Epilogue

WE HOPE THAT this book has met its key objectives which were to be clear and simple. The plethora of perspectives from such a wide range of stakeholders (including patients, their families, health professionals, researchers, NGOs, Pharma, policy makers and charities) underline the difficulties faced by the rare disease community. Those working in the area already are all too familiar with its inherent frustrations—we envisage that this book will act as a guiding beacon to new arrivals to the field whilst offering support to others. Any newcomers should be under no illusions: this is an extremely challenging area littered with multiple obstacles (be they financial, legal, regulatory, clinical, technical and perhaps even ethical and moral).

With challenges, come opportunities. The proliferation of contemporary IT tools (the web, email, social media—the core of Health 2.0) has allowed patients and their families to be connected in a way that could simply not be imagined a few years ago. The diagnosis of a rare disease is harrowing enough—to hear that “sorry, we have no answer” must come as a hammer blow to patients and their families. Patient-led advocacy is perhaps strongest when answers are not forthcoming from the (professional) healthcare community and the power and passion that radiates from our vignettes attests to this perspicacity of spirit. “No” certainly does not mean “no”.

In discussion with many of the chapter and vignette authors, there was an overwhelming consensus regarding rare diseases: patients and families could not care less about regulation, policy and procedure. They want answers, they want a cure and they want it now. Stigmatization, lack of awareness, ignorance and other difficulties are not limited to any one rare disease—it could be argued that many of the challenges of one rare condition are replicated among the others. It is almost a case of “insert name of rare disease here”.

The very nature of rare diseases means that there is a limited number of patients and a paucity of expertise. The use of Health 2.0 has been very important in connecting patients and their families with one another. As exemplified by several of our chapters, patient-led advocacy and information groups can help drive
new research avenues as they act as an invaluable knowledge base. The rarity of a disease often means that patients themselves know more about their condition than the healthcare professionals, especially the challenges of living with it on a daily basis. Patients and their families are all too willing to be involved in research activities once they learn of new initiatives.

As communication and dissemination of promising rare disease research is of paramount importance, we trust that this book has offered new insights and a reassurance that multiple stakeholders are working on fundamental issues in the field. We look forward to increased patient-centric approaches and coordinated efforts in order to make better sense of the situation, which unify currently dispersed knowledge centers and which push for reform and action.

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