



Alström NEWS

With your help we have hope

NEWSLETTER OF THE ALSTRÖM SYNDROME UK SUPPORT GROUP

Winter 2012 Issue 11

Another story to inspire anyone

Liam is the perfect model

Liam Mackin has set a new marker for all those suffering from rare diseases – by becoming the positive role model for disability in the National Diversity Awards!

Liam gained his most prestigious award yet – and, in doing so, introduced a whole new influential audience to the great work being done by Alström Syndrome UK.

Liam's Mum, Liz Little, nominated him and accompanied him to the Midland Hotel in Manchester in late September for the black tie awards ceremony sponsored by The Diversity Group – one-stop shop that delivers a wide range of multi-media services to reassure and encourage people from the most disadvantaged areas to further their careers.

"In all, there were over 8,000 nominations over all the categories and Liam was in the final three for his. We really didn't think he stood much of a chance. In our typical fashion we got changed in a services toilet! We went in our scruffs and came out all dolled up!

"It was just like an award ceremony you see on TV and afterwards Liam did a bit of networking with a lady who works at an Asian newspaper, we enjoyed a three-course course meal, then it was award time.

"We were over the moon when Liam won, I walked him to the stage where he received his award from Jodie Cundy - paralympic star- then Brian Dowling (host of Big Brother) brought over the microphone for Liam to say a few words.

"Liam thanked Alström Syndrome UK, Living Paintings and The Children's Society for all their support.

"Ruth Brown (the Voice) then sang a few songs and we got home around 2.30 in the morning," explained Liz.

Liam had been nominated for the work he does for the three charities - work which includes street collections, training volunteers who want to work with children with disabilities, working with a group of young people with hidden disabilities to gain skills, confidence and independence and going round the country giving speeches.



Pictured above: Liam with the national diversity trophy presented by Paralympic star Jodie Cundy

**...and this is mobility
- the Mackin way**

Liam has set up his own sighted guide training course which comes highly recommended by Kerry Leeson-Beevers.

Training sessions are two hours long and can cater for between 1 and 8 people. For more information contact Liam on 07748 695900 or email lmackin@newcollegeworcester.co.uk or little_dean@sky.com

For more inspirational stories and to find out what the AS family have been up to - Just look at our stories inside

Alström Europe gets underway



Together at the first meeting of Alström Europe in Strasbourg, France - with, in the centre, ASUK Chief Executive Kay Parkinson and John Parkinson. Thirteen countries in the European Union signed up to start the ball rolling.

The first Alström Europe meeting took place in Strasbourg, France, on 27th September, organised by ASUK Chief Executive, Kay Parkinson.

ASUK funded a welcome dinner and then the first ASEU Directors' meeting was held on the 27th when Kay was given the honour of being the first President. The meeting was hosted by Prof. Helene Dollfus and her team. Thirteen countries have signed up to starting up Alström Europe.

"We hope the meeting in Strasbourg will generate a sharing of information on Alström Syndrome and provide the stimulus for joint

research ventures as well as providing more patients for potential clinical trials," says Kay.

"Alström Europe (ASEU) will comprise physicians and researchers who will collaborate on research projects for the benefit of patients. It will not be a patient support organisation - but will try to encourage patient support groups to start up in countries where there is no family support.

"ASEU is an independent charity consisting initially of two representatives from each European country. We aim to work collaboratively with ASI to avoid duplication of scarce resources. Our new organisation will

seek to complement the excellent work that has been undertaken by ASI."

ASEU will be able to access European grants which are not open to countries outside of the EU, but can, in some circumstances, include International partners - thus enabling ASEU to extend research capacity into Alström Syndrome.

The aim In the coming months is to develop a European research strategy which encompasses each country's strength, and jointly with ASI help develop a global plan.

- ASEU are delighted that American researcher Dr Jan Marshall has agreed to act as an advisor.

Fantastic news about Centre for Rare Diseases

As we go to press Queen Elizabeth Hospital Birmingham have confirmed that plans for a new Rare Disease Centre have received the green light.

This will raise the profile on rare diseases and provide a forum for fruitful discussion and co-ordinated initiatives and so the idea is no longer a distant dream.

ASUK held a meeting in August where the first steps to establish a centre which could encompass expertise and specialist support across all disciplines in one place were discussed.

We are trying to achieve something similar to Frambu, a National Resource

Centre for Rare Diseases, in Norway which looks holistically at all aspects of care and support for people affected by a rare disease. More info can be found here - www.frambu.no

If you have any ideas or suggestions about what you would like to see or want from such a centre - or would like to be involved in some way then please contact Kay Parkinson on email:

kay.parkinson@alstrom.org.uk

You can keep up-to-date with the progress through the monthly e-news. To subscribe contact catherine.lewis@alstrom.org.uk

Human spirit

"The Paralympic Games is about changing our perception of the world. We are all different; there is no such thing as standard or run-of-the-mill human being, but we share the same human spirit. What is important is that we have the ability to create.

This creativity can take many forms, from physical achievement to theoretical physics. However difficult life may seem, there is always something you can do and succeed at..."

- Quote from Professor Stephen Hawking

Cycle ride raises £16,000

The ASUK Cycle Team raised a fantastic sum of over £16,383.25 easily topping their £10,000 target for cycling 200 miles from Torquay to Birmingham.

"On behalf of ASUK I would like to thank the cycle team for taking on this challenge to help raise funds for adults living with Alström Syndrome," says Fundraising Co-ordinator Catherine Lewis.

"A big thank-you to everyone who took part and showed their support to this inspirational group – with particular thanks to Nina and Mark who supported the entire cycle ride. Without them it would not have been possible.

"Please rest assured that we will put the money raised to good use and will make sure we provide opportunities for people to have fun and new experiences.

"This event has not only raised a great amount of sponsorship but has also inspired others to participate in events, which they didn't think possible. You are all truly inspirational!"

Flying start

The cycle challenge began on 22nd June with patients with Alström Syndrome on tandems, their families and doctors from Torbay and Birmingham Hospital riding to raise funds for ASUK.

They pedalled 50 miles per day and reached Birmingham Queen Elizabeth Hospital on the 25th June - a great finish.

The cyclists taking part were: Kevin, Dave, Dan, Richard, Tarek, Kion, Scott, Alexi, Adam, Simmie, Rose, Matthew, Julie, Andreas, Rick, Arthur, Katherine, Christopher and Sarah.

The ride literally got off to a flying start – with a cheque for £250 from the Dartmoor Vale Rotary group to support the work of Alström Syndrome UK being presented at the start by president Ken Newall to Catherine.

Lord Clifford, of Ugbrooke House, Chudleigh, also gave £150. Our grateful thanks to them both.

Our pictures show the cycle challenge...from start to finish. A cheque from Dartmoor Vale Rotary sends the team on their way from Torbay Hospital - to the finish at the Queen Elizabeth Hospital, Birmingham



"This event has not only raised a great amount of sponsorship but has also inspired others to participate in events, which they didn't think possible."

Pictured right and proud after their fantastic ride - Kevin Goodwin, Kion Leeson-Beevers, Alexi Griffiths Rayson and Simmie Gould



It's a summer of fun,



It has been a fun-filled summer for many of our Alström family. The weather wasn't always great but the experiences were. A host of events have given our youngsters a chance to experience an exciting range of activities, providing fun, relaxation and confidence. All of our breaks and outings are only possible due to the continued efforts of our supporters so a HUGE thank you to everyone who has supported our great cause.



A group of young adults supported by John Parkinson had a fun-filled week at the Calvert Trust in Exmoor. They enjoyed many new activities including horse riding, canoeing, archery, sailing, abseiling and much more...

Calvert Trust Exmoor is an award-winning, fully accessible centre, located in an area of outstanding natural beauty on the edge of Exmoor National Park.



fun, fun all the way

families. The
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This year ASUK have organised many outings and holidays for families throughout the summer - including a trip to Snoezelen Sensory Centre for a very messy art workshop followed by a sensory session and a Harry Potter visit



Make a date for Children's Week

There was lots of fun at Children's Activity Week in Torbay - including a bat mobile experience!

Every year the Children's Week is organised for the last week in August. In 2013 ASUK would like to offer accommodation for families to come down to Devon to join in the fun.

If you would like to secure your place please contact Catherine Lewis on 07970 071675 or Catherine.lewis@alstrom.org.uk

Don't miss out on next year's fun from the 19th - 23rd August 2013. Book now to secure your place!



Flamingo Land!

A family trip to Flamingo Land, even though it was very wet, did not dampen the families' spirits on their fun day at the theme park. There are still half price tickets available if you would like to go please contact Kerry Leeson Beevers on 01709 210151 or kerry.leeson@alstrom.org.uk

Family Theatre Trips

ASUK are booking tickets to see the Christmas pantomime Cinderella, starring Billy Pearce at The Alhambra Theatre in Bradford. A number of families have already secured their place but we have room for more. Even if you don't live near Bradford we are organising theatre trips throughout the UK, please let Kerry know if you would like to be involved.



Fundraising CORNER

The Great Big Small Charity Car Draw

Alström Syndrome UK are taking part in The Great Big Small Charity Car Draw where you can sell raffle tickets to raise money for our great cause – and have a chance of winning a brand new Kia Picanto!

For every £2 raffle ticket sold ASUK receives £1.84! You can buy tickets online at www.smallcharitycardraw.org and you will find our information in the Participating Organisation section of the website.

Please contact Catherine Lewis

directly if you would like to sell paper tickets and she will get these in the post to you. For small charities, fundraising events can consume time and resources that would otherwise be directed to that charity's beneficiaries, yet fundraising income is much-needed to ensure the continued delivery of services.

To confront this problem the Foundation for Social Improvement (FSI) launched the The Great Big Small Charity Car Draw in 2009 to take the hard work

out of fundraising events and support charities right across the UK.

Thanks to the support of Kia Rotherham and the FSI trustees, the draw is back in 2012. It's a lottery for supporters of small charities (and the public) to buy tickets to support their cause with the chance to win a brand new Kia Picanto.

Get your friends, family and colleagues involved and raise money for ASUK, not to mention that you will be in with a chance of winning a stunning new Kia Picanto!

Helping hands

ASUK are now able to provide collection buckets for your fundraising events and also ASUK wristbands for sale at £1.00 each.

ASUK T-shirts are now 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.

The Plantation Olympic Rug raised £200 following charity auction on Ebay for small charity week.

Iram Shah raised £84.50 at a recent Asian Mela event for ASUK. Also her local dental surgery is going to display an ASUK collection bucket to raise funds for our great cause.



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Ready, get set for the new running season

The running season for marathons and half-marathons reaches a peak in the Spring of next year - so would you like to run for the charity?

At the time of going to press, we had two places available in the Virgin London Marathon - **21st April 2013** - and five places in the BUPA London 10K run - **27th May 2013** - and further five places in the Silverstone half marathon - **3rd March 2013**.

Please read on for further information about each event:

The **Virgin London Marathon** is the largest annual fundraising event on the planet - runners have raised over £500 million for good causes since the race began in 1981. If you'd like to get involved and run for ASUK we have two places available. Please contact Catherine Lewis on **07970 071675** or email:

catherine.lewis@alstrom.org.uk

The **BUPA London 10,000** will take place at 10.00am on Monday May 27th 2013. It starts and finishes in St

James's Park and uses Green Park as its assembly area.

The Race starts (and finishes) in The Mall and follows a clockwise route around the City of Westminster and the City of London.

Runners pass many of London's famous sights including, Admiralty Arch, Nelson's Column, St Paul's Cathedral, Mansion House, Bank of England, Leadenhall Market, The Monument, Millennium Bridge, Tate Gallery, Cleopatra's Needle, London Eye, Big Ben, Houses of Parliament and Westminster Abbey.

It is also hoped that Buckingham Palace will act as the backdrop for both the start and finish.

The **Adidas half marathon** takes place on 3rd March 2013, at the historic Silverstone motor racing circuit.

Whether you are attempting your first half marathon, or using this event as a training run for the Virgin London Marathon, this race could be for you.

Thank-you for every penny raised...

Alström Syndrome is a very rare genetic condition that leads to progressive blindness and deafness. It can also lead to diabetes, heart and kidney failure, liver dysfunction and associated problems.

Alström Syndrome UK support individuals affected, their families, carers and professionals and endeavour to raise awareness of this very rare condition.

Please get in contact with Catherine Lewis on 07970 071675 or email Catherine.lewis@alstrom.org.uk if you would like any help organising a fundraising event.

A huge **THANK YOU** to everyone who continues to support ASUK - our great cause. As a small charity, every penny raised does truly make a huge difference.

Rotary raises £1,000 with a little help

Dawlish Water Rotary Club in Devon will be handing a cheque for £1,000 to ASUK in the near future as they were the charity of choice for Club President Bruce Pell (who with Phil Pell designs this newsletter).

The money was raised at a special concert at The Langstone Cliff Hotel in Dawlish Warren with a Welsh male voice choir and through the Rotary club's charity pub quiz.



How to connect world-wide

The website is currently being developed and is looking really good with positive family stories and inspirational fundraising articles.

Please send in your articles or a note of up-and-coming events along with photographs and Catherine Lewis can put them onto the website. You can now check our events page for planned family outings and fundraising events.

There is also a BIG Lottery page which will be regularly updated so you can all keep track of the progress of this exciting research grant.

Please visit www.alstrom.org.uk to have a look around the new website.

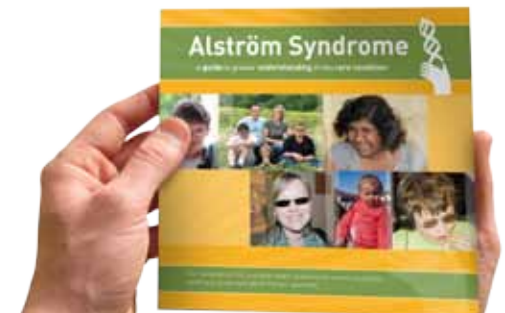
Alström Syndrome is now "live" as part of the EURORDIS/NORD Rare Connect project which links rare disease communities world-wide. The link is:

www.rareconnect.org/en/community/alstrom-syndrome

The website has interpretation facilities built in. The more people who use the site the more languages we will get interpreted.

Please ensure that your patients are aware that they can now connect with patients with Alström Syndrome world-wide. Professionals have the opportunity to submit papers and see also what other professionals have submitted.

RareConnect was created by EURORDIS (European Rare Disease Organisation) www.eurordis.org and NORD (National Organisation for Rare Disorders) www.rarediseases.org so that individuals and families affected by rare diseases can connect with each other, share vital experiences, and find helpful information and resources.



The Alström medical handbook, which has been well received in many quarters, is being revised and will contain some new sections - for example on research - and updated sections on the child and adults' clinics now held in Birmingham.

The handbook provides a valuable reference for health professionals and for families seeking more information about Alström Syndrome.

A flying visit to big Xmas party

When U.S. Army Air Forces first arrived at RAF Duxford, Cambridgeshire, in 1943, they decided to hold a Christmas party for 500 local children.

Duxford is now a museum, but 492nd Fighter Squadron still hosts a Christmas party for children with complex conditions and would like to invite families to a special Christmas Party on Saturday 15th December from 11am - 3pm.

There will be a special visit from Santa who will be arriving by helicopter! And there will be a vast array of presents, games and activities including a magic show and face painting. There will also be guided tours of the museum and food will be served during the day.

Please contact Catherine Lewis if you would like to attend this special event as places are limited.



The incredible story of ASUK

The incredible success of ASUK - from modest beginnings in a backroom in Kay Parkinson's house to a charity that has initiated major research and established a leading role in Europe for both Alström and rare diseases generally - was told in a large publicity poster displayed at European research conference this year.

Find out more in the Parent Workshop

ASUK's first Parent Workshop was a great success. Families discussed what additional information they feel they need about Alström Syndrome and we will be inviting professionals along to the next workshop to discuss this further. Please contact Iram Shah on 07988 237664 if your family would like to take part; the workshops are not only about learning more about Alström Syndrome but are a great opportunity where families can share experiences and learn from one another.

JESSICA: AN AMAZING STORY

After Jessica Rowlinson was found to have dilated cardiomyopathy, it took another four years for the wobbly eyes (nystagmus), heart condition and small size to combine into a diagnosis of Alström Syndrome.

Meanwhile, Alder Hey's Dr Ian Peart battled with a slowly deteriorating heart function and developing mitral valve leak. This got to the point where the valve had to be repaired. On Friday, January 22 2010 Jessica went for her pre-op assessment for having the valve repaired the following Wednesday. However, on Sunday January 24 disaster struck. Jessica had developed a

clot inside her heart which dislodged and caused her to have a large stroke.

This delayed the surgery while she was stabilised and had some rehabilitation; during this time she was put on warfarin.

Jessica had the valve repair surgery on 9th May. She then spent the next few months in Alder Hey being closely monitored. She was then discharged with weekly visits to clinic.

On one such visit in August, Dr Peart confirmed the worst of our expectations - the leaky valve had been providing a pressure release and now the heart was dilating rapidly and Jessica was readmitted to hospital. During this time she was taken to the Freeman Hospital in Newcastle, where she was assessed and accepted on to the heart transplant list. She then returned to Alder Hey to wait.

Her condition continued to deteriorate quickly, the decision was then made to transfer her to the Freeman Hospital to make use of mechanical support. On the 18th November, she underwent surgery to install the Berlin Heart device; while on this device the patient MUST be anticoagulated.

During this time Jessica had some severe problems with bleeding, she had

Doctors found she was awake and so began the journey to an astonishing recovery and heart transplant

a persistent nose bleed, which required multiple surgical interventions, and which "seemed like enough blood to fill a horse." During this time she also needed extra help breathing, this was when she needed a tracheostomy.

She continued this way until the 12th December when she then suffered cataclysmic stroke. Over the next few days in discussions with the doctors we came to the point where we felt all was lost. It was at this point that we called family and friends to her bedside, including Jessica's older sister Stephanie. After what was about to happen was explained to Stephanie, we went to visit Jessica. Following this we made preparations to turn off the Berlin Heart and the ventilator.



'Fairy princess' 3 months after transplant

Our daughter's fight for life by Dave and Karen Rowlinson

It was at this point that one of Jessica's doctors came in looking confused. He then said we need to rethink our course of action, when we asked why he told us "SHE'S AWAKE".

From this point Jessica made an "ASTONISHING" recovery. By the time the neurologist came to see her she was well enough to pull on his beard. We then made the decision with Jessica's doctors that to protect the brain to stop the heparin.

However this ran the risk of causing clots in the Berlin Heart.

Jessica was then put back on the active list for transplant, while all the staff watched hawkishly for any signs of blood clots. Seven days later we got the call we had been waiting for. We had a donor.

Jessica was prepared for surgery as we explained to her that the doctors were going to swap her poorly heart for a new one. Strangely while she was down in theatre, we felt the least stressed we had been for a long time, we supposed that with all the events of the previous week there was just no more stressed feelings to feel.

The transplant liaison staff kept us up to date with each stage of the operation and then with the all important news that all had gone well and she was back on intensive care Jessica made a rapid recovery and on the 31st December she had some neurosurgery to remove blood and clots from the brain. Once all the drains had been removed the difference in

Jessica was unbelievable. Where she was exhausted by watching TV 3 weeks before, she was now more than making up for it.

Now 22 months down the line Jessica is going from strength to strength and is flowering under the care of St Vincent's School for Blind and Partially Sighted Children.

We have regular checkups with both the team at Newcastle, where Jessica loves telling jokes with Terry (one of the transplant liaison nurses) and bending Julie's (the other liaison nurse) ear about anything, and with Alder Hey where we are never sure who is going to get the first tickle in - Jessica or Dr Peart.

It is safe to say that the hospitals still have their hands full with Jessica but in the nicest possible way....

However, if not for the expertise and determination of Dr Peart and his team at Alder Hey and Dr Kirk and Mr Massimo Griselli and their team at Freeman hospital we would not be here today.



STORY OF SURVIVAL

When she was just a baby...



Happy together... Jessica gets a hug from her older sister Stephanie

Jessica was born on the 16th September 2005. She seemed to be a healthy baby, although dad had noticed she had a slight wobble to her eyes.

When Jessica was 3 weeks old she started projectile vomiting and was diagnosed with pyloric stenosis (PIE). This was surgically repaired and all seemed fine afterwards.

In December 2005 Jessica suddenly became short of

breath, her breathing was weak and wheezy. She was rushed to her GP who took one look at her and phoned for an ambulance. She was then rushed to Whiston Hospital where she was diagnosed with suspected bronchiolitis put on a ventilator and transferred to the nearest ICU bed which was in Stoke-on-Trent.

Shortly before Christmas, Jessica was transferred to Alder Hey Children's Hospital and was put under the care of Dr Ian Peart. Here she was given a confirmed diagnosis of Dilated Cardiomyopathy.

Successful operation holds the promise of a better life...

The first successful heart transplant on little Jessica provides fresh hope for our Alström families with the promise of a better quality of life for the most severe cases.

It also means that Jessica's story has offered new experiences that will help the medical profession in planning, monitoring and preparing for such cases in the future.

We have had other success in terms of transplants too and a number

of patients are on waiting lists. See how Johanne Foster's life has changed for the better. Turn to page 10.

Transplants, of course, need to be carefully thought through with the expert multi-disciplinary teams at the Queen Elizabeth Hospital, Birmingham, so that all the conditions patients with Alström Syndrome have can be monitored closely.

Help us to plan your future...



Dear Families,

Welcome to my new column as Chair. Through the newsletter, I hope to keep in touch with you all regularly and I will be contactable by phone and email.

As a start of my new term as Chair I want to hear from YOU!

As a Board, we are starting work on our five-year strategy plan and cannot do this without hearing about the things that you all want and need from your charity. The Board will work on a questionnaire which will go out to you all through our family support workers who will be able to help you complete them if necessary.

The questions will be designed to help us find out what you want from Alström Syndrome UK, how you would like to help if you can and your thoughts as parents, carers and patients for the future. We also want to hear about anything you do not like and what you would like to change if you could.

We have new Board members joining established members who all gave up a weekend recently to see how Alström Syndrome UK will develop in the future.

The charity has grown considerably from its foundation in 1998. The main goal then was to establish multi-disciplinary clinics which all patients now enjoy. We now need to set new goals and keep ASUK at the forefront of research, care and support.

Please help us see the new vision through answering the questionnaire as best you can.

Best Wishes
Michelle Hough

michelle.hough@alstrom.org.uk
Tel: 01872 553067

'I feel extra close to my dad now,' says Johanne after transplant

Johanne Foster, who has AS, was struggling for years on a dialysis machine.

She was exhausted and everything centered around her treatment - but now she has her life back thanks to her dad, Paul Foster!

He offered to donate a kidney. "I feel extra close to my dad now," says Johanne.

"Kidney transplants are life transforming," says Paul. "Johanne used to live her life around dialysis. She had no energy- even holidays were planned around centres where there were facilities for transplants.

"Johanne's kidney transplant changed everything. It is a big emotional experience - live donor transplants between relatives can cause big conflicts but ultimately can bring everyone closer together."

Both Johanne and Paul are independent again and have regained their own lives back. Johanne still has AS but she is not ill now.

Johanne tells us: "I was very ill and tied to a dialysis machine for three years... Fiona Biggins at Preston Hospital told me about going on the kidney transplant list. She asked if



Johanne Foster now has much more energy and says she is living life to the full, after her father Paul donated a kidney

anyone would like to donate a kidney in my family.

"I said I do not know... I will hint. My brother offered to donate his kidney but it was not suitable. My dad had gone with him and so when my brother's kidney was not suitable my dad offered. I feel extra close to my dad now.

"I was grateful and happy that I had got a live donor but I was very scared going into theatre. When I came round I was very sleepy... Mr Campbell my surgeon said the kidney was "awake" when it was put in

which was a good sign. On the ward I had some initial rejection and had a biopsy and was treated for this.

"I was in hospital for two weeks following the 29th February - my 'leap year' transplantation operation date.

"I now feel I have much more energy, I am much happier and able to get out more, I have got my life back and I am living it to the full."

- *Johanne says she would be happy to talk to anyone else in the same situation as she knows the dilemmas only too well.*

The lessons in dealing with diabetes

All of the UK Alström team contributed to an article in the journal *Practical Diabetes* which is received by all medical, nurse and dietetic professionals involved in diabetes care in the UK, and many in Europe.

In *Practical Diabetes*; Volume 28, Issue 8, pages 340–343, October 2011), the article is entitled "Rare disorders presenting in the diabetic clinic: an example using audit of the NSCT adult Alström clinics". A summary is below:

"We describe the audit of 11 cases of Alström Syndrome diagnosed as adults, eight in the context of diabetes clinics who were referred to the National Specialised Commissioning Team (NSCT) adult Alström clinic at Torbay Hospital. All have severe insulin resistance, altered blood fats and a variable degree of cardiac, renal and musculoskeletal involvement

– features not associated with a unifying diagnosis until referred to their local diabetic clinics in eight of them.

"Obesity and young onset type 2 diabetes are increasing and it is important to be aware that some cases will have associated rare recessive conditions such as Alström Syndrome, Wolfram Syndrome, lipodystrophies, Bardet-Biedl Syndrome (LMBBS), Prader Willi Syndrome or occult cystic fibrosis.

"Early recognition of Alström families will facilitate prompt recognition and treatment of complications and genetic counselling."

Two other articles of interest from Canada and Italy have been published this year and concern development of diabetes during childhood and adolescence [1-2]. The progression from obesity to type 2 - Diabetes in Alström Syndrome.

Bettini V, Maffei P, and others: *Pediatric Diabetes*. 2012 Feb;13(1):59-67 and 2 - Mokashi and Cummings Presentation and course of diabetes in children with Alström Syndrome. (*Paediatric diabetes* 2011;12:270-275)

A few of you may have met Beth Cummings from Nova Scotia and many will remember Pietro Maffei from the 2011 family conference. Their two articles highlight that keeping body weight down and exercising do help prevent diabetes in Alström Syndrome.

- A highly-regarded reference source for rare genetic disorders is called *GeneReviews*. The section on Alström Syndrome has been compiled by Jan Marshall, Pietro Maffei, Cathy Carey, Tim Barrett and myself since 2008 and we update this every year.

By Dr Richard Paisey, Torbay

Community award for sporty Alexi



Alexi Griffiths Rayson has featured in a top community award ceremony for her part in the big cycle ride challenge (as featured on Page 3).

Alexi became runner-up in the Community Award in Sport which formed part of the Canterbury and District Sports Award event 2012 – that encourages organisations to participate in sport. It was held on October 26 in Herne Bay.

Many congratulations Alexi!

Cool Chris records his own brand of rock and blues on CD

Chris Lyon has written a song called "Nothing's Right" ...appropriately about what was going on in his life and what he was thinking about when he got his diagnosis of Alström Syndrome.

It is 60-70's rock style music which he has produced with three other friends. Chris, who plays bass guitar, is very proud of his music - especially as had a music teacher who did not agree with self - taught musicians!

Others in the group play guitar, keyboard and drums. The group is called "Bursting the Norse." Two projects have been rolled into one, one with six tracks and a cover project with eight tracks.

"Lazy Days" track is all about going with the flow... about times in life when you find things difficult to accept; the style is funky blues. The covers' project took 18 months and there is one more track to master.

The original tracks are still being worked on before the CD is cut and the cover should be ready by the New Year.

Chris is funding everything himself as he wants to keep his independence. He believes it is



not about being the biggest or the smallest but about keeping your own identity. Chris's music tutor is helping with the cutting of the CD, with recording and tracking now complete. They are now at the mastering stage, listening back and making sure they are happy with the results.

The cost for 100 copies will be £19 including artwork and copyright for Chris to produce his own CD.

You can find Chris on Facebook under **Chris Pythia Lyon** – UK melodic hard rock band from London. You can also listen to Chris on YouTube.

Our calendar will put you in the picture

ASUK has showcased your truly special and precious children to create a 2013 event calendar. To do this, we had sought your favourite, cutest and most precious photographs.

Winning photographs have been published in the calendar which is now on sale.

We have received so many lovely photographs - thank you to everyone who has contributed.

The calendar, which also incorporates charity event dates, is priced at £10. For orders contact Catherine Lewis on 07970 071675 / Catherine.lewis@alstrom.org.uk or use the order form here.

Thanks to the very kind support from our sponsors:

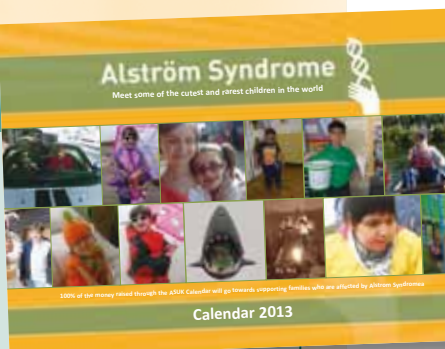
- New College Worcester
- RNIB Pears Centre, Hear First
- Frankham Consultancy Group Ltd

CALENDAR ORDER FORM

Name

Address

Tel No



No of copies

Total price (£10 each)

Please fill in your details and make a cheque payable to 'Alström Syndrome UK'. Send to Catherine Lewis, Alström Syndrome UK, 49 Southfield Avenue, Paignton, South Devon, TQ3 1LH



Brothers prove they are at the top of their game

It's another triumph for members of our AS family! Curtis and Taylor Vasey - with their friend Charlie Woodhead - won the national novice goalball tournament held at Nottingham University. Well done!

In the Goalball Domestic league, Curtis and Taylor and Charlie played all six games and ended up with eight points, two ahead of their nearest rivals.

They represented Hull outreach novice team which is made up of players from Scarborough.

Seven teams took part. They each played each other in a 'round robin' competition. Hull outreach team finished top winning five out of their six games.

This was only their second tournament after previously finishing fifth out of nine teams. All the details are on Goalball UK web site which also has details of all local clubs www.goalballuk.com

Goalball was originally developed as a rehabilitation programme for visually - impaired World War II veterans.

Within the next few decades it evolved into a competitive sport and



Goalball champions - brothers Curtis and Taylor Vasey with the help of their friend Charlie - celebrating after their victory in the national novice tournament, winning five of their six games

was officially included within the Paralympic movement in the 1980 Summer Games.

Goalball is a game played by two teams of three players with a maximum of three substitutions on each team. It is open to both male and female visually-impaired athletes, and sighted players can also play domestically. All players wear eyeshades so that they are totally

blindfolded. The indoor court has tactile markings and the ball contains internal bells, so the ball position can be located.

- British Blind Sport publish an events calendar which includes forthcoming sporting events, which people may be interested in joining. Contact details for each event can be found next to the event information on... www.britishblindsport.org.uk

Clinic dates for 2013 →

The 2013 Clinic dates for the Birmingham Children's Hospital and Queen Elizabeth Hospital in Birmingham are:

Children's Clinic

11th Feb
29th April
23rd Sept
11th Nov

Transition Clinic

10th and 11th April

Adult Clinics

9th and 10th Jan
12th and 13th June
11th and 12th Sept
20th and 21st Nov

Join the telephone conference

Catherine Lewis is inviting family members to join her on 30th November 2012 at 11am for an informal family telephone conference.

This is a great opportunity for our families from across the UK to come together to discuss their own experiences. There will be open discussions about any difficulties you are facing at the moment

and ways to tackle these. Share your experiences, learn from each other and help one another.

If you would like to take part, please contact Catherine on 07970 071675 or email Catherine.lewis@alstrom.org.uk

Families are given a number to call and a special code to use.

Looking to next year's conference

Next year's conference will take place at the Queen Elizabeth Hospital in Birmingham on Sunday 28th April 2013

It has been another exciting year for ASUK and the conference will be a good opportunity to learn about new developments and listen to talks from a variety of different people. There will be an update from the clinics and a presentation about the findings of the Big Lottery Medical and Scientific Research Project

and the Euro WABB Project, as well as a selection of workshops and a Medical and Scientific conference for medics, scientists and researchers. Download a copy of the full conference agenda nearer the time on our website at www.alstrom.org.uk If you would like any additional information about the conference or if you feel there are other topics we should be covering please contact Kerry on 01709 210151 / 07716135940 or kerry.leeson@alstrom.org.uk