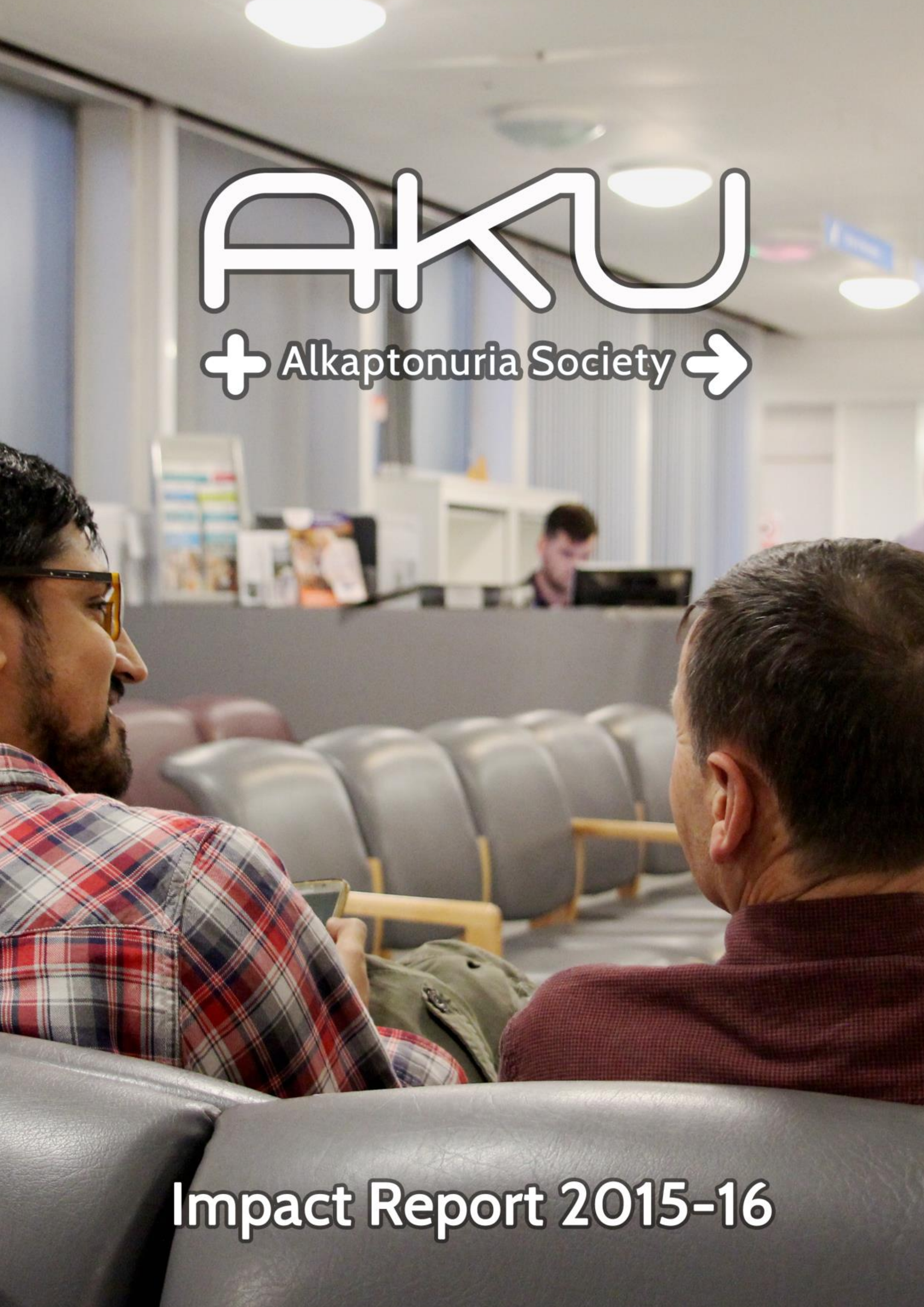


AKU

Alkaptonuria Society

Impact Report 2015-16

"It is great to know that you are not alone and there are others that you can learn from. There are people that care about you and this rare condition. One day there will be a cure for it and you are part of this research to help further generations"

Contents

On the cover: Two patients at the Robert Gregory National Alkaptonuria Centre (NAC)

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 variety of life-changing disabilities. Patients develop early onset osteoarthritis, destroying every joint in the body. The immobilising nature of the disease leads to social isolation, unemployment and chronic pain.

> " It feels as if your bones are 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

Meet Ann Kerrigan

I would be completely lost without the AKU Society. I didn't know for so long what was going on and it was very frustrating and upsetting. I felt isolated, I hadn't a clue what was going on. Then, when I first got the diagnosis, that was a relief, but then the next thing was, well, what does that mean?

I feel so privileged to be able to access the National Centre in Liverpool because I know what it was like before we had the specialist centre. I tried three times to get nitisinone and was turned down three times by my NHS trust. I was low at that stage, and pretty soon after I found out we had the specialist centre.

The AKU Society is a wonderful resource that I am able to access for health checks and so many other things. I can phone about anything. It's an amazing Society when you look at everything that has been done –it's got to be a model of best practice. It's just a wonderful team that has been put together. The Society has helped me cope with my disease, I feel so privileged that I've had such access, it's all very positive.

"It's an invisible disability because you look OK on the outside so people don't always believe that there is anything wrong with you. That's really upsetting, especially when it comes from your family"

A Message From the CEO

Welcome to the AKU Society's Annual Review of 2016. We had a great year, achieving our mission statement of: 'Transforming the lives of AKU patients; through patient support, community building and medical research'.

Patient Support

National AKU Centre patients attended their 5th annual visit, accessing expert care and treatment for half a decade. Our Patient Support Manager, Lesley, offered excellent patient support. She met the most in-need patients one-on-one in their homes and supported Welsh patients in their attempts to access the same level of care as patients in England and Scotland.

Community Building

We held our largest ever workshop: the first International AKU Patient Workshop. This was a truly global meeting, with one patient travelling over 7,000 miles from Argentina to Liverpool. We received fantastic feedback, with everyone in attendance agreeing that it was valuable to meet other patients and learn new methods to live with AKU.

We launched brand new AKU Society and DevelopAKUre websites, based on patient feedback from the old sites. Along with our social media channels, anyone who needs help has a way to contact us and find AKU information online. In November, we welcomed a new Admin and Events Officer, Robin, as our main contact online.

Medical Research

The DevelopAKUre clinical trials, our major research programme studying nitisinone in AKU patients, celebrated its four year anniversary.

Our superb patient support is led by our Clinical Trials Officer, Ciarán. Thank you to everyone who has taken part in our clinical research. I hope the final two years of DevelopAKUre continue as successfully.

Looking Ahead to 2017

In spring 2017, we will organise a patient workshop supporting adults with AKU, and for the first time, a separate children's workshop. We hope to support all children affected by AKU, either directly, or through their parent's or sibling's diagnosis.

Our fundraising events will include one runner at the London Marathon (on 23 April 2017), and 10 runners at the Cambridge half-marathon (on 5 March 2017). These events help us raise much-needed funding to support AKU patients, so please consider sponsoring them. If you want to plan your own fundraising events, please do contact our new Admin & Events Officer, Robin.

The AKU Society will continue to grow and I want you to be involved in that process. If you have any ideas for future goals, want to discuss our past work, or have any comments at all, please do contact me at oliver@akusociety.org or call our office on +44(0)1223 322897.

Yours faithfully,

Oliver Timmis, CEO

Patient Support

AKU is such a rare disease that patients often feel isolated and alone. The condition degrades their quality of life and self-esteem. 71% of our patients said that they accomplish less than they would like because of depression or isolation. There is a lack of information available to them about how to cope with pain and disability, about how to handle the disease.

This is why the AKU Society holds patient workshops and 2016 saw our first international workshop. Patients attended from 13 different countries, including Jordan, Argentina, Slovakia, Poland, Holland, Spain, Sweden, Canada, USA, UK and Belarus. In many of these countries, little is known about AKU, so the workshop was their first opportunity to meet and learn from AKU experts. For some, this was also their first time meeting other AKU patients.

There were specialist sessions on topics such as physiotherapy, nutrition, mental health and pain management. Patients came together, to reduce their isolation and build a supportive AKU community. Our workshops offer patients the opportunity to talk to others with the same condition. We closely monitor feedback so that we can improve with each event. After our workshop in 2016, 98% of attendees rated their knowledge of AKU as good or excellent.

“This workshop is a cherished opportunity to learn more about ongoing treatment and rehabilitation from recognised leaders in AKU research”

We now offer home support visits as an extension of the National AKU Centre. The face-to-face visits provide an opportunity to discuss anything around AKU, their care, treatment or anything else they may want to discuss. The value of these home visits cannot be underestimated, it is a clinically useful intervention, an invaluable teaching tool and an opportunity to provide support. It gives our Patient Support Manager the opportunity to meet patients in a more relaxed and informal setting, where it is often easier to talk about any personal issues or problems.

We are continually looking to develop and improve our home support service and more recently offered chair-based exercise during these visits. One of our patients Ann found the visit really helpful, and especially enjoyed the chair-based exercise session. The session taught her exercises to do at her own pace in the comfort of her home. This may help to increase or maintain her independence, help with strength and balance, and most importantly, aid with activities of daily living.

> "I've become an expert in my own condition, I definitely feel less isolated and I know that there is a wonderful team of people trying so hard to make things better"

Advocacy

Dissemination is a key aspect of 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. Also, with a drug that effectively stops the build-up of the harmful acid, it is important for people to get diagnosed and start treatment before any more damage can occur.

Most of our dissemination focuses on the unusual work we do: from the ground-breaking patient-focused DevelopAKUre clinical trials, to the dedicated National AKU Centre, and our many patient workshops and home support visits; we offer a range of services that are unique.

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

Over the past year we have:

Spoken at over 23 events across 6 European countries travelling 14,500 miles.

We have exhibited to GP’s, nurses, doctors and other charities at 7 major conferences across the UK.

The Coffee Morning is an important day in the AKU Society's calendar. On November 25th, we celebrate Garrod Day, which marks the birthday of Sir Archibald Garrod, the man who first described AKU. This important anniversary is celebrated by holding annual Coffee Mornings. This year we went global, holding coffee mornings in Australia, Jordan, Germany and all over the UK.

The Coffee Morning allows us to spread the word about AKU, whilst raising crucial funds. The most common comment from participants is:

"I hadn't heard of AKU before"

"Recent discussions with the WHSSC have been far more positive, with possible ways forward being pursued. We can only hope for a positive outcome"

In 2013, Welsh AKU patients were able to visit the National AKU Centre for the first time. For Welsh patients with rare diseases, accessing treatment can be a challenge and it is often difficult for them to gain timely and effective access to the right specialised services and treatments. The application process can be complicated and in the case of our patients, access to full treatment is not always granted.

We work alongside other rare disease groups in Wales to raise awareness and ensure equitable access to services for Welsh patients in the future. We continue to work closely with the Welsh Health Specialised Services Committee (WHSSC) to improve and simplify the process. We are also liaising with the All Wales Therapeutics and Toxicology Centre, looking at how Welsh AKU patients might be able to access nitisinone in the future. This piece of work is still very much in its early stages, but seems promising.

NAC–Treatment

The National Alkaptonuria Centre (NAC) is based at the Royal Liverpool University Hospital. The NAC was established by the Department of Health (National Specialised Services Commissioning Group) and opened to patients on 18th June 2012. It was set up to provide assessments and treatment for all people over the age of 16 who have been diagnosed with AKU. Patients who attend the NAC have a week long annual visit where they receive a full health assessment, off-label access to the drug nitisinone and advice on living with and managing AKU.

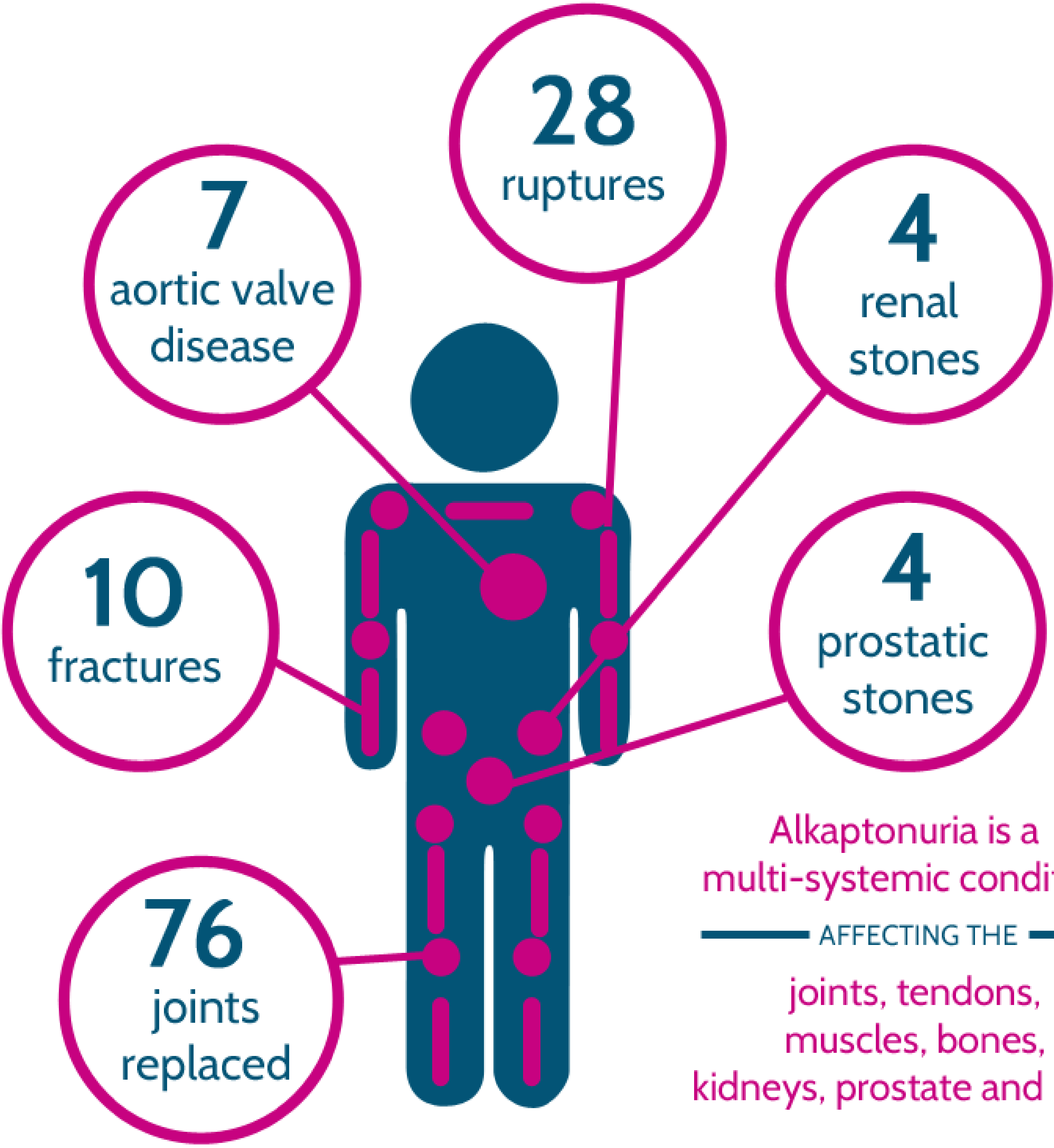
> “The centre gives vital information that supports further research into AKU, which is so important in trying to find treatments and maybe even a cure for this disease”

In 2016, we celebrated 16 patients’ 5th visits to the NAC. Since 2012, the NAC has gone from strength to strength and now sees 55 patients every year as opposed to 21 during its first year, with more patients applying every month. The NAC gives AKU patients expert care and access to a drug, off-label, that has the potential to reduce the levels of Homogentisic acid in their bodies by 95%. Over the past 4 years, the NAC has changed dramatically. During their first visits, patients were required to stay on the ward overnight at the hospital but after listening to patient feedback, this was changed in 2015. Now patients enjoy overnight stays in a local hotel, allowing them to relax out of the hospital and socialise with other AKU patients in an informal environment. An NAC patient returning for their 5th visit said;

“Being on nitisinone has meant that I am no longer producing the harmful acid in my body, so my body is ageing at a more normal pace. Joints won't get better but at least their deterioration is at a normal rate. I have appreciated having access to the pain clinic and Dr Jones' injections helped my

The effects of alkaptonuria

based on a study of 44 UK patients



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 running a project supported by the Big Lottery Fund's Reaching Communities grant scheme, to develop our patient communities. The BLF is a fantastic funder who has supported several past projects from the AKU Society, including the initial development of our website.

Our current project aims to use our online work to continue to support AKU patients, to hopefully have a positive impact on physical and mental health, through access to a supportive online community.

What do we hope to achieve?	What have we done to reach those achievements?
AKU patients involved in the project will have improved physical health.	A third of patients who took part in our 2015 quality of life survey rated their health as much better since 2012. Most of this is due to the care received by the 50+ patients who now attend the NAC each year.
AKU patients and carers involved in the project will have reduced isolation and an increased sense of community.	39,000 users have accessed our website, and around 8,000 are returning users. Over 90% of patient workshop attendees rate our meetings as good or excellent. 1,131 users follow us on Facebook, 1,174 follow us on Twitter, 170 patients engage on PatientsLikeMe and 191 engage on Rare Connect.
AKU patients, family members and carers involved in the project will have improved mental health.	We have organised several sessions on mental health, including for the 88 patients and families at our 2016 International Patient Workshop, and for the 170 users who joined our PatientsLikeMe mental health week in 2015.
Medical professionals and the wider rare disease community will have improved understanding of AKU and provide better policy, research and care for AKU patients.	We exhibited at 7 medical professional conferences in 2016, and have continued to identify new AKU patients in the UK and abroad. Our RCGP training module on AKU diagnosis and treatment has been seen by 5,000 medical professionals in the UK, and a third of patients who took part in our 2015 quality of life survey rated local care from their GP as improved since 2012.

This year, we have done a major review

and update of our AKU and DevelopAKUre websites, ensuring all content is relevant and accessible to our users. The website is now mobile friendly; an important feature, with over 40% of our users accessing it via mobile.

We aimed to create a website that was easy to use, accessible, modern and responsive.

User feedback has been very positive, with reports that it is easier to navigate around the site and access relevant information.

Since launching the new site in July 2016, we have had positive feedback from all over the world, from the United States to Australia.

On the website we also aim to publish one blog every two weeks. These can be an update on research, tips for living with AKU or the latest on our fundraising activities.

Social Media in numbers

- 1174 followers on Twitter
- 1131 likes on Facebook
- 191 members on RareConnect
- 170 members on PatientsLikeMe

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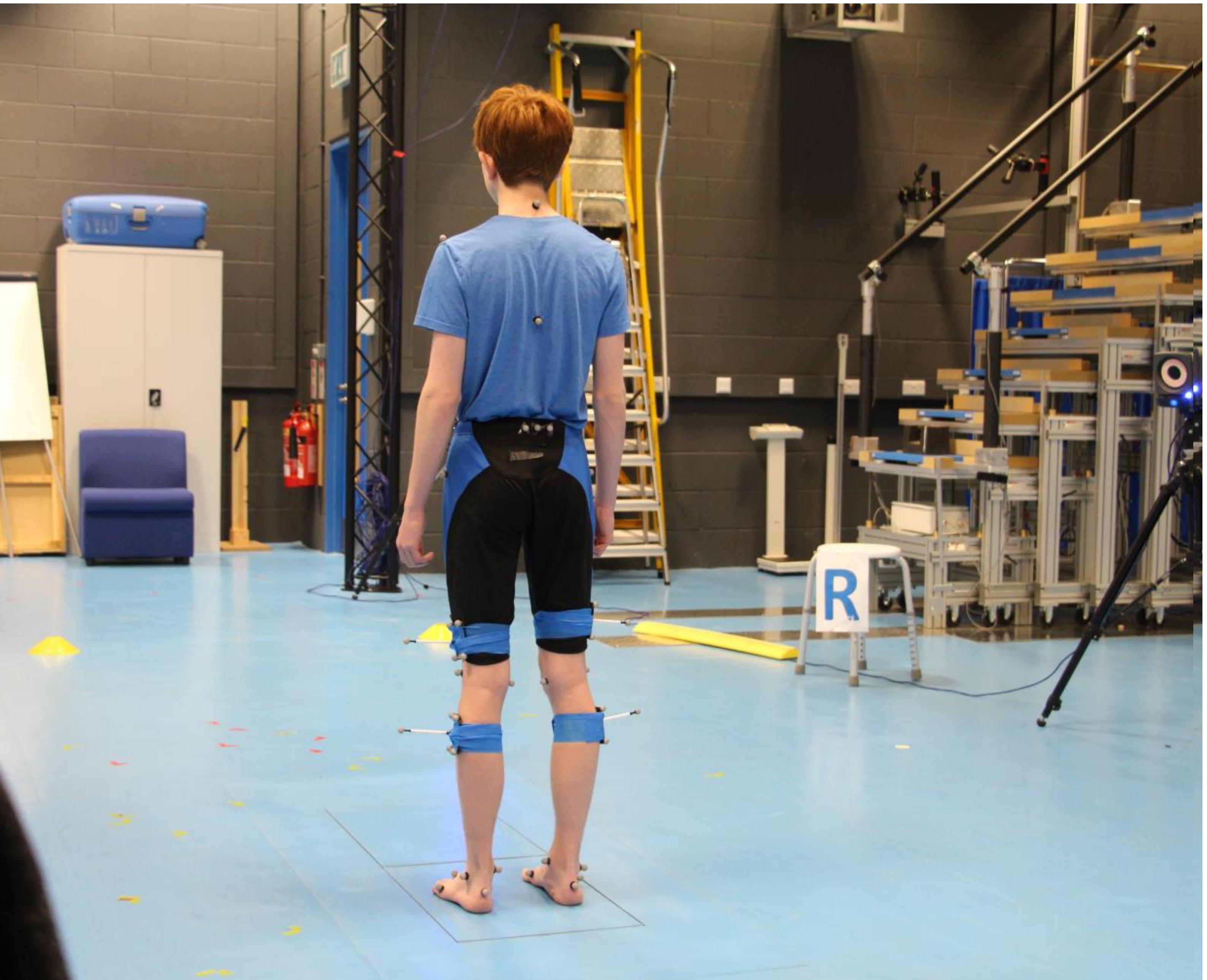
DevelopAKUre

DevelopAKUre is a series of clinical trials, run by a consortium of 12 European partners. It aims to study a new drug called nitisinone, and assess its potential effectiveness in treating the rare disease alkaptonuria.

November marked 4 years since we helped gain the staggering £8 million funding needed to start the trials. This includes £4.3 million received from the European Commission.

Thought to be the first time a UK patient group has been the driving force behind a major international drug research trial, we are extremely proud of how far we have come and how close we are to finding an effective treatment for AKU, after striving so hard over the past 11 years.

> “AKU is such a rare genetic disease that I felt the obligation to help the researchers with everything I could. I have gained more knowledge about AKU”



DevelopAKUre is 3 groundbreaking clinical trials, called SONIA 1, SONIA 2 and SOFIA. SONIA 1 was designed to ascertain the best dosage of the trial drug nitisinone. This finished in 2014 and the resulting dosage was used in the next trial SONIA 2. This trial is currently ongoing and will test the safety and effect of the drug in AKU patients. Currently half way to completion, the 1-year analysis of the data gathered from the trial so far is expected to be released in early 2017. This will hopefully show a reduction in the levels of HGA, and that the drug is safe for use in treating AKU. The SOFIA observational study is designed to underline at what age the most damaging symptoms of the disease present in those with AKU. This allows future doctors to prescribe nitisinone at the correct age, so patients in the future may live without the devastating damage that accumulates today. The study will finish in February 2017, with DevelopAKUre ending in 2019.

> "It will bring hope to thousands of people that there is a disease modifying treatment for this condition for the very first time. I absolutely love working in DevelopAKUre"

Professor Ranganath is the chief investigator of the DevelopAKUre clinical trials. With the continued success of the trials, he shared his views on working with AKU patients and what motivates him to keep going.

"I really value working with people where my contribution will hopefully make a difference to their lives. The patients are such a mixed and interesting bunch of people who value what myself and others are doing for them and it makes it all worthwhile. If we are successful in getting the European Medicines Agency to license nitisinone for people with AKU, it will bring hope to thousands of people that there is a disease modifying treatment for this condition for the very first time. I absolutely love working in DevelopAKUre as this is a fantastic, enthusiastic group of people who have come together and dedicated themselves to ensure that nitisinone treatment becomes available to all."

2016 in highlights

We worked tirelessly for AKU in 2016, from comedy nights to International Coffee Mornings. Last year was an amazing time for the Society. With so much going on, here are our highlights of 2016.

February

An amazing £2,427 was raised by 8 fantastic runners at the Cambridge Half marathon. Touring the historic and flat streets of Cambridge, this was a brilliant start to the fundraising year.

April

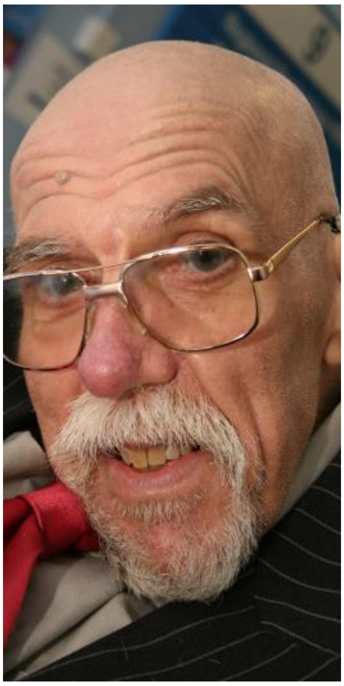
People travelled from over 13 countries to take part in our hugely successful International Patient Workshop. Held at Everton's ground Goodison Park in Liverpool.

June

Our first ever Stand up for AKU Comedy Night raised an incredible £556.15. Over 80 guests paid to see some of the cream of British comedy, including members of the infamous Cambridge Footlights .

September

Eve, our online communities guru, left us after 2 years with the Society. She left to go to Nepal to help rebuild after the devastating 2015 Earthquake.



October

We remembered AKU patient, pioneer and co-founder of the AKU Society, Robert Gregory, who died 2 years ago. Robert's inspiring work still motivates everyone at the Society.

Rare Disease Day

We travelled all over the country representing AKU at the receptions for Rare Disease Day. Tony Esmond gave a moving speech about his experiences of AKU to the Welsh Senedd.

Oliver's sixth year

Our CEO Oliver Timmis celebrated his 6th year at the Society. In that time he became our CEO and has been at the helm as the Society has gone from strength to strength.

4 years of the NAC

June saw the 5th anniversary of the first visits to the National Alkaptonuria Centre at the Royal Liverpool University Hospital. The NAC opened its doors to AKU patients based all over the UK.

August

In August, we said a fond farewell to Lydia, our first dedicated fundraising officer. She planned some amazing community fundraising events and is sorely missed. Rory filled her shoes in September and is continuing the good work.

November

We raised almost £1,000 thanks to everyone who bought and sold cakes and coffee for our Garrod Day Coffee Mornings. We held events all over the world.

DevelopAKUre is 4

November marked the 4th anniversary of successful funding for the ground breaking DevelopAKUre clinical trials. Designed to test the effectiveness and safety of nitisinone.

Thank You

Trusts and grant giving organisations make up a large portion of our income.

Without such generous benefactors, we would not be able help AKU patients in the way that we do. So thank you to those who donated to the AKU cause. This is a list of the trusts and institutions who have supported us this year;

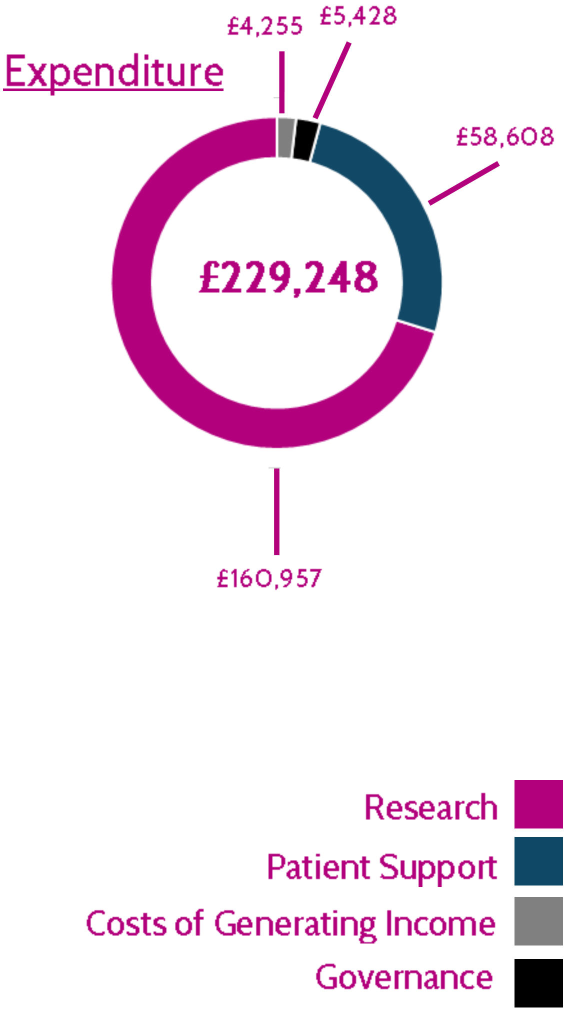
Big Lottery Fund - Online AKU project, overcoming the lack of information for AKU Patients project.
Childwick Trust - 3 years of funding for research studies at the University of Liverpool and the Royal Liverpool University Hospital.
European Commission - FP7 DevelopAKUre project. This is our clinical 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.
Sobi– Sponsored International AKU Patient Workshop
Vitaflo– Funding for a 2017 AKU Dietary Advice
Wixamtree Trust–Support AKU patients in Bedfordshire

Another important revenue stream is that of our community fundraisers. We would like to thank everyone who took part in an event to raise money for AKU Patients in 2016. A special thanks goes out to the brave runners at different events. We had 8 participants in the Cambridge half marathon, who raised an amazing £2897. A big thank you also goes to Katie Smith, the daughter of AKU patient Richard, who ran the Cardiff half marathon and raised £280.

Thank you to everyone who donated, everyone who volunteered, and everyone who got involved in one of our events. Thank you from AKU patients and everyone here at the AKU Society!

2016

Thanks to our generous donors and grants from several trusts and foundations, the AKU Society accounts are in good shape, enabling us to support patients for another year. - Nick Sireau, Chair of Trustees



	Unrestricted Funds	Restricted Funds	Total Funds
INCOMING RESOURCES	**£**	**£**	**£**
Voluntary income	17,083	55,741	72,824
Investment income	1,374	-	1,374
Research	-	213,028	213,028
Other incoming resources	935	1,475	2410
Total Incoming Resources	**19,392**	**270,244**	**289,636**
RESOURCES EXPENDED			
Costs of generating voluntary income	127	4,128	4,225
Research + Patient Support	1,691	217,874	219,565
Governance Costs	5,428	-	5,428
Total Resources Expended	**7,246**	**222,002**	**229,248**

HELP US CURE BLACK BONE DISEASE

AKU
Alkaptonuria Society

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Cambridge
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Call us: 01223 322897
Contact us: info@akusociety.org
Visit us: www.akusociety.org

Registered Charity in England and Wales: 1101052

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The Royal Liverpool and Broadgreen University Hospitals NHS Trust
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