



# Whose Voice

# Is It Anyway?

## Meeting Report

On Thursday 27 May 2021, a number of individuals from an array of backgrounds – all with rare diseases at the forefront of their minds – attended a meeting called **'Whose Voice Is It Anyway?'** in order to discuss the UK Rare Diseases Framework English Action Plan.

Together, Sondra Butterworth, CEO of RareQoL and founder of the **#WhoseVoice** campaign and Lucy McKay from **Medics4RareDiseases (M4RD)** hosted an engagement session on the UK Rare Diseases Framework on behalf of NHS England & NHS Improvement (NHSE & I).



# Participants



**Aisha Seedat** - Equality, Diversity and Corporate Social Responsibility lead for The Bushra Ali Group Ltd.  
I'm 23 years old. I have a rare disease myself and am an ambassador for the MPS Society UK.



**Andrew Stewart** - Communications Manager for Specialised Commissioning, NHSE & I

I am a Communications Manager for Specialised Commissioning at NHS England and NHS Improvement covering rare and ultra-rare diseases. Before this, I worked in the NHS Genomics Unit where I supported the roll-out of the NHS Genomic Medicine Service. Before joining the NHS, I was a senior adviser to an MP and former Government minister and an international journalist and TV Producer.



**Carole Knowles** - Epidermolysis Bullosa (EB) Clinical Nurse Specialist at Heart of England NHS Foundation Trust

I am a Senior EB Clinical Nurse Specialist currently working at the Heart of England NHS Foundation Trust.



**Dan Lewi** - Cure & Action for Tay-Sachs (CATS)

My wife and I founded the Cure & Action for Tay-Sachs (CATS) Foundation in the UK, after our daughter Amélie was diagnosed with Tay-Sachs. The CATS Foundation has helped pharma develop treatments for Tay-Sachs and Sandhoff diseases; from gene therapy programs to drug repurposing studies. I have over ten years of experience in medtech and advocacy, and have also served as Chairman of the European Tay-Sachs and Sandhoff Charity Consortium since 2012. I am fully aware of the patient voice, and wish to consistently put patients and their needs at the forefront of clinical discussion.



**David Rose** - Rare Disease Advocate

I have an ultra-rare condition called Occipital Horn Syndrome. I have been giving talks on life with a rare disease for the last 8 years at various events and conferences. For the last 3 years, I've been working for Rare Revolution Magazine – a magazine focusing on rare diseases. I have met so many fantastic rare disease advocates and industry professionals from all over the globe. I also volunteer for a cause close to my heart - Great Ormond Street Hospital. I recently joined Mitrofanoff Support as one of the trustees in January. I have been using a Mitrofanoff since the age of 7; my operation was carried out by Great Ormond Street Hospital in London.



**Dawn James** - Senior EB Clinical Nurse Specialist in association with DEBRA Birmingham Children's Hospital

I am a Senior EB Clinical Nurse Specialist with a background in general paediatrics. I have worked within dermatology for over 17 years and specialise in children with a particular rare skin condition. I am passionate about children and young people having the means to help them understand their health, care and social needs which would aid their empowerment towards making informed decisions that may help them for the future.



**Fiona Marley** - NHS England

I head up the Highly Specialised Commissioning Team in NHS England which commissions over 80 highly specialised services. The portfolio includes a number of transplant services, as well as services for patients with rare cancers and other rare diseases. We ensure that patients have: excellent clinical outcomes, optimised through interactions with international partners; equitable access to services regardless of where they live; and access to clinical trials. I have worked for the NHS since 1991 in a number of roles, including in primary care and health improvement. I also have a university degree in Biochemistry from Imperial College, London.



**James Brooks** - RARE Youth Revolution

I am the youth coordinator for RARE Youth Revolution, which works to give young people with rare disease a voice. I am also a rare disease patient myself, living with Niemann-Pick disease type B.



**Jo McPherson** - M4RD Operations and Finance Manager

I have been in the rare disease field for over a decade. Earlier this year I moved from the International Gaucher Alliance to M4RD, and am currently an Operations and Finance Manager for M4RD. I am also Patient Advocacy Group liaison lead for M4RD.



**Katie Callaghan** - RARE Youth Revolution

I am 19 years old and work with the RARE Youth Revolution team. I live with two rare diseases (Ehlers-Danlos and chronic intestinal pseudo-obstruction) and am passionate about improving the lives of those with rare disease and chronic illness. I work within multiple youth forums giving the patient/adolescent voice and have spoken at events, been part of many studies, podcasts and more. I'm particularly interested in the topic of transition and mental health! Along with this I run my own organisation Cards for Bravery.



### **Kerry Leeson-Beevers - Alstrom Syndrome UK / Breaking Down Barriers**

I'm a parent of an adult son with a rare condition, a partner in delivering a highly specialised service and I facilitate a network of patient organisations, support groups and community networks to improve engagement with people from diverse communities and reduce health inequalities.



### **Laurence Woollard - On The Pulse Consultancy**

I am the director of On The Pulse – an independent, strategic consultancy providing specialist insight to UK and global healthcare providers on the development and rollout of patient activation campaigns in haemophilia and rare diseases. I am highly driven by my own journey and challenges of living with severe haemophilia and the impact on the family dynamic, to campaign for and effect real change. I am representing the haemophilia community.



### **Lucy McKay - CEO of M4RD**

I'm a UK trained doctor and the CEO of M4RD. Growing up my brother had a rare disease, and I was around various other people with rare diseases. I studied Human Genetics at The University of Nottingham and then medicine at QMUL. At medical school I started Barts and The London Society of Rare Diseases, and subsequently Students4RareDiseases (S4RD). S4RD became M4RD in 2018 and a UK Registered Charity in 2019. My role of CEO combines my unique personal experiences alongside my medical training to improve the lives of those living with rare diseases.



### **Matthew Miles - Engagement Manager for Specialised Commissioning, NHSE & I**

I am an Engagement Manager for Specialised Commissioning at NHS England and NHS Improvement, working to ensure people and communities are engaged with and involved in commissioning of specialised services. Before this I worked in patient and public involvement for various charities, most recently in women's health for the Royal College of Obstetricians and Gynaecologists.



### **Pushpa Hossain - HCD Economics**

I'm a medical doctor by background with special interest in public health systems and rare diseases. I previously worked for a rare disease charity before moving to consultancy.



### **Rafael J. Yáñez-Muñoz - BSc PhD FHEA FRSB**

Professor of Advanced Therapy. Rafael Yáñez-Muñoz is the Director of the Centre of Gene and Cell Therapy in the Department of Biological Sciences, Royal Holloway University of London, UK.



### **Sheila Richards - Dermatology Clinical Nurse Specialist Birmingham Women's and Children's NHS Foundation Trust**

I am a semi-retired General Dermatology Nurse who has worked in dermatology centre for 17 years, some of that time working with children with EB. I'm very interested in genetic skin diseases and will deliver care for children with other rare skin diseases including Ichthyosis. I also provide support to the Ichthyosis Support Group charity.



### **Shwetha Ramachandruppa - Unique and Genes and Health cohort based at QMUL**

I work as a Consultant Clinical Geneticist at Guys Hospital. I am also a trustee at Unique, a rare disease charity representing children with rare genetic conditions. In a research context I work with the Genes and Health Cohort in East London who have recruited almost 50,000 people to a community based genetic cohort. I am looking at basic genetic literacy within the cohort and use this work to develop resources to improve genetic literacy.



### **Sondra Butterworth - RareQoL**

I am the founder of RareQoL, from a rare disease family, a carrier of a rare disease and I have worked with the rare disease community for over 10 years. I completed a PhD study which was focused on the quality of life and social support of people living with rare diseases. It was from this work and my lived experience that I developed the perspective that there are many sectors of the community who feel that their voices go unheard. I am participating from two perspectives: representing the rare disease and BAME communities, also as a researcher and Community Psychology Specialist.



### **Talia Coren - Senior Medical Copywriter at emotive**

I have a background in Biomedical Science and joined emotive, a global healthcare communications agency who create inspiring medical communications after graduating from Bart's and The London School of Medicine and Dentistry with an MSc in Forensic Medical Sciences. I specialise in rare diseases and have worked alongside Lucy and M4RD for the last few years.



# Introduction

**'Whose Voice is it Anyway'** is a RareQoL campaign promoted through their Rare Community Network to draw attention to the voices of those who are often under-represented in discussions about issues that affect the rare disease community. In an effort to provide a forum for those who may have been under-represented so far during consultations around the UK Rare Disease Framework and an Action Plan developed by the Department for Health and Social Care alongside NHSE & I, M4RD is proud to support the **#WhoseVoice** campaign in drawing attention to the lack of diverse voices being heard. Together, they have a shared aim to drive discussions in order to make real change happen.

“One of the key findings [from Sondra's PhD study] was that the participants were feeling that their voices were not being heard...there was a focus on the physical effects of the rare conditions but the psychosocial impact was largely ignored. A holistic approach to care and support was often absent. The aim of this session is to listen to the rare disease community whose voices are often unheard”

- Dr Sondra Butterworth

Lucy kicked off the meeting with a warm welcome, thanking everyone for their participation and sharing her excitement to be facilitating such an event in collaboration with Sondra Butterworth and the team from NHSE & I.

“This meeting is about representation”  
- Dr Lucy McKay

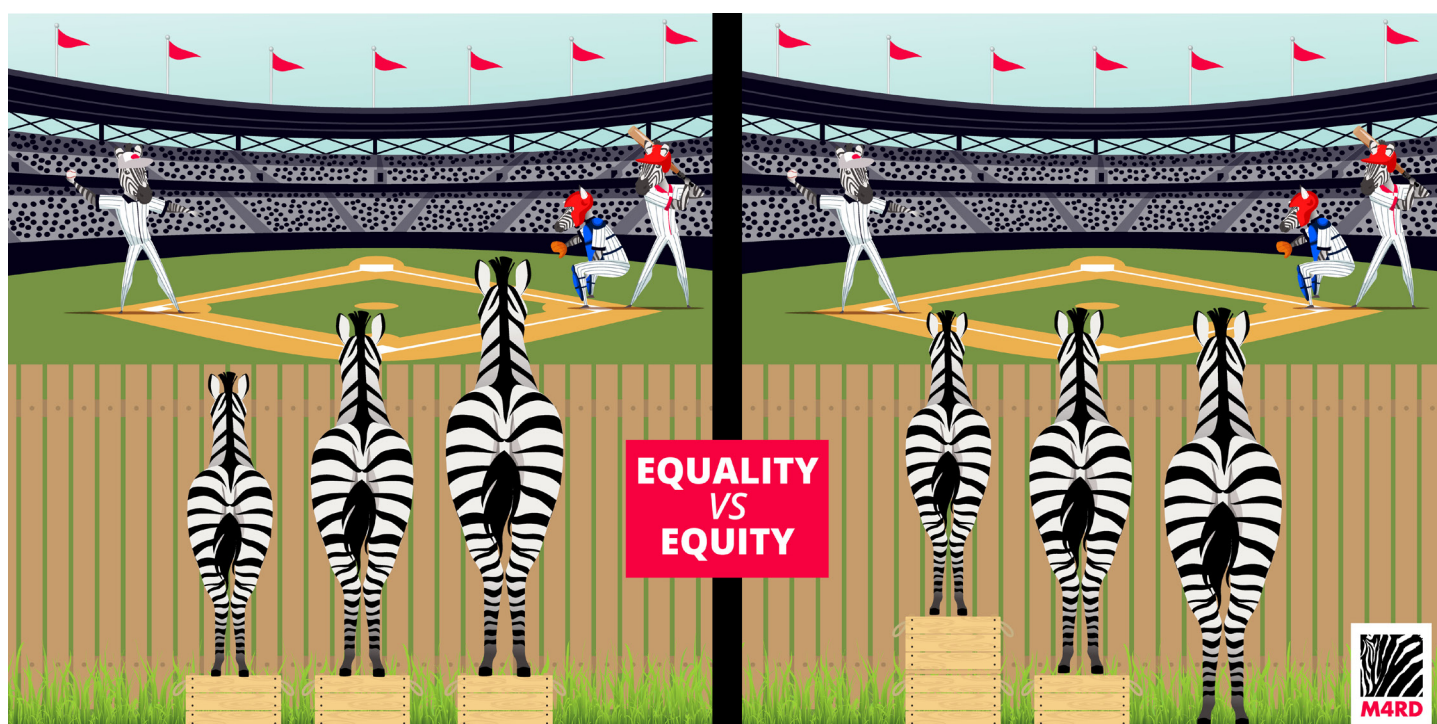
Sondra, as Chair of the meeting, then introduced the delegates to the #WhoseVoice campaign and the 'theories of need'. Sondra discussed 'expressed need', which is associated with the patient voice, and then compared this with 'assessed need', which is associated with the healthcare system. Sondra spoke about the fact that there is currently a mismatch in society, with

assessed need taking priority over expressed need, and that, going forward, a co-production approach is key in trying to reach equilibrium.

“The psychosocial needs of the rare disease community are not being met but the focus was very much the physical aspects of the conditions. There seems to be an imbalance in power with medical professionals having most of the power and patients having less”

- Dr Sondra Butterworth

The first set of slides was presented by Fiona Marley, Head of Highly Specialised Commissioning for NHSE. Fiona presented all delegates with a background on the UK Rare Diseases Framework and NHSE's role in delivering the 'Action Plan'. As attendees came from different backgrounds, including healthcare professionals (HCPs), patients, carers and patient advocacy group representatives, knowledge of the Framework and how it is made up was variable and thus had to be catered for. Fiona explained that each UK nation develops its own action plan and that this meeting was being run to discuss and identify themes and how improvements can be made and taken into consideration for England's action plan.



## NHSE'S EMERGING THEMES:

1. Ensuring a good fit of rare disease services with the proposals set out in the White paper – Integration and innovation: working together to improve health and social care for all
2. Reduction in health inequalities
3. Improving access to drugs
4. Patients with undiagnosed conditions
5. Provision of good quality virtual care where clinically appropriate and other initiatives that support care close to home
6. Continued innovation in genomics

These emerging themes aren't the only themes that NHSE are exploring. Various other themes that have been raised during alternate engagement activities and are also being explored.

Fiona then took delegates through the four key priorities highlighted within the Framework and what each comprises.

### The priorities are as follows:

**Priority 1:** Helping patients get a final diagnosis faster

**Priority 2:** Increasing awareness of rare diseases among HCPs

**Priority 3:** Better coordination of care

**Priority 4:** Improving access to specialist care, treatments and drugs

Andrew Stewart, Communications Manager for Specialised Commissioning for NHSE & I, then took the floor and further elaborated on the requirements to develop a successful Action Plan. Andrew explained how a 'tag-team' approach between themselves and the Department of Health is necessary in order to establish the best possible Action Plan for England, and that everybody in attendance had the same common goal: to create change and ensure the Action Plan encompasses specific recommendations that are informed by those who may not otherwise have a platform on which to have their experiences or voices heard.

Fruitful discussion started early when Laurence Woolard asked, "Thinking about reduction in health inequalities - how come that wasn't positioned as improvement in health equity?". Equity was subsequently a dominating theme of the remainder of the meeting.

Breakout rooms were next on the agenda for delegates. Before this, Sondra reminded attendees that it is the patient voice that must underpin the four priorities and the action plans created by each nation. Similarly, M4RD believes that HCP awareness (Priority 2) is fundamental to success in the other three priority areas and should thus be woven into discussions for all Priority groups. Consequently, attendees were separated into three breakout groups for Priorities 1, 3 and 4, respectively. Each group was facilitated by either Lucy, Sondra or Fiona respectively, with support from a member of either the NHSE or M4RD team.

*"Awareness amongst healthcare professionals needs to be integral to all areas of the UK Rare Diseases Framework and action plans, just like the patient voice"*  
- Dr Lucy McKay

In each of the breakout rooms, the delegates were asked to provide their insight into the Priorities outlined in the Framework and to discuss the emerging themes presented by Fiona in the introduction.

### Each group were given the following questions to think about whilst engaging in discussions:

- Do these themes reflect the expressed needs of the rare disease community?
- Is anything crucial missing from these themes?
- How will HCP awareness benefit this priority and these themes?
- What specific ways can be suggested to use the patient voice in the designing of services?

### Priority 1: Helping patients get a final diagnosis faster

For the first priority group, key themes for discussion included the speed of diagnosis, experiences along the diagnostic pathway, continued innovation in genomics, concentration on undiagnosed patients, management of being given a diagnosis and the aftermath, and reduction in health inequalities and awareness amongst HCPs.

The participants in this group engaged in fruitful dialogue, using their backgrounds and experiences to contribute their viewpoints to the discussion. The group concluded that, in healthcare, we may need to go back to basics in order to create health equity for everybody. Instead of considering some groups of patients as being 'hard to reach', we are likely to have to shift our mindset and think of it as a flaw in the system in terms of accessibility.

*"The Bangladeshi and Pakistani communities that I work with in London are often labelled hard to reach, but that's based on assumptions rather than knowledge"*  
- Dr Shwetha Ramachandrapa



*“There is a whole movement towards understanding of intersectionality...Essentially, the more issues or complexities there are for a person, the further down the social strata someone comes. So, if someone has a rare condition... They may have mobility problems which requires the use of a wheelchair; they may have different ethnic backgrounds or religious differences which affect the care and support they require. The intersection between these factors can mean that they can have increased challenges or face health inequalities”*

- Dr Sondra Butterworth

*“Striving for health equity doesn't mean providing the same services for everybody, it's about having appropriate services for our diverse community”*

- Kerry-Leeson Beevers

*“I've worked in the department for 17 years...skin really, in the medical world is neglected...but I've spent my whole life dealing with problems that could have been prevented”*

- Sheila Richards

Discussions continued, and the concept of cultural competence alongside HCP education was raised. The group had a conversation around HCP education and the fact that there is unconscious bias woven into society; they highlighted how, in order to combat this, HCPs could receive further training on the matter in order to confidently approach patients.

*“It's important to think about communities and cultural competence and ensure that things are presented to people in a way that is appropriate to the social context. And I think we have a responsibility to make sure we do that and to make sure services are designed in a way that's accessible to people”*

- Dr Shwetha Ramachandrapa

In this country, we are lucky to have people from a wide range of countries and cultural backgrounds, and HCPs could be offered further education to help them successfully fulfil their duties. One delegate asked, “How are HCPs expected to help their patients if there are language barriers and cultural differences that they can't currently cater for?” The group agreed that further training was necessary and that we have a responsibility to design services that are accessible to all. Following this, the group agreed that a ‘one size fits all’ approach in the diagnostic journey doesn't work and that those with skills in communicating should be at the forefront of conversations; letters should be replaced with human contact and patients should be given the opportunities to participate in discussions about their health.

*“If you look at it from the patient's perspective, they're being sent a letter saying “you need to have this test in 6-months time, call us to book an appointment” but what is this test for? And if you add into that the language barrier and into that the whole stigma about rare disease...sometimes people do not know why it's important. What will it lead to? What will it mean? Who do I ask these questions to?”*

- Dr Pushpa Hossain

The group then shifted their discussions towards diagnosis itself and asked the rhetorical question, “Who is the diagnosis for?” The group commented on the fact that parents are often the ones receiving the primary diagnosis; however, it is the child who then has to grow up learning what their condition is and what their diagnosis means, and the implications this may have on their own family. The question “Is speed the most important thing for patients?” was also asked. The group discussed how diagnosis should be looked at as a lifelong journey or experience. It is not only at the point of diagnosis that people need support and information; patients and their families need different information at different periods throughout life. Over time, the priorities of each individual changes; diagnosis inevitably begins as the most important priority and then, over time, other parameters become more important. The delegates spoke about the need for services to be designed to respond to patients as and when they need them and to ensure they are designed in a way to provide continuity of services throughout life. It is important to have structures in place to support patients and their families both when a diagnosis is reached and immediately afterwards, and for any potential mental health implications that may be experienced as a result.

*“There's a lot to be done to engage these communities on even a very basic level even before you think about finding a diagnosis faster or any of these issues. Just general trust and engagement I think is something that we have a responsibility to work towards”*

- Dr Shwetha Ramachandrapa

*“It's not just about getting a diagnosis as fast as possible, it's also about acknowledging the experience of getting that diagnosis and acknowledging that services like genetic counselling are not just needed at the point of diagnosis. Family's needs will differ and people will need information at different periods, not just in the first year”*

- Kerry Leeson-Beevers

To conclude, the patients in the group touched on how the system and agenda could be informed by the patient voice.

*“We need an open and plain channel of communication that goes both ways between patients and healthcare professionals. It needs to be very clear. It needs to be very well understood and not just a tick box exercise”*

- Dr Pushpa Hossain

The way to successfully overcome some of the issues discussed is to directly speak to those it affects. Patient advocacy and peer support were also discussed, and how important these voices can be throughout each and every journey. The group discussed the potential for collaboration between patient advocacy groups with a good record of excellent care and authors of the White paper and how this could be an exciting collaboration, with real change being made as a result.

### Priority 3: Better coordination of care

In the Priority 3 group, key themes for discussion included the provision of mental healthcare, ensuring a good fit of rare disease services (with a focus on integration and innovation to help improve health and social care for everybody), the provision of good quality virtual care where clinically appropriate, reducing health inequalities, coordination of health and social care, peer support and ensuring the stability of patient advocacy groups, the transition from paediatric to adult care, and awareness amongst HCPs.

The participants of the group had plenty to talk about and many were able to share their personal experiences with coordination of care within the healthcare system. The group discussed the enormous requirement for those in primary and secondary care to communicate with each other directly instead of using the patient as a conduit. The patient is always the consistent party within discussions, so more work could be done to ensure that patients become equal partners in their care but aren't further burdened by the administrative side of coordination of care.

*“We need a tool that helps people communicate with each other, and this needs to be patient-friendly. Where services can communicate with each other plainly that is patient-centred so patients can see what is being communicated and what is being dropped”*

- Dr Pushpa Hossain

As it stands, patient centricity is lacking from existing communication channels, and more could be done to ensure these changes take place.

These discussions linked directly to the group's next set of discussions around the transition of young patients with chronic conditions from paediatric-centred to adult-orientated healthcare systems. The group discussed how transition should be as smooth as possible and requires successful communication between paediatric and adult teams of doctors. The participants discussed how the transition should be based on individual patient cases and their families and not just clinical decisions.

*“Successful communication and having a patient-centred approach [are key to successful transition of care]”*

- Katie Callaghan

Multiple attendees raised the point that the transition should happen slowly and start at an earlier age, rather than patients being 'thrown in at the deep end' aged 16 to 18. A first-hand account from one of the attendees emphasised the importance of this, as he shared his own experience with transition and how it took time for the changes to come to the surface. He commented that the amount of admin required by himself, and communication needed once he transitioned to adult services, was overwhelming and something that had not been sufficiently explained. Another attendee, who works as a Clinical Nurse Specialist, suggested that adult care nurses should go into paediatric appointments during the transition to meet patients and help them

settle into the adult hospital. This model is currently very centre- and community-specific, and the group agreed that this model could be more commonly utilised. This conversation evolved into further discussion on where the problems arise; is the problem with the transition itself, or is it that adult services divide patients into multiple body systems rather than using a holistic approach?

*“Youth participation in rare and chronic condition healthcare settings is fundamental to service design and delivery... these should be evidence-informed”*

- Laurence Woollard

Further discussions brought to light personal accounts of the burden of explanation during transition and how a large number of adult specialists aren't necessarily trained on how to work with young adults. Currently, services are designed for either young children or adults, and there is a clear and notable gap for young adults during the transition.

*“I think quite a lot of adult specialists aren't necessarily prepared enough on how to work with young adults”*

- Katie Callaghan

*“It feels like Rare Youth Revolution needs to be part of the Transition Management plan”*

- Dr Lucy McKay

*“Examples of good practice should be shared, and the voices of young people heard...It's important if we're thinking about the patient's voice that those who are designing services speak to the people who are experiencing those services or talking specifically about transition services at the moment”*

- Katie Callaghan

Compared with paediatric doctors, adult doctors are very system-specialised, and patients are unsure who to approach when the doctors don't have the knowledge to help them. This gap in transition is something that patients struggle with, and the delegates agreed that more could be done to help combat this. To conclude conversations, the group agreed that successful transition is based on care coordination between services and that having a care coordinator is vital to support patients both during and after the transition.

This group also discussed coordination of care in the light of virtual care and how, in their opinion, phasing within the Framework of 'where clinically appropriate' isn't entirely accurate.

*“Not everybody can access virtual care...we need to be careful that these decisions aren't made on a purely clinical basis. It needs to be right for the patient as well”*

- Kerry Leeson-Beevers

As the world of digital consultation remains a fairly new concept, the group discussed that decisions could be integrated and that patient choice needs to become a big part of those decisions. Patients should be given a choice of whether they are happy to have their appointments virtually or whether they would prefer a home visit. The group noted that funding is a current issue but that, in an ideal world, all services could be provided as a choice so that patients are comfortable in the way they are receiving their care.

Finally, Sondra led the group's conversations on mental health and social prescribing. The group discussed how there is currently no general approach to mental health within the rare disease community and that disparity in mental health service provision is enormous. Rare disease-specific mental healthcare is uncommon and, considering so many patients with rare diseases suffer from mental health issues, more should be done. One delegate explained how he was lucky enough to receive rare disease-specific mental healthcare and highlighted how it felt far more tailored to his needs.

*“I advocate whole-heartedly, coming from a RD family, for the biopsychosocial needs management, treatment and care has to be important. You can not have one without the other”*

- Dr Sondra Butterworth

#### Priority 4: Improving access to specialist care, treatments and drugs

For the Priority 4 group, key themes for discussion included physical access to a specialist centre, remote access to specialist input, access to treatment, access to research and clinical trials and ensuring a good fit of rare disease services, with a focus on integration and innovation to help improve health and social care for everybody.

The participants in this group opened the discussion by talking about the tendering process and how tender activities appear to be solely cost-based. They highlighted that if there was user involvement throughout the whole process, a different conclusion may be reached. The concept of cognitive diversity was also brought to light and the attendees discussed how diversity of thought and the involvement of diverse patient populations during the tendering process could help introduce real change. Secondly, discussions centred around the culture whereby a small biotech company or organisation might be bought out by a big pharmaceutical giant and the impact this has on price. The price of rare disease treatments was discussed in detail; attendees considered how expensive rare disease treatments are and asked the simple question, “Do the costs of medicines and treatments need to be that high?” The group spoke about how research is fundamentally carried out by public organisations in academia, funded by national programmes and charitable funds and that it is only when clinical trials demonstrate positive results that pharmaceutical companies tend to intervene and take over.

*“The big concern for us is access to treatment. We've been watching our colleagues and the fights they've been having trying to establish things. It always worries me about the cost of drugs, the whole approval process and how long it takes. And then access to medicines. How can we implement a programme for these drugs? If we can get approval and be approved quickly there should never be a delay in access to treatment. Unfortunately, this is what's been happening”*

- Dan Lewi

The group concluded that the pricing of therapies must evolve while we search for new models to look at affordability and access. As the bulk of research is in the public sector, shareholders should not be in a position to profit in the way they currently are.

Delegates offered to speak about their individual experiences with clinical trials, access to treatment and lack of specialist care. One attendee discussed how expensive clinical trial enrolment is and that if funding were to stop, it would be incredibly upsetting for patients and their families, whilst also expressing her frustration around the lack of understanding from her doctors.

*“We're going into review again and it could be that the funding would stop and that's upsetting for both patients and their families”*

*“I live in Leicester, and I agree that healthcare professionals here don't know much about my rare disease. I felt like they knew more about it when I was a younger child but since I turned 18 it's like the whole process of explaining my condition over and over again”*

- Aisha Seedat

When trying to access healthcare for problems unrelated to her rare conditions, Aisha Seedat reports:

*“Doctors are confused on the management of the condition and will refuse to communicate with the metabolic team. It makes me think, 'are they not doing their research properly?' I'm 23 now and I've been seeing them since I was 18. You would think that they would have grasp, some knowledge and understanding and would have educated themselves long ago. It's me as a patient. I'm having to educate them”*

Discussions around the lack of patient populations and access to treatment then evolved. Delegates highlighted the fact that there are few treatment centres in the UK that can administer the therapies needed for their conditions, meaning they have to travel for a few hours in order to receive them. Conversations around how the COVID-19 pandemic has exacerbated this issue were also explored; the group concluded that the issue of ease of access to treatments for those who need it should be tackled as soon as possible. The group made it clear that approvals, time, cost and access should never result in delays to life-changing care and posed the idea that a framework to speed up approvals and ensure access to treatment is more readily available should exist.



The group also discussed that the Framework is lacking priority about searching for novel treatments and agreed that rare disease treatments should be seen as more of a priority. This may require defining rare diseases as a specific area of disease research rather than splitting them up into specialties.

**“We want to get the Government, Medical Research Council and Wellcome Trust to acknowledge that rare diseases should be a research priority”**

- Professor Rafael Yáñez-Muñoz

The group engaged in a conversation around the fact that, beyond diagnosis, there is little other than palliative care for most patient populations. One delegate was confident in making the statement:

**“In my view, while genomics is critical and will deliver a diagnosis which is really important for the patient, it won't deliver them a treatment. So, for me one lacking priority [of the UK Rare Diseases Framework] is the development of novel treatments for rare diseases and have research programs specifically focused on those”**

- Professor Rafael Yáñez-Muñoz

The group agreed that for many conditions we have the potential to diagnose patients effectively, but it's a shame that we don't have enough treatments in the pipeline to improve their outcomes. The group collectively discussed this statement and agreed that rare disease treatments should be incentivised where research is required, and we should be making this more attractive. Related to this, the delegates went on to discuss the lack of awareness of these treatments.

**“Patients were open about the fact that their doctors didn't know treatment existed. Healthcare professionals need to know what's out there when treatments are approved”**

- Talia Coren

The group concluded that more work is needed regarding treatment uptake to help close this gap. We should learn where access is limited and establish whether processes can be built on to ensure this doesn't happen in future. Fiona spoke to the group about how uptake rates could be looked into and compared with incidence rates, with any identified mismatch being geographically investigated in further detail.

## Summary and close

At the end of the breakout discussions, all delegates simultaneously returned to the main meeting room to discuss and summarise each groups' findings. A spokesperson for each breakout group ran through the key themes and discussions from their priority group to share with the other members. All attendees had the chance to comment and bring up any points they wanted to raise or add to discussion points raised in other breakout groups. Sondra then wound up discussions and brought conversations around the breakout groups to a close.

Andrew and Lucy then took the floor and concluded the meeting by saying thank you from NHSE and M4RD, respectively. Lucy highlighted how incredible it was to have everyone in one room (albeit it a virtual room) and how she hopes that there are further opportunities for the group to meet again in the future.

M4RD, Rare QoL and NHSE are all extremely grateful for the proactive nature of all delegates and are hopeful that these themes can be fed through into England's Action Plan.

**“Pass the power and you will hear the voice”  
#whosevoice**

## Appreciation

On behalf of all meeting facilitators, we would like to extend out a huge thankyou to all the delegates that participated in the 'Whose Voice Is It Anyway?' meeting. Every delegate brought their experience and expertise to the table and helped us engage in such fruitful, constructive, and open exchanges to help provide insight into the Priorities outlines in the Framework.