Alström Syndrome UK
Strength for today, hope for the future

Alström Syndrome UK (ASUK) is an award winning charity who provide information, support and advice for individuals affected, their families, carers and professionals. Alström Syndrome is a very rare genetic condition which can cause progressive blindness, hearing loss and can lead to; heart and kidney failure, type 2 diabetes, liver dysfunction and associated problems. The symptoms arise at different stages making diagnosis difficult. We endeavour to raise awareness, conduct pioneering research and enable better treatments and monitoring through the AS multi-disciplinary NHS clinics.
Further information about our work can be found at **www.alstrom.org.uk**

A huge thank you and a very

Merry Christmas!

to our friends and supporters

A heartfelt thank you to all our friends and supporters who have worked tirelessly in 2016. As many of you know this is a difficult time for many charities and we have worked hard to ensure we continue to offer a high level of support for families; at clinic and on a more personal level at home, in schools and colleges and in the community.

We have supported individuals who have generously given their time to take part in the clinical trial and worked with others to raise awareness of Alström Syndrome.

We have organised fun weekends away for families and continued to ensure families have access to equipment and financial support. To all our supporters; families, clinicians, researchers, volunteers, the team at ASUK and all, an enormous thank you for your continued support and the valuable contribution that you make.

In 2017 we will be recruiting more family support staff to ensure that individuals and families can be supported wherever they live in the UK. We will continue to encourage research and we are hopeful that real progress can be made in finding potential new treatments and discovering more about Alström Syndrome for the benefit of this and future generations. On behalf of us all at ASUK I would like to wish you all a Merry Christmas and a very happy and healthy New Year.

Ann Chivers, Chief Executive at ASUK

Our vibrant new logo is revealed!

Firstly, a huge thank you to everyone who completed questionnaires about the new logo and strap line and gave feedback during face to face meetings. We received lots of responses and suggestions about the colours, the look and the image we were trying to project. Kerry and Marie led a consultation with families who came up with a lovely positive strap line, Jane asked everyone she met to comment on the logo and Catherine analysed lots of feedback.

Alström Syndrome UK
Strength for today, hope for the future

This new logo and strap line represents ASUK: promoting fresh and innovative ways of working that empowers individuals to reach their potential and supports families on that journey. It is vibrant and captures a picture of working together and a pictorial representation that nods to the genetic source of Alström Syndrome. The use of 'happy colours' can be used in a variety of ways throughout our promotional materials and we are sure it will stand out on our website.

The logo is made up of 3 'figures' each in a different colour with 2 tones of each colour. The middle figure is green, a grass green and a lighter green tone, the figure to the right is 2 shades of orange, the deeper colour is a burnt orange, the figure to the left is 2 tones of blue, sky blue and the other shape is a lighter blue. The umlauts are green, the same colour as the middle figure. The lettering is black and bold.

Orange is the colour of innovation, modern thinking and health, green is often used to reflect ethical credentials, organic growth and nature and blue is often used to imply professionalism, integrity and tranquillity. The overall effect is bright and colourful, it speaks of boldness and strength.

We hope you all love the logo as much as we do and we hope it makes you all smile!

First Clinical Trial for AS in the UK

In December 2015, The Queen Elizabeth Hospital, Birmingham (QEH) opened as a clinical trial site for the first clinical trial for people affected by Alström Syndrome in the UK. Working in partnership with Alström Syndrome UK (ASUK) and the Canadian Pharmaceutical Company ProMetic Bio therapeutics Ltd, Dr. Tarek Hiwot, Consultant in Inherited Metabolic Disorders, is leading this trial as the Principal Investigator and is supported by the specialist research team at QEH. The first patient with a diagnosis of Alström Syndrome was recruited in February 2016.

The clinical trial drug (PBI-4050) aims to halt the progression and possibly reverse some of the effects of fibrosis (scarring) which is one of the main problems associated with Alström Syndrome. The trial drug may also have the potential to improve sugar and fat levels within the body, improving diabetes and obesity and reducing insulin resistance.

The primary aim of the trial is to evaluate the safety and tolerability of PBI-4050 and its effects on inflammatory, fibrosis, diabetes and obesity biomarkers in people affected by Alström Syndrome.
All patients recruited to the trial will take the trial drug in capsule form. There will be no placebo, which means everyone taking part will receive the trial drug.

The trial has received ethics committee approval and is only open to people affected by Alström Syndrome who are over the age of 16 and meet the trial's eligibility criteria. Dr. Hiwot has ensured all safety measures are in place and that all patients will be closely monitored throughout the duration of the trial.

ASUK are working closely with colleagues at QEH and will be providing support to patients and their families, organising accommodation, travel and any assistance required throughout the trial to make it as easy as possible for those who want to participate.

While ASUK is hopeful that this trial will be a success, it is important to remember that this trial is for research purposes only and there is no guarantee that patients will have any clinical benefit. However, ASUK believe this to be an exciting opportunity and are hopeful that this trial will enable us to further develop our understanding of Alström Syndrome and further contribute to the management and treatment of this ultra-rare disease.

For further information on this trial you can visit www.clinicaltrials.gov using the trial reference number NCT02739217.

Further information about ProMetic can be found on their website www.prometic.com

Please get in touch with ASUK National Development Manager, Kerry Leeson-Beevers if you would like to learn more about this trial. You can call her on 01709 210151 or email kerry.leeson@alstrom.org.uk

Dates for your Diaries

Children's Clinics

To be held at Birmingham Children's Hospital in 2017
13th February 2017
8th May 2017
11th September 2017
13th November 2017

Children's Outreach Clinic in Leeds

9th November 2017

Adult Clinics

To be held at the Queen Elizabeth Hospital, Birmingham in 2017
11th and 12th January 2017
12th and 13th April 2017
12th and 13th July 2017
11th and 12th October 2017
8th and 9th November 2017
13th and 14th December 2017
If dates need to be changed we will let you know as soon as possible.

SAVE THE DATE!

We are delighted to announce that our next conference will be taking place in Birmingham on Friday 6th and Saturday 7th October 2017.

The conference will bring together individuals, families, researchers, clinicians and professionals. We are planning this exciting event now so please get in touch to register your interest today.

Please call Catherine Lewis on 01803 613117 or email catherine.lewis@alstrom.org.uk

Please keep an eye on our website for further information

Annual General Meeting 2016

The ASUK Annual General Meeting took place on the 5th November at the Aston Conference Centre in Birmingham. This was a lovely opportunity to meet the team and hear about the charities developments over the past twelve months.

Michelle welcomed everyone to the AGM after her re-election as ASUK Chair for a further three years.
ASUK Finance Manager, Steve Scoffield presented the accounts that demonstrate that ASUK continues to be prudent in its financial management which is stable and sustainable. If you would like to view our accounts these can be found on our website www.alstrom.org.uk

Michelle thanked the team for their tireless work for the charity and thanked the Trustees for their continued time and hard work. Michelle thanked the former CEO of ASUK. In 2015/16 Martin Henwood, acted as an Interim CEO and provided support to the team and developed new procedures and policies. Martin has now left ASUK and Michelle on behalf of ASUK wished him well in his new ventures.

Kerry, Catherine and Marie gave short presentations on achievements and challenges throughout the year:

- Kerry gave an overview of the multi-disciplinary clinics and spoke about the conferences and events where she represents ASUK and is often invited to speak. She highlighted ASUK's involvement with other organisations who are actively campaigning for developments within the field of rare diseases. Kerry described the 'Breaking down Barriers' project that she is leading and talked briefly about the research developments which are helping to understand the complex nature of Alström Syndrome.

- Catherine gave an overview of the years events, fundraising achievements, continuing to be a member of the Information Standards following a successful audit and raising awareness through the website, social media, campaigns and discussed the exciting re-brand of the charity.

- Marie gave an overview of the Children in Need transition project, explaining the resources that have been developed with children and young people. Marie described the many and varied, social and fun activities organised throughout the year and identified the positive outcomes for children and young people. For example, improving social skills, improving relationships with food and improving confidence. Marie also talked about the personalised family support she has been able to provide through home and school visits.

- Michelle said that she is proud of the diverse work being achieved by the team. The families who attended wanted to reiterate that they appreciate the work that the team does. Ensuring we are engaging with families, using the website and social media effectively to shout about the work we are doing. Looking also at a 'you said, we did' feedback section of the website.

After 3 years Julie Beck stood down as a Trustee, she stated that she would like to contribute in future and thanked everyone for their support. Michelle on behalf of everyone at ASUK, stated that we would like to say a HUGE THANK YOU to Julie for her time, effort and hard work.

ASUK will be advertising for more Trustees to join the Board. If you, or anyone you know maybe interested in knowing more about the role of a Trustee please get in touch, we would love to hear from you.

Getting to Know You Weekend

In August 2016, families enjoyed a 'Getting to Know You' activity weekend in Tamworth funded by Children in Need. A chance for families to get to know one another and try new fun activities. Families visited Tamworth Castle, enjoyed bowling, swimming and for the brave, had a go at driving a car!

"I enjoyed watching my daughters driving. The whole weekend was fabulous. Thank you for arranging this Marie."

"I liked driving the car. Being in charge of the car was quite good. I felt excited and confident."

In September, families enjoyed a residential activity week-end at Whitemoor Lakes, Staffordshire generously funded by Children in Need. Challenging activities included Abseiling, zip wiring, canoeing, fencing, and the quieter activity of arts and crafts. Mums and Dads, brothers and sisters got to know one another, learn from each other's experiences, and got to try out new activities whilst having lots of fun!

As part of the Children in Need project we have been introducing activity sessions focusing on nutrition and wellbeing. We have now expanded on the 'Foodie Friends Clubs' to look at other sessions we can provide which would be useful for families. To coincide with the children's multi-disciplinary clinics in November, we arranged for, John Pemberton BCH Paediatric Dietitian to run a sleep and relaxation session. John discussed the impact of sleep on your body, personal sleep habits, good sleep hygiene and the impact of poor sleep on physical impact, behaviour and emotions.

We then ran a workshop for families about portion sizes, where we worked in groups to decide on the appropriate portions for a typical, breakfast lunch, dinner, drinks and snacks throughout the day. The children and young people chose the portion size (not the parents) with the parents being encouraged to discuss choices with them, once each group had chosen the appropriate size with drinks. The dietitian discussed the choices as a group and then moved onto portion sizes. This was a really good way for everyone to see which foods need to be certain portion sizes to encourage a healthy lifestyle. These workshops will be developed further in 2017 to incorporate more useful information and guidance.

The Children in Need funding has enabled ASUK to provide children and their families, fun activities and opportunities. We are planning more activities in the New Year. In January we will begin taking a group of young people to visit the QEH to familiarise themselves with the Hospital and team before moving across from children's services. Please get in touch with Marie McGee if you would like to be involved, her email address is marie.mcgee@alstrom.org.uk she would love to hear from you.

NHS Stay Well This Winter: Don't put off your vaccinations!

The 'Stay Well This Winter' NHS campaign encourages vulnerable people including people with long-term health conditions and anyone aged over 65 to get a free flu jab.

Alström Syndrome specialists also advise that the pneumococcal vaccine should be given every 5 years if you are affected by a complex condition such as Alström Syndrome. The pneumococcal vaccine protects against serious and potentially fatal pneumococcal infections. It's also known as the "pneumo jab" or pneumonia vaccine. Further information about this vaccine can be found via the NHS website
http://www.nhs.uk/conditions/vaccinations/pages/pneumococcal-vaccination.aspx

Flu is a highly infectious disease and can lead to serious complications if you have a long-term health condition, for example: chronic respiratory disease, such as chronic obstructive pulmonary disease, bronchitis, or emphysema; diabetes; heart, kidney or liver disease; chronic neurological diseases, like multiple sclerosis or cerebral palsy; or have suffered a stroke. People with these and other long-term health conditions are eligible for a free flu jab through their GP or pharmacist.

The free vaccine is also offered to other groups at particular risk of infection and complications, anyone aged 65 and over; residents of long-stay care homes; carers; and pregnant women. Children aged 2, 3 and 4 and in school years 1, 2 and 3 are also being offered a free flu vaccination.

Those eligible should contact their GP or pharmacist, to arrange a vaccination. If you are the main carer of an older person or someone with a disability you may be eligible for the free flu jab. The advice is to ask at your GP's surgery
Visit www.nhs.uk/staywell for more information

Genetic Disorders Campaign Launched

We have partnered with Mediaplanet UK for the 2016 Genetic Disorders Campaign which launched in The Guardian newspaper on the 23rd September 2016. This also raised awareness online through the health awareness website. You can read about inspirational twins Katie and Hannah who are affected by Alström Syndrome via their website:

http://www.healthawareness.co.uk/genetics/genetic-disorders/our-genetic-disorders-and-us-read-our-true-stories

ASUK Bike Club

ASUK currently has spare tandems, trikes and tag-alongs which all need a new home. If you are interested in getting out and about and experiencing the joy of cycling please get in touch to find out more information.

Are you getting enough Vitamin D?

People who are affected by Alström Syndrome will stay out of the sun to avoid the bright light which could mean that they are not getting the recommended amount of vitamin D to remain healthy.

The NHS advice is that everyone should consider taking a daily supplement containing 10mcg of vitamin D in the winter. During the spring and summer months the majority of people should be able to get all the vitamin D needed from sunlight on the skin. So you may choose not to take a vitamin D supplement during these months. Please follow the link below to visit the NHS website for full information.

http://www.nhs.uk/conditions/vitamins-minerals/pages/vitamin-d.aspx

Awareness Raising

In September, ASUK National Development Manager, Kerry Leeson-Beevers attended the EURORDIS - European Rare Diseases Organisation Round Table of Companies workshops in Barcelona. The focus was "Bringing solutions to young rare disease patients". Kerry was delighted to be asked to lead a workshop at the event which focused on how to engage with young people. You can find further information via the EURORDIS website http://www.eurordis.org/ertc

On the 22nd October, Kerry and ASUK CEO, Ann Chivers attended the Midlands Rare Disease Showcase at Birmingham Children's Hospital. The event celebrated advancements in the rare disease community by bringing together patient groups, pharmaceutical and biotech companies, clinicians and researchers to discuss current and future developments.

In October, Kerry attended the National Institute for Health Research (NIHR) 'Think Research' Rare Disease Patient Day at the Barbican Centre in London. The day was a joint collaboration between NIHR Rare Diseases Translational Research Collaboration (RD-TRC) and NIHR BioResource – Rare Diseases. The day featured many research sessions and presentations about patient information, ethics, protocols and provided many more interesting topics throughout the day. You can find further information including videos and summaries from the day via the NIHR website http://rd.trc.nihr.ac.uk/think-research-patient-day2/

On the 30th November, Kerry was invited to attend the Sparks Winter Ball. The purpose of the event was to highlight the work of Sparks, to thank the people who have contributed funding towards the development of paediatric research and to raise funds to enable them to continue this vital work. The evening raised over £300,000 and it was hosted by an array of sporting stars. ASUK continue to collaborate with organisations like Sparks to ensure we are aware of new developments in research and we are always seeking new opportunities to further develop our knowledge and understanding. You can find further information about their work via their website http://www.sparks.org.uk/

Genetic Alliances' New Project, Get Involved Today!
Genetic Alliance UK are looking for people to help shape the way that information is produced in relation to genome editing. They would like people to take part to develop information, which is clear for everyone to understand and they need your help to do this.

In the New Year, they will be arranging four workshops in central London and if you are selected to take part you would need to commit to attending all four workshops. All reasonable expenses will be reimbursed.

If you are over 18 years old and if you or a member of your family is affected by a genetic or non-genetic condition or suspected condition, they would like to hear from you. Follow the link to find out more information and register your interest: https://www.surveymonkey.co.uk/r/8N8CTHT

Helpful Hints and Tips

The Care Act 2014
– are you getting what you are entitled to?

ASUK Family Support Officer, Jane Biglin has a wealth of knowledge and experience regarding benefits and entitlements. She has produced the first 'Helpful Hints and Tips' for families which we hope you find useful. Please let us know any topics which would be most helpful for you and your family and we will compile information onto our website and in each newsletter.

The Care Act 2014
– your right to specialist assessment

Many families and individuals who are affected by Alström Syndrome know what they are entitled to by law. Others are not so sure, especially after the law changed last year. So we wanted to give you brief information which may help you decide whether your needs have been properly looked into and provided for by your own Local Authority. With apologies to those outside England this information relates to English law only – Wales has its own guidance and we can advise accordingly.

This information is for those over 18 years of age

– if your child/young person has not been assessed please let us know if you need support, as the guidance is slightly different, particularly around the level of expertise the assessor holds. For example, in England any assessment of a deafblind/multiple sensory impaired child must link up with an assessment of Special Educational Needs. For those approaching 18 years old the Care Act now includes 'transition assessment'.

If you need this information in another format, or if you have any questions or issues please ask us, as we recognise every person and family is unique.

How does an assessment work?

The assessor will meet you at your home (this may be once or several times). They should discuss and look at your access to the local environment. The assessor must be either a deafblind qualified social worker or a qualified assessor from an outside organisation, commissioned by the Local Authority. The assessor will usually use an assessment form – you can ask for a copy in suitable format (large print, Braille) before the meeting, to help you consider the questions in advance. We always encourage a positive attitude around what you can do, however you may need to speak specifically about any difficulties you have on 'bad' days. The assessment should look at your needs in the following areas and recognise that this may change at different times (within a day, a month, at different times of year or weather conditions):

- Communication
- One-to-one contact with people
- Social interaction and accessing the community
- Support with mobility
- Assistive technology
- Rehabilitation / mobility
- Your current and possible future needs

The assessment should be 'carer-blind' which means that needs are assessed as if the person, usually a family member, is not available so that if, for example, the family member is taken ill there is still sufficient support in place.

The assessment should also look at how Alström Syndrome affects various areas of your life and how this affects your general wellbeing. For example, if in the past you have been unable to join activities you wanted to do outside the home, then this must be part of the assessment and care plan process, to see if there is a way to do things with the necessary support / equipment.

The Care Act 2014

The Care Act 2014 replaced previous laws (often referred to as Section 7 Deafblind Guidance) and is considered to be stronger for people who have sight and hearing loss, as well as the other complications associated with Alström Syndrome.

Under the Care Act, special deafblind guidance has been issued to Local Authorities called 'Care and Support for Deafblind Adults and Children'.

We know that the word 'deafblind' can be difficult for some people – it doesn't mean someone has lost all their sight or all of their hearing – so most people who are affected by Alström Syndrome may be described as deafblind within the law. The important point is that even a small loss in sight and hearing together can give challenges in everyday living, especially with communication, accessing information and mobility.

What does the Local Authority have to do by law?

The most important areas for you to know are that Local Authorities have to:

- Make an assessment of need for support – needs may include providing care, equipment, access to the community (Personal Assistants, Communicator Guides, intervenors) and information and advice
- Make sure the person carrying out the assessment MUST be suitably qualified and deafblind trained in line with the Care Act 2014. Generally, it is not acceptable to use separate assessors such as one related to hearing impairment and another for visual impairment from two separate teams. The authority may not have suitably qualified staff themselves so may need to use outside organisations with suitably qualified staff. It is very important to check this
- Ensure when services are provided they may need to be specific to dual sensory loss, rather than hearing loss or vision loss as single sensory impairments
- Carry out an assessment, if you request it (subject to being eligible through dual sensory loss and other complex needs).The authority cannot say that it does not have sufficient funding or that your financial situation is too high for them to do an assessment. If you are refused an assessment, please let us know so that we can check or help you check that the authority has a good reason

The assessor writes a report which shows the full extent of your needs. The assessor reports on your 'eligible needs'. This may include:

- Specifically trained one-to- one support workers (Personal Assistants, Communicator Guides, Intervenors)
- Home care help with things like cleaning and shopping where visual loss makes this difficult
- Equipment for the home – this should allow for sight and hearing loss rather than separate provision which doesn't take account of your eligible needs. For example, we hear of Deaf Services giving a doorbell with a flashing light or a Visual Impairment team giving a doorbell with a loud alert when together they should have given a bell with a tactile alert.
- Technology equipment and support
- Recommendations and information about local service provision and networking
- Mobility training
- Teaching daily living skills over a suitable time period e.g. 6 weeks
- Communications skills

If you would like one of the ASUK team to be there with you for the assessment, please talk it through with us in advance of the meeting to enable us to plan the visit.

The Care and Support Plan

A budget will have been given which is an estimate of meeting your care and support needs before the care plan is put together. This may be low if a Local Authority uses what is called a Resource Allocation System (RAS) to decide an available budget – the Care Act says that this may not be used where needs are complex or costly to meet, because the person is deafblind. If the estimated budget seems impossibly low let us know as we can help check if RAS has been used.

Together with you, the qualified person who carried out the assessment should be involved in helping to draw up your care and support plan. This is so that you have control and influence over the plan and may use the person's expertise to come up with solutions you may not know about, for example technological equipment.

After the Assessment Report?

A suitable qualified person then decides for the Local Authority whether your needs meet the eligibility criteria. The eligibility criteria includes the following outcomes – you may be able to do some, but not always in a safe, timely way or without stress. You will have an eligible need if two or more of the following outcomes are not met:

- Managing and maintaining nutrition – for example if you have difficulties shopping, cooking, identifying food labels and instructions
- Being appropriately dressed – for example choosing clothes by colour, dress appropriate to the weather, clothes that need washing or mending
- Being able to make use of the home safely –for example you may need vibrating smoke/fire alarms or doorbells; improvement to lighting to avoid falls and safe movement around your home
- Maintaining a habitable home environment – e.g. keeping the house clean, hygienic and hazard free; being able to manage paying your bills
- Developing and maintaining family and other personal relationships – for example is it difficult for you to communicate or get out to meet friends; difficulty in using the phone or email
- Accessing and engaging in work, education, training and volunteering. For example are you receiving support and information in ways that are suitable for you to understand and join in?
- Making use of necessary facilities or services in the local community including public transport and recreational facilities and services. For example can you go swimming, are there ways for you to keep fit and active?

Decision On Funding

The care and support plan MAY go to a panel which must include your assessor (or a similarly qualified person).
The Local Authority will then decide on a Personal Budget to meet your needs.

If the cost of services is higher because they do not have qualified staff, such as dual sensory qualified Personal Assistants, then they should increase the budget. This is instead of expecting you to take fewer hours or a lower paid unqualified worker who may provide an inadequate service if, for example, they do not know how to sight guide safely.

They will either pay for some or all of the services themselves or offer you Direct Payments where you make your own decisions about equipment and services.

What are the Advantages?

New rules in the 2014 Care Act allow you to take the lead in explaining what care and support you need to make life easier for you. The Care Act places your wellbeing at the centre of your support needs.

If you have your own finances, over the thresholds there may be no financial contribution from the Local Authority. However, the assessment will give you the chance to discuss your needs with a health or social care professional who can advise you on what help is available.

What are the Disadvantages?

- Being assessed can feel intrusive. However, our experience is that if someone is suitably qualified they have a real interest in your needs and you can tap into their expertise for ideas even if, in the end, there is no provision of service
- It can be a battle - we are very aware that Local Authorities are varied in their provision and some ignore the spirit of the Care Act or are under enormous financial pressure. We do not know what the effects of recent political changes may be but we have to work in the present. We believe in the need for Care Act provision for people who are affected by Alström Syndrome. Some families are very good at making their own cases (any tips welcome!), while for others we are very happy to support you in seeking the correct levels of service
- The amount the Local Authority is willing to pay depends on your finances. Once your Local Authority has carried out your assessment and worked out what services you need, they'll carry out a financial assessment. This is called a 'means test'. This looks at your regular income (pensions, benefits or earnings) and your capital (cash savings and investments, land and property and business assets). However, your home won't be counted as capital if family or close relatives (meeting certain conditions) live there as well as you. The threshold in England is £23,250. This will help you work out if you need to contribute towards the cost of your care and support plan, and whether the Local Authority will pay for all or some of your costs.

How to get an Assessment?

If you have already had an assessment it may have been a standard social care needs assessment but not a specialist deafblind assessment. If you are in doubt, contact us for advice.

If you have had an assessment in the past few years but your needs have altered, perhaps because your sight has changed or another condition has affected your needs, then you are entitled to ask for a new assessment. In any event, any existing care plan should be reviewed annually and within the Care Act guidance.

Contact your Local Authority adult social services department, if you need support to do this let us know. Request a deafblind specialist assessment which is compliant with the Deafblind Guidance issued under the Care Act 2014. Remember, the Local Authority cannot refuse to do this on the grounds of your finances or their idea of your need prior to assessment.

Personal Health Budgets (PHBs)

We are currently awaiting guidance on a new option which MAY apply to people who are affected by Alström Syndrome. A personal health budget is an amount of money to support a person's health and wellbeing needs, planned and agreed between the person and their local NHS team. The idea is to enable people with long-term conditions and disabilities to have greater choice, flexibility and control over the health care and support they receive. Personal health budgets are very new so the process is still being developed. We are very fortunate that Jane Lodwig, formerly the clinical nurse at the Alström Syndrome adult clinics is now working directly with PHBs and we hope to receive updates from her and will let you know if this will apply.

If you would like to share your own experience of the assessment process, please email us. It is helpful to share ideas, especially about how people have argued their case and obtained a good care package. We know it is easier in some areas of the country than others and we can all learn by sharing. This is about rights not about continually having to fight for basic human rights.

This information is part of a series that we will be producing to help individuals and families. If you would like to contribute or suggest ideas for other information sheets please let us know.

You can do this by either emailing Catherine at:
Catherine.lewis@alstrom.org.uk or calling 01803 613117.
We are always pleased to hear from you.

Rare Disease Day 2017

Research brings hope to people living with a rare disease

In preparation for Rare Disease Day 2017, EURORDIS have published the below information about the theme for next year's Rare Disease Day and why this awareness is vitally important.

'Rare disease research is crucial to providing patients with the answers and solutions they need, whether it's a treatment, cure or improved care.

On 28 February 2017, the tenth edition of Rare Disease Day will see thousands of people from all over the world come together to advocate for more research on rare diseases. Over the last few decades, funds dedicated to rare disease research have increased. But it can't stop there. Rare Disease Day 2017 is therefore an opportunity to call upon researchers, universities, students, companies, policy makers and clinicians to do more research and to make them aware of the importance of research for the rare disease community. Rare disease patients and families, patient organisations, politicians, carers, medical professionals, researchers and industry will come together to raise awareness of rare diseases through thousands of events all over the world.

Rare Disease Day 2017 is also an opportunity to recognise the crucial role that patients play in research. Patient involvement in research has resulted in more research, which is better targeted to the needs of patients. Patients no longer solely reap the benefits of research; they are empowered and valued partners from the beginning to the end of the research process.

Background on rare disease research

The US Orphan Drug Act (1983) and the EU Regulation on orphan medicinal products (2000) both stimulated rare disease research. This is the result of determined advocacy efforts from the rare disease community to ensure that policy makers, companies and researchers understand the benefits of rare disease research.

In EU programmes alone, €620 million were assigned to rare disease research between 2007 and 2013. Money continues to be allocated to rare disease research through the EU with €200 million assigned between 2014 and 2015, making rare disease research continuously attractive to scientists. In parallel, EU Member States have grown their research budgets.

Due to the rarity and diversity of rare diseases, research needs to be international to ensure that experts, researchers and clinicians are connected, that clinical trials are multinational and that patients can benefit from the pooling of resources across borders. Initiatives such as the European Reference Networks (networks of centres of expertise and healthcare providers that facilitate cross-border research and healthcare), the International Rare Disease Research Consortium and the EU Framework Programme for Research and Innovation Horizon 2020 support international, connected research.

Rare disease research reduces costs for healthcare systems. As a result of research, a rare disease patient who is diagnosed or properly treated no longer needs irrelevant tests, ineffective treatment or hospital visits. In addition, research on specific rare diseases often shines a light on more prevalent diseases. Pioneering approaches in rare disease research often benefit the much wider public affected by more common diseases.

There are various types of research and frameworks that are important to the rare disease community, including:
Infrastructures such as registries (databases of patients with their clinical and genetic information) and biobanks (catalogues of human biological samples), which researchers need to do their work. Initiatives such as RD-Connect connect databases, registries, biobanks and clinical bioinformatics for rare disease research.

At present only few European countries fund research on rare diseases through specific dedicated programmes. The E-Rare consortium links responsible funding organisations and ministries that combine scarce resources for national rare disease research and enables the participation of many researchers in transnational projects.

Fundamental research to identify the cause and mechanisms of rare diseases. Translational research, which focuses on using the conclusions of basic research to develop therapeutics for patients living with a rare disease.
Clinical research, when medicinal products are tested in humans through clinical trials.

Not only is clinical research important, research into quality of life, working conditions, social needs, integration at school, and multidisciplinary education of social service providers is key and supported through projects such as INNOVCare.

Rare disease research changes the lives of the millions of people living with a rare disease across the world and their families.'

Visit the Rare Disease Day website and Get Involved with Rare Disease Day 2017 http://www.rarediseaseday.org/

Remembering a very special young lady

Maariyah Iqbal, sadly passed away on the 2nd December 2016, shortly before her 17th birthday.

She was a truly inspirational young lady who was always smiling and laughing and she never let anything stand in her way. She loved socialising with her family and friends and she thoroughly enjoyed the time she spent with members of ASUK.

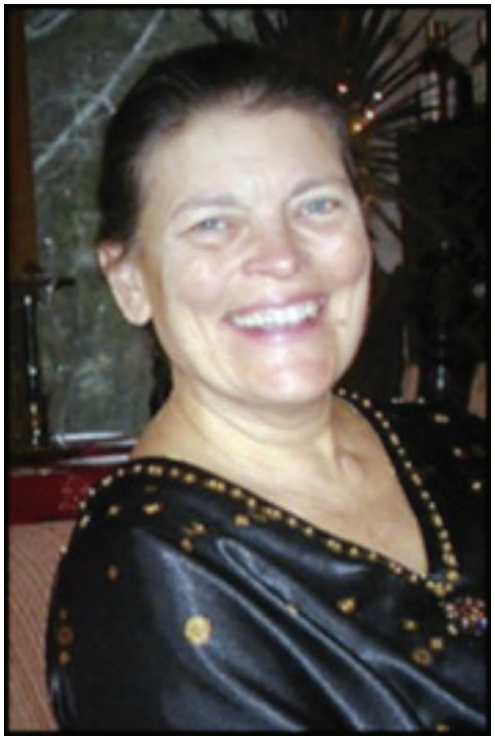
Maariyah was an active member of our Hear My Voice youth forum and she was an excellent advocate. She was never afraid to stand up and speak her mind to remind others of the importance of listening to the voices of young people.

Maariyah was extremely brave and courageous and she was supported by her wonderful parents, sister and extended family members. Everyone at ASUK sends their deepest condolences to all of her family and friends.

Tribute to a very special lady

Jan Marshall, sadly and unexpectedly passed away on the 6th September 2016.

On behalf of ASUK, Dr. Richard Paisey, ASUK Trustee has written a tribute to Jan, who was an inspiration to all.



No praise is too much for such a wonderful person. She embraced the cause of Alström families worldwide and laboured tirelessly on their behalf.

Having had the privilege of collaborating with her for 18 years I can say, without reservation that she was kind, courteous and thoughtful of others at all times. Yet she possessed a steely determination to uncover the genetic cause of Alström syndrome and put that discovery to further the cause of families.

I can remember occasions when her humanity and selfless caring for families left me in tears, not least the group hugs with Alström friends at conferences.

All of us in the UK, who met Jan have been aware of the presence of profoundly caring, intelligent and welcoming person who always put others first.
God has laid a finger on her brow.

Her spirit of friendship and most ardent scientific endeavour will not be forgotten. We must continue her work in our own lives. In fact she is a role model for family support and investigation of all rare disorders.

By Dr. Richard Paisey, ASUK Trustee on Behalf of Everyone at ASUK
Please follow the link below to view Jan's obituary from her local newspaper.

http://obituaries.bangordailynews.com/story/jan-marshall-829342971

Alström Syndrome UK
Strength for today, hope for the future

Alström Syndrome UK (ASUK) is an award winning charity who provide information, support and advice for individuals affected, their families, carers and professionals. Alström Syndrome is a very rare genetic condition which can cause progressive blindness, hearing loss and can lead to; heart and kidney failure, type 2 diabetes, liver dysfunction and associated problems. The symptoms arise at different stages making diagnosis difficult. We endeavour to raise awareness, conduct pioneering research and enable better treatments and monitoring through the AS multi-disciplinary NHS clinics.
Further information about our work can be found at **www.alstrom.org.uk**

ASUK offer a help-line during office hours and an answer phone service at all other times where calls are promptly returned by our experienced Family Support team;
Please call 01803 613117 / 01709 210151

A huge thank you and a very

Merry Christmas!

to our friends and supporters

Please feel free to contact any member of the team

Kerry Leeson-Beevers
Tel: 01709 210151 / 07716135940
Email: Kerry.leeson@alstrom.org.uk

Catherine Lewis
Tel: 01803 613117 / 07970071675
Email: Catherine.lewis@alstrom.org.uk

Marie McGee
Marie works part-time, normally on a Friday offering transition support to young people and their families
Tel: 07812 173953
Email: marie.mcgee@alstrom.org.uk

Jane Biglin
Jane works part-time, normally on a Tuesday and Wednesday offering family support to families in the South
Tel: 07714 798413
Email: jane.biglin@alstrom.org.uk

Contact over the Christmas period:

If you need to get in touch with us urgently over the Christmas period please dial the office telephone number 01803 613117 and we will endeavour to get back to you within 24 hours.

A heartfelt thank you to all our friends and supporters who have worked tirelessly in 2016. As many of you know this is a difficult time for many charities and we have worked hard to ensure we continue to offer a high level of support for families; at clinic and on a more personal level at home, in schools and colleges and in the community.

We have supported individuals who have generously given their time to take part in the clinical trial and worked with others to raise awareness of Alström Syndrome.

We have organised fun weekends away for families and continued to ensure families have access to equipment and financial support. To all our supporters; families, clinicians, researchers, volunteers, the team at ASUK and all, an enormous thank you for your continued support and the valuable contribution that you make.

In 2017 we will be recruiting more family support staff to ensure that individuals and families can be supported wherever they live in the UK. We will continue to encourage research and we are hopeful that real progress can be made in finding potential new treatments and discovering more about Alström Syndrome for the benefit of this and future generations. On behalf of us all at ASUK I would like to wish you all a Merry Christmas and a very happy and healthy New Year.

Ann Chivers, Chief Executive at ASUK

Our vibrant new logo is revealed!

Alström Syndrome UK
Strength for today, hope for the future

Firstly, a huge thank you to everyone who completed questionnaires about the new logo and strap line and gave feedback during face to face meetings. We received lots of responses and suggestions about the colours, the look and the image we were trying to project. Kerry and Marie led a consultation with families who came up with a lovely positive strap line, Jane asked everyone she met to comment on the logo and Catherine analysed lots of feedback.

This new logo and strap line represents ASUK: promoting fresh and innovative ways of working that empowers individuals to reach their potential and supports families on that journey. It is vibrant and captures a picture of working together and a pictorial representation that nods to the genetic source of Alström Syndrome. The use of 'happy colours' can be used in a variety of ways throughout our promotional materials and we are sure it will stand out on our website.

The logo is made up of 3 'figures' each in a different colour with 2 tones of each colour. The middle figure is green, a grass green and a lighter green tone, the figure to the right is 2 shades of orange, the deeper colour is a burnt orange, the figure to the left is 2 tones of blue, sky blue and the other shape is a lighter blue. The umlauts are green, the same colour as the middle figure. The lettering is black and bold.

Orange is the colour of innovation, modern thinking and health, green is often used to reflect ethical credentials, organic growth and nature and blue is often used to imply professionalism, integrity and tranquillity. The overall effect is bright and colourful, it speaks of boldness and strength.

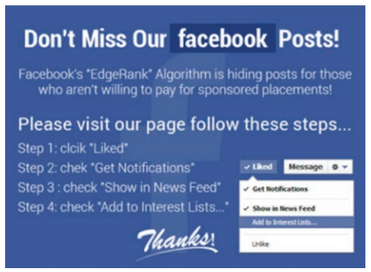
We hope you all love the logo as much as we do and we hope it makes you all smile!

Alström Syndrome UK
Strength for today, hope for the future

Registered Office address: 31 Shearwater Drive, Torquay, Devon TQ2 7TL
Please visit our website www.alstrom.org.uk to keep up to date with our latest news.

ASUK now have a dedicated You Tube page
http://www.youtube.com/alstromsyndromeuk

Please keep up to date with Alström Syndrome UK via twitter @AS_UK and also via our dedicated facebook page:
www.facebook.com/alstromsyndromeuk,
please click 'like' to show your support and spread the word!



Registered Charity no 1071196 Registered Company Limited by Guarantee 3557191

Design & Print Ambinet - www.ambinetcreative.com