The Unusual Suspects Report 2021

On Wednesday 24 February, emotive were in attendance at this year's Medics4RareDiseases (M4RD) Annual Symposium, "The Unusual Suspects: Rare disease in everyday medicine". This year, the event could not take place at the Royal Society of Medicine (RSM) due to the ongoing COVID-19 pandemic, so instead, it took the form of an online interactive meeting. Senior Account Executive Claire Mitchell, Junior Account Executive Charlotte Roe and Graduate Medical Writer Emily Keizer all attended the event in order to tell us more about what was discussed this year.

Meeting report

Based on recent data gathered globally, there are currently over 400 developers active in gene therapy for rare disorders, and over 1200 ongoing regenerative medicine and advanced therapy trials worldwide, representing a constant growth of nearly 30% from the year 2000 up to now. The main indication for gene therapy trials is oncology, followed by monogenetic diseases, haematology and ophthalmology. It was clearly highlighted that such growth in the industry is based on fast-moving science. Over 19 billion dollars were raised in 2020 by the industry, shattering previous annual financial records (e.g. 9.8 billion dollars in 2019). Despite 2020 being a tumultuous year, the M4RD Annual Symposium has been a popular event in the rare disease calendar for many years, allowing healthcare professionals at all levels, trainees and students to come together to learn more about the importance of understanding rare diseases as a whole, with a large focus on improving awareness and management. In previous years, the event has had a UK focus; however, due to the meeting being held virtually this year, the reach has been bigger and has allowed delegates to attend from across the globe.

Dr Lucy McKay, CEO of M4RD, started the meeting by welcoming delegates to the online event and discussed the agenda for the evening, which this year had a focus on diagnosis. Lucy also reflected on the very different circumstances in which the event was being held in comparison to previous meetings and considered how the unknowns we have all faced with COVID-19 can be used to better understand what uncertainty feels like in the rare disease space. Lucy also thanked the RSM for their continued support and efforts to bring medical education opportunities to a virtual audience.

Following her introduction, Lucy went on to discuss Rare Disease 101 – a free, online, interactive course of e-learning modules created by M4RD. Rare Disease 101 is aimed at medical professionals in the early stages of their careers to educate them on the fundamentals of rare diseases, all in the hope of speeding up the journey to diagnosis whilst improving care for patients with rare diseases. Lucy also explained how the series of modules helps to address some of the issues highlighted in the Department of Health and Social Care's UK Rare Diseases Framework, specifically increasing awareness of rare diseases amongst healthcare professionals. Rare Disease 101 has been worked on by rare disease experts and organisations, including emotive who have supported M4RD with the development of this project.

The first talk was given by Dr Gareth Baynam, a clinical geneticist from Australia. Dr Baynam focused on "Y and How?", discussing why healthcare professionals should care about rare diseases. He considered how although individually these diseases are rare, collectively, they are common and have a large impact on healthcare systems. Dr Baynam also touched on a wider outlook in terms of finding treatments for rare diseases and how they can have advantages for patients with other illnesses. He used the example of statins, a class of drugs that were originally used for the treatment of hypercholesterolaemia but which now have a much wider use. Finally, Dr Baynam shared his thoughts on how to better approach the management of rare diseases and how to raise awareness within the healthcare system. He encouraged us all to consider system-wide thinking alongside the idea that one size doesn't fit all: we need to utilise different approaches and be more agile in order to improve the doctor–patient relationship and speed up the journey to diagnosis.

The next talk was given by Georgina Morton, founder of the ArchAngel MLD Trust and parent to Ava, who has been diagnosed with metachromatic leukodystrophy (MLD). Georgina discussed her experiences from a parent's perspective, including initial misdiagnosis, having to push for further tests and the lack of awareness of MLD from healthcare professionals, even after diagnosis. She also shared valuable insights on the process of finding a clinical trial, which Ava was accepted on to, and seeing other children who appeared to be in a similar condition to Ava but who did not get accepted. Reflecting on her family's experience, Georgina considered that the time it took for Ava to get a diagnosis

(10 months) was quick in comparison to other children who have been diagnosed with MLD. She summarised her talk by sharing the work that ArchAngel MLD Trust are doing to raise awareness among GPs, encouraging them to question the warning signs, and also gave an insight into the work the ArchAngel MLD Trust are doing in the campaign to push for a more in-depth newborn screening programme for all children in the UK.

Following on from Georgina's talk was a talk by Prof. Bobby Gaspar, Honorary Clinical Professor at Great Ormond Street Hospital and the UCL Institute of Child Health, and CEO of Orchard Therapeutics. Prof. Gaspar spoke about his work with haematopoietic stem cell (HSC) gene therapies and his ambition to find a treatment for MLD. He shared his insights into MLD as a disease – a fatal and rapidly progressing disease affecting the central nervous system – caused by a single gene defect in the ARSA gene. He discussed the concept of putting a working copy of the defect gene into HSCs and how cells with the correct copy of the gene will eventually distribute to the brain and treat patients with neurometabolic disorders like MLA. Prof. Gaspar concluded his talk by going through some of the treatment efficacy data with us, which demonstrated why the treatment has been approved for use in the EU.

Lucy then held a panel discussion with Georgina and Prof. Gaspar, where they reviewed the importance of MLD awareness and the impact that improved awareness could have on the families of children diagnosed with MLD.

Following this discussion, Abie Epstein and David Rose gave a talk on the "I am number 17" campaign, which was initiated and funded by the company Takeda. Abie from Takeda and David, a person living with an ultra-rare disease and an advocate at Rare Revolution magazine, started off with a brief video outlining the objectives of the campaign. The campaign focuses on 17 'changemakers', whose aim it is to show that although people may have had very different experiences reaching their diagnosis, there are still so many shared challenges that people who live with rare diseases face. Each changemaker was paired with an artist to produce a piece of work showing what it is like to live with a rare disease, and the artworks created will be exhibited in a gallery. Finally, David touched on what his experience of getting a diagnosis was like and the impact this can have on a person's life.

Lucy then introduced the winner of the 2020 Student Voice Prize, Catriona Chaplin, who spoke about her essay "Unmasked: An insight into three patients with rare disease in the COVID-19 pandemic". Catriona discussed what it was like to talk to three people who have been diagnosed with, or care for someone with, mastocytosis, a systemic disease which appears to affect only the skin but can cause a range of symptoms dependent on where mast cells proliferate in the body. She reflected on the challenges faced as a result of the COVID-19 pandemic, including shielding, conflicting advice from GPs and the general feeling of lack of support. Reflecting on this, Catriona considered that lack of awareness can increase the burden on people with rare diseases, and how talking to patients and understanding their needs is of utmost importance.

The meeting closed with a second panel discussion, led by Lucy with five of the meeting's speakers. Together, they reflected on what had been said throughout the event and answered any questions raised by the delegates. Topics mentioned included the importance of healthcare professionals knowing their limitations, discussions around what their individual wishes are for the rare disease space and how patients can properly benefit from support groups. There was overall agreement that the greatest tool that can be used by healthcare professionals is their ability to listen.

Reflections

Reflecting on the meeting, Claire said "It was amazing to hear from such a range of speakers covering many aspects in the rare disease space. Listening to Georgina's story was particularly moving and highlighted the value that Rare Disease 101 can bring to doctors, patients and families in the future."

Charlotte gave her thoughts on the evening: "Hearing directly from people whose lives have been affected by rare diseases, whether it be by themselves like David or a family member like Georgina, is hugely important in helping us understand the impact of rare diseases on people's lives. The personal touch that M4RD added complemented information from experts like Prof. Gaspar and really motivates me to get up every day and put every ounce of effort into the work that I am producing for M4RD and for my other clients in the rare disease space."

Emily reflected on Georgina and David's stories, "Hearing the experiences of people living with rare diseases really emphasises why it is so important to continue to raise awareness of rare diseases and the importance of patient groups working with healthcare professionals on this issue. Attending this meeting has shown me why the work M4RD and emotive do is so important and the impact it can have on the lives of patients and caregivers."