Health Equity & The UK Rare Diseases Framework

Focus Group Findings
21st November 2023

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INTRODUCTION

About Alström Syndrome UK

Alström Syndrome UK (ASUK) was established in 1998 and is a registered charity providing information and support to individuals and families affected by Alström Syndrome (AS) and to the service providers working with them. ASUK works in partnership with Birmingham Women's and Children's Hospital and the Queen Elizabeth Hospital, Birmingham to deliver a highly specialised service, funded by NHS England. As a patient led organisation, the needs and wishes of people affected by AS remain at the heart of everything we do. We aim to; provide personalised support, raise awareness, conduct pioneering research, and enable better treatments and monitoring through the AS multi-disciplinary clinics.

ASUK co-founded Breaking Down Barriers in partnership with The Sylvia Adams Charitable Trust.

About Breaking Down Barriers

Breaking Down Barriers is a network of over 70 organisations providing support to people affected by rare and genetic conditions. We work together to learn, develop, and share good practice. Promoting equity, diversity and inclusion (EDI) is at the centre of what we do. Our Experts by Experience Advisory Group help us to understand the lived experiences of people from diverse and marginalised communities who are affected by rare and genetic conditions. We create safe spaces for learning and collaboration, deliver training and run a community outreach project. We unite and strive for equitable access to services and support for all.

www.alstrom.org.uk

www.breaking-down-barriers.org.uk

ADDRESSING HEALTH EQUITY WITHIN THE UK RARE DISEASES FRAMEWORK

Breaking Down Barriers (BDB) and the Department of Health and Social Care (DHSC) have been working together to make sure health equity is addressed within England's Rare Diseases Action Plans, following the publication of the UK Rare Diseases Framework.

The UK Rare Diseases Framework was published in January 2021, setting out a national vision on how the UK will aim to improve the lives of people living with rare conditions. The Framework was developed based on the findings from the National Conversation survey which was launched by Baroness Blackwood in November 2019. 6,293 people responded to the survey, including over 5,000 patients, families, and patient organisation representatives.

There are 4 key priorities and 5 underpinning themes, and each nation of the UK developed an Action Plan to show how these priorities will be met.

4 Key Priorities

- Helping patients get a final diagnosis faster
- Increasing awareness of rare diseases among healthcare professionals
- Better coordination of care
- Improving access to specialist care, treatments, and drugs

5 Underpinning Themes

- Patient voice
- National and international collaboration
- Pioneering research
- Digital, data and technology
- Wider policy alignment

When producing England's Action Plans, work has been carried out by England's Rare Diseases Framework Delivery Group to understand the health inequalities experienced by people affected by rare conditions, and to identify ways to address this through specific actions.

In 2021, BDB and DHSC worked in partnership to run a workshop to understand the health inequalities experienced by people from diverse and marginalised communities, who are also affected by a rare condition.

The following recommendations were identified:

People living with rare conditions need:

- A holistic approach to care and support
- Accessible and inclusive resources
- Be able to develop and maintain trust in healthcare providers

In December 2022, BDB and DHSC organised the first focus group meeting, looking at the progress that had been made in terms of health equity, and plans for future actions within England's 2023 Action Plan.

The 2022 focus group report can be viewed via this link:
Health-Equity-Focus-Group-Report-Dec-22-Final.pdf (breaking-down-barriers.org.uk)

In November 2023, BDB and DHSC organised a second focus group meeting to discuss progress against England's 2023 Action Plan.

The virtual focus group included a diverse group of 9 people with lived experience, plus representatives from BDB and DHSC. The previous Health Equity focus group report (December 2022), background information about the Rare Diseases Framework, and a briefing document provided by DHSC, was shared with the group ahead of the meeting to help everyone prepare for the discussion.

The focus group was chaired by Kerry Leeson-Beevers, CEO of ASUK and BDB and patient representative on England's Rare Diseases Framework Delivery Group.

Kerry discussed the importance of this focus group; to ensure the voices of people from diverse and marginalised communities who live with rare and genetic conditions is included throughout policy and practice. The findings from these focus groups are shared with England's delivery group partners, to support learning and encourage the development of inclusive and representative actions.

During the focus group, a representative from DHSC gave a presentation on progress and plans and asked the group to discuss 4 key areas:

- Health Equity
- Patient and Public Involvement & Engagement (PPIE)
- Mental Health
- Transition

The group explored some of the key challenges, examples of good practice and opportunities for further improvement.

DISCUSSION POINTS

Health Equity and PPIE

S discussed her previous input into PPIE groups and how at times she felt like she was just part of a tick box exercise. There needs to be more consistency and meaningful engagement and involvement with clear actions and feedback. PPIE representatives should be valued, respected, and treated as equal partners. Patients often fear that negative feedback will affect their care. The fear of offending patients can sometimes prevent healthcare professionals from engaging with people from diverse and marginalised communities.

J gave her professional and lived experience of the importance of ensuring easy access to research opportunities. This is particularly important if we are to gain a better understanding of the genomic diversity within the UK population. After her cancer diagnosis, *J* recalls wanting and asking to be part of research, but nothing was acted upon either at the time or at any point since. There needs to be more awareness of the importance of a diverse and representative data set. People need to have access to transparent and accurate information, that is sensitive and representative of our diverse population. Healthcare professionals need to work with patients and communities to build trust and confidence in research.

H gave his experience of wanting to take part in research projects, but how the initial information provided to him was not accessible. He then went on to say how helpful it was when he was introduced to a researcher who could speak Urdu.

K talked about how important raising awareness is and connecting with grass root communities and voluntary organisations, that communities trust. *K* shared his experience of the barriers he experienced within his own family when his son took part in research. He found many challenges to overcome such as religion, culture, and language barriers.

D reiterated the need to engage with grass root community organisations, as often they are undervalued. PPIE should be implemented at every stage, right from the start of any new development and not just at the end when it is often too late.

CC gave an example of a recent conversation with a GP who said that in 20 years of practice they had never referred a patient for genetic testing. They explained that they just treat each symptom as they appear.

A is deafblind and has recently had cochlear implants fitted. She gave an insight into her experience with the health service and how the hospital had provided accessible information, discussed everything with her and included her in all decisions. She shared how this positive experience made her feel empowered and less isolated.

J gave an example of the impact of not being given information or included in decision making. She was diagnosed with cancer at the age of 18 and the Doctor told her there was nothing to worry about. Following the appointment, the Doctor then called her parents to share the diagnosis

and discuss how concerned they were. *J* was upset at the breach of confidentiality but also that she had been made to believe that everything was going to be ok. This had a negative impact on the Doctor-patient relationship and *J*'s trust in healthcare professionals.

On a more positive note, *J* was then treated at a hospital with a young person's unit for people aged between 18-25 years old, which gave great emotional support provided by CLIC Sargeant (now Young Lives v's Cancer). *J* had to travel an hour to get to the hospital but felt it was worth it due to the care and support she received. However, she is concerned about what happens to those who are not able to travel or who are diagnosed older than 25. Will they receive an equitable service?

L shared her negative experience of being a young adult on a ward with elderly people who sadly passed away.

SO shared her experience of supporting families accessing a highly specialised service. The multi-disciplinary clinics are fantastic, but it can be challenging for a family/individual to get a referral into the service. There is a lack of understanding regarding referral pathways, and local specialists are sometimes concerned about the cost implications of referring patients to specialist services. This can cause a delay in patients receiving the expert care they need. ASUK (patient organisation) provide a letter that patients and families can give to their GP explaining the referral process, and giving reassurance that patients in England can access the service without any funding requirements. Additional information is provided for people living in devolved nations.

SO also discussed the challenges some families experience in accessing carrier testing. When a genetic condition is identified within a family, parents and wider family members are often concerned about how this will impact future family planning. While efforts are being made to provide information to support families to make informed decisions, it can be difficult for potential carriers to access genetic services. This may be due to long waiting times, referral criteria, lack of Genetic Counsellors or the availability of genetic testing on the NHS. *L* shared her experience of being diagnosed with Sickle Cell disorder and her concerns that it is often just thought of as a 'black disorder', which isn't true. People of any race or ethnicity can be diagnosed with Sickle Cell disorder, therefore there is a risk that the condition will not be considered or tested for, in some communities.

L talked about the importance of communication. Patients need to have confidence in their healthcare providers. *L* feels that test results and feedback should only be shared by those who have expertise, so that they can respond to questions. *L* also shared some of the negative communication she has experienced from healthcare professionals – *"Oh people with your condition don't live long."*

*CC** discussed the initial findings from the BDB community outreach project. During workshops, people from diverse communities shared their experience of stigma, blame and shame when they or a family member were diagnosed with a genetic condition. In *C*'s experience, there is a real appetite for more information about genetics and rare conditions from people living within diverse and marginalised communities. Feedback has demonstrated how people want to improve their knowledge and understanding to support their own families and others within their wider community. *CC* gave an example of a 15-year-old girl who was born with a cleft lip, her Mum asked her GP for support and was given a phone number to call. English is not her first language,

and therefore she had no idea how to make contact, so has not been able to access any further support. Tools such as a 'Please Be Kind' card are being developed with the support of the BDB Experts by Experience Advisory Group. This card can be presented at a GP/health practice which explains that English isn't a person's first language and asks people to please be kind and patient. The card helps a person to request an interpreter for a specific language. Awareness and tools like this are needed to get this basic communication right.

"They are brave enough to come forward, then the system lets them down with the basic support".

Community Outreach Worker* *(relating to language barriers)*

MENTAL HEALTH

K discussed the stark inequalities in mental health support. He shared his personal experience of living with someone who has learning disabilities and experienced psychosis. It took 7 weeks for him to receive a diagnosis and to access the support he needed. *K* also highlighted the importance of sleep and how sleep deprivation can have an impact on mental health. This can impact people with rare conditions plus parents, carers, and siblings. Some families require additional support from healthcare professionals to establish positive sleep routines.

S discussed how there needs to be a referral and diagnosis pathway. Families wait too long for a diagnosis without any help, support, or any timeline in place of when the result will be received. During this time their child's condition is often progressing. This anxious time of waiting has a negative impact on the mental health and wellbeing of the whole family. This is also often the case for people who do not have support with care coordination.

SO explained how there is a national shortage of Clinical Psychologists, *"we have been without one for the AS multi-disciplinary, highly specialised adult clinical service for 2 years, despite having the funding in place."* ASUK have now started a pilot, wellbeing programme that is led by someone with lived experience. A consultation is taking place to learn more about needs in relation to wellbeing and to look at ways to co-design a wellbeing programme.

CC talked about the stigma associated with mental health in some communities and how this can prevent people coming forward for help. *CC* shared her experience of mental health services and explained that even where services for mental health may be available, they aren't tailored to rare diseases. Professionals often lack the understanding of how a rare disease can impact their physical and emotional health and wellbeing.

"Psychologists don't need to know about all 7,000 + rare diseases but they do need to have an understanding of the impact of having a rare condition and how physical and emotional wellbeing affects our mental health".

Parent

TRANSITION

A gave her insight into how important it is to have support when growing up, and how she shared her own experiences to help develop the T-KASH resources (**T**ransition – **K**nowledge **A**nd **S**kills in **H**ealthcare). *A* discussed how important these resources are for young people.

Kerry explained that through her experience of working with young people and their parents/carers, transferring to adult services is often described as 'falling off a cliff'. There is clearly a general lack of support and preparation. Although there are policies and guidelines around transition, we felt more was needed to support young people living with rare conditions, parents, carers, and healthcare professionals. In partnership with people with lived experience, ASUK developed T-KASH, these resources are freely available to support people through the transition/growing-up process.

H gave his experience of transferring to adult services. With the support of a patient organisation and Transition Coordinator, he was able to build on his skills and confidence to see professionals on his own. He described how scary transition can be, particularly if English isn't your first language. *H* is now a very independent young man and has since used his own experience to support others through the transition process. He recently supported families attending a transfer clinic and was able to provide information and support. The families found this incredibly helpful, especially as *H* was able to speak with them in Urdu which is their first language. Peer to peer support can be incredibly important and impactful.

H reiterated the importance of advocacy when transitioning to adult services to build confidence to go into appointments and ask questions independently.

C explained how she had been able to adapt the T-KASH resources into a booklet for a conference/workshop in America. Participants really liked the straightforward way they could look at all elements of their life and think through what they wanted and needed now and in the future. *C* felt that her transition was good, but it was only through talking about her own experiences with others, that she realised she wasn't supported well at all.

KEY POINTS

Society needs to stop thinking of people from diverse and marginalised communities as being hard to reach. In some cases, services can be incredibly difficult for people to access. More work is needed to improve the accessibility and inclusivity of services and support. We need to become more representative and design services based on the diverse needs of the population if we are to achieve equity.

Genomics has advanced over the last few years, and this will continue with further developments in technology and diagnostic capabilities. While one of the key priorities of the UK Rare Diseases Framework is to get a final diagnosis faster, it is essential that **all** families receive appropriate support throughout this process. Unfortunately, feedback from families highlights that there is much more work to be done to improve support and create appropriate pathways that are timely and equitable.

"It isn't unusual to have a family call our support line asking for further information as they have just received a genetic diagnosis but had no signposting or support, often googling to find further information."

Care Coordinator (Patient Organisation)

Getting the basics right - if we can't get the basics right in terms of communication and access to services, then how are people who encounter barriers on a daily basis expected to navigate the complex systems, and requirements associated with living with a rare condition.

Further training is needed to improve communication with patients. Information should be clear, concise, accessible, and produced in different formats. Translation and interpreting services should be made available when required.

To **address stigma and bias**, people within the rare disease community need to be aware of their own unconscious bias and how this can impact others. We need to strive to create a non-judgemental environment.

Patient education – empowering patients to take an active role in their own healthcare.

To **establish trust**, we must show empathy, compassion, and respect, be honest and transparent in interactions and demonstrate active listening.

While developments in technology and virtual consultations is a welcome addition for some, there is a risk that the use of technology in healthcare services could further widen the health inequalities gap, due to accessibility. In-person interactions is essential for some people within the rare disease community.

Discussions during this focus group and other BDB Experts by Experience Advisory Group meetings, highlight that although people are diagnosed with different rare conditions, so many of the experiences, issues and challenges are the same.

RECOMMENDATIONS

- Education and support to embed EDI throughout the Rare Disease community.
- Develop good practice guidelines for PPIE in Rare Diseases.
- Systemic changes – with input from PPIE to enable learning from existing barriers and to make meaningful and representative changes.
- Clearly defined pathways supporting people throughout their whole journey – including signposting to support services/patient organisations and to specialist services.
- Co-production of resources to provide further education around genetics, rare conditions, and research.
- Community outreach to build relationships, exchange information and establish trust.
- Improvements in mental health support and referral pathways – appropriate level of practitioners who understand the impact of living with a rare condition.
- Increase the use of T-KASH and other transition resources to support young people, parents, carers, and healthcare professionals.
- Ensuring genetic support services keep up with the ongoing development of genomic medicine into everyday clinical practice.

"Times are changing, and communities are wanting information and to be involved in all stages of their healthcare more than ever".

Person diagnosed with a rare condition.

NEXT STEPS

T-KASH resources and information to be sent to NICE as they are currently updating the NICE Transition Guidelines.

The findings from this focus group will be shared with the England Rare Diseases Framework Delivery Group and included in England's 2024 Action Plan.

BDB and DHSC will continue to work together to review the progress being made to address health equity and strive for continuous improvement. Engagement and involvement of people with lived experience will remain central to this work.

FUTURE DEVELOPMENT

Dissemination of BDB Training

- Introduction to EDI
- Walking on Eggshells
- Getting Comfortable with Uncomfortable Conversations

Dissemination of BDB Resources

- 'Please Be Kind' cards
- Good practice guidelines
- T-KASH tools

Thank you…

ASUK and BDB would like to thank everyone who contributed to the focus group, and all members of the BDB Experts by Experience Advisory Group for sharing their insights and experiences and for their continuous support.

We also give thanks to the DHSC and delivery partners for the efforts being made to address health equity for all people affected by rare conditions. We appreciate the recognition given to the importance of the patient voice, including the voices of people who often go unheard.

REFERENCES

UK Rare Diseases Framework
https://www.gov.uk/government/publications/uk-rare-diseases-framework

2022 Focus Group Report
Health-Equity-Focus-Group-Report-Dec-22-Final.pdf (breaking-down-barriers.org.uk)

T-KASH Resources
T-KASH Transition Tools | Breaking Down Barriers (breaking-down-barriers.org.uk)

BREAKING DOWN BARRIERS
Understanding genetics together