



Alström NEWS

With your help we have hope

THE NEWSLETTER OF ALSTRÖM SYNDROME UK

Summer 2015 Issue No.16

Alström Syndrome UK provide information, support and advice for individuals affected, their families, carers and professionals working with them. Alström Syndrome is a very rare genetic condition which can lead to progressive blindness and deafness and can also lead to heart and kidney failure, type 2 diabetes, liver dysfunction and associated problems. The symptoms arise at different stages making diagnosis 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
Registered Charity no: 1071196 Registered Company Limited by Guarantee 3557191

Exciting Research Times Ahead!

On the 11th May 2015, ASUK Chair Michelle Hough and ASUK National Development Manager Kerry Leeson-Beevers attended a meeting with Dr. Rob Semple at the Biomedical Campus in Cambridge. The aim of the meeting was to discuss our research projects and the best ways to move forward.

ASUK are forging ahead to gain further funding as there is such an emphasis on rare disease and research in this area. Alongside the teams at the Queen Elizabeth Hospital and Birmingham Children's Hospital, Dr. Semple at Cambridge University has been able to collect skin samples from patients to learn more about this very rare condition. This was made possible in 2010 through Big Lottery funding of £350,000 which has enabled further insight into this condition.

Professor Phil Beales and Professor Tim Barrett have also been developing research into Alström Syndrome which can now be developed further through a NIHR Translational Research Collaboration grant "Deep phenotyping in Bardet-Biedl and Alstrom Syndromes" which has

received £465,000 in funding.

A further research meeting has been scheduled to take place in Birmingham on the 15th July 2015. ASUK will be re-establishing the Medical and Scientific Advisory group which will be discussed during this meeting. This will bring together members and professionals alike to discuss research projects which are in the best interests of everyone involved. This will enable specialists to collectively look at research projects available to determine what would be in the best interests of patients to take forward and develop further.

The ASUK team are very excited to be working alongside the teams at Cambridge University, the Queen Elizabeth Hospital, Birmingham and the Birmingham Children's Hospital as well as the expertise from our new Trustee Dr. Richard Paisey. This team work will enable future developments and research projects to be achieved.

ASUK Research Facts

- Big Lottery funded £350,000 in 2010 for a 3 year research project to learn more about this rare condition, skin samples were collected and clinical patient database compiled.
- 23 sets of cells were originally collected and stored at Cambridge University.
- 5 were then batched and sent to the tissue bank at BCH as part of EUWABB project.
- Skin samples sent in 2015 to Prof. Phil Beales for further research.
- Prof. Beales and Prof. Barrett "Deep phenotyping in Bardet-Biedl and Alstrom Syndromes" research started.
- The Euro-WABB project is an initiative, to investigate Rare Diabetes namely Wolfram, Alström and Bardet Biedl. ASUK are actively involved in this work with Prof. Tim Barrett.
- Potential collaboration with Prometics to research anti-fibrosis treatments.

Farewells and Greetings

Alström Syndrome UK has some big news. After founding and piloting the charity for many years, Kay Parkinson is now moving on. For the past 24 months, Kay's work has taken her across Europe and further afield while the Board of Trustees and our contractors have continued to drive ASUK forward and have ensured that our families are supported throughout the UK. Our families will always be at the heart of everything we do, now and in the future and we would like to reassure you that there will not be a change to the level of support you receive.

ASUK will continue to offer a 24 hour help-line, covered by our very experienced team:

Kerry Leeson-Beevers: Tel: 01709 210151 / 07716135940 or email Kerry.leeson@alstrom.org.uk

Catherine Lewis: Tel: 01803 613117 or email Catherine.lewis@alstrom.org.uk

Iram Shah (fluent in Urdu and Punjabi): Tel: 07988 237664 or email: iram.shah@alstrom.org.uk
John Parkinson: Tel: 01803 409952 or email john.parkinson@alstrom.org.uk

On behalf of our Board of Trustees and the team at ASUK, we wish to give our warmest thanks to Kay for everything she has achieved for our families and we look forward to the continuing developments within the Alström community.

Curtis Vasey

who has been an ASUK Trustee has decided to step down from his role but he would still like to remain involved in the work of ASUK. Especially in developing transition services and activities for young people affected. Curtis has been a dedicated Trustee who has always had the needs of our families at the heart of everything he has done. ASUK would like to sincerely thank him for his commitment and support over the years.

At QEH, Psychologist Robin Paijmans who has been involved with our AS clinics is sadly leaving. We are working with QEH to ensure another Psychologist will be part of our service moving forward and we hope to have another Psychologist in place soon.

Martin Henwood

Martin Henwood has been appointed ASUK's interim Chief Executive Officer and has come on board to develop our governance, ensuring we are compliant in all areas. ASUK will soon be advertising for a part-time Chief Executive Officer to develop the charity further.

Dr. Richard Paisey

We are delighted to announce that Dr. Richard Paisey has joined the ASUK Board of Trustees. Richard has been part of the ASUK's journey since it began, helping develop the ASUK multi-disciplinary clinics at Torbay Hospital, before they relocated to the Queen Elizabeth Hospital in Birmingham. Richard has been involved in our research projects, including monitoring members who took part in the ASUK bike ride in 2012. His expertise and experience of this very rare condition will be a major boost for ASUK as we progress our future research work and ongoing developments.



Introducing, National Transition Coordinator

Marie McGee

We have pleasure in introducing a new member to our team, Marie McGee who will be our National Transition Coordinator. The post has been kindly funded by Children in Need as part of a three year transition project. Interviews took place on the 1st May 2015 in Birmingham and we would like to say thank you to our members, Curtis and Maariyah for their excellent contributions to the interview process.



The Children in Need Transition project will work with young people and their families affected by Alström Syndrome. By providing fun activities and one to one support with transition, ensuring that young people who are affected are less isolated, healthier and able to better manage change.

Marie explains about her previous work and achievements below:

My early career saw me work in a number of inner city Primary and Secondary schools where I supported children and young people (CYP) to develop connections with their emotions, skills and abilities using a variety of activities. These include nurture groups, anger management sessions, 1:1 mentoring, playground buddies, family skills and youth parliament. I believe in widening CYP opportunities in life so have arranged involvement in activities such as: the opening of Water Hall Art Gallery, Birmingham (with Cherie Blair) organising theatre productions around themes including gang/gun culture, and organising careers events with artists who delivered speakeasy activities (building confidence to speak up for yourself). The role also saw me lead on statement reviews, attending child protection meetings, child in need reviews and family representation at police interviews.

I moved on from there to work for Birmingham Children's Fund, which became part of Birmingham City Councils Children and Young Peoples Commissioning Team. Here I commissioned a variety of services with a budget of £150,000. My creative talents have allowed me to also contract a number of additional events including anti-bullying workshops in a variety of locations across the city. The events included dancers, street artists and bakers to give families and professionals a platform to discuss/respond to aspects of bullying in a safe and fun manner. I also worked with Birmingham City Councils Communications Team to produce city wide leaflets to inform families about child protection in relation to Victoria Climbié and the new government framework for assessing family needs. In addition I also worked with the Children's

Safeguarding Team to make the Common Assessment Framework more accessible, using actors and media to create a short film and poster resource for Birmingham's families and professionals.

In 2007 I eventually started my own mentoring consultancy known as Achieving Together. This is where I could develop a more creative range to my delivery which includes projects like Lunchtime Heroes, where I developed a range of characters to support behaviour issues in Primary schools. (See the website at: ambinetdemos.com/lunchtimeheroes/)

Balanced with my mentoring work I am also employed part time at Birmingham Children's Hospital as the Transition Co-ordinator for the Rheumatology and Rare Diabetes Teams. Within the role, I build the skills and knowledge of young people with long term chronic health conditions. This helps to build their resilience to manage their condition, which in the long term helps them to reach their best potential. These young people often feel they have become invisible or fear being different so using creative artists, rappers, dancers and singers to give them a different outlet, has a very positive impact on them.

In mid-2014 I took a sabbatical for four months in Tanzania. I became involved in a country wide project to produce resources to raise the awareness of the early signs of childhood cancer. I managed to get funding from a Dutch Construction Company, coupled with a reduced rate for graphic design and printing rates from a Birmingham based Media Company.

I am excited to be involved in this transition role within ASUK and I look forward to supporting both young people and the family as a whole to ensure their voice is heard and transition is a positive experience for them.

Please feel free to contact Marie McGee for further information:
marie.mcgee@alstrom.org.uk

ASUK

Raising Awareness at Prestigious Events

Kerry Leeson-Beevers recently attended a Leadership Symposium organised by Genetic Disorders UK in partnership with Global Genes. The purpose of the event was for charities to come together and share best practice. The impressive line up of speakers highlighted the excellent work and projects that are happening throughout the UK and how new projects such as the 100,000 Genomes Project are bringing new hope for a greater understanding of genetic conditions. Pictured below, Kerry with guest speaker, Actor Warwick Davis.



Kerry went on to attend the Oxford Rare Disease Conference 2015 from the 6th - 7th May 2015 at St. Anne's College in Oxford. This was a great opportunity to learn more about current and future medical advances, research projects and network with similar organisations. The objective for the conference was to support the development of new therapies for rare diseases by creating a forum where representatives from academia, industry, patient organisations, and funders could come together to network and exchange ideas. Professor Dame Kay Davies, Chair Oxford Rare Disease Initiative thanked Kerry for attending this prestigious event, "This could only be achieved if all attendees

contributed and I would like to thank you for helping to make this event a great success with your presence and your enthusiasm."

From the 6th - 9th June 2015, Kerry also attended the European Human Genetics Conference in Glasgow. The conference brought together all workers in human and medical genetics to review advances and develop research collaborations. ASUK were delighted to have an exhibition stand at the event and with over 3,000 delegates attending this was a great opportunity for researchers and professionals to learn more about this ultra rare condition.

Family Stories

You may remember from our winter 2014 newsletter, our article about our members becoming increasingly interested in Judo as a fantastic sport to be involved in. Since then one of our members Millie (pictured below) who is 7 years old has gone on to win a bronze and gold medal in a local competition, well done Millie!



Information & Activity Day 19 SEPTEMBER 2015 SAVE the DATE

ASUK will be holding a more informal conference this year on Saturday 19th September 2015. This information and activity event will take place at New College Worcester, where families and professionals will be able to hear about updates regarding the charity and pioneering research opportunities. The AGM will be held before lunch and there will be activities and workshops being held in the afternoon to keep everyone entertained and informed. There will also be an opportunity to tour the school and college facilities throughout the day. Please find booking form and further information enclosed. Further information can be found at:

www.alstrom.org.uk

or contact Catherine Lewis to reserve your place on:

01803 613117

email: catherine.lewis@alstrom.org.uk

We look forward to seeing you all there!

Patient Engagement Group

ASUK National Development Manager, Kerry Leeson-Beevers is now an active member of the Patient Empowerment Group, coordinated by Rare Disease UK. The purpose of the group is to monitor the implementation of the UK Strategy for Rare Diseases, ensuring the voices of patients and carers are heard. The group is now also being used as a sounding board for Public Health England during the development of the Congenital Anomaly and Rare Disease Registration Service. For more information please follow the link below or contact:

Kerry - 01709 210151 or
email Kerry.leeson@alstrom.org.uk
www.gov.uk/the-national-congenital-anomaly-and-rare-disease-registration-service-ncards

Otley Sailing group – 28th May 2015

On the 28th May 2015, families affected by Alström Syndrome had a fun-filled day out at Otley Sailing Centre. This was a great opportunity for families to try sailing in a relaxed atmosphere with the help and support of Otley Sailing Club who kindly provided volunteers for the day and supported by our National Development Manager, Kerry Leeson-Beevers. Otley Sailing Club promote dinghy sailing in the West and North Yorkshire regions providing sailing, power and safety boat training from absolute beginners to the highest level. They are committed to working with sailors of all abilities, have a Sailability Centre of Excellence award and are an RYA OnBoard Centre offering sailing experiences to young people in the local area. You can find out more about their fantastic work at:

www.otley-sailingclub.co.uk

We are currently organising a horse riding activity in the Midlands area and a Calvert Trust break in the South of England so please get in touch if you would like further information.



More photos - PTO

ASUK Family Sailing Trip May 2015



Guide Dog Experience Day

Family Support Manager, Iram Shah attended a Guide Dog Experience Day at their training centre in Leamington Spa on the 1st April 2015 with one of our members (family pictured below). There were lots of activities during the day including a touch tour of the centre and an Easter egg hunt.

More information about Guide Dogs and their work can be found via their website:

www.guidedogs.org.uk



Let's get Moving with the ASUK Bike Club!

The ASUK Bike Club is now stronger than ever, through Sport England funding we have been able to provide another 8 tandems and specially adapted trikes for families.

Cycling is a great way to exercise and exercising has many benefits - it can ease muscle aches and pains, reduce stiffness, and increase flexibility, balance, strength and mobility. It can also help you get fitter, have more energy, lose weight, lift your mood and most importantly, it can be fun! Alström syndrome makes it especially difficult for you to maintain a healthy weight so regular exercise is really important for you. Exercise programs work



best when tailor-made for an individual, taking into consideration specific needs, abilities, and difficulties. Tandem cycling is ideal as it brings people together of all abilities and enables people with dual sensory loss to cycle alongside their peers. The other big advantage of a tandem is togetherness. Riders of differing strength don't lose one another, you can keep up the chat –

including when the road is too narrow or busy to ride side by side – every mile of the ride and every hill is a joint experience and a joint achievement.

We currently have spare tandems and tag-alongs available if anyone is interested in getting involved.

Please get in touch if you would like to know more.

The Seeing Ear Poetry Competition

You may remember in our November e-newsletter we published an article about the Seeing Ear Poetry Competition. Many of you entered and your poems are going to be published in the anthology, well done! You can see all the winners at the link below:

www.seeingear.org/home/the-seeing-ear-poetry-competition/

The poems were written to commemorate the 100 year anniversary of the start of the 1st World War and are published below. Twins Katie and Hannah Beck (photo above.) are also having their poems about 'winter' published, which we will be published in our ASUK winter newsletter.



Who will?

*Who will...March up the hill?
Who will...Fight the men?
Who will...Win the war?
Will you my laddie?*

By Hannah Beck, aged 8 years old

Who will?

*Who will...March all day?
Who will...Dig trenches all night?
Who will...Wear wet dirty clothes?
Will you my laddie?*

By Katie Beck aged 8 years old

The Unidentified

*Rain poured harder that night as if it was
sad to see innocent brave men die.
They laid in lines of ten, men young and
old were at their fatal end.
Telegrams sent to every family with the
bodies of the brave.
Some were left. They were on their own*

*now, in the pouring rain in a foreign field.
All because no-one could see or even
remember a glimpse of who they once
were.*

*They lay in holes and bomb shell holes
alike, all in foreign mud and drowned in
their own blood.*

*They were far from their true home
where their loved ones and friends
remembered the soldiers.*

*But one was given the chance. To return
and show in the glory of the whole of
Britain that he and only he will represent
the lost men.*

*Ones who died in their duty to England,
who never came back.*

*He is always there. Now, in the Abbey
for his spirit is the one shoulder everyone
can cry on.*

**By Hannah Bromley-Challenor aged 14
years old
(pictured with Mum, Susan)**



THANK YOU to Our Fantastic Fundraisers!

A HUGE THANK YOU to everyone who continues to support our great cause! The numbers of people being diagnosed with Alström Syndrome is increasing so please get in touch if you would like help to arrange a fundraising event; we also have spaces available in the Silverstone Half Marathon and the Bupa 10K Marathon. These funds raised really do make a huge difference and ensure we can support families throughout the year. You can donate to Alström Syndrome UK securely through our Just Giving page www.justgiving.com/alstromsyndromeuk/ or send cheques made payable to Alström Syndrome UK to our registered office 31 Shearwater Drive, Torquay, Devon TQ2 7TL. Well done to our fantastic runners Brian Vasey and David Lunt who took part in the Virgin London Marathon on Sunday 26th April 2015 in aid of ASUK. Jointly they have raised over **£2,000**. It is still not too late to show your support through Just Giving.

www.justgiving.com/BrianVasey/
www.justgiving.com/David-LuntAlstromuklm2015/

Brian (pictured below) gives his



thanks to everyone who supported him "Hi you amazing people I completed the London Marathon in 4hrs & 33Mins. It was a brilliant experience and I'd like to thank

all of you for donating. Alström's Family Rock! xxxxx"

Thank you to Kayleigh Lewis who continues to make and sell candles and crafts in aid of ASUK, raising a wonderful **£125.00** well done Kayleigh!

Thank you to the Hough family and the St. Agnes Community in Cornwall for helping raise funds for ASUK during the Victorian Fair on the 25th May 2015. The cute and cuddly giant Bolsters were on sale at £15.00 each. Profits go to our great cause so please get in touch with Michelle Hough today to own your very own Bolster and make a huge difference to the lives of people affected by this devastating condition.

CONTACT:
michelle.hough@alstrom.org.uk

01872 553067



The Hough family

The Giant Bolster now has his very own Facebook page!



ASUK Chair, Michelle Hough and her Daughter Millie have been busy creating the cute and cuddly Giant Bolster!

You can now keep up to date with his exciting travels at:

facebook.com/GiantBolsterappeal

The Giant Bolster is a legendary character said to live near the cliffs of St. Agnes in Cornwall and is a popular character in village parades.



You can now own your very own limited edition 25cm Giant Bolster priced at **£15 each**.

Coach Seats for Great Causes



A HUGE THANK YOU to Williams Travel who have chosen ASUK as their charity to support through their Coach Seats for Great Causes Scheme. Their Summer 2015 holiday brochure is out now and for every holiday booked £1 will be donated to ASUK.

They have already raised awareness through a coffee morning and raffle which raised £52.50
<http://williams-travel.co.uk/>
All profits will support the ASUK appeal.

For more information or to place an order please contact:
Michelle Hough on:

01872 553067

Email:

michelle.hough@alstrom.org.uk

Cardiomyopathy Association is Changing

The Cardiomyopathy Association is changing its name to Cardiomyopathy UK and is having a new look.

The changes are being introduced over the next few months and people affected by cardiomyopathy will notice differences in all their communications, including their

website, cardiomyopathy booklets, their magazine and promotional items. You can find out more via their website :

[www.cardiomyopathy.org/
Cardiomyopathy-change.html](http://www.cardiomyopathy.org/Cardiomyopathy-change.html)

Book now for the Cardiomyopathy Information Day in Leeds on the 31st October 2015.

Find out about heart conditions, treatments and latest research as well as meeting families who are

affected. Call Sarah Dennis from Cardiomyopathy UK on:

01494 791224

to secure your place today.

CardiomyopathyUK
the heart muscle charity

Charities Registered Office: 31 Shearwater Drive, Torquay, Devon TQ2 7TL

Tel: 01709 210151 / 01803 613117

Email: info@alstrom.org.uk Web: www.alstrom.org.uk Facebook: www.facebook.com/alstromsyndromeuk

Twitter: @AS_UK Donate: www.justgiving.com/alstromsyndromeuk - **With your help, we have hope**
Alström Syndrome UK is a registered charity, number 1071196 - Registered Company number 3557191