Penning hope: The impact of medical writing for rare conditions

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Abstract

In this article, I explain my route into the world of rare conditions, my experience of working as a clinician, and my passion for raising awareness to all involved. I achieve this by providing education to healthcare professionals, patients, and patient advocates in low- and middle-income countries. I also discuss the vital role that medical writers have played in this mission.

Introduction

I qualified as a medical doctor in South Africa in 1985 from the University of Pretoria, where I also completed my master's degree in Sports Medicine. Following a rare disease diagnosis of my youngest child, I moved to the United Kingdom to continue my studies, becoming an expert in inborn errors of metabolism in paediatric and adult care.

I became a Member of the Royal Society of Physicians (MRCP) and a Fellow of the Royal Society of Paediatrics and Child Health (FRCPCH), with subspecialty registration after training at the world-renowned Willink Unit in Manchester, UK.

After spending more than 20 years as an academic, clinician, and researcher, I am now the Chief Community Impact Officer for A Rare Cause, a non-profit organisation based in England that educates clinicians on rare disease management in more than 50 countries, with the list growing annually. This creates hope for those with the least chance of being recognised.

Misunderstood and of no importance

Rare diseases and medical writing are often misinterpreted and undervalued. This article will explore the significant yet frequently overlooked impact that both have on the scientific community and society, highlighting my personal journey with each.

My more than 20 years of experience in rare disease management was triggered by the journey with our youngest child, who has a rare disease. During this journey, I made many acquaintances who became true friends, made up of medical professionals, patient organisations, pharmaceutical industry colleagues, and medical writers (MWs).

My first introduction to medical writing was very early in my career after I presented an awareness programme to a public audience. The MW connected with me afterward and said that they had really enjoyed my presentation, and that, although it was not their place, they wanted to provide some constructive feedback. I learned about MWs' skills in making presentations more visually attractive and using infographics to explain complex concepts. Their advice became very important later when I wanted to develop patient-friendly communications.[1]

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Through my experience, I've found that the most effective MWs are those who have been exposed to the rare disease community and understand its unique issues.[2] The unique challenge of medical writing for the rare disease community lies in the rarity of these conditions and the lack of extensive evidence. Consequently, the patient's perspective and input become especially important. The patients and families are also usually very knowledgeable about their condition and the impacts of the disease on family life and the wider community. MWs will usually have a science background, which would help them engage with the community, but this is the time to be humble, listen, and get a true insight into the frustrations and challenges of the community. Then follows the understanding of working with small groups, unique clinical trial designs, and the lack of any minimally clinically important (MCI) outcomes or validated tools, which are usually required in scientific publications. MWs are in a unique position to highlight these issues and help develop publications to support the MCI differences (MCID) in rare disorders.[3] Being involved in this community is becoming part of the solution. I will share examples of how this became a reality in my journey through rare diseases and the benefit of rare disease medical writing champions.

While working for the National Health Service, our interactions with MWs were primarily related to post-approval registry studies. Our medical writing colleagues brought valuable experience from the pharmaceutical industry to these projects. They had a vital role in the management of the registries by creating templates and online tools that helped streamline communication and collaboration, ensuring that the projects progressed smoothly. MWs may also be involved in setting up the kick-off and comment resolution meetings, developing the timelines, and ensuring alignment with industry guidance, for example, the International Committee of Medical Journal Editors' criteria for determining authorship.[4] They play a crucial role as intermediaries between the pharmaceutical industry and the egos of key opinion leaders. Their goal is to ensure that data is collected and presented in an accessible format, ultimately benefiting patients and non-expert readers.

In industry and early clinical development, regulatory medical writers specialising in rare diseases are once again invaluable. Some of the routine tasks, such as template development, formatting, submission guidance, and compliance with copyright, are no different from those for more common disease areas. Still, a regulatory MW experienced in the field will frequently identify gaps for developers. Some simple examples are: the submission for orphan designations and the tools and methods to calculate prevalence data for rare diseases; the ability to obtain Rare Disease Paediatric Vouchers and how these support pipeline development;[5] and posing critical questions during the synopsis/ protocol development phases, for example, "Have you heard about this option?" or "Have you seen this before working with another partner?". This interaction between MWs and early clinical trial developers brings added value, which can only be provided by those with expertise in the rare disease space.

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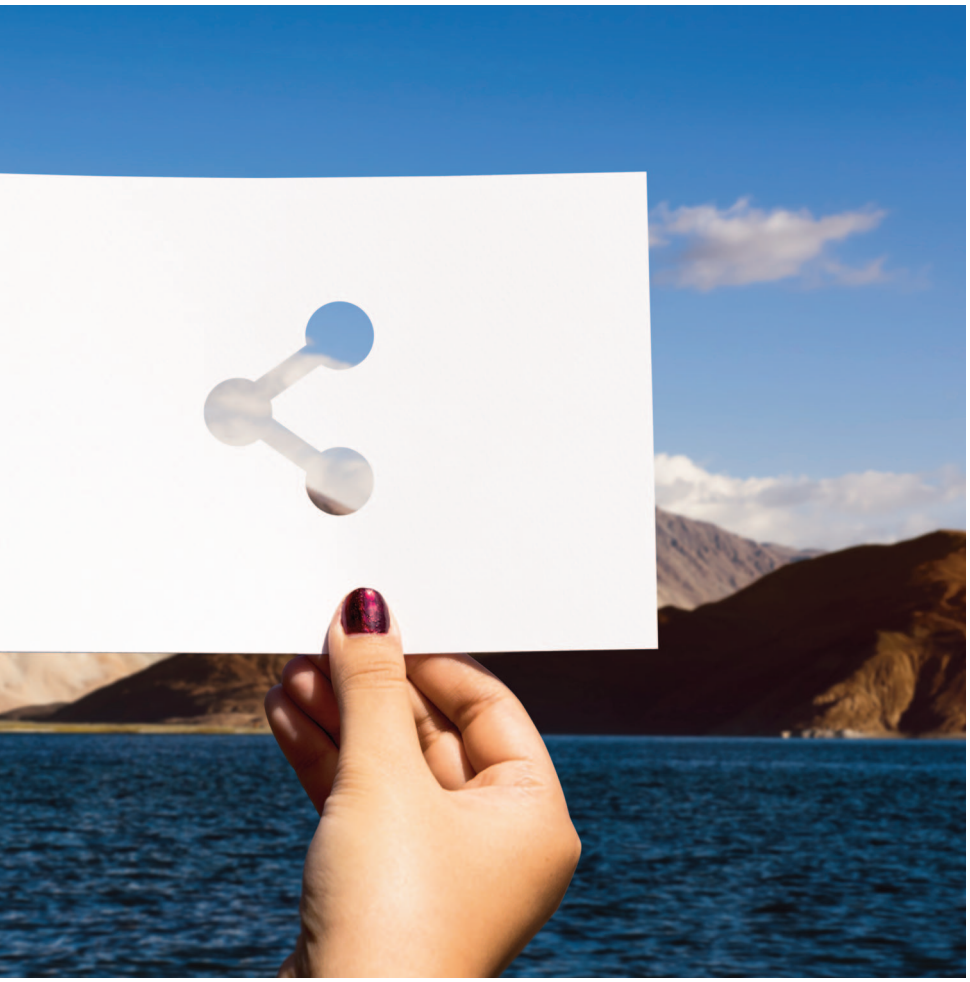
Medical writers can be patient advocates

Working on rare diseases in low- and middle-income countries (LMICs) is complicated, and having the voice of an impartial medical writer brings perspective and additional benefits. MWs can bring knowledge about working on different platforms and how low-cost measures like free or charitable collaboration platforms and virtual tools can be used to connect groups to build networks.

We started a very ambitious project called the Africa Roadmap project to build a clinical diagnostic network for those in Sub-Saharan Africa.[6] This was a joint project between multiple partners, including academia, patient organisations, laboratories, and charitable programmes. There was no funding to support medical writing, but on a pro bono basis, our MW colleagues provided advice and guidance on strategies to support the project. The Africa Roadmap project has now connected 10 Sub-Saharan countries, with more to come. Although the need for medical writing is well appreciated by those in high-income countries, it is still perceived as a luxury add-on in this context rather than an integral partner, and this is a genuine gap that needs to be filled in the future. Having MW experts help develop culturally appropriate medical guidelines and patient-friendly medical communications will become more important as this field develops. We have been able to create patient-friendly guidelines for high-income countries, but this should also become a reality for those less privileged. We frequently push the pharmaceutical industry to show some social responsibility by supporting charitable access programmes, but as there is no perceived financial benefit from these programmes, only a select few are helping to change this. This is a call to action for MWs and their organisations to examine social responsibility and how they can contribute to the United Nations Resolution.[7]

Lastly, don't underestimate the value MWs



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bring from their own connections and interactions with charitable organisations. Rare disease MWs tend to become proud advocates for our patients and can unlock unexpected pathways. Due to complex logistical pathways and infrastructure challenges, we have been struggling to fund appropriate measures to collect high-quality samples for our Africa Roadmap project. One of the project's MWs, who has over 20 years of experience in this field, shared with me that she attended a conference where she met with a charity interested in remote sample collection. She challenged them on whether they are considering working on this with LMICs. This led to an introduction where I learnt a lot more about the benefits of remote sample collection and all the tools that have been developed.[8] This quickly led to multiple additional introductions, with a project now planned for completion in early 2025, which is a potential game changer. This was just another example of the influence of rare disease MWs who are part of the winning team.

On multiple levels, medical writing and rare diseases are undervalued, but in LMICs they are truly forgotten. However, this can only change if we all feel our social responsibility to help those less fortunate than ourselves. Medical writing for rare diseases is a "Marmite" option, as it is frequently described; you either hate it or love it so much that it becomes part of your everyday life. Please consider joining Team Marmite. First, start by reading public social media posts on rare disease initiatives. Once you become interested and start engaging with the content, take on the challenge when a rare disease project arises. The rare disease community is wonderfully supportive and understands that everyone starts with no knowledge. Alternatively, attend rare disease sessions at conferences. Engage in conversations with attendees, and you'll quickly find yourself in meaningful discussions that may shift your work focus and definitely change your life.

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Disclaimers

The opinions expressed in this article are the author's own and not necessarily shared by his employer or EMWA.

Disclosures and conflicts of interest

The author declares no conflicts of interest.

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