BREAKING DOWN BARRIERS

Understanding genetics together

Health Equity & The UK Rare Diseases Framework

Focus Group Findings, 7th December 2022

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 60 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 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.

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:

- a holistic approach to care and support
- accessible resources
- developing and maintaining trust in healthcare providers

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

The virtual focus group included a diverse group of 9 people with lived experiences, plus representatives from BDB and DHSC. A draft confidential progress report and a list of further actions was shared with the group ahead of the meeting to help 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.

During the focus group, a representative from DHSC gave a presentation on progress and plans and asked the group to consider 3 questions that had been submitted by the Rare Diseases Framework Delivery Group:

1. Do you have any examples of current experiences that relate to addressing health equity?

2. What do you believe are the urgent challenges for addressing health equity?

3. What education and training resources do you think would be helpful for healthcare professionals to help improve the way that genomic medicine services are being delivered within underserved communities?

Discussion Points

J discussed her experience of being diagnosed with cancer at a young age and the challenge she had in seeking a referral and diagnosis. She openly discussed the need to exaggerate her symptoms to get the GP to take her concerns seriously and make a referral.

L talked about her involvement in the CONCORD study and the importance of care coordination. There is no one size fits all, and experiences of care is dependent on the way services are delivered for people with different rare conditions.

S shared her experience of losing 5 babies due to a rare condition. She talked about the work she is doing in the West Midlands to raise awareness of the increased chance of babies being born to parents who are close blood relatives. S is working with a local authority task force and with communities and healthcare professionals to improve cultural competence, improve awareness, reduce stigma, and increase support.

H talked about the importance of role models and highlighting that health conditions and disabilities don't define who we are. He talked about how important his religion is to him and how he would like to be a leader within his community. However, his disability is seen as a barrier and people within his religious community often tend to have low expectations with regards to his capabilities. H is working hard to break down these barriers and show just what is possible.

H shared his experience of diagnosis and how much more information is available now. He remembers how it felt 10 years ago to get a diagnosis and how difficult it was to accept. He remembers how challenging it was for his parents, especially as English is not their first language. He now enjoys having access to a highly specialised services and is supporting other young people and parents as they transfer from paediatric into adult services.

L said how important she feels it is to have charity representatives who have lived experience. Having people represent their condition and showing others in the community what can be achieved, particularly given the shortened life expectancy. Being able to find others who have some understanding of what you are going through and experiencing similar challenges. L also feels that people with rare conditions need better access to mental health services and cognitive behavioural therapy tools. People not being able to access support contributes to the mental health and wellbeing of people with rare conditions.

K agreed and shared the experience of her son. Despite having access to a highly specialised service where funding is in place for a psychologist, they have not been able to recruit, as there appears to be a shortage of mental health practitioners.

C&J explained how involving people with lived experience in their work can make a huge difference. Creating a safe space and sharing experiences often enable others to open up, share their journey and seek support.

C&J discussed the issues faced by young people affected by rare conditions and the importance of them having their voices heard and being able to develop the skills, knowledge, and confidence to make their own decisions.

C shared her experience of delivering 'Genes, Family History & Health' workshops within diverse, marginalised, and under-represented communities in West Yorkshire. C is using arts and crafts to share information about genetics and genetic inheritance. The response has been positive with participants giving excellent feedback about this inclusive approach that is being delivered in partnership with community groups and people with lived experience. Barriers identified within these workshops include language, stigma, blame, shame, the need for safe spaces where people feel able to open up and share their concerns and experiences. Also, knowing where to turn to for support when a genetic condition is suspected in the family. There is an appetite for people wanting to learn about health and genetics even before a genetic condition is suspected in a family. Improving genetic health literacy amongst the general public is important and is different to when this is forced on people when a genetic condition is suspected or diagnosed.

L shared information about the Journey to Diagnosis research that is currently underway at Alstrom Syndrome UK (ASUK). ASUK have taken a sample of people that were diagnosed within a specific period of time. Interviews and data analysis highlighted the experiences of individuals and families and some of the areas of good practice such as earlier diagnosis and the role that patient organisations play in signposting families to appropriate information and support. It also highlighted some key challenges including stigma, blame and shame experienced by parents, lack of care coordination, provision of accessible information and disparities throughout different locations across the UK. While findings do highlight that the time to diagnosis is reducing, more needs to be done to improve the experience of diagnosis, especially following the mainstreaming of genetics.

Next steps

The findings from this focus group were presented to members of the England Rare Diseases Framework Delivery Group ahead of the publication of England's 2023 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.

Thank you…

ASUK and BDB would like to thank 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.

Reference:

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


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