The Unusual Suspects Report 2024

On 21 February 2024, emotive attended Medics 4 Rare Diseases' (M4RD) 11th Annual Meeting, 'The Unusual Suspects'. This meeting is held in association with the Medical Genetics Section of the Royal Society of Medicine (RSM) in London and after overwhelmingly positive feedback in 2024, the meeting was held in hybrid format giving attendees the opportunity to join face to face and online. Senior Account Manager, Charlotte Roe, and Associate Medical Writer, Daniella Butler, joined the event to tell us more about what was discussed this year.

Who are emotive?

emotive are a global medical communications agency who partner with life science companies across medical & scientific affairs and brand & patient engagement. emotive has supported M4RD for 6 years, providing commercial, creative and strategic expertise to support their mission to raise awareness of rare diseases within the medical curriculum and profession.

Meeting report

In the 11 years since the event first took place, the M4RD Annual Meeting has become a very popular event where healthcare professionals, medical trainees and students, patients and carers gather to raise awareness and emphasise the importance of a holistic approach to rare disease, providing tips to improve the diagnostic journey and management.

This year, the focus of the event, held at the RSM, was on communication and inclusivity in rare disease.

The event was opened by Dr Shwetha Ramachandrappa, Consultant Clinical Geneticist, President of the Medical Genetics Section at the RSM and Chair at Unique, a charity supporting, informing and networking families and individuals affected by rare chromosome and gene disorders. Shwetha opened the event by reflecting on the idea that each person wears many hats, taking on various roles and identities. However, connecting with others beyond those various roles is about **sharing our unique stories, practising good communication, actively listening and being vulnerable**. By approaching interactions on a personal level, authentic and understanding connections are created.

Dr Lucy McKay, CEO and Founder of M4RD, welcomed face to face and virtual attendees, introduced her team at M4RD and provided an overview of the afternoon's agenda and speakers.

‘Rare Disease 101’ provides healthcare professionals with the tools to recognise, support and manage patients living with rare conditions

Lucy began by sharing M4RD’s vision – **a world in which no one faces inequality in healthcare based on the rarity of their condition**. She highlighted that just because a disease has a low prevalence, this shouldn’t impact a patient’s access to basic healthcare. Lucy described their mission to build a healthcare workforce that is trained and supported to meet the needs of those impacted by rare conditions, so that patients, carers and families feel listened to, believed and involved in their care.

Lucy then gave a whistle-stop tour of M4RD’s **‘Rare Disease 101’** e-learning course, including the basic principles of what rare disease is, the challenges faced by those living with a rare condition and how healthcare professionals can support patients and their families.

Rare disease is one experienced by <1 in 2000 people. However, there are more than **7000 rare diseases**, affecting 3.5 million people in the UK, with an estimated **1 in 17** people affected in their lifetime – so, although the conditions are individually rare, they are collectively common. A high proportion of rare diseases present in childhood and 80% have a genetic origin; however, only nine rare genetic conditions are screened for at birth. Lucy explained that while healthcare professionals cannot know all 7000 rare diseases, they should be **‘Rare Aware’** and expect to encounter patients with them.

Diagnosis of a rare disease takes an average of **4–6 years** and patients are frequently sent from one specialist to another, sometimes encountering disbelief, misdiagnosis, inappropriate treatment or no treatment at all. Lucy emphasised that this difficult journey of diagnosis is such a common experience in the rare disease community, that it has its own term known as the **‘diagnostic odyssey’**. The diagnostic odyssey leaves patients just wanting answers and a diagnosis, which can take its toll on patients and families.

Lucy discussed the overarching goals of M4RD – to ensure people living with a rare condition receive a timely diagnosis, mental health support and holistic specialist care, including excellent communication, as well as support from patient advocacy groups and hope from ongoing research projects.

Closing her talk, she reminded the audience of the impact of words, noting they can “either hurt or help” patients and families. She emphasised that communication is a tool that can be honed and that we all have the capacity to improve our communication skills.

Find more resources in M4RD Module:

Rare Disease 101 ›

RARE 101

The power of words

The first invited speaker was Jono Lancaster, author of ‘Not All Heroes Wear Capes’, public speaker and Co-founder of Love Me Love My Face Foundation.

Jono opened his talk by asking the audience to form a heart by raising their index finger and thumb, which resulted in the room instantly uniting in a display of love. This moving gesture served as a reminder that words have power, even to make or break an individual’s heart, something everyone should consider in their interactions.

Jono was diagnosed with Treacher Collins syndrome at birth, a condition characterised by a range of distinctive craniofacial differences that can affect the eyes, ears, cheeks, palate and jaw. At just 2 weeks old, he was placed in foster care. Jono shared with the audience the story of how Jean came into his life and later adopted him. Jono reflected how, as a young child, he would share his personal stories with pride, telling other children how his mum Jean had specifically chosen him. He explained that he would refer to his smaller ears as his ‘go-faster ears’ and would excitedly tell other children that they made him run faster than anyone else. However, as Jono grew older, life became harder and he struggled to see where he fit in the ‘image-obsessed’ world. Jono described how he started to listen and believe the hurtful words and negative conversations around him. He explained how the words used about him by others as a child, he later repeated to himself, and these interactions profoundly influenced his self esteem.

After starting therapy, Jono began a journey of self-love and acceptance. Jono introduced the audience to Zippy doll, who represents ‘Little J’, wearing one of his childhood rugby tops. He discussed how he is learning to visualise his inner self, using Zippy to ask ‘Little J’ what he needs and how he made him feel. He explained how he is learning to love ‘Little J’ and continues to work on the relationship with his inner self. Jono explained the **power of the words we have with ourselves** and how they stay with us, reminding everyone to be a little kinder to themselves.

‘Not All Heroes Wear Capes’ tells Jono’s story about how he found his way out of difficult times. The book talks about some of the heroes who changed his life through conversations. He expressed how important the conversations we have with strangers can be and how they can either put out an individual’s spark or give them a little more light to keep going. He reminded the room that we are all strong enough to become our own hero (although the cape is optional)!

Jono closed his talk by leaving the room with a final thought – “The next interaction that you have with another human being can change someone’s life forever and that is the power of words and communication.”

Listen to Jono chat to Lucy about his experiences with Treacher Collins syndrome and what he’s learnt over the years

Listen now ›

Core communication skills

The subsequent talk was given by Dr Lisa Kauffman, Consultant Community Paediatrician and Associate Medical Director for children's community health services for Manchester Local Care Organisation and communication skills trainer.

Lisa asked the audience "What do patients and families need from us?" – responses included patience, honesty, time and to be believed and listened to. Lisa then took the audience through the skill of **listening**, breaking it down and exploring how healthcare professionals can make conversations more supportive, successful and patient/family centric.

1. Know how to get your patient and family to tell you **what is important to them**

- ✓ Ask an open question – "How are you?" "What do you want to talk about today?"
- ✓ Let the patient and family lead

2. Silence

- ✓ Stop and listen to the patient and family; you don't need the answer ready, you just need to listen

3. **Reflect** on what you've heard

- ✓ "What you've told me is" "What you're struggling with is"
- ✗ Do NOT say "I hear what you say"; instead, repeat back what the patient has told you

4. Acknowledge and validate

- ✓ Hear, acknowledge and validate what you have heard
- ✓ This will allow the patient to listen to what you have to say

5. **Summarise** what you have learnt

6. Empathy

- ✗ Do NOT say "I understand"; instead, "I can't imagine" or "It must be really difficult"

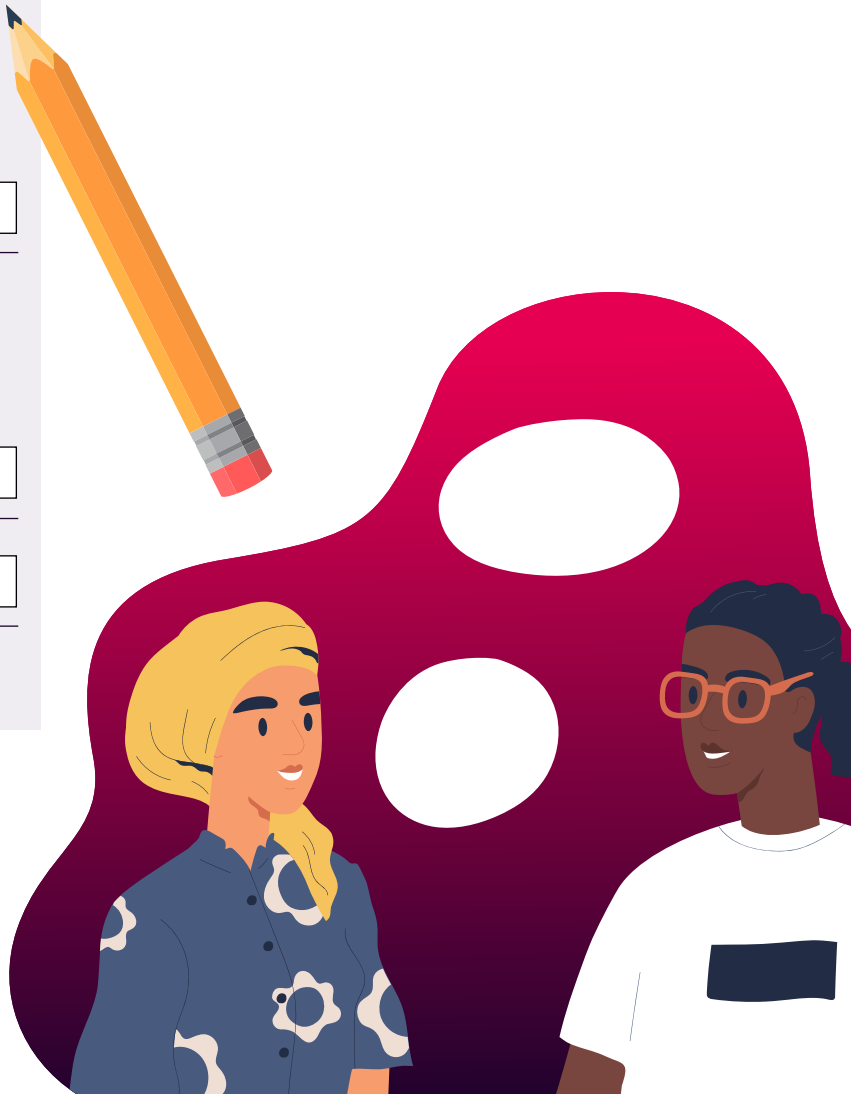
Lisa reflected on the meaning of **empathy** – imagining what someone is experiencing and feeling, without thinking you **are** them. She described how healthcare professionals cannot understand or know a person's condition; however, they can see their battles. Lisa emphasised the importance of listening and reflecting on the words families use to navigate the conversation, letting the patient and family take the lead.

Lisa highlighted how important it is to give one piece of information at a time, so that the conversation can be digested and processed. Furthermore, when giving information, to pause and ensure the patient and family understand and are ready to move on, considering the effect difficult conversations can have on their mental health.

Lisa reflected on conversations being like a recipe, and how the skills of listening are ingredients for supportive, helpful and positive conversation, which are not damaging to mental health. Lisa closed her talk by emphasising how patients **value being seen, listened to and treated with empathy**. With this, conversations can be patient and family-led; therefore, more successful for both the patient and healthcare professional. Furthermore, if healthcare professionals can get this right in the rare disease community, this can pave the way forward throughout practice.

Learn more about Reach communication skills training.

Reach communication ›



Breaking down barriers

Lucy then introduced Kerry Leeson-Beevers, Alström Syndrome UK (ASUK) and Breaking Down Barriers (BDB) Chief Executive (CE), and patient representative on the England Rare Diseases Framework Delivery Group.

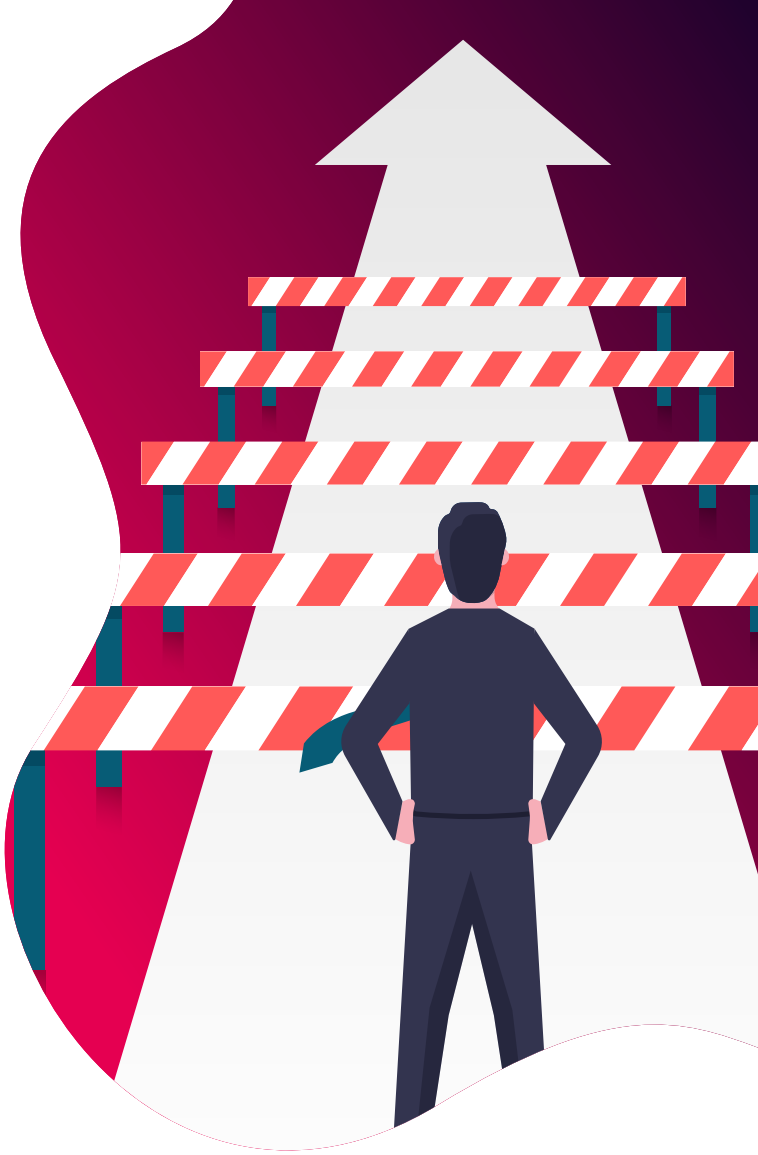
At only 3 weeks old, Kerry's son went into heart failure. After being given some possible diagnoses, Kerry researched online, determined to find answers and support for her son, where she came across and joined ASUK. Her son's diagnosis of Alström syndrome (AS), an ultra-rare, complex and progressive condition, was later confirmed following genetic testing. Kerry has had several roles throughout her time at ASUK and is now the CE. The non-profit organisation provides support for 92 patients with AS and their families.

Kerry introduced the BDB project, a network of organisations working together to improve the lives of people from diverse and marginalised communities affected by rare/genetic conditions, so they have equal access to health services. Kerry explained that **"there are no such things as hard-to-reach communities, only services and organisations that can be difficult for people from diverse groups to access."** She explained how they work to find ways in which health services can be shaped to become inclusive and accessible, and work with families to encourage and support them in accessing those services. This includes ensuring the creation of safe conversation spaces where families and healthcare professionals can connect and access those services.

Kerry then shared some of the learnings from their BDB 'experts by experience' group, particularly the importance of language and how, if used incorrectly, it can result in parents feeling blame, helplessness, shame and guilt. She explained how healthcare professionals must consider the most appropriate time to ask questions, and how to communicate with families and patients in a supportive and empathetic manner.

Kerry summarised the key findings that arose from the focus group, which are shared with The England Rare Disease Implementation Group's partners, to support learning and encourage the development of inclusive and representative actions.

- **Improve the accessibility and inclusivity of the services and support available**
- **Ensure services are more representative and based on the needs of the diverse population**
- **Provide safe spaces to refresh on communication skills**
- **Address stigma and bias people in the rare disease community experience**
- **Demonstrate active listening**



Kerry emphasised the importance of getting the basics right, including communication. As language changes so much, healthcare professionals need to have a safe space to refresh their communication skills. She urged healthcare professionals to be open and honest in conversations and to be aware of any unconscious bias, highlighting that trust, empathy and active listening are the foundations for meaningful and supportive conversations.

Kerry introduced Transition – Knowledge and Skills in Healthcare (T-KASH) resources, which are free resources designed to support young people, families and healthcare professionals understand the transition journey and the things that are important to young people as they grow up. The resources have been designed to ensure they are inclusive and representative of the needs of young people within the rare disease community. Kerry closed her talk by reflecting on a thought from young people experiencing rare conditions – **"Don't just see me and my rare condition, see me and the things that impact me."**

Learn more about the Breaking Down Barriers network

Read the report – BDB focus group findings ›

Explore the T-KASH transition tools ›

Embracing the unknown

The **Student Voice Prize** (SVP) is an annual international essay competition run in collaboration between Beacon and M4RD, with support from Orphanet Journal of Rare Diseases and this year, sponsored by emotive. Each year, the SVP team pose three questions centred around rare diseases and the patient experience to medical students, and undergraduate/masters students in health sciences. The questions encourage students to reflect on the impact on patients and families of rare disease experience. In 2023, there were **100 entries** – the largest number to date!

After congratulating the runners up, Chandan Singh Sekhon, Iman Muzafar and Oreoluwatonu Oduwole, Lucy introduced the winner – Leisha Devisetti from the University of California, Berkley. Leisha's essay recounted her family's experience with rare disease and how the communication of uncertainty can be improved in the medical field, especially with diagnoses.

Leisha began by sharing her family's experience of the diagnostic odyssey. She described her mother's 10-year journey of misdiagnosis and eventual diagnosis of ameloblastic carcinoma (AC), a rare, malignant tumour in the jaw. There have been less than 100 cases of AC reported since 1948. With conditions this rare, there is a large data deficit and Leisha explained how this deficit made it difficult to find resources to help them during their diagnostic journey. Furthermore, she explained how, as a family, they found it difficult to find concrete information and came across conflicting evidence. Leisha highlighted that it is not only difficult for patients to navigate this uncertainty, but also challenging for healthcare professionals. With immense uncertainty, it can be hard for a healthcare professional to convey information and, therefore, difficult for patients and families to get their questions answered.

AC can arise de novo or from long-standing lack of proper treatment. Due to her mother's long diagnostic journey, Leisha expressed that this is something they will always question; whether AC could have been avoided if mother's condition recognised earlier. However, it will remain unanswered. Leisha shared how her family took comfort in focusing on what they did know, even if it did not all make sense.

Leisha explored two types of uncertainty that are heavily associated with rare disease. Inherent uncertainty comes with the variability with each patient's condition and presentation, including their future impact. Epistemic uncertainty refers to uncertainty attributed to missing information or expertise. Leisha explained how she and her family found it difficult to put trust in the treatment plan, given the immense uncertainty they faced. She explained that addressing these uncertainties requires not only systemic or research-based changes, but also transparent communication with patients. This includes healthcare professionals conveying what they do know and not communicating what they think someone wants to know or see. Overall, this can create a more supportive journey for those dealing with a rare condition.

Leisha then took the audience through key areas related to communication, highlighting areas she identified as needing improvement, especially in the face of uncertainty.

- **Challenges** – address diagnostic challenges and explicitly acknowledge gaps in the evidence
- **Affirm** – give optimism by highlighting successful outcomes
- **Support** – provide emotional support for patients and families
- **Significance** – view every rare disease as significant; each case can inform the next one
- **Research** – identify areas for future research
- **Clarity** – clarify the main types of uncertainty

Establishing good communication is pivotal for patient support. Leisha highlighted that healthcare professionals must openly acknowledge knowledge gaps in rare conditions. Rather than concealing or misdiagnosing, they should actively look for solutions and create a transparent and supportive environment for patients and their families.

Leisha ended her talk by sharing her family's latest travels, highlighting that, while their journey is still ongoing, her mother is now doing well.

Read Leisha's essay here ›

SVP 2024 gallery ›

What matters to you?

The final talk was given by Kevin Ward, Area Teams Manager, Healthcare Improvement Scotland, centred around the 'What matters to you?' (WMTY) campaign. WMTY is an international person centred care movement, encouraging more meaningful conversations to improve the way health and care services are delivered and designed.

Kevin began by taking the audience through the four principles of patient-centred care:

1. Ensure everything is framed by **dignity, compassion and respect**

Ensure care is:

2. **Personalised**
3. **Coordinated**
4. **Enabling**

He explained that these principles underpin WMTY and emphasised that patient centred care entails engaging in meaningful conversations, not only with individuals, but also with their families and care givers.

Kevin reflected on the importance of **kindness** and how this basic skill must be the foundation of the healthcare environment. He described how being attentive to an individual's needs enables trust to be built and how this relationship can allow for therapeutic alliance and better outcomes for the patient.

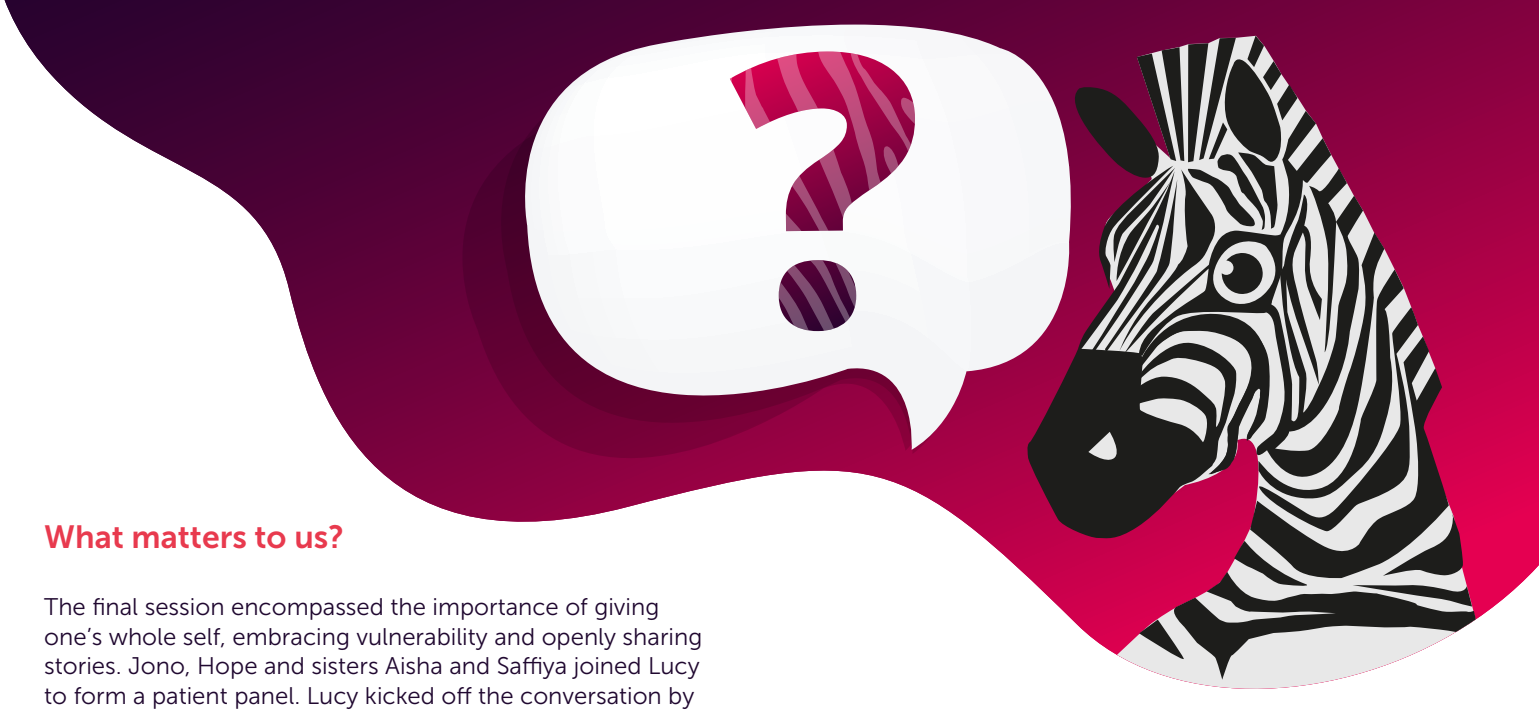
What matters to you? Kevin shared there are many ways to approach this to enhance patient experience and outcomes. He highlighted that the experience of a rare condition is very much a journey, so it is important to ask a patient WMTY questions and ensure these are tailored to the individual. He urged healthcare professionals to take a step back and consider the unique needs of the individual, and showed the audience some of the WMTY templates and conversation matrix.

Kevin emphasised the importance of what it means to **'really listen'**. On average, 80% of a conversation is taken up by the healthcare professional. It takes approximately 12–18 seconds for a healthcare professional to interrupt a conversation; however, if an individual is not interrupted, they will speak for 30–90 seconds. Therefore, it is important for healthcare professionals to take a moment to pause and actively listen to both the patient and family. Kevin highlighted those conversations can shape decisions regarding an individual's health and care decisions. So, when healthcare professionals truly listen to the patient, this can lead to improvement in the patient's well-being and their outcomes.

Kevin closed his talk by urging healthcare professionals to be compassionate and kind, encouraging them not to make assumptions and to witness the positive impact unfold.

Explore the WMTY template

WMTY template ›



What matters to us?

The final session encompassed the importance of giving one's whole self, embracing vulnerability and openly sharing stories. Jono, Hope and sisters Aisha and Saffiya joined Lucy to form a patient panel. Lucy kicked off the conversation by sharing her own healthcare journeys, creating a platform to discuss and explore what matters to everyone openly.

Lucy asked the panel **"How do you feel you've received patient-centred care or good communication?"**

Hope explained this is when she leaves an appointment feeling less anxious and more in control. She emphasised the importance of personal connections with healthcare professionals and how this allows patients to feel recognised as individuals and feel well supported. Aisha also reflected on the importance of the bonds she has formed with her doctors. She explained how strong bonds can help guide professionals in understanding what the patient wants if they are unable to advocate for themselves. Jono added that healthcare professionals are integral for the well-being of an individual with a rare condition. Saffiya described, from a sibling's perspective, the importance of professionals showing empathy toward both the patient and family, emphasising the need to reassure the entire family, taking into consideration everyone's mental health.

Lucy then asked the panel's thoughts on **"assumptions."**

Hope stressed the importance of empowering young people to be their own advocates and underscored the significance of professionals addressing them as individuals. The theme of being recognised as an individual was reflected upon throughout the conversations, with the panel each reflecting on their connections with healthcare professionals who have understood this aspect and recognised them beyond their medical diagnoses.

Jono highlighted how healthcare professionals should not assume all patients have had the same life experiences. Aisha discussed the crucial need for society to not stigmatise individuals with rare conditions and avoid biased judgements based on their upbringing.

Each story shared was unique, yet collective experiences emerged, reflecting that while these conditions are individually rare, they collectively shape a shared reality within the rare disease community.

Final thoughts from our panel:

"Include patients in communication and strengthen bonds"

– Aisha

"Treat both the patient and family with reassurance and empathy, considering everyone's mental health"

– Saffiya

"Words can make or break our heart"

– Jono

"Rare disease is a sexy topic to study!"

– Hope

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