

IMPACT REPORT 2020



AKU

+ Alkaptonuria Society →

Who We Are

The AKU Society is a patient-led support group. We work to improve the lives of those with alkaptonuria (AKU) and those who support them while researching for a cure.

Our Mission - Transforming the lives of AKU patients through patient support, community building and medical research.

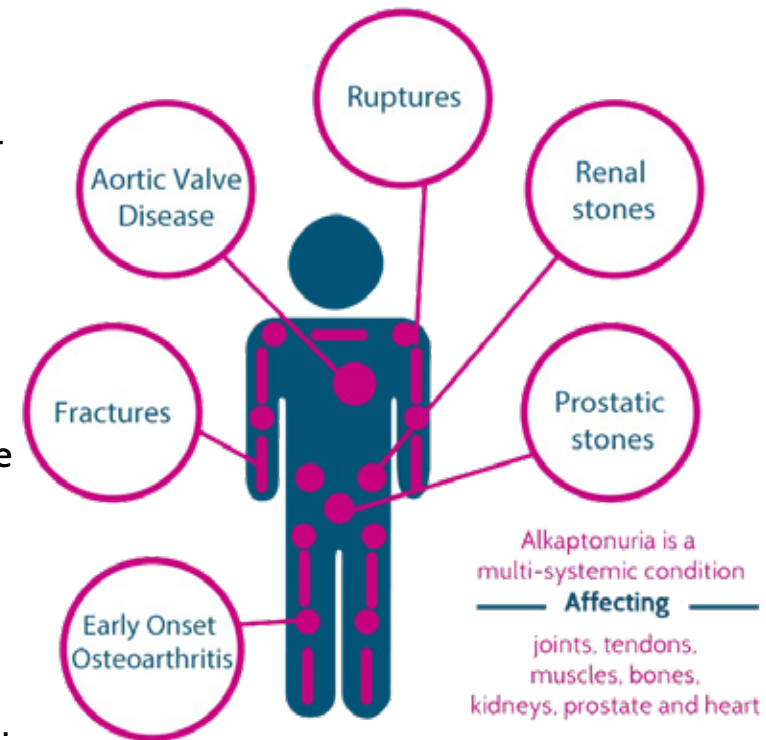


On the cover: The last meeting of the DevelopAKUre consortium in Siena, Italy, 2019



Alkaptonuria is the first identified genetic disease. It causes a build-up of toxic acid in the body that eats away at the patient's bones, cartilage and tissue, turning them black and brittle. This leads to several life-changing disabilities.

Patients develop early-onset osteoarthritis, destroying every joint in the body. The disease is immobilising, which can lead to social isolation, unemployment and chronic pain. Since 2012, the AKU Society has been supporting patients at the National Alkaptonuria Centre (NAC) whilst undertaking groundbreaking medical research into potential treatments. In 2020, nitisinone was licensed as a treatment for use in AKU.



A Message From The Chief Executive

Nick Sireau, CEO and Chair of the Trustees at the AKU Society, takes some time to reflect on a challenging but successful year at the charity. COVID-19 to nitisinone, 2020 has been an interesting.

It's been a tough year for everyone, and the world of AKU is no exception. The pandemic led to the temporary closure of the National AKU Centre (NAC) in Liverpool as well as the scientific labs there. This had a definite impact on our patient care and research, with both being delayed considerably.

Yet it's also been a successful 12 months for AKU, not least because nitisinone was approved as an official treatment for AKU following an eight-year clinical development programme. The European Medicines Agency considered all the data from the SONIA 1 and SONIA 2 trials in record time and gave its positive opinion in September 2020, paving the way for the European Commission's marketing authorisation the following month.

This is major good news for AKU patients in Europe and the UK, but also around the world. For the first time since AKU was initially identified as an inborn error of metabolism by Sir Archibald Garrod in 1901, there is now an approved treatment for AKU based on strong biochemical and clinical results. This is a huge achievement for the DevelopAKUre consortium under the leadership of Prof Ranganath at the Royal Liverpool University Hospital.

The AKU Society's mission is now to help AKU patients around the world obtain access to the drug. We are working with local AKU societies in many different countries to help them make the case to their health authorities to approve nitisinone and provide it to them. This is not an easy task as rules and regulations are very different from country to country. Nevertheless, we consider it as our duty to see this through.

If you are an AKU patient and want to access nitisinone in your country, please do get in touch and we will try to help as much as we can.

As this impact report shows, we also worked on many other projects over the past 12 months. For instance, we launched the Body and Mind programme to support the mental health of AKU patients in the UK. We worked with other patient groups on the Breaking Down Barriers project to help minority communities in the UK affected by rare diseases. We also have big plans for further research to develop a gene therapy as an ultimate cure for AKU.

So despite the Covid pandemic, the future is starting to look brighter for AKU patients. Please do continue supporting us through these challenging times.

“There is now an approved treatment for AKU based on strong biochemical and clinical results.”



Nick Sireau, Chair and CEO of the AKU Society

Our Response To COVID-19

2020 was a difficult year for the AKU Society. Covid-19 changed the way we offered support to those with AKU and how we worked to continue that support.

COVID-19 has changed the way a lot of organisations operated in 2020. Charities have been no different. Luckily thanks to the support of our patients, community and funders, the AKU Society has weathered the storm a lot better than less fortunate charities.

The main impact to our service this year was the unfortunate temporary closure of the National AKU Centre (NAC) in the UK. The NAC is based at the Royal Liverpool University Hospital and sees each UK AKU patient once a year, normally at the start of the month. At the outset of the pandemic, it became clear that the strain on all hospitals throughout the country and the need for space and staff to assist with the fight against the virus meant that the NAC would have to stop seeing patients. Apart from a month in the summer when the restrictions were temporarily lifted, this has been the case since the start of the crisis. To continue the support the NAC offers, together with the NAC we started exploring the use of virtual consultations. This is a way that AKU patients can still talk to Dr Milad Khedr, the clinical lead of the NAC, and keep track of the progression of the disease and the use of nitisinone.



Everyone's mental health was negatively affected by the lockdown and the anxiety associated with the virus. To combat this, we embarked on an innovative project designed to offer all AKU patients in the UK access to free virtual counselling. This project is covered in more depth on page 9. Research has continued as much as possible during the COVID-19 pandemic.

Although the situation was undeniable grim across the world, we learnt that nitisinone had received a licence for the treatment of AKU in Europe, which is potentially life-changing for AKU patients. Since then, we have been working with partners and governments to give AKU patients access to the drug. To learn more please head to page 7. Other research into an ultimate cure and treatments has continued and is currently being planned. To learn more, head to the future section on page 13.

The AKU Society office had to shut its doors temporarily in early March. Since then, the team has been working just as hard from their home offices. Due to this, the office phone number

has not been used as a way of contacting people. Although based at home, we continue to fight hard for the rights of AKU patients to access the support they deserve while championing ground-breaking research.

“The AKU Society has weathered the storm a lot better than less fortunate charities.”

Duncan's Story

Patient Dr Duncan Batty tells his AKU story from two diagnoses to 11 years on nitisinone and five joint replacements. Duncan became a trustee of the AKU Society in 2018.

My parents knew that there was something different with me when my nappies turned brown/black. I was child number 4 and none of my elder siblings had colour changing nappies. The family GP sent off samples of both my nappies and urine and the diagnosis came back as alkaptonuria. My parents were told by a Consultant that there was “unlikely to ever be a cure or treatment for AKU unless it comes from treating another disease” and “to treat him exactly the same as their other children”. My younger brother was also diagnosed with AKU as my parents understood the implications of the stained nappies.

Fast forward to 1996, I was 34, married with three children, and had been suffering from lower back pain for several years. I finally went to see a GP but they could only work on what they had in front of them. All my paediatric notes had been lost when I went to university. The GP prescribed a short course of painkillers and physiotherapy for the back pain. I queried whether the pain could be caused by AKU. Like most GPs she had no knowledge of AKU but did write to a geneticist to get information but no further action was taken.

Over the next couple of years I had a change of job and home, and consequently a change of GP. When I went to see the new GP about my back problems and mentioned AKU, he decided that I needed to see a specialist and referred me to Prof Tim Cox at Addenbrooke's Hospital. Prof Cox formally re-diagnosed me with AKU in 2000 and actively managed my treatment. In 2008 he was able to get ethical approval to prescribe nitisinone for

me and I have been on it ever since.

It was only when I attended my first AKU Society patient workshop in 2008 that I realised how fortunate I was in having both a diagnosis and a consultant that actively managed my worsening condition. As a consequence of this meeting I have given my support to the AKU Society whenever I could. I readily took part in the clinical study in 2009 and have attended the National Alkaptonuria Centre since its creation in 2012. I have spoken at several conferences and in 2018 I became a patient trustee of the society.

Even though I have been on nitisinone now for over 11 years, life hasn't been plain sailing. Like a lot of patients in their late 50s I've had various joint replacements; 5 to be exact, with another one waiting in the wings. Some have questioned why I needed the surgery as I've been taking nitisinone for so long now. The easy answer is that the damage had started long before 2008 and that is why the AKU Society and the NAC recommend nitisinone for all adult AKU patients.



“Even though I have been on nitisinone now for over 11 years, life hasn't been plain sailing.”

Our 2020 Highlights

February

Our brave runners raised a fantastic £1,923.75 at the Cambridgeshire Half Marathon. A huge thank you for the amazing effort and supporting our work!



September

We found out the news that the European Medicines Agency (EMA) indicated that the license for use of nitisinone should be extended for treating AKU.



October

We received £45,000 from the National Lottery COVID-19 fund to help us support AKU patients' mental and physical health through the pandemic.



March

The AKU Society team moved to home working because of the COVID-19 pandemic. We began to look at ways to support AKU patients during the pandemic.



October

Along with our counselling service Rareminds, we began to offer free online counselling to all UK based AKU patients who wanted mental health support.



November

COVID-19 couldn't stop Garrod Day, our virtual coffee morning which saw 30 people from all the world over sign in and listen to the latest AKU news.



Nitisinone

A treatment for AKU has been a goal that we have been striving for since we started our journey all those long years ago. In 2020, we finally proved nitisinone was an effective treatment for the disease.

2020 was a landmark year for the AKU Society and AKU patients everywhere.

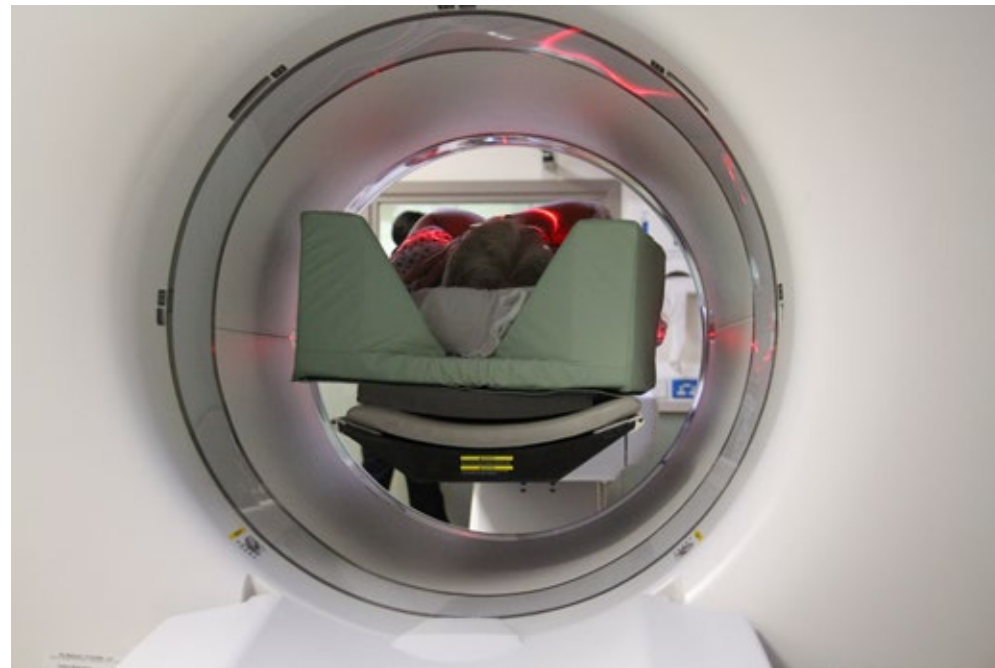
Since we were first made aware of a potential treatment for AKU, a drug called nitisinone, most of our research focus has been proving that the drug can lower homogentisic acid, or HGA, in the bodies of those affected by AKU. As HGA causes the damage to AKU patients' bones and cartilage, nitisinone fast became the biggest hope for sufferers of the diseases. In October, we found out that after years of hard work, dedication, and sacrifice, nitisinone had finally been licensed as a treatment for AKU.

Nitisinone was initially licensed for use in another rare disease called hereditary tyrosinemia type 1 (HT1). Scientists quickly realised that it could also treat AKU as it blocks the same part of the metabolic pathway that causes both diseases.

In 2012 the AKU Society founded a pan-European consortium called DevelopAKUre. This was made up of 12 members, including hospitals, pharmaceutical companies, universities, biotechs and national AKU patient

groups from all over Europe who shared one aim: to prove that nitisinone works to reduce HGA AKU and that it had a positive impact on clinical symptoms.

Together, DevelopAKUre applied for funding through the European Commission's Seventh Framework Programme (FP7) to develop and run the trials that would prove the drug works. The funding secured for this programme included €6 million from the European Commission, with an additional €4 million in co-financing (for in-kind costs such as the drug supply)



Armed with this funding, we launched three studies into the efficacy and safety of the drug. The last, SONIA 2 (Suitability of Nitisinone in Alkaptonuria 2), was designed to determine if nitisinone can be used in AKU to reduce the acid that causes the damage to bones and cartilage and reduce AKU clinical symptoms.

The trial was successful and showed that the drug reduced the acid by over 99%. Due to this, in 2020, the European Medicines Agency (EMA) and the European Commission (EC) extended the license of nitisinone to show it can be used as a treatment for AKU.

This is the first time an effective treatment for AKU has been found. If given early enough, the drug could prevent the symptoms from developing at all. The AKU Society will spend the next few years encouraging countries in Europe and further afield to begin using

DevelopAKUre

Breaking Down Barriers

The AKU Society is proud to be involved with the Breaking Down Barriers Project. The funding we receive from them allows us to offer innovative projects designed to target hard to reach communities.

We continue to be involved with the Breaking Down Barriers Project (BDB), which has now grown into a network of 30 organisations working together to improve the lives of people from marginalised communities, including those from BAME backgrounds. This project enables us to work with other organisations and patient groups to come together and address the challenge of supporting diverse communities and addressing health inequalities. We have received funding from BDB for three quite different projects.

Our first year of funding aimed to revolutionise the support we offered to AKU patients from ethnic minority communities. We delivered this project in stages. Firstly by surveying our patient group to find out what information they wanted and to explore more effective ways to raise awareness of AKU in different communities. We held a focus group session at one of our patient workshops and worked with the patient group to review and update our existing information leaflets. These leaflets were then translated into some of the more common languages spoken within the UK ethnic minority communities that we work with.

“Those family members being offered a test will be given education on the genetics of AKU and how it is inherited.”

The second year of funding has helped us to continue developing relationships with our patient group and to offer enhanced support at the National AKU Centre (NAC) in Liverpool. We created a short video about AKU that can be viewed in GP practices throughout the UK, helping to raise awareness of the condition.

We also integrated BrowseAloud to our website to improve the accessibility and reach of our online information.

The third year of funding received from the BDB project will allow us to offer genetic testing to the extended families of some of our patients who attend the NAC in Liverpool. The need for this project was identified during discussions with AKU patients who attend the NAC. Several have asked about testing partners, children, and other family members, especially those who already have two or more family members diagnosed with AKU or where there is an increased risk of the AKU gene being inherited.

The project involves collaboration between the AKU Society, NAC and laboratory team at The Royal Liverpool University Hospital. A testing kit using a simple finger-prick tool that can be used at home and returned for testing has been developed and will allow family members to identify if they have AKU or carry the AKU gene. This will initially be a pilot project and depending on the outcome, our long-term plan is to offer such testing to the extended members of all our NAC families. Those family members being offered a test will be given education on the genetics of AKU and how it is inherited. The service we plan to provide is about ensuring those who are carriers or even found to have AKU are better informed about genetics and being a carrier and, for those with AKU, they are better prepared for living with AKU, aware of treatment options and how to access the NAC.



Counselling Service

Covid-19 impacted everyone's mental health negatively. For already isolated AKU patients, it got worse. The AKU Society is now proud to be in a position to offer all UK patients free virtual counselling.

In October 2020, we received £45,000 from the Coronavirus Community Support Fund, distributed by The National Lottery Community Fund, to fund a pilot project to provide virtual counselling to AKU patients.

Our 'Body to Mind' project funded virtual counselling for AKU patients and we hired external admin support to contact all of our UK patients it.

Some AKU patients live with anxiety because of the uncertainty of living with the disease. During the COVID-19 pandemic, this anxiety has increased so we started this project to protect high-risk patients and the rest of the patient population.

We worked with the company 'Smart PA' who allocated two members of staff to the project. The AKU Society team met with Smart PA to highlight what was needed from the project and they quickly assigned Natalie & Nige to work with us. We had introductory meetings with them to explain the project, the work of the charity and what their tasks will be. By the end of 2020, they had contacted all UK patients about the service and completed a survey to understand patients' mental health.

"The counselling is being a great help, thank you. They have made a huge difference."

Once we received the funding from the National Lottery in October 2020, we advertised for counsellors on the BACP (British Association for Counselling & Psychotherapy) website. We decided to partner with the non-profit organisation 'Rareminds: Mental Health for the Rare Disease Community'.

Nick interviewed founder Kym Winter, who is the lead counsellor for Rareminds. Kym has over 20 years of experience as a psychotherapist and counsellor, supervisor and consultant. She has presented on the psychological impact of rare disease internationally. She works closely with patient communities and health care professionals to help them define and deliver the most appropriate mental health care for each stage of the rare disease experience.

We chose Kym and her team for their lengthy experience and specific knowledge of counselling those with a rare disease. We started offering counselling from November 2020 onwards and have had regular meetings with Kym on the project.

So far, three AKU patients have undergone counselling. One AKU patient's husband passed away during the COVID-19 restrictions; Lesley Harrison, our Head of Patient Support & Welfare, signposted her to the counselling service, which she then accessed.



Mental Health for the Rare Disease Community

She said this about our counselling service:

"The counselling is being a great help, thank you. They have made a huge difference and helped me through a difficult time. He (the counsellor) asks how I'm feeling with my AKU and even helps me through the frustrations."

Our counselling service has been vital in supporting patients during COVID. We want to say a huge thank you to the National Lottery Fund for awarding this grant and Rareminds for their excellent service.

Fundraising

COVID-19 had the potential to harm our fundraising efforts in 2020. However, thanks to the support of our funders and funds set up during the pandemic, our fundraising continued to be successful.

Fundraising for 2020 focused primarily on COVID-19 support for AKU patients. In April, we received £4,750 from the Cambridgeshire Community Foundation Coronavirus Support Fund to help us fund calls to our UK AKU patient population and funded part of our virtual counselling to UK patients.

Following this, we received £45,000 from the Coronavirus Community Support Fund, distributed by The National Lottery Community Fund, to turn this counselling service into a pilot project. We spent the last quarter of 2020 using external admin support to contact all of our UK patients about the counselling service and teamed up with Rareminds to deliver this service virtually. Rareminds offer mental health services specifically for the rare disease community.

The AKU Society also received €30,000 from the European Joint Programme of Rare Diseases (EJRPD) to fund an international AKU scientific conference. However, due to COVID restrictions, we decided to postpone this conference until 2021. At the meeting, AKU researchers will discuss and plan future research such as gene therapy to cure AKU.

We continued to receive funds from smaller trusts and foundation for projects such as our international patient workshop. We want to thank all of them for their support and patience with delayed projects because of the pandemic.

While COVID did disrupt some of our

community fundraising plans, we did manage to have Juliette, Rob and Dave participated in the Cambridge Half Marathon in March (Dave completed the race around his village). In total, they raised £1,923.75 for the AKU Society – well done! For the rest of 2020, our community fundraising went digital, with virtual coffee mornings planned and our annual Garrod Day went virtual. Garrod Day is an important day for AKU, on the 25th November, the birthday of the late Sir Archibald Garrod, who was an English physician who first described AKU. We celebrate this by hosting coffee mornings all over the world, raising money for the AKU Society.

While we would usually hold face-to-face coffee mornings around the globe, this year, we promoted virtual coffee mornings instead and sent our supporters a virtual coffee morning kit. The AKU

Society held our own coffee morning on the day. We had much to celebrate this year due to the recent European Commission approval for Nitisinone. Over 30 people attended our coffee morning from all around the world. We had supporters from Canada, Greece, Russia, India and other countries .

To round off the year, we ran our annual Christmas appeal. Our appeal was dedicated to raising money for our online counselling service. Overall, we raised £2,691; we would like to thank everyone who donated to the appeal and made it such a success. It was a great way to end 2020.

While 2020 was an unforeseeable and challenging year for many, at the AKU Society we are grateful for our supporter's ongoing support and donations that helped so many AKU patients during the COVID pandemic – thank you.



Our Funders



The National Lottery
Community Fund (Covid-19)

£45,000 – Body to Mind
Programme



Cambridgeshire Coronavirus
Community Fund

£4,749 – Body to Mind
counselling & Covid Support



Cycle Pharmaceuticals

£5,000 – International Patient
Workshop



European Joint Programme of Rare
Diseases

€30,000 – 2021 International
AKU Scientific Conference



Genetic Disorders UK

£3,416.66 – Unrestricted



The Hospital Saturday Fund

£2,000 – SOFIA-Paediatric

Finance

Income



● Research & Support
 ● Voluntary

Expenditure



● Research & Support
 ● Cost of Generating Income
 ● Governance

Incoming Resources	Unrestricted Funds	Restricted Funds	Total Funds
Voluntary Income	£42,105	-	£42,105
Other	£939	-	£939
Research & Support	-	£269,756	£269,756
Total Incoming Resources	£43,044	£269,756	£312,800

Resources Expended	Unrestricted Funds	Restricted Funds	Total Funds
Research & Support	-	£214,425	£214,425
Cost of Generating Income	£28,099	-	£28,099
Governance	£4,428	-	£4,428
Total Resources Expended	£32,527	£214,425	£246,952

The Future

After a year of COVID-19 disruption, the future is looking bright for AKU patients. Now nitisinone is a licensed treatment for AKU, we want to move forward with research to cure the disease forever.

Last year was a challenging one for the world, and AKU was no exception. The National AKU Centre at the Royal Liverpool University Hospital was put on pause while the pandemic raged and the AKU research labs at the University of Liverpool were also temporarily closed.

Nevertheless, our work continued: we took the opportunity to spend time on our plans for the future both on the research front and our global campaign to help AKU patients access nitisinone.

On the research front, we have two key areas of focus: first, developing a gene therapy as the ultimate cure for AKU, and second, developing a means of reducing tyrosine in AKU patients who are on nitisinone to prevent the side effects of the medication.

Preparations for the gene therapy are underway. The scientists at the University of Liverpool, led by Prof George Bou-Gharios and Prof Jim Gallagher, have developed excellent mouse models of AKU that represent the disease effectively. We will be able to test the gene therapies in these models to see if they cure the AKU mice.



We are now at the stage of raising funds so that we can go ahead with these experiments. This is not easy – particularly during the current economic crisis caused by the pandemic – but we are hopeful that we will succeed in raising the necessary funds in the coming months.

The research into the tyrosine lowering therapies is already happening thanks to funding from the AKU Society. We have a new doctoral student called Dominic Rutland who is working closely with our post-doctoral researcher Dr Brendan Norman on this. We will keep you updated about progress.

More broadly, following the ground-breaking approval of nitisinone for AKU by the European Medicines Agency and the European

Commission in autumn 2020, we are now starting a project to work with local AKU patients and physicians around the world to help them access nitisinone. This is not an easy task as rules and regulations in each country are often very different, but it is our long-term goal to make sure that no AKU patient is left behind wherever they live.

Please do keep supporting us as the journey is far from over. Your help is very much appreciated in whatever form it takes.

The AKU Society



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