



# Alström Syndrome UK

*With your help we have hope*

**Support Group**

[www.alstrom.org.uk](http://www.alstrom.org.uk)

Summer 2012 Issue 10

Charity No. 1071196 Company No. 3557191

## Rare, but stronger together...

**Rare Disease Day, February 29 was celebrated across the world by a joining of hands expressing the theme of solidarity "Rare but strong together."**

The huge 'show of hands' was supported by patients and patient groups, doctors and other health professionals in countries as far apart as the Ukraine, South Africa and America.

People of all ages with a wide-ranging number of rare diseases took the opportunity to briefly tell their stories using the internet and Flickr for photos.

Kay and John Parkinson were in Brussels for the EURORDIS gathering and Kay was one of the speakers at the event. Other speakers included Ms. Nessa Childers, Member of the European Parliament, Mrs. Sandra E. Roelofs, Honourable First Lady of Georgia, Ms. Paola Testori Coggi, Director General DG SANCO, Ms. Ruxandra Draghia Akli, Director Health, DG Research and Dr. Ségolène Aymé, Chair of EUCERD.

Rare Disease Day put the spotlight on "Solidarity" to focus on the importance and the need for collaboration and mutual support in an area where patients are rare, expertise is scarce, and people affected face similar challenges.

The theme is in line with efforts by the European Commission to harness collaboration in the field of rare diseases and promote rare diseases as a model of EU solidarity.

## Now we set our sights on Europe



*Joining hands... a worldwide show of solidarity. ASUK Chief Executive Kay Parkinson is seen (centre) at the Brussels meeting. Photo © Eurordis, Brussels Rare Disease Day Symposium 2012*

**Alström Syndrome is set to gain a higher profile across Europe under a new co-operative venture championed by ASUK Chief Executive Kay Parkinson.**

Professionals and patient support groups working with Alström families in France, Italy and the UK are getting together to set up Alström Europe.

The first steps were taken on April 10 following an arranged telephone conference to work out the details for establishing the new umbrella organisation.

"We would like to hear from anyone interested in joining the group in any capacity, either at Board level or simply as a member," says Kay.

The idea has already attracted funding from Eurordis, the European Organisation for Rare Diseases, which acts as the voice of rare disease patients in Europe.

Eurordis have a 'Rare Together' project which aims to help European

Rare Disease groups to set up their own European Federation. You can find more on their website: [www.eurordis.org/content/council-european-rare-disease-federations](http://www.eurordis.org/content/council-european-rare-disease-federations)

There was also a workshop on "How to create a European Rare Disease Federation" at the next Eurordis membership meeting held on May 23 in Brussels and there is an opportunity to join the Council of European Rare Disease Federations.

If you are interested in joining or would like more information then please contact Kay Parkinson on 01803 524238 [kay.parkinson@alstrom.org.uk](mailto:kay.parkinson@alstrom.org.uk) [www.alstrom.org.uk](http://www.alstrom.org.uk) / [www.euro-wabb.org](http://www.euro-wabb.org)

*For more information turn to the back page...*

### Conference success

*Alström Syndrome UK held another successful family and professional conference in March, in Birmingham.*

*More on the conference - centre pages...*

Inside your newsletter: Read about some fantastic fundraising including the big cycle challenge

# New Asian Mentoring Project

**The Asian Mentoring scheme is a new project for families affected by Alström Syndrome.**

The aim of the project is to work specifically with Asian communities to raise awareness of the condition and to support families to develop a greater understanding of the complex nature of Alström Syndrome.

Alström Syndrome UK has welcomed Iram Shah and Shamila Kafait who are both supporting Kerry Leeson Beevers with the Asian Mentoring project.

If you would like further information about the project or to request an Asian Mentoring leaflet please contact

Kerry on 01709 210151 or email [kerry.leeson@alstrom.org.uk](mailto:kerry.leeson@alstrom.org.uk)

If you would like to speak to someone in Urdu or Punjabi then please contact Iram Shah, Project Assistant on 07988 237664 or email [iram.shah@alstrom.org.uk](mailto:iram.shah@alstrom.org.uk)

## It's time for the big cycle challenge



**A cycle ride challenge from Torbay Hospital to Birmingham Queen Elizabeth Hospital in aid of ASUK was due to take place from June 22 – June 25 to raise vital funds and raise awareness of the multi-disciplinary specialist clinics which have moved from Torbay to Birmingham.**

Cyclists were set to cover 200 miles, around 50 miles per day, and included

patients on tandems, professionals and family members, please show the team your support at [www.justgiving.com/teams/ASUK](http://www.justgiving.com/teams/ASUK) or you can show your support through Just Text Giving.

All you need to do is text ASUK01 followed by the amount you wish to donate to 70070. For example ASUK01 £10.

Riders Alex Griffiths-Rayson (pictured right) and her partner Adam Line organised a pub quiz and charity raffle / auction at the White Hart pub in Canterbury, Kent, and raised an amazing £250.

It was a great evening filled with laughter and joy with a superb range of prizes such as books, DVD's, a food hamper, a scooter, MP3 player, iPod speakers. "A brilliant start to get our challenge on the way," says Alex.

The whole ride was self-funded, in order to raise as much money as possible for the charity – which means everyone

paid for their own travel to and from the ride, for accommodation, food and any entertainment along the way. To try and minimise these costs, riders were staying as much as possible in youth hostels.



## Liam scoops another top award

**Liam Mackin won the Worcester Young Citizen of the Year Award recently and was featured in an article on the Worcester Standard website.**

Liam's love for poetry is gaining him further notoriety as he was featured in Sense magazine talking about his passion for song writing and having his poetry published. He was also nominated in the category Young DeafBlind Person of the Year at Sense's awards ceremony late last year.



## Alstrom.org.uk update

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 website.

Email your news to [catherine.lewis@alstrom.org.uk](mailto:catherine.lewis@alstrom.org.uk) and please visit [www.alstrom.org.uk](http://www.alstrom.org.uk) for a look around the new website.



## Helping you to eat more healthily...

**ASUK would like to do more to encourage children and young people to eat healthily - and this could be done in a fun and creative way. So... what about growing your own vegetables?**

ASUK are offering vegetable and salad kits for you to grow at home with the whole family being involved. ASUK can also provide stands for you to use in case you do not have access to a garden.

Families can then photograph the end product for display in the next newsletter and a prize would be given for the biggest grown vegetable! Contact Catherine on [catherine.lewis@alstrom.org.uk](mailto:catherine.lewis@alstrom.org.uk) or 01803 613117 and start growing today!





# Fun-Filled Family Days Out

## **Drayton Manor Park** Saturday 24th March 2012

While parents attended the family and professional conference, we took 29 children off to Drayton Manor Theme Park for an exciting day out.

The weather was glorious and all the children had great fun exploring the theme park, zoo, Thomas land and having a go on the scary rides.

The children really enjoyed the day and were thoroughly exhausted when we returned to the hotel.

Thank you to Day Trippers who contributed £240 towards the trip and also a huge thank you to all the volunteers who attended. Without their help and support the trip wouldn't have been possible.



## **Bocketts Farm family trip**

On the 12th April 2012 two families had fun at Bocketts Farm near Surrey.

The weather wasn't great but that did not dampen our spirits. The children had great fun horse-riding, tractor rides, animal feeding and handling and holding more exotic creatures such as a snake, hedgehog and bearded dragon!

The children also got to cuddle a four day old lamb which was fantastic; it was lovely to see the children having fun and making friends.

Jane gives her thoughts about the day trip: "John-James and I would like to say thank you for a great day out today. We may not have got to see a lamb being born but got to hold a 4 day old one lamb and the horse riding which J-J thought they both made it a good day.

"Was lovely to see J-J and Hannah chatting away together with Catherine on places to go to."

We are planning more trips throughout the year so please keep an eye on the website events page for more information or contact Catherine Lewis on 01803 613117 or email [Catherine.lewis@alstrom.org.uk](mailto:Catherine.lewis@alstrom.org.uk)



## **Calvert Trust Exmoor residential break**

Friday 27th July 2012 - Monday 30th July 2012.

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, next to Wistlandpound Reservoir and a short distance from the North Devon Coast.

All activities are on site and specifically designed and equipped to enable people with physical, sensory and learning disabilities of all ages and levels of ability, together with their families & friends, to experience exciting, challenging and enjoyable outdoor activity adventures.

The activities they provide include; Sailing, canoeing, kayaking, horse-riding, archery, climbing, challenge courses, abseiling, zip-wire, giant swing, accessible cycling and much much more... They also have a swimming pool, jacuzzi, games room and sensory room. All activities, meals and accommodation will be funded by Alström Syndrome UK. There is still availability.

**The Bendrigg Trust** is based near the Lake District and the Yorkshire Dales. It is a residential outdoor activity centre for children with disabilities.

Each group is given a programme that is specific and tailored for them. They have a separate lodge which is available from the 27th until the 29th July 2012

They do outdoor activities and watersports as well as social evenings. For both of these trips all activities, accommodation and meals will be funded by ASUK.

## **Independent Living Workshops - Beginning of the Summer holidays at New College Worcester.**

All children and young people are invited to take part in the day sessions which will be focused on building confidence through independent living skills and finding solutions to using everyday equipment as well as adapting the home environment. A fully qualified rehabilitation officer and experienced support staff from New College Worcester will be available to help families throughout the day. Accommodation can be provided for families outside of the area.

## **Birmingham Sea Life Centre family trip**

On the 14th February 2012 six families including sixteen children attended a fun outing to Birmingham Sea Life Centre.

The families enjoyed learning about the different species of sea life and got to hold a crab and star fish. They also got wet when they experienced a 4D Movie of Happy Feet.

The children had great fun exploring the centre and running around the soft play area. It was great to see the families getting out and having fun!

I am organising activity breaks for both adults and young people throughout the year so please feel free to contact me if you would like to attend an activity break or if you have any ideas or places you would like to go to.

Contact Catherine Lewis on 01803 613117 or email [catherine.lewis@alstrom.org.uk](mailto:catherine.lewis@alstrom.org.uk)

# 14th Alström Family Conference 2012

Alström Syndrome UK held their family and professional conference on the 24th March 2012 in Birmingham.

This was a great opportunity for everyone to meet the team from the Queen Elizabeth Hospital where the adult clinics are now based and also have a tour of the Hospital.

The conference consisted of many interesting talks and information stands. In the evening there was the much anticipated dinner and auction. Thank you to everyone who brought items to donate and ASUK raised £1,532.70 which is excellent.



## Round-up of other useful topics

**Kay Parkinson, CEO and founder of Alström Syndrome UK** spoke about the difficulties of finding a diagnosis for her two children, Matthew and Charlotte.

She gave a parent's perspective on living with Alström Syndrome and then looked back at how the charity has evolved over the years.

**Professor Tim Barrett**, Project Leader, discussed the Euro-WABB Project and the aim to support efficient diagnosis, treatment and research into three specific conditions Wolfram, Alström and Bardet Biedl. Alström Syndrome UK is a partner in the project.

**Alison Shaw**, Senior Research Fellow in Social Anthropology, discussed genetic and cultural factors effecting people with rare conditions. She discussed the sensitive issue of consanguinity and how the myths surrounding this can impact on people's understanding.

**Nick Meade**, Policy Analyst, discussed the work of Genetic Alliance UK and their role in supporting families wishing to have pre-implantation genetic diagnosis. There is currently no licence for this procedure for Alström Syndrome but once a family decides this may be an option for them, they will be fully supported by Genetic Alliance UK and Alström Syndrome UK.

**Tarek Hiwot**, Adult Clinical Lead, introduced the new Alström team at Queen Elizabeth Hospital. The team is very

enthusiastic about working with us and the 'state of the art' hospital will hopefully provide access to additional specialists to benefit our patients.

**Marie McGee**, Transition Coordinator discussed her work at Birmingham Children's Hospital in developing transition plans. Marie's involvement will hopefully ensure that our young people are fully prepared for their transition to adult care.

The conference delegates then divided into two groups:

**Professor Barrett** and the team from the children's hospital discussed the Paediatric clinics and families gave feedback.

He discussed the additional tests that have been carried out recently including 24-hour blood pressure monitoring and early morning urine tests.

Professor Barrett plans to audit these results and feed the findings back to families. He also gave details of the new psychologist who has recently joined the team.

**Hazel Riggall**, Dietitian, spoke about sweeteners.

**Dr Hiwot** and **Dr Richard Paisey** discussed the clinics and chaired a question-and-answer session between patients, families and staff at QEH. Although there may be a little anxiety about the move, everyone appeared

positive. Families and staff of Alström Syndrome UK are extremely grateful to the team from Torbay Hospital for their support in the development of our clinics and for the specialised care they have given to our patients.

**Jackie Elshaw**, Engagement Officer, discussed the benefits of being a Guide Dog owner. She also informed everyone that the Buddy Dog Scheme is no longer a pilot project and that everyone will be considered for a Buddy Dog on an individual basis.

**Ray Perry**, Benefits Advisor, discussed the proposed changes to the benefits system and how this may impact on our families. There will be a great deal of uncertainty, change and re-assessment over the next few years.

We would also like to thank the exhibitors from Humanware, St Vincent's School in Liverpool and Royal National College for the Blind in Hereford, Guide Dogs, Euro-WABB and Marie McGee.

Plans are already in progress for next year's conference so we look forward to seeing you all there!

By Kerry Leeson-Beevers



# Hope for speedy diagnoses

**The wide-ranging Medical/Scientific discussions at the Family Conference centred on the latest information and developments in Alström and other rare diseases.**

Many encouraging signs were debated including further advances in genetic science that could speed up diagnoses.

The first talk was given by Richard Paisey from Torbay Hospital. Richard told us about the metabolic complications of Alström Syndrome, particularly about the problems associated with obesity and diabetes.

He also discussed how different diets, exercise, and possible surgical interventions could help with these issues.

Cathy Carey, also from Torbay Hospital, continued by reviewing her personal experiences of studying the heart in Alström Syndrome. This led to a discussion of the advantages of new technologies that are available at Birmingham, and the best methods for heart monitoring in patients over long periods.

Tim Barrett from Birmingham Children's Hospital gave a review of how the Syndrome affects children, and of how there is a huge variability in the symptoms displayed by patients.

In the second half of the conference talks were given by guest speakers. Alison Shaw from Oxford University expanded on her talk from the morning session about the impact of consanguineous (blood relation) marriages on the occurrence of rare genetic diseases.

Colin Johnson from the University of Leeds, an expert in molecular medicine, reviewed his lab's work in various rare diseases. He focuses particularly on Meckel-Gruber Syndrome, another ciliopathy, and so the genes he studies are likely to be very relevant in the search for a better understanding of Alström Syndrome.

Colin highlighted new developments in genetic science that he believes will revolutionise the way we think about rare

genetic syndromes, and will remarkably speed up the way we can analyse genes and reach correct diagnosis for patients.

There was unanimous agreement that the afternoon had been very helpful to everyone in gaining a better understanding of the Syndrome, and that there should be regular get-togethers for the sharing of ideas and to plan further research. On behalf of ASUK I would like to thank everyone who attended the conference, particularly the speakers for their excellent talks.

*Dr Dan Jagger, Senior Lecturer,  
University College London*

For a more in depth look at individual speakers' presentations go to [www.alstrom.org.uk/conferences/conference-2012](http://www.alstrom.org.uk/conferences/conference-2012)

## Your comments continue to help us to improve our service to you

**Thank you to everyone who took the time to fill in an evaluation form at the conference.**

We really do value your opinions and each year we aim to ensure that the conference is designed around what people with Alström Syndrome and their families need.

The forms were split into five sections covering the hotel, the conference, the charity, information and the clinics. Here is a summary of responses:

### **The Hotel**

Views on the Menzies Strathallan in Birmingham were very positive, with comments on the helpful staff and healthy food available.

**On the negative side** - Small rooms, no pool or sauna, not enough food choices.

### **The Conference**

The general feeling was that the conference was a great success and enjoyed by all.

**Positives included** - very well organised, new information and updates, networking opportunities, and an address on the impact of consanguinity and some of the myths surrounding it.

**Negatives included** - lack of clarity on path from research to clinical benefits, no update from Rob Semple and no opportunity to write down their questions for speakers.

### **Recommendations for the next conference:**

- More practical speakers
- Advice on issues around education and the impact of multi-sensory impairment
- Talk from a respiratory specialist
- Research updates, the development of any new treatments and more information on stem cell research
- A conference over two days
- Self-help groups to cope with depression
- Counselling for parents and patients
- Sighted Guide Training
- Practical sessions with Guide Dogs

### **The Charity**

**Only positive feedback** - a feeling that the charity was fantastic with excellent support. And much appreciated. There was a feeling that charity could focus on how research can lead to treatments and promote family outings where children get to spend time together.

However if there are areas you think we could improve on then please contact us at any time.

### **Information**

ASUK has received the Quality Assurance Kite Mark for the information we produce and we will constantly be reviewing all of our materials so thank you for your input. This includes material that is easy to understand, productions in Braille and Audio, more use of social media and networking sites.

### **The Clinics**

Our families appear very satisfied with the clinics - "long days but necessary"- with appreciation for the "continuity of consultants." There were also comments about possible access to counselling via phone or Skype, holistic care and the desirability of additional professionals to cover for sickness and holidays.

If you have any questions about the evaluation forms then please do not hesitate to contact me. We will continue to use the full document throughout the year to help us to improve the service we offer.

*By Kerry Leeson-Beevers*

[Kerry.leeson@alstrom.org.uk](mailto:Kerry.leeson@alstrom.org.uk)  
01709 210151 / 07716135940

# Move to 'super hospital' - a new milestone for Alström patients



*The team at the Queen Elizabeth Hospital, Birmingham*



## **A new milestone was reached when Alström adults became a recognised patient group at the state-of-the-art Queen Elizabeth Hospital, Birmingham (QEHB).**

In May the first adult clinics were held in Birmingham. The services patients will receive include:

- 24-hour blood pressure monitors fitted
- Bloods will still be done by phlebotomist Carol Wardle from Torbay Hospital
- Cardiology
- Audiology
- Ophthalmology
- Echo Cardiogram
- Consultation with dietician and diabetes advice
- MRI scans
- Physiotherapy session and exercise, healthy-eating advice
- Hepatic ultrasound and partial vein dopplers

Torbay Hospital will continue to provide some services and liaison over the next year or so. Dr. Richard Paisey and Kath Williams (dietician) from Torbay will jointly see patients with Dr. Tarek Hiwot, Sarah Boocock (dietician) and Jayne Hodgkiss (Diabetic Nurse Lead) for their consultation.

The first adult clinics followed an earlier transition session for those young people ready to move up into the adult clinics.

The new Queen Elizabeth Hospital boasts a purpose-built environment and the latest technological advances with services ranging from a single outpatient appointment to a heart transplant.

The Trust is a regional centre for cancer, trauma, neurosciences, burns and plastics, and has the largest solid organ transplantation programme in Europe.

It is internationally recognised for the quality of its specialist medical and surgical training and has forged relationships with hospitals in many different countries to provide training and share clinical expertise. Many of the world's liver transplant programmes are headed up by doctors who have been trained there.

The hospital is designed specifically around the needs of patients and visitors, making coming to hospital easier and more pleasant. For example, there are dedicated patient and visitor lifts.

The design also greatly improves the ability to prevent infection, as it gives staff greater ability to protect or isolate patients who have a higher risk of picking up a bug or passing one on to others.

Wards have clinical handwash stations at their entrances to highlight the need to wash hands before entering and leaving.

The hospital is kitted out with the latest medical advances available to the NHS. This means the latest diagnostic scanners, new theatre equipment and even robots deployed in the pharmacy and laboratories to make drug dispensing and testing faster and safer.

The Queen Elizabeth is one of the leading clinical research institutions in the country, being adjacent to the School of Medicine at the University of Birmingham and is a regional and national centre for specialist clinical services.

With the children's clinics being held at the Birmingham Children's Hospital, there is greater scope for closer links and a more seamless transition for children moving up to the adult clinics.

The Plaza Restaurant is located on Level 2 of the hospital and is open seven days a week from 7.30am to 8pm serving hot and cold drinks, snacks, confectionery, sandwiches, hot meals and cooked breakfasts. The Plaza Restaurant also operates a trolley of snacks, sandwiches and drinks, which goes around the Outpatients' department.

There are approximately 3,000 car parking space on site for staff, patients and visitors. There are 240 free disabled spaces with 33 of these in open-air car parks located within close proximity to the Main Entrance at the Queen Elizabeth Hospital Birmingham.

The nearest car park for QEHB is car park A. The drop-off spaces outside the hospital may be used by disabled drivers of high-top vehicles if they are required and car parking attendants are on hand to assist patients and visitors. Disabled parking is free in designated disabled areas.

All routes inside and outside the hospital are suitable for wheelchair use. Wheelchairs are available in the Main Atrium Entrance. Support is on hand for anyone who requires assistance with their wheelchair.



# Fundraising CORNER

## London Marathon success

On Sunday 22nd April, Alström Syndrome UK had two very courageous runners, **Nichola Martland and Stacey Chaplin**, who took part in the Virgin London Marathon.

Combined they raised almost £6,000 for us. Many congratulations to Nichola and Stacey for taking on the challenge in aid of ASUK. Taking part in another race, the London Bupa 10K run, was Amy Farmer (pictured right) who raised £150. Runner David Lunt has raised a magnificent £1,000 for Alström Syndrome UK, which is great news!



**Nichola Martland did the Yorkshire Three Peaks Challenge in aid of Alström Syndrome UK.** Thank you to everyone who showed their support, she raised £470.

**Laura Martland did a sponsored walk of 8.6 miles to raise vital funds for Alström Syndrome UK on 25th February 2012.**

Through her JustGiving website donation page Laura managed to raise £155. Well done Laura!

The two sisters have also both been doing lots of fundraising including sponsored events, pub quizzes, auctions and even waxing a poor friend's chest! All for the great cause of Alström Syndrome.



## School Fundraising

**Millie Hough's school in Cornwall has raised a magnificent £414.56** (including Gift Aid) in donations for ASUK. A collection was taken at the Nativity performances at St. Agnes School. We are grateful to all who contributed and touched by the support of this small school.



## A Wizard Collection it's Liam again

**Liam Mackin and his Step-Dad dressed up as the tin man and lion on Christmas Eve to do a street collection in aid of ASUK.** In just a couple of hours they had raised £136.09 – Well done Liam!

If you are interested in doing a street collection in aid of ASUK please contact your local council as you will need a street permit. These are straightforward to get, but can take around six weeks.

## A dinner date

**Raise money for ASUK by doing something you enjoy – having friends around for dinner!**

Dinner4Good take care of everything, including the Gift Aid application, so



all you need to do is eat, drink and enjoy yourselves. Over dinner you can then tell your friends about ASUK and they may want to hold their own Dinner4Good of their own – so the good goes on!

Please visit our Dinner4Good web-page to see how easy it is to host a dinner party and raise money for our great cause at the same time [www.dinner4good.com/Alstrom](http://www.dinner4good.com/Alstrom)



# New appointments for ASUK Board

**Michelle Hough**, well known as **delightful Millie's mother**, to be the **new Chair of ASUK**.

"I am very excited at the prospect of working with Michelle and think that the added kudos of being Chair will add considerably to her already proven fundraising skills" says Chief Executive Kay Parkinson.

**Lesley Porter** will be deputy chair and help with administration, employment and transition issues. She has already been a big help with providing templates for management guidelines, grievance procedure and other documents based on NHS guidelines.

**Kevin Goodwin** will act as spokesman if we need someone out and about to represent the charity. "I am very proud of the way he always puts the charity interests first- he will make an excellent ambassador," says Kay.

New Director, **Liz Little** will take on education- helping with tribunals, appeals etc. as well as getting feedback from



*Pictured above: New Chair Michelle Hough with her daughter Millie and new Director Curtis Vasey*

families about clinics and other issues which may develop.

New Director, **Curtis Vasey** will be mentored by **Alexi Griffiths Rayson** and help with the young adults' activities.

**Mark Millais** will continue to develop his research into home adaptations and help with any building projects.

**Jane Entwistle** is resigning having served the Board faithfully for around 8 years and we will miss her very much.

"We have an excellent Board who all give of their time freely and I appreciate their help, support and dedication very much indeed. I think we will have an exciting future together and will really make a difference to our patients' care and treatment, which is what we are all about," says Kay.

- The next Directors and staff meeting will be on Sunday 21st October 2012 at the Menzies Strathallen, Birmingham.

## New roles to boost position in Europe

*[continued from page 1]*

**Important changes are being proposed as ASUK develops its role in the UK and Europe.**

Chief Executive Kay Parkinson has proposed that ASUK appoints a Scientific Chair and a Medical Chair - heading two Boards that could arrange to meet at least once a year together.

Dr Rob Semple from Cambridge University and a partner in the Big

Lottery research project, has agreed to become ASUK's Scientific Chair - a non-executive position.

Dr. Tarekegn Hiwot, Consultant in Inherited Metabolic Disorders in Endocrinology at the University Hospitals' Birmingham NHS Foundation Trust will Chair the ASUK Medical Board.

Dr Richard Paisey from Torbay Hospital and Dr Hiwot are the UK

representatives and Directors of Alström Europe.

An Alström Europe meeting is being arranged for 27th September in Strasbourg for Directors only - so far nine countries have agreed to join.

Kay, accompanied by John, chaired a session in Brussels on the EU Cross Border Directive and also spoke on the session of patient involvement in clinical management.

## Working together for a positive future

**Advances in our understanding of Alström Syndrome and other rare diseases may happen faster if scientists and clinicians interested in all the associated conditions work together, to share information and plan joint research.**

The Cilia 2012 1st International Conference on "Cilia in Development & Disease", organised in May and supported by the Ciliopathy Alliance ([www.ciliopathyalliance.org](http://www.ciliopathyalliance.org)) and many other organisations did just that by attracting 300 delegates including scientists and clinicians from 20 countries.

There are big gaps in our understanding about what causes Alström Syndrome. The best held theory is that mutations in the ALMS1 gene affect the function of a small structure called the "cilium", which is an important part of many of the cells in the body (for more detail go to: [www.alstrom.org.uk/scientific-info](http://www.alstrom.org.uk/scientific-info)).

Professor Joe Gleeson told delegates how his group in California have been developing ciliopathy therapies based on their findings in the lab. Delegates also got the chance to meet ciliopathy patients and the support groups based in the UK.

There were a total of 40 talks and 140 poster presentations, making it a very hectic and information packed schedule! Feedback from participants has been very positive.

*By Dr Dan Jagger, University College London*

