs.uk, Consultant Clinical Scientists, Liverpool Clinical Laboratories.

for the science inside us