



# Alström NEWS

With your help we have hope

THE NEWSLETTER OF ALSTRÖM SYNDROME UK

Winter 2013 -14 Issue No. 13

## Hannah and Katie ... such devoted sisters

### Seven-year-old Hannah and Katie Beck are the only known twins in Britain with Alström Syndrome.

There are 60 known children and adults with AS in the UK, and 800 worldwide.

Hannah and Katie, who were diagnosed when they were four, are learning to use white canes and Braille.

They both attend a mainstream primary school where they get special one-to-one teaching to help them live as normally as possible.

Their parents Julie, 41, and Ian, 44, from Harrogate, North Yorkshire, both carry the Alström Syndrome gene but are unaffected themselves. As we know AS is a rare, recessively inherited genetic disorder.

However, they are determined to give their daughters as full a life as they can.

Mrs Beck said: "We took them on their first holiday abroad to Lanzarote in February and Hannah particularly loved it and begged to go back again so we're going back next year."

"Katie loves all the Disney princesses, so we want to take them to Disneyland.

"She loves singing and has loads of confidence, while Hannah is really quiet and a lot more shy.

"But really they are just two normal, seven-year-old girls who love horse riding and swimming, which is great for their posture and will benefit them greatly.

"They're mad about playing with dolls and love school, and the other pupils have really taken to them and treat them just like everyone else.'

She added: "They are doing so well



*Above: Twin sisters Hannah and Katie - singing in the rain on a windswept beach*

and they are so strong-willed. I'm proud of their determination and their wish to be independent. They also have a fantastic bond, like all twins really, but I think they have a bit of a sixth sense about when the other really needs them.

"Ultimately they really lean on each other and spur each other on, and I'm glad they have each other for that support."

Mrs Beck, herself a primary school teacher, echoes the experiences of many other families - where experts struggled to identify what the girls were suffering from. Since diagnosis the family say they have received life-changing support from Alström Syndrome UK (ASUK).

Mrs Beck is also a trustee of ASUK, which is attempting to raise £1m for research - see <http://www.justgiving.com/ChildinaMillion>



The 'Living with Alström Syndrome' DVD, produced by RKD Films and kindly funded by Genetic Disorders UK and Jeans for Genes, can now be watched in English, Urdu, Arabic, Hindi and Bengali. Alongside the 'Living with Alström Syndrome' video you can also see videos about our latest conference and a training video. Please contact Kerry on 01709 210151 or email [Kerry.leeson@alstrom.org.uk](mailto:Kerry.leeson@alstrom.org.uk) for a hard copy of the DVD. The videos can be viewed via <http://www.youtube.com/alstromsyndromeuk>

## Board members take more active role

ASUK now have a dynamic and skilful Board of Trustees who will be more involved and proactive in the running of the charity, as they will now take on the Executive role. Kerry Leeson-Beevers who has been doing an excellent job

as Acting Chief Executive Officer will become ASUK's Development Manager.

The Board of Trustees expresses their grateful thanks to Kerry for holding the fort while they instigated the changes

needed to take the charity forward. Catherine Lewis has now become ASUK's Office Manager and Iram Shah becomes Family Support Officer, to work together with the Trustees as a dynamic and strong team.

# Search for treatments

BY KAY PARKINSON

**Having lived with Alström Syndrome for over 30 years now and run the charity for 15 of those years, I was becoming frustrated with the lack of treatments available and progress into fundamental understanding of the disease.**

Having met Dr Vincent Marion from Strasbourg, who is pioneering new treatments it became clear that without more patients, possible clinical trials for new drugs would not happen.

So two years ago now Alström Europe was created, we have just held our second meeting in Milan where we discussed possible grant applications for the EU Horizon funding programmes which will be launched early next year.

There are two grant programmes we are working towards one which will be co-ordinated by the Dr Vincent Marion under the Innovative Medicine Initiative 2 (IMI 2) and the other by Queen Elizabeth Hospital, Birmingham, under the Horizon Health programme. Both will have patient interests and involvement central to their projects and work Pan-European and globally.

Whilst in Milan, John and I (with help from Professor Barrett ) manned the EuroWABB (Wolfram, Alström and Bardet Biedl) stand at the 9th International Endocrinology Society Meeting; we met quite a number of doctors working with all three conditions and were pleased that all diagnostic criteria handbooks were given



**ASUK Founder and Director Kay has worked tirelessly for families affected by Alström Syndrome and this continues with Kay now forming exciting links within Europe**

out. The conference was a great awareness raising success.

I was fortunate to attend a EURORDIS training workshop in Paris which gave more information on the grants, which we are hoping to apply for. Magda Gunn, Scientific Project Manager, talked about the Innovative Medicine Initiative (IMI) which is jointly funded by the EU Commission and European Federation of Pharmaceutical Industry Associations (EFPIA)

### INNOVATIVE MEDICINES INITIATIVE

The Innovative Medicines Initiative (IMI) is Europe's largest public-private initiative aiming to speed up the development of better and safer medicines for patients.

IMI supports collaborative research projects and builds networks of industrial and academic experts in order to boost pharmaceutical innovation in Europe. IMI is a joint undertaking between the European Union and the pharmaceutical industry association EFPIA.

### HORIZON HEALTH, DEMOGRAPHIC CHANGE AND WELL BEING

These topics will improve our understanding of the causes and mechanisms underlying health, healthy ageing and disease; improve our ability to monitor health and to prevent, detect, treat and manage disease; support older persons to remain active and healthy; and test and demonstrate new models and tools for health and care delivery.

In doing so, support will be provided to research and innovation performers, including significant, tailored support to small and medium-sized enterprises.

November saw John and I in Geneva at the World Orphan Drug Conference where we presented a poster on the "Cellular Modelling of Alström Syndrome in Primary Dermal Fibroblast and Derived Cells from Affected patients". This is the work that was undertaken with the Big Lottery fund.

Following this I was asked to talk at the Hands on Biobank Conference in The Hague on experience of developing a Patient Registry and Biobank

Life is hectic, but we are enjoying immensely the opportunities which have really opened up since we were awarded the EURORDIS Patient Organisation Award.

● Kay has accepted EURORDIS' invitation to join the Rare Connect Patient Advisory Council (PAC). This is a newly-launched European project called RD-Connect in which EURORDIS' role is to integrate the patient perspective. The objective of RD-Connect is to create a platform and framework for integrating research data (patient registries, biobanks and genetic data). <https://www.rareconnect.org/>

## Publication selected for prestigious event

Kay Parkinson has had her publication selected for a prestigious event: DIA (Drug Information Association Conference "Increasing Health Literacy and making Doctor-Patient Communication more effective" has been selected for inclusion at the 26th Annual Euro Meeting in Vienna on the 25th - 27th March 2014. This will include a 20-25-minute presentation on this topic at this influential meeting.

# Yes, I was a Princess for a day

Maariyah Iqbal's wish of being a Princess for the day was granted by 'Rays of Sunshine' at Dudley Zoological Gardens back in August.

Maariyah was dressed as a beautiful Princess and had a personal tour around the castle accompanied by her own personal guard.

She was given a bag full of goodies on behalf of the zoo and was treated with the utmost priority, just like a Princess.

She had a close encounter with the amazing animals at the zoo and enjoyed the

personal experience of touching the birds. Maariyah's joy was visible to see through her beaming smile which lasted all day.

Maariyah's family were pleased to see Maariyah having a fantastic time as she has recently spent lots of time in Hospital.

Maariyah was crowned queen and a scroll was honoured to her on behalf of the castle.

The best part of the day was the arrival of the horse and carriage to take Maariyah home. A perfect ending for a truly special princess day.



## Partnership will increase support

Recently The Royal Blind Society have started to work in partnership with VICTA, the organisation helping blind and partially-sighted children. This relationship has enabled them to significantly increase the support services which they offer to families, children and adults who are affected by a visual impairment.

VICTA also run a number of activity breaks throughout the year and can help with grants for equipment and holidays. Please get in touch if you need any help completing these forms.

Further information can be found at <http://www.victa.org.uk/>

## Fancy a fantastic holiday?

Torbay Holiday Helpers Network (THHN) now has a fantastic new luxury static caravan that is situated in the brilliant Haulfryn Finlake Holiday Park in South Devon.

The caravan sleeps up to 4 and this luxury caravan has a very large private decking space with a huge hot tub, outside seating/eating area and a top of the range BBQ. The caravan does have ramp access, however there is no disabled facilities within the caravan and the entrance door is not adapted in any way. The caravan overlooks a peaceful lake and is surrounded by dozens of woodland walks.

The park facilities include fishing; golf; tennis courts; gym; indoor and outdoor swimming pools; sauna and steam room which families can access free of charge and a huge amount of other activities and services are available at a discounted rate.

THHN will be offering future holidays all year round at "Memory Maker" using rolling Saturday - Friday system.

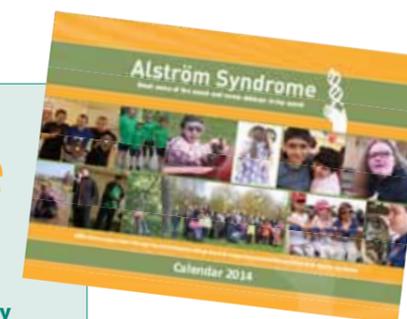
Please get in touch if you would like ASUK to refer your family for this fantastic break.

Contact: Catherine Lewis, Office Manager, Tel. 01803 613117 Mobile 07970071675 or email [catherine.lewis@alstrom.org.uk](mailto:catherine.lewis@alstrom.org.uk)

## It's time to order the new ASUK calendar

The ASUK 2014 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 will make great Christmas presents!

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



We are grateful to all our sponsors for the new calendar - Hearfirst, New College, Worcester, Colour Digital Print and on-line store specialist Retro Box

HearFirst  
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ncw

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PRINT LIMITED



# We're having fun!



**ASUK organised a family week in Torquay from the 18th – 25th August 2013 which incorporated 15 adults and 15 children.**

Families had a fabulous time in the resort being involved in lots of fun activities and benefiting from sharing each other's

experiences. We would like to say a HUGE thank you to Torbay Holiday Helpers Network who provided each family with an activity pack which included a free pass into many fun attractions. Also a HUGE thank you to Brunel Manor who organised complimentary accommodation for all

families. Without this support the family week wouldn't have been able to take place. Also Dawlish Water Rotary club provided a grant to make sure all families had a wonderful time in Torquay, so thank you to everyone who made the family week truly special – we are hoping we can do this again next year!



Also late in August many families enjoyed a fun-filled trip to Chessington World of Adventures with its zoo and sea-life centre. Our thanks go to Merlin's Magic Wand who enabled families to gain access to the park free of charge. See below for some of our photos from the trip.



## Action-packed family trip



Families had a great time at the Calvert Trust adventure centre in the Lake District, although the weather wasn't the best, this didn't dampen their spirits as they had a fabulous time doing gladiator challenges in the pool. They also enjoyed horse-riding, archery, canoeing, climbing and also enjoyed the opportunity to socialise together. A HUGE thank you to Bradford Short Breaks Fund which enabled families from Bradford to attend the trip.

## Special party with Santa



Maariyah Iqbal, Kion Leeson-Beevers and their families enjoyed a wonderful Christmas party at Drayton Manor, thanks to the National Blind Children's Society. "It really was a great day and all of the children really enjoyed themselves. NBCS staff were on hand to offer support and discuss the services they provide. We had time to enjoy the rides and Ayisha and Cole even got me and Javed on the roller-coaster which was great fun! We had a very nice lunch and even a visit from Santa with lovely gifts for every child and the day ended with a firework display. This was also a good opportunity to meet other families," says Kerry Leeson-Beevers.

## Chair's Report



Dear Families,

2013 has been a very exciting year for Alström Syndrome UK. There have been many positive changes within ASUK and the Board of Trustees are now taking a more proactive and executive approach to move the charity forward.

They will continue to work with Kerry, Catherine, Iram and John to ensure families receive the support and help they need alongside promoting awareness and looking at exciting new research projects.

With the new approach, Kay Parkinson has been able to move her focus onto collaborating with specialists, researchers and funding partners across Europe and the rest of the world.

We hope this will lead to better treatments, research and clinical trials which could improve the lives of people with AS. Kay has already made a impact in starting Alström Syndrome Europe and discussions have started into moving our research project forward and expanding upon this vital work.

The ASUK Family Week (see more on these pages) was great fun and enabled families from across the UK to get together and support one another. Next year's Family Week will give more families the chance to come together as it will combine the ASUK Family and Professional conference. I hope to see you all there!

ASUK are currently looking for people with a passion for our work to serve on the Board of Trustees. Please feel free to contact me if you are interested in knowing more about this role or if you have any suggestions or ideas for the charity's work.

Best Wishes  
Michelle Hough  
Michelle.hough@alstrom.org.uk  
Tel: 01872 553067

## Now raise your hands!

Alexion Pharma UK is asking people to be 1 in 2,000 by raising their hand as a symbol of Rare Disease Day (Feb 28th). Your hand will become part of a powerful, contemporary art installation that will be designed by an aspiring British artist. The installation will bring 2,000 hands together as one. We're supporting this new rare disease awareness campaign at [www.raiseyourhand.co.uk](http://www.raiseyourhand.co.uk) <<http://www.raiseyourhand.co.uk/>> - please sign up and 'Raise a Hand' against rare disease.

## World of adventure



## More opportunities at the new-style family conference

The ASUK Family and Professional conference 2014 will take place at Brunel Manor in Torquay on Saturday 16th August 2014.

There will be a dinner and charity auction in the evening and both families and professionals will have the opportunity to meet and network. The Family

and Professional conference and our AGM will be held on the 16th August 2014 and this will enable updates to be shared regarding the pioneering research we are currently undertaking. Please contact Catherine Lewis for further information or to reserve your place on 07970 071675 or email

catherine.lewis@alstrom.org.uk Please also get in touch if you would like to know more about the company sponsorship packages available for this event. We look forward to seeing you all there! Please note there is limited accommodation at Brunel Manor and Brunel Lodge so families will

need to book this accommodation early in the New Year if they would like to stay here. Brunel accommodation rates are £46 per person per night on a B&B basis, children 0-5 year olds are free, 6 - 12 year olds are £11.50 and 13 - 17 year olds are £23.00 per child per night. You can book this

accommodation by contacting Brunel on 01803 329333 and mention you are attending the ASUK conference. Off site the Travel Lodge, if you are quick, offers rooms from £32 per night and there are many hotels in Torbay if you would like to incorporate a holiday.



# News on the Fundraising front..

## T-shirts are one way you can help...

ASUK T-shirts are available in small, medium, large, ex large and 2 XL. Families affected by AS may have a complimentary T-shirt per person affected. Additional T-shirts are £10 each. ASUK now have a dedicated You Tube page <http://www.youtube.com/alstromsyndromeuk> Please keep up to date with Alström Syndrome UK via twitter @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! You can also find more information at <https://www.rareconnect.org/en/community/alstrom-syndrome> - connecting rare disease patients globally. 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. If you would like to register for any of the ASUK fundraising events, please contact Catherine Lewis on 01803 613117 or email [catherine.lewis@alstrom.org.uk](mailto:catherine.lewis@alstrom.org.uk) Here is the address where you can donate [www.justgiving.com/alstromsyndromeuk](http://www.justgiving.com/alstromsyndromeuk)

## Thanks to everyone

A HUGE thank you also to everyone who continues to support our great cause. Thank you to all the families who fundraise for us in particular Liz and Dean Little who donate 10% from their table top sale to ASUK and to the Lewis family who have raised £40 from saving 5 pences, the Haslingdon High School, Lancashire who donated £100 following their school charity day and Bramhall Methodist Thrift charity shop who donated £500. Julie Ryder's (Hearfirst Training) daughter Annie Ryder raised £98.57 by having a stall at her school charity day and the Corporation Club in Scarborough raised £120 for ASUK to enable young adults and children to take part in fun activities - through a 70s night and a quiz.

In December the Dragon Meet Convention met for London's foremost single day of role playing, board games and collectable card gaming. ASUK were honoured to be chosen as their charity and offer a HUGE THANK YOU as they raised a magnificent total of £2,274.93.

## New project offers opportunities

**Why not get involved in our new PossAbility project? ASUK have received funding from Awards for All to enable individuals who have been diagnosed with AS to participate in voluntary opportunities and activities in their local area.**

ASUK can support you through this process and help make this possible. We will also support the organisations involved to help them offer young people and adults the chance to make these dreams a reality and hopefully lead to paid employment.

Some ideas could be animal care, school/college assistant, gardening and many more...If you have an area of interest where you would like to develop your skills or if you are an organisation who would like to help then please contact Catherine Lewis on 01803 613117 or email [catherine.lewis@alstrom.org.uk](mailto:catherine.lewis@alstrom.org.uk).

## Farewell and a big thank you to Julia!

A HUGE THANK-YOU to our fundraiser Julia Scott who is leaving after Christmas. In difficult times, Julia has done an outstanding job tracking down various charitable trusts to help raise money for ASUK. Like many other charities, we have seen donations decline in the current economic climate making it more important than ever that we all work together to try and develop effective ways to raise money for our families.

## How you can support our Marathon team

Please support Lucy Leeson, Rik Bennett and Brian Vasey who have joined the ASUK running team and will be running in the 2014 Virgin London Marathon.

You can support Lucy via her **Just Giving page** <http://www.justgiving.com/Lucylovespugs>

Also Lucy has been able to design her own Juicy Lucy Cocktail for a local bar, in Yorkshire who are going to donate £1 to ASUK for every cocktail which is sold.

Their website is <http://www.theshedbeverley.com/>

Luke Waiting will be doing the Manchester Marathon in aid of ASUK and we still have places available in the BUPA London 10K Marathon and the Adidas Silverstone Half Marathon. Please get in touch if you would like to join our running teams!

## Choir concert to boost funds

Exeter Police and Community Choir are to stage a concert in Paignton on Thursday, March 6 2014 in aid of Alstrom Syndrome UK.

The concert will be held at St. Paul's Church, Paignton, and tickets are likely to be in the region of £6.00. The Police and Community Choir give concerts all over Devon and help churches, community

and charity groups to boost their funds. The choir's repertoire includes popular songs, hymns and spirituals. A soloist will also be joining the choir so a great evening of music is in store.

For enquiries about tickets, please contact Catherine Lewis, Office Manager, Tel. 01803 613117 Mob 07970071675 or email: [catherine.lewis@alstrom.org.uk](mailto:catherine.lewis@alstrom.org.uk)

When Alström Syndrome was first granted funding to develop the AS multi-disciplinary clinics this was based on 30 patients attending. We now have 60 patients who attend these clinics and we have received no increase in our budget. Therefore it has become unsustainable that we are able to offer an evening meal as part of these clinics. Families will still be able to receive the Hospital meal voucher whilst attending the clinic and all accommodation will be provided on a B&B basis, but we will be unable to provide an evening meal for future clinics.

## Real progress in Asian communities

As many of you may be aware, I began working specifically within Asian communities five years ago. In 2011, Kay Parkinson and I developed the Asian Mentoring Scheme.

This project has attracted a phenomenal amount of money, thanks to the fundraising efforts of Julia Scott.

We recently received a further £18,722 from the Sylvia Adams Charitable Trust to enable us to complete the final year of the project. We are extremely grateful for the support that the Trust

have given us throughout the project and to all of the other organisations and individuals who have supported us.

The Asian Mentoring Scheme goes from strength to strength and we are extremely proud of our achievements. We feel we are meeting the aims and objectives of the project and are improving outcomes for families. Iram Shah has been a wonderful addition to the team and her ability to relate to families and converse in Urdu and Punjabi has ensured that the support we

are able to offer to families is accessible to all. We have some magnificent families and as a team we feel privileged to work with each and every one of you!

A full analysis of our work on this project will soon be available on our website. If you would like any additional information, please contact me on 01709 210151 / 07716 135940 or email [kerry.leeson@alstrom.org.uk](mailto:kerry.leeson@alstrom.org.uk)

**Kerry Leeson-Beevers,  
Development Manager**

## Garden makeover

Gardening guru Alan Titchmarsh is searching for special individuals or families anywhere in the country who would really benefit from a new garden... as long as they are special people, and it would enhance their lives then Alan wants to hear from you. We would need photographs of your family and of the garden in its current state!

We think all our members are truly special and if you need a garden makeover get in touch with Catherine Lewis today...

## 2014 clinic dates

Children's Clinics to be held at Birmingham Children's Hospital

17th February 2014

19th May 2014

22nd September 2014

17th November 2014

Adult Clinics to be held at the Queen Elizabeth Hospital, Birmingham

8th and 9th January 2014

7th and 8th May 2014

11th and 12th June 2014

24th and 25th September 2014

19th and 20th November 2014

If dates need to be altered for any reason you will be informed as soon as possible.

## Seek advice before you try out any new diet

Fruit and vegetables - as we reported before - are part of a balanced diet and can help us stay healthy. That is why it so important that we get enough of them.

There are no individual foods or pills that will miraculously burn fat. Some diets and ingredients in supplements and herbal products can even be dangerous. 'Fad Diets' are often temporary

weight loss which can lead to additional health problems. Before any diet is undertaken ensure you speak to your health care provider for advice.

Alström Syndrome UK and the Birmingham Children's Hospital recommend a healthy balanced diet alongside regular exercise. We understand that this can sometimes be very challenging to achieve but it is very

important for optimal health.

Please get in touch if you need any help or support or if there is any information which you would like to see included in the next e-newsletter.

● The information here is provided by Hazel Riggall RD, Clinical Specialist Paediatric Dietitian from Birmingham Children's Hospital.

## Wobbly Wednesday raises awareness

The very first International Nystagmus Awareness Day 'Wobbly Wednesday' was held in November.

People around the world are being encouraged to organise awareness and fund-raising events in their communities to support the work of Nystagmus Network. Event organisers can register for a supporters pack by emailing [info@nystagmusnet.org](mailto:info@nystagmusnet.org).

Through Facebook supporters can interact with each other <https://www.facebook.com/groups/325221670943004/>. You can buy wristbands and badges from the Nystagmus Network website <http://www.nystagmusnet.org> <<http://www.nystagmusnet.org/>>

## Social Media and Website development

Please keep up to date with Alström Syndrome UK via twitter @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! You can also find more information at <https://www.rareconnect.org/en/community/alstrom-syndrome> - connecting rare disease patients globally.

The website is still developing and carries positive family stories and inspirational fundraising articles. Please send me your articles or up and coming events along with photographs and Catherine Lewis can put them onto the website. Please visit [www.alstrom.org.uk](http://www.alstrom.org.uk) <<http://www.alstrom.org.uk/>> to have a look around.

# They're top of league

**ASUK members, brothers Curtis and Taylor Vasey won the novice goalball league and Liam Mackin won the intermediate league. Taylor ended up as leading goal scorer!**

This is a great achievement - well done Lads!! Liam has been playing goalball for four years and usually he trains 3-4 times a week.

Liam gives his thoughts: "I absolutely love goalball and although it's a great physical workout I can play for hours as

my mind is focused on the game and my technique helps to stop me getting tired." Liam enjoys this sport so much he has even tried coaching!

Are you interested in playing goalball? Or becoming a coach? Goalball UK are launching new clubs around the country. A number of free 'have a go' sessions are being held for potential players, coaches, officials.

You can find more information about goalball at <http://www.goalballuk.com/>



If you want to learn more about Liam's 'Mackin Mobility Training' and Sighted Guide Support you can contact him on 07748 695900 or email [liam\\_mackin@hotmail.co.uk](mailto:liam_mackin@hotmail.co.uk)

## Six take Liam's training course

**Industrious Liam has trained six people in Scarborough with his Sighted Guide Training.**

Congratulations also to Liam who achieved excellent results in his A-Levels - with an A\* in French an A in German and a C in History. Also John - James Entwistle achieved excellent grades in his GCSE results and is now able to study A Levels in History, Government, Politics and Sociology at New College Worcester. Well done indeed!



## Iram heads fight to help more families

This year ASUK have been very busy raising awareness of Alstrom Syndrome and our work throughout the UK and across Europe. We have attended many prestigious conferences and events including attending Sight Village Conferences in Birmingham, Edinburgh and London. Iram Shah (pictured left) has been raising awareness throughout the UK and is the ASUK

Family Support Worker she is going to be visiting families in the New Year to do Special Needs Assessments to ensure all families are getting the support and assistance that they deserve.

Iram also speaks Urdu and Punjabi and you can contact her for support on 07988 237664 or email [iram.shah@alstrom.org.uk](mailto:iram.shah@alstrom.org.uk)

## Strategy for Rare Diseases gets under way

**The first UK Strategy for Rare Diseases has been published to drive changes in the way rare diseases are dealt with. The strategy aims to put the patients' needs first.**

*This is something ASUK has been trying to achieve for some time through greater awareness. The strategy intends to bring together the talent, skills and*

*professionalism of all relevant sectors to enact real, positive change in how people with rare diseases and complex conditions are dealt with.*

*There are between 5,000 and 8,000 rare diseases. Each one affects less than 0.1% of the UK's population, but together they affect the lives of 3 million people. The UK is a recognised leader in research, treatment and care for rare diseases. It is*

*at the forefront of the genomics revolution which could radically transform the way we diagnose and treat patients with rare diseases.*

*Moves are already starting to drive this work forward, through initiatives such as the programme that will map the whole genome of 100,000 NHS patients which has rare disease as one of its key priorities.*