



Rare disease education is key to enabling medics to **achieve the best for their patients**

The rare disease revolution may have arrived, but medical education is still playing catch up.



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In an ideal world, a patient with a rare disease would have a timely diagnosis, mental health support, care in a specialist centre, excellent communication from their local hospital and GP, and receive support from a patient advocacy group in the hope of research providing a cure.

The reality is that most people with a rare disease wait longer than five years for a diagnosis, spending a lot of that time in the revolving door of GP and hospital appointments, endlessly repeating their health story. For many this “diagnostic odyssey” leads to social isolation, loss of income or education and worsening mental health.

Improving patient advocacy

Around 3.5 million people in the UK live with a rare disease, which is defined as a disease affecting no more than five in every 10,000 people. Most rare diseases have a genetic origin and will present in children, three in 10 of whom will die before age five.

One in 17 people will be affected by a rare disease in their lifetime. Yet in Europe, fewer than one in five clinicians say they feel confident in their ability to correctly diagnose a rare disease. Hansa Bhargava, M.D, Chief Medical Officer at Medscape Education points out: “It may take many years before a patient receives their diagnosis. This delay can often be attributed to the lack of familiarity among clinicians about the signs and symptoms associated with rare diseases.”

Disease heterogeneity, patchy infrastructural support and lack of local clinical expertise underlines the need for better diagnosis of rare disease, which is why M4RD, a charity registered in England and Wales, was set up. Its founders are doctors with a personal interest as well as first-hand experience of the limitations of rare disease medical training.

The charity aims to reduce the diagnostic odyssey and improve patient experience through advocacy aimed at medical professionals.

Resources include its online education module, Rare Disease 101, the basics of rare disease. Further information on the challenges patients and their caregivers face is also available via Medscape’s Rare Disease Learning Centre, which M4RD chief executive, Dr Lucy McKay, describes as “a strong signal to the global medical community that rare disease is relevant and it needs their attention”.

Transforming the odyssey

Dr McKay believes that all doctors have the potential to transform patients’ experience of rare disease. Its social media hashtag #daretothinkrare aims to inspire doctors to improve their own skills and care offerings.

Dr McKay feels that every doctor needs to understand the relevance of rare disease to their clinical practice, can work alongside their “expert patients” and develop a real sense of curiosity and compassion for what patients with rare and undiagnosed conditions are going through.



Even if there is no treatment, a diagnosis can at least help the patient and family understand what they are facing. They can start to focus on how to manage the condition and their ‘new normal’.

Effective symptom management

The charity believes that advances in genomics, diagnostics and therapeutics have transformed rare diseases into an exciting area of medicine for doctors, with a great potential for their development. For patients, better care coordination and more effective symptom management can offer quality of life improvements even for an incurable disease.

Dr McKay says: “Even if there is no treatment, a diagnosis can at least help the patient and family understand what they are facing. They can start to focus on how to manage the condition and their ‘new normal’. A management plan including symptom control and peer-support is vital, just as for other long-term conditions. There is no reason why people with rare diseases should be denied the chance to live life to the fullest. The hope is that in 10 years’ time, that fullest will be even fuller still.”

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