Alström NEWS

With your help we have hope

THE NEWSLETTER OF ALSTRÖM SYNDROME UK

Winter 2014 Issue No.15

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

The Alström Syndrome UK Team would like to wish you all a Merry Christmas and a Happy and Healthy New Year

CHILDREN IN NEED FUNDING SECURED!

Supported by
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ASUK Office Manager, Catherine Lewis and ASUK National Development Manager, Kerry Leeson-Beevers have developed a transition project to ensure young people and their families who are affected by Alström Syndrome receive the support they need alongside establishing fun activities throughout their journey into adult services.

We are delighted to announce that the Children in Need application to run this three year transition project has been successful.

The 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.

Curtis Vasey and Kerry Leeson-Beevers are already busy planning support activities for the Transition Project. Our first regional support group is likely to take place in Scarborough as we do have a number of families within easy reach of this location. This will be a weekend break where we run workshops for young people, parents and siblings. We are also looking to plan an activity such as bowling and the Vasey's will help us to run a goalball session on the Sunday morning. We will be planning similar events at different locations throughout the UK.

ASUK is delighted to be awarded this grant and we look forward to starting this exciting project in the New Year.

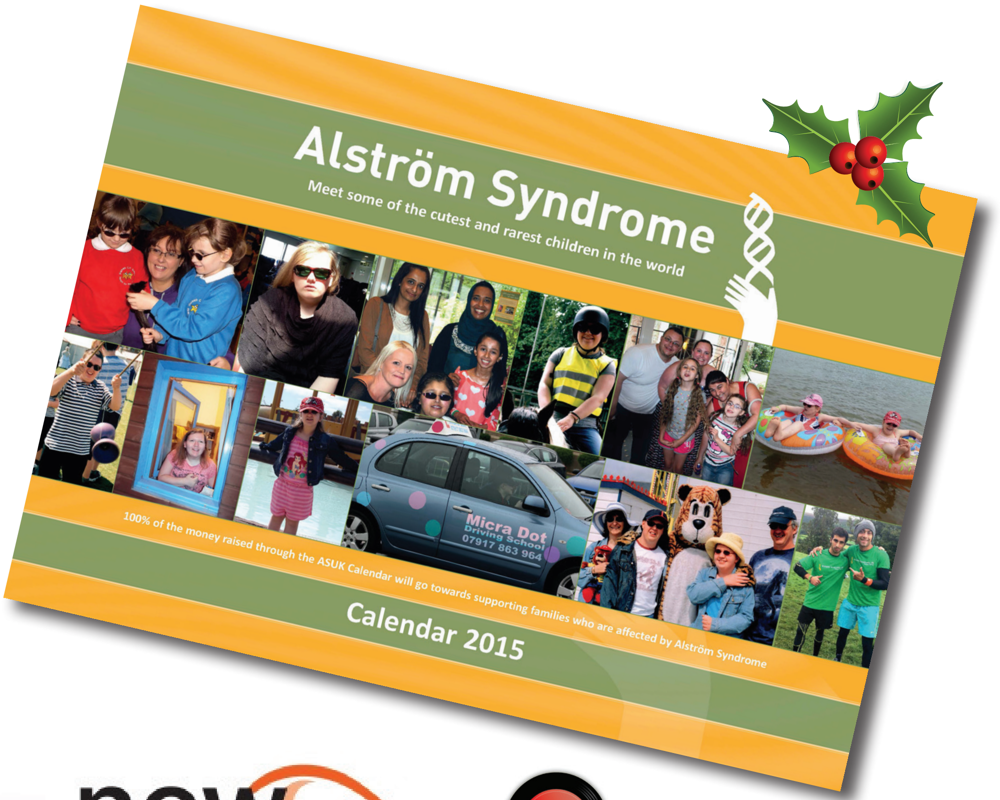
If you have any ideas for this project we would love to hear them. Please contact Curtis and Kerry on 01709 210151 or email curtis.vasey@alstrom.org.uk and kerry.leeson@alstrom.org.uk

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.

The ASUK 2015 calendar is now ready!

It showcases your truly special photographs. Please contact Catherine Lewis if you would like to order a calendar, they are priced at £10 each and 100% of the money raised will support families with Alström Syndrome.

Tel: 01803 613117 or email Catherine.lewis@alstrom.org.uk
Cheques can be made payable to 'Alström Syndrome UK' and sent to 31 Shearwater Drive, Torquay, Devon TQ2 7TL
Alternatively please make your payment through our Just Giving page https://www.justgiving.com/alstromsyndromeuk/



Thank you to our calendar Sponsors:

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Passionate about Diversity and Equality Training.
Specialists in Deaf, Disability and British Sign Language training

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ASUK RAISING AWARENESS THROUGHOUT THE WORLD

ASUK National Development Manager, Kerry Leeson-Beevers had the privilege of speaking at the prestigious World Orphan Drugs Congress in Brussels from the 12th – 14th November 2014. Kerry's presentation was about living with a rare disease and this journey. This was a great chance to network and learn from one another to develop solutions in the rare disease sector. Kerry is pictured far left with BBC Health Correspondent Fergus Walsh and Campaigner Alan Thomas at the event. You can learn more at http://www.terrapinn.com/conference/world-orphan-drug-congress/index.stm

Kerry has also attended the Acquired Deafblindness Conference in Belfast, where we had an information stand and ran a workshop to raise awareness of Alström Syndrome. 14 different countries were represented and many delegates were from specialised Deafblind Centres. There was a great deal of interest in the condition and many useful contacts have now been made.

On the 16th November 2014, ASUK Founder, Kay Parkinson gave a talk on the patient experience to SOBI Pharmaceutical's top 60 Executives at their strategy meeting held in Cap Ferrat France. She received a standing ovation for her talk and was presented with a book illustrating other patients' experiences and many of those present were moved to tears on hearing what her children had experienced. SOBI are a Swedish Pharmaceutical firm specialising in Rare Diseases and they would now like her to give the talk to their Board of Directors. Kay was also invited into their CEO's strategy round up which was a great experience and learning opportunity.

Kay then travelled to the Cilia Conference in Paris from the 18th – 21st November 2014. This event was attended by 390 delegates, which brought together patients, patient groups, clinicians and scientists.

ASUK Family Support Manager, Iram Shah attended the Family Fund Conference on Learning Technology and Disability in Cardiff on the 19th November 2014. The Conference was looking at how advances in technology are enabling disabled children to make great strides in their learning, communication and social skills. Family Fund, Chief Executive Cheryl Ward presented a short film of how new technology has made a difference to the development of two children. The workshops centred around accessible devices, physical and motor challenges and accessible learning and literacy.

Alström Syndrome Research Project – Have your say!

Sense are funding a research project which will be looking at the experiences of people with Alström Syndrome (and other rare syndromes) who attend specialist clinics. If you (or a relative) attend the QE in Birmingham or Birmingham's Children's hospital and would like more information about taking part in this research project please contact Liz who is a researcher at the University of Birmingham on l.ellis.2@bham.ac.uk or 0121 414 4392 07745 244597

Dates for your Diaries

Please find below the dates for next year's AS multi-disciplinary clinics. The Children's clinics will take place at Birmingham Children's Hospital

16th February 2015
18th May 2015
21st September 2015
16th November 2015
19th November 2015 – Children's Outreach Clinic in Leeds

The adult's clinics will take place at the Queen Elizabeth Hospital, Birmingham

7th/8th January 2015
20th/21st May 2015
23rd/24th September 2015
11th/12th November 2015

The UK is leading the way in the development of Genomic Medicine

The aim of the 100,000 Genome Project is to sequence 100,000 genomes by the year 2017. We all have around 20,000 genes made from our DNA and these are what make up our genome which is present in almost every cell of the body. The DNA of a person contains around 3 billion letters and the technology used to sequence these genes in recent years has been very slow and expensive.

As technology advances, the cost of gene sequencing has reduced and it now costs around £1,000 to sequence a whole genome and takes a matter of days to complete.

The purpose of this project is to develop an understanding of the role of different genes within the body and the impact they have.

The more samples collected, the greater the knowledge scientists hope to develop, hence the need for a sample size of 100,000.

It is also important to study the person and their medical history alongside the samples they collect. This will help to make links between the science and the experiences of individuals and how they are personally affected by a particular disease. The hope is that a greater understanding of genomics will result in personalised medicine which will enable medics to predict how a person may respond to particular treatments. If this is possible then genomic medicine may be made available as part of the NHS care we all receive in the UK.

Genomics England will initially focus on rare diseases and cancers. Ciliopathies will be covered under rare diseases and therefore there is a strong possibility that this will include Alström Syndrome, however, this will depend on those who consent to taking part. The samples collected will be from patients and two close blood relatives.

Rare diseases collectively are not rare and therefore genomics may have a major impact on both of these groups of diseases.

A key message being relayed is that patients are extremely important throughout this whole process and that informed consent is of prime importance. Genomics England has an Independent Ethics Advisory Committee to review all patient information and consent forms. They are running a series of patient engagement activities and ASUK have already been consulted.

There is a possibility that when studying a person's genome, researchers may discover 'incidental findings'. Only specific information will be fed back to the patient and they will be given the opportunity to opt in or out of receiving this information.

The information gathered from this project will be stored securely and anonymously by Genomics England. Medical professionals and commercial companies will be required to submit a research proposal and undergo an ethical review if they wish to gain access to this information.

The project may directly benefit some of those taking part by receiving new or improved treatments. However, the majority may not benefit directly but it is expected that their information will be used to develop knowledge and understanding and help to create additional research and development. There is still so much to learn about genomics but it is believed that this project is a major step in the right direction. ASUK will continue to follow this project closely and will update you regularly with any new developments. If you require any further information, please contact Kerry Leeson-Beevers, ASUK National Development Manager at Kerry.leeson@alstrom.org.uk

Further information can be found at Genomics England, The 100,000 Genomes Project, 2014 http://www.genomicsengland.co.uk/the-100000-genomes-project, (accessed 8 December 2014)

There is a great video 'understanding genomics' which you can view via the Genomics England website.

Let's Get Moving!

Let's think of those New Year's resolutions and get active and moving!

Exercise 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.

For further information you can get in touch with Nicky Cartwright (Physiotherapist at Queen Elizabeth Hospital Birmingham)
nicola.cartwright@uhb.nhs.uk

Your doctor can be a useful resource in accessing community 'health and fitness' initiatives at your local leisure centres through an 'exercise referral'. This can provide the one to one support you may need to 'get going' or to try new ways of exercising.

You will need to visit your GP to speak further with them about their 'exercise referral schemes'. I will be really pleased to hear about how you get on and what you've found out that is available to you. If who would like support accessing local health and fitness initiatives in your own area please contact me. Good exercising!

Have a Go Sports Day!

Action for Blind People and British Blind Sport would like to invite children and young people aged 8 to 17 with sight loss to a FREE have a go day to try out different sports. This day is taking place on the 10th February 2015 at the Doug Ellis Sports Centre, 150 Wellhead Lane, Birmingham, B42 2SY. As this event is during school time please ensure that you get permission from your school to attend. They will also need a parent to stay for the duration of the day. Various sporting activities will be on offer for the young people to try, with exit routes for these sports for them if they really enjoy them and want to continue them at a club.

Please contact Liz Williams for details on 0121 665 4200 or email elizabeth.williams@actionforblindpeople.org.uk

A HUGE THANK YOU to everyone who continues to support our great cause!

Thank you to **Les Wilkins** and the **Lewis family** who organised a quiz night in their local pub, this raised a fantastic £300 – well done!

Sue Vasey asked for donations to go towards ASUK rather than receive Birthday presents which raised a fabulous £250 – well done and Happy Birthday!

The **St. Agnes art group** held an art and craft exhibition in St. Agnes, Cornwall and raised £235

Kayleigh Lewis (pictured here) has been very busy making crafts and candles and selling them at craft fayres to raise funds for ASUK, thank you Kayleigh.

Please support **Melanie Davison** and her Brother **Fabian** who are bravely doing the Bath Half Marathon on the 1st March 2015 in aid of ASUK. Please show your support via her Just Giving page http://www.justgiving.com/Melanie-and-Iwe

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.

Kayleigh Lewis

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

Ice Bucket Challenge

Get involved in the fundraising craze which has gripped the world and is sweeping across social media – The Ice Bucket Challenge!

It's simple, just challenge a friend to have a bucket of ice-cold water poured over their head in exchange for a donation to ASUK, then they will nominate someone else to do the challenge. Make sure you film it and share it via Facebook to raise awareness of this ultra rare condition and raise funds for our great cause.

You can see the brave souls who have already taken on the challenge via our facebook page www.facebook.com/AlstromsyndromeUK - The videos are hilarious!

You can Text ICED12 £10 to 70070 to donate to Alström Syndrome UK and make a difference today.
You can also donate via our Just Giving page www.justgiving.com/alstromsyndromeuk/

FAMILY ADVENTURES!

Goal Ball Success!

Curtis, Taylor and **Rio Vasey** have been competing in a recent Goal Ball Tournament and they came 2nd – Well done lads!
You can learn more about Goal Ball and how you can get involved at http://www.goalballuk.com/

Taylor has not only been having fun doing Goal Ball but bravely did a sky dive – showing that anything is possible!

Stephen Thacker travelled to Torquay to enjoy one of his passions; to compete in a Chess Tournament, Stephen gives his thoughts below:
I lost the fourth round game and drew the remaining three to finish with 3.5 out of 7 which in a strong sighted tournament is a creditable score for me: chess is divided into three sections, the opening which is played fairly fast, the middle game where all the tactics take place and is played at the slowest speed, and the end game where time is sparse and sometimes it's a race to reach the time control as if you don't, you've forfeited the game! There were three time controls in this tournament: 40 moves in two hours for each player, two moves in the next hour for each player and finally, 30 minutes to finish the game. In the sixth game especially I blundered a piece in the opening only to fight back and almost win the game! I finished joint 15th but 21st on tie break with 3.5 and there were 45 in the tournament.

Judo Achievements!

Many of our members are finding that Judo can be fun and is a great way to build confidence and self esteem. Millie who is 7 years old (pictured LEFT) has just received her yellow belt in Judo and her Mum Michelle has seen her confidence and enthusiasm for the sport grow. Also John James (pictured RIGHT) who has just turned 19 years old has been doing Judo now for 4 years and he has achieved his Orange belt in Judo – well done to you both!

ASUK Bike Ride, Come and Join the Fun!

The planning for the Second Alström Syndrome Bike ride has begun. The first bike ride was a great success and took place in 2012 from Torbay Hospital, Devon to the Queen Elizabeth Hospital, Birmingham – a distance of over 200 miles – this signified the move of the medical team and sponsorship raised over £16,000.

This time the bike ride is likely to be a coast to coast route (TBC) and we are hoping to cycle from the 1st – 4th May 2015. We are aiming to raise money and awareness as well as socially interacting with each other and having a great time!

Anyone can take part and whether you have a bicycle, tandem, tricycle or tagalong you will be able to do as much or as little as you want. Any support that can be given will be greatly appreciated. Please get in touch if you would like to take part or if you would like to help by being part of the medical/support team.

Please contact either Alex Griffiths-Rayson or Curtis Vasey to register your interest
Alex.rayson@alstrom.org.uk or
Curtis.vasey@alstrom.org.uk

Hands-up who wants a Tandem or trike?

It's not too late to get in touch, so we can help you source and fund a tandem or trike for you and your family to get out and about.

Following funding from Sport England we can now fund tandems and trikes and support you through ensuring the support is in place for you to get out and about safely. Please get in touch if you would like to know more by calling Catherine Lewis on 01803 613117 or email catherine.lewis@alstrom.org.uk

Fun-Filled Residential Trips on Offer

Young People aged between 18 to 25 years with a visual impairment from across England can attend this exciting Action for Blind activity trip which is taking place from Tuesday 24th to Friday 27th March 2015 at the Windermere Manor Hotel, Lake District, Cumbria. The cost is £50.00

The Cost Includes: 3 nights accommodation on a full board basis, travel costs via train using a disabled railcard and all planned activities. You will also have the opportunity to use the hotel's leisure facilities such as the heated indoor swimming pool and sauna, plus make new found friends. All participants must be willing and able to take part in Workshop based sessions, around Education & Employment, Assistive Technology and Confidence Building including planned Social & Leisure Activities. To express an interest please email: philip.wagstaff@actionforblindpeople.org.uk or call 0161 787 9252

Victa is now open for applications for their February youth weekend for people aged 18 to 29 years old. After the success of their November weekend they will be returning to the Thorpe Woodlands Adventure Centre in Thetford Forest. On offer will be a range of thrill-seeking activities such as advanced high ropes, kayaking, raft building and caving. It will take place from the 6th – 9th February 2015 and will cost £35.00 The VICTA youth weekends are a great chance to meet new people from a similar age group and to gain new experiences.

If you would like to take part please apply or for more information please call Victa on:01908 240831 or email admin@victa.org.uk

Gap Year Adventure!

Liam Mackin decided to take a gap year, look at his options before going to University and fulfil his dream of travelling – and boy he did it in a big way! Liam shares his gap year adventure…

In August 2013, I received my A-level grades and, unfortunately, they were not high enough for me to attend my first choice university. I spoke to Kay who advised me that, rather than accepting my second choice, it would be better for me to have a gap year which would give me time to consider my options. So I did.

What did I do in my gap year then? Well, firstly I made sure to get a good look around at good, red-brick universities so I had a much better idea when it came to applying. Secondly, I picked up some leaflets and prospectuses from local colleges to see if there were any courses which would be suitable for me.

At Shipley college I enrolled on advanced French and German courses which I hoped would help me to keep up on my level of French and German (the subjects I will study for my degree). These courses have been really helpful, not only in maintaining my language skills but in improving them as we have general conversations about a range of different topics. It's also not a bad thing that the German class always ends up in the pub!

At Bradford college I found a community interpreting course. I enrolled and had to take an entrance exam. I was told that I would have to take the level 2, rather than the level 3 course. I wasn't happy with this because the level 3 course gives you the possibility of getting the necessary qualifications to become an interpreter, whereas level 2 doesn't. With help from my family, we contested it with the course tutors and their manager. After a lot of arguing and some help from the visual impairment team, they relented and allowed me to take the entrance exam again which I passed and I was finally accepted onto the level 3 course. I have a support worker and have also been lent a laptop and a voice recorder to help me with the course and I am delighted that in May this year I qualified as an Interpreter.

With all these on a regular basis I then looked for something else to fill my time with.

I asked ASUK Support Officer Catherine to ask around at a few schools as part of the PossAbility project to see if I could get a placement. She found me one, Dixon's City Academy, which were willing to accept me as a language assistant for a few hours per week. I work helping out with conversational classes for year 8's and 9's who are new to German and also with a 6th form student, giving him extra help to prepare for his oral exams. I'm really enjoying this and it is a valuable experience for me as teaching is the career I want to pursue after university.

Finally, in February, came my most recent piece of work. I have worked with the Children's Society's disability department on a regular basis for three or four years, so when I was approached by a member of the refugees and asylum seekers team, informing me of a scheme which they were running I was interested to find out more.

The scheme was based around young refugees, each of whom would receive one to one mentoring on subjects which they felt they need to improve. We had three training days to learn the difficulties faced by refugees and ways of overcoming communication difficulties as well as safeguarding.

I was matched with a girl from the Democratic Republic of the Congo (a French-speaking country!) who wanted help with conversational English. When I initially met her, her sister asked if she could also join us and they now both take part in the weekly sessions where we practice conversational English and I also learn and practice my French!

On top of all this I have managed to factor in some travelling, something I have always wanted to do.

In January I went to India for twelve days with a company called Traveleyes, who organise holidays for blind and partially sighted people.

We flew to Delhi where we visited India's largest mosque and temple, rode through the old town on rickshaws and saw Mahatma Gandhi's house and other historical and religious sites.

We then travelled to Agra where we visited the amazing Taj Mahal and were able to have our photographs taken on the bench where Princess Diana sat, as well as to enter the building. We also visited the fort of Agra and the Marble Art Palace where artifacts are made from the same marble of which the Taj Mahal was constructed.

Our final journey took us to Jaipur, India's cultural capital, where we rode on camels and elephants, visited the Amber Palace, fought our way through a typical Indian market and saw how paper is made from cotton in the village of Sanganir. It was a truly amazing experience and one which I will never forget.

I also visited Dublin on St.Patrick's day and stayed for four days which was a great experience. We lined the streets for the St.Patrick's Day parade, visited Trinity College and Dublin Castle and, of course, sampled some Guinness.

In June I decided that I wanted to try and fulfil an ambition I had had for a long time and apply for a TV quiz show. I went onto the BBC website and found a new show which they were recruiting for called Two Tribes. I filled in my application and thought nothing of it. A couple of weeks later I had a phone call from one of the producers, they wanted me to take a short test over the phone to see if I was suitable for the show. I was asked 10 general knowledge questions and was told I might hear back from them if I was successful.

A little while later I was asked to go to Manchester for an audition. We had to answer a series of written questions, role play the show and do a short piece to camera. A week later I heard that I was wanted on a recording of the show. I had to go down to London and take part in the recording.

The game show was aired on TV on the 28th August 2014 and you can see how I did via the You Tube video:
http://youtu.be/U9MGl88HikY

I thought that that was the end of my gap year but in August I was selected from a list of applicants to travel to Latvia to take part in the International Computer Camp. A group of 6 people from the UK joined up with representatives from over 15 different countries to create an international network of young blind people. We took part in several workshops including woodwork, judo and cookery and visited some of the tourist attractions. It was a wonderful 12 days where I got to meet lots of new friends, so now when I want to go travelling I've always got somewhere to stay.

I still have some more travelling planned with trips to Malta, Italy and China before I begin at Nottingham University, where I have been offered an unconditional place.

On top of all of this I have also found time to improve my mobility skills by doing lots of train travel around the country, met lots of new people and won another gold medal for goalball. I don't regret my decision to take a gap year, in fact I think it's one of the best choices I have made and I wouldn't have done it without Kay's advice!

Until next time!

Merry Christmas and a Happy New Year

Did you know?

The Charities Registered Office address has changed to:
31 Shearwater Drive, Torquay, Devon TQ2 7TL

Tel: **01803 613117**
Email: info@alstrom.org.uk

ASUK T-shirts are available in sizes small, medium, large, Extra Large and 2 XL.
Families affected by AS may have a complimentary t-shirt per person affected.
Additional t-shirts are priced at £10 each.

ASUK now have a dedicated ***You Tube*** page - www.youtube.com/alstromsyndromeuk
Please keep up to date with Alström Syndrome UK via twitter @AS_UK

We also have a dedicated ***Facebook*** page - www.facebook.com/alstromsyndromeuk
Please click 'like' to show your support and spread the word!

You can also find more information at:
https://www.rareconnect.org/en/community/alstrom-syndrome
Connecting rare disease patients globally.

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

Please show your support or get involved in one of our fundraising challenges.
Every penny which is donated supports children and adults with Alström Syndrome.

You can donate via our ***Just Giving*** page
https://www.justgiving.com/alstromsyndromeuk/

With your help, we have hope

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