



Journey to Diagnosis

This year's conference brought families together to map their diagnostic journey. Our aim was to learn more about how and when children and adults have been diagnosed, so we can take positive action to target our future awareness campaigns to ensure families affected are diagnosed sooner.

We want to ensure people affected by Alström get the support and medical treatments that they need to live a fulfilled life. The longer the wait for a diagnosis; the longer families wait for support and specialist treatments.

We want to change this!

Ann Chivers (ASUK Chief Executive, pictured below) opened the conference with a warm welcome to everyone who had travelled far and wide to attend. Ann gave an insight into why understanding families journey to diagnosis is vitally important.



Ann facilitated a workshop which involved families sharing their stories about their journey to diagnosis; not one story was the same! For some their diagnosis was quick, but for many the journey was a long and difficult process, symptoms mis-diagnosed or missed all together.

Personal stories have the power to make a difference now and in the future.



Dr. Tarek Hiwot (*Consultant in Inherited Metabolic Disorders*) presented the remarkable history of Alström over the last 21 years, describing the progress that has been achieved by working together with European and International professionals. Tarek talked about the current clinical trials in the UK and ongoing research initiatives with European partners, as well as the current thinking on future research projects.



Tarek spoke about Alström affecting multi-organs and as a result of this patients should be treated holistically with a multi-faceted approach. He stressed the importance of a healthy lifestyle for everyone with Alström.

Tarek highlighted how following a healthy, balanced diet and taking regular exercise, can help to manage the condition. ASUK are here to empower families to lead a healthy lifestyle and offer support to access activities in their local area. Funding applications can be considered by ASUK for exercise equipment or activities such as gym membership.

One of the highlights of the day was dancing to a newly created exercise routine called **'the Stroove'** this was great fun and it was fantastic to see families, the ASUK team as well as clinicians taking part & dancing along to the music!

"What a great conference. I've learnt so much that I didn't before, it was one of the best rare disease conferences I have been to. The atmosphere, bond with the families and the team was wonderful."

Key points from Tarek's presentation:

- The UK has a world class Alström clinical service
- It is important to maintain a healthy lifestyle to manage the condition
- Holistic, multi-faceted approach is needed
- Remarkable progress has been achieved in the last 21 years
- Intense research is now needed to find further treatments.

Tarek offered possible hope for the future through gene therapy treatments. Although, due to the size of the ALMS1 gene further research is needed.

Let's gain a deeper insight into Alström.

Liz Loughery, ASUK Project Manager (*pictured below*) introduced the **Alström Syndrome UK Patient Registry**.



Liz explained how families will be able to get involved and thanked those who had contributed towards the registry development. The aim is to create a registry which families will be able to participate in and regularly update with their information. This will help to build a complete picture of Alström, where scientists are able to develop research projects, which we hope will lead to improved treatments and new initiatives.



Strength for today, hope for the future

Interactive workshops were provided throughout the day.

Kerry led an invaluable workshop for new families, answering questions and introducing ASUK. ***“For Kerry to share her experience, helped us understand not just the condition but the challenges and barriers that others feel and how much stronger it makes you. Kerry is an inspiration and I really benefited from her workshop.”***

Members Alex and Haris (pictured below) shared their insights into **useful technologies and gadgets** that support them to live independently. Alex described the RNIB pen friend, which is an audio-labeller so you can label anything and everything. This sounded very useful, especially with items which feel similar such as medicines, CD's and clothing. They gave insights into the everyday kit they wouldn't be without, such as the liquid level indicator, which you attach to the side of a cup and an alarm will beep when the liquid is close to the top. Their workshop went down such a storm, that they are considering doing a regular top tech article for the ASUK website and newsletter, so watch this space!



Hassan and Kez (pictured below) discussed their **healthy lifestyle** and how they keep themselves motivated. Hassan has had a new lease of life since joining his local cycling group. He now travels all over the place seeking new cycling adventures! Hassan is a tremendous inspiration to us all to look at ways we can adapt our lifestyle to help our health.



On the healthy living theme, Gayle from Birmingham Children's Hospital lead a **yoga workshop** to help us learn techniques which can be used anywhere at anytime to relax both the body and mind.



Hazel, from **Guide Dogs** discussed the range of services that they provide. Did you know? You don't need a guide dog to access their services. They provide services for children and young people which include CustomEyes books, assistive technology, grants, mobility training, family support and events and a free sighted guide service. As well as providing Guide Dogs.



Strength for today, hope for the future

The tiger came for tea!



The one thing that families always say they enjoy at the ASUK events is to meet other families and socialise together. Well this conference certainly did just that! The day had a relaxed feel to it, families and professionals had plenty of time to chat, share experiences and reminisce on 21 years of Alström Syndrome UK.

In the evening, the socialising continued over a delicious dinner which was followed by lively music and dancing long into the evening.



Congratulations to Mary and Paddy who won the large cuddly tiger. Mary's shock at winning was priceless! (pictured above right) Along with the raffle this raised **£200**



Thank you

A HUGE THANK YOU to families, professionals and our volunteers who travelled from around the UK and across Europe to attend the conference, supporting youngsters, sharing knowledge and the lived experience of Alström, offering insights and understanding to help us know more!

Thank you to our supporters and generous sponsors, Liminal BioScience and Rhythm Pharmaceuticals. ASUK is grateful for all the support and help in raising awareness of Alström Syndrome and finally thank you to the presenters and exhibitors for taking part and making this conference a truly memorable event—**see you at the next one!**



Full conference information can be found
on our website
www.alstrom.org.uk


Alström Syndrome UK
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