



Alström Syndrome UK

Strength for today, hope for the future

Alström Syndrome UK (ASUK) is a registered 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 2023 ASUK Newsletter

Hello Everyone,

I hope you and your family and friends are all keeping well and enjoying a lovely summer. It has been great to see so many of you getting out and about and having fun. I love to hear and read about your adventures and see some of the new experiences and activities you are taking part in.

I can't tell you the excitement that the team and I felt when the news came in that Lexi and Adam were expecting a baby. You can find more at the bottom of this page. Welcome to the world baby Amelia and huge congratulations to Lexi and Adam!

I don't think we have ever had as many uplifting family stories to share and celebrate with you. Read on to hear about; Hassan's latest challenge as he raises £1,500 after climbing Scafell Pike in the Lakes, Hannah flourishing at University, Millie setting up her own pop-up shop, Melissa completing her studies, J-J enjoying his judo camp and many of you going off to your proms in style!

We are also celebrating at ASUK too, as we reach 25 years of supporting people with this ultra-rare condition. Families will receive a memento – a small pin badge as a keep sake of this important milestone.

Fond farewell

Sadly, Toni left ASUK in January due to family commitments. We are thankful to Toni for all that she did during her time at ASUK and wish her all the best.

Dr Richard Paisey stepped down from his role as ASUK Trustee in May. Many of you will remember Richard as the first adult Clinical Lead when the first clinics were set up in Torbay. When the adult clinics then moved to Birmingham, Richard kindly agreed to join the Board of ASUK and has continued to provide us with his support, knowledge, and expertise over the years. We would all like to give a HUGE thank you to Richard for his dedication and commitment to Alström Syndrome UK and the many individuals and families he has supported along the way. We are so pleased that Richard has agreed to stay involved as our Medical and Research Advisor.

A warm welcome to ASUK

We hope you will join us in giving a very warm welcome to a new member to our team. We are pleased to have Clair Pudaruth joining us as our new Alström Syndrome Care Coordinator and will be working alongside Sarah Oliver. You can learn more about Clair on the next page.

As we approach winter, please remember that our AS doctors recommend that everyone eligible should have the flu, Covid and pneumonia vaccinations and boosters.

The cost-of-living crisis is likely to continue to hit our community hard so please get in touch with us if you need any support. We have provided further information and guidance on our website www.alstrom.org.uk/family-support/

Remember that ASUK grants are available for activities and equipment and to help you through the cost-of-living crisis so please contact us to find out more.

Thank you

As always, a big thank you to all our families, friends and supporters and to the medical teams in Birmingham for your continuous support.

We are stronger together so let's stay in touch.

Stay safe and look after yourself and your loved ones.

We are here for you.
Kerry and the ASUK team

Baby Amelia is born!

After 54 days in the neonatal intensive care unit, beautiful baby Amelia Elizabeth Line has been brought home by Mum Lexi and Dad Adam. Amelia was born early on the 7th June 2023 weighing a tiny 2lb 3.5 oz.

I know you will join us in welcoming baby Amelia to the world and sending her the biggest hug and best wishes to the family!



Over to you... family news!

Celebrity in our midst!

Millie and her Mum Michelle, have opened a **pop-up shop** in their garden after, struggling to find a summer job for Millie. The family have been loaned a shipping container and gifted many of the artisan products to sell from their home.

This fabulous initiative has had them featured in the press, on the radio and even on the ITV news... they even roped in ASUK Office Manager, Catherine Lewis who was interviewed about the issues facing young people looking for work.

Michelle says "it's wonderful to see Millie learning new skills and gaining confidence".

Don't worry if you missed, Millie, Michelle or Catherine, you can catch up on the ITV news story with the link below:

<https://www.itv.com/news/westcountry/2023-08-02/teen-runs-up-pop-up-shop-from-garden-after-struggling-to-get-a-job>



Adventures Galore!

You may remember reading in our previous newsletter that Hassan was off cycling across Europe... well he has been off on another adventure...

He has raised £1,500 after climbing Scafell Pike in the Lakes - it took over 13 hours to complete!

Hassan said: "I feel proud and privileged to have now most recently completed the highest Mountain in England, Scafell Pike."

Where will his next adventure take him...

Well done Hassan!



Join us

Hear about our work and developments over the last year, at the ASUK Annual General Meeting. Join us virtually on zoom on the 22nd November 2023 from 7-8pm (UK time).

Get in touch to register
with Catherine at catherine.lewis@alstrom.org.uk

Fun for All!

It has been a busy Summer and we know that families have been out and about enjoying the glorious weather. ASUK has been busy supporting children and young people to take part in activities and opportunities which have been funded through a BBC Children in Need grant.

Whitemoor Lakes Residential – Getting to Know You Weekend, April 28th-30th 2023



At long last ASUK were happy to bring families together once again, for a fun-filled residential weekend in April 2023. There were lots of opportunities to take part in all sorts of activities such as abseiling, canoeing, arts, crafts, and a campfire sing song! The activities were aimed at children and young people from age 8 upwards.

Located in the heart of glorious Staffordshire, Whitemoor Lakes Activity Centre provided the backdrop for the weekend, which was full of adventure, laughter, and fun!

21 families aged 8 plus were invited to attend the residential, which was funded through BBC, Children in Need (CinN). 8 families attended along with siblings, carers, and family friends. They were supported by ASUK Staff and volunteers from SENSE and ASUK Trustee's.

The CinN funding aims are:

- ♥ to support families to come together
- ♥ for young people to have fun
- ♥ to speak to someone new
- ♥ to try something new or something they have only tried once before.

The week-end was full of smiles and laughter. It was clear to see the bond that ASUK families feel when they come together to share experiences and build resilience to live their lives in the best way possible!

Some fabulous quotes from our families who attended:

"X has been on the residentials before but he would not have a go on the zip wire or abseiling. This time he tried everything; you have no idea how proud I am of him- he's becoming so much more confident. ASUK have really supported him to grow and change in his abilities!"

"I enjoyed being all together and talking to the other parents. The activities were great confidence builders for the kids!"

"Well done, Marie - You did an amazing job, as always! You always go the extra mile, but it really paid off! A huge thank you to you and the volunteers, for all the support you provided throughout the weekend. Great to see so many people interacting in shared experiences. Peer to peer support is so important for people living with AS. We get our strength from one another."

If your child or young person wants to visit the theatre, take part in day trips, try driving a car or a fire engine, build on their health and fitness, is looking for personalised resources, or to take part in other activities please contact Marie McGee, ASUK National Transition Coordinator by email;

marie.mcgee@alstrom.org.uk



Meet our New Alström Syndrome Care Coordinator!!

We would like to warmly welcome Clair Pudaruth to our team. Clair is joining us in August as our new Care Coordinator, working alongside Sarah Oliver. Clair will be helping to coordinate the clinics and providing family support.

Clair has extensive knowledge and experience as she has worked for 13 years with Retina UK. Retina UK is a national charity that supports people living with inherited sight loss conditions, including complex syndromes.

We are sure it won't be long before you meet Clair in person at one of the AS multi-disciplinary specialist clinics.

Clair can be contacted by email
clair.pudaruth@alstrom.org.uk

or by phone or text **07825 384362**



Hann the Historian – talks university, clubbing and purple hair!

By Hannah Bromley-Challenor

When I left Sixth Form in the summer of 2020, I was socially isolated and had been confined at home for over a hundred days, which at the time, felt like a hundred years. I had taken my 'A' levels in isolation and had no idea how the results were going to turn out, or what I was going to do next. University had not really been discussed at my last school, especially since I had been home since March and was not sure if it was even an option.

Once the exams were done, I spent my time listening to audiobooks on historical fiction, classical literature, and comedy, to keep myself occupied until the results arrived. Finally, they did, and I was pleased to see that I had passed. Still, with no idea where I was going next, one day that August I decided to enter clearing to see if I could get any offers to study history at university. I was interested in Ancient, Classical or Medieval History, and I was pleased to see that by lunchtime I had received about 6 offers from universities all over the country, from Bristol, Leeds, Bangor and Swansea in Wales, as well as a few closer to home. I finally decided on Medieval History at Winchester, which was a bonus on two counts – it was only an hour or so away from home, and it is a beautiful city drenched in history, from its Roman connections, with the walls still visible today, to being the capital of England under Alfred the Great. Winchester's cathedral is full of Saxon tombs, and it was also the location where the Anarchy during King Stephen's reign ended with the Treaty of Winchester, in 1153. I couldn't believe that I was going to get the opportunity to study in a city so filled with the history of my favourite period.



When I accepted my course, I decided that I wanted to do a Foundation Year. I chose to do it this way for a few reasons, but mostly because of the uncertainty of more lockdowns being imposed, I did not want to spend all my first year of my course at home. As it turned out, this was a good choice, because most of that year was spent in lockdown at home, but I had my Foundation Year lectures and seminars on zoom. Despite not being physically at the university for much of the time, I really enjoyed the Foundation Year because I got to meet a lot of other students who were also passionate about history, as well as students from other Humanities subjects. A Foundation Year is also helpful for learning transitional skills and you get to see what sort of assignments you will be doing in your actual degree. It also gives you time to assess whether you have made the right choice, so, if you want to change courses, it is not as disruptive as if you changed during your first year.

Studying over zoom, and having lectures and seminars this way, worked out quite well for me whilst I was at home because I could adapt the room to my needs with regards to my light sensitivity and hearing impairment. Lectures were also pre-recorded, so I could take notes at my own pace, as well as go back over things I might have missed in the live sessions. I also found that the live sessions worked better with my hearing impairment, as I could contribute to the conversations a lot easier than I sometimes can in lecture rooms. My fellow students were very considerate about my sight loss, describing images or videos to me, or sometimes telling the : ˙ ˙˙ :˙˙˙˙˙˙˙ (lecturer) at the beginning of a session, on my behalf, which resulted in one of them very kindly describing what the Domesday Book looked like.

So, in September 2021 I was physically at the university full time. Now I was able to properly start getting my teeth into Medieval History, doing two modules on early Medieval Britain and Europe in the first semester and two on late medieval Britain and Europe in the second semester. I also chose a case study module which focused on the reigns of Richard II and Henry IV. It was great to learn from lecturers who are so passionate about their subjects and specialties. For instance, one of my lecturers is an expert on the Wars of the Roses, so when he did a lecture on it, you could really see how much he enjoyed teaching it, which made it even more enjoyable to learn.

Exams at university, and essay writing, are completely different from 'A' levels. This was the first year that I had exams which I was worried about, to the point I was considering if I could take a different type of assessment, but the Disability Team at Winchester were brilliant with putting a learning agreement together to help me get through them with reasonable adjustments like having a reader and scribe, rest breaks and extra time to complete the exams in, and I have had some of my best results from the exams, which is something I didn't think would happen!

Socially, I have tried to get involved too. During my first year I joined the History Society, who were great at making sure things were accessible to me. We had a Christmas meal out, quizzes, history talks, and we did a trip to the British Museum.

My favourite year at university so far must be the year I have just finished, my second year. I really enjoyed the modules, because the choices were so interesting, and there were some unusual ones that were great fun. For instance, looking at society through the food and drink in the medieval and early modern period which roughly covered from after the Black Death to just before the Industrial Revolution. I also got to study the reign of King John and the Vikings, which I have always enjoyed.

I also joined the new Taylor Swift Society who I found at Welcome Week last September. I absolutely love spending time with so many fellow Swifties who have been brilliant with guiding, describing music videos, putting audio description on for a film, and they took me to my first ever nightclub in Southampton, as well as a Halloween party on campus. I also go to a wellbeing café on campus, where you can relax with other students and get a free Costa drink. It has student listeners who are brilliant, and I've made a lot of friends here.

I even went to my first protest in December to support the staff going on strike. The following month this empowered me to start up a petition, to get all products sold in the UK to have clear Braille labelling. This includes toiletries like hair dye, as since coming to university I have started dying my hair, making purple my new signature colour! (Hannah rocking her purple hair right)

Next year will be mainly doing my dissertation, which will be on Robert of Gloucester and the Anarchy. This will involve studying plenty of chronicles and I can't wait to dive into them, to see how history was portrayed by the writers at the time. I'm also doing modules on chivalry and an in-depth study of the Vikings.

In the future I hope to write a book, The Medieval Calendar. I got this idea from last Christmas when I posted an historical event or fact for every day in December up until Christmas Day. People said they enjoyed it, so I thought it might be good to extend it to a full year.

I have really enjoyed my time so far at university, as it not only gives me a chance to study something I'm passionate about, but it has given me new friends, and helped me grow into the person and Historian I would like to be.



Let's dance the night away!

It has been fantastic seeing your prom pictures on social media – especially watching Jai be carted off by the police so he could arrive in style in a police car!

What a fantastic experience and don't you all look fabulous!



Out and about... at Sight Village

QAC Sight Village is described as ‘the UK’s leading exhibition for blind and partially sighted people and for those experiencing sight problems’. This year ASUK had an information stand to raise awareness and also to present to attendees about the newly developed T-KASH resources (see page 9 to find out more about these vital transition tools.)

The event attracts thousands of people to explore the latest technology, products and support services.



“Oh my goodness, I can’t believe it, I’ve passed”

I exclaimed happily to my Mum, staring in disbelief at my phone. This was the culmination of 3 years of studying for my Humanistic Therapeutic Diploma in Counselling. I was filled with excitement and joy in finally qualifying to practice the career I had been preparing for.

After completing a master’s degree in social work in 2018, I had decided that I wanted to work closely with individuals enabling them to explore their own thoughts and feelings. This would help them develop insights into themselves; allowing them to use these insights to better understand and advocate for themselves.

I have always been passionate about working with people, especially enabling, and promoting individual’s independence and positive wellbeing. Currently I work for a Mental Health charity as a Crisis Recovery Mentor, working with individuals who are experiencing distress and despair. I also work for my local university in the Social Work Department in several roles, teaching students, assessing students before they go on social work placement and working with placement providers to ensure that placements are accessible for disabled students. Both jobs are extremely fulfilling for me and give me the opportunity to change the shape of people’s lives.

Over the past 3 years I have had many experiences which have led to growth in my self-development and confidence. These experiences have been both good and challenging, but as I like to say; all have provided me with new lessons and have contributed to the person I am today. I like to believe that if things go wrong (and they do because that’s life) these experiences provided me with the experience to grow and develop my resilience.

This article will focus on tips and advice for those looking at progressing their studies. It will discuss some of the challenges I faced, and how I overcame them. I strongly believe that you can do anything you put your mind to, even if you might have to do things in a different way, there is ALWAYS a work around.

Firstly, know your needs: having a think before you start your studies is extremely important as then you can explain these to the educational institution or course tutor. This will ensure that when you come to study, your course can be as accessible as possible. For me, this meant thinking about how course materials could be adapted, which included having all resources electronically before each session. I also had a support worker who would read out class materials from the board and explain other visual elements on the course, as well as providing sighted guiding around the college. Being able to discuss your needs with your support worker is also extremely important, as then they will be able to deliver this in class. I had a really good relationship with my support worker, but creating the best support for me took time.

At the beginning of my course, I had 2 support workers, one for the visual aspects of the course, and one for any issues that may arise due to my hearing impairment. I felt that this was overwhelming and made me feel doubly disabled, plus I felt that this isolated me from my peers, having 2 people sat either side of me meant that there was a barrier between my classmates and myself. It took multiple conversations with the college to persuade them that I only needed one support worker at a time, but once this was implemented it worked great. I formed a close relationship with my support worker and I felt that as a result of this my course was much more accessible.

Although I am aware that we do not always need to raise awareness about Alström Syndrome to everyone, explaining my condition to classmates was useful. It allowed them to understand what impact the condition had on my life, but also ways in which they could interact with me. This made me feel like a fully participating member of the class. On previous courses, I did not have the confidence to fully discuss my condition which meant that sometimes fellow students were unsure of how best to include me and as a result I felt isolated.

The students on my course also stated that because I was open about my sight and hearing this allowed them to empathise and learn how they could work in a more accessible way for their clients in the future. Be as open as you can, explain how you best communicate with people. Do you have a better side for hearing? Or does it help if people state their name when talking to you? Understanding this allowed me to explain my needs and was really beneficial. If you don’t feel confident enough explaining this yourself, you could ask a tutor or support worker to do this in your class.

Getting to grips with any technology and systems that your course uses is also extremely beneficial. When I started level 3 of my counselling studies in the middle of lockdown, it’s safe to say that I had NO idea how to use Microsoft Teams. This made it difficult for me to submit assignments and interact with my class. Over time I had some training from the college’s IT department on how to use the platform, but it may also be useful receiving training with an organisation that understands screen reading software. I used Voice Over on my iPhone for the duration of my course, but it’s important to remember that there are different commands depending on which kind of software you are using. It’s also important to remember that different buttons are in different places depending on what kind of device you are using. Getting to grips with this before your course starts is crucial, I didn’t, and it added on so much extra time and complications to my life that I didn’t need.

If there is a placement element to your course, you can also liaise with your college or university to ensure that reasonable adjustments are made to your placement, so it is as accessible as possible. This may include having a support worker on placement, all resources being in a format you can access, having an office layout that you can safely navigate and many more. I was lucky enough to be able to complete my 100-hour placement with the organisation I work for. This was beneficial as they already understood my needs. They allowed me to complete my placement remotely over the phone, so there was no need to navigate the office or get to grips with any new systems or technology. They also knew my worth and were excited to be able to enhance my skills.

Having a placement which champions equality and diversity is also extremely beneficial and will improve your wellbeing whilst on placement. If everyone is working together to ensure that you thrive, you will feel confident in fulfilling your duties. Being in a place that championed my autonomy and independence allowed me to focus on the work that I was doing. Working in a place that believed I was capable really increased my confidence and self-worth. If this proves a problem for you, you should definitely contact the placement team at the institution you’re studying with, as they can liaise with placement and work to change this situation.

Finally, it's important to choose a course that you're passionate about and interested in. This will help to remove any barriers or challenges you may face as you will be enjoying what you are learning.

I loved every minute of my time studying both for my master's and my diploma. This made me determined to overcome any issues that I faced along the way on the course. As I said at the beginning, you CAN overcome any obstacle you may encounter. It takes knowing what you need and being able to communicate it which will allow course providers to ensure that your course is accessible.

Under the Equality Act (2010) institutions and course providers must make reasonable adjustments so that students with disabilities can access both course material and other elements of their courses.

Sometimes it may feel like a solution is impossible, but please believe me there is a solution to every problem. You just have to think outside the box (and sometimes you have to put that box in recycling and make your own!). Nothing should ever stop you from doing anything you want to do, be that in life, your career, studying or having fun.

So, in conclusion, all I want to say is go out and do whatever it is you want to do.



Believe in yourself and you WILL be amazing!!!

Help us develop your wellbeing service

Melissa is now working with the team at ASUK to run a pilot project looking at how best we can support people with wellbeing. We have developed a questionnaire to discover your understanding of wellbeing, and what, if any, wellbeing service you would like to receive from ASUK.

We also hope to do some interviews with you to discuss this in more detail and get your views.

You will be receiving an email shortly with the questionnaire which should only take around 10 minutes to complete. You can always get in touch if you would like a hand with any of the questions or if you would like to take part in an interview.



Ticket Closure – Consultation extended to 1st September 2023

Rail operating companies have announced plans to close 974 ticket offices across England. A public consultation has been launched and closes on Friday 1st September 2023. Take action now to stop these plans.

Without specific measures to protect independent travel, these closures will add another barrier to an already inaccessible rail network.

- Existing alternatives, such as ticket machines and many online booking apps are inaccessible to blind and partially sighted people.
- Ticket offices provide a readily accessible and easy point of assistance for blind people. Until an improved alternative is in widespread use, then they must remain.
- Poor station design, failing audio announcements, outdated wayfinding, inaccessible technology, inconsistent tactile navigation and Victorian platforms, already make navigating rail travel a challenge for blind people. Ticket office closures will simply add an extra barrier.

The Pocklington Trust have provided further information about how you can have your voice heard and also useful templates to use:

<https://www.pocklington-trust.org.uk/news/countdown-to-closures/>



Judo Dreams! By John-James Entwistle

ASUK were delighted to award an adult empowerment grant to J-J so that he could attend 'The Lakes Summer School, Judo Course' in Scotland!

J-J tells us more below:

Some of the judo terms that may be helpful as you read the article. Uke (Player receiving opponent's attack) maintains the correct posture, and allows Tori (Player executing technique) to easily execute the Waza without resisting.

The first session of the day started at 10 in the morning and was taken by former GB Olympic Medallist 7th Dan Neil Eckersley. It began with balance exercise where you had to balance on one leg and one where you step though you're entering for a foot sweep called De Ashi Bari. We also did an exercise around control with the hands and how to move your opponent. We then worked on a turnover where you grab the waist and place the arm to grab the sleeve therefore trapping the arm and then roll Uke over the top of you.

Then we broke for lunch and in the afternoon, we focused on Tachi-waza or standing techniques. We started with a technique called Come De Ashi Bari. With normal De Ashi Bari you take 3 steps to the side and end up with your leg slightly ahead of Uke's foot you are attacking. The difference with the technique we looked at you attack both legs rather than one.

Then we worked on what techniques to do if your partner is able to step off which involves resisting the technique. The first technique we used was going round into Tai-otoshi which is known as body drop in English. Where you step across Uke's legs and pull them across with the arm but have the elbow at the chest level. We then worked on how best to break balance when entering for Osoto Gari. This is major outer reap. Where you step forward with the left foot if you're right-handed. You take a high collar lapel grip to have control of the head. You then pull down onto the leg that you are attacking and place your other leg slightly above the mat and attack with a sweeping action. But there are many ways to break your partner's balance. What I was shown was you grab the sleeve but close to the wrist. We then worked on groundwork where you twist around the body and end up in an armlock called Juji Gatame. Where you are in a position similar to the hold down Kesa-Gatame which means scarf hold.

Unfortunately, I can't describe the armlock in more detail as I'm not sure how to describe it. We then finished for the day.

On the Saturday evening there was a social event evening. Where there's a quiz and a subsidised bar and food.

On the Sunday we started on groundwork again. We started with Mune Gatame which means chest to chest hold down. This is a technique where you are chest to chest with your partner and lie as flat to the mat as possible with your legs almost outstretched and you place your hands over the head and trap the arm furthest away from you. We started with if Uke tries to roll away from you. The way to counter this is by placing your head on the mat. We then worked on what to do if Uke rolls towards you. This is where you place your legs though you were standing and walk them around Uke's body. We then recapped what we'd gone over the last 2 days.

To thank one of the coaches who organise the course in the Lakes, Mum bought Sylvia flowers on my behalf to show my appreciation for all that her and her husband John who both organize the course for all they do for me. Mum also bought one of their players who'd worked with me a bottle of wine as a thank you present.

Also most of the coaches who worked with me did so because they wanted to get a better understanding how to coach a blind

player like myself. Then on Monday it was time to visit Clyde a Judo club I know in Paisley. Who are very useful if you wish to be noticed by Scotland. Since selection for Scotland is one of my long-term ambitions. The reason they are good to know for Edinburgh is because their head coach is part of the Scotland elite setup and has many of her own players selected for Scotland. They are based in an industrial unit with their mats on a wooden platform. Sussie their head coach paired me with one of her female players who was a Brown belt called Harriet.

First, we did balance and movement work where we are stepping in for De Ashi Bari. Then stepping in for Ouchi Gari. Major inner reap. Where you step into the inner leg while making a circle. I then worked with Paul the other coach who Sussie runs the Monday Senior session with went through a technique called Sassi. This is where you enter with your left foot at an angle and pull up and out with your left hand and then you pull round with the lapel and end up propping the ankle. We then worked on techniques to use if your partner steps off. Initially we worked with Osoto Gari. But I was advised to get more power out of the technique I had to place my forearm on the top of Uke's chest.

We then worked on a variation where you hook the leg but drive backwards. We then worked on turnovers where you grab the furthest sleeve from you and the nearest trouser leg. You then raise yourself and end up flipping Uke onto their back. I was then shown an armlock where you place your knee in the armpit and have the arm straddle your leg and place your wrist on top of Uke's wrist and press. It's one I found very affective. We then finished off with both groundwork and standing Randori. In Judo this is similar to sparring in other combat sports. When we did groundwork Randori I was first paired with Harriet and found her a difficult opponent. Partly because she was able to stop me applying a strangle where you take a deep cross grip in the collar. Also, I was unable to turn her on to her back. I said at the end that she was a good groundwork fighter. I also thanked Susie for the interest she takes in me.

On Tuesday the senior session was taken by Kirsty a coach I know who pointed me in Clyde's direction. I worked with Harriet again. We did Sassi again but into either Tia Otoshi or Harai Goshi which are a major hip technique with a leg sweep or Uchi Mata. This is a similar technique to Tia Otoshi. I chose Tia Otoshi since Harriet was about 4 to 5 inches shorter than me. But I kept having problems around gripping the sleeve to break Harriet balance. Kirsty got me to feel what she wanted me to do. I felt this went well considering she'd never taught a blind player like me before. We then had a standing randori session where half the mat worked on Sassi and how to use it on the move. We then had general standing Randori. I started by being paired with Harriet and found her difficult. I couldn't break her balance. I also had the same experience on the ground. I said that she was a difficult opponent to turn onto her back and she said I'd done well all the same.

Overall, I was worked very hard and at the end I thanked Kirsty for introducing me to Clyde something I really appreciate.



T-KASH (Transition— Knowledge And Skills in Healthcare)

T-KASH (Transition - Knowledge and Skills in Healthcare) are FREE resources which were originally created in 2015 by young people from the Alström Syndrome UK (ASUK), Hear My Voice Youth Forum. The resources are designed to support young people, families, and healthcare professionals, to help everyone understand the transition journey and the things that are important to young people.

At the end of 2022, we began the process of updating the T-KASH resources to ensure they are inclusive and represent the needs of young people within the rare disease community. With the support of the Breaking Down Barriers (BDB) Experts by Experience Advisory Group and our youth and young adult advisors, we have updated T-KASH and created brand new resources, including an animation that we hope will be used throughout the BDB network and beyond.



The resources aim to:

Draw attention to the knowledge and skills young people may be able to develop to manage their healthcare, while having the best life possible

Prepare young people/families for identifying and coping with change at key points in their lives

Supporting young people to plan for their future and cope with their eventual transfer/settling into adult services

The T-KASH resources are based around 12 logos that identify key areas of a young person's life. These logos can be used in any health/speciality or education setting e.g. Hospital, GP, or school to signify different aspects of transitional care. Transition in health covers all aspects of a young person's life and supports them as they mature and enter adulthood.

Launch time!

The new logos and creative animation will be launched from the 11th September 2023.

You can join us virtually to hear more about how to use them on the 13th September 2023:

BDB T-KASH Launch - Let's Get it Right for Young People
(Afternoon Session)

1:00 – 2:00 pm

Register for the afternoon session:

<https://us02web.zoom.us/meeting/register/tZ0ocOirqzluGtcS1PlscorKRjGRhjeux6hF>

BDB T-KASH Launch - Let's Get it Right for Young People
(Evening Session)

7:00 – 8:00 pm

Register for the evening session:

https://us02web.zoom.us/meeting/register/tZYvcOCqrDkuHNNuXbBzXSqC_B_zj5OJYwJu



<https://breaking-down-barriers.org.uk/t-kash-transition-tools/>

AS Global updates

Alström Global Community Wellness Club

Usually on the last Thursday of the month the AS Global community have been coming together virtually to discuss a range of useful topics.

Don't worry if you have missed any of the sessions, you can view the recording and top tips from the session on the ASUK website

<http://www.alstrom.org.uk/as-global/>

You can also read our latest Impact Report on the website too.

AS Global Patient Registry

Be part of history—let's find all those pieces!

The ASUK Patient Registry enables you to provide your health information in an easy and straightforward format by completing questionnaires about your health in a safe and secure way.

This information will be available (anonymised) for researchers and clinicians worldwide to learn more about Alström Syndrome and develop treatments into this rare condition. It is free to join for both you and professionals. This information will help build a worldwide picture of Alström Syndrome, improving knowledge and understanding of this rare and complex condition.

You have the potential to make a real difference to everyone living with Alström.



How to join?

Check out the CoRDS Connect website and complete the CoRDS activation form, it is quick, secure and easy to do, just go to the website below:

<https://cordsconnect.sanfordresearch.org/BayaPES/sf/screeningForm?id=SFSFL>

Don't delay, enrol TODAY!

A big thank you to those who have already completed the registry. If you need any support to join the registry or if you would like any further information, please get in touch with ASUK Office Manager, Catherine Lewis on 07970 071675 or email catherine.lewis@alstrom.org.uk

ASI Conference will be taking place in the USA from the 5th – 9th October 2023. This will be a great opportunity to meet up with families and professionals and learn from one another.

On the 26th July AS Global hosted a virtual Q&A session where people could virtually meet up to have their conference questions answered.

Don't worry if you missed the session, you can find the video on the AS Global website, alongside further information <https://www.alstrom.org.uk/as-global/#International-Conference-2023>

If you fancy going... you can get a discounted rate at the hotel if you book by the **4th September 2023!**

Approval granted for Pre-implantation Genetic Testing (PGT) for families affected by Alström Syndrome

The Human Fertilisation and Embryology Authority (HFEA) have approved Pre-implantation Genetic Testing (PGT) for families affected by Alström Syndrome. This is where genes or chromosomes can be checked in embryos for Alström Syndrome. This involves IVF and embryos that do not have Alström Syndrome can be placed back in the womb. There is a strict criteria for this procedure and further information can be found on the following link <https://www.hfea.gov.uk/treatments/embryo-testing-and-treatments-for-disease/pre-implantation-genetic-testing-for-monogenic-disorders-pgt-m-and-pre-implantation-genetic-testing-for-chromosomal-structural-rearrangements-pgt-sr/>



If you or a member of your family has a diagnosis of Alström Syndrome and you are concerned about the chance of passing the gene on during pregnancy, you can speak to your healthcare provider about the possibility of PGT.

ASUK is planning a webinar to discuss genetics in more detail later this year. This will include a detailed explanation of PGT and other genetic testing that is available. We will share the dates with you shortly.

Exciting developments in Rare Disease Research!

LifeArc

LifeArc, a self-funded, non-profit medical research organisation and charity is launching a new programme called The Rare Disease Translational Challenge. They plan to invest more than £100 million by 2030 to help improve the lives of people living with rare conditions.

Firstly, they plan to invest £40 million to create up to five Translational Rare Disease Centres across the UK. These centres will bring together experts to collaborate and share knowledge and expertise to improve diagnosis and develop new treatments.

Through our work at ASUK and within Breaking Down Barriers, we are pleased to be involved in a number of applications that have been submitted and we will keep you informed as the programme develops. Further information about LifeArc can be found by going to their website <https://www.lifearc.org/strategy/rare-disease-translational-challenge/>

UK Rare Disease Research Platform

The Medical Research Council (MRC) and The National Institute for Health and Care Research (NIHR) have set up a UK Rare Disease Research Platform that will invest £14 million over five years to improve diagnosis and treatment for people living with rare conditions.

11 specialist nodes will be based at universities throughout the UK with a central platform hub that will pull together expertise and deliver wider activities.

The team at ASUK and Breaking Down Barriers are pleased to be involved in the specialist node being led by the University of Birmingham – Changing clinical practice in rare diseases through innovative trial designs (CAPTIVATE). Further information about the UK Rare Disease Research Platform can be found on the following website <https://www.ukri.org/news/new-14-million-investment-targets-rare-diseases/>

Welcome Sandra

"I recently joined the dynamic Inherited Metabolic Disorders team in May 2023 and I am enjoying gaining experience as the new Clinical Nurse Specialist. I am currently sharing the role with Sarah and Helen, who have been working with you in Alström clinics for a while.

I am learning a lot about Inherited Metabolic Disorders in order to deliver the best care and gain a better understanding into rare conditions like Alström Syndrome. Most of you will already know me from the Centre for Rare Diseases, where I've worked since 2019. Gaining experience in performing fibro scans, spirometry, ECG's and doing bloods for all patients that come to the department.

I have worked in various settings and in different hospitals that has prepared me for my new role.

I have over 20 years of experience in nursing and completed my nursing training in Dundee in Scotland in 2003, before moving to Bath in Bristol and now fully settled in Birmingham.

I always strive to provide excellent care and support to patients and enjoy meeting and helping people from all walks of life. I learn a lot from listening to people telling me their unique individual stories, and this makes the job really interesting for me.

I am looking forward to meeting you all in the future and being a part of your journey of care."

Mary, pictured above with Sandra who many of you know from the AS adult clinics, will still be supporting those attending as a volunteer.

How to get in touch with the clinical experts by Kelly Watkins, Service Coordinator / Office Manager - Inherited Metabolic Disorders/Lipid

"It has been a challenging couple of years with covid and the recovery in terms of clinic settings but I think we are finally back to where we were prior to covid with face to face appointments and regular follow ups. Since covid we've been able to include additional contact and follow up with Dr Ali regarding Semaglutide. This has proven valuable with getting additional information and for you to share any concerns with Dr Ali, who relays back to the team.

We are now looking to improve direct communication with yourselves and the IMD team here at the Queen Elizabeth Hospital. One way of improving the service is being able to directly communicate with you or your parents/carers or other family members in regards to medical issues or passing on information you feel the team need to be informed of. We do have this set up with many of you but there is always room for improvement. I have added our contact details for you to contact us directly either via email or telephone should you need to and also our out of hours emergency contact, should this be needed."



Telephone Numbers

0121-371-6987 – Lindsey Fletcher-Jennings, Team Medical Secretary and P.A to Professor Hiwot and Dr Dawson

0121-371-6984 – Kelly Watkins, IMD Service Coordinator and Office Manager

0121-371-6985 – Sarah Steeds, Helen Gallagher and Sandra Munyoro – Clinical Nurse Specialists

Emergency out of hours contact

0121-627-2000 – this number can be used out of hours from 4pm until 8am weekdays, or at any point over the weekend.

Please note, this number will take you to our switchboard, please ask for the consultant on call for IMD and you will be put through.

Email addresses

alstrommonitoring@uhb.nhs.uk – This email address can be used to contact myself or Lindsey for anything that needs to be shared with the consultants as well as your monthly monitoring results and appointment queries.

imdurses@uhb.nhs.uk – this email address is monitored by all 3 of the IMD nurses and can be used for any general enquires, support letters and advice.

I hope you all have a lovely summer and look forward to seeing the next group in September.

Research Update from Dr Sadaf Ali, Clinical Fellow - Metabolic Medicine

At University Hospital Birmingham, we care for patients with Alström Syndrome, which is a rare metabolic syndrome. We have carried out groundbreaking research in the past to help us get a good understanding of the condition. We are actively recruiting for an ongoing study to help us learn even more about the condition. We hope that this will help us to find new treatments and improve quality of life. We hope to recruit until the end of 2023 and will have exciting information to share with you by the end of 2024.

Some adult patients have been taking a medicine called Semaglutide. This is a type of medicine called a GLP-1 analogue and it is used for the treatment of type 2 diabetes. We have been looking at how effective Semaglutide is for people with Alström Syndrome, and we are pleased to report that we have seen some benefits such as significant weight loss and improvements in diabetes control.

The team will talk to you about research when you come to clinic, but you can also contact us if you would like to find out more.

Unfortunately, as you may have heard in the news, there has been a worldwide shortage of Semaglutide since 2022. We know that some of you may be having a few problems getting your repeat prescriptions. Due to the weight loss effect, it has become a very popular and on demand medicine and the problem has been made worse by the media advertising the weight loss effect. Should you find yourself in the position where you cannot get the medication, please contact the IMD team either via email or telephone and we will get a clinician to contact you to discuss alternative medication until the supply issue is resolved.

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

Meet the ASUK Team



Jas



Sharon



Kerry



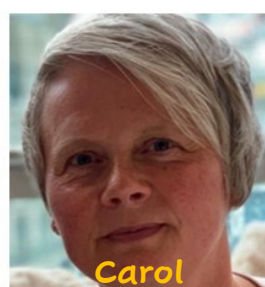
Catherine



Kez



Trevor



Carol



Lexi



Melissa



Haris



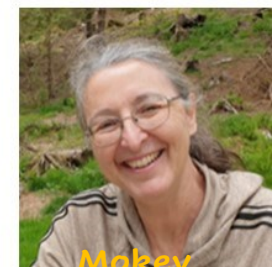
Vanessa



Clair



Ann



Mokey



Jane



Tim



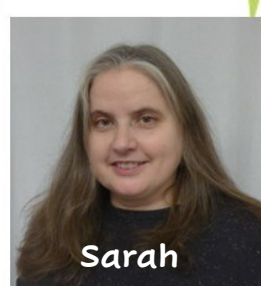
Steve



Marie



Katie



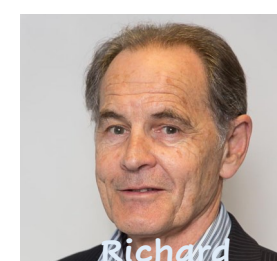
Sarah



Derek



Cathy



Richard

Keep in touch

Remember we are only an email or phone call away

Registered Office address: 4 St Kitts Close, Torquay, Devon TQ2 7GD

For all enquiries please get in touch on 07970 071675 / 01709 210151

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 twitter [@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>

