

# Preface

Rare diseases impact more people than AIDS and cancer combined.  
[www.globalgenes.org](http://www.globalgenes.org)

The Twin Aims of this book were simple: to be clear and to be useful. Clear and useful not only to an academic audience, the typical target group of this sort of text, but also to be of relevance to the most important people of all: patients. We have departed from convention somewhat in not just assembling another textbook, one written primarily for academics and healthcare professionals. Instead, we have elected to present chapters, stories, and vignettes in as accessible manner as we could manage. In this way, we hope that the book will act as a landmark and key resource in the field of rare and orphan diseases, useful not just to professionals but also to patients and their families. We are confident that this book meets a few essential requirements:

- It should meet a long overdue need—specifically, to act as a much needed resource (for academics, health professionals, patients, and their families) which combines essential elements of health, empowerment, new social media, and judicious management.
- Be relevant and accessible—we wanted this text to be different in that it combines the essential competencies and perspectives of clinicians, managers, and patients, all presented in an easy-to-read manner.
- Be globally inclusive—we have included chapters, cases, and vignettes from around the world, sadly proving the prevalence of orphan and rare diseases.

In trying to ensure relevance and usefulness for a wide readership, we instructed contributors to use as few clinical terms as possible (other than those deemed absolutely essential). To ensure accessibility for all stakeholders, we have included as many patient vignettes and case studies as we could. These examples may echo your own questions and concerns. Many chapters either include or end with a relevant insight into patient conditions and needs, often written by patients (or their advocates). These cases are at once intensely emotional, insightful, and inspirational. Most importantly: they are real.

The complexities associated with rare and orphan diseases require a complex response. Thankfully, this task is made somewhat easier thanks to the continued and rapid progress of technology; the phrase *Health 2.0* attests to this fact. The advent

of new social media, such as *Facebook*, *Twitter*, *YouTube* (and similar outlets) has perhaps done more to *organically* enable “problem sharing” between affected disease communities than previous, targeted, initiatives. The ability to simply discover that there are “others like me” (via a web search) must give some hope and motivation. Connecting like-minded individuals together results in exchange of ideas, care practice, advice, and, most importantly, support. “People Power” is alive and well.

The multifaceted problems faced by the rare disease communities demand the respect of a similarly multi-layered response. Readers should be satisfied that the first such book of this type is edited by a team whose collective skills span medicine, integrated healthcare, knowledge sharing, and patient advocacy. Editors Bali, Bos, and Gibbons were particularly keen and subsequently pleased to convince a passionate patient advocate to join the team. Simon Ibell (founder and CEO of the *iBelieve Foundation*—a Canadian charity established to find a cure for Mucopolysaccharidosis II (MPSII, or Hunter’s Syndrome—and convener of the *Be Fair 2 Rare*<sup>TM</sup> public outreach campaign) provides the team with a powerful and credible voice when communicating with the rare and orphan disease communities. Simon’s story features amongst the vignettes within the book. Any lingering doubts that this is merely “just another academic text” should be dispelled.

It should be noted that all proceeds from the sale of this book are to be donated to the iBelieve Foundation ([www.ibelieve.com](http://www.ibelieve.com)).

The book is split into four interlinked sections: *Rare and Orphan Diseases*, *Health 2.0*, *Patient Perspectives and Empowerment Issues* and *Closing Gaps: Promising research and future considerations*. Each section contains a set of chapters which, together, contain key definitions and concepts, applied research and development projects, opportunities and challenges in the field, and, as previously mentioned, patient-focused case studies for many chapters. Building on our collective, extensive and wide-ranging experience, we trust that we have produced a book which presents a consolidated perspective of the intricacies involved. We hope that readers enjoy the book and trust that it achieves what it set out to do: to convey an important message...simply.

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