The Unusual Suspects Report 2022

On Wednesday 9th February, emotive attended this year's Medics4RareDiseases (M4RD) Annual Symposium, "The Unusual Suspects: rare disease in everyday medicine".

The meeting was held in association with the Medical Genetics Section of the Royal Society of Medicine (RSM), virtually as an interactive meeting.

Senior Medical Copywriter Talia Coren, Senior Account Executive Charlotte Roe and Graduate Medical Writer Ledicia Botas Pérez joined the event to tell us more about what was discussed this year.

Meeting report

A very popular event in the rare disease calendar, the M4RD Annual Symposium encourages healthcare professionals, medical trainees and students to come together and learn more about the importance of a holistic approach to understanding rare disease, with a focus on providing practical tools and pragmatic tips for improving diagnosis and management. Last year, the online presence of the symposium meant that a higher proportion of international delegates could take part and this year was no different.

The evening started with Dr Lucy McKay, CEO of M4RD, who briefly introduced the speakers participating in this year's event and provided an overview of the evening's agenda. Lucy began by introducing M4RD, a registered charity driving an attitude change towards rare disease amongst medical students and doctors in training to fulfil the unmet needs of the rare disease community. Lucy went on to explain how insufficient medical education and training in rare disease as a collective group of conditions has partly contributed to the current unmet need we're seeing in the rare disease space. She discussed how M4RD has been working to increase rare disease awareness, reduce the so-called 'diagnostic odyssey' and improve patient experience by providing resources to the medical community. Lucy reflected on the progress made since M4RD started as a student society in 2011 by highlighting how rare diseases have become a national health priority, focusing on the UK Rare Disease Framework published by the Department of Health and Social Care. She reminded the audience that further work is needed, describing the case of Evan Nathan Smith, who died after an unrecognised sickle cell crisis in 2019 and the subsequent findings of the No-One's Listening Report. Lucy reiterated that this event offers an opportunity to reframe rare diseases in a way that is helpful for both doctors and patients.

Following her introduction, Lucy explained that despite the large collective impact rare diseases have – with an estimated 3.5 million people living with a rare disease in the UK – newborn genetic screening of only 9 conditions remains "a drop in the ocean" and the average time to diagnosis remains long at 4–6 years. A lack of information and consensus amongst specialists and an underestimation or disbelief in the presence of the condition often lead to the diagnostic process ending prematurely and treatment being inappropriate. Lucy called attention to the fact that many patients face a struggle when entering adulthood as their complex care is no longer coordinated by a Generalist Paediatrician. To help medical professionals integrate rare disease into everyday practice, Lucy shared some advice from the M4RD Rare Disease 101 e-learning course, which was partially developed in collaboration with emotive. Lucy reminded medical professionals that "common things are common and so are rare diseases" and encouraged them to "think rare" and become a rare disease ally. Other recommendations included considering rare diseases during diagnostic workups and making use of specialist resources and free tools, such as FindZebra and Red Flags for Genetic and Rare Disease. Lucy emphasised the importance of listening to patient stories from beginning to end and including at least three generations when researching family history.

Amongst her top tips, Lucy reinforced the fact that we need to remember the basics; knowing when to have a high level of suspicion of a rare disease and reviewing and revisiting diagnoses periodically. Final tips included knowing limitations, working in collaboration with other specialists and reaching out directly to relevant patient advocacy groups. The overarching goal of M4RD is to ensure people living with a rare condition receive a timely diagnosis, mental health support and holistic specialist care as well as support from patient advocacy groups and hope from ongoing research projects.

The first speaker was a patient with Morquio syndrome, Aisha Seedat. Aisha is a De Montfort University graduate, an ambassador for The Society for Mucopolysaccharide Diseases (MPS) UK, and a rare disease advocate through various channels. Aisha opened her talk by introducing herself and the UK Rare Diseases Framework, a policy paper established early in 2021 that aimed to improve the lives of those living with rare diseases. She shared her own experiences and insights into the rare disease community in parallel with the priorities set out in The Framework.

Regarding Priority 1, 'Achieving a final diagnosis faster', Aisha had an early diagnosis at the age of 6 months, which gave her access to adequate care during childhood. However, she acknowledged the diagnostic journey can be a long one for less fortunate patients, and when they finally reach a diagnosis, it may be too late to treat the condition or prevent its progression. Aisha herself has experienced difficulties accessing specific treatments due to complications identified following her initial diagnosis.

In relation to Priority 2, 'Increasing awareness of rare diseases amongst healthcare professionals', Aisha shared her belief that medical education and awareness constitute the foundation of medical engagement, enabling healthcare professionals to play an active role in identifying what can be done for a patient with a rare condition and not giving up when there are treatment or intervention options available.

As for Priorities 3 and 4, 'Achieving a better coordination of care' and 'Improving access to specialist care, treatments and drugs', Aisha and her family have witnessed a lack of communication between care teams at her local and specialist hospitals. They have also repeatedly encountered hurdles within the healthcare system concerning the distance travelled to receive care and access to specialist consultants via her GP. Aisha strongly believes that patients with rare conditions need regular, well-coordinated multidisciplinary specialist care, as well as dignified, priority access to urgent care and reassurance that everything possible is being done at what can be an important turning point in their lives.

The subsequent talk was given by Dr Denise Williams, Consultant Clinical Geneticist at Birmingham Women's and Children's Hospital NHS Foundation Trust. Denise opened her talk by highlighting reports over the last 20 years warning about infant mortality of genetic origin in the West Midlands and publications recommending the enhancement of genetic services in England. The National Genomics Healthcare Strategy – which has recently included the complete restructuring of clinical genomic laboratories in England to establish seven Genomic Laboratory Hubs and medical alliances – aims to provide the best genomic healthcare service in the world by integrating the latest genomics advances into routine healthcare and thus improving the diagnosis, stratification and treatment of genetic diseases. Measures such as 'mainstreaming' genetic testing – where patients are given direct access to testing through their specialist without the need to consult a clinical geneticist – contribute to a more streamlined and efficient experience for the patient, including earlier access to genetic-based treatments and relevant research.

A change in clinical practice from forward to reverse genetics, where a patient's genotype is tested before an attempt to establish a diagnosis, and the use of whole-genome sequencing, where every gene sequence is analysed before focusing on those most relevant to the clinical picture, are also speeding up the diagnostic process and reducing costs. Denise pointed out that patients need to be well informed about the possibility that genetic testing can fail to detect disease variants and/or pick up unintended genomic information. Healthcare professionals should also inform patients about the existence of a classification system for the severity of detected variants. Denise then moved on to discuss the lack of diversity both in genomic databases and the latest rare disease genome study, 'The 100,000 Genomes Project', posing a barrier to research and clinical practice. Denise closed her talk by emphasising the need to embrace inclusion and reach out to under-represented communities to develop databases that reflect real-world populations and, hence, tackle healthcare inequality.

Denise's talk was followed by a talk by Dr Sondra Butterworth, Community Psychology Specialist, founder of RareQoL and guest speaker on the M4RD podcast. Sondra opened by highlighting the fact that people living with rare conditions and their families tell their stories not just about their illness but also through their illness, sharing their personal perspective. She explored the concept of narrative-based medicine, which uses a holistic approach to study a patient's perspective through their diagnostic 'odyssey' and gain knowledge that can positively influence the patient's diagnostic journey and quality of life. Narrative-based medicine takes into consideration the fact that patient intersectionality – the complex, cumulative combination of individual characteristics – can lead to social and health inequalities. Therefore, a mixed methodology, which includes evidence-based quantitative medicine and narrative-based qualitative medicine before and after clinical considerations, could translate into improved quality of life beyond clinical outcome measures. Sondra then reviewed the red flags – identified in a survey run by M4RD – that get in the way of achieving an accurate diagnosis for patients with rare diseases; these include a range of issues, from multisystemic

and potentially complex physical and psychosocial problems to having to consult multiple specialists in search of a diagnosis. Sondra closed her talk by emphasising that patients must now be "at the front and centre of any rare disease implementation plan" and that patients will benefit from evidence-based medicine integrating narrative-based data.

The next talk was given by Dr David Adlam, Associate Professor of Acute and Interventional Cardiology at the University of Leicester, who plays a key role in the advocacy of spontaneous coronary artery disease (SCAD). David began his talk by describing SCAD in more detail; he explained that young to middle-aged women may suffer from SCAD before or after birth, when a bruise or bleed within an artery wall compresses the artery, diminishes blood flow and can lead to a sudden heart attack. David shared the story of Rebecca Breslin, who was a patient of his and a winner of the British Heart Foundation (BHF)'s Heart Hero Award 2016 after securing funding for the Beat SCAD foundation. He described how Beat SCAD has funded studies on accurate diagnosis, patient awareness campaigns, medical education for healthcare professionals and partnerships with larger organisations. He went on to tell the group that the foundation has also enabled international research identifying rare and common genetic risk factors and currently support important work at the Glenfield SCAD clinic in Leicester, where patients are referred from all over the world. David closed his talk by outlining the challenges that remain for the SCAD community; a lack of research support from funding bodies means research relies solely on patient contribution and a lack of support from specialist commissioning means specialist SCAD clinics risk being overburdened and under-resourced. The backlog generated by the COVID-19 pandemic also constitutes a challenge in maintaining the research and care needs of the rare disease community.

Lucy then introduced the evening's final speaker, Miss Zainab Alani, winner of the The Student Voice Prize 2021, who spoke about her essay "Putting the 'I' in Intersectionality: The Unspoken Pandemic". Zainab told the group a little about herself and explained that she is a first-year medical student at the University of Glasgow and a rare disease patient and advocate. Zainab shared that she suffers from myasthenia gravis, a rare disease that causes muscle weakness. She explained that her diagnostic journey started with uncertainty from her GP, disbelief from her optician and a subsequent game of 'ping-pong' that involved jumping from one specialist to another and back to square one. Her diagnosis was eventually communicated to her over the phone when she was alone at the vulnerable age of 15. Zainab reflected on her intersectionality and the role her age played in her diagnostic odyssey: "As a patient, my concerns were taken lightly, but I was also expected to be an adult [when receiving the diagnosis]". She highlighted how each patient's uniqueness can be a target for discrimination and encouraged the next generation of medical professionals to "act upon issues when they arise". Like Aisha, she emphasised that "we're not making enough of what we have".

Following Zainab's talk, the meeting was closed by a panel discussion led by Lucy. The host and all five speakers gave their thoughts on what had been discussed throughout the meeting and answered questions posed by the delegates. They discussed the importance of data collection and the creation of a rare disease patient registry, the relevance of applying a holistic approach to research, diagnosis and management and the need for funding. There was a consensus that medical education and training, with a focus on understanding the genetics of disease, are key factors that will significantly improve quality of life and care for patients with rare diseases.

Reflections

Reflecting on the meeting, Talia said:

"I truly think that every year, this event gets better and better. Listening to such a diverse panel and hearing the perspectives of so many different people in the healthcare space is so enlightening. I love how easy to digest the information is, and I always leave feeling extremely motivated to continue my work in the rare disease space with both emotive and M4RD."

Charlotte also gave her thoughts on the evening:

"M4RD's annual symposium is always a pleasure to attend. The range of speakers is incredible, and they cover all aspects of the rare disease community; HCPs, patients, relatives, students and more. My highlights this year had to be hearing from Dr David Adlam discussing his work with Beat SCAD and Dr Sondra Butterworth discussing intersectionality within the rare disease space."

Ledicia reflected on Zainab's story:

"Listening to Zainab's story really opened my eyes to the reality of the unconscious bias, inequality and contradictory expectations existing in our healthcare system. Her story clearly highlights the importance of educating healthcare professionals and places immense value on the work M4RD and emotive do to change the lives of those affected by rare conditions."