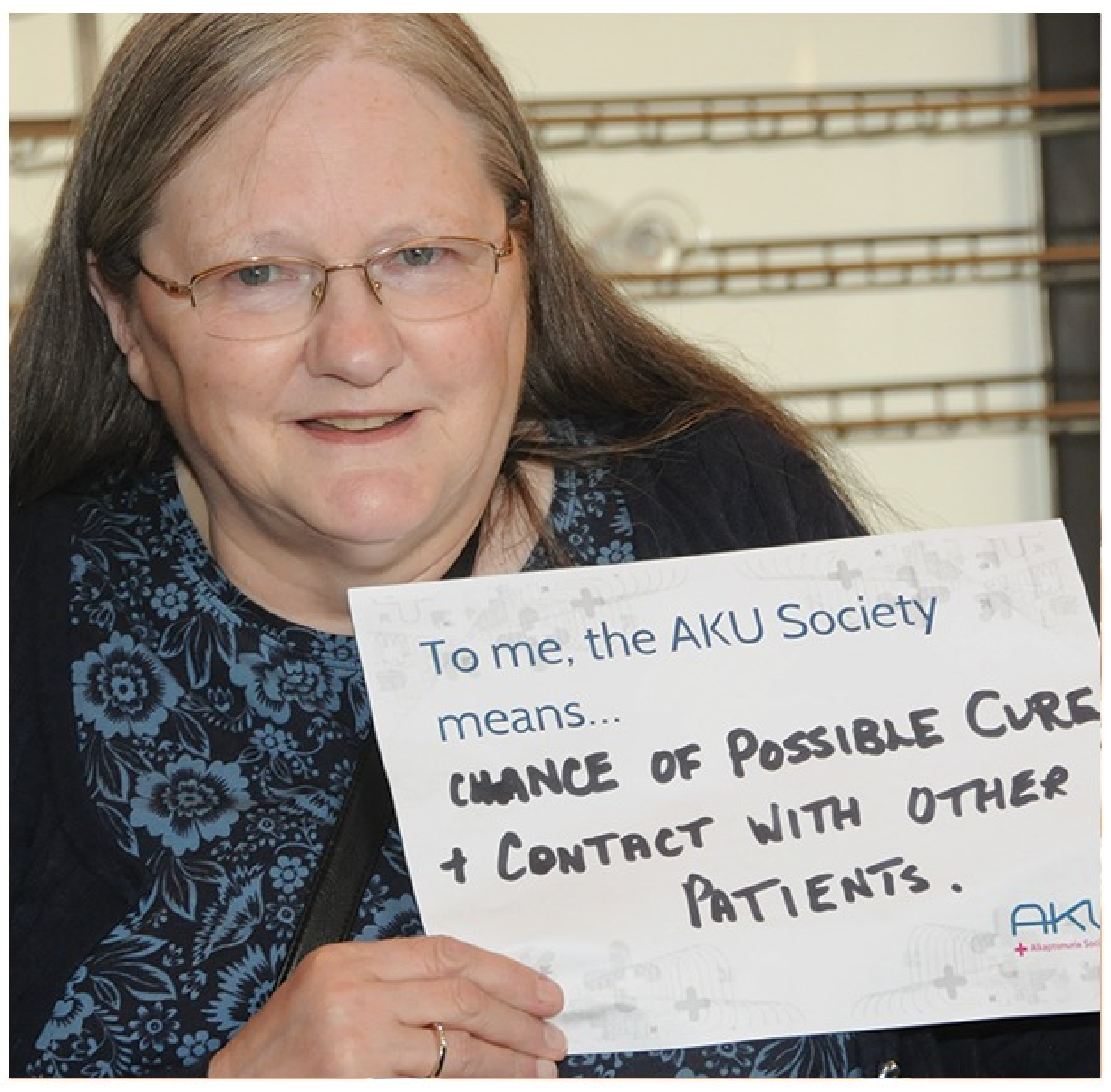
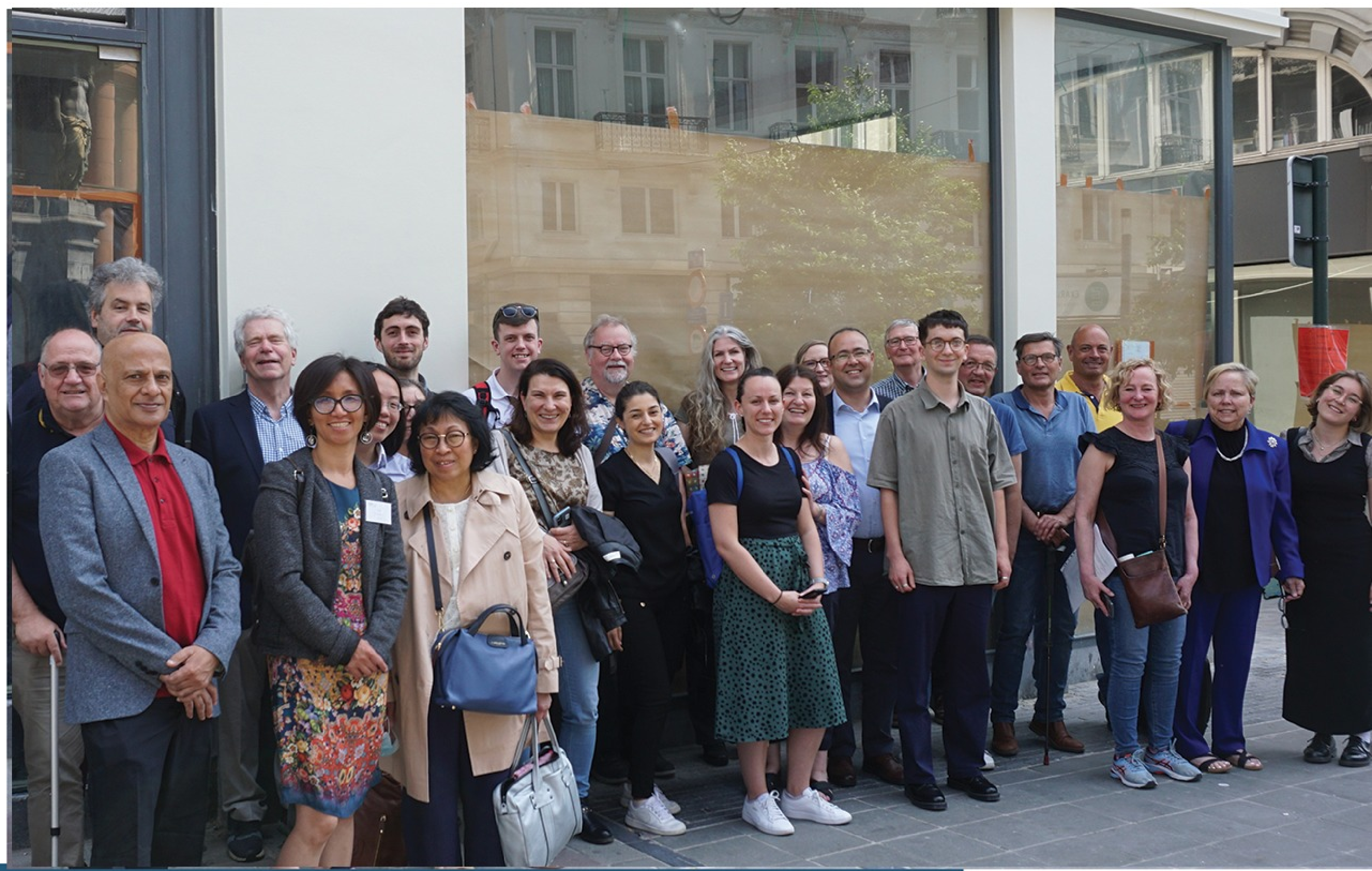
AKU
Alkaptonuria Society

Impact Report

2022





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Who We Are

The AKU Society is a patient-led support group. Since 2003, we have worked to improve the lives of those with alkaptonuria (AKU) and those who support them while researching for a cure.

Our mission statement is: 'transforming the lives of AKU patients; through patient support, community building, and medical research'

What is alkaptonuria?

Alkaptonuria (AKU), also known as 'Black Bone Disease', is a rare inherited condition that causes the cartilage on joints to become black and brittle. It's caused by a problem with a gene and leads to a build-up of a substance called homogentisic acid (HGA).

HGA builds up in connective tissue, especially cartilage, and over time causes it to darken and become brittle. This leads to early onset osteoarthritis and means multiple joint replacements are needed.

HGA is secreted in large quantities in the urine, making the urine turn dark when left exposed to air. Because of this, AKU can also be called Black Urine Disease.

A Message From The CEO

Nick Sireau, CEO and Chair of the Trustees at the AKU Society, takes some time to reflect on the past year at the charity.

As we have all emerged from the pandemic in the last year, the AKU Society has had another busy year supporting and uniting AKU patients. I would like to say thank you to the AKU community for your continued support and we look forward to maintaining this momentum moving into the next year.

This year we celebrated two anniversaries. June marked the 10th anniversary of the National Alkaptonuria Centre (NAC) opening it's doors as well as Lesley Harrison's, our Head of Patient Support & Welfare, 10th year at the AKU Society. The NAC has been one of the key achievements of the AKU Society and we are incredibly proud of the work which the NAC team has invested into the service and the impact they have had on the AKU community. Lesley has been at the forefront of our work at the NAC, assisting patients on their annual visits to the centre. Her support to patients has been invaluable to the AKU community and contributed to the smooth running of the NAC.

In May, we were once again able to host a scientific conference on AKU. We brought together researchers and clinicians from all over the world to discuss current and future research into AKU. The collaborations we formed will help to drive forward our work towards developing treatments and cures for AKU.

Looking ahead, we have a lot of exciting projects coming up in the next year. Our SOFIA-Paediatric study, investigating the way in which AKU begins to damage connective tissue is scheduled to begin early next year, while we are also launching our AKU patient registry in 2023, and hope to move closer to beginning a study into developing a tyrosine-reduction therapy.

Nick Sireau

CEO and Chair of the Trustees at the AKU Society

2022 Recap

10th Anniversary of the National Alkaptonuria Centre

June 2022 marked the 10th anniversary of the National Alkaptonuria Centre (NAC), which has played a pivotal role in supporting and caring for the AKU community.

In 2012, the AKU Society collaborated with the Department of Health to establish the NAC at the Royal Liverpool University Hospital. UK AKU patients attend the NAC once per year, where they receive a full health assessment, access to the drug nitisinone, and advice on living with and managing AKU. The NAC has now provided treatment to 89 AKU patients.

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

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

12th International Scientific Conference on AKU

In May 2022, we hosted our 12th International Scientific Conference on AKU at Vrije Universiteit, Belgium. After two years of disruption from COVID-19, we were finally able to reunite the AKU scientific community.

We heard presentations from 23 speakers, coming from eight different countries, who discussed current and future research into AKU. More than 100 people joined us in person or virtually across the weekend, and we connected with academics, clinicians, and AKU patients from all around the world.

We held sessions on 'Alkaptonuria in 2022', 'Gene therapy in AKU', and 'Combatting tyrosinaemia'. The conference was a great success. We were able to unite the AKU community and forge new collaborations into scientific research going forward.

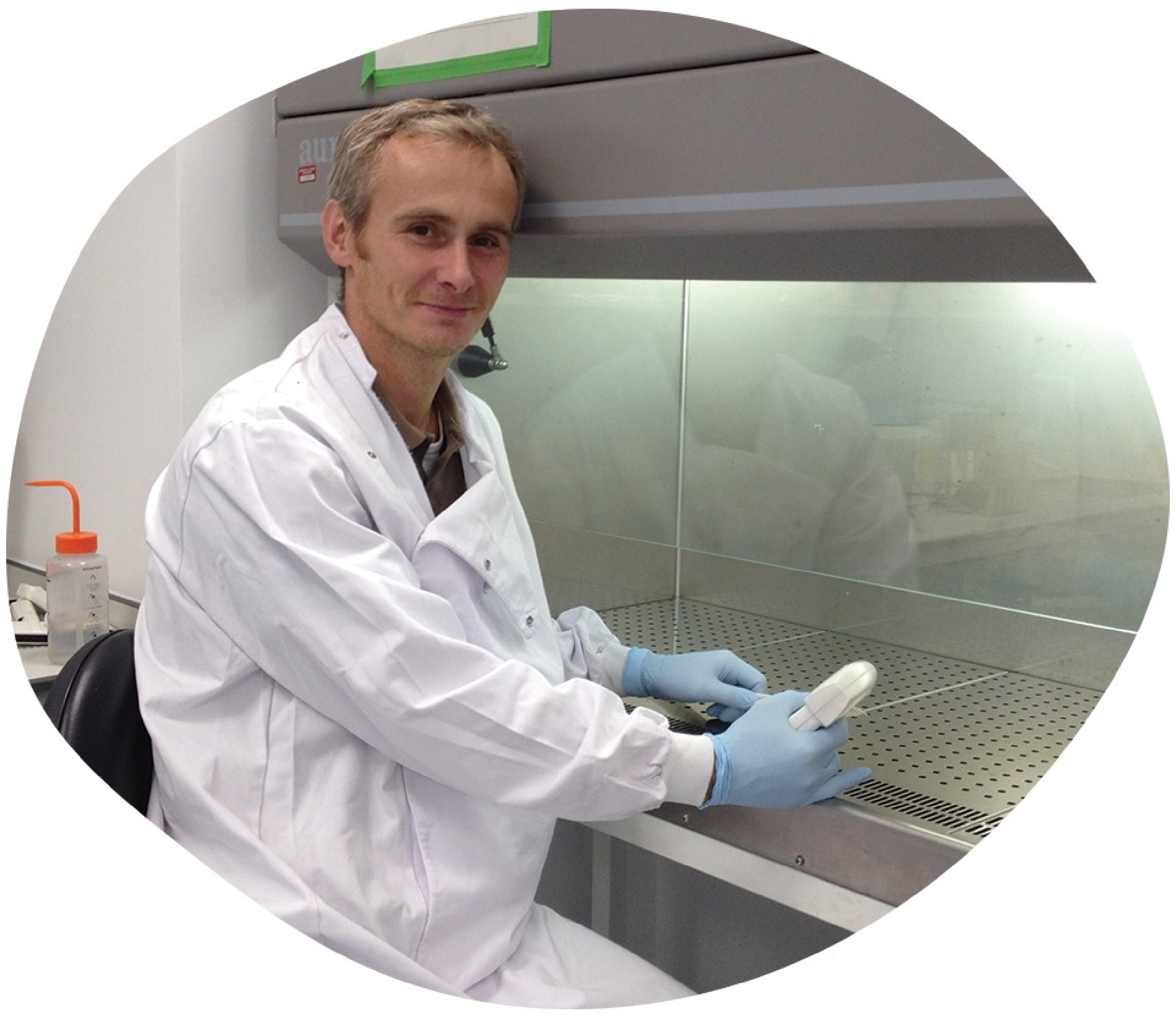
AKU patient registry

In December, we launched a crowdfunding campaign to raise funds for a global AKU registry. This registry will allow us to gather information on AKU, the way the disease progresses, and diagnose new patients. Crucially, this registry will allow us to provide support to AKU patients in developing countries and help us to establish more AKU Societies and work towards universal access to nitisinone.

During our campaign we were met with overwhelming support and generosity from our supporters and we raised over £30,000 towards our registry. Having recently finalised our plans for the registry, it will be launching in the UK in early 2023 before expanding to encompass the entire world.

SOFIA-Paediatric study

After years of delays due to COVID-19, our SOFIA-Paediatric study is finally due to start in early 2023. Nitisinone has been a life-changing treatment for AKU patients, however the side-affects and the need to follow a protein-controlled diet means that, currently, it cannot be prescribed to children. This study will identify the age at which AKU begins to cause damage to the bones and joints of children with AKU. This information will allow us to determine the safest and most effective age at which to begin nitisinone treatment in children to minimise the damage AKU causes.

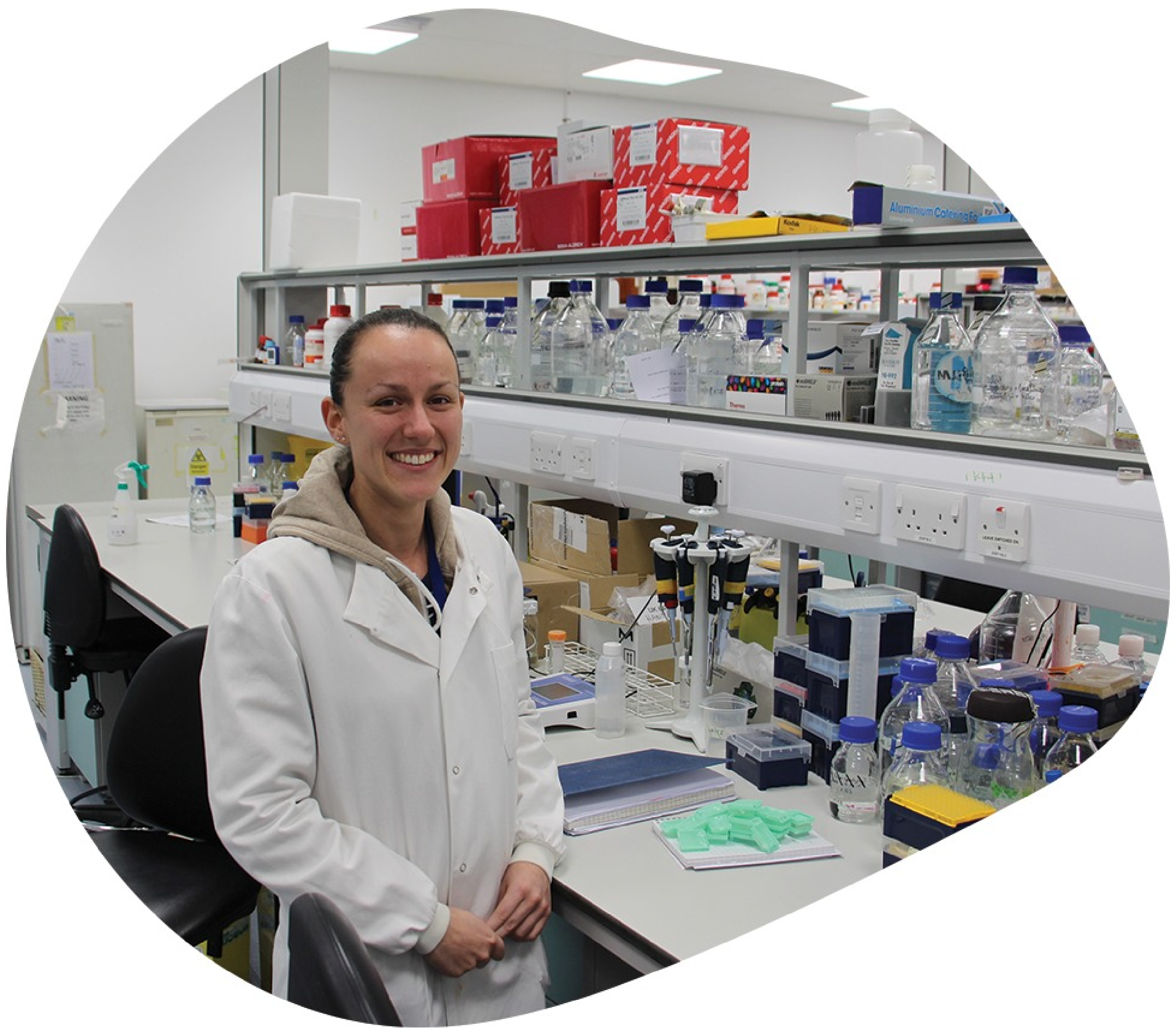


Tyrosine-reduction study

We are currently in the process of fundraising and planning for a tyrosine-reduction study. This study will identify a co-therapy to be used alongside nitisinone therapy which will control the levels of tyrosine which become elevated following nitisinone treatment. This co-therapy will significantly improve the safety of nitisinone treatment and allow patients the freedom of an unrestricted diet.

Other scientific research

We are also looking ahead to the future of AKU research. These include, an AKU gene therapy, an mRNA therapy, and a gait analysis study. These studies are many years away from commencing, but we are excited by the positive impact they could have on the AKU patient community.



Patient Support in 2022

Due to the COVID-19 pandemic and ongoing rail disruption over the past 12 months, we were forced to postpone the two face-to-face workshops we had planned. Our workshops offer a variety of sessions addressing both the physical and psychological impact of AKU. In place of the postponed workshops, we hosted two virtual low protein cooking demonstrations and a number of virtual sessions on mental health and mindfulness practices. We are hoping to return to in-person workshops in the summer of 2023.

Lesley, our Head of Patient Support and Welfare, has accompanied patients to the NAC each month for their annual reviews. She has offered help and assistance when needed and provided one to one support to help them better understand and manage their condition. Regular telephone support is provided to our patient group and home support visits are offered to all patients who require additional support outside of their annual visit to the NAC.

We have continued to offer a remote counselling service to AKU patients in the UK. Several patients have independently accessed this service as well as some being signposted to the service by Lesley. This service has played an invaluable role in supporting our patients to build their resilience to the devastating impacts that AKU can have.

Financial Report

Income

	Unrestricted Funds	Restricted Funds	Total Funds
Voluntary Income	£20,029	£284,849	£304,878
Investment Income	£330	-	£330
Research & Support	-	£281,500	£281,500
Total Incoming Resources	£20,359	£566,349	£586,708

Expenditure

	Unrestricted Funds	Restricted Funds	Total Funds
Raising Funds	-	£600	£600
Research	£49,781	£244,818	£294,599
Governance	£5,152	-	£5,152
Total Resources Expended	£54,933	£245,418	£300,351

Our Funders

We would like to say a huge thank you to the following trusts, foundations, and companies for supporting our work:

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