



Liverpool University Hospitals

NHS Foundation Trust

Robert Gregory National Alkaptonuria Centre

Patient information



Royal Liverpool University Hospital

PIF 1630 V2

What is AKU?

Alkaptonuria (also known as AKU or black bone disease) is a rare inherited condition that causes the cartilage of bones to become black and brittle. This can lead to early onset of arthritis in the spine and problems with weight bearing joints.

AKU results from a deficiency in one of the enzymes that break down the amino acid tyrosine. Amino acids are the building blocks of protein and tyrosine is present in the body from both dietary protein and body proteins (such as muscle).

Because of this enzyme deficiency, a chemical called homogentisic acid (HGA) accumulates in body fluids and tissues. This accumulation of HGA causes the discoloration of tissue (also known as ochronosis). It can also cause the urine to turn black. Blue/black tinge to the ears and dark spots in the eyes could be noticed too.

Diagnosis

One of the earliest signs of the condition is dark-stained nappies caused by HGA in the urine. If this is missed AKU can go unnoticed until adulthood as there are usually no other noticeable symptoms until the early 30s when joint symptoms typically appear.

This occurs more in the weight-bearing joints (the spine, hips, knees, and shoulders). Diagnosis of AKU is usually confirmed by undergoing specialised laboratory testing of urine to confirm the presence of HGA.

Symptoms

People with AKU can experience some of the following, although not everyone will experience all of them:

- Arthritis – in weightbearing joints (spine, hips, knees, and shoulders).
- Bones become more brittle.
- Heart valves become blackened, brittle and narrow.
- Stones formation in the gall bladder, kidneys, and prostate.
- Tendons, muscles, and ligaments damage.
- Hearing problems.
- Dark pigment in the eyes and ears.
- Urine becomes dark after exposure to air.

AKU can be managed successfully using the drug nitisinone. Restriction of dietary protein is required to mitigate the risks from rising tyrosine levels in the blood after starting nitisinone.

Robert Gregory National Alkaptonuria Centre

The National Alkaptonuria Centre (NAC) was set up in 2012 to provide patients with a single, specialist centre for the management and treatment of AKU at the Royal Liverpool University Hospital.

The NAC is named after Robert Gregory who was an AKU patient and co-founder of the AKU Society along with Professor L Ranganath.

Robert realised the lack of provision for AKU patients and was responsible for setting up the AKU Society for patients as the world's first charity exclusively dedicated to the disease.

The NAC provides:

- A single national centre of expertise for comprehensive assessment of AKU patients.
- Expert one-stop care – monitoring and treatment of the disease by an experienced multidisciplinary team.
- Effective disease management - modifying therapy and use of the drug nitisinone.
- Access to a range of expert surgical and medical assessments.
- Reviews of AKU patient health problems (if required).
- Holistic management plans for patients and their local healthcare providers.
- On-going expert advice to maximise the efficiency of local clinical management.

Referral

Patients who are over 16 and have a proven diagnosis of AKU are eligible to attend the NAC. Referral will usually be through a GP.

Management and visits to the NAC

Following referral, the first visit to the NAC in Liverpool spans a wide range of assessments and therapy taking place over four days. Patients visit together with a small group of other AKU patients.

Depending on individual requirements, patients are then offered attendance to the NAC on an annual basis (which usually lasts for three days) as part of their on-going follow up.

The NAC provides a comprehensive report of results, investigations and recommendations for the patient, their GP and local services. Where appropriate, suggested referrals will be highlighted in the report for effective ongoing patient care.

Assessments at the NAC typically consist of consultations with AKU team clinicians and other specialist assessments including:

- Blood tests.
- Radiology (e.g. MRI, X-ray, bone scan).
- Physiotherapy.
- Gait (walking studies).
- Joints (Rheumatology).
- Eyes (Ophthalmology).
- Ear, nose and throat (ENT).
- Cardiology.
- Photography.
- Psychology (Psychometrics).
- Diet consultation.
- Pain management.

The AKU Society

The AKU Society is a charity that supports AKU patients and funds research into finding treatments. The AKU Society works closely in partnership with the Royal Liverpool University Hospital.

The society provides additional support including:

- Working alongside the NAC liaising with local services for referrals and coordination.
- Meeting with patients during the NAC clinics.
- Emotional support and care to patients, their family and carers during the NAC clinics and between visits.
- Publicising and raising awareness of AKU and the NAC to local GPs and other medical professionals.

The NAC and the AKU Society work in partnership to continually review and improve the NAC through research, patient and clinician surveys and interviews.

Feedback

Your feedback is important to us and helps us influence care in the future. Following your discharge from hospital or attendance at your Outpatient appointment you will receive a text asking if you would recommend our service to others.

Please take the time to text back, you will not be charged for the text and can opt out at any point. Your co-operation is greatly appreciated.

Further information:

NAC

**4th Floor Edwards Building
Royal Liverpool Hospital
Prescott Street
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**(Monday - Friday 9am – 4pm)
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AKU Society

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All Trust approved information is available on request in alternative formats, including other languages, easy read, large print, audio, Braille, moon and electronically.

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