The Increasing Importance of Clinical Education in the Rare and Orphan Disease Sector

Diagnosing and treating rare and orphan diseases remains one of the life science sector's great challenges – there are effective therapies for less than five per cent of 7000 known rare diseases and it often takes over five years to reach a diagnosis. These problems are compounded by small patient populations, which are difficult to access and lead to significant challenges for the healthcare industry in rare disease management and support of these patient populations.

Rare disease patients require a more personalised approach than those with common conditions, and often these patients extensively self-educate about their condition. This is making patient support even more challenging as those with a rare condition are often very knowledgeable and have set ideas and perceptions before they come into contact with health professionals.

To respond adequately, the industry must not just support patients but extensively educate health professionals and other key stakeholders, such as patient HUB service providers and payor groups, that are involved in the ecosystem of care around a patient. Education ultimately empowers everyone to better understand the disease, make more informed and better choices and improve patient outcomes.

Tailored clinical education is a vital tool in the care for those with a rare disease. Here Nagore Fernandez, Head of Patient Solutions, Ashfield Europe and Canada (EUCAN) discusses its increasing importance and positive effect on care in the rare and orphan disease sector.

Recognition

Estimates vary, but the number of rare diseases the medical community has identified could exceed 7000. A rare disease is defined as affecting fewer than 1 in 2000 people in Europe and fewer than 200,000 Americans at any given time.

Rare diseases have a devastating impact on families. Of the approximately 7000 rare diseases, only a small proportion have an approved treatment and about 50% of rare disease patients are children. Tragically, rare diseases are responsible for 35% of deaths in the first year of life and 30% of children with rare diseases won't live past their fifth birthday.

Unfortunately, it is common that primary care physicians either do not recognise specific symptoms or don't make the link between common symptoms and a rare disease. As a result, the patient journey to an accurate diagnosis can be long and difficult. For people living with a rare disease, their quality of life is typically greatly affected by the chronic, progressive and frequently life-threatening nature of the disease.

Investing in clinical education raises awareness and encourages the healthcare industry and decision-makers to understand the needs of those living with rare diseases.

Treatment

Whilst there is generally no cure for a rare disease, there have been huge advances in treatment options.

The goal for first-generation treatments was survival, but second-generation treatments are now helping patients live a more 'normal' life and enjoy a better quality of living. Just last year we saw landmark breakthroughs in advanced genetic therapies, such as viral mediated gene transfer and CAR-T therapies.

Today, thankfully, adults and children with rare diseases can often live much longer and continue to enjoy the milestones of life through childhood, adolescence and adulthood.

Clearly treatment can be life-saving and life-changing. However, the lack of awareness around rare diseases, and the diagnosis challenge, still remains. And, as patients live longer, this creates whole new challenges to make sure they receive the level of care they need.

Diagnosis and Detection

Diagnosis is not always an easy journey – sometimes primary care physicians do not link the initial signs and symptoms to a rare disease because often the symptoms suffered by the individual mimic other more common diseases – this can result in patients being misdiagnosed. 40% of rare disease patients are misdiagnosed at least once, making it a very difficult journey for the individual. It is astonishing to learn that it takes 4.8 years on average to get an accurate diagnosis – and this can be even longer depending on the disease.

Because of this, investing in and supporting the identification of patients with rare diseases is very important. We know that early detection of genetic disorders is key for the survival of rare disease patients, even more so when dealing with children. For example, in lysosomal storage diseases like Fabry, a young patient can suddenly suffer from stroke or develop terminal kidney injury without early detection. Genetic testing is available, but awareness of the disease and genetic testing amongst physicians in contact with these patients is key.

Advocacy

Advocacy, including patient-driven advocacy is becoming increasingly important in driving research into and awareness of rare diseases.

Working with advocacy groups and raising awareness amongst the healthcare professional community via targeted campaigns in their practice or through social media is an important part of industry activity and now, thanks to advocacy, learnings and treatments derived from research are evolving.

Working Together

It is important to offer personalised support to patients with rare diseases and their families. This can be delivered in many different forms, including; informative campaigns for patients or their carers or education around the administration of the often very

complex therapies that may have been prescribed by specialist centres.

Partnering with pharmaceutical companies to deliver patient support programmes to the patients affected as well as their families and carers is also an effective way of garnering positive outcomes when considering support options.

Medical information is often complex, but it can be broken down to make it relevant and easier to understand, both in the content, style and language and the format it is delivered in. Dedicated writers, who are experts in both the therapy area and writing for people without medical training or expertise, are essential to this.

The industry needs to talk to patients to understand their experiences and challenges, continuing to get their feedback along their journey, and understand that their needs will change as they transition from early childhood, through to adolescence and adulthood.

This helps with understanding how a difficult journey to the right care increases stress for the patient and family and that disease-specific, multichannel awareness campaigns are key to addressing this challenge.

The objectives of these campaigns are to:

- Raise awareness of the symptoms with patients/families and encourage them to visit their primary care doctor and ask the right questions.
- Educate the primary care doctors.

Hands-on Intensive Support

Highly-trained, specialised clinical educators can make a huge difference to a patient's experience and their outcomes of treatment by providing:

- Supplemental training on treatment administration
- Support materials on managing side-effects
- Support using health psychology techniques shown to help patients cope
- Someone at the end of the phone to answer questions and talk through concerns.

Conclusion

The industry is quickly recognising that the challenges a rare disease patient meets throughout their journey should be met with a different skill set than those treated for common illness. Clearly, treatment can be life-changing, however the lack of awareness around rare diseases, and the original diagnosis challenge, needs to be addressed.

Post diagnosis, and in receipt of a medical prescription, patients, families and carers will still need support to find specialist care and navigate the maze of diagnosis problems.

Listening to these patients and their care givers and advocates and finding ways to identify with their unique concerns and struggles will ultimately benefit all.

Nagore Fernandez

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