AKU

Alkaptonuria Society

Impact Report 2017

Who We Are

Alkaptonuria (AKU) was the first identified genetic disease, also known as 'black bone 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 a number of 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.

> "It feels as if your bones were wrapped in barbed wire."

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

Medical Research

To understand and treat the disease

> “Just because a disease is rare shouldn’t mean it’s overlooked - patients often lack access to effective medical care and information which leaves them feeling isolated and misinformed. The AKU Society has worked and will continue to work hard to combat this.”

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On the cover: Gábor Barton with a young patient at the Children’s Workshop

Meet Bob Montague

I wouldn't know anything about alkaptonuria if it wasn't for the AKU Society. The surgeons who replaced my aortic valve in 2013 found black spots in my chest cavity. Interested, they sent out for a diagnosis, which meant very little to me. I understood the conjunctive words – 'and', 'but', 'because' – and practically nothing else. But Googling 'ochronosis' led me to your website. I saw pictures of people with symptoms like mine, including the black eye spots which had confused both my optician and an optical cancer specialist back in 2010. It all started to make sense.

I was spurred into action a few years later by seeing Nick Sireau on the BBC. I remembered that the AKU Society existed to help potential AKU patients like me. I spoke to Lesley, your Patient Support Manager, on the phone. She told me how to check for ochronosis in my urine. I checked. I had AKU.

I've just concluded my first visit to the National Alkaptonuria Centre in Liverpool. I've received my first batch of nitisinone, which will stop further degeneration. The help I'm getting from you is second to none. Your enthusiasm is infectious. It gets us all going. Thank you so much.

> "After my heart surgery, I didn't want to go to any more hospitals. It took me a year to find your website and even longer to speak to you. But I'm glad to be getting treatment now."

A Message From the CEO

A Good Year for AKU

The last year has been positive for AKU. First, our international DevelopAKUre clinical trial for nitisinone for AKU has been progressing well: very few patients have dropped out and the focus is now on successfully completing the final year of the trial, which ends in early 2019.

Second, the National AKU Centre (NAC) at the Royal Liverpool University Hospital has continued to deliver an outstanding service for AKU patients. They frequently give us good feedback, and the data from the past five years of the NAC show that nitisinone is having a positive impact on reducing the damage caused by AKU.

Third, we've successfully delivered a five-year programme funded by the Big Lottery Fund to help build online AKU communities and bring AKU patients together. The project ends in April 2018 and has allowed the AKU community to grow online and through social media.

I also want to take this opportunity to say thank you to Oliver Timmis, who left as CEO in July and whom I am replacing, for his seven years of hard work at the AKU Society. Oliver played a key role in the development of the funding proposals for DevelopAKUre and for the NAC. He will be missed.

Planning for the Future

We're now looking to the future and asking ourselves what still needs doing once the DevelopAKUre clinical trial is over. We had an awayday with trustees, scientific advisors and staff where we analysed the challenges that continue to face us.

A key challenge is how to help AKU patients around the world, not just in Europe where DevelopAKUre is taking place. We're frequently contacted by AKU patients from Asia, Latin America or Africa who ask us how they can access treatment and care with specialists who understand AKU.

Having a rare disease is very isolating, even in countries such as the AKU where treatment exists. Having a rare disease in a developing country, where there is little access to good healthcare, is even more difficult.

That's why we're determined to help AKU patients around the world, wherever they are. Working closely with our key partners at the Royal Liverpool University Hospital and the University of Liverpool, we are planning a project that will build stronger links between scientists, clinicians and patients in developing countries such as India and those in the UK.

Can You Help?

That will, of course, rely on whether we can secure funding for such a programme. That's why I'd like to encourage you – whether you're a patient, a carer, a friend or a supporter of the AKU Society – to join in our community fundraising activities. Do get in touch if you'd like to find out more.

So 2018 promises to be another good year for AKU, building on the work of 2017 and previous years. In April, we're organising our second international patient workshop, in Liverpool, bringing together patients from around the world. We also have our international scientific workshop taking place just afterwards, where all the latest research on AKU is presented. Do contact me at nick@akusociety.org if you'd like to find out more.

Nick Sireau, CEO and
Chair of Trustees

Sophia Hajnorouzi, a young patient, with her mother at the Children's Workshop

Patient Support

Because AKU is such a rare disease, patients can often feel isolated. They might never meet another AKU patient over the course of their normal lives. Moreover, medical professionals are often badly informed about AKU, and are unable to supply patients with accurate information. Quality of life and self-esteem can suffer. 71% of our patients report that they are affected by depression and feelings of isolation, and accomplish less than they otherwise would.

This is why the AKU Society holds patient workshops, and this year we held three. Our Children's Workshop at ZSL London Zoo, the first of its kind, brought together young AKU patients and their families from all over the country.

The day started on an exciting note, with the children being introduced to two meerkats. After the meerkats had been fed, the children learned about their own diet from Lesley Harrison, the AKU Society's Patient Support Manager. The next talk, from Liverpool John Moores' Gabor Barton, explained the principles of gait analysis. This was done through a game in which the children had to fly a dragon using their body movements.

Professor Ranganath, Director of the National Alkaptonuria Centre (NAC), also spoke to the families. He explained that young AKU patients will attend the NAC as soon as they reach adulthood, and receive treatment there.

The workshop also saw the launch of our first AKU Children's Booklet. We are very grateful to SOBI for sponsoring it and to Vitaflo for their assistance in its production. The booklet explains, in accessible language, what it means for an individual to have a genetic disease like AKU, and provides information on some of the AKU symptoms. We hope that it will be a useful tool in teaching children and their families about AKU.

Adult patients were invited to two workshops this year. One was held at London's Alexandra Palace, the other at Liverpool's Bluecoat, in order that we might be accessible to as many patients as possible. There were specialist sessions on scientific research, nutrition, online patient care management and gait analysis.

A particular highlight from the Liverpool workshop was the 'patient story' segment. Three AKU patients, Antony, Brenda and Paul, told us about their pain-free childhoods and their recent struggles coming to terms with their condition.

Patients were also able to meet the AKU Society team and ask questions. On the stalls, there were a number of living aids and dietary supplements on display for patients to try for themselves. They were very popular.

We monitor feedback so that we can improve each event. One idea that was raised was the possibility of 'organised socialising' in future, so that NAC patients can meet each other outside of the formal workshop setting. We will be incorporating this into the April 2018 International Patient Workshop, which will bring together patients, families and carers from around the world for two packed days in Liverpool.

> "I got to meet other people with AKU, which was good as it made me feel I am not alone in this. Having AKU has extended my family."

Rory Munro at the patient workshop in March

Dissemination

Dissemination is key to all our work. Not only is it a requirement of our funding from the European Commission, but it often leads to useful new partnerships and new ideas. Furthermore, as we are dealing with a drug that effectively stops production of a harmful acid, it is important for people to get diagnosed and treated before further damage occurs.

Much of our dissemination focuses on the unusual nature of the work we do. We are, after all, involved in a range of unique activities, including the innovative patient-focused DevelopAKUre clinical trials, the dedicated National AKU Centre, and our many patient workshops and home support visits.

Everything we do is achieved through collaboration, which is key to developing new ideas. A great example of this is our DevelopAKUre consortium, which includes several organisations.

> "Our role is to disseminate the project, to spread information about the clinical trials to help recruit new patients, and generally tell the story of how the AKU Society began research into a possible new treatment."

EURORDIS membership meeting in Budapest. Credit: EURORDIS

In May, Oliver Timmis, then our CEO, and Ciarán Scott, our Clinical Trials Officer, travelled to Budapest to represent us at the EURORDIS (Rare Diseases Europe) membership meeting. This brings together patient groups from all over Europe for an annual general meeting and a day of useful talks. This time, the meeting coincided with EURORDIS' 20th anniversary. Talks were focused on the achievements of the last 20 years, and hopes for the next 20.

Oliver was invited to speak in the panel discussion on the role of patient organisations, where he talked about the success we have had in pulling together the DevelopAKUre consortium. He also helped to organise the workshop on practical advice, called the 'Survival Kit for Patient Organisations'. This covered issues like branding, mission statements, staff hire and fundraising. We tried to give smaller patient groups ideas to develop and become more effective patient advocates.


Oliver, then CEO, in Budapest. Credit: EURORDIS

The AKU Society was also represented at the Royal College of Nursing Congress, in May, and at the Best Practice 2017 event for the primary care sector, in October. Both events allowed us to advertise ourselves and the work that we do to healthcare professionals, whom we hope will be more likely to recognise AKU in the future.

Costello Medical, our medical consultants, also presented a poster about NAC research at April's InSPIRe (Innovation in Small Populations Research) conference. We are very grateful to them.

In April, our then Admin and Events Officer, Robin Marshall, gave a talk on local community fundraising at the Findacure Community Fundraising Workshop.
Robin described the events he had been involved in organising – the Garrod Day Coffee Morning and the Stand up for AKU Comedy Night – and gave attendees some useful fundraising tips: think about your target audience when planning an event, and don't forget the importance of event 'after-care' after your guests have left!

> "When planning an event, always think about whom you want to come. My key message is: even if you are a small organisation, it does not mean you have to have a small audience."

NAC–Treatment

The National Alkaptonuria Centre (NAC) is based at the Royal Liverpool University Hospital. Established by the Department of Health (National Specialised Services Commissioning Group), it opened to patients on 18th June 2012. The service was set up to provide evaluation and treatment for patients with AKU.

It provides yearly one-stop care to assess and detect complications, as well as formulating a shared care management plan with local health providers. The NAC gives patients access to the drug nitisinone, thought to be effective in AKU treatment, on an off-label basis. Patients attending for the first time are assessed over four days; those returning for follow-up visits, over three.

> “I always benefit from attending. If there are any health problems, these are always flagged up, so an appropriate medical intervention can take place.”

Richard Smith, NAC patient, with an ophthalmologist

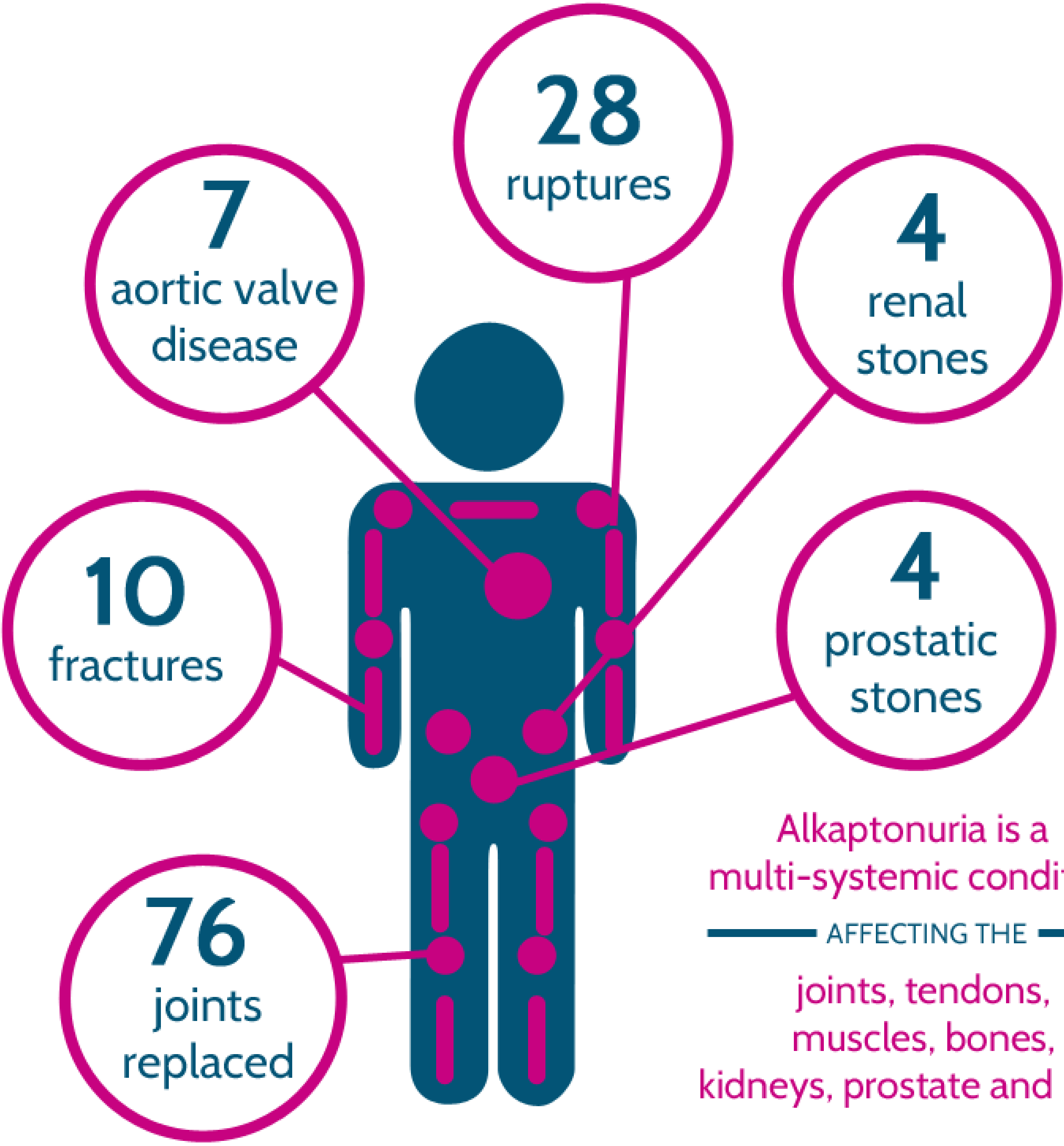
When the NAC started in 2012 it was an inpatient-based treatment service. In response to feedback, however, we now put patients in a local hotel. This makes for a much more enjoyable trip, where they can socialise in an informal and relaxed environment. Many say that they welcome the chance to catch up with old friends and meet new patients. Though patients still find their visits tiring, they understand their value.

In 2017, we saw 15 patients return to the NAC for their sixth annual visit. In 2018, there will be five new patients attending and two more new referrals, so it’s going from strength to strength.

The service which the NAC offers is continually being reviewed so that we can improve it. For example, in 2017 blood spot testing was introduced. When they take nitisinone, our patients are monitored through specialist blood tests, and samples are then sent to the NAC. It has previously been difficult for local healthcare providers to take patients’ blood. Consequently, patients are now shown how to take small blood samples on to special cards. These can easily be posted to the NAC. Monitoring patients on nitisinone is now much easier. Patients have more control over the management of their condition.

The effects of alkaptonuria

based on a study of 44 UK patients



28
ruptures

7
aortic valve
disease

4
renal
stones

10
fractures

4
prostatic
stones

76
joints
replaced

Alkaptonuria is a
multi-systemic condition

AFFECTING THE

joints, tendons,
muscles, bones,
kidneys, prostate and heart

The Big Lottery Fund

Since 2012, the AKU Society has been responsible for a project funded by the Big Lottery Fund (BLF)'s Reaching Communities grant scheme, which will last until June 2018. The BLF has been a generous supporter in the past, and has supported many previous projects, including the initial creation of our website.

The project aims to develop our patient communities through online communication. In doing so we hope to impact positively the mental and physical health of AKU patients, who will have better access to a supportive web-based community.

What are we aiming for?	What have we achieved?
AKU patients involved in the project will have improved physical health.	20/57 patients who took part in our 2015 quality of life survey rated their health as better. 17/57 rated it as the same, which is good as AKU is progressive. The number of NAC patients has gone up to 59.
AKU patients and carers involved in the project will have reduced isolation and an increased sense of community.	We have 1,161 likes on Facebook and 1,356 followers on Twitter. We have 261 members on PatientsLikeMe and 256 on RareConnect. We have had over 124,298 on our website. After a recent patient workshop, 87% of respondents rated the support and sense of community that we provide as good or excellent.
AKU patients, family members and carers involved in the project will have improved mental health.	88 patients and family members attended our 2016 patient workshop, which included a session on mental health. 41/44 respondents subsequently rated this as good or excellent. We have also secured funding for patients in the Bedfordshire area to receive counselling.
Medical professionals and the wider rare disease community will have improved understanding of AKU and provide better policy, research and care for AKU patients.	In our 2015 quality of life survey, 41/41 of those who attended the NAC rated confidence in their medical care as better. 65 medical professionals attended our scientific workshop in Copenhagen in October 2015; 50 attended meetings in Stockholm (2014), Liverpool (2013) and Slovakia (2012). 879 medical professionals have completed our e-learning module through the RCGP. The University of Cambridge has also funded 2 PhD research projects studying the social experiences of AKU patients.

Patients at November's Patient Workshop

To make our website more accessible to visitors with diverse needs, we have installed some software called Browsealoud from a company called Texthelp. Every visitor from our website can access this by clicking an icon on the corner of the screen. This activates (among other things) a range of functions, available in a number of languages, to assist the visually impaired.

We are very grateful to Breaking Down Barriers for funding this project. It is part of a broader effort to make the AKU Society's information more accessible to patients from minority communities.

Our blog is also going strong, with one item published on the website each week. Each blog post is accompanied by a flurry of promotional activity on social media. The write-ups of our patient workshops, as well as human interest stories about individual patients and employees, were particularly popular in 2017.

Social Media in numbers

- 1361 followers on Twitter
- 1163 likes on Facebook
- 255 members on RareConnect
- 263 members on PatientsLikeMe

BIG LOTTERY FUND

NATIONAL LOTTERY FUNDED

Research

It's been five years since the groundbreaking DevelopAKUre clinical trials started and we are now nearing their end. 2017 saw the fifth visits for SONIA 2 across the three trial sites: Piestany (Slovakia), Paris and Liverpool. We also saw the end of the exciting SOFIA observational study. We are now within touching distance of finding an effective treatment for AKU.

DevelopAKUre continues to be a shining example of how a patient group can be instrumental in the planning and execution of an international clinical trial. We are incredibly proud of all those brave patients who have volunteered to help their patient group, and we remain hopeful that we will see a generation of AKU patients grow up without the damaging symptoms of their disease.

> "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."


Bob Montague having a PET scan

SOFIA ('Subclinical Ochronotic Features in Alkaptonuria') was an observational study designed to find out the age at which ochronosis, the damaging blackening of cartilage and bones, starts in AKU patients. The end goal of SOFIA was to highlight the age at which nitisinone should be administered, potentially to prevent a generation growing up with the symptoms of the disease.

30 AKU patients, aged 16 to 70, travelled to the Royal Liverpool University Hospital last year to take part in the groundbreaking study, which saw its last patient in summer, and, by winter, had highlighted its findings. The hospital also recruited some brave non-AKU patients, including some of the AKU Society staff, who donated their own blood for a comparison.

Symptoms were recorded in some of the youngest patients and led to the clinical need for an exciting paediatric study to look at AKU in the very youngest AKU patient. This study has yet to be planned, but promises to expand our knowledge of young people and AKU's effect on them.


Professor Ranganath, Chief Investigator

> "In other words, it might help me and other AKU patients to take back the parts of our lives that we thought had been lost to our pain for ever."

Excitingly, the AKU Society has also helped the Royal Liverpool Hospital to undertake a study into tyrosine levels in AKU patients,

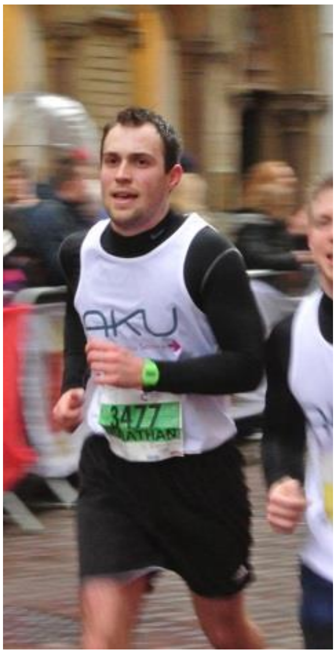
Dr. Milad Khedr explains,

"Nitisinone can halt the production of HGA, but it leads to a rise in tyrosine levels. This can cause eye symptoms such as eye dryness and pain, itching, watering of the eye and irritation.

"This study will investigate how nitisinone affects the tyrosine levels in alkaptonuria. This will eventually help to improve the dietary advice given to AKU patients, as well as the use of nitisinone in treating AKU. We want to investigate how healthy adults and patients with AKU break down tyrosine differently. A comparison of results between the two groups will help us understand this better."

Highlights

So much happened at the AKU Society in 2017. It was a year that saw us hold our first Children's Workshop, climb part of the way up Mount Everest, and travel around Europe promoting AKU research. Here are some of the highlights.



March

7 runners from the AKU Society ran the Cambridge Half Marathon, raising an impressive £1609 to support our work. The runners were great ambassadors for the charity and helped us start the year on a positive note.

June

We held our second Stand up for AKU! Comedy Night, at the Hot Water Comedy Club in Liverpool. The headline act, Ross Leslie, had the audience in stitches. We raised £802 in total.

July

We held our first Children's Workshop at ZSL London Zoo. The young patients got to meet the animals and hear from a range of experts about AKU. The workshop also saw the launch of our first Children's Booklet.

October

Lesley, our Patient Support Manager, and Carol, an AKU patient, went to the Best Practice 2017 conference in Birmingham. It was the primary care sector's flagship event and a great opportunity to publicise our work.

November

Hannah Shepherd, gait analyst for the NAC at Liverpool John Moores University, trekked to Everest base camp in 15 days. This took her to 5,364m above sea level. She raised £1170 through her climb, for which we are very grateful.

April

Marathons seemed to be becoming a theme in AKU Society fundraising. Joe England successfully ran the London Marathon in just under four hours, raising £1725 in the process.

May

We travelled to Budapest for the EURORDIS– European Organisation for Rare Diseases membership meeting, which brings together patient groups from all over Europe to share best practice. Oliver, then our CEO, gave a talk about our clinical trials.

Staff Changes

We said farewell to our CEO, Oliver Timmis, in July. Oliver had worked with us for seven years and had been CEO for the last two. He was replaced by interim CEO Nick Sireau, who is enjoying getting back to grips with the charity.

July also saw the departures of Rory, our Fundraising Officer, and Robin, our Admin and Events Officer. We wish them the best of luck in their future careers. In September we were joined by Reece (Admin and Comms.), who is carrying on their great work.

Garrod Day

In November we marked the birth of Sir Archibald Garrod, the Victorian doctor and scientist whose discovery of alkaptonuria in 1902 was a milestone in modern genetic science. We baked, sold and ate cake to celebrate.

Sociology Project

We met Tanisha Spratt, a PhD student at the University of Cambridge, who is studying AKU patient experiences in the USA. She is especially interested in how the physical manifestations of AKU affect patents socially and psychologically.

Thank You

Trusts and grant-giving organisations make up a large portion of our income. Without the generosity of our benefactors, we would not be able to help AKU patients in the way that we do. Thank you to all those who have donated to the AKU cause. This is a list of the trusts and institutions who have supported us, and what their money has been spent on:

Big Lottery Fund - Online AKU project, aimed at informing AKU patients.
European Commission - FP7 DevelopAKUre project. This is our clinical trial 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.
The Hospital Saturday Fund - Our first Children's Workshop.
The Strangward Trust - The continuation of our BLF project 'Reaching Communities'.
The Hedley Foundation - Our first Children's Workshop.
London Catalyst - The continuation of our BLF project 'Reaching Communities'.
Sylvia Adams Charitable Trust - Funding for Breaking Down Barriers patient identification project.
Sobi - AKU Children's Booklet
Vitaflo - AKU Dietary Advice booklet
Wixamtree Trust - Support for AKU patients in Bedfordshire

We are also immensely grateful to our community fundraisers. We had seven runners in the Cambridge Half Marathon, who raised £1609 this year. Thanks, too, to Joe England (who raised £1725 running the London Marathon), Hannah Shepherd (who raised £1170 trekking to Everest base camp) and the Pedwar Pawen Training Club (whose charity dog show raised £200).


Patients at the March patient workshop

Thank you to everyone who donated, everyone who volunteered, and everyone who got involved in one of our events. We're all very grateful for your support.

Financials

We are very grateful to all of our supporters over the past year. Their generosity has left the AKU Society finances in good shape and funded the important work we do supporting patients.

Nick Sireau, Chair of Trustees and CEO


Income


Expenditure

	Unrestricted Funds	Restricted Funds	Total Funds
INCOMING RESOURCES	**£**	**£**	**£**
Voluntary income	10,096	9,122	19,218
Other	1,178	-	1,178
Trusts and institutions	-	316,693	316,693
Total Incoming Resources	**11,274**	**325,815**	**337,089**
RESOURCES EXPENDED			
Costs of generating voluntary income	1,494	23,567	25,061
Research and support	18,902	216,045	234,947
Total Resources Expended	**20,396**	**239,612**	**260,008**

AKU Society

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Twitter: @AKUSociety

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