Rare diseases

doi: 10.56012/nvvg4559

Medical writers are vital partners in raising awareness of rare diseases and advancing research.

Rare diseases affect around 1 in ten people, or an estimated 300 million people globally, with over 7,000 rare conditions. National definitions of a rare disease vary from a prevalence of 5 to 80 per 100,000 population. Due to low prevalence and lack of knowledge in the healthcare profession, this community often has a long journey to diagnosis, termed the "diagnostic odyssey", or even has several misdiagnoses along the way.

The zebra is the symbol that represents rare diseases. This originates from a quote from an American physician, Dr Theodore Woodward: "When you hear hoofbeats, think horses, not zebras". He taught medical students to think of more common conditions before the rare diagnosis. Thankfully, new rare disease training initiatives in medical schools and continued professional development programmes are beginning to change this. They emphasise that rare diseases are common, and rare diagnoses should always be considered.

In the complex landscape of rare diseases, medical writers serve as crucial gatekeepers of information between science, healthcare professionals, and patients. There are unique challenges for communicating about rare diseases. The terminology is often complex, with limited research, data from small clinical trials, or case studies. Our role is synthesising information from diverse sources, including clinical trials, patient-reported outcomes, and, where available, patient registries and real-world data.

There are unique challenges when communicating about rare diseases.

However, our profession's contribution to rare disease research and patient care extends far beyond document preparation. We require a particular set of skills, including emotional intelligence and the ability to understand the patient's perspective.

We become essential partners in advancing knowledge and raising awareness and hope for rare disease communities. We must also be familiar with communicating with diverse stakeholders, including regulatory bodies, pharmaceutical representatives, payers, and patient organisations. We could argue that medical writers who write in the field of rare diseases focus on patient-first priorities more than any other medical writing discipline.

Regulatory approval for rare disease treatments poses a unique set of challenges. We must develop expertise in emerging regulatory pathways specific to rare diseases and embrace new information technology to communicate

GUEST EDITORS

Sarah Milner
sarah_milner1983@hotmail.co.uk

Heather Mason
heathermason80@outlook.com

complex data. Crafting narratives to present evidence from much smaller data sets and often alternative study designs can significantly impact the success of rare disease drug applications.

About 80% of rare diseases have a genetic origin, and about half affect children. Therefore, other demands in writing for the rare disease population include crafting text to present information in a way that must be accessible to children, those with cognitive difficulties, or those who may be neurodivergent. Thus, as medical writers, we must be mindful of the audience and write with ethics and inclusivity in mind. We balance scientific accuracy with clarity.

Information can be scarce and complex; developing patient education materials, informed assent and consent documents, and lay summaries empowers patients and their families to make informed decisions. There is often an under-recognised impact on rare disease research and patient care. We must continue to evolve our skills to meet the ever-changing landscape of rare disease research.

First and foremost, we wanted to recognise the incredible rare disease community. Through necessity rather than choice, the patients and caregivers become experts in their condition, and yet, so often, their voices are silenced. In an honest and open patient interview, **Richard Farquhar** talks about the day-to-day challenges of living with a rare disease, how the community can help, and what he hopes for in the future.

Designing and executing a trial in rare diseases is full of predictable and unforeseen quandaries. From the identification of study endpoints and

Author information

Sarah Milner is currently the Director of Medical Writing at Eliquent Life Sciences, where she specialises in serious and life-threatening rare diseases in children. She joined the medical writing industry in 2009 and has held positions in both the pharmaceutical and consultancy sectors. She holds a degree in Biomedical Sciences and a PhD in Cell and Molecular Biosciences.

Heather L. Mason is a freelance medical writer with over 14 years of experience, specialising in rare diseases and specifically inborn errors of metabolism. She also has a passion for patient advocacy and inclusion.

EUROPEAN MEDICAL WRITERS ASSOCIATION

Submissions:
For instructions to authors, go to the journal section of EMWA's website (www.journal.emwa.org).
All manuscripts should be submitted to mew@emwa.org.

MIX
Paper | Supporting responsible forestry
FSC® C126839

limited patient population to finding investigators, **Bregje Mommaas, Mary H. Ryan, and Neha Agarwal** discuss the impact these have on studies and how they impact regulatory writers directly and indirectly.

An interview with **Kelley Hill** discusses the unique challenges and potential solutions when writing regulatory documents for rare disease indications for Europe and the United States.

Understanding the regional requirements for regulatory documents is also challenging, particularly in orphan drug development. **Katie Brooks, Pauline Haleux, and Montserrat Cuadrado** provide an overview of regional requirements for orphan drug applications and summarise considerations from the pre- to post-approval stages, emphasising the fundamental role of multifunctional collaboration throughout the process.

Writing clinical trial protocols for rare disease trials also offers unique demands and can often be extremely complex, time-consuming, and burdensome. **Philip Burridge** and **Julie Eastgate** describe an innovative approach using master- and sub-protocols to promote efficiency in the clinical investigation process and provide clarity to both study investigators and regulatory reviewers.

In recent years, model-informed drug development (MIDD) has come to the forefront, especially in rare diseases in children where data is scant. In an article by **Natalie Brine, Clare Dyer, and Kelly Smith**, the emerging role of MIDD in drug development and assessment is introduced. As the use of MIDD grows, the authors describe the importance of regulatory writers learning how to understand these outputs to facilitate translation into clear, strategically messaged, and impactful statements.

Ensuring population diversity in clinical trials is crucial but difficult to achieve, especially for rare diseases. **Cheryl Roberts** discusses the evolving regulatory framework aimed at encouraging diversity, the unique challenges in the rare disease landscape, and sustainable solutions. Regulatory guidelines emphasise the need for diverse participant representation, but implementation is complex due to small, dispersed populations. Strategies like adaptive trial designs, community engagement, and decentralised trials can help increase diversity. However, recent federal changes in the US raise concerns about the commitment to diversity and health equity in clinical trials.

The ultimate goals are to reduce the current challenges around diagnosis time, approve new therapies, and improve understanding of rare diseases' impact on healthcare systems.

The potential of real-world data (RWD) and patient registries in addressing research and knowledge gaps in rare diseases is discussed in an article by **Sara E. Mole, Emily Gardner, and Heather Mason**. It highlights the importance of early diagnosis, understanding disease mechanisms, and developing effective treatments. RWD, collected from various sources, including patient registries, can provide valuable insights into disease prevalence, natural history, and treatment outcomes. They also emphasise the benefits of patient registries in gathering comprehensive data, supporting clinical trials, and improving patient care. It advocates for the integration of RWD and digital health technologies to enhance research and treatment for rare diseases.

Chris J. Hendriksz, a medical doctor and Chief Community Impact Officer for A Rare Cause, shares his journey in medical writing for rare diseases. After his own child was diagnosed with a rare disease, he became an expert in inborn errors of metabolism. He describes his experience collaborating with medical writers and their essential perspectives in creating patient-friendly communications and supporting rare disease management. MWs play a crucial role in bridging gaps between patients, clinicians, and pharmaceutical industries, especially in low- and middle-income countries. Hendriksz calls for greater social responsibility and collaboration to support rare disease communities.

Roseline Favresse, the Research Policy and Initiatives Director at European Organisation for Rare Diseases (EURORDIS), discusses the rare disease landscape in Europe. EURORDIS represents over 1000 rare disease patient organisations in 74 countries, aiming to improve the lives of 30 million people with rare diseases. The European Rare Disease Research Alliance (ERDERA), a new alliance, aims to address research and funding gaps with a €380 million budget. The ultimate goals are to reduce the current challenges around diagnosis time, approve new therapies, and improve understanding of rare diseases' impact on healthcare systems. Advocacy for an EU Action Plan and a World Health Assembly Resolution is ongoing.

Judit Mészáros/EMWA Creative Team