

ALKAPTONURIA (AKU)



WHAT IS AKU?

In AKU, an inherited gene mutation causes a compound called homogentisic acid, or HGA, to build up in tissues, bones, and cartilage.

This can cause joint pain, heart problems, and brittle bones. Most people need several joint replacements over their lifetime, in addition to surgery for heart problems.

The earliest and most obvious signs of AKU are black urine and colour changes to the eyes and ears. Around 1 in every 250,000 people are affected.

BUILDING A TEAM

Nitisinone was already used to treat a condition called Tyrosinemia type 1 (HT1). Both HT1 and AKU are caused by gene mutations which affect the breakdown of a compound called tyrosine. Because of the similar disease mechanism, researchers hypothesised that nitisinone could be repurposed for AKU.

The AKU Society and its director, Nick Sireau, were instrumental in forming the DevelopAKUre consortium. The consortium ran clinical trials of nitisinone across Europe involving over 100 patients.

The successful trials and unwavering determination of the patient group and consortium led to nitisinone being approved for use in AKU in the UK, and EU.

STAYING MOTIVATED

DEALING WITH SET BACKS

A previous study into nitisinone for AKU was unsuccessful, making it difficult to get started. However, the innovative DevelopAKUre consortium developed a new trial design which suited patients and regulatory requirements better and encouraged funders. The trial was a success.

NEGOTIATING WITH INDUSTRY

Drugs that are candidates for repurposing often have a marketing authorisation held by a pharmaceutical company. The consortium needed permission from the pharma company to use or test nitisinone. Nick built personal connections with management and worked tirelessly to secure their support.

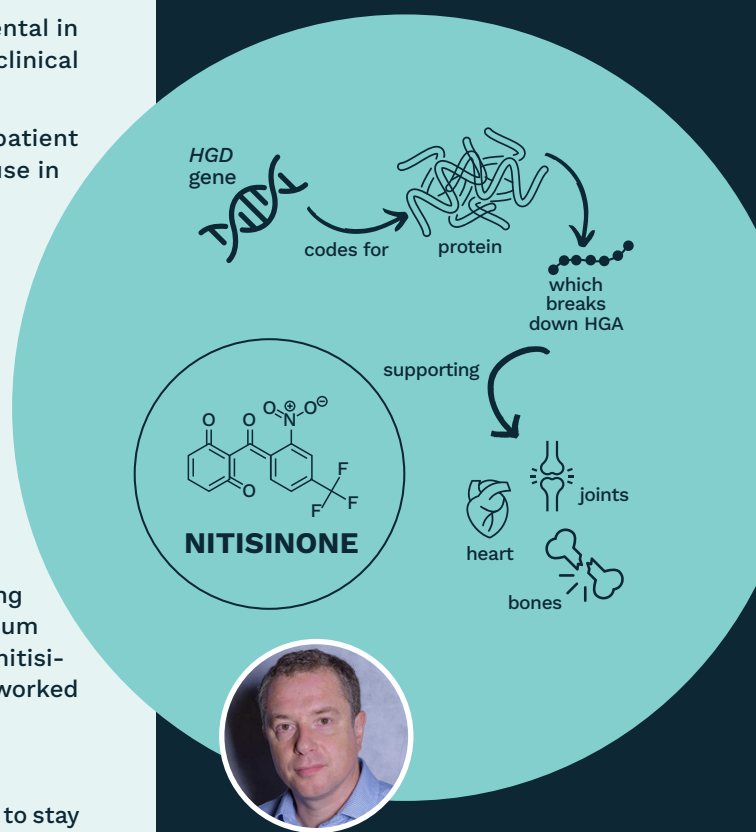
PEER SUPPORT

As a parent of two children with AKU, Nick relied on peer support to stay positive. Through the AKU Society, he connected with other families and patients. Talking to others in a similar position to yourself can be helpful.

AT A GLANCE

- AKU causes a compound called HGA to build up in cells.
- This causes joint and heart problems, often needing surgery. Nitisinone was already used in a similar condition called HT1.
- The DevelopAKUre consortium conducted clinical trials to see if it worked in AKU as well.
- Nitisinone is now approved for AKU in the UK, and EU.

HOW IT WORKS



SPECIAL THANKS TO NICK SIREAU

Nick has two son's with AKU.

He runs the AKU Society and helped set up a UK treatment centre. He continues to support patients and families to manage symptoms and access nitisinone.



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