

Alström Global Conference

3rd December 2022



ASS.A.I. ONLUS



Be Inspired Be You Be Together



Bringing the Alström Community Together

Be Inspired. Be You. Be Together

In true virtual style, with over **100 registrations...** the Alström community came together; to learn from one another and share their stories at the 2022 AS Global conference. This year it was bigger than ever with families and professionals from **24 countries** joining online.

The aim was not only to bring the community together but provide a global insight through presentations from families, researchers, and clinicians from around the world.

We know that having an ultra-rare condition like Alström Syndrome can sometimes be a lonely place; feeling that no one really knows what you are going through. The global conference is an opportunity to talk and learn together. It shows us all the strength, resilience and support for each other throughout our ultra-rare community.

On Saturday 3rd December 2022, the day started with a warm welcome from **Ann Chivers**, AS Global Executive Director this was followed by presentations from Germany, Italy, the UK, and America who shared their knowledge and experience.



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Alström Syndrome UK

Strength for today, hope for the future

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The first session of the day, centred around a Dad's ambition to provide support and research opportunities in Germany. **Bernd** shared his story of how his 10 year old Son Ben, creates artwork that is joyful; inspiring others to follow their dreams.

Bernd explains that he felt he couldn't accept doing nothing after his Son, Ben was diagnosed at the age of 4. So he shared his emotional journey to find more families affected by AS in Germany to try and support them through research, awareness raising and building a supportive network.

Bernd said that Ben's art captures peoples imagination, raising awareness to empower others to be ambitious and do what they love doing.



BEN.

BEN'S ART



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Dr Pietro Maffei, Consultant Endocrinologist from Padua University in Italy led next with his popular session 'Live Life on the Med!', all about the benefits of following a Mediterranean diet. Pietro shared his top tips of how discovering the delicious and colourful fresh food of the Mediterranean can help keep you healthy.



The diet originated from Southern Italy and Greece in the 1960's, where Physicians began to recognise the link between fewer cardiovascular disorders in those who had fresh, good quality foods.

They studied the diet; it centred around an abundance of fresh fruit and vegetables, wholemeal pasta, bread, grains and olive oil as their main source of fat. Dairy and cheese was eaten in limited amounts. Fish and white meat was eaten several times a week, whereas red meat was restricted to only a few times a month. Sugary sweets and sweet bakery products were very limited.

The concept was simple, no calorie counting or complex tracking and no foods were forbidden. Using herbs, spices, garlic, onion and lemon to flavour food.

Pietro shared some statistics which showed the importance of education and a whole team and family approach to eating well.

Pietro also stressed the importance of physical activity alongside a Mediterranean diet to look after your health and wellbeing.



Next we welcomed **Professor Rick Steeds**, Consultant Cardiologist, Queen Elizabeth Hospital, Birmingham, Deputy Director of Clinical Research, Institute of Cardiovascular Sciences and the Chair of the AS Medical and Scientific Advisory Group.



Rick took us on a tour of the research happening across Europe.

Rick emphasised the amount of growth in published papers about research into AS. This is mainly limited to single centre studies; either describing clinical manifestation or in basic research. Few studies are analysing therapy or disease.

It was fascinating hearing all the developments and interest about AS around the world, the main points from his session were:

- Heart transplants may have many limitations but it still remains an option
- Future drug therapy may assist weight reduction for those with AS
- Basic research into the ALMS1 protein indicates that this protein has an important role in the cell cycle
- Identification of a potential new therapy for insulin resistance in France
- One study showed a higher bone density on those with AS, related to reduction in bone turnover and this may be related to insulin resistance
- Audiology research indicated that 94% of those with AS had dual-sensory loss, their hearing deteriorated with age at a rate of 1.23 dB per year
- Research indicated 74% of those with AS had buccofacial apraxia which may be why patients often struggle to give the puff on a lung function test.

Rick discussed the development of the AS Medical and Scientific Advisory Board, who had met twice in 2022 to look at worldwide collaborative work and identify gaps in research so we can look at ways to improve diagnosis, care and treatment for all affected by AS now and in the future.



The next presentation was given by **Professor Rob Semple**, Chair of Translational Molecular Medicine at the Centre of Cardiovascular Science, University of Edinburgh and **Eleanor McKay**, PhD student in Cardiovascular Science. They gave an overview of their research into the role that unhealthy fat tissue plays in diabetes, heart and liver problems that are often seen in people affected by AS.



Eleanor explained the use of mice to study heart failure and diabetes for those affected by AS.

The first part of the research looked at understanding heart failure. When they looked at the mice with the ALMS1 gene they had all the symptoms apart from heart failure, only the mice who were over 30 years old and female showed some markers. Therefore mice aren't a viable option at the moment for investigating heart failure.

The second part of the study looked at why many people affected by AS have early onset and severe diabetes. The evidence showed that fat tissue looks damaged in AS and the importance the fat plays in health and energy. The results showed that if AS mutations are only in the fat tissue, the mice would still develop diabetes and they still had a fatty liver, caused by 'fatty failure'.

Two journal articles are planned to be published looking at the heart and metabolism.



The next presentation came from **Jonny Eintracht**, PhD student from Great Ormond Street Hospital, who gave an exciting overview of the development in eye research and how this may benefit people with AS in the future.



Jonny explained the important role that cilia have in our bodies and that eyes can't send signals to the brain without cilia. Unfortunately, these cilia don't work properly in people with AS and currently there is no treatment to manage this. With 3 billion letters that make up our DNA, the gene mutation is like a spelling mistake. 40% of ALMS1 mutations are 'stop' mutations and two drugs have been tested on skin cells in the laboratory to see if they can read through these stop signals.

The drugs Ataluren and Amlexanox were tested and it was found that these drugs may be able to rescue the protein and make the cilia work again.

The study has been published in the eBio Magazine, which is part of the Lancet Discovery Science publications.

Future studies will look at expanding the treatment to more patients and looking at clinical trials to build upon the existing data.



The next three talks shared some insights about living with Alström:

First to present was **Melissa (ASUK Trustee) and Alisha** with their presentation called 'Don't wrap me in cotton wool!' – looking at a young person's experience of Alström Syndrome. Alisha suggested some advice for Moms, Dads, and all...



They both gave some insightful top tips:



Top tips to help children or young people (CYP)

- Tell CYP about Alström when a parent or carer thinks they can understand some things about it
- If the CYP are seeing lots of doctors, they should know why they are seeing them
- If the CYP are asking questions, tell them the answers and be honest! You can't hide everything from the CYP all of their life!
- If a CYP gets upset, you have to learn to comfort them - sometimes that means leaving them to think about things on their own
- Find ways to help the CYP - anything is possible to do - find a way round things, it may take longer! Stay upbeat and positive. Talk to other people with the condition, listening to them can be helpful.

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A young person's perspective of Alstrom Syndrome

Don't wrap me in Cotton wool!

TOP TIPS for someone newly diagnosed

parents
VI specialist
ASUK
school or college

♥ Be yourself!

♥ Have lots of people to support you

♥ Share your feelings don't let them build up

♥ Get a medi-dose box

♥ Get a FIT-BIT or similar to encourage activity

♥ Keep healthy: eat well & drink plenty

♥ Don't let A.S. hold you back - set small realistic goals

for mums + dads

♥ Give me space + time to talk

♥ Explain AS to me

♥ Make others aware of AS

♥ Connect me and the people who work with me

♥ Let me take control and do things for myself

♥ STAY STRONG!

we meet online

CULTURE SPARKLE SISTERS

Alisha

Melissa Crowland

thinkbigpicture.co.uk

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Dr Richard Paisey, Honorary Consultant from Torbay Hospital and Alström Syndrome UK Trustee, explored the evidence behind why exercise is vitally important to keeping fit and healthy with AS. Richard, was joined by our very own **Trevor** (Advisor to the Board of ASUK) who shared his experience of using a fitness tracker and how this has motivated him to exercise more.



Dr Paisey explored the global context of diabetes and asking the question **'can a culture stop a pandemic?'** well, from his presentation you would come up with the conclusion that it can! Looking at how our culture, around what we eat and drink has a huge effect on our health and wellbeing.



Dr Paisey used the example of Cuba; when the Russians withdrew in 1990, austerity hit and people ate much less, China also donated 1 million bicycles for the country. In just 5 years obesity, type 2 diabetes, coronary disease and cancer all reduced. But from 1996-2005, aid stepped in and provided more rice and beans per family and this had the effect over these years of a decline in the country's health once again. When you contrast families affected by AS in Italy and Canada, Italy have a lower BMI and only 13% of people develop diabetes by 18 years old whereas in Canada the BMI is much higher and there is a nearly 100% chance of diabetes by the time you are 18 years old.

Dr Paisey reiterated the good results of the Mediterranean diet and combined with exercise can help sustain your health and wellbeing and remission from diabetes may be possible.

Trevor and Dr Paisey also discussed the use of a fitness tracker and how this technology can motivate and encourage you to exercise.



Next, we heard from **Hassan**, sharing his story about riding his tandem to trek across Europe. Hassan was joined in his presentation by his Dad, **Kez** who shared his insights into how difficult it can be to let go as your child grows into an adult, but how it can be positive for the whole family if this can be done in a supportive way.



Hassan gave an inspirational talk about his journey to Calais and his tandem bike ride from Bruges to Amsterdam and, although it was very hot and lots of sore bums, they had a great time!

Hassan's motto is "feel the fear and do it any way".

Hassan and Kez explained that this is only possible due to the wonderful PA's that are in place to ensure that Hassan can achieve his dreams and also Kez as a parent, can feel reassured and confident for his Son to be independent too.

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A young person's perspective of Alstrom Syndrome

cycling across Europe

Trekking across the universe!

BRADFORD - DOVER - CALAIS - BRUGES - AMSTERDAM

100 miles!

27

Kez + Hassan Hayat

set off with 3 friends

FUNDING £ ASUK funded my TANDEM!

my bum is sore!

overcoming this beast of a disease

It's exciting & fun!

we spent 3 HOT days cycling

HASSAN + ADAM

I love cycling

It keeps me fit

helps to manage my diabetes

Now Hassan is a super young man

Kez:

as a parent it's so important to have faith in them....

and know they have great support

and can use technology to keep safe

It's not EASY to let go

but it's POSITIVE for the whole family

thinkpicture.co.uk

Bringing the Alström Community Together



We were delighted to welcome **Chase Palmer** from Alström Syndrome International (ASI), **Marina Valenti** from ASSAI in Italy, **Bernd Rosenbichler** from AS Germany, and our very own **Kerry Leeson-Beevers**, Chief Executive from ASUK to share their updates on what is happening in their corner of the world.

Chase started by explaining that he would be taking over from Robin Marshall, and after Robin's 30 years at the helm, they will be big boots to fill! Chase discussed their work with the Food and Drug Agency USA, to learn more about the experiences of people affected by AS, and also their work with AS Spain to support families with medication recommendations. Their next conference will take place in Baltimore, USA in October 2023.

Next we heard from **Marina**, who showed fabulous images of a recent family get together over 3 days to celebrate Fabio's life, who sadly passed away. Fabio was AS Italy's Secretary and his vision was live life to the best. The DeafBlind Association also provided a Haptic Communication workshop to show how useful signs on the body can be for people who have dual sensory loss. Their next family conference will be on the 28th October 2023 in Lake Como/Lecco.

Bernd explored his AS Germany dream and how only a year ago, he was starting from scratch, looking for families, organising families and looking for research initiatives and collaborating together. The recent meeting with ASEU in Tübingen in Germany was an insightful day of learning from one another and exploring future research initiatives.

Kerry spoke about the ongoing developments at ASUK from the AS multi-disciplinary clinics, the research initiatives and working more broadly through the Breaking Down Barriers project and working with policy makers and pharma to implement real change for all rare diseases and diverse communities. Kerry reiterated that collaboration is key to all this work and it is only by learning from one another and working together that the true progress will happen

'AS is too small to work in isolation'.



Lastly, **Ann Chivers**, Executive Director, AS Global thanked all of the presenters and emphasised the commitment of AS Societies and Groups around the world to work and learn together to further research and support services for all those affected by Alström Syndrome.



Kerry and **Ann** shared the exciting news that ASUK will be having a silver anniversary. **ASUK will be 25 years old next year!!** – it is going to be a year of get togethers and celebrations starting with a family gathering in the UK in April.

Ann described that there will be celebrations and activities throughout the year, including a residential for children and young people in April 2023. Ann also encouraged everyone to get involved, by sending in your creations; your pictures, paintings, collages, tactile art and share your **'Rare Moments'** by the 31st March 2023. This creative celebration is open to everyone, including parents, carers, siblings and professionals and the creations will be displayed in an online art exhibition on the 1st May 2023 to celebrate **ASUK turning 25!**



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Thank you

On behalf of ASUK, ASSAI, AS Germany and ASI we would like to say a big thank you to all the presenters who contributed in making this third virtual conference such a success; and of course a huge thank you to everyone who attended, the more we know the better the treatment and management of Alström Syndrome. **Alone we are strong, together we are stronger!**

Why not listen again? — you can find further information, including the conference recordings on the ASUK website: <https://www.alstrom.org.uk/as-global/#AS-Global-Conference-2022>

What's next?

Well, we will be collaborating with our friends, families and professionals from around the world to host webinars throughout the year on topics to bring the AS community together. From discussions around ear health, to staying healthy, through to ways to stay independent and ways to keep everyone connected around the globe. If you would like to be involved, present or send us an article then please get in touch with **Catherine by email catherine.lewis@alstrom.org.uk**

We look forward to seeing you all again soon!

“Thank you all. Great conference and brilliantly organised.”

“The scientific information was very interesting and helpful”

“Thank you for putting this together, it was a great experience and an honour to present at the event”

It's all about team work!





The numbers!



106 Registrations



From **24** Countries



17 Presenters



8 Family 'experts by experience' speakers



1 Medical & Scientific Meeting



10 Hours of collaborating knowledge & experience

100%

Would or may attend another AS Global Conference



9/10

Average score for how much you enjoyed the conference

