



# Alström Syndrome UK

Strength for today, hope for the future

## The Newsletter of Alström Syndrome UK

### Issue 21 – Summer 2018

Alström Syndrome UK (ASUK) is a charity who provide information, support and advice for individuals affected, their families and carers and professionals. Alström Syndrome 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. We endeavour 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](http://www.alstrom.org.uk)

## Welcome to the 21st edition of the ASUK Newsletter



It's lovely to introduce to you the 21st edition of the ASUK Newsletter. Putting together articles and snippets for this edition of the Newsletter has given us all a chance to reflect on the enormous strides that have been made over the last 20 years to understand, improve and share our knowledge of Alström Syndrome. Families have connected with each other and shared some ups and downs. What ties us together is the eternal hope of improving treatments and minimising the impact that Alström syndrome has on people's lives.

As ever we are grateful for the support and generosity from all of those that continue to give their time and donations, so that we can give grants to families and individuals. We are delighted to have received a further grant from Children in Need and excited to have launched the 'Big Activity Challenge' for adults with Alström. We're looking forward to your applications on some wild and wonderful activities!

As we head into cooler days and nights here's an update on what families have been doing over the summer and the work of the dedicated ASUK team.

### Family Gathering – Identifying the Diagnostic Journey

We are just finalising the November date for this year's family gathering where the focus will be on the 'Diagnostic Journey'. We want your help to address questions such as; what led to your diagnosis, how and who recognised AS, what professionals were involved and at what age did you or a family member receive a diagnosis? We also want to hear your ideas and views on what we can do to improve the rate of diagnosis, to try and make it easier for families and individuals in the future.

Meanwhile, I hope you enjoy reading about the work and family fun!

I look forward to seeing you in November.

**Ann Chivers** -Chief Executive at ASUK

## **Raising awareness and working with our partners**

Collaborating with families and professionals internationally helps us to expand our knowledge and understanding, so we have been discussing joint projects, attending conferences and listening to patient's stories about their journey and their challenges

### **Bardet Biedl Conference 20-22nd April**

Kerry attended the annual Bardet Biedl conference which was a fantastic weekend full of learning, networking and socialising opportunities. We're pleased to work with Angela Scudder from Bardet Biedl UK and Tracey Lynch from Wolfram Syndrome UK.

### **Cardiomyopathy UK information day Sheffield, 7th April 2018**

As you may be aware, cardiomyopathy is one of the conditions that some people with Alström Syndrome are diagnosed with. Therefore, Kerry went along to the Cardiomyopathy Information Day to learn more about dilated and restrictive cardiomyopathy and to learn about new developments in research, medication and support. The charity also provides lots of useful information documents, including fact sheets on the different types of cardiomyopathy, medication, travel and life insurance, alert cards and many other issues. For more information and guidance on living with cardiomyopathy, you can download or order their fact sheets and publications from their website <https://www.cardiomyopathy.org/cardiomyopathy-information/cardiomyopathy-information>.

You will also find details of other information days throughout the country that individuals and families are welcome to attend.

### **Genetic Disorders Leadership Symposium UK – 10 – 11th March 2018**

This was a fantastic opportunity to join with patient organisations and others working within the field of genetic conditions to share examples of good practice, learn about new developments and initiatives and to discuss the current challenges faced by people living and working with genetic conditions. Here is a snap of Kerry with Angela Scudder from Bardet Biedl UK and Tracey Lynch from Wolfram Syndrome UK.



### **Council for International Organisations of Medical Sciences (CIOMS) – A working group**

ASUK have been asked to join the CIOMS Working Group IX to work alongside patient organisations, industry, regulators and academic researchers to develop guidance and recommendations on patient involvement in the development and safe use of medicines. The document will highlight global knowledge and initiatives and will address ongoing challenges. This work is predicted to take between 2 – 4 years to complete.

### **NIHR Think Research Rare Diseases Patient Day - 21st March 2018**

Kerry also went along to the National Institute for Health and Research (NIHR) Think Research Rare Diseases Patient Day in March. This event was aimed at rare disease patients, carers and people who work for a rare disease group. The day included presentations and workshops on topics such as using social media, working

with industry, genomics, data and research, it was a useful opportunity to meet others from the rare disease community.

## QAC Sight Village Birmingham July 2018

The ASUK team attended the QAC Sight Village event in Birmingham in July.

A fantastic event for people who are visually impaired, for professionals and for organisations wishing to improve their services for the visually impaired.

It was also a great opportunity to meet new people and catch-up with friends we haven't seen for a while! More information on the QAC upcoming events can be found on their website: <http://www.qac.ac.uk/exhibitions.htm>



### Thank you Chris Lyon-spreading the word about Alström

A special thanks to Chris Lyon who many of you will know.

Chris gave an excellent presentation about AS at Sight Village, talking about his lifestyle and his experience of specialist care assessments and how specialist assessments can help in daily living. Chris was filmed being interviewed about AS by another organisation – click on the following link to view the fabulous short film: <https://vimeo.com/283655026>

Chris is pictured left with Mum Carrol, one of our Family Support workers.



## A huge THANK YOU to **Children in Need!**

In 2015, we were fortunate to receive 3 years funding from Children in Need to support young people through the sometimes-difficult transition from children's clinics at Birmingham Children's Hospital to the adult clinics at the Queen Elizabeth Hospital. We know that for many young people and their families this can be a challenging time. Working with young people and their families we wanted to help to ensure that young people who are affected with AS are less isolated, healthier and able to manage what can be a big change. We worked with young people to develop resources called TKASH (<http://www.alstrom.org.uk/growing-up/#1489163131413-c6cb09c8-c0eb7c38-ea07>) aimed at helping young people navigate this transition stage. We consulted with young people and their families and organised a whole range of events, meet-ups and fun stuff for young people to try out new activities such as: a driving experience and weekend's away. The most recent being a weekend at the Pioneer Centre in Kidderminster. Here families had the chance to socialise, try something new, relax and have fun! Families tried abseiling, zip wiring, archery, bouncy castles, arts and crafts and high ropes. One of our young people has written a very special account of her experience at the Pioneer Centre:

## How I found the ASUK activity weekend by Hannah Bromley Challenor

- “I have to say when I first got told I had a mixture of thoughts but the lowdown of it was I was excited and happy to meet other people with Alström that I hadn’t seen since the last clinic the previous year. I’m not a confident rock climber but I think that the weekend had something for everyone of all ages meaning both young people and parents enjoyed it.



As I have put above we went rock climbing with a few people getting to the top. Some people did the high ropes which are like a high up rope bridge with bin like tunnels and spider like webs of ropes to get through to the end and back. With the people who did it they were scared at first but by the end they were all trying to do cool tricks with the ropes they had got used to it. We also went on a very muddy walk avoiding golfers that seemed pointless for all the young people who have Alström as I made the point of saying our parents were literally dragging us through the mud to get a nice view, but we enjoyed the nature, mud and collecting of pine cones. We also went on a bouncy castle assault course that ended up being massive fun and creative trying to explore through it and how to muck about. If I had to describe it, the main one was going through a blue or red whole which you had to pull your body through like a worm then go up and under these rungs and then go up a hill that is almost impossible to climb unless you have support on the other side because I admittedly started it and fell off with giggle! You then had to go down a slide and then find these large pillars that are like the brush bit of a toothbrush that you can start walking through but I almost got pushed onto the outside wall and got easily stuck so I find it is best to slide through like a snake through the undergrowth just to get the stomach muscles pumping.

The reason I was honoured to write this was to give my top tips on one of the more intimidating activities – zip wiring. It’s one of those things you love to hate. I personally don’t like it sometimes it’s because with the small amount of sight I have there is sometimes a contrast between the strip of wood you’re stuck as a statue on and the whiteish gravel covered the ground very far below. Luckily at the weekend, thanks to my colour blindness, it was practically camouflaged. What is nice is with the experience they had people overseeing the activity so your safety is in good hands. They’ll tell you what ropes you can grip onto with your life and what they want you to do at the end. I find once they do this with you the only and best thing you can do is to lean into your harness and with your bent legs, walk to the edge until your feet are about half way off, and when you can feel yourself going with it then go with it. It’s all you can do. It makes you feel like you’ve left your stomach up there even more with the strange jerk near to the end but at the end of the day you do enjoy the thrill of it deep down. This way definitely works because it was the first time this weekend I did it

twice. I had a great time with everyone and I looked forward to the next opportunity to seeing other people with Alström”.



For many young people and their family’s weekends like the one we organised at the Pioneer Centre make a real difference to people’s lives. One Mum said ‘I didn’t believe it was possible to watch my son whizzing down a zip wire on his own-I now know he can do so much more than I thought!’

The project has helped young people to develop their own confidence and speak up for themselves.

### The future

Building on the success of this project we are delighted to have received a further 3 years of funding from Children in Need to continue working with young people and families through the transition stage. It

also means we can continue activities on Sunday evenings prior to children’s clinics where we will be exploring fun ways to get fit and active with the help of John Pemberton, the Dietitian at Birmingham Children’s Hospital. We are also thrilled to announce that Marie McGee will continue her role as National Transition Coordinator.

### Partner Projects

The Breaking down Barriers project which has kindly been funded by the Sylvia Adams Charitable Trust, continues to gather momentum. <http://breaking-down-barriers.org.uk/>

We are pleased to welcome new members to the project this year and we now have a total of 15 member organisations who are all carrying out individual projects under the Breaking Down Barriers banner to further develop the services they offer to their members.

The next Breaking Down Barriers workshop will take place in Birmingham on the 19th and 20th September 2018. We will be joined by representatives from the member organisations, the Trustees of the Sylvia Adams Charitable Trust, clinicians, community development workers and academic researchers who all have an interest in developing supportive and inclusive services for people from ethnic minority communities.



## Welcome Asya!

Please join us in welcoming Asya Choudry to the ASUK team. Asya is the new Community Engagement Manager for the Breaking Down Barriers project managed by ASUK. Asya's previous experience in genetics, her work within the voluntary sector and her understanding of some of the barriers experienced by people within South Asian communities will be a great asset to the project and to ASUK. You can find out more about the project here:

<http://www.alstrom.org.uk/breaking-down-barriers>



## Farewell

For those of you that attend the adult Alström clinic you will be familiar with Shanat Baig, a Clinical Research Fellow who specialises in cardiology and inherited metabolic disorders. Shanat continues his training at another hospital and therefore will be stepping down from his role at the QE hospital for 12 months.

ASUK would like to say a huge thank you to Shanat for all the hard work, support and dedication he has provided for our adult patients over the years. We would like to wish him all the very best over the next 12 months, we will miss him!

## The Big Activity Challenge

**The Adventure starts here!**



### What is the Big Activity Challenge?

Are you an adult with Alström Syndrome? Do you want to try a new activity.? You can apply for between £50-£500 to pay for an activity, all we ask you to do is provide some photographs and write an article for publication in the ASUK newsletter, Or, if you would rather tell us about

your experience, we can help you produce a podcast for the ASUK website. Who knows, you may inspire others to try out new activities too!

### How you can apply!

You can find the application form on our website <http://www.alstrom.org.uk/asuk-big-activity-challenge/> and send it to Liz at [liz.loughery@alstrom.org.uk](mailto:liz.loughery@alstrom.org.uk) or if you would like any further information or support completing the form you can contact her on 07517 278 946.

## **Alström Syndrome multi-disciplinary clinics 2018**

### **Adult clinics at the Queen Elizabeth Hospital, Birmingham**

26<sup>th</sup> and 27<sup>th</sup> September 2018

7<sup>th</sup> and 8<sup>th</sup> November 2018

### **Children's clinics at Birmingham Children's Hospital**

17<sup>th</sup> September 2018

12<sup>th</sup> November 2018

### **Outreach Clinic in Leeds**

November 2018 – Exact date tbc

We have created a new document to explain more about the clinics and the tests involved, for access simply click on the icon below.



AS Clinic Document -  
Final.docx

ASUK will be in touch with individual families prior to each clinic they are due to attend. Should dates need to be altered for any reason then everyone shall be informed as soon as possible. Please get in touch if you would like further information.

## **ASUK Family Support Service**

All those diagnosed with Alström Syndrome are allocated a Family Support Worker depending on where you live in the UK. Our Family Support service supports children and adults, their families and carers.

We do this by:

- Supporting individuals and families at the AS specialist clinics. We can organise overnight accommodation, offer to accompany patients in appointments and help people understand medical terms
- Liaising with professionals to improve diagnosis and treatment, sharing evidence based approaches to improve daily living
- Supporting individuals and families to become 'experts by experience' in managing their condition and treatments
- Promoting healthy living. We run workshops for children and families, loan tandems, provide grants for fitness activities and equipment and signpost to specialists
- Connecting families together to share their experiences, support each other and socialise
- Organising short breaks, residential holiday weekends and activities to have fun
- Supporting individuals and families to improve access to financial support, benefits and specialist advice. We help families to complete forms, signpost to benefits, complete health, education and social care plans and help wade through the administration maze that can so often be a barrier to getting your rights

- We focus on working together, to enable and empower individuals to lead the life they choose
- We support people to become self-advocates
- We act as a catalyst and facilitator to further clinical research into this ultra-rare condition

### How we work

We work as a virtual team, supporting people throughout the UK. We make home visits, visit schools and colleges and liaise with health, educational and social care professionals.

If you or a family member has been diagnosed with Alström Syndrome or if you require the information in an alternative format then please get in touch.

We are here to support you.

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AND FINALLY, **A HUGE thank you** for all your donations over the last 6 months or so and to everyone who continues to support our great cause! Special thanks go to:

- David Lunt
- Keith Coombs Trust
- Wickersley Old Village cricket club
- Albert Hunt Trust
- The Sylvia & Colin Shepherd Trust
- Ulverscroft Foundation
- KET Charitable Trust
- The Lynn Foundation
- And to all those who have donated through Just Giving and would like to remain anonymous.

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Please visit our website [www.alstrom.org.uk](http://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](http://www.facebook.com/alstromsyndromeuk) please click 'like' to show your support and spread the word!

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<https://www.justgiving.com/alstromsyndromeuk>



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ASUK has been assessed as meeting the standards of The Information Standard and continues to be a certified member.

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