



Alström NEWS

With your help we have hope

THE NEWSLETTER OF ALSTRÖM SYNDROME UK

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Now we look to setting up Alström Asia

We are setting our sights on the East - ASUK Family Support Manager Iram Shah and ASUK Trustee Kez Hayat alongside the support of ASUK Founder and Director Kay Parkinson are looking into starting up Alström Asia.

We are currently assessing the roots where families affected by Alström Syndrome originate from in Asia, as this

will provide us with a bigger picture of specific locations to target.

Iram and Kez are planning on collaboratively working with other organisations in Asia and we would appreciate any information and contacts with families or medical professionals in Asia. Please contact Iram.shah@alstrom.org.uk with any further information.

Also, due to the rising numbers of Asian families within the charity, we are seeking Asian Trustees to be part of our ASUK board.

We are open to consider applicants from any cultural background, gender or religion as we strongly believe equality and diversity is crucial in any leading organisation.

'5m Euro bid' hope

We recently put forth a five million Euro bid with partners from across the EU but have been unsuccessful on this occasion.

The bid for Horizon 2020 was masterminded by ASUK Founder and Director Kay Parkinson.

It specifically focused at empowering patients through 'real time' monitoring of their personal health needs, by mobile health applications.

ASUK and ASEU are meeting on the 25th and 26th September to discuss re-applying to PHC 27 next year - now with the benefit of detailed feedback from the EU and a great collaborative working team in place.

8 EU countries would all link "virtually" to our UK multi-disciplinary clinics, increasing expertise and knowledge for all.

The bid was written and submitted in collaboration with Dr Tarek Hiwot who may lead this state-of-the-art project based from the Queen Elizabeth Hospital, Birmingham in the future.

Thank you to everyone who participated in the London School of Economics (LSE) survey, kindly conducted by the MSc students as an evidence base for the Horizon 2020 grant.

The students supplied a very detailed report for ASUK to show the financial implications of having Alström Syndrome and the cost effectiveness of the multi-disciplinary clinics which we coordinate. It also highlighted the importance of the ASUK community in providing support, helping to tackle isolation and creating an environment for free discussion of experiences and problems.

- Story continued on the Back Page



Liam Mackin decided to take a gap year, look further at his options for university and fulfil his dream to travel to amazing places - and, boy, did he do that in a big way! Here he is relaxing with his family in India. You can read more about Liam's adventures in our next issue!

Gaining influence around world

Kay and John Parkinson have crossed half the world in their continued quest to seek influential international contacts and new sources of funding and research.

Their travels have already fostered valuable links as they spread awareness of Alström Syndrome and carry the flag for all those with rare diseases.

In late April they headed for Washington - funded by Dr Pierre Laurin from the Canadian group Prometics, which has developed an anti-fibrosis treatment which is potentially life saving for those with AS. Negotiations are under way see if he can be allowed access to the skin samples taken from the BIG Lottery research - or to acquire new skin samples.

Washington's 4th annual World Orphan Drug Congress is where industry, government and key players descend to develop new strategies and partnerships to advance orphan drug development and patient access.

Not long after returning from Washington, Kay and Family Liaison Officer John left for Berlin for the European Rare Disease Conference (ERDC) where Kay was involved in panel debates on Rare Disease Policy as well as chairing a session with top International professors on "Stimulating Research and Removing Barriers." John was manning the EURO-WABB stand and distributing the new guidelines on Alström, Wolfram and Bardet Biedl Syndromes



Professor Tim Barrett, who is leading the EURO-WABB project, pictured with Kay and John Parkinson when they met up in Berlin for the European Rare Disease Conference

and Professor Tim Barrett who has led on the EURO-WABB project thanked John for his fantastic work in helping disseminate EURO-WABB information.

In March Kay spoke at short notice on Patient Registries in Switzerland at the RE(ACT) research of rare and orphan diseases international congress 2014 and John manned a stand there. This high-profile conference, funded by the Black Swan Foundation, explored cutting-

edge technologies and key issues that need to be addressed if we are to see new and promising therapies and treatments delivered rapidly around the world.

Kay was also invited to address the Drug Information Association's (DIA) annual conference in Vienna on 'Increasing health literacy and making doctor-patient communication more effective' – an opportunity to showcase patient empowerment.

Key role in rare disease strategy

National Development Manager Kerry Leeson-Beevers has had the honour of being asked to become a member of the UK Rare Disease Patient Forum to help oversee implementation of the UK Strategy for Rare Diseases. Kerry has also been successful in her application to

become an alternate member of the Paediatric Committee (PDCO) at the European Medicines Agency on behalf of EURORDIS. The role of the PDCO is to evaluate paediatric investigation plans and consider the quality and safety of medicines to be used within the paediatric population.

All joining together .. for better care



Rare Disease Day this year was all about everyone *Joining Together for Better Care!*

ASUK was represented at many events. Kay Parkinson flew out to Heidelberg in Germany to attend the RD-Connect conference. RD-Connect is a European Commission sponsored project led by Eurordis.

Its long-term plan is to collect medical information and tissue samples from rare disease patients to increase understanding of diseases, new treatments and improved care.

Family Support Manager Iram Shah (seen left) attended the Rare Disease UK event at the House of Commons - a great opportunity to find out more about the latest developments in the rare disease sector, including the Rare Disease Strategy.

Development Manager Kerry Leeson-Beevers attended the Science Museum in London for the unveiling of an art piece, depicting the support of the general public- and Alexion Pharma UK asked people to be 1 in 2000 by raising their hand as a symbol of Rare Disease Day.

Wow! Kion takes his first drive

Kion Leeson-Beevers enjoyed his first driving lesson - in a Blind Drive Experience thanks to Guide Dogs for the Blind.

Kion and his family went along to the Heritage Motor Centre in Gaydon, Warwickshire, for this exciting venture.

He drove a Nissan Micra and was guided by a qualified driving instructor.

"Kion did extremely well and really enjoyed the day.

"Cars have always been one of his great passions in life and he finds the thought of never being able to drive a car on the road very difficult.

"Scott and I also had a blind drive experience but Kion did far better than us!" says Mum Kerry.

As part of the day, Kion also had an off-road Land Rover Experience which was great fun!



This is a great adventure - Kion takes the wheel of a Nissan Micra

Important changes for families

ASUK has been closely involved with two particularly important issues that will directly impact on our families.

National Development Manager Kerry Leeson-Beevers has attended a number of consultation events concerning the new Children and Families Act which came into force in September.

Education, Health and Care (EHC) plans replace Statements of Educational Need and cover those

aged up to 25. With more emphasis on involving children, young people and parents and taking into account personal education, health and social care needs, it is hoped there will be a more co-ordinated approach and an improvement in the support and services available.

Each local authority will plan how it introduces changes between now and April 2018. We urge you to attend any consultation events in your area. For more information and support, contact Kerry on **01709 210151**.

Kerry also attended the European conference organised by Rare Disease UK. This was an opportunity to see how the UK Rare Strategy for Rare Diseases is progressing in England, Ireland, Scotland and Wales.

As the health system is developing differently in each devolved nation, work is ongoing to ensure they are closely linked and implemented effectively. For more information please contact Kerry on **01709 210151** or visit the website www.raredisease.org.uk

You can help to support research

ASUK needs your help - we need 15 followers to unlock Alström Syndrome on the Re(Act) Website which brings together researchers, patients and supporters to work together.

Please follow the link to register and follow Alström Syndrome to show your support for research. Here it is....

<http://react-community.org/diseases/1074>

The online RE(Act) Community is organised around four main axis dedicated to research on rare and orphan diseases: Learn, Meet, Share and Support.

Award marks 'fantastic achievement'

On behalf of ASUK, Office Manager Catherine Lewis was delighted to collect the Blue Shield Partnership Award in recognition of the partnership between South Devon Healthcare Trust and Alström Syndrome UK in setting up NHS highly-specialised multi-disciplinary clinics in 2006 and expanding research opportunities for people affected by AS. ASUK was the first charity to be funded as equal partners in this initiative. The organisers wanted to congratulate ASUK on all the fantastic achievements they have made and wanted to wish us well and continued progress now that the adult multi-disciplinary clinics have moved to the Queen Elizabeth Hospital in Birmingham.



Looking to a future w

Pioneering and exciting new developments and opportunities that could potentially change the lives of everyone living with AS formed the main themes at the ASUK Family and Professional Conference 2014.

Leading researchers and scientists led discussions at the conference which was chaired by Trustee Kez Hayat and ASUK National Development Manager Kerry Leeson-Beevers.

The conference took place at the historic and beautiful Brunel Manor in Torquay, Devon on 16th August.

Morning sessions were centred around current and future research projects. **Dr. Rob Cramb**, Consultant Endocrinologist at Queen Elizabeth Hospital in Birmingham, gave updates about the AS Clinics and current research projects in which families have been involved.

ASUK Founder and Director, **Kay Parkinson** then explained how potential funding from the Horizon project would work at a European level.

This was followed by **Hans Erik Frolander** who travelled all the way from Sweden to discuss the Theory of Mind research study and gave an insight into how the interdisciplinary teams in Sweden work.

The AGM which followed was chaired by ASUK Chair **Michelle Hough** who gave an overview of the charity's activities over the last year and gave the Trustees and Managers an opportunity to explain more about their roles.



Our picture above shows Massimiliano and Marco from Itex Technology, who travelled all the way from Italy, to learn more about the technology needs of our families as Marco (seen on the right) is a partner in Horizon 2020 grant application and he is responsible for developing M health applications.

Mark Millais, who has been an ASUK Trustee for many years, decided to step down from his role but he would like to remain involved in the annual ASUK bike ride which is currently being organised (see Back Page).

Mark has been a dedicated and committed Trustee, always putting the needs of our patients first and ASUK would like to thank him sincerely for his commitment and many years of service.

The afternoon sessions focused on family support and advancements.



Kerry Leeson-Beevers and Kez Hayat who chaired the conference

ASUK Family Support Manager, **Iram Shah** explained how ASUK can help and support families and her own personal story of living with the condition.

Marco Gregnanin, CEO Itex Technology, gave an overview of technology advancements and **Katie Barron** from Contact a Family gave an insight into how the new SEN reform will affect families across the UK.

This was then followed by Support and Advice sessions which gave a wonderful overview of organisations throughout the UK who are there to help.

The sessions were presented by **Jackie Ekers**, from the Citizens' Advice Bureau, who gave an overview of welfare and benefits; **Lorna Mackie** gave an overview of the services which Sense offer; **Michelle Woolf** showcased the services which Action on Hearing Loss offer families; and **Josh Allan** from the Calvert Trust, Exmoor, gave a presentation about their fabulous activity centre.

Great auction and a birth

The evening, which included the ever-anticipated charity auction, was a fabulous chance for families to get together and catch up.

This year we also ran a pop-up bar and asked attendees for donations for our great cause. The auction and bar raised an excellent £925.00.

Thank you to everyone for making our 2014 conference such a success. Thanks to the speakers who were excellent and gave very informative presentations and special thanks must go to those who travelled from Italy and Sweden to be with us.

Thank you also to Simmie Gould who gave massages to delegates throughout the day.

ASUK would like to say a HUGE THANK YOU to Brunel Manor in Torquay who provided complimentary accommodation for families to stay in Torquay and have a great time.

Most of all, thank you to our families who travelled to Torquay. We

Conference a from Swede



The Torquay conference at Brunel Manor gave families to meet up, relax, have fun



During the evening we also celebrated Rida's 9th birthday with a fabulous princess cake (see above).

have received very positive feedback and the ideas and contributions you gave us will be used to develop the services we offer and will help shape our five-year strategy.

Unfortunately, we did not have as many families and professionals as in previous years and we do accept that holding the conference in Torquay in

With fresh hope

attracts guests
from all over the world
to the UK and Italy



Manor offered a great chance for
to meet and develop friendships.



Friday treat

In the middle of summer may not have
been ideal.

The reason for this decision
was that we have seen a decline
in donations to the charity due to
the current economic climate and
we have needed to make some
savings. However, we do understand
how important the conference is
to our families and will take all your
suggestions on board when planning
our next one.

As part of this evaluation, we would
like to consult those who did not attend
this year to gather additional feedback.
A questionnaire will be circulated
shortly.

Our charity continues to grow from
strength to strength and we firmly
believe this is because we continue to
remain patient-led and focus on issues
that are important to those living with
Alström Syndrome.

Thanks again and we look forward
to seeing you all at our next conference!



**Ah - now this is
more like it!**

During the day, whilst their parents attended
the conference the children had a fun-filled
day out at Crealy Great Adventure Park.

The children joined in the exciting rides,
getting wet on the water slides and fun in the
indoor ball pools.

The children really enjoyed their
experience and were returned to their
parents exhausted after their exciting day.

A HUGE THANK YOU to all the volunteers
who helped on the day, without your support
the outing wouldn't have been able to take
place.

Following the Conference many families
stayed in Torquay for a fun-filled ASUK
Family Week.

Families had a great time exploring
South Devon including fun-filled days at the
beaches, cultural explorations and days out
at many of the tourist attractions including
Bygones in Babbacombe, Torquay, which
was a trip back in time including a life-size
Victorian Street. Thank you to Bygones who
enabled families to visit and explore their
wonderful attraction for free.

Why genetic counselling is so important

**Families and individuals are becoming
more aware of the genetic contribution
to conditions.**

Patients and relatives at risk of
conditions, which may be inherited,
are advised of the consequences of the
disorder, the probability of developing or
inheriting the condition and Information
about new developments that may help
families in their decision process.

Shagufta Khan is a Genetic Counsellor
at the Birmingham Women's Hospital,
West Midlands Regional Clinical Genetics
Dept and at the Birmingham Children's
Hospital.

She offers specialised genetic
counselling and support to families at
the Alström clinics in Birmingham and
throughout the West Midlands.

There are many friendly genetic
counsellors around the UK; families
can be referred by their GP or other
specialists. Some genetic counsellors
are also bilingual and can speak in
Urdu, Punjabi and Hindi as well as other
languages. An interpreter can also be
booked for your appointment.

Recent advances such as Pre-
implantation Genetic Diagnosis (PGD)
have assisted couples through specialist
NHS centres to have a baby that is not
affected by the family genetic condition.

PGD is not available for all genetic
conditions: the centre must have a licence
in place for the genetic condition in your
family and PGD can only be performed if
the gene alteration has been confirmed
in your family.

New study - my condition, my DNA

The Genetic Alliance UK are looking for patients
and their families who would be willing to take
part in a new study, My Condition, My DNA.

A new way of reading our genes means that
it is becoming faster and cheaper to sequence
large amounts of genetic information.

At the same time, researchers are finding
out more about how our genes can affect our
health and how we can use this information to
improve healthcare through better diagnosis
and treatment.

The study will use a series of interactive
sessions, where participants can take part in

the comfort of their own home, to find out what
patients and their families feel about the use of
genomic sequencing in the NHS.

The findings of this research will be fed
straight back to those making decisions about
how genomic sequencing could be used by the
NHS in the future in the form of a 'Patient and
Family Charter'.

If you have any questions about this project
then please feel free to contact Alice at alice@geneticalliance.org.uk or 020 7704 3141.

Huge thank-you everyone for helping us

Marathon runs bring rewards

Thousands of runners took to the streets of London in April for the Virgin London Marathon. This year we had three runners taking part and raising funds for our great cause,

It is still not too late to show your support through their Just Giving pages:

Lucy Leeson, whose nephew is affected by Alström Syndrome, has raised £1,410

www.justgiving.com/Lucylovespugs

Rik Bennett, who knows the Hough family in Cornwall (see story below) has also been holding many fundraising events in St. Agnes to raise funds totalling £1,800.

www.justgiving.com/Rik-Bennett

David Lunt who was running for a number of worthy causes including ASUK - his 20th Marathon - raised £750.00.

Luke Waiting whose uncle sadly passed away from Alström Syndrome, ran in the Greater Manchester Marathon on the 6th April and raised £162.00.

www.justgiving.com/Luke-Waiting1

If you are up for a challenge - and would like to raise funds for our great cause at the same time - why not join our running team. We still have places available in next year's BUPA London 10K and the Silverstone Marathon.

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 £10 each.

That's hair-raising!



Lorna Hythe from Starbeck primary school in Harrogate, Yorkshire, raised £510.00 from her Extreme Hair Cutting Challenge! – Look at how much hair she cut off!!

Sue Vasey continues to sell horse manure to her customers and has raised £120.00

The Manhattan Movers, a line dancing group also from Harrogate, have raised £595.00 to help families with AS.

The Lewis family continue to save 5ps and have raised £40.00 while the **Little family** have raised another £61.15.

In memory of **Safaa Manzoor**, her family have raised a further £190.00.

A special concert organised by the **Exeter Police and Community Choir** concert in Paignton, Devon, raised £506.10 and a local Rotary group gave £100, which was gratefully received.

Purity Dental in Birmingham have donated £200 from staff and customer collections - Thank you!

Mud challenge



Ready to go... Andrew Puttick (left) who raised £327.61 from an Extreme Tough Mudder Challenge!

Meet Millie's Jolly Giant

The story of seven-year-old Millie Hough and her family again raised great awareness of AS through an article in the Daily Mail.

The Daily Mail reported the severe effects of AS and how Millie, from St Agnes, Cornwall, was coping. Although Millie still attends school four days a week, and loves practising swimming and judo, her parents know that she will need an organ transplant in the future.

Her mother, Michelle, who is Chair of the Board of Trustees, said: "We try and do everything that you would do with a normal child and we appreciate every minute with her. She just brings so much to my life."

Michelle has created a cute and cuddly Giant Bolster - a legendary giant said to live near the cliffs of St Agnes and a popular character in village parades.

● You can now own your very own Limited Edition 25cm Giant Bolster priced £15 each.

All profits will support the Alström Syndrome UK Appeal. For more information or to place your order, please contact Michelle Hough on 01872 553067 or email michelle.hough@alstrom.org.uk



Millie and the Giant Bolster

How we keep on spreading the word

National Development Manager Kerry Leeson-Beevers and Family Support Manager Iram Shah have been promoting our great cause at a number of interesting events. Kerry attended:

Patient and Public Involvement in Research - to discuss the findings from the RAPPOR study. We were one of 22 case studies - findings shortly.

Navigating the NHS Conference - to develop our understanding of the new health system in the UK. Please get in touch with Kerry if you would like to receive a copy of the presentations.

Joining Together for Better Care - a discussion on the implementation of the UK Rare Disease Strategy. At the event Earl Howe, Parliamentary Under Secretary of State for Quality, Department of Health, launched the podcast of children living with rare conditions.

Kerry also attended the Findacure Scientific Workshop which highlighted the same optimism which ASUK have promoted for a long time that if we find treatments and cures for rare conditions such as AS, this may then benefit the wider population suffering from conditions such as diabetes, cardiomyopathy etc.



In May, Iram attended the 'Working Together for Change Forum'. Change is an independent group for parents and carers to voice their opinions on the SEN (Special Educational Needs) reform and cuts in education.

At the event, Iram met with the Deputy Mayor of Dudley Councillor Aston (above) - another great opportunity to raise awareness of Alström Syndrome.

Other events attended by Iram included the VIEW conference and the Disability Awareness day in Birmingham in March and at the Kidz in the Middle Exhibition in Coventry which showcased over 100 information stands and free seminars around disability, equipment and SEN.

If you would like more details - please contact Iram:

iram.shah@alstrom.org.uk



Finding time for fun at clinic

ASUK would like to say a HUGE thank you to all the families who have participated in the young adult questionnaire.

This enabled us to plan specific support at the young adult clinic in May and tailor workshops to everyone's needs.

The clinic was spread over four days to incorporate many support services and workshops to highlight both patient and carers' needs.

The clinics, normally run over two days at the Queen Elizabeth Hospital in Birmingham, were extended to four days to offer additional services and social activities. In addition to the usual tests, young adults had the

opportunity to share their experiences and discuss any areas of concern.

This was carried out in a safe environment and facilitated by Robin Paijmans, psychologist for the adult clinics.

We also offered parent workshops and fun activities such as goalball, bowling and a boot camp to show that exercise can be fun!

Young adults and their families enjoyed the chance to network with others and we hope to repeat this again in the future.

ASUK would like to thank our families, QEH staff and Goalball UK for making this such an enjoyable week!

You can pre-order your ASUK calendar today

You can now pre-order the ASUK 2015 calendar which will showcase your truly special photographs. Priced at £10 each - they will make great Christmas presents!

Cheques payable to Alstrom Syndrome UK can be sent to Catherine Lewis at 31 Shearwater Drive, Torquay, Devon TQ2 7TL. Alternatively please pay through our Just Giving page

www.justgiving.com/alstromsyndromeuk/

Our successful journey

ASUK produced a 'Journey' poster, which depicts how the charity started and our journey to start AS Europe and how lead Clinicians and Researchers now collaborate together to enable pioneering research.

The poster was chosen to be displayed at the European Conference on Rare Diseases & Orphan Products (ECRD) event in Berlin in May.

ASUK exhibits at Sight Village

ASUK exhibited at a recent Sight Village events in Birmingham and Leeds and will also be exhibiting on 4th November in London.

Remembering smiling Chloe



Chloe Cornwall sadly passed away in August. Here's a photo of Chloe with her Ipad which was funded by ASUK. We didn't know Chloe for long but she was an inspiration to us all, she was always smiling and singing and she will be truly missed by everyone.

Our next big bike ride

Planning for the second Alström Syndrome bike ride - *The Goodwin Cycle Ride* named after Kevin Goodwin - has begun.

Kevin who was an enthusiast for many activities, including the cycle ride in which he took part, died last year.

The first bike ride - from Torbay Hospital, Devon, to the Queen Elizabeth Hospital, Birmingham - was a great success and covered 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 and we are hoping to cycle in the summer of 2015.

We are aiming to raise money and awareness as well 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 will be greatly appreciated.

Please get in touch if you would like to take part - or 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
Curtis.vasey@alstrom.org.uk

Wonderful example

Continued from Page One

Luca Giacovazzi, who took part in the LSE study, has raised awareness of Alström Syndrome in the largest government hospital in Africa.

Luca's wife works as an Audiologist in South Africa where they celebrated Jeans for Genes Day. What a wonderful example of how ASUK is creating awareness worldwide!

The wide-ranging study itself highlighted the importance of the ASUK multi-disciplinary clinics in managing and more than halving treatment times, the support of the ASUK community in reducing emotional impact and explored the possible benefits of ehealth (through wireless networks).

For more go to: www.alstrom.org.uk



Jubilant scenes from the last big bike ride at the finish line in Birmingham.

The event held in 2012 raised over £16,000

Would YOU like to try a tandem?



It is always a good time to enjoy the great outdoors, so don't forget 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 acquire tandems and trikes and help you through ensuring the support is in place for you to get out and about safely.

For details, please get in touch with Catherine Lewis on Catherine. lewis@alstrom.org.uk or call 01803 613117

Fight for young people

RLSB's Youth Forum (RLSB empowers blind and partially-sighted children and young people to live life without limits) is made up of visually-impaired people aged between 16 and 25 years old.

It was set up to act as a voice for vision-impaired young people in London and the South East and to unearth potential solutions to challenges that visually-impaired people face, such as employment, transport and accessible technology.

During 2014, the RLSB Youth Forum will be making sure that the views of vision-impaired young people are heard by politicians, companies and decision-makers, through the RLSB manifesto.

This is the first manifesto representing such views and written by blind young people. Find out more information and pledge your support at <http://www.rlsb.org.uk/youthmanifesto>

Here are clinic dates for 2015

The clinic dates for AS children for 2015 are:

16th February
18th May
21st September
16th November

Dates for the adult clinics at the Queen Elizabeth Hospital, Birmingham, will be confirmed in the Christmas edition.

Should any dates be changed, everyone will be informed of alternative arrangements as soon as possible.