**Alström Syndrome UK, the inspirational Paignton-based rare diseases charity, is to receive another accolade to mark its pioneering work in Europe.**

Founder and Chief Executive Kay Parkinson will receive the EURORDIS *Patient Organisation* Award 2013 at a special Gala Dinner in Brussels on February 26th – two days before Rare Disease Day on the 28th which aims to raise awareness with policy makers and the public of rare diseases and of their impact on the lives of patients.

“This is a great honour – and underlines all the achievements made by the whole Alström family - patients, families, staff and directors. It also strengthens our resolve to keep on fighting to gain more recognition for rare diseases in Europe and around the world,” says Mrs. Parkinson, who lost two children to Alström Syndrome.

EURORDIS, the European Organisation for Rare Diseases is a non-governmental patient-driven alliance of patient organisations and individuals dedicated to improving the quality of life of all people living with rare diseases in Europe.

The EURORDIS Patient Organisation Award recognises “extraordinary work and commitment to patient support and advocacy for people with Alström Syndrome, which serve to improve living conditions and inspire others working in the field of rare diseases.”

Nominations for the award were submitted by the EURORDIS member patient organisations, volunteers, staff and members of the EURORDIS Round Table of Companies. The final decision was made by the EURORDIS Board of Directors.

EURORDIS appreciates the dedication of the many volunteers, patient organisations, civil servants, policy-makers, researchers and entrepreneurs so passionate about this cause and says “ Together we are making a difference.”

The Award will be presented on February 26 at the second EURORDIS Black Pearl Gala Dinner, *Solidarity and Hope for Rare Disease Patients throughout Europe*.

Picture:

ASUK Director Kay Parkinson and acting CEO Kerry Leeson-Beevers travelled to Brussels to attend the EURORDIS Black Pearl Gala Dinner: Solidarity and Hope for Rare Disease Patients throughout Europe.

Whilst there Kay was awarded the EURORDIS Patient Organisation Award 2013. This prestigious award is given in recognition of extraordinary work and commitment to patient support and advocacy for people with Alstrom Syndrome.

The picture shows Kay collecting  the award from Avril Daly, EURORDIS Vice Chair.

* Kay Parkinson has also been instrumental in setting up Alström Europe and has been given the honour of being the first President. Thirteen countries have signed up to Alström Europe.

“We hope to 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 Mrs. Parkinson.

“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.”

Publicity….1..

Alstrom Syndrome UK secured half a page (Feb 2013) in the Independent supplement which went out on Rare Disease Day 28th February 2013 when the “official”announcement that Alstrom Syndrome UK have been awarded EURORDIS Patient Organisation of the Year was made.

We were allocated 600 words enabling us to also raise awareness of the Big Lottery grant that has helped AS UK gain experience of  research and produced a database and tissue resource.

Liam…1… (picture awaited here??)

Liam Mackin won another prestigious award for his outstanding work with charity The Children’s Society for helping to transform the lives of disadvantaged children.

The 18-year-old, from Shipley, Yorkshire, proudly received his award at the fifth annual ‘Over the Rainbow’ Awards, held in Gaydon, Warwickshire, where he goes to school.

The event, organised by The Children’s Society, formally recognises the outstanding work by children and young people who are involved with the charity.

Using money from The Children’s Society’s Pot of Gold scheme – where young people manage and allocate funding to other children and young people – Liam worked with The Children’s Society’s LEAP project in Leeds to raise awareness about disability. He has also been running life skill workshops for disabled young people and has set up his own company offering training to parents, carers and others who help the blind or partially-sighted.

Liam said: “Working with The Children’s Society has been an amazing experience and one which has allowed me to give something back to the people who have made such a difference to my life.” Last year he was presented with an award for Best Positive Role Model for Disabilities at the National Diversity Awards 2012.

A keen Scrabble player, he also won a charity competition in Shipley last year using braille letter tablets. Mathew Reed, Chief Executive at The Children’s Society, presented Liam with his latest prize and said: “Liam is an outstanding ambassador for the charity and should be extremely proud of what he has achieved.

“His tireless work is making a real difference to the lives of other children and young people. It was an honour to present Liam with his much-deserved award.”

**More to come on Sean and Kevin…..**

I have to send very sad news, on the 12th January 2013 Sean Waiting sadly passed away, Sean has actively been involved in the charity. In previous years he was an ASUK Director and Editor of the ASUK newsletter.

Nina Griffiths-Rayson, who was Alexi’s Grandmother had been battling cancer for some time, Nina sadly passed away with her loved ones around her. Nina had always supported ASUK and provided much needed support during the 2012 ASUK cycle ride.Our thoughts are with Sean and Nina’s family they both will be greatly missed.

Fundraising News

Luke Waiting whose uncle sadly passed away in January is going to run the Bupa 10K London Marathon in aid of ASUK please show your support and encouragement at [www.justgiving.com/Luke-Waiting](http://www.justgiving.com/Luke-Waiting)

**Re-structure of ASUK**

ASUK Founder and CEO, Kay Parkinson has worked tirelessly for families affected by Alström Syndrome and this is going to continue with Kay now forming exciting links in Europe.

Due to this increased workload, Kerry Leeson-Beevers was appointed ASUK Acting Chief Executive Officer from Monday 11th February 2013. This will enable Kay to provide more support to the new Board and take up many of the exciting UK and EU possibilities.

 *“I have every faith that Kerry will prove a dynamic and innovative acting CEO and continue to progress the charity. She will be able to keep a firm eye on patient’s needs and aspirations and develop new and interesting ideas together with our new Chair, Michelle.  I would like to be the first to congratulate her on this promotion.” – Kay Parkinson*

*To go with conference….*

Thank you to everyone who completed the ‘Patient Service Review’ questionnaire. We are currently compiling all of the information received and the ASUK Board of Directors will be meeting to discuss the findings. They will then feed back to you all at our Annual General Meeting which will take place at the conference.

**Jeans for Genes Award**

We have been successful in our grant application to Jeans for Genes!

They have awarded ASUK just over £12,000 to produce a DVD entitled ‘Living with  Alström Syndrome’. The DVD will be produced in a variety of different languages and will be a useful resource for all families, especially those who are newly diagnosed.

For more information about this project or to discuss any ideas you may have about content for the DVD please contact Kerry on 01709 210151 or email Kerry.leeson@alstrom.org.uk

Thank You Jeans for Genes! <http://www.jeansforgenesday.org/>

**Fundraising News**

Last year, through the dedication of many families and individuals affected by Alström Syndrome over £33,000 was raised for ASUK.

ASUK would like to say a HUGE THANK YOU to everyone who showed their support in 2012, ASUK needs this level of support to continue so we can maintain the high level of support and progression of the charity.

This year we have already raised over £1,000 from calendar sales and over £400 from the car draw raffle. Updating here??

ASUK still have places available in the Virgin London Marathon, the Bupa London 10K Marathon and the Silverstone Half-Marathon, you can raise money for ASUK and get fit at the same time!

**Family Adventures!**

Through your excellent fundraising events John Parkinson has been on a Tall Ship adventure, sailing from Malta to the coast of Sardinia and Sicily with two young adults... updates to follow.

ASUK are planning a week of fun from the 19th August 2013 in Torbay, we would like as many families as possible to join in, so please get in contact for further details.

Be sure to let your ASUK family support workers know what your wishes are....so they can help you plan exciting adventures.Don’t forget to keep up with the excellent fundraising so that our young people can seek new adventures and make fantastic memories.

**Optional item unless required….ASUK Calendar Sale**

If Santa didn't bring you a calendar, then meet some of the rarest children in the world in the ASUK 2013 calendar. Calendars are now at a reduced price of £5 each and can be ordered by contacting me and sending a cheque made payable to Alström Syndrome UK.  The full amount is donated to ASUK.

Thank you to a number of companies who have sponsored the calendar these include the; RNIB Pears Centre, New College Worcester, Hear First and Frankham Consultancy Group Ltd.

Please get in touch if you would like to know more about the sponsorship opportunities we currently offer.

**Pre-Implantation…1..**

The Medical Research Council is running a small project on pre-implantation genetic diagnosis (PGD).

The aim is to put more detailed information about PGD into the public domain, and ensure that the patient voice is central to this information.

PGD is a technique that enables couples with a particular inherited condition in their family to avoid passing it on to their children. PGD is used when one or both parents has a known genetic abnormality and diagnostic testing is performed on an embryo to determine if it has also inherited the abnormality.

### Background

One of rthe egular tasks at Genetic Alliance UK is to monitor the list of conditions that are awaiting a licensing decision at the Human Fertilisation and Embryology Authority and ensure that there is a statement from the perspective of patients submitted in time for the licence committee meeting. The committee must decide if the condition proposed for PGD is sufficiently serious to warrant a licence.

The aim of the statements is to describe the effect of the genetic condition under consideration on the family as a whole, including the inheritance pattern. Genetic Alliance UK have made more than 100 statements in support of PGD for individual conditions.

The statements would be useful evidence in making the case for innovative therapies such as PGD, and the emerging technology of mitochondrial replacement, to continue to be supported.

For more about inheritance patterns here: www.geneticalliance.org.uk/education2.htm

### The project

The next stage is to get in touch with the relevant support group. We will contact all those who worked with us to develop the statements and discuss whether they support the document’s publication. We will work with them to bring the statements up to the highest standard and ensure that the caveats placed on the cover of the documents are clear that these documents are related to PGD and that information about the relevant condition is best accessed through the individual patient support group.

You can contact me about this new project by phone or by email.

Nick Meade, Policy Analyst

nick@geneticalliance.org.uk, 020 7704 3141

Rare Disease update here

The Rare Disease Centre which once seemed a distant dream is now a step closer. The Queen Elizabeth Hospital Charity in Birmingham has agreed to start raising money for a Rare Disease Centre, which is great news! Please have a look in the next ASUK Newsletter and e-news updates which will have further information.

Conference round up to come – write up to do

**The ASUK Conference**

On the 28th April 2013 we held our family and professional conference at the Menzies Strathallan Hotel, Birmingham. Children who attended the conference had a fun filled day out at Drayton Manor theme park, thank you to all the volunteers who kindly gave their time to help on the day.

The conference was a great success and was filled with interesting research talks in the morning and family workshops in the afternoon. Families really appreciate the time together and they really enjoyed the presentation by award winning Liam Mackin who is affected by AS, many families thought his presentation was truly inspirational, well done Liam!

Thank you to everyone who joined in the charity auction and tombola, on the night we raised £1,034.00

Family News

During the ASUK conference many youngsters were given awards for their special achievements in 2012.

Maariyah received inspirational young person of the year award.

Ayisha received supportive sibling of the year award

Liam received special achievement award.

All cyclists received medals for taking part in the 2012 sponsored cycle ride which was from Torbay Hospital to the Queen Elizabeth Hospital in Birmingham.

Well done to everyone who continues to smile through adversity and thank you to everyone who continues to support ASUK.

We have organised lots of exciting trips throughout the summer, including the ‘summer of fun’ family week in Torbay. Please get in touch if you would like to take part in our events or if you have any suggestions for future trips.

 Update from Dan Jagger?

Ciliopathy Alliance Conference – ‘Speaking Up for Children with Ciliopathies’

The Ciliopathy Conference will take place from the 31st May – 2nd June 2013 at the Hilton Hotel in Northampton.

Through a Jeans for Genes grant the Ciliopathy Alliance have a limited number of funded family places. Please have a look on their website for more information

<http://www.ciliopathyalliance.org/>

Birmingham Retinitis Pigmentosa (RP) Awareness Day

On Thursday 30th May 2013 the charity RP Fighting Blindness are holding a one-day conference on retinal dystrophies aimed at people with RP, their families, and others that might want to learn about the impact of RP. The event is free of charge and includes a light lunch.  Topics covered will include the genetics clinic, genetic counselling and testing, medical research and social research, as well as a Q&A session. AS UK will also be having an information stand at this event, so come along and learn more about retinal dystrophies.

The event will take place at Council Chamber, Sandwell Council House, Freeth Street, Oldbury B69 3DE \*

**Family Service Review**

Thank you to everyone who participated in this year’s family service review; this is to ensure that ASUK are providing an excellent service and enable the charity to remain user-led.

If you didn’t get chance to complete the survey, you can download a copy on our website and you can also find the published results from the survey at <http://www.alstrom.org.uk/news/asuksurveyresults>

Please feel free to get in contact if you have any questions relating to the questionnaire or if you would like any help to complete the survey.

Social Media

Please keep up to date with Alström Syndrome UK via twitter @AS\_UK and also via our dedicated face book 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.

 Website Development

The website is currently being developed and is looking really good with positive family stories and inspirational fundraising articles. Please send me your articles or up and coming events along with photographs and I can put them onto the websitePlease visit [www.alstrom.org.uk](http://www.alstrom.org.uk/) to have a look around the new website

 *If you have any articles for our next e-newsletter or would like to register for any of the ASUK fundraising events, please contact me on 01803 613117 or email* *catherine.lewis@alstrom.org.uk*

*The deadline for the next issue is the 28th June 2013*

*………Draft @28.5.13*