AKU
Alkaptonuria Society

20 YEAR IMPACT REPORT

2003-2023

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INTRODUCTION

What is AKU?

Alkaptonuria (AKU), also known as Black Bone Disease, is an ultra-rare genetic condition which causes severe damage to the bones, cartilage, and tissues of those affected. AKU normally affects one in every 250,000 people.

AKU stops patients' bodies from breaking down a chemical called homogentisic acid (HGA) which the body naturally produces during the digestion of food. This causes HGA to build up over time and leads to black and brittle bones and cartilage, ultimately resulting in severe early onset osteoarthritis.

Symptoms of AKU:
- Black urine
- Osteoarthritis
- Black spots in the eyes
- Discolouration of ears
- Heart complications

The AKU Society

The AKU Society is the only charity supporting people and their loved ones living with AKU. We work to transform the lives of AKU patients; through patient support, community building, and medical research.

Based in Cambridge, we work to support all AKU patients across the UK. We work closely with the University of Liverpool to drive research into understanding, treating, and curing the condition. Over the past 20 years, we have invested over £1 million into researching AKU. We also work to establish AKU sister societies around the world and have helped 11 groups to form to ensure that all patients are able to access support and care for their condition.

A MESSAGE FROM OUR CEO

“This year we celebrate the 20th anniversary of the AKU Society, marking two decades of relentless dedication to AKU. I am both honoured and humbled to share this milestone with you. The impact we have made together against this ultra-rare disease is nothing short of extraordinary.

Two of our key successes include our DevelopAKUre consortium, which resulted in the approval of nitisinone across Europe, and the founding of the Robert Gregory National AKU Centre in Liverpool, named after the co-founder of the AKU Society and led by inaugural director Professor Ranganath.

Our global network of AKU patient groups has expanded, with 11 groups established around the world. We have successful united the AKU community from all corners of the world, offering them the support and information they need. As we look back on these 20 years, we have countless stories of resilience, hope, and determination from our community. Together, we have made a profound impact on the lives of those affected by AKU. The journey ahead is filled with exciting possibilities, and with your continued support, we will strive for even greater accomplishments in the years to come.

Thank you for supporting our journey and helping us to become a beacon of hope for those living with AKU. Here's to the next 20 years of progress, discovery, and transformation. Together, we can achieve a future where AKU is no longer a burden, but a condition that can be managed and treated effectively”.

Nick Sireau

Nick Sireau
Chair and CEO, AKU Society

MEDICAL RESEARCH

AKU is an iconic disease, as it was the first disease to be identified as inherited by Sir Archibold Garrod in 1902. The ultimate aim of the AKU Society is to cure the condition.

Since founding in 2003, we have invested over £1 million into researching the condition. Working closely with the University of Liverpool (UoL), we have a proven track record of conducting world-leading research into understanding the condition and developing potential treatments and cures for the condition.

Research at UoL has been led by Professor Lakshminarayan Ranganath, Professor Jim Gallagher, Professor George Bou-Gharious, and Dr Brendan Norman.

Over the past 20 years our collaboration with UoL has resulted in the university becoming a global leader for research into AKU. Some of our most significant breakthroughs include, developing an AKU mouse model, proving that nitisinone could be repurposed to treat AKU, and identifying a link between AKU and cataracts and Parkinson’s disease.

Our research is continuing to increase our understanding of AKU and is aiming to develop potential treatments and cures for the condition. Our current projects include a tyrosine reduction co-therapy study, gene therapy, and mRNA therapy.

We have also built an extensive scientific research network across the world who we work with to drive our understanding of AKU forward and collaborate with to develop new treatments and cures. We have held 13 international scientific conferences where we invite our partners to present their research and discuss how we can collaborate moving forward.

NATIONAL AKU CENTRE (NAC)

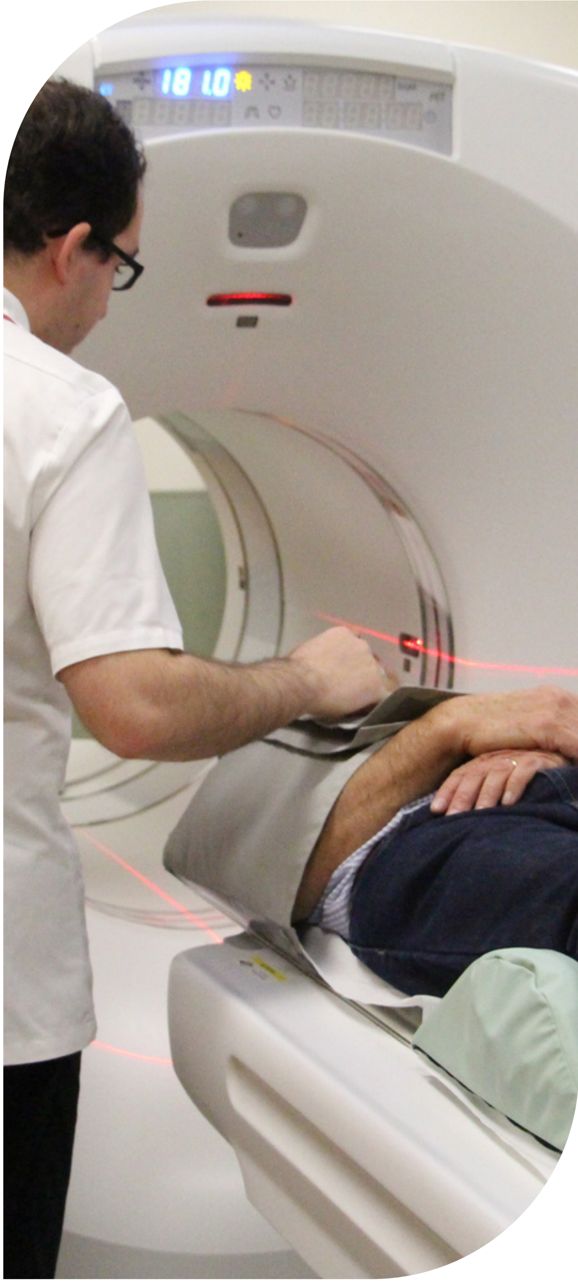
In 2012, we lobbied NHS Highly Specialised Services to establish the National AKU Centre (NAC) at the Royal Liverpool University Hospital. UK AKU patients attend the NAC annually and receive world leading treatment and care.

Under the guidance of inaugural director Professor Ranganath, the centre soon became a world-leading hub for treating AKU and assisted the UoL to conduct ground-breaking research into the development of treatments and cures for AKU.

One of the key achievements of the NAC has been providing patients with access to nitisinone treatment. This treatment has been proven to reduce the toxic-acid that causes AKU by 99.7%, effectively slowing and even halting the progression of the disease. Thanks to the NAC and nitisinone treatment, AKU patients have been able to maintain a higher level of mobility and independence.

Current NAC clinical director Dr Milad Khedr said: 'AKU is such a rare disease that patients were left to fend for themselves before the NAC was created. It has completely changed their lives with a one-stop shop where they can access world experts in the field and a life-changing treatment.'

June 2022 marked the 10th anniversary of the NAC which has now provided life-changing support and treatment to 95 patients.



PATIENT SUPPORT

Providing support to all people living with AKU and their loved ones is a key aim of the AKU Society. Over the past 20 years we have worked to ensure that no one has to face their AKU journey alone.

Our patient support is led by Lesley Harrison, Head of Patient Support and Welfare, who reflects back on the past 20 years of patient support at the AKU Society:

"As a patient-led support group, we strive to empower all those affected by AKU; helping them to understand and manage their condition and build a strong and supportive community. The patient support we offer is vitally important and at the centre of the work we do. We are the main point of contact for patients and the link between local healthcare providers. Central to the support we provide is ensuring that all are treated fairly, have equal access to treatment, and empowering them to shape the vision of the AKU Society.

Over the past 20 years, our patient support has evolved to meet the changing needs of the patient group. The depth and nature of our support has grown immensely to fill the socioeconomic gaps and barriers left unaddressed by the broader healthcare system. We aim to provide a holistic approach to patient support which includes providing financial assistance, care navigation, practical help with non-medical needs, education and resources, and access to counselling services.

We are committed to ensuring the patient voice remains central to the work of the AKU Society and are constantly looking for new ways in which we can support our community to manage the impacts of AKU more effectively".

PATIENT TESTIMONIES

Brenda, UK

“I was diagnosed with AKU in 2002, following my first joint replacement surgery. I remember walking into a sea of faces with the surgeon saying the operation had gone well but, at that moment I feared cancer, he told me in 20 years of operating he had never seen it but he believed I had a rare disease called AKU. He sent me to a rheumatologist to take tests to confirm the diagnosis and was eventually given the only treatment at the time, vitamin C.

I found out about the AKU Society purely by chance as the National AKU Centre (NAC) is a mere 15 miles from where I live! In 2014, my husband John was reading the newspaper when he came wafting the paper excitedly to show me. It was an article that would change my life. The article explained how a father had given up his job to fund research into a rare disease affecting his two sons, it was our very own Nick Sireau. Nick joined forces with Professor Ranganath and AKU patient Robert Gregory to found the AKU Society in 2003. I contacted the newspaper who put me in touch with Lesley, Head of Patient Support and Welfare at the AKU Society. It felt like I had won the lottery, I had access to a wealth of information and support on AKU and was invited to the upcoming AKU patient workshop in Liverpool which was my first time meeting other AKU patients and then attended the National AKU Centre for the first time in 2015.

Each year I attend the NAC with the special friends I met on my first visit, Kevin, Freddy, and Dave. Through all the conferences and workshops, I have made so many friends across the AKU community from all around the world. Having all these friends who know the struggle with daily life and speaking with them helps to lessen the burden. Meeting and listening with the AKU community is a great help and I am so happy to be part of this community to help others”.

Carolyn, Canada

"I first connected with the AKU Society in 2007. I was able to meet with Robert Gregory and Professor Ranganath in Liverpool and that same year I attended my first patient workshop. For the first time ever, I met others who understood what I was going through or what I may eventually have to cope with.

Through the AKU Society I began to communicate with other AKU patients from around the world via conferences, workshops, and social media groups. I found the support and encouragement from the community so helpful in coping with my condition. It helped me to feel less alone and answered some of my uncertainties. Understanding what each of us faces and sharing our experiences and personal learnings has been invaluable. I've made some lovely friends through the AKU Society.

The AKU Society has been a wonderful support. They have happily answered so many of my questions and have shared countless resources. The conferences and workshops they organise have been incredibly beneficial. Thanks to the information and support of the AKU Society I was able to obtain nitisinone treatment in Alberta, Canada.

I admire the AKU Society and the professionals that work within it. The strides they have made in the past 20 years have been truly incredible for patients like me. I am very grateful for all their hard work. They bring about hope to so many of us with AKU. Like so many, I have advocated strongly for myself over countless years and with the ongoing support of the AKU Society I have had some positive results.

I am so grateful for the global AKU community. A disease that is not kind has brought about a lot of compassion. There are so many new patients that have joined the conversation about AKU over the years and the one thing that has not changed is the support shown to each other".

Simon, UK

"I first became aware of the AKU Society through conversations online with Robert Gregory, back in the early 2000's. I had been taking part in the US National Institute of Health nitisinone study at the time and, like many others, I was disappointed when the study ended and was inconclusive.

As I connected with the AKU Society they were beginning their own study into nitisinone. Thanks to the determination of Robert Gregory, Professor Ranganath, Nick Sireau, and the rest of the team the DevelopAKUre trials began. I jumped at the opportunity to take part and help out in any way I could.

I was able to attend the NAC which was founded at the Liverpool University Hospital and has become a second home for people living with AKU. I have always looked forward to my annual visit to the NAC. The staff have always been supportive and friendly and I have made many friends through my visits. I am very grateful for all of the medical facilities I have been given access to over the last 13 years and for all the new friends I have made. Sadly, some of those friends are no longer with us but I know how much the AKU Society and NAC meant to them. They will be missed and remembered by the AKU community.

After decades of looking for help from all over the world, little did I know that the answer would be found right here in the UK. Not only has the AKU Society been involved in finding a treatment for AKU, but it has also strived to involve every patient and hold regular events to support and unite the community. They support and follow up on all aspects of AKU, whether it be medical or personal, and have always been there when needed.

I am grateful for everything the AKU Society has done for me and my family over the years and look forward to what the next 20 years has in store".

LOOKING AHEAD

Our ultimate aim is to cure AKU. We are continuing to fundraise and commit funds to conduct medical research into understanding AKU and developing potential treatments and cures for the condition

We are currently working on a number of projects including developing a tyrosine reduction therapy, a gene therapy, mRNA therapy, and researching the link between Parkinson's and AKU. We are also in the final stages of planning a paediatric study which will investigate the age at which AKU begins to inflict damage in children and determine the best age to begin nitisinone treatment to minimise the symptoms children experience as they grow up.

In early 2024 we are planning on launching our global AKU patient registry. Our registry will be open to all AKU patients around the world who will be invited to track the progression of their condition. This will allow our patients to view how their condition has progressed over time but it will also allow us to track the progression and prevalence of AKU across the world. This will provide us with a wealth of information which will accelerate our research into treating and curing the condition while it will also help us to achieve universal access to nitisinone.

ACKNOWLEDGEMENTS

The AKU Society would like to thank all members of the AKU community, our funders, and researchers who have contributed to our work over the past 20 years.

We are extremely proud of the progress we have made into treating and curing the condition, raising awareness within the medical community, and uniting and providing support to the AKU community.

We are looking forward to what we can acheive as a community moving forward.

AKU

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