

# Impact Report 2018



Patient Support  
Community Building  
Medical Research

**AKU**  
+ Alkaptonuria Society →

The AKU Society is a patient-led support group. We work to improve the lives of those with 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.



## Patient Support

Reliable information and support



## Community Building

Uniting AKU patients in the UK and globally

“It feels like your bones are wrapped in barbed wire.”



## Medical Research

To understand and treat the disease

On the cover: Prof Ranaganath, AKU trustee and Clinical Director of the National Alkaptonuria Centre (NAC). Photo: Findacure/Arnab Ghosal



## Simon's Story

Having a rare genetic disorder can be a pretty daunting thing. It can leave a person prone to feeling frustrated and isolated. I know this from personal experience, after spending over ten years searching for information and other people with Alkaptonuria.

Before the AKU society, I found it hard to get the right diagnosis and treatment for the excruciating pain that I was experiencing at the time. Each of my many symptoms were treated separately and, because of my young age, often mistaken for various other conditions.

I was diagnosed as a baby, but my medical notes were destroyed by a fire at my local hospital. As my condition was so rare no one ever looked into it, I didn't know what AKU could do to a person who was unfortunate to be born with it. Thanks to the Alkaptonuria Society and the National Alkaptonuria Centre (NAC), the mystery [of AKU] is becoming a thing of the past. More and more

medical professionals are now aware of this little known about disorder. The AKU Society and NAC have given hope to people who have and are still being diagnosed with AKU.

I am fortunate enough to visit the NAC every year and receive multiple medical investigations. Many of the tests I would not be able to get at my local hospital. I am kept up to date with tests and findings and receive copies of all the reports. These findings are also sent to my medical team back home, which can then treat me accordingly. I can also contact the NAC/AKU Society if any problems arise and this can then be worked out between me, the NAC and my local medical team.

The NAC was set up for the benefit of people with AKU and their families. They have become a sort of extended family to me and others. Without the AKU Society and the NAC, I would still be struggling to get help and treatment and I personally owe them my gratitude.



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“Just because a disease is rare shouldn't mean it's overlooked.”

# The Chief Executive's Statement



“DevelopAKUre was a monumental undertaking. It brought together 13 partner organisations from across Europe and the Middle East.”

Research into AKU progressed fast in 2018. The seven-year DevelopAKUre programme, funded by the European Commission, came to an end, with results expected by September 2019. If positive, we will be applying to the European Medicines Agency for marketing authorisation for the drug nitisinone to treat AKU. This would be the culmination of more than 15 years of work by the AKU Society and would mark a significant milestone in the treatment of the disease.

DevelopAKUre was a monumental undertaking. It brought together 13 partner organisations from across Europe and the Middle East: hospitals, universities, a biotech, a pharma company, and the AKU patient groups in the UK and France. It has been heralded within the wider rare disease community as one of the most pioneering, patient-driven drug development programmes ever seen.

I would particularly like to thank Prof Ranganath, who led DevelopAKUre throughout the seven years and without whom none of this would have been possible. Ranga has shown an exemplary dedication to AKU patients, also through his leadership of the National AKU Centre in Liverpool, which is recognised as the leading global centre for the treatment of AKU.

We had three new trustees join the board of the AKU Society in 2018: Prof Jim Gallagher, who has been leading the AKU basic science programme since 2006 at the University of Liverpool and has been a key driver behind many of the scientific

breakthroughs that have helped us understand and treat AKU better; Dr Duncan Batty, who is an AKU patient and a former pharma executive who has been volunteering for the AKU Society for more than a decade; and Tony Esmond, who is an AKU patient who used to work as a nurse before entering senior management in NHS Wales.

As you'll see in the pages of this impact report, the AKU Society was also active on many other fronts in 2018: from patient support to fundraising and community building.

For this, I'd like to thank our three members of staff: Ciarán Scott, who was Clinical Trials Coordinator in 2018 (and is now Communications Manager), for providing excellent support to European patients during DevelopAKUre; Lesley Harrison, Patient Support Manager, for always being there for our UK patients at the National AKU Centre; and Juliet Rowe, Fundraising Officer, for driving our fundraising plans as we continue to scale up AKU research and support patients.

There is still so much to be done to help AKU patients as they fight this ultra-rare disease. Please continue to support us.

A handwritten signature in blue ink, appearing to be 'Nick Sireau'. The signature is fluid and stylized, with a long horizontal line extending to the right.

Nick Sireau, Chair and CEO of the AKU Society

“Nitisinone is a source of great hope. It might enable me to play golf with my son in ten years’ time, kick a football with my daughter, or even take my wife dancing.”

# Patient Support

The National Alkaptonuria Centre (NAC) is based at the Royal Liverpool University Hospital and was established by the Department of Health (Highly Specialised Services Commissioning Group). It was set up to provide assessments and treatment for anyone over the age of 16 who has been diagnosed with AKU.

The NAC provides one-stop care to assess and detect disease complications and formulate a shared care management plan with local health care providers. Anyone living in England and Scotland diagnosed with AKU is offered an annual review at the NAC and off-label access to the drug nitisinone, as well as advice and support on managing and living with AKU on a daily basis. People living in Wales and Northern Ireland can also access the NAC once the relevant NHS service has agreed funding.

In 2018 we saw 58 patients attend the NAC, 15 returning for their 7<sup>th</sup> visit. As awareness of AKU and the NAC increases amongst health care professionals, more patients are being diagnosed and referred to the service. 2018 was no exception, with five new patients at the NAC. For many, this is the only time they meet other people in a similar situation to themselves and have the opportunity to discuss their condition with experts in the field. All our patient group are asked for feedback on the service and changes where appropriate are made to improve and develop the service for the future. A patient attending the NAC during 2018 said; 'The best part is knowing we are being looked after and monitored by specialists in AKU. They understand what is happening to our bodies and provide us with lots of information and updates on the condition. It gives me peace of mind when the test results come through and I can discuss them with the team at the NAC.'



An NAC patient during a gait analysis examination

2018 saw our 2<sup>nd</sup> year working as part of the Breaking Down Barriers project (BDB). This project allows us to work with other organisations and support groups to come together and share examples of best practice. This project is helping us strengthen our ability to develop inclusive information and services for individuals and families affected by AKU. During the two years on the project, we have worked with our service users to review and change our patient information leaflets. These have now been translated into several languages ensuring we are reaching out to and supporting diverse communities. Funding from the project has enabled us to develop an educational video for the Envisage GP waiting room system. This short health education video raises awareness of AKU and the NAC and targets a broader audience than we might typically reach. Statistics received are positive with the video

receiving a high number of plays on the system during 2018, as many as 873 in September. BDB has funded the addition of 'Browsealoud' to the AKU Society website, making it more accessible and reducing the barriers between the content and all our audiences. We hope to continue working with the BDB project for a third year, developing information and services for the extended families of people living with AKU.

In 2013 we started to offer home support visits as an extension of the NAC in Liverpool. These face to face visits provide an opportunity to discuss anything about the NAC, future visits, nitisinone, diet, blood and urine tests. It can be whatever the patient needs it to be. The value of these home visits cannot be underestimated and can make a huge difference to the life of one of our AKU patients.



NAC managerial lead Hollie and research nurse Giovanna

“I feel that my health is better and I feel confident and have a much more positive attitude for the future.”

62 Patients  
Attended



This year at  
the NAC

Over 40000 mg of  
Nitisinone  
Prescribed



Over 800 Tests  
Completed







An NAC patient receiving an eye test

# Community Building

Over two days 89 people – AKU patients, supporters, researchers and staff – from around the world travelled to Liverpool for our 2nd international patient workshop. They came from as far off as Canada, the United States, Belarus, Jordan, Brazil and La Réunion.

Lesley Harrison, our Patient Support Manager, organised everything. In each hotel room were eight branded AKU chocolates, presented in elegant bags. On arrival, Lesley greeted each delegate and handed them their delegate pack, which was full of extra information for the event. Our three newly designed leaflets ('What is Alkaptonuria', 'How Alkaptonuria is Inherited' and 'The Alkaptonuria Society') were very popular.

From genetics to diet and exercise to cutting-edge research, patients left the event with up to date information and a rounded insight into their disease. Two of the most popular talks from the days were Prof Ranganath's explanation of the

progress of the DevelopAKUre clinical trials and Dr Andrew Jone's experience treating AKU patients' pain and how patients can help themselves alleviate as much as possible.

Day one ended with a fantastic three-course meal, served in the Bluecoat's restaurant. While eating delicious food, delegates got to hear some personal stories. Jim Fish, from Canada, and Flavia Mayrink, from the AKU Society in Brazil, talked about growing up with AKU. Nick Sireau, our CEO, reminded everyone how he threw himself into the AKU Society when both his sons were diagnosed with the disease.

Nick said that we are living through a golden age of AKU research. Never have the medical and scientific worlds been so interested in AKU. At the heart of what we do, though, are still the patients. As they left, many said they felt less isolated. They were not alone with their condition, and there was hope for the future.



Attendees of our 2nd International Workshop

“When we were introduced to the AKU Society, we had a huge weight lifted off our shoulders. It’s not just a charity, it’s a community, where people can reach out to each other.”



A patient asks a question at our 2nd International workshop

We had a busy year raising money. We held events, sold cakes and had a fantastic fundraiser run all for AKU. In May, Jamie Rumbelow ran for the AKU Society up in Edinburgh and he raised £363.85, an impressive amount! We held our annual Stand Up for AKU Comedy Night at the Portland Arms, in Cambridge, which was a huge success. The outrageous Pam Ford hosted and we had five comics perform who had us in stitches. We also held a raffle after the acts were done. Prizes included champagne, handmade fudge, theatre tickets and many other goodies. We sold 155 raffle tickets and raised £469.19. We are now planning our next Comedy Night which will be held in Liverpool in 2019 at the Hot Water Comedy Club.

“I feel more in control of my life now and taken seriously by medical professionals.”

We also held our Garrod Day Coffee Morning in November; this was great fun. We have held this event for years, so the whole office was excited for it to come around again. The day marks the birthday, 25<sup>th</sup> of November, of Sir Archibald Garrod who first described AKU way back in 1902. We sent off coffee morning kits to people to host their own Garrod Day Coffee Morning. Our Fundraising Officer, Juliet, held the coffee morning here in our office in Cambridge. We had cupcakes, Malteser tiffin, pumpkin pie and lemon drizzle on offer, which all sold. We sold some AKU Christmas cards as well to get the office into early Christmas spirit. We also held a raffle on the day, with fantastic prizes on offer, including wildlife park and cinema tickets! We would also like to say a massive thank you to Carol Day for hosting her own coffee morning, raising £112.10, selling delicious cakes and her own books.



An AKU patient shares her thoughts about the AKU Society

Our 2nd International Workshop saw patients come from:



“For me, it is very important to participate in such research because I do not want future generations to go through the same thing as me. I feel it is my duty to contribute to this important research.”

# Medical Research

2018 was a busy year for research at the AKU Society. It was the last year of the SONIA 2 clinical trial (Suitability of Nitisinone In Alkaptonuria 2) and the end of the DevelopAKUre programme. DevelopAKUre was a series of trials run by a consortium of 13 European partners all with one ambition, to prove that nitisinone works in treating AKU.

SONIA 2 was designed to test the effectiveness and safety of nitisinone in treating AKU. We have to wait until later in 2019 for final results; however, the one-year report and a paper published by the team at the NAC have left us feeling positive. Once we receive the results, and if they are positive, we will approach the European Medicines Agency (EMA) for a license to use the drug for AKU across Europe. We hope that if this is successful, it will then start to become available across the world.

With the help of SOBI (Swedish Orphan Biovitrum), we also surveyed 120 AKU patients from across the world. The answers will be used to understand how patients receive a diagnosis and how many doctors they see before they get one, along with how AKU symptoms affect patients. The findings will help us if and when we ask the EMA for a license for the use of the drug in AKU. We are also very close to starting a study, called the SOFIA Paediatric study. This will look at what age the symptoms in AKU begin in children with the disease. This will be used to work out at what age nitisinone treatment could start in children. We hope this will lead to a generation of children growing up without any of the symptoms of AKU.

We also continue to support other research projects and PhDs that continue to advance our knowledge of AKU and the way it affects patients. These studies are not just academic; it continues at the National Alkaptonuria Centre in Liverpool.



AKU PhD student Juliette Hughes

But there is still more research to be done for AKU.

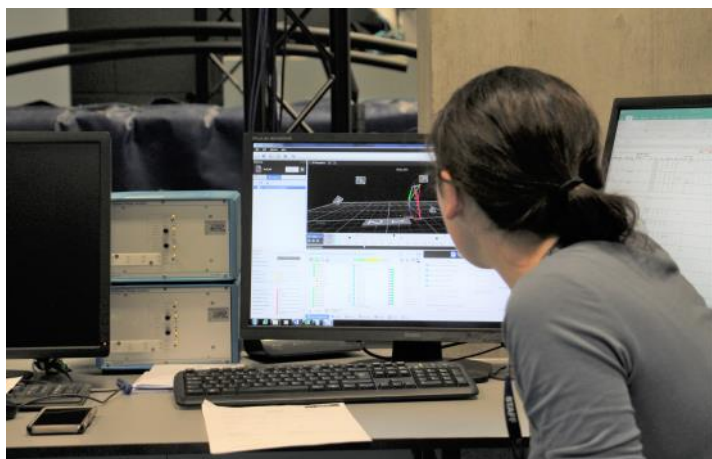
Nitisinone has side effects, namely a massive increase in tyrosine levels and the accumulation of tyrosine crystals in the eyes, which can cause problems. We're also unsure whether nitisinone is safe to provide to children, as there is some evidence that it may cause learning difficulties at that age.

That's why our team in Liverpool has been developing a new treatment that would hopefully resolve these side effects. This new treatment is a plant enzyme that breaks down tyrosine in the gut. If we can prove that this works, this will mean that patients will have a combination therapy: the nitisinone to reduce the homogentisic acid that causes all the damage in AKU, and the plant enzyme to reduce the levels of tyrosine and avoid

the side effects from nitisinone.

Other projects we are looking at include a gene therapy for AKU and a sociological study of AKU. The gene therapy is still at the theoretical stage and we are fundraising to test it in our lab models. It will take many years to develop and will need to prove it's 100% safe before we can enter any human clinical trials. Nevertheless, if successful, it would ultimately be a cure for AKU.

The sociological study is run by a student from Cambridge University who is looking into the experience of being an AKU patient and the practices of AKU treatment. She will be interviewing patients, carers and clinicians at the National AKU Centre as well as the team at the AKU Society. She will look at how existing approaches to treatment can be improved. We look forward to seeing the results.



Gait analysis researcher Hannah Shephard

“I want it for all of us; for the children with AKU and the adults who battle with it every day.”

140 AKU Patients

4 years

3 trial Sites

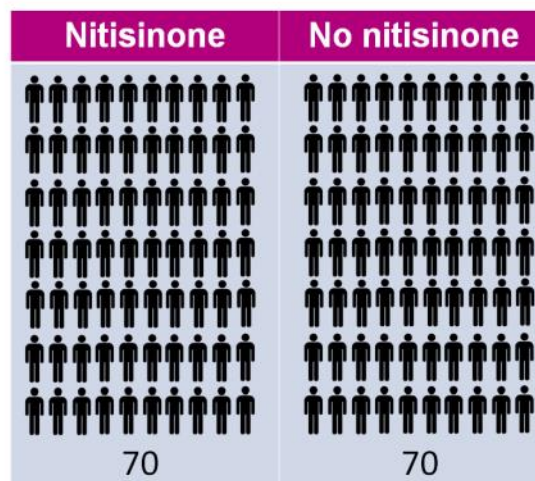
Paris, Piešťany, Liverpool

70 on Nitisinone

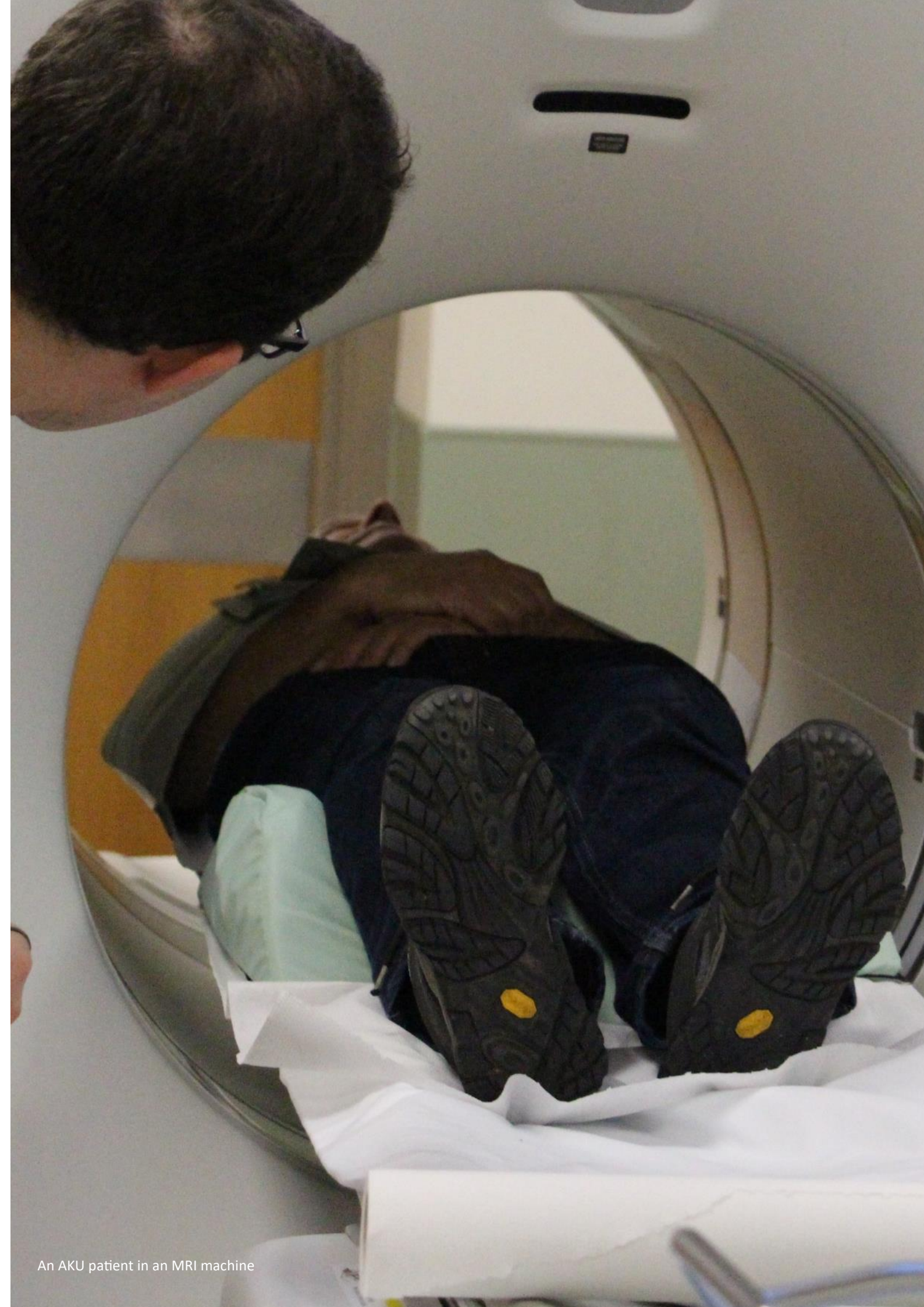
70 not on Nitisinone

DevelopAKUre

SONIA 2







An AKU patient in an MRI machine



Stand up to AKU host Pam Ford

The AKU Society would like to say a massive thank you to our funders, fundraisers and supporters. Trusts and foundations have made up a large portion of our income and have enabled us to host patient workshops and vital medical research. Below is the list of funders who have supported us over the year and what their grant was used for:

**Childwick Trust** – SOFIA Paediatric study

**European Commission** – FP7 DevelopAKUre project. Our clinical trials into the drug nitisinone

**National AKU Centre** – Patient support before, during, and after NAC clinics

**Rosetrees Trust** – Research funding at the University of Liverpool and Lancaster University

**Sylvia Adams Charitable Trust** – Breaking Down Barriers ethnic minority communities project

**Swedish Orphan Biovitrum (SOBI)** - 2nd International Patient Workshop

We are also very grateful to our community fundraisers this year; Jamie Rumbelow, who ran the Edinburgh half marathon; and Carol Day for hosting her fabulous Garrod Day Coffee Morning, raising £112.10. Well done!

A massive thank you to everyone who has donated, volunteered and everyone who has been involved in our events. The AKU Society is extremely grateful for your support.

## Income



## Expenditure



	Unrestricted Funds	Restricted Funds	Total Funds
<b>Incoming resources</b>	<b>£</b>	<b>£</b>	<b>£</b>
Voluntary Income	9,903	92	19,218
Other	498	-	1,178
Research and Support	1,750	214,778	316,693
<b>Total Incoming Resources</b>	<b>12,151</b>	<b>214,870</b>	<b>227,021</b>
<b>Resources Expended</b>			
Costs of generating voluntary income	-	28,270	28,270
Research and support	20,041	165,286	185,327
<b>Total Resources Expended</b>	<b>20,041</b>	<b>193,556</b>	<b>213,597</b>



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