From the Editor

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Rare experiences

My first experience with rare diseases was about 10 years ago, while working on a marketing authorisation for a gene therapy for a rare form of congenital blindness. We used the same regulatory guidances and document templates (i.e. ICHE3, ICHM4) like any other submission. What made this experience different were the trial participants – visually impaired children as young as 3 years old.

Clinical researchers on the Sponsor side do not get to know the participants in a clinical trial. It is part of the ethical and regulatory standards to protect patients and the integrity of the trial. But in a clinical trial of 12 participants, anonymity was difficult to keep. The participant was more than just an ID number on a form. They were very real kids. And I was a mother.

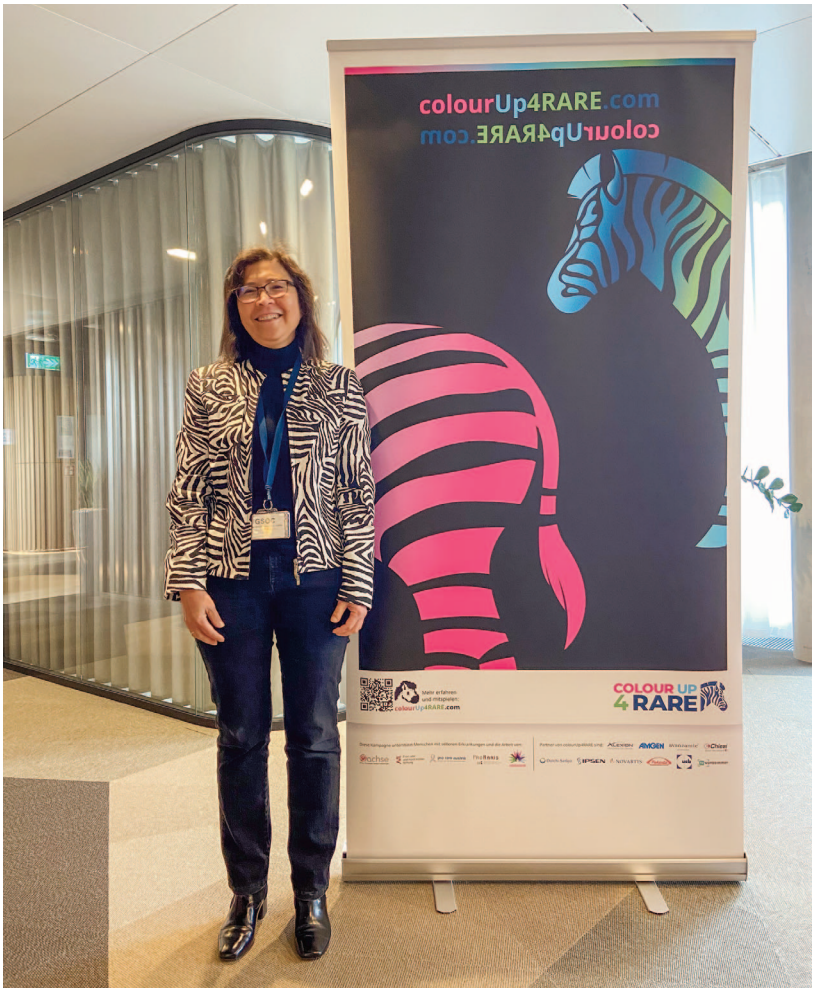
Sometimes we may inadvertently get to know our patients through many different channels, such as personal testimonials on TV shows and social media. Over the years, I got to see how those visually impaired children grew up and led normal, or even extraordinary lives, be it riding a bike, getting a driver's license, seeing a rainbow, or participating in America's Got Talent.[1,2]

It was just another job. We were not supposed to get emotionally attached to a project. But when I started writing those individual patient narratives based on audiovisual testimonials "*Yes, I can see the star, Mom!*", detachment went out of the window. And the requirement of at least 15 years of follow up[3] made it hard to completely forget these extraordinary subjects and move on.

It may sound banal, but that was the most rewarding (not financially, but emotionally) medical writing project I have ever worked on.

Many years later, I moved on to work for a rare disease company and personally met other rare disease patients and their families. I also had learned about orphans and zebras (see p. 51) and regularly celebrate Rare Disease Day on the last day of February.

My heartfelt thanks to Heather Mason and Sarah Milner for taking on the task of compiling this issue. Thank you to all our contributors who shared their rare experiences. And to people out there with rare diseases – this issue is dedicated to you.



Observing Rare Disease Day on February 28, 2025

References

1. Creed's Story: Seeing A Rainbow. Available from: https://www.youtube.com/watch?v=Jd40tbaTULk
2. Gene Therapy Restores Sight for Blind Patients with Hereditary Eye Disease. Available from: https://www.youtube.com/watch?v=LTWqFEOsmAk
3. US FDA. Long term follow-up after administration of human gene therapy products; Guidance for industry, January 2020. Available from: https://www.fda.gov/media/113768/download