**With your help we have hope**

**THE NEWSLETTER OF ALSTRÖM SYNDROME UK Summer 2015 Issue No.16**

Alström NEWS

Alström Syndrome UK provide information, support and advice for individuals affected, their families, carers and professionals

working with them. Alström Syndrome is a very rare genetic condition which can lead to progressive blindness and deafness and

can also 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

Registered Charity no: 1071196 Registered Company Limited by Guarantee 3557191

Exciting Research Times Ahead!

On the 11th May 2015, ASUK

Chair Michelle Hough and ASUK

National Development Manager

Kerry Leeson-Beevers attended a

meeting with Dr. Rob Semple at the

Biomedical Campus in Cambridge.

The aim of the meeting was to

discuss our research projects and

the best ways to move forward.

ASUK are forging ahead to gain

further funding as there is such

an emphasis on rare disease and

research in this area. Alongside

the teams at the Queen Elizabeth

Hospital and Birmingham Children’s

Hospital, Dr. Semple at Cambridge

University has been able to collect

skin samples from patients to learn

more about this very rare condition.

This was made possible in 2010

through Big Lottery funding of

£350,000 which has enabled further

insight into this condition.

Professor Phil Beales and

Professor Tim Barrett have also

been developing research into

Alström Syndrome which can now

be developed further through

a NIHR Translational Research

Collaboration grant “Deep

phenotyping in Bardet-Biedl and

Alstrom Syndromes” which has

received £465,000 in funding.

A further research meeting has

been scheduled to take place in

Birmingham on the 15th July 2015.

ASUK will be re-establishing the

Medical and Scientific Advisory

group which will be discussed

during this meeting. This will

bring together members and

professionals alike to discuss

research projects which are in the

best interests of everyone involved.

This will enable specialists to

collectively look at research projects

available to determine what would

be in the best interests of patients

to take forward and develop

further.

The ASUK team are very excited to

be working alongside the teams at

Cambridge University, the Queen

Elizabeth Hospital, Birmingham and

the Birmingham Children’s Hospital

as well as the expertise from our

new Trustee Dr. Richard Paisey.

This team work will enable future

developments and research

projects to be achieved.

ASUK Research Facts

• Big Lottery funded £350,000

in 2010 for a 3 year research

project to learn more about this

rare condition, skin samples were

collected and clinical patient

database compiled.

• 23 sets of cells were originally

collected and stored at Cambridge

University.

• 5 were then batched and sent to

the tissue bank at BCH as part of

EUWABB project.

• Skin samples sent in 2015 to Prof.

Phil Beales for further research.

• Prof. Beales and Prof. Barrett

“Deep phenotyping in Bardet-Biedl

and Alstrom Syndromes” research

started.

• The Euro-WABB project is an

initiative, to investigate Rare

Diabetes namely Wolfram, Alström

and Bardet Biedl. ASUK are actively

involved in this work with Prof. Tim

Barrett.

• Potential collaboration with

Prometics to research anti-fibrosis

treatments.

Farewells and Greetings

Alström Syndrome UK has some

big news. After founding and

piloting the charity for many years,

Kay Parkinson is now moving on.

For the past 24 months, Kay’s work

has taken her across Europe and

further afield while the Board of

Trustees and our contractors have

continued to drive ASUK forward

and have ensured that our families

are supported throughout the UK.

Our families will always be at the

heart of everything we do, now

and in the future and we would like

to reassure you that there will not

be a change to the level of support

you receive.

ASUK will continue to offer a 24

hour help-line, covered by our very

experienced team:

Kerry Leeson-Beevers: Tel: 01709

210151 / 07716135940 or email

Kerry.leeson@alstrom.org.uk

Catherine Lewis: Tel: 01803 613117

or email Catherine.lewis@alstrom.

org.uk

Iram Shah (fluent in Urdu and

Punjabi): Tel: 07988 237664 or

email: iram.shah@alstrom.org.uk

John Parkinson: Tel: 01803 409952

or email john.parkinson@alstrom.

org.uk

On behalf of our Board of Trustee’s

and the team at ASUK, we wish to

give our warmest thanks to Kay for

everything she has achieved for

our families and we look forward

to the continuing developments

within the Alström community.

Curtis Vasey

who has been an ASUK Trustee has

decided to step down from his role

but he would still like to remain

involved in the work of ASUK.

Especially in developing transition

services and activities for young

people affected. Curtis has been a

dedicated Trustee who has always

had the needs of our families at the

heart of everything he has done.

ASUK would like to sincerely thank

him for his commitment and

support over the years.

At QEH, Psychologist Robin

Paijmans who has been involved

with our AS clinics is sadly leaving.

We are working with QEH to ensure

another Psychologist will be part of

our service moving forward and we

hope to have another Psychologist

in place soon.

Martin Henwood

Martin Henwood has been

appointed ASUK’s interim Chief

Executive Officer and has come on

board to develop our governance,

ensuring we are compliant in

all areas. ASUK will soon be

advertising for a part-time Chief

Executive Officer to develop the

charity further.

Dr. Richard Paisey

We are delighted to announce that

Dr. Richard Paisey has joined the

ASUK Board of Trustees. Richard

has been part of the ASUK’s journey

since it began, helping develop

the ASUK multi-disciplinary clinics

at Torbay Hospital, before they

relocated to the Queen Elizabeth

Hospital in Birmingham. Richard

has been involved in our research

projects, including monitoring

members who took part in the

ASUK bike ride in 2012. His

expertise and experience of this

very rare condition will be a major

boost for ASUK as we progress our

future research work and ongoing

developments.

Introducing, National

Transition Coordinator

Marie McGee

We have pleasure in introducing

a new member to our team,

Marie McGee who will be our

National Transition Coordinator.

The post has been kindly funded

by Children in Need as part of

a three year transition project.

Interviews took place on the 1st

May 2015 in Birmingham and we

would like to say thank you to our

members, Curtis and Maariyah for

their excellent contributions to the

interview process.

The Children in Need Transition

project will work with young

people and their families

affected by Alström Syndrome.

By providing fun activities

and one to one support with

transition, ensuring that young

people who are affected are less

isolated, healthier and able to

better manage change.

Marie explains about Safeguarding Team to make the

Common Assessment Framework her previous work and more accessible, using actors and

achievements below: media to create a short film and

poster resource for Birmingham’s

ASUK

families and professionals.

My early career saw me work in

a number of inner city Primary

and Secondary schools where

I supported children and

young people (CYP) to develop

connections with their emotions,

skills and abilities using a variety

of activities. These include nurture

groups, anger management

sessions, 1:1 mentoring, playground

buddies, family skills and youth

parliament. I believe in widening

CYP opportunities in life so have

arranged involvement in activities

such as: the opening of Water

Hall Art Gallery, Birmingham (with

Cherie Blair) organising theatre

productions around themes

including gang/gun culture, and

organising careers events with

artists who delivered speakeasy

activities (building confidence to

speak up for yourself ). The role

also saw me lead on statement

reviews, attending child protection

meetings, child in need reviews

and family representation at police

interviews.

I moved on from there to work

for Birmingham Children’s Fund,

which became part of Birmingham

City Councils Children and Young

Peoples Commissioning Team.

Here I commissioned a variety of

services with a budget of £150,000.

My creative talents have allowed

me to also contract a number of

additional events including anti-

bullying workshops in a variety

of locations across the city. The

events included dancers, street

artists and bakers to give families

and professionals a platform to

discuss/respond to aspects of

bullying in a safe and fun manner. I

also worked with Birmingham City

Councils Communications Team to

produce city wide leaflets to inform

families about child protection in

relation to Victoria Climbie and the

new government framework for

assessing family needs. In addition

I also worked with the Children’s

In 2007 I eventually started my own

mentoring consultancy known as

Achieving Together. This is where

I could develop a more creative

range to my delivery which includes

projects like Lunchtime Heroes,

where I developed a range of

characters to support behaviour

issues in Primary schools. (See the

website at: ambinetdemos.com/

lunchtimeheroes/)

Balanced with my mentoring work

I am also employed part time at

Birmingham Children’s Hospital

as the Transition Co-ordinator

for the Rheumatology and Rare

Diabetes Teams. Within the role,

I build the skills and knowledge

of young people with long term

chronic health conditions. This

helps to build their resilience to

manage their condition, which in

the long term helps them to reach

their best potential. These young

people often feel they have become

invisible or fear being different

so using creative artists, rappers,

dancers and singers to give them a

different outlet, has a very positive

impact on them.

In mid-2014 I took a sabbatical for

four months in Tanzania. I became

involved in a country wide project

to produce resources to raise the

awareness of the early signs of

childhood cancer. I managed to get

funding from a Dutch Construction

Company, coupled with a reduced

rate for graphic design and printing

rates from a Birmingham based

Media Company.

I am excited to be involved in this

transition role within ASUK and I

look forward to supporting both

young people and the family as

a whole to ensure their voice is

heard and transition is a positive

experience for them.

Please feel free to contact

Marie McGee for further

information:

marie.mcgee@alstrom.org.uk

Raising Awareness at

Prestigious Events

Kerry Leeson-Beevers recently

attended a Leadership Symposium

organised by Genetic Disorders UK

in partnership with Global Genes.

The purpose of the event was for

charities to come together and

share best practice. The impressive

line up of speakers highlighted

the excellent work and projects

that are happening throughout

the UK and how new projects

such as the 100,000 Genomes

Project are bringing new hope for

a greater understanding of genetic

conditions. Pictured below, Kerry

with guest speaker, Actor Warwick

Davis.

Kerry went on to attend the Oxford

Rare Disease Conference 2015

from the 6th - 7th May 2015 at St.

Anne’s College in Oxford. This was

a great opportunity to learn more

about current and future medical

advances, research projects and

network with similar organisations.

The objective for the conference

was to support the development

of new therapies for rare diseases

by creating a forum where

representatives from academia,

industry, patient organisations,

and funders could come together

to network and exchange ideas.

Professor Dame Kay Davies, Chair

Oxford Rare Disease Initiative

thanked Kerry for attending this

prestigious event, “This could

only be achieved if all attendees

contributed and I would like to

thank you for helping to make this

event a great success with your

presence and your enthusiasm.”

From the 6th - 9th June 2015, Kerry

also attended the European Human

Genetics Conference in Glasgow.

The conference brought together

all workers in human and medical

genetics to review advances and

develop research collaborations.

ASUK were delighted to have an

exhibition stand at the event and

with over 3,000 delegates attending

this was a great opportunity for

researchers and professionals to

learn more about this ultra rare

condition.

Family Stories

You may remember from our winter

2014 newsletter, our article about

our members becoming increasing

interested in Judo as a fantastic

sport to be involved in. Since then

one of our members Millie (pictured

below) who is 7 years old has gone

on to win a bronze and gold medal

in a local competition, well done

Millie!

Information & Activity Day

19 SEPTEMBER 2015

SAVE the DATE

ASUK will be holding a more

informal conference this year on

Saturday 19th September 2015.

This information and activity event

will take place at New College

Worcester, where families and

professionals will be able to hear

about updates regarding the

charity and pioneering research

opportunities. The AGM will be

held before lunch and there will be

activities and workshops being held

in the afternoon to keep everyone

entertained and informed. There

will also be an opportunity to tour

the school and college facilities

throughout the day.

Please find booking form and

further information enclosed

Further information can be found at:

www.alstrom.org.uk

or contact Catherine Lewis to

reserve your place on:

01803 613117

email: catherine.lewis@alstrom.org.uk

We look forward to seeing

you all there!

Patient

Engagement

Group

ASUK National Development

Manager, Kerry Leeson-Beevers

is now an active member of the

Patient Empowerment Group,

coordinated by Rare Disease UK.

The purpose of the group is to

monitor the implementation of

the UK Strategy for Rare Diseases,

ensuring the voices of patients and

carers are heard.

The group is now also being used as

a sounding board for Public Health

England during the development of

the Congenital Anomaly and Rare

Disease Registration Service. For

more information please follow the

link below or contact:

Kerry - 01709 210151 or

email Kerry.leeson@alstrom.org.uk

www.gov.uk/the-national-congenitalanomaly-

and-rare-disease-registrationservice-

ncardrs

Otley Sailing

group – 28th

May 2015

On the 28th May 2015, families

affected by Alström Syndrome had

a fun-filled day out at Otley Sailing

Centre. This was a great opportunity

for families to try sailing in a

relaxed atmosphere with the help

and support of Otley Sailing Club

who kindly provided volunteers

for the day and supported by our

National Development Manager,

Kerry Leeson-Beevers. Otley Sailing

Club promote dinghy sailing in the

West and North Yorkshire regions

providing sailing, power and

safety boat training from absolute

beginners to the highest level.

They are committed to working

with sailors of all abilities, have

a Sailability Centre of Excellence

award and are an RYA OnBoard

Centre offering sailing experiences

to young people in the local area.

You can find out more about their

fantastic work at:

www.otley-sailingclub.co.uk

We are currently organising a horse

riding activity in the Midlands area

and a Calvert Trust break in the

South of England so please get

in touch if you would like further

information.

More photos - PTO

Guide Dog

Experience

Day

Family Support Manager, Iram Shah

attended a Guide Dog Experience

Day at their training centre in

Leamington Spa on the 1st April

2015 with one of our members

(family pictured below). There were

lots of activities during the day

including a touch tour of the centre

and an Easter egg hunt.

More information about Guide

Dogs and their work can be found

via their website:

www.guidedogs.org.uk

ASUK Family Sailing Trip May 2015

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ASUK Family Sailing Trip May 2015

Let’s get Moving

with the

ASUK Bike Club!

The ASUK Bike Club is now stronger

than ever, through Sport England

funding we have been able to

provide another 8 tandems and

specially adapted trikes for families.

Cycling is a great way to exercise

and exercising has many benefits

- it can ease muscle aches and

pains, reduce stiffness, and increase

flexibility, balance, strength and

mobility. It can also help you get

fitter, have more energy, lose

weight, lift your mood and most

importantly, it can be fun!

Alström syndrome makes it

especially difficult for you to

maintain a healthy weight so

regular exercise is really important

for you. Exercise programs work

The Seeing

Ear Poetry

Competition

You may remember in our

November e-newsletter we

published an article about the

Seeing Ear Poetry Competition.

Many of you entered and your

poems are going to be published in

the anthology, well done!

You can see all the winners at the

link below:

www.seeingear.org/home/theseeing-

ear-poetry-competition/

The poems were written to

commemorate the 100 year

anniversary of the start of the 1st

World War and are published below.

Twins Katie and Hannah Beck

(photo above.) are also having their

poems about ‘winter’ published,

which we will be published in our

ASUK winter newsletter.

Who will?

Who will…March up the hill?

Who will…Fight the men?

Who will…Win the war?

Will you my laddie?

By Hannah Beck, aged 8 years old

Who will?

Who will…March all day?

Who will…Dig trenches all night?

Who will…Wear wet dirty clothes?

Will you my laddie?

By Katie Beck aged 8 years old

The Unidentified

Rain poured harder that night as if it was

sad to see innocent brave men die.

They laid in lines of ten, men young and

old were at their fatal end.

Telegrams sent to every family with the

bodies of the brave.

Some were left. They were on their own

now, in the pouring rain in a foreign field.

All because no-one could see or even

remember a glimpse of who they once

were.

They lay in holes and bomb shell holes

alike, all in foreign mud and drowned in

their own blood.

They were far from their true home

where their loved ones and friends

remembered the soldiers.

But one was given the chance. To return

and show in the glory of the whole of

Britain that he and only he will represent

the lost men.

Ones who died in their duty to England,

who never came back.

He is always there. Now, in the Abbey

for his spirit is the one shoulder everyone

can cry on.

By Hannah Bromley-Challenor aged 14

years old

(pictured with Mum, Susan)

best when tailor-made for an

individual, taking into consideration

specific needs, abilities, and

difficulties. Tandem cycling is ideal

as it brings people together of all

abilities and enables people with

dual sensory loss to cycle alongside

their peers. The other big advantage

of a tandem is togetherness. Riders

of differing strength don’t lose one

another, you can keep up the chat –

including when the road is too

narrow or busy to ride side by side

– every mile of the ride and every

hill is a joint experience and a joint

achievement.

We currently have spare tandems

and tag-alongs available if anyone

is interested in getting involved.

Please get in touch if you would

like to know more.

THANK YOU to Our Fantastic Fundraisers!

A HUGE THANK YOU to everyone www.justgiving.com/BrianVasey/ all of you for donating. Alström’s

who continues to support our great www.justgiving.com/David-Family Rock! xxxxx”

cause! The numbers of people LuntAlstromuklm2015/

being diagnosed with Alström Brian (pictured below) gives his Thank you to Kayleigh Lewis who

Syndrome is increasing so continues to make and sell

please get in touch if you candles and crafts in aid of

would like help to arrange

ASUK, raising a wonderful

a fundraising event; we also

£125.00 well done

have spaces available in the

Kayleigh!

Silverstone Half Marathon

and the Bupa 10K Marathon.

Thank you to the Hough

These funds raised really

family and the St. Agnes

do make a huge difference

Community in Cornwall for

and ensure we can support

helping raise funds for ASUK

families throughout the year.

during the Victorian Fair on

You can donate to Alström

the 25th May 2015. The cute

Syndrome UK securely

and cuddly giant Bolsters

through or Just Giving

were on sale at £15.00 each.

page www.justgiving.com/

Profits go to our great cause

alstromsyndromeuk/ or

so please get in touch with

send cheques made payable

Michelle Hough today to

to Alström Syndrome UK

own your very own Bolster

to our registered office 31

and make a huge difference

Shearwater Drive, Torquay,

to the lives of people

Devon TQ2 7TL

affected by this devastating

Well done to our fantastic

condition.

runners Brian Vasey and

David Lunt who took part in

the Virgin London Marathon

on Sunday 26th April 2015 in aid of

ASUK. Jointly they have raised over

£2,000.

It is still not too late to show your

support through Just Giving.

thanks to everyone who supported

him “Hi you amazing people I

completed the London Marathon

in 4hrs & 33Mins. It was a brilliant

experience and I’d like to thank

CONTACT:

michelle.hough@alstrom.org.uk

01872 553067

The Hough family

The Giant Bolster now has his very own Facebook page!

ASUK Chair, Michelle Hough and

her Daughter Millie have been busy

creating the cute and cuddly Giant

Bolster!

You can now keep up to date with

his exciting travels at:

facebook.com/GiantBolsterappeal

The Giant Bolster is a legendary

character said to live near the

cliffs of St. Agnes in Cornwall and

is a popular character in village

parades.

Coach Seats for Great Causes

A HUGE THANK YOU to Williams They have already raised awareness

Travel who have chosen ASUK as through a coffee morning and raffle

their charity to support through which raised £52.50

their Coach Seats for Great Causes http://williams-travel.co.uk/

Scheme. Their Summer 2015 All profits will support the ASUK

holiday brochure is out now and appeal.

for every holiday booked £1 will be

donated to ASUK.

You can now own your very own

limited edition 25cm Giant Bolster

priced at £15 each.

For more information or to place an

order please contact:

Michelle Hough on:

01872 553067

Email:

michelle.hough@alstrom.org.uk

Cardiomyopathy

Association is Changing

The Cardiomyopathy Association

is changing its name to

Cardiomyopathy UK and is having a

new look.

The changes are being introduced

over the next few months and

people affected by cardiomyopathy

will notice differences in all their

communications, including their

website, cardiomyopathy booklets,

their magazine and promotional

items. You can find out more via

their website :

www.cardiomyopathy.org/

Cardiomyopathy-change.html

Book now for the Cardiomyopathy

Information Day in Leeds on the

31st October 2015.

Find out about heart conditions,

treatments and latest research as

well as meeting families who are

affected. Call Sarah Dennis from

Cardiomyopathy UK on:

01494 791224

to secure your place today.

Charities Registered Office: 31 Shearwater Drive, Torquay, Devon TQ2 7TL

Tel: 01709 210151 / 01803 613117

Email: info@alstrom.org.uk Web: www.alstrom.org.uk Facebook: www.facebook.com/alstromsyndromeuk

Twitter: @AS\_UK Donate: www.justgiving.com/alstromsyndromeuk With your help, we have hope

Alström Syndrome UK is a registered charity, number 1071196 - Registered Company number 3557191