



Alström Syndrome UK

Strength for today, hope for the future

Alström Syndrome UK (ASUK) is a charity providing information, support and advice for individuals affected, their families, carers and professionals. Alström Syndrome (AS) is a very 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

Welcome to the Summer 24 ASUK Newsletter

Hello Everyone,

I hope you are well and enjoying the warmer weather. Our Summer newsletter is packed full of information and top tips, so I hope you enjoy catching up on everything that has been going on within the Alström Syndrome (AS) community.

Our small but mighty team have been supporting families, developing new connections, attending award ceremonies, raising awareness at conferences, providing an AS Global virtual conference, planning the ASUK conference and welcoming new families and team members.

We have had to say a fond farewell to our Global Executive Director, Ann Chivers. We are incredibly grateful to Ann for all the great things she has achieved during her time at ASUK. Her vision, guidance and support along with her fabulous personality really helped to shape and develop the team and strengthen the strategic direction of ASUK. Ann will be missed by the ASUK and AS Global community but we wish her all the very best. Catherine and I had the pleasure of enjoying afternoon tea with Ann before she left (photo below). **Thank you Ann!**

As one door closes, another one opens as they say... I am delighted that Melissa Crowland has joined our team as the new Wellbeing Coordinator. Melissa has settled in very well and is already making a difference. You can find out more about Melissa and the Wellbeing Service on page 5.

There have also been a couple of changes to the Board of Trustees. Melissa Crowland has stepped down and Alex Line has re-joined the Board. I would like to take this opportunity to thank all Trustees past and present for the support and direction they provide.

It's an exciting time as funding is made available to support rare disease research. Find out more

about the new LifeArc Centre for Acceleration of Rare Disease Trials on page 8, our work with Genomics England to address Equity in Health Research and our partnership with the Open University and an event planned at the British Science Festival on page 9.

In celebration of World Alström Syndrome Day, the AS Global community came together on the 2nd May 2024 for this year's annual lecture all about Alström Syndrome. Prof. Pietro Maffei from Alström Syndrome Association Italy presented his insights into what we have learned and how, through collaborations and networks, our knowledge and understanding of Alström Syndrome is progressing. Marina then spoke about her daughter's experience of education and what she wishes she had known when her daughter was at school. She gave us lots of helpful top tips along the way. Don't worry if you missed the session, check out the recording on the ASUK website

www.alstrom.org.uk/as-global/#World-AS-Day-2024

We are so excited to be planning the ASUK Conference 2024! For many of us, this will be the first time we have had the opportunity to be together in person since before the pandemic. The **'Together Again'** conference will take place on the **11-13th October 2024** in West Bromwich, near Birmingham. It will be fantastic to bring everyone together once again to all learn from one another, quiz the healthcare professionals and researchers and enjoy an evening of entertainment. We can't wait to see you all again.

Finally, I would also like to say a big thank you to all our friends, families and supporters who have given so generously to ASUK throughout the year. Check out page 4 where you can get involved in fundraising activities, without your support our work would not be possible.

Take care and have a lovely summer everyone.

We are here for you
Kerry and the ASUK team



9 years of Achievements

We have been very fortunate to secure Children in Need funding for 9 years. This funding has meant that we have been able to provide a dedicated National Transition Coordinator, Marie McGee to support children and young people throughout all stages as they grow up. This also included providing lots of fun-filled activities throughout the year, where families could come together and discover activities they never thought possible, such as driving a car or flying a plane. These activities and experiences were based around three key areas of their lives:

- Developing a healthy lifestyle.
- Improving social skills and friendships.
- Managing change at key times in a young person's life, this is also known as transition.

This project has now come to an end, so we are taking some time to reflect and shine a light on the achievements of our children, young people, and their families.

Let's see what difference the project has made to them.

Foodie Active Friends Club (Developing a healthy lifestyle)

In consultation with our young people and their families we provided creative activities that highlighted a healthy lifestyle and wellbeing in a fun and accessible way.

This grew into the Foodie Active Friends Club which forms the basis of the pre-clinic activities. Everyone involved had great fun with these hands-on activities, such as making fruit kebabs, creating pizzas, baking healthy hedgehogs with dough or making hot, healthy wraps with the George Foreman Grill as well as learning about portion sizes.

'The fruit kebabs were great to make. I got to hold fruit that was not in a package - I didn't know how some of them felt in their skins, like a pineapple or a melon!' young person aged 14.

During the activities, families have tried accessible cooking equipment and the dietitians from the Birmingham Women's and Children's Hospital who are part of the Alström Clinical Team have provided their healthy top tips. Everyone involved had a chance to try out cutting, rolling, chopping, and baking. Whilst learning how to use a knife safely, using timers for the ovens or grating cheese with several types of graters. The funding also provided items that enhanced the children and young people's access to outdoor spaces to encourage health living. This included providing tents



to be used at home or on holiday, play equipment such as balls of varied sizes with bells in them, and trampoline sessions as part of the Food Active Friends Activities.

'It's a good idea to do trampolining after a long journey to the hotel the kids loved it - so did I.' Mum

All the families took part in the sessions and enjoyed the learning aspect of the pre-clinic activities. Other aspects of healthy living and wellbeing we have covered included sessions about relaxation, sleep hygiene, managing periods and more recently oral hygiene. The feedback we have received has shown that the Foodie Active Friends sessions have made a significant difference to our children and young people. They continue to learn more about themselves, while being more aware and more informed about making healthy choices. They have tried a variety of foods, herbs, and spices which they may not normally eat and have learned how to measure food portions using their hands.

'I have never seen him so interested in different textures and smells; this is really good for him!' A sibling of a child commented- 'I like this activity I can help my sister and make a pizza myself.'

We need to have fun and a good laugh (Improving social skills while developing friendships)

This is where our children and young people had the chance to make new friends while jumping, skipping, climbing, running, and having a good laugh! This has been delivered through day trips, residential weekends or meeting each other during the AS clinics. Families from across the country, even as far away as Jersey and Ireland came together to enjoy each other's company. Parents and carers draw strength from each other, siblings find that they are not alone as young carers, and the children and young people meet those in their peer group where they bond and share their experiences. Family friends sometimes took part in the activities too, as they provide vital support to many of our families. We have been able to fund residential weekends where families have taken part in activities such as canoeing, sailing, caving and abseiling.

Young people have eagerly told us 'The obstacle course was a little bit hard and a little bit easy. Aisha laughed with delight as she said, 'I liked the zipwire, it was like flying!'. Not forgetting the campfires and sticky marshmallows, while singing songs and being silly with each other! One of the young people reported 'I give the residential 10/10, I liked it so much. I really liked the canoeing! While some dads even found time to unwind 'The yoga session for the dads was a good idea. It really helped me to relax'. Dad

Celebrating the achievements as the Children in Need project comes to a close

Recently, the younger children took part in a fun-filled residential weekend where they went to a farm, which included time to bounce, jump and run around an indoor play barn. They also met Dippy the Clown who squirted them with a large water pistol and taught them circus skills!

The weekend, which was a resounding success, was another chance to be silly and have some fun!

Eleanor declared, 'I liked making friends, playing and the animals.'

While Ayaan preferred the clown, 'Dippy was the best because he was funny'. Parents and carers also enjoyed the trip 'I absolutely loved the farm but most of all enjoyed meeting other families.'

Lock-down and the pandemic did not stop us from thinking of ways to get everyone together. Moving to online activities such as The Teddy Bears Tea Party helped to keep everyone's spirits up.

The ASUK Culture Family Forum (CFF), started to develop their meetings into an online group of young people aged 12 -18, who meet every month. Parents and carers also played an active role as they supported some of the young people to take part in the meeting. One of the CFF members Ibrahim says 'I like talking online and listening to the other CFF members. The CFF have been involved in testing out products, listening to guest speakers and giving their advice or opinions to the ASUK Board of Trustees. We are also delighted that some of the CFF members will be actively taking part in the ASUK Conference from the 11th-13th October 2024.

Growing up and trying new things (Managing change at key points in the young people's lives)

Our young people aged 11 onwards need support to cope with many changes in their lives. Over the years, we have supported them to take part in many exciting activities that helps to build their confidence, self-esteem and learn to take control of their lives.

Driving an adapted car was extremely popular, with many of the young people taking part in driving sessions across the country.

Amber shouted from the wheel of her car:

'This is amazing, I'm going to try and go very fasssst!'



Flying a plane is something that many people in life don't get a chance to do, however we made it possible for our young people to fly a plane. We have Hannah and Emma who have already flown a four-seater plane and Jai is booked to fly one in August 2024.

Work experience plays a big part in getting young people ready for the world of work. While staying with the theme of flying, Jai had a day at Birmingham Airport where he spent

time with the Customer Service Team to understand how an airport looks after their passengers, and Katie and Hannah have been working with a textile and print company. When ASUK takes on new staff some of them may be interviewed by a panel of young people and parents. When Hassan and Owais were younger, they interviewed Jane, who went on to be one of the Family Support Workers (FSW). Although Jane has left her role as a FSW she is now an ASUK Trustee.

During the AS clinics is another time when lots of education and opportunities takes place to build on the young people's skills. This includes supporting them to make decisions, speaking up for themselves or knowing when to disclose. This is where T-KASH (Transition – Knowledge And Skills in Healthcare) is used to underpin their knowledge and skills to manage their own health care. Other activities the funding has allowed us to provide were theatre trips, with Peter Pan, Snow White, The Lion King and Shrek, all giving us many laughs. Having a go at horse riding, cinema experiences, hair styling and swimming sessions. As well as buying items requested by the children and young people to enhance their everyday lives. This included items such as sandpits, swing chairs, male grooming products, bath lights or liquid level indicators.



ASUK can't possibly capture all the tenacity, bravery and resilience of our children and young people over the last nine years. Each one of them strives every day to cope with the challenges of Alström Syndrome. They have coped with the pandemic and came out the other side, ready to throw themselves back into their active lives again. They get up each day and they get on with their lives, supported by parents, carers, siblings, wider family members and friends who walk beside them offering daily support and guidance...

Continue on the next page...



A huge thank you to BBC Children in Need, who have given ASUK the opportunity to provide experiences and opportunities which we would never have been able to provide without this funding.

We would also like to give a special thanks to **Anne Cheesbrough**, a Senior MSI Practitioner from the charity Sense. Anne has attended many clinics and residentials, whilst delivering some of the activities herself. She has been a wonderful supporter and champion of ASUK. We would like to take this opportunity to give her a massive thank you for all she does and look forward to working with her for many more years to come.

Marie gives her thoughts:

'From my point of view, it's been a privilege to get to know every one of you, whilst having fun together. I've been so proud to see so many of the you mature, develop confidence, make decisions without support and eventually move into adult services. You have been my joy; it's been a blast! We will keep our fingers crossed that Children in Need will fund us next year.'

Marie will continue to develop and raise awareness of the T-KASH (Transition – Knowledge And Skills in Healthcare) and supporting young people as they grow

up. We will be seeking further funding so we can continue to provide activities for our community.

How you can help?

We have seen a reduction in the funding available to charities since the pandemic. Many of the patient organisations and support groups we work with are experiencing the same. We can no longer rely on grants and are having to look for alternative ways to bring in funding to ASUK. We are now managing a number of contracts and becoming increasingly involved in patient and public involvement in research. Our priority will always be to support people living with AS and we will continue to work hard to bring in funds to support health, wellbeing and happiness.

We are always hugely grateful for the funding we receive and would kindly ask the AS community to consider helping raise funds for ASUK. The possibilities are endless, you could hold a quiz with your friends and family, do a sponsored walk, run or even a danceathon!

Every penny really does make a difference . Please contact Catherine Lewis for your fundraising pack, catherine.lewis@alstrom.org.uk or pop along to our JustGiving Fundraising page to set-up your fundraising page: www.justgiving.com/alstromsyndromeuk



Up, Up and Away!

One of our families shares their once in a lifetime experience to actually fly a plane—this trip was made possible through the BBC Children in Need funding.

"I was gifted a voucher by Alström Syndrome UK to take part in a 30min flying experience. On the morning, I was due to do it, we got a phone call asking if I would like a 2hr flying experience as they needed to fly up to near Birmingham to change a plane over, I jumped at the chance!

I was a little nervous before we got there, however the instructor soon made me feel ok. He showed me a model of the plane we were going to fly and explained how it all worked. He then took me around the outside

of the actual plane and let me feel the wings and the tail comparing it to the model he had shown me. I found this really helpful to know, so that when I was flying it, I knew what was happening with the plane. On the way to Birmingham, he showed me how the flying instruments worked and how to move the plane from side to side and up and down. He then let me do a bit of flying before he took over to land the plane. On the way back, he let me fly the whole way back (he just took off and landed the plane), and just advised me when I needed to straighten up or go higher or lower. I really enjoyed the experience and I would do it again if I got the opportunity and I would really recommend it to others to do, it's a once in a lifetime opportunity."

Family Support Team gets a wellbeing boost! - Melissa joins the ASUK Team

We have some fantastic news - Melissa Crowland has joined our ASUK team as our Wellbeing Coordinator. Melissa has stepped-down from the Board of Trustees and will now work more closely with the AS Community to promote wellbeing.

ASUK carried out a survey at the end of 2023, to try and understand more about mental health within the AS community, and to look at how we may be able to offer more support.

Comments in the survey showed that people diagnosed with AS have experienced feeling low and isolated, this was especially relevant during the pandemic. The survey illustrated that participants believed that maintaining their wellbeing was important with over 80% rating its importance over five out of ten.

With this need in mind, we have now set up a new **'Wellbeing Service'**. We will develop 1-1 and group wellbeing sessions, webinars,, round table discussions prior to the AS multi-disciplinary clinics and signpost to relevant local organisations, as well as providing wellbeing resources and top tips.

"I also wanted to say, how lovely it was to speak with Melissa the other day! Speaking to someone doing so well that has Alström was just amazing. More of that would be wonderful. I genuinely am thankful that we have Alström UK, what would we do without you." (Parent)

Melissa (pictured above tells us a bit about herself;

"I am very new to Alström Syndrome, beginning my journey with the condition in 2018. It has been a very interesting journey but I'm very grateful for all the support I've had along the way which has given me insights into both my health conditions and myself. I qualified as a Social Worker in 2018 and have always been passionate about promoting and empowering independence both for myself and others. Since graduating with my Masters, I have dedicated myself to working with people with a range of disabilities, which has allowed me to achieve all that I have wanted to.



In addition to being the Wellbeing Coordinator for ASUK, I also work as a Counsellor and Crisis Recovery Mentor at a mental health charity. I pride myself on providing a safe, non-judgemental, person-centred environment for everyone I work with. In 2020, I decided to pursue a career in Humanistic Therapeutic Counselling which I completed in June 2023. I believe that everyone deserves a safe space, where they can speak freely, open and honestly without fear of judgement. This is what I endeavour to provide for the people I work with.

I also work for my local University as part of their Carer Advisory Participation Group (SUPA group). As well as being part of the interview process for prospective students, I also provide teaching support for lecturers and have created and taught sessions. I have also collaborated with colleagues to produce guides for both students affected by disabilities and placement providers to enable independence and participation, as well as a piece for the British Association for Social Workers about online working during lock down.

I have worked over a number of years facilitating a group for young people with Alström Syndrome, the condition which I have. This group provides a sense of community for the young people, where they can speak openly about how the syndrome affects them and be supported by both peers and myself. I also developed talking groups for adults with Alström during the lock down, again to provide a sense of community.

I believe that Mental Health is still stigmatised in our society, with individuals feeling reluctant to discuss any issues. I feel that in order for greater acceptance it is the job of leading organisations such as ASUK, to shine a spotlight on Mental Health provision, so that our families can be supported to flourish not only physically but psychologically. Research states that when an individual has good physical health then their mental health is improved. It is important to me that we create the right environment, so that people who are impacted by Alström Syndrome are supported so that they feel empowered to live their best life and develop insights into their thoughts, feelings and emotions."

ASUK at the Oscars!

The Let's Get it Right for Young People, T-KASH film was nominated for an award in the Charity Smiley Film Awards – the Oscars of the charity world.

Catherine Lewis, our Operations Manager and her husband Steve (pictured right) attended the night and gives her thoughts:

"Well... what a night at the Charity Film Awards... tissues were at the ready with so many amazing charity films, all with extremely emotional stories to showcase. Our T-KASH film unfortunately didn't win... but what an achievement to have our work recognised in this way.

The awards also generated an additional 126,000 views of the film and 2,800 visits to the ASUK website!

Well done to everyone involved."

Why not take this opportunity to shout about these vital resources for young people and check out the film too on the Breaking Down Barriers website:

www.breaking-down-barriers.org.uk/t-kash-transition-tools/



Grants for Holidays and Day Trips

Summer is finally here and we know how expensive it can be, so we have put together some information that may help. Some of these grant applications are only open for a short time, so please be quick if you would like to apply.



Orchard Holiday Park in Clacton-on-Sea in Essex. Families that meet their eligibility criteria can apply for a free week-long break, see the link below for more information

www.sunnydaysfund.org.uk

SEND Local Offers

For all children and young people with special educational needs (SEND) your local council must have a Local Offer. A Local Offer gives children and young people with SEND, and their families, information about what support services the council thinks are available in their local area. Councils are responsible for writing a Local Offer and making sure it is available for everyone to see. In Scotland this is called Additional Support for Learning (ASL) and in Wales Additional Learning Needs (ALN). Northern Ireland do not have Local Offers instead, they have the Special Educational Needs Advice Centre (SENAC), who provide independent advice to children and young people with SEND.

The link below will take you to the Well Child website. Follow the link to the Local Offer for the area where you live and you should be able to find out about any activities or clubs in your local area.

www.wellchild.org.uk/get-support/information-hub/send-local-offers-in-the-uk/

Family Fund

Family Fund offer a wide range of grants if you meet the eligibility criteria. You can apply online.

In England and Northern Ireland, you can currently apply for a grant every 24 months this includes holidays with Inspire, Butlins and Haven. In Scotland you can apply every 18 months. Unfortunately, the programme is currently closed in Wales.

Family Fund also manage some other grant programs, including equipment for adults aged 18-24 years. The site is regularly updated so it's worth checking regularly

www.familyfund.org.uk/grants/schemes/

Happy Days Children's Charity

Happy Days fund a variety of day trips and self-catering short breaks for families on lower incomes.

These include Haven, theme parks, Centre Parcs, cottages breaks and Butlins.

Further details and application forms can be found on their website www.happydayscharity.org

Sunny Days Children's Fund

Sunny Days have two holiday homes at the Haven

VICTA

VICTA is a national charity that provides support to children and young adults from 0 to 29 years, who are blind or partially sighted and their families. VICTA run a range of different holidays, activities and courses both for families and individuals to attend independently. These are often subsidised or for a minimal fee. You may be able to use direct payments or a short breaks payment to fund the break if you receive this support from social care. You would need to speak with your local authority to check if you can use your payments to fund this. www.victa.org.uk/our-services/

ASUK Adult Empowerment Fund

A reminder to those of you who have not previously applied, or who have not had a grant recently, ASUK offer adults (18's and over) with Alström Syndrome, the chance to try something new. You can apply for between £50-£500 to pay for an activity, all we ask you to do is write an article for publication in the ASUK newsletter (within a month of the activity) with a few photographs to publish too if possible. If you would rather tell us about your experience, we can help you produce a podcast for the ASUK website.

BBC Children in Need Emergency Essentials Programme

for children and young people aged 17 years and under. ASUK can access the BBC Children in Need Emergency Essentials Programme which supports children and young people who are facing exceptional difficulty. It can deliver or fund critical items such as: large home appliances, furniture, kitchen equipment, small appliances, children's beds and bedding (including cots), baby equipment and clothing. So, if you face a break down of a critical item please get in touch with Sarah Oliver and we will see what we can do Sarah.oliver@alstrom.org.uk

Freebie Activities and Meals

Money Saving Expert have brought together some free activities and meals to help you throughout the Summer holidays www.moneysavingexpert.com/deals/things-to-do-with-the-kids/

For a full list of grants pop along to the ASUK website for full information

www.alstrom.org.uk/family-support/

Enjoy yourself, have fun and smile!

Our National Transition Coordinator, Marie McGee catches up with one of our families, and asks them about specialist residential schools, independence and supporting one another (photo right of Ummi, Aisha and Muhammed at one of our residential week-ends).

Mum, Ummi gives her views on residential schools.

'Residential school is a place to learn independence and life skills that are essential for future success.'

Why did you chose a residential school for Aisha?

I have wanted Aisha to be in a space where she will become independent within a short time and be in the midst of people like her. She will feel a sense of belonging more, to connect with her peers and share similar experiences and challenges. Residential school is a place to learn independence and life skills that are essential for her future success.

How did you decide it was a good place for her?

I have observed that out of instinct and too much love, I do a lot of things for her, I cook for her, serve her, clean for her and do loads for her. So my dream and ambition for Aisha to become a successful young lady, with a good career and a good life made me, send her to a residential school. As I know she will learn life skills that are very important for her confidence and future.

How did you prepare for her to leave home?

Aisha was very excited as she understood the need to be independent. So the preparation was very easy. We went shopping and she bought all the things she wanted, like toys, radio, books, and toiletries and a brand new suit-case that is purple for her. She said bye happily to me when I dropped her off and I cried throughout my way home.

What is the difference being there, is making to you and her?

Her staying at residential school is life transforming for her. It wasn't easy for me from the beginning as I was too attached to her. But when I started seeing her handling her things without my prompt and she is always eager to leave my house on Monday, it became a part of me for her to only be around during the week-ends. She is so happy and comfortable at school. **She loves it.**

Aisha talks about what it's like to live in a residential school.

Mum interviewed her daughter Aisha about her thoughts on her time at residential school.

What is it like living at a residential school, Aisha?

Well, it's eye opening and exciting at the same time, because you get to do a lot of activities and be independent. You learn how to do dishes, use the dishwasher, you learn how to make your own bed and cut your own food. You can do lots of things there and they also teach you how to serve up your own dinner.

What sort of things do you do in the evenings?

Art, relaxation, cooking, scouts and a few other things like talking to my friends, and having karaoke and

parties.

Aisha, when you go home are you more independent at school or at home?

I think in school, because I do more than just sit around and at home my mum does most things for me. I could be independent at home too as I know how to use an induction hob safely and know about kitchen safety rules.

How has your independence improved?

I am quicker in walking to a lesson, and I am quicker at doing things like downloading documents that the teacher sends. I don't need any sighted guides.

What top tips would you give someone who is thinking of going to residential school?

Enjoy yourself, have fun, be yourself and smile the biggest of smiles, because if you share your biggest smile with everyone it will leave them with a big smile as well.

Aisha's brother Muhammad talks about how he supports his sister.

Having a sibling with Alström Syndrome (AS) can be tough at times, but it really teaches me how to deal with certain situations; like when my sister is having a meltdown, I have to try to calm her down etc... I normally have to help her with things like her food, tying her laces, turning the tv on, helping her charge her phone, making sure she knows routes of our house by describing where everything is. Also, try to be an even better brother by the day in a myriad of ways, by making sure she enjoys her time with me so her mental health will be very stable and steady.

Why its important to help someone with AS?

I understand AS comes with a lot of difficulty such as visual & hearing impairment, diabetes, it affects organs like heart, kidneys a lot of things. So it is important to help people with AS to make life easier and the pain they go through bearable. I also want to see my sister successful and happy and for her to reach her potential. **I am happy when she is happy.**

Have you leant any new skills?

The main skills I have learned is emotional intelligence and patience.

What advice would you give to other brothers or sister who have to help someone with AS?

Tips for anyone who has a sibling with AS: always be understanding when they are in a rage or are sad. Try to spend and cherish your time with them to make sure they enjoy being around you.

Best of all, be their favourite!



Research News

Rare Disease Centres get the Green Light
LifeArc launches £40m research centres to hopefully unlock new tests and treatments for people living with rare conditions.

We are pleased to announce that funding has been approved to develop The **LifeArc Centre for Acceleration of Rare Disease Trials** with expertise from Newcastle University, Queens University Belfast, and University of Birmingham, led by Prof Tim Barrett. ASUK is actively involved and will help lead patient and public involvement and engagement and make sure the needs and views of people living with rare conditions is embedded throughout.

Kerry, CEO of Alström Syndrome UK/Breaking Down Barriers, says:
“We have no specific treatment for Alström Syndrome and when my son, Kion, was a baby, I was told it could take around 10 years for any treatment to be developed. 20 years later, we are still waiting. People living with rare conditions don't have the luxury of time and the mainstream way of delivering healthcare and drug development rarely works for people with rare conditions. As a mum and the Chief Executive of Alström Syndrome UK, having a centre that will deliver a coordinated, inclusive and supportive approach to accelerate clinical trials gives me great hope.”

Thank you to LifeArc and everyone involved in the development of these new research centres.
www.lifearc.org/2024/lifearc-launches-40m-research-centres-that-will-unlock-new-tests-treatments-and-cures-for-people-living-with-rare-diseases/

One of our young members, Edward gives us his thoughts about research and how to get involved (pictured below, at his recent prom).

“Hi, I'm Edward, I'm 16 years old and I have Alström Syndrome. I was diagnosed when I was aged 11, now I am multi-sensory impaired, but this does not stop me from going out and being independent. I have started to be interested in research and find out a lot more things about Alstrom Syndrome. I like to find out what causes it and how it can be treated.



I found out about Alström Syndrome research from the ASUK team when I visited the Birmingham Women's and Children's team for my first medical appointments. The ASUK team share their latest research with young people by sending emails or through the email newsletter. Young people can ask their GP about research into Alström Syndrome. They can also ask if there are any medical trials happening and how they can take part in them. I am interested in speaking to other people about Alström Syndrome and collaborating with professionals, young people and adults about future research into Alström Syndrome.”

All about the Alström heart - series is out now!

THE HEART is a clever PUMP! an extraordinary organ! goes up when you exercise. NORMAL RATE 60-100 beats per minute. electrical system. from your lungs. valves. squeeze muscle.

RESEARCH Be Inspired Be You Be Together. A 54 AGEING preliminary work PUBLISHED. people with AS seem to AGE quicker... The HEART can have a huge IMPACT. immunity? cells? fat metabolism? Alms1 protein? what drugs may help? seeking funding for 3-5 years. TRIALS

HEALTHY HEART Be sensible! Know your limits! rate up! (220-year age) x 50-70%. STEPS: ideally 12000 a day. BENEFITS START WITH 5 minutes a day. healthy & balanced diet. Your doctor can advise. START NOW! IT'S NEVER TOO LATE. AIM FOR 2.5 HOURS A WEEK. UNDER 18? AIM FOR 5 HRS!

LOOK OUT FOR hard! out of breath! heavy! (NORMAL ACTIVITIES) DO WHAT YOU ENJOY

The Alström Heart Prof. Rick Steeds in conversation with Ann Chivers

...may be THICKER, HEAVIER, STIFFER due to... kidney function "bad cholesterol" DIABETES INSULIN RESISTANCE. CAN AFFECT babies heart problems ADULTS serious impact

NT Pro BNP Echo Cardio Gram ECG MR1. CARDIOLOGIST sees you every 12 months

thinkbigpicture.co.uk www.alstrom.org.uk

AS Global Director, Ann Chivers and AS Expert Cardiologist, Professor Rick Steeds talks all about the heart in a series of 6 episodes. Pop along to the ASUK website to watch:
www.alstrom.org.uk/as-global/#All-About-The-Heart



Alström Syndrome UK

Strength for today, hope for the future

Equity in Health Research

ASUK have been supporting Genomics England to address Equity in Health Research. Information held in databases is not representative of the ethnicity of the UK population. With the development of personalised medicine comes the real risk that some communities will be left behind. This will widen the health inequalities gap that already exists. We are working with Genomics England to develop a framework to improve innovation and research practices with the voices of people from underserved communities at its heart.

Genomics England are working with a number of community groups to gather insights from people with lived experience. They are also engaging with a panel of professionals working in this field to further understand challenges, barriers and opportunities. Findings will then be used to create a new framework and strategy.

ASUK/Breaking Down Barriers have been working with Genomics England on this important topic and supporting them with the panel discussions and engagement workshops. Bringing together community groups and professionals

in this field to learn, discuss, share insights and experiences and develop next steps.

We are also working with many organisations who have been developing good practice in this area of work for many years, so we can capture the existing tools and practices currently being used.

<https://breaking-down-barriers.org.uk/equity-in-health-research/>



Explore the Tree of Life

Explore the Tree of Life at the British Science Festival 2024. Join Breaking Down Barriers, Central and South Genomic Medicine Service (NHS), and the GRACE project (The Open University) in a series of creative workshops designed to uncover the missing pieces in our data and ask what it really means to be representative.

Celebrating the people, stories and ideas at the heart of science.

The British Science Festival is Europe's longest standing science Festival, travelling to a different place in the United Kingdom each year. The Festival aims to connect people with scientists, engineers, technologists and social scientists.

Each year, they offer an inspiring programme of free events to the public over five days, bursting with exciting opportunities to get involved in. The talks, workshops and drop-in events span a diverse range of subjects that encompass science in the broadest sense, promising something for everyone.

The British Science Association and the University of East London (UEL) are pleased to announce that the British Science Festival in 2024 will take place in London across five days. The Festival will be held between **Wednesday 11th – Sunday 15th September 2024.**

For access information and directions to this venue, please visit the website below:

<https://britishsciencefestival.org/british-science-festival-2024-venues/>



There are no Strangers Here, Only Friends You Haven't Met Yet

Melissa spoke with two young adults about the important role that friendships play in our lives. "Friendship is hard to find, harder to leave and impossible to forget" (G. Randal).

Friendship plays an important function in our lives; whether it's putting the world to rights, laughing until it hurts or having someone walk beside you. Life is more fun with a friend at your side. Being able to share experiences with other people is important as it can reduce feelings of loneliness and isolation.

This article explores friendship from the Alström perspective and will discuss how friendship impacts those with Alström Syndrome. After all, "let us be grateful to those who make us happy, they are the charming gardeners who let our souls blossom" (Proust. M.)

For some people it has been difficult making and maintaining friendships because of their condition. For example, it can be difficult understanding facial expressions and making eye contact and if you have a hearing impairment you don't always know when you're being spoken to. Also, the complex nature of Alström Syndrome can mean that you have multiple conditions to deal with, where the symptoms or treatment you're receiving may make you feel poorly.

"It has been challenging for me to make friends because when I was in mainstream school it was hard for me to explain my syndrome to others because I wasn't diagnosed with Alström Syndrome until I was 30. I was sick lots, with colds, flu, digestive problems, and headaches so I missed loads of school which was hard to explain. When I started going to the deaf and blind school in Middle School, it became easier for me to make friends because everyone there was either deaf or blind or deaf blind like me so that was helpful."

"I feel like being with younger kids or kids with disabilities it's easier to make friends but when I'm around people my own age it's really awkward because they have jobs, are at college or dating or are married or doing something fun".

Although there can be some significant challenges for people with Alström Syndrome when making friends; this article will now focus on how people with Alström Syndrome have found friendships, and how these relationships have flourished and thrived.

Alisha and Alisha have been friends for just over a year. They met as the result of a YouTube video Alisha made focusing on the voices of young people.

"She seemed really open and cheerful and was around my own age, so I wanted to talk to her and see if we had anything in common. I asked one of the Family Support Workers at the Alström clinic if she could put us in touch and our friendship's blossomed from there" the other Alisha says.

They enjoy hanging out together, watching videos, chatting and sharing tips about perfume and jewellery and generally having a good catch-up, whether it's in

person or on the phone. "It's good to have someone who has the same interests as you, but it can take forever to get to know a person properly".

They have built up a strong friendship over the past year, but it has taken time and effort on both their parts: "it's important to get to know each other and make sure you've got some rules in place. So, your friend knows how often you're available to speak to them. It can also sometimes be difficult when your friend has a lot of medical issues going on, you want to be there for your friend, but at the same time it's not up to you to fix that problem. You can just be there to listen".

Both Alisha's stated the significance of having Alström Syndrome and the impact that this has had on their friendship: "she makes me laugh when I'm down and understands what I am going through as she has experienced similar things so I know I can speak openly to her about what is going on with me medically". "Alisha has always been very supportive of my physical and mental health. I can tell her what I'm struggling with, and she can tell me about what's helped her through that situation".

They stated that their shared condition made them feel closer as friends as they understood some of the issues that they were experiencing, and they could work through coping strategies together. "You're stronger when you have your friend by your side as she can be there for you to talk to about anything".

They have both spoken in depth about the importance of boundaries and speaking honestly about what they can bring to the friendship "I do try to say when something is overwhelming for me and when I need my own space". They say it is also important to be transparent about whether you do want to be someone's friend and how it is crucial to have appropriate friendships. "If you don't want to be friends you should just say and then you're not wasting anyone's time", "you're not going to be friends with everyone, but that's ok and it's important to have things in common with your friends".

Alisha and Alisha live relatively close to each other, and they can meet up and do things face-to-face. They appreciated that friendship can also be a long-distance thing "although it's nice to physically see your friend, you don't always have to be face-to-face. Talking over the phone or on social media is great too. But when talking to someone online you must be careful and make sure you are safe".

They both spoke about the positives they had gained from speaking to individuals who have Alström from around the world, "it's nice to speak to more people and learn about their experiences of Alström". "Talking to people from around the world reminds me that I'm not alone with this condition, and sometimes they have helpful tips about what's helped them".





Both Alisha and Alisha had some positive things to say about the importance of friendship, and why they appreciated each other “When you’re happy or sad or have any news to share it’s good to have someone who’s your own age to talk to”. “She’s really funny”, “she’s happy and makes me laugh all the time and there when I need her, she’s so sweet”.

Having friends isn’t always sunshine and rainbows though, and it’s important to create healthy boundaries with your friends. When asked what they would like to change about their friendship they answered: “have a bit more time for me, sometimes life can get busy, and you don’t have time for your friends. It’s important that your friend reminds you to check in with them”.

What are the Alisha’s top tips for making friends?

“Try to find out about each other and keep asking questions, meet up, or speak regularly if you can, get to know their likes and dislikes, and don’t be shy and be open with them about what’s going on in your life. Even though it’s not their job to help, talking to someone does make you feel better about things”.

This article has discussed the importance of friendship. How, having someone who can listen and empathise with the complexities of Alström Syndrome is beneficial, as it helps individuals with the condition feel listened to and heard.

This article has also discussed the importance of transparency and honesty in a friendship. How being open with our friends about what is going on in our lives is important as it allows other people to know what you need from a friendship and what they can expect.

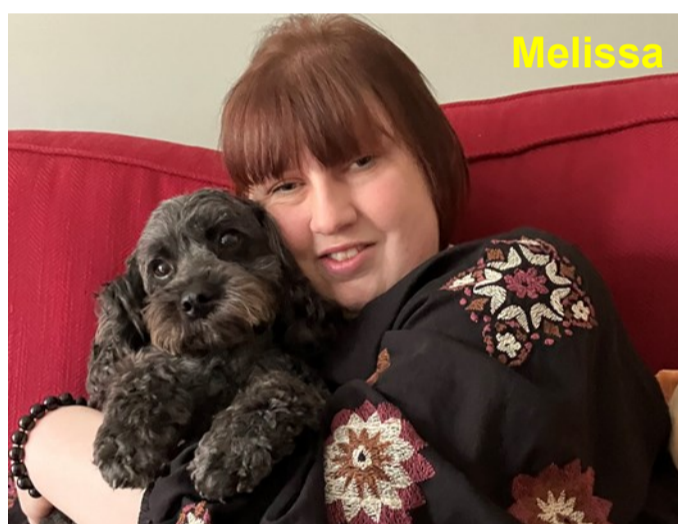
The Alisha’s also spoke about how having mutual interests and committing to regular catchups, whether this be face-to-face, or over the telephone is crucial to develop a friendship.

Friends come in all shapes and sizes, some are human, and some may be furry (referring to pets here, although sometimes I guess humans can be furry too!).

In the words of A. A. Milne’s beloved Winnie, the Pooh “a friend is someone who helps you up when you’re down, and if they can’t, they lay down beside you and listen”.



Alisha



Melissa



Alisha

Invitation



Dear Families and Friends,

You are invited to our Family and Professional Conference 'Together Again' on the 12th October 2024 at the Mercure Birmingham West Hotel

Come and join us for this informative day of research developments, family stories, interactive workshops and get the chance to quiz the clinicians and researchers, this will be followed in the evening by dinner and entertainment. This will be a unique opportunity to hear from leading Alström Syndrome experts, access personalised support and enable invaluable time to socialise together, catching up with old friends and have fun making new ones.



Reserve Your place today call or email Catherine Lewis on email: Catherine.lewis@alstrom.org.uk or call: 07970 071675

If you need accommodation, you will have to be quick to take advantage of the **early bird rate** of £109.00 per room, per night on a B&B basis, (sleeps 2 adults and 1 child) and if you need a second hotel room, this will be charged at the lower rate of £70. Grants are available.

Check out the website for more information, or scan the QR code for the booking form.

<https://www.alstrom.org.uk/book-now-as-conference/>

We look forward to seeing you all there!



ASUK offer a help-line during office hours and an answer phone service at all other times where calls are promptly returned by our experienced team;

Please call: 07970 071675 / 07716 135940

Email: Catherine.lewis@alstrom.org.uk

Kerry.leeson@alstrom.org.uk



THERE FOR US

Meet the ASUK Team



Jas



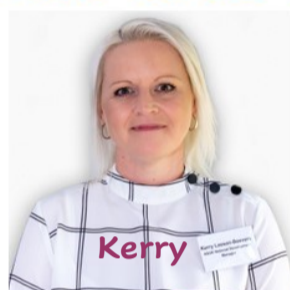
Sharon



Carol



Lexi



Kerry



Catherine



Kez



Trevor



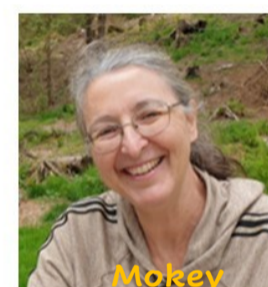
Melissa



Haris



Clair



Mokey



Jane



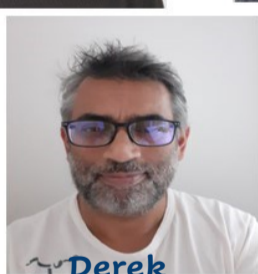
Tim



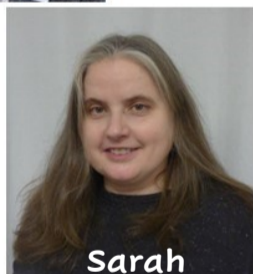
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Please visit our website www.alstrom.org.uk to keep up to date with our latest news.

ASUK has a dedicated You Tube page <http://www.youtube.com/alstromsyndromeuk>

Please keep up to date with Alström Syndrome UK via X [@AS_UK](https://twitter.com/AS_UK) and also via our dedicated Facebook page www.facebook.com/alstromsyndromeuk please click 'like' to show your support and spread the word!

You can donate securely online, visit our Just Giving page to make a difference today, <https://www.justgiving.com/alstromsyndromeuk>

