



Alström Syndrome UK

Strength for today, hope for the future

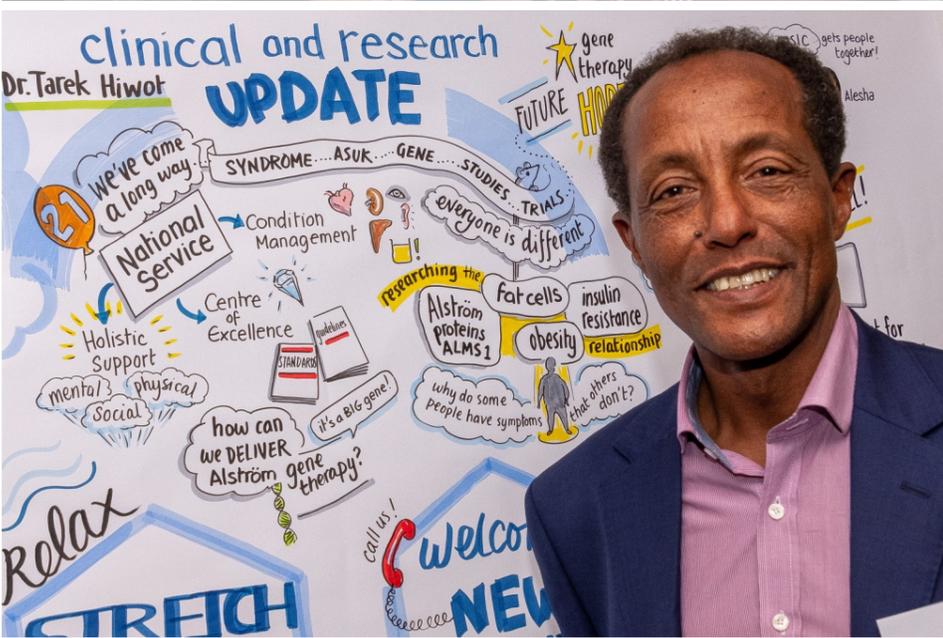
Alström Syndrome UK (ASUK) is a charity who provide information, support and advice for individuals affected, their families, carers and professionals. Alström Syndrome (AS) is an ultra-rare genetic condition which can cause progressive blindness, loss of hearing and can lead to; heart and kidney failure, type 2 diabetes, liver dysfunction and associated problems. The symptoms arise at different stages making diagnosis very difficult. Our mission is to raise awareness, conduct pioneering research and enable better treatments and monitoring through the AS multi-disciplinary NHS clinics. Further information about our work can be found at www.alstrom.org.uk

Community Impact Report 2019-20 Advancing Diagnosis, Care, Treatment and Support for those living with Alström Syndrome in the UK

ASUK is an ambitious and highly focused charity, determined to improve the lives of those affected through personalised family support, research initiatives and raising awareness.

86 people (currently known to us) in the UK have Alström Syndrome (AS).

We have supported **80** families throughout this financial year.



Most professionals will never meet a patient with this ultra-rare condition. It is likely that there are more individuals with AS, who remain undiagnosed — we are working hard to change this!

ASUK strive to empower families to reach their potential and dream big!

- **54** children and adults attended **9** Alström Syndrome (AS) specialised clinics, enabling them to seek individualised, medical expertise.
- **4** new referrals, learning more about Alström Syndrome, how it will affect their lives and seeking our support.
- **12** Team Around the Family meetings attended, leading to robust Education, Health and Care plans and further support for young people.
- **53** professionals received Alström Syndrome training from our team, to improve support services and aid awareness raising.
- The Breaking Down Barriers project worked with **29** patient organisations, support groups and community networks to develop resources to improve engagement with people from ethnic minority communities, as well as sharing good practices to ensure everyone can access healthcare services.
- Families and professionals unite together at the ASUK conference to map their journey to diagnosis.
- The Family Support team visited **46** families at home and **31** children in school to ensure they have the right support and are receiving everything they are entitled to.
- Supported one young adult who was feeling isolated to achieve their dream of going to university and assisted another in securing a sixth form place at a specialist residential college. They are both thriving and the increased independence of getting out and about on their own is enabling their confidence to flourish.
- We launched the AS Patient Registry which we hope will lead to learning more about this rare condition.
- **100%** of families who completed evaluation forms told us they were **'happy'** or **'very happy'** with the AS clinics and ASUK Support Services.

“What a great conference. I've learnt so much that I didn't before. It was one of the best rare disease conferences I have been to. The atmosphere, bond with the families and the team was wonderful.”



7 families took part in the Big Activity Challenge and **44** families took part in fun filled activities including:

- What goes on behind the scenes? Theatre trip
- Trip of a lifetime, Game of Thrones fan visits set in Ireland
- Horsing around with personalised horse-riding lessons
- Along for the ride, cycling challenge from Bruges to Amsterdam
- Grrrrreat big cat feeding experience
- Relaxing spa day in the Scottish Highlands
- Free goody bags at Growing up and Puberty workshops
- The fun at the fair and thrilling theatre sessions
- How high can you go, trampoline sessions
- Bowled over, cricket experience in Barbados!
- Ello, Ello, Ello, Birmingham Bobbies make dreams come true as youngsters are on the Beat for the day!

What a day!

“The ASUK big activity challenge wanted to provide a fun filled activity or adventure, well this day certainly delivered for Adam. He had the opportunity to experience things that under normal circumstances he never could. He travelled on a water taxi and in a cab. He visited a London theatre for the first time and got to spend time on the streets of London, experiencing all the different sounds, smells and hustle and bustle. More importantly, he was able to make his own choices on all aspects of his day, where we went, how we got there and where he went to eat.

Perhaps the best indicator of the impact that the experience had on Adam was the excitement with which he explains what he did to colleagues and family.

8 weeks later and he still talks about the trip every day!”



Let the Battle commence!

For one of our members Simmie, living alone was a dream come true. The independence and the confidence this gave was immense and with the right support this is totally achievable. This is where the system often lets us down, not giving people enough social care support so they can manage their day to day tasks effectively. This can lead to isolation and feeling let down and this is where ASUK Family Support workers often have to step in.

Our Family Support team realised that Simmie was only getting the basic level of direct payments, just 21 hours. She was at a high risk of becoming isolated, and the potential to get into financial difficulty as she was spending more on her care hours, than the Local Authority were giving.

“This mornings mission... helping the Local Authority understand that this independent lady is entitled to a Specialist Deafblind Assessment and this had to be completed by an appropriately trained assessor, easier said than done!”

To put this in perspective, the average specialist assessment is a 40-50 page document. The average social care assessment is around 4 pages of tick boxes. Therefore, there is a huge difference in the quality and depth of the specialist assessment, covering every area of support.

After many months of battling with the Local Authority an assessment was carried out and it determined that a minimum of 35 hours of support would be needed.

The assessment also revealed a need for specialist equipment to access information and communicate effectively, as well as equipment in the kitchen to become more independent and less reliant on others. This is where ASUK were able to provide a tailored package of equipment for the kitchen, through their Family Assistance Grants. This provided immediate support, by providing a Combination Talking Microwave and a Penfriend Audio Labeller for labelling items. This made a huge impact, giving Simmie independence in the kitchen and empowered her to start cooking for herself. She even made some muffins and sent the picture to ASUK. This featured in one of our newsletters.

Can you imagine not being able to communicate?

One of our members struggles to communicate with the outside world. She is a selective communicator and although she is happy to chat with her family, she has been reluctant to communicate with anyone else. ASUK worked with this young lady over many years and encouraged her to use her I-pad to record messages about her views on services. Over time the young lady has felt empowered to have independent consultations and the biggest achievement for our team was the day she sent a message, hearing her voice was an incredible moment. She now joins in conversations and has her voice heard!

‘My sister got to know all the staff at ASUK and she had fun with them. She likes to go to the residential and the transfer weekend, they have all helped to build up her independence and confidence. She trusted all the ASUK staff because she has got to know them and they know how to encourage her and not give up on her.’ older sibling

Unfortunately, it took many more months for the Local Authority to actually agree to the extra 14 hours needed. After many months; 2 specialist assessments, frequent home visits, and countless emails and phone calls to the Local Authority the extra 14 hours of support were agreed.

This delay and uncertainty impacted her mental health and wellbeing, but ASUK were there for her every step of the way, offering her support and reassurance.

With ASUK’s support Simmie successfully interviewed her Personal Assistant’s (PA’s) and we were also able to offer Alström Syndrome awareness training. This enabled the PA to better understand Simmie’s needs and the complexities of Alström Syndrome. This was welcomed by everyone involved and the relationship is flourishing!

Simmie is now enjoying her life to the full with time for exercise and social activities. She has support to go shopping and with the extra support she is more confident in her own home. She has a regular weekly music lesson, enjoys trampolining each week, is progressing at a regular dance class and has many more social opportunities than a year ago. She is no longer in danger of isolation which so many of our family members experience whilst living with deafblindness. Simmie is also offering her skills to others at a local day centre and has formed friendships with people at all these activities.

It has been a long journey, with stresses and frustrations along the way however, the courage and perseverance of our family member has resulted in a more enriched, healthy life. Simmie is pictured right at the ASUK conference enjoying a yoga session with her trusted guide dog.



Rambling Away - with ASUK support one of our young people is feeling empowered with an increase in his direct payments from 9 hours to 20!

You Cane Do It! - ASUK challenged a social care decision and gained an extra 15 hours of specialist support for one of our young people to develop his cane skills. His independence is now flourishing!



Around the world with Alström

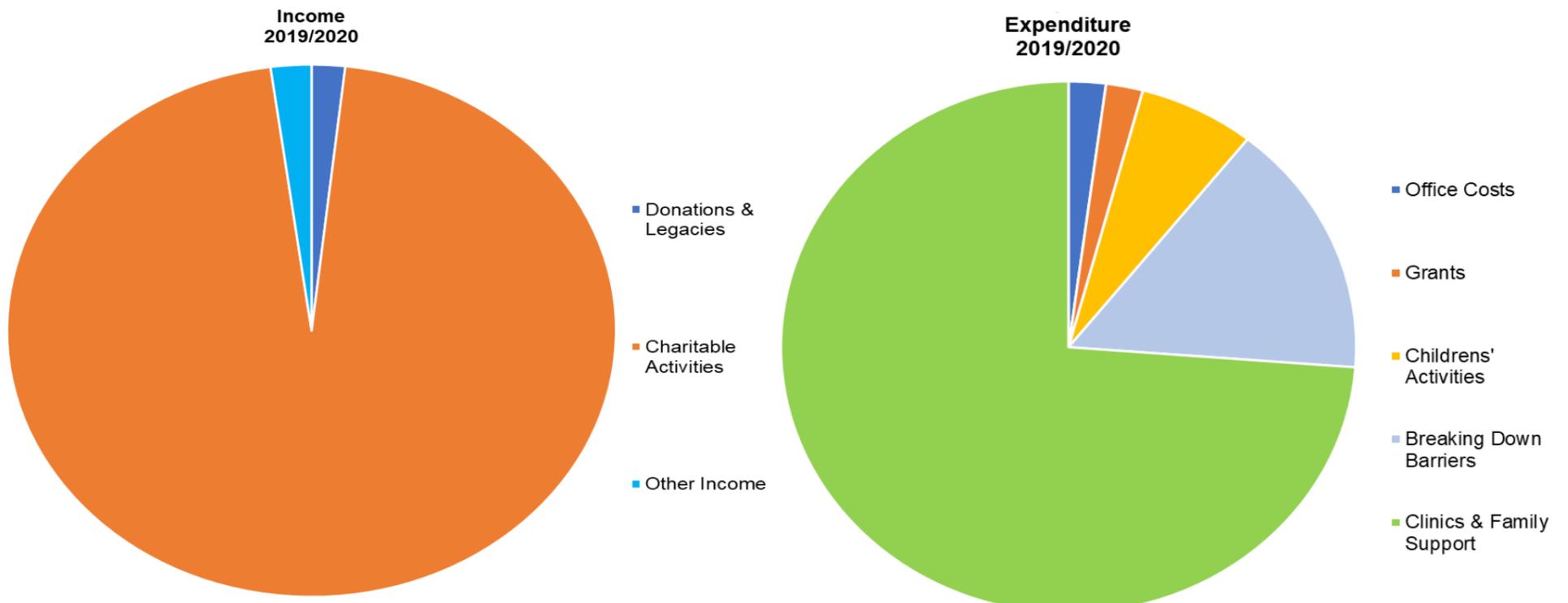
Awareness is vital to find those who remain undiagnosed. Highlighting the complexities of this ultra-rare condition to clinicians and researchers to find further treatments and aid better management.

- Alex our Vice Chair, attended and presented at the Alström Syndrome International conference in Texas, USA
- Families and professionals joined together to raise awareness on Rare Disease Day (pictured below)
- Connecting with families and professionals at Sight Village and other relevant events



Behind the scenes

- For the year 2019/20 there were **8** Trustees on the ASUK Board
- **8** people work for ASUK, supporting families, promoting research and raising awareness
- ⇒ Our website reached **42,540** views from **136** countries
- ⇒ Our 'Living with a Rare Disease' animation helped to raise awareness of AS reaching an incredible **1,265** views.



All information is provided from the financial period 31st March 2019—1st April 2020. The full annual report and accounts can be found on the ASUK website www.alstrom.org.uk

Thank you!

We are always immensely grateful to everyone who continues to support ASUK each and every year, whether it's through donating, holding a charity event, or giving generously from a Trust Fund. We make every penny count to ensure that we learn more about Alström Syndrome, that families and individuals get help when needed and have the opportunity to meet up and share stories and experiences.

We would also like to thank all the medical professionals at the Queen Elizabeth Hospital and Birmingham Women and Children's Hospital who work tirelessly to seek new treatments and ways to further manage Alström Syndrome.

