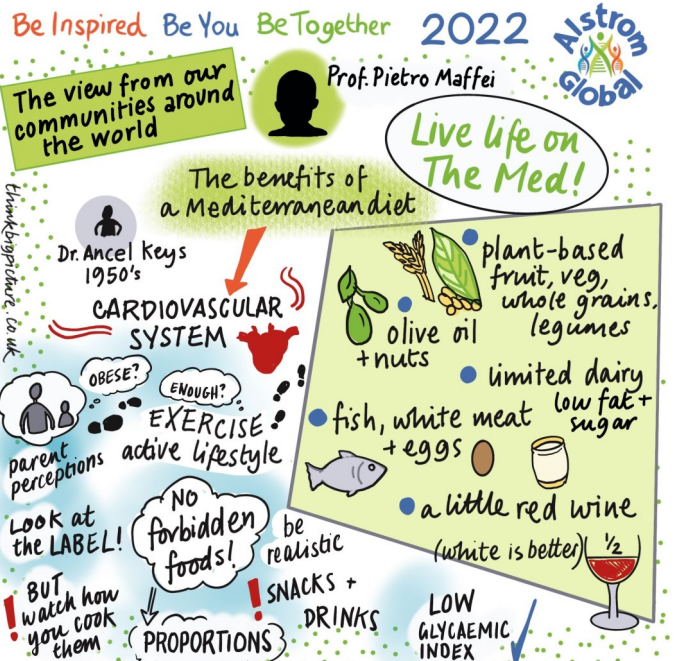
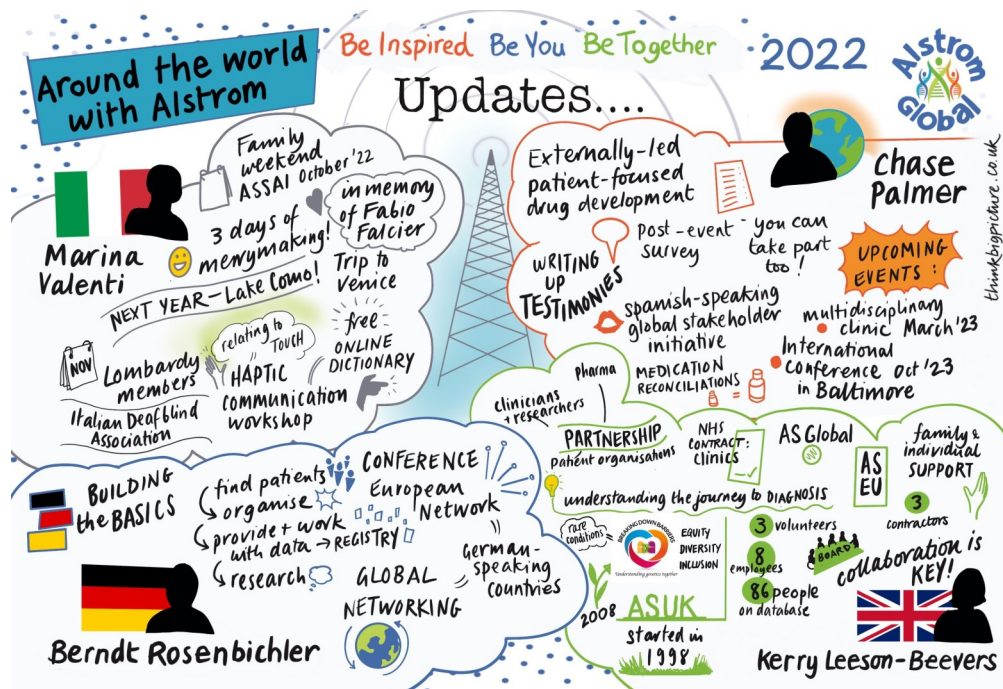




Impact Report
The Knowledge Exchange
2022-2023



Introduction

From humble beginnings...

In 2023, ASUK is proud to celebrate 25 years as a patient support charity.

From humble yet determined beginnings we have developed into a dynamic, family focussed organisation with our people at the heart of all we do.

Alström Syndrome

Alström Syndrome is a very rare genetic condition which can cause progressive blindness, loss of hearing and can lead to; heart and kidney failure, type 2 diabetes, liver dysfunction and associated problems.

The symptoms arise at different stages making diagnosis very difficult.

A diagnosis of Alström Syndrome can be devastating, isolating, and bewildering for parents and care givers. ASUK has an understanding of what it is like, as we are run by families and people with Alström as well as with

the extraordinary support of Clinicians. Which is why ASUK's aims are to provide personalised support, raise awareness, facilitate pioneering research, and improve treatments and management through our UK based AS multi-disciplinary clinics.

We have come a long way in 25 years; from AS clinics and consultations in hotel rooms, to the development of highly specialised multi-disciplinary clinics for children and young people and adults at two leading hospitals.

Reflecting on this anniversary, highlights the huge strides forward that have been made in accelerating the diagnosis for individuals with AS. In 1998, we knew only of approximately 7 individuals affected, to now in 2023, knowing of 92 people affected in the UK. We have seen more people being diagnosed over the past few years than ever before, enabling them to get the specific treatment and management guidance from AS Experts.

Key points:

7 people known to us with AS in the UK in 1998

92 people known to us with AS in the UK in 2023



The art and science of collaboration

In 2021 we helped to develop a global patient registry, enabling the collection of information and data from individuals and their families. The patient registry was achieved in collaboration with CoRDS Coordination of Rare Diseases at Sanford and is slowly gathering momentum with more and more individuals adding their details each year. We are hopeful that this registry will go a long way in helping researchers from all over the world, learn more and understand this complex condition. Our purpose is to improve the treatment and management of AS for every individual affected by this ultra-rare disease.

Following on from this successful international collaboration and with the learning from our virtual working throughout the pandemic, the Board of Trustees decided that it was important to expand and reach out to all of those affected by Alström and their supporters to see what we can learn from each other locally, nationally, and internationally.

With the generous support of Alström Canada, ASI (Alström Syndrome International), ASSAI (Associazione Sindrome di Alström Italia) and as well as the enthusiasm of families, clinicians, researchers, and speakers, we launched the very first virtual Alström Syndrome Global Conference and Scientific Symposium.

We were learning as we went and fortunately, with not too many technical hitches, the response from the AS community was very positive with 183 delegates attending from 23 countries. Not only did it give families the chance to 'virtually meet up', it provided an open forum for Clinicians and Researchers from all over the globe to share updates and evidence about the research, management and treatment of AS.

One of the highlights of the conference was welcoming Qianwen Zhang and Dr. Xiumin Wang, Chief of Department of Endocrinology, Genetics and Metabolism at the Shanghai Children Medical Centre in China.

Qianwen explained about their work, research insights and how they have found 57 patients in China, who are mainly under the age of 18.

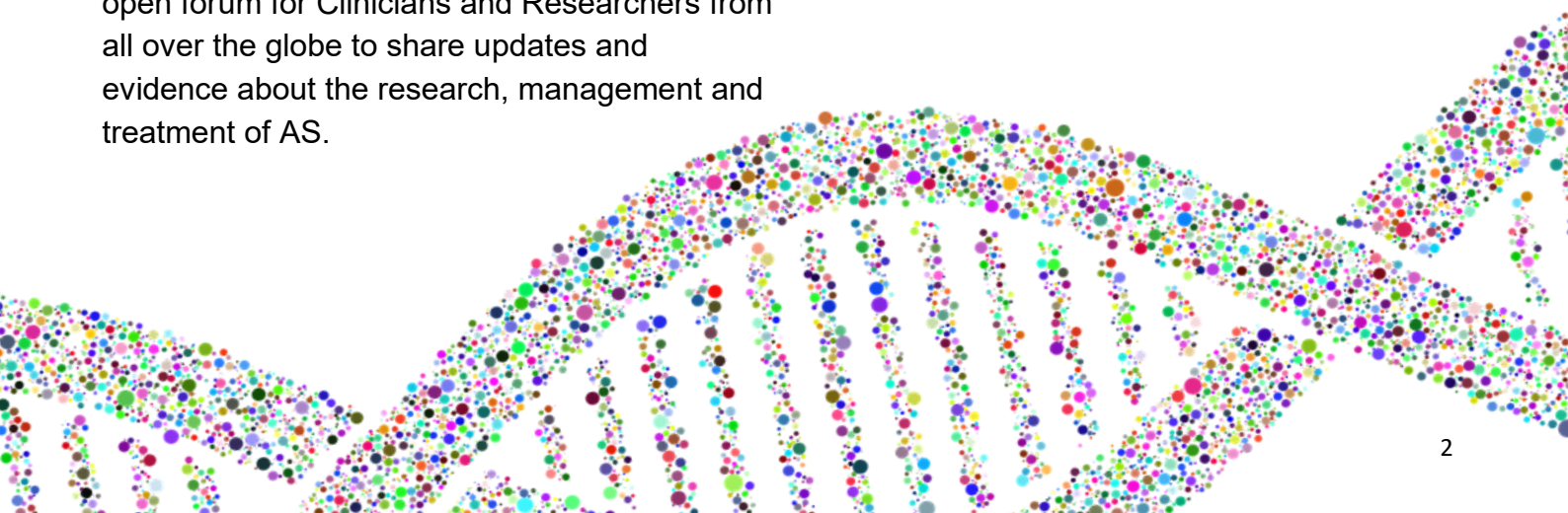
This collaboration has now meant that Qianwen is now coming to the UK to study for six months.

Families were pleased to hear from Bernd, 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 is spearheading getting families in Germany to connect as well as facilitating meet ups for Clinicians.

Building on this promising foundation of collaboration, in 2022 we held our third virtual conference, this time with 117 registrations from 24 countries. Squeezing in so many great presenters and topics led us to work with families to identify what other topics mattered to them and what they wanted to share and learn more about.

With this enthusiastic remit from families all over the world Team Global Alström began work in earnest.



Worldwide Reach

The AS Global Conference in 2022, had attendees from 24 countries:

- Australia

Canada

China

Denmark

France

Germany

Greece

India

Ireland

Italy

Jersey

Norway
- Oman

Peru

Portugal

Republic of Moldova

Romania

Singapore

Spain

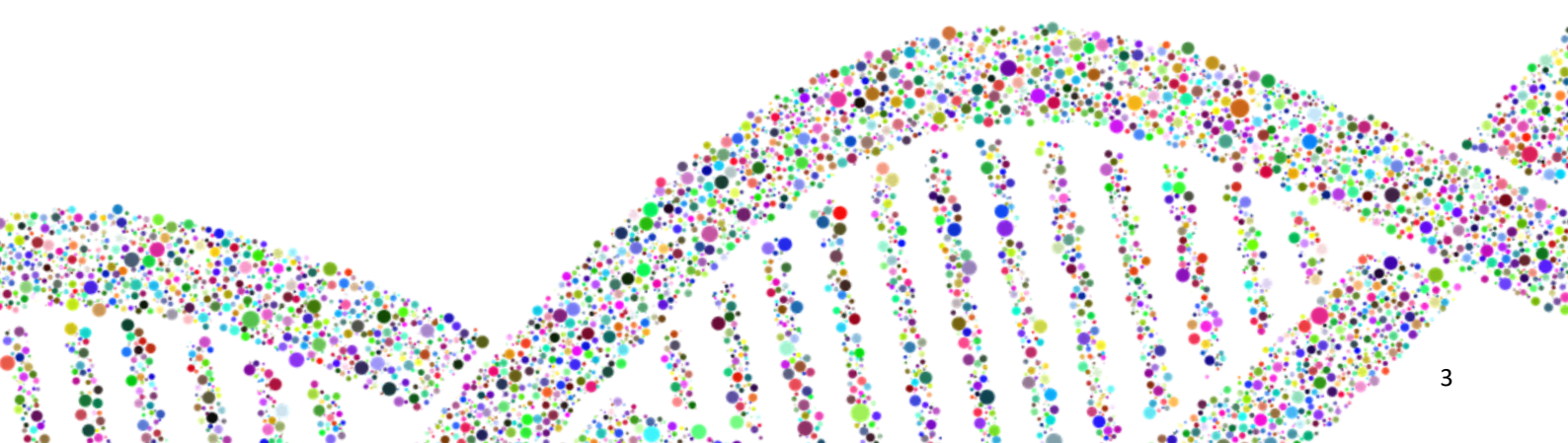
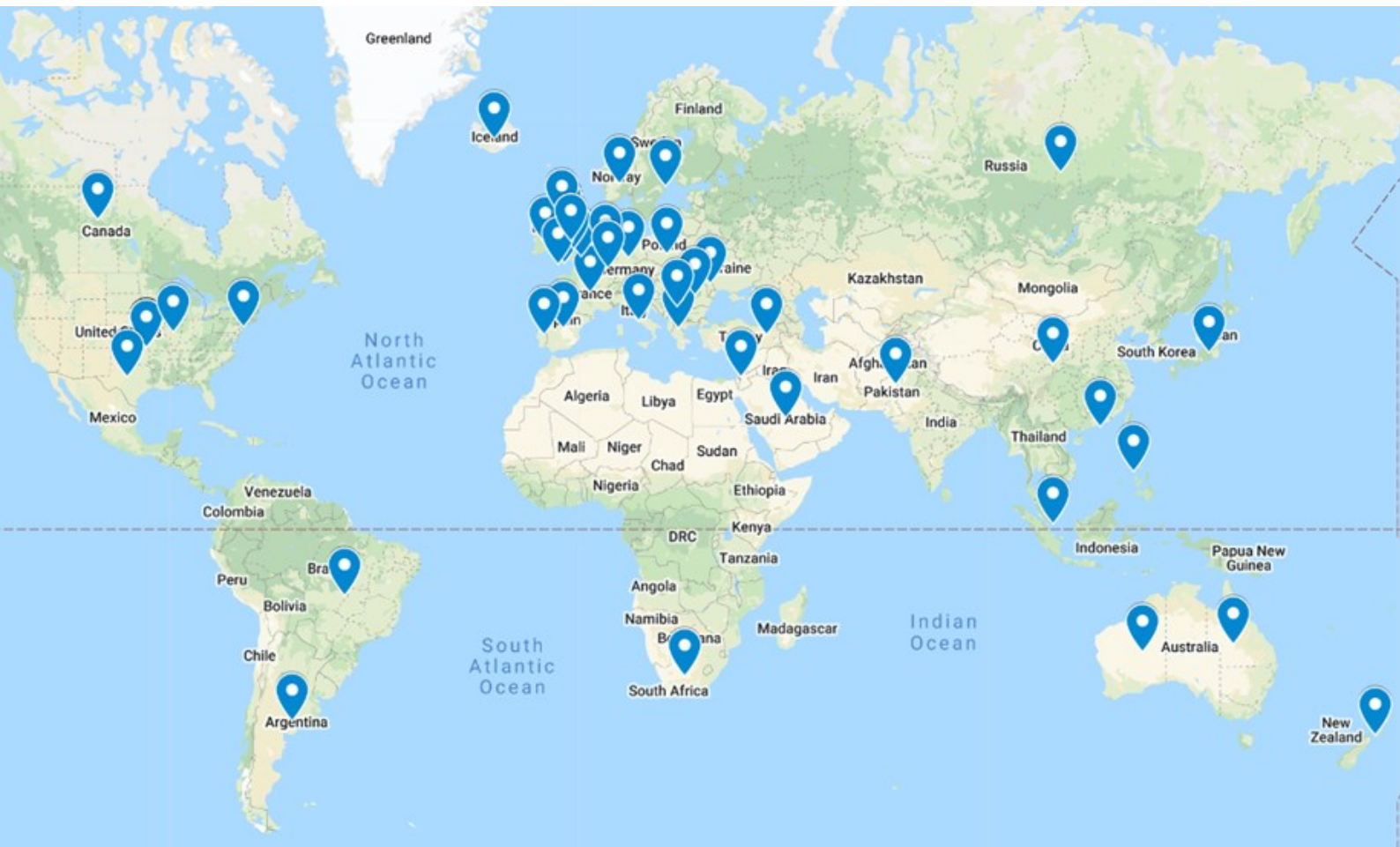
Switzerland

Sweden

Turkey

UK

USA



Designing the programme

With a wing and a prayer, we hosted the first global webinar in February 2022. The topic for our first webinar was 'How I Manage My Diabetes' presented by one of our Experts By Experience Lexi Line and Dr Richard Paisey. We were delighted that 12 families from 12 different countries joined the webinar.

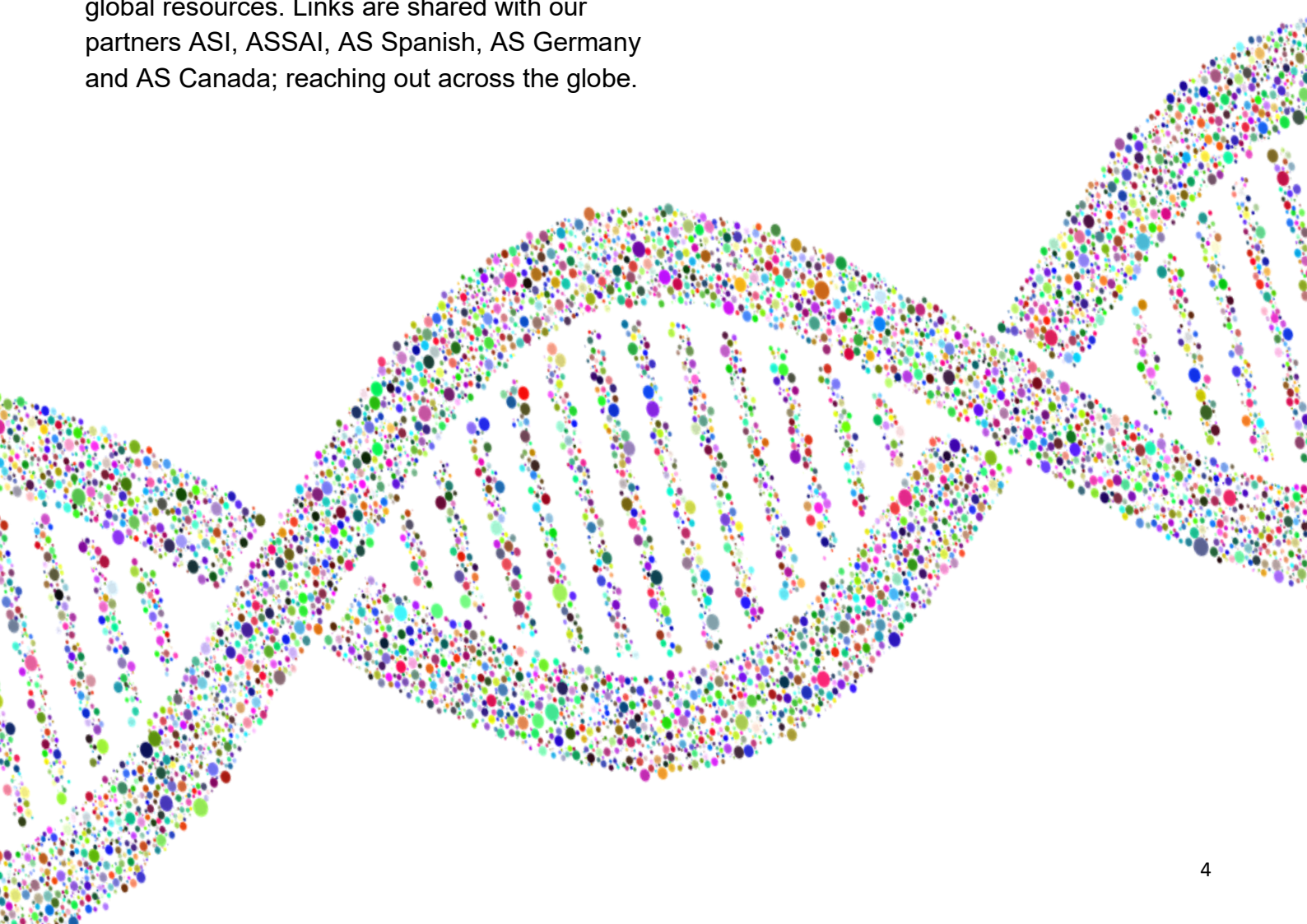
Based on suggestions from families, educators, and clinicians we began what has now grown into a series of monthly global webinars; inviting people to attend, present and encouraging everyone to share information about the programme via social media channels.

The global webinars have developed into stand-alone learning modules that can be shared amongst families, clinicians, and educators and anyone who has an interest in Alström Syndrome. As we continue to learn more about Alström, these bite-sized modules can be updated and accessed immediately on the AS website under AS Global - a dedicated area for global resources. Links are shared with our partners ASI, ASSAI, AS Spanish, AS Germany and AS Canada; reaching out across the globe.

The webinars bring the AS Global community together, no matter where they live in an accessible, inclusive way at a time when people can sometimes feel isolated and lonely. With the global picture of Alström differing from country to country and many people not receiving the expert guidance and support that they need; the webinars are proving an invaluable resource for people to stay in touch with the AS experts and people affected from around the world. Sessions are interactive, with time for questions and the sharing of experiences; the good, the bad and the ugly!

"Today was great. Understandable for parents and patients. If we can't meet in person, this is the next best thing."

Family feedback



Key Challenges

The IT system

At the start of the global initiative, we believed that the technology would be one of the key challenges; the speed or lack of, the accessibility and the platform itself not enabling us to be able to see and talk directly to each other. We experimented with different platforms. Led by two Trustees who are Experts by Experience. Lexi and Melissa tested out different sites to find the most accessible platform that worked for most people. They found the 'zoom infrastructure', that can send invites and enables the sharing of presentations is working well for most people. As the IT develops so will our ability to continue to update and use what works best for our community.

Getting the timing right- International time zones

To be able to give as many people as possible the opportunity to join live webinars we agreed that a regular day and time would work best. Scheduling at 7pm (UK time) on the last Thursday of the month has meant that those countries that have an active AS organisation can reach out to their families, clinicians and educators at a reasonable hour in the day and join online.

For those families in time zones that mean they can't join the sessions live, we record every webinar and develop it into a 'Learning module' that includes; a short film of the online presentation, a Top Tips Information page and links to other related resources.

The Language barrier

One area we are currently developing is how we can provide webinars or more effectively manage the IT platform for AS groups; families and the professionals who do not speak English. We continue to experiment with google translate but wanted to go one step further...

We have collaborated with the AS Spanish Association to provide the platform and administrative support for them to host their first webinar in Spanish.

The first Spanish webinar was all about 'reviewing your medicines', it certainly helped that both Specialist Pharmacists Antonio Ochoa-Ferraro at University Hospital Birmingham and Chase Palmer President of ASI speak Spanish!

This successful webinar will be shared with other Spanish speakers. It offered a trustworthy platform for the Spanish Association to come together virtually.

Continuous Learning

We have plans to host a webinar in Italian, with our friends from the Alström Italian Association and we will continue to invite feedback to ensure webinars can be as inclusive as possible for everyone in the global community.

"Thank you for all your hard work, not only to enable the conference, but to be a source of information and support for Alström families. We loved it all from listening to the Doctor's and dietician to listening to Lexi and Jamie." Family feedback.



Key Learning Points

At the beginning of the webinar programme, we needed to support lots of individuals and families to be able to get online and manage the technology.

As more and more people, have adjusted to virtual meetings and appointments, we now know that the webinars are accessed in different ways; both by PC, tablet and by mobile 'phone. This allows us to make any adjustments to resources that may only be read on a tablet or 'phone. We continue to be advised on what works well for people with AS.

We have developed recognisable templates and short briefings for the webinar presentations, striving towards keeping it simple and as accessible as possible. There are short films as part of the learning modules, these can be accessed online or downloaded, saved and easily shared.

Topics that are suggested for webinars can sometimes be difficult subjects to talk about in a group, therefore where possible we have endeavoured to follow up on a one-to-one basis so that an individual or family can talk about issues privately. Privacy and data protection is important and as each session is recorded it allows chance to edit, ensuring that where an individual or family member doesn't want to appear on screen or be identified we can easily achieve that. Attendees are told at the start of

each webinar that it is recorded so that everyone has the chance to state their own preferences. This has worked well.

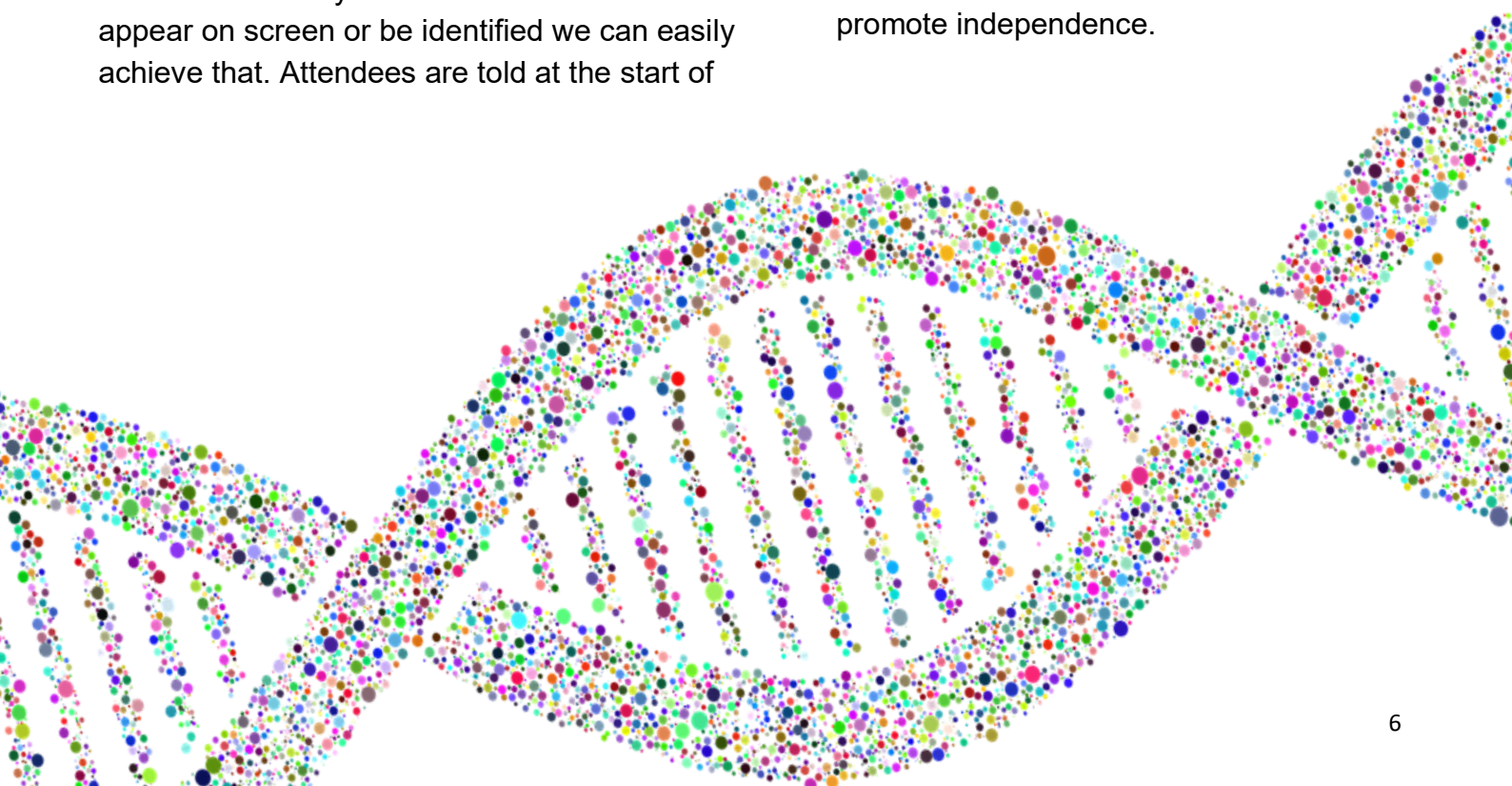
Health, Social Care and Education systems worldwide.

What does good look like?

As these differ throughout the world there has been enormous opportunities for families to learn from each other and consider good practice. This has led to some of the family leaders to begin challenging systems and local services in their own country, armed with knowledge of what good practice can look like. There are no quick fixes but the sharing of information is a small but vitally important step forward to improving our collective knowledge and understanding and giving families the resources to be able to challenge systems.

Resources include:

- Consensus clinical management guidelines for Alström Syndrome
- The Alström Syndrome Medical Handbook
- Top tips from families and professionals around common topics, such as healthy eating, exercise and gadgets to help promote independence.



We didn't know, what we didn't know!

There have been many unexpected consequences of this global initiative and how the AS community are collaborating to make an impact.

The Knowledge Exchange

We call this impact 'The Knowledge Exchange' and cite examples of how, collectively, we are making a difference.

- Through the promotion of the webinars and the Scientific Symposium and Conference, we 'discovered' that there are a high number of related families affected by Alström in what was an isolated village in Palestine.

We have since worked with Dr Reham Khalaf-Nazzal, Associate Professor from Arab American University in Jenin and Doctor Emma Baple Clinical Senior Lecturer (Genomic Medicine) and Consultant in Clinical Genetics from Royal Devon & Exeter NHS Foundation Trust linking them together with Professor Tim Barrett, AS paediatric Expert in the UK. These fruitful discussions have enabled Professor Barrett to offer advice and information to Clinicians and their teams in Palestine to share learning and knowledge.

- Attendees at webinars have found AS Ambassador leaders in Europe and elsewhere of Alström initiatives, (mostly Moms and Dads) and joined the associations.
- Many of these AS Ambassadors are now forging ahead; raising awareness, sharing information, challenging services. Working together and hopefully feeling less isolated as they support their family member with AS and other siblings.
- AS Leaders have linked together Clinicians in Europe organised to meet up and exchange knowledge and expertise.
- Through the Global conference we learnt about the experience in a Chinese province which has led to a Research Doctor from China coming to the UK to study for six months.
- Over the past year, (March 2022 – March 2023) over 1,000 people have visited the dedicated AS Global website page on the ASUK website.
- To celebrate ASUK turning 25 and Worldwide Alström Awareness Day, the first AS inaugural lecture was provided by Leading Clinical Expert Dr Richard Paisey 'Alström Syndrome: The Past, Present and Future' and was attended by 45 people from 7 Countries.

We're delighted that so far the webinar and conference films have been viewed by around 1,300 people.



Conclusion

Collaboration is key

With the expanse and development of worldwide research initiatives, genome sequencing, awareness raising and the increasing numbers of experts with a passion for AS, the global network is beginning to flourish.

As Doctor Richard Paisey stated in his Global Lecture:

“It is only by working together that we can improve the diagnosis, research, treatments, and management of Alström Syndrome”.

Working and learning together with families, people with Alström, Clinicians, Researchers and Educators is vital to discover more about this ultra-rare condition.

Thank you

Our thanks go to everyone who has helped shape this first phase of this global initiative; Experts by Experience, presenters, collaborators, families, charity representatives and attendees from all over the world. They have helped co-create and contributed their time so generously to help each other and improve our shared understanding.

Thank you to all from all of the partners in Team AS Global:

Chase, Eric, Robin, Palmer, Randy, Susana, Bernd, Pietro, Marina, Ann and Catherine.

Finally, to everyone who has joined in, please keep those ideas coming in!

We hope this snapshot will shine a light on what we can achieve together and how we are collectively and with kindness and generosity developing a truly collaborative global network.

Further Information

To find out more or catch up on any webinars you have missed, then head across to the AS Global website: <https://www.alstrom.org.uk/as-global/>





Be Inspired Be You Be Together



ASS.A.I. ONLUS



AS Canada



ASEU

Alström Syndrome Europe



Alström Syndrome UK

Strength for today, hope for the future





www.alstrom.org.uk/as-global/