Communications in Medical and Care Compunetics

Rajeev K. Bali
Lodewijk Bos
Michael Christopher Gibbons
Simon R. Ibell *Editors*

Rare Diseases in the Age of Health 2.0

ICMCC
The international council on medical & care compunetics

Springer

Communications in Medical and Care Compunetics

Volume 4

Series Editor

Lodewijk Bos
International Council on Medical and Care Compunetics
Utrecht, The Netherlands

For further volumes:
http://www.springer.com/series/8754

This series is a publication of the International Council on Medical and Care Compunetics.

International Council on Medical and Care Compunetics (ICMCC) is an international foundation operating as the knowledge centre for medical and care compunetics (COMPUting and Networking, its EThICs and Social/societal implications), making information on medicine and care available to patients using compunetics as well as distributing information on the use of compunetics in medicine and care to patients and professionals.

Rajeev K. Bali · Lodewijk Bos
Michael Christopher Gibbons · Simon R. Ibell
Editors

Rare Diseases in the Age of Health 2.0

Springer

Editors
Rajeev K. Bali
BIOCORE
Coventry University
Coventry
UK

Lodewijk Bos
International Council on Medical
and Care Compunetics
Utrecht
The Netherlands

Michael Christopher Gibbons
Johns Hopkins Urban Health Institute
Baltimore
MD
USA

Simon R. Ibell
iBellieve Foundation
Toronto
Canada

ISSN 2191-3811 ISSN 2191-382X (electronic)
ISBN 978-3-642-38642-8 ISBN 978-3-642-38643-5 (eBook)
DOI 10.1007/978-3-642-38643-5
Springer Heidelberg New York Dordrecht London

Library of Congress Control Number: 2013944887

Printed on acid-free paper

Springer is part of Springer Science+Business Media (www.springer.com)

Never doubt that a small group of thoughtful, committed, citizens can change the world. Indeed, it is the only thing that ever has.

Margaret Mead

For our families and friends.

All proceeds from the sale of this book are to be donated to the iBellieve Foundation. (www.ibellieve.com)

Foreword

We continue to move very quickly through the period following the human genome mapping project, an international research effort to sequence and map all of the genes—together known as the genome. Newer methods of molecular diagnoses based on whole genome sequencing are utilized more frequently. Appropriate interpretation of the outcomes of sequencing efforts will become the norm for future diagnostic procedures. Expanding research, evaluation of potential therapies, and introduction of precision or personalized medicine interventions are now available for a few rare diseases. The rare diseases community anticipates the arrival of even more products into the market place for genomics identified subpopulation of both common and rare diseases.

Past research advances have made possible the molecular and enzyme replacement therapies for many diseases such as Gaucher disease and Fabry disease. Current research reveals a glimpse of the future with dramatic treatment effects from gene therapy and stem cell approaches to treatment. Regenerative medicine research offers hope to millions of patients worldwide. A greater emphasis will continue to be placed on the repurposing of investigational and approved products for uses other than the original intended patient population. High throughput screening facilities will provide access to products with potential application to other diseases through a more systematic approach to screening procedures for potential products for further development.

The patients, their families, and the patient advocacy communities have gained a more appropriate status as research partners to the traditional basic and clinical research and development efforts of the academic, federal, and biopharmaceutical research communities. Arrival and full utilization of the capabilities of the internet and world-wide-web approaches to information gathering, development, and distribution have provided ready access to reliable and useful information. Patient registries are considered useful tools to obtain a better understanding of the rare diseases across the lifespan. Developing an extended knowledgebase about rare diseases is now possible through a well-curated patient registry for an individual disease or a group of related disorders. The usefulness of a patient registry is dependent upon the quality of the data entered and the sustainability of a registry for a number of years to enable growth of the data sets for rare diseases.

More recent advances offered by social media and social networking provide the opportunity for gaining access to many more patients and families through

crowd sourcing data gathering efforts unknown in the past. Rapid access to thousands of patients connected by common interests is now possible. Using these communication techniques and development of more useful patient registries will increase patient recruitment and entry into clinical trials. The goal remains to gather significant information from as many patients as possible on a systematic global basis to provide for pan-diseases analyses and to increase the knowledge-base for the individual rare disease. For the research investigator and the pharmaceutical industry, more ready access to patients should speed-up the opening of a clinical trial, accrual of patients, and completion of the investigation. Delays in recruiting sufficient participants for clinical trials have always been a major barrier to rare diseases research and development.

Increased research opportunities exist for most rare diseases based on the greater availability of patients worldwide to participate in clinical trials, the improved willingness of the research community to develop multinational research teams, the heightened interest of the biopharmaceutical industry in orphan products and rare diseases, and access to global markets. Multidisciplinary and multisite collaborative partnerships are required for research of rare diseases and the development of orphan products. The rare diseases community is responding to the needs and scientific opportunities offered by rare diseases and in particular, if there is potential usefulness in the more common diseases. The extent of the rapid response to these needs and opportunities will determine how quickly we eliminate the disparities in access to treatment of rare diseases by orphan products. The uniqueness of the book provides perhaps the most valuable research baseline of information—the patient perspective. The growing acceptance of this perspective resulted in the empowerment of the patient, family, and caregiver as a necessary and major partner in the clinical research environment. Individual patient vignettes provide a history of their experiences with a rare disease and how they were able to develop a treatment program to live with a rare disease, even when there are no apparent therapeutic interventions. Increased communication among all of the partners in the rare diseases community is required if optimal care is to be provided to the millions of patients around the world who have received a rare disease diagnosis. Even greater efforts and support are required for the large number of patients who have not received the appropriate diagnosis and remain as patients with undiagnosed diseases.

Stephen C. Groft
Director, Office of Rare Diseases Research
National Center for Advancing Translational Sciences
Department of Health and Human Services
National Institutes of Health
Bethesda, MD, 20892-4874, USA

Preface

Rare diseases impact more people than AIDS and cancer combined.
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 *iBellieve Foundation*—a Canadian charity established to find a cure for Mucopolysacharridosis II (MPSII, or Hunter's Syndrome—and convener of the *Be Fair 2 Rare*™ 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 iBellieve Foundation (www.ibellieve.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.

August 2013

Rajeev K. Bali
Lodewijk Bos
Michael Christopher Gibbons
Simon R. Ibell

Acknowledgments

One of our key objectives for this book was *accessibility*. We hope we have achieved this by way of the numerous patient-focused case studies and vignettes and we are therefore indebted to the many kind folk who contributed to these, as listed below:

Patrice F. Band: *Autoimmune Polyendocrine Syndrome Type I* (*APS1*)
Jason Barron: *Recessive Dystrophic Epidermolysis Bullosa* (*RDEB*): *Sibling experiences*
Angela Covato: *Beta-Thalassemia Major*
Jocelyn Gardner: *Living with NOMID*: *Michael's Story*
Adrian (Ed) Koning: *Hope—Overcoming Fabry*
Marie Ibell: *A Giant of a Man*: *Simon Ibell* (*MPSII*)
Grainne Pierse: *The Blessings and Curse of Diagnosis—Myasthenia Gravis*
Deb Purcell: *The Journey of a Lifetime*
Jeneva Stone: *The Wilderness* and also *Route 125* (*October 2009*)
Roy and Zezee Zeighami: *MPSIIIA*(*Sanfilippo*)

We of course say a big thank you to all chapter authors for their wise and expert input. Between the editors, we have had the great pleasure of either meeting in person or conversing with many of the contributors. Such encounters were invaluable in shaping the book and we would like to thank everybody for giving up so much of their time for these discussions. Their constructive contributions have made this book much more relevant than it would otherwise have been.

We thank our publishing team at *Springer*, in particular Christoph Baumann, for their helpful assistance during the publication process. We are grateful to our team of reviewers (and, in particular, to the speedy dedication of Dr. Jacqueline Binnersley) for their comments and suggestions.

Finally, we reserve particular thanks for Dr. Stephen C. Groft, Director of the *Office of Rare Diseases* at the *National Institutes of Health* (*NIH*) for his insightful foreword and accommodating discussions.

About the Editors

Rajeev K. Bali, B.Sc (Hons), M.Sc, Ph.D., PgC, SMIEEE is a Reader in Healthcare Knowledge Management at *Coventry University* (UK). His main research interests lie in healthcare and clinical knowledge management, management of change, and integrated healthcare. He founded and leads the *Knowledge Management for Healthcare* (KARMAH) research subgroup (working under the *Biomedical Computing and Engineering Technologies* (BIOCORE) Applied Research Group). He is well published in peer-reviewed journals and conferences and is regularly invited to deliver presentations and lectures internationally. He is an invited reviewer for several journals and publishers and has been involved with the authoring and editing of several well-received books. In addition to overseeing various projects in the UK, he is leading (or has led) several international development and research projects (or short courses) involving partners from the UAE, the USA, Canada, Malaysia, Singapore, Nigeria, Oman, and Finland.

Lodewijk Bos, MA is founder and president of the *International Council on Medical and Care Compunetics* (ICMCC), an international foundation operating as the knowledge center for medical and care compunetics (COMPUting & Networking, its EThICs and Social/societal implications). He is series editor of *Communications in Medical and Care Compunetics* (Springer-Verlag). He maintains one of the leading websites providing information on health IT-related news and science articles. He is co-Editor-in-Chief of the recently founded IUPESM/Springer journal *Health and Technology*.

M. Chris Gibbons, MD, MPH is Associate Director of the *Johns Hopkins Urban Health Institute* and an Assistant Professor at the *Johns Hopkins Schools of Medicine and Public Health*. He is an urban health expert and informatician who works primarily in the area of consumer health informatics where he focuses on using health information and communications technologies to improve urban healthcare disparities. His work is forming the foundation of the emerging field of Populomics which represents the fusion of the population sciences, medicine, and informatics. Prior to coming to the *Urban Health Institute*, Dr. Gibbons was a Senior Policy Fellow at the *Centers for Medicare and Medicaid Services (CMS)*.

Simon Ibell is the founder of the *iBellieve Foundation* as well as the *Be Fair 2 Rare*™ campaign. A truly remarkable and inspirational individual, Simon is an advocate for Human Potential, a Role Model for Persons with Disabilities, and a crusader for the rare disease community. As one of 30 people in Canada (and approximately 2000 worldwide) born with a condition known as MPS II (mucopolysacharridosis) or Hunter Syndrome, living with MPS II has taught Simon that every moment of life is precious. He continues to learn how people perceive and handle disabilities and he uses this knowledge to lower the barriers and help bring about change. Receiving his degree in Sports Administration at the *University of Victoria* in June 2002, Simon is also a sought after speaker on human potential and overcoming obstacles. He has been awarded the *Spirit of Sport Story of the Year Award* at the 2003 *Canadian Sport Awards*, the *Queen's Golden Jubilee Commemorative Medal for Her Majesty Queen Elizabeth the Second* as well as the 2012 *St. Michaels University School Distinguished Alumni Award* and 2012 *University of Victoria Distinguished Alumni Award*. For more information on Simon, please visit: www.ibellieve.com, www.simonibell.com and http://nsb.com/speakers/view/simon-ibell.

About the Authors

Dr. Brendan Allison has been active in EEG research for about 20 years, most of which involved brain–computer interface (BCI) systems. He earned his Ph.D. in Cognitive Science in 2003 at *UC San Diego*, where he focused on BCIs based on visual attention (primarily P300) and imagined movement. He has since worked with several top researchers and institutes, including Prof. Wolpaw at the *New York State Dept of Health*, Prof. Polich at *The Scripps Research Institute*, and Profs. Pfurtscheller and Neuper at *Graz University of Technology*. He recently returned to his alma mater and is again with the Cognitive Science Dept. at *UCSD*. Dr. Allison's main interests involve making BCIs more practical, flexible, and robust. Despite considerable progress recently, most BCIs use simple interfaces that do not function well for disabled users outside of laboratory settings without ongoing expert support. These problems can be alleviated by combining BCIs with other communication devices and intelligent systems, such as hybrid BCIs, which is Dr. Allison's main research interest. He has also been active in addressing policy and infrastructural issues within the BCI community, such as surveying researchers and companies to assess their needs and analyzing and recommending specific directions.

Patrice Band lives in Toronto with his wife Jennifer, their daughter Julia, Julia's service dog Tory, Nibbles her rat, and Liberty her fish. When he is not spending time with his family and critters or playing hockey, he practises criminal law.

Jason R. Barron is Associate Director of Public Policy for the National Organization for Rare Disorders (NORD). His current focuses include a variety of operational activities that support NORD's efforts in advocacy, government affairs, and scientific affairs. Jason is a graduate of Dartmouth College, class of 2010. His previous work experience includes 2 years as a laboratory assistant in the Genetics and Molecular Biology Department of Dartmouth Medical School, as well as a 6-month internship as a junior legislative correspondent for health affairs in the legislative office of Patrick Kennedy, former Congressman of Rhode Island's 1st District. As the sibling of a young adult with Recessive Dystrophic Epidermolysis Bullosa, Jason brings his personal experiences as a rare disease patient advocate to NORD as he seeks to improve the lives of all affected by rare diseases.

Dr. Norman Barton's biomedical research career spans 30 years with leadership responsibility for clinical research programs in both the academic and industrial sectors. The unifying principle throughout has been the development of effective first-in-class therapeutics for genetic and metabolic disorders such as Gaucher, Fabry, and Niemann-Pick Disease, intractable gout, Duchenne Muscular Dystrophy and bronchopulmonary dysplasia in premature infants. Serving in the capacity of Chief, Clinical Investigations and Therapeutics Section at the National Institute of Neurological Disorders and Stroke, Dr. Barton conceived and executed the clinical development program that led to the approval of Ceredase as the first safe and effective enzyme replacement product for Gaucher Disease. Recognition for this contribution included the Meritorious Service Medal from the Public Health Service, the Outstanding Achievement Award from the National Gaucher Foundation and the Distinguished Alumnus Award from Pennsylvania State University. Dr. Barton earned a Ph.D. and MD at Pennsylvania State University. He was a resident in medicine at Albany Medical College Hospital, and a resident in neurology at Cornell University, the New York Hospital.

Dr. Cindy Bell joined *Genome Canada* in August 2000. From 2000 to 2008 she held the position of Vice-President, Genomics Programs in which she was responsible for providing policy and strategic advice on scientific and other aspects of *Genome Canada*'*s* programs. This included overseeing and managing the peer review process used to establish the research program of *Genome Canada*. In her role as Executive Vice President, Corporate Development she provides leadership in the development and implementation of strategic initiatives and approaches to enhance *Genome Canada*'*s* business model and secure funding to support genomics research in Canada. Prior to joining *Genome Canada*, Dr. Bell was a Deputy Director in Programs Branch at the *Canadian Institutes of Health Research* from 1994 to 2000. At CIHR she managed a number of research programs and was involved in policy development and implementation. From 1986 to 1994, Dr. Bell was a researcher at the University of California, Riverside. Her research focused on investigating the basic defect in the genetic disease, Cystic Fibrosis. She obtained her Ph.D. in Genetics from McGill University in 1986.

Dr. Raymond Bond completed a B.Sc (Hons) in Interactive Multimedia Design and a Ph.D. in Computing Science in 2007 and 2011 respectively. He teaches and carries out his research in the School of Computing and Mathematics. His research interests include health informatics, computerized electrocardiology, patient safety, and usability engineering.

Dr. Kym M. Boycott received her Ph.D. in Medical Genetics and subsequently studied Medicine and specialized in Medical Genetics at the University of Calgary, Canada. She is now a Clinical Geneticist at the *Children's Hospital of Eastern Ontario (CHEO*), Investigator at the *CHEO Research Institute*, and an Associate Professor in the Department of Pediatrics at the University of Ottawa. Dr. Boycott's research, bridging clinical medicine to basic research, is focused on elucidating the molecular pathogenesis of rare inherited diseases using next-

generation sequencing approaches. She is the Lead Investigator of the 'Finding of Rare Disease Genes in Canada' (FORGE Canada) project which is investigating the molecular etiology of almost 200 rare pediatric diseases.

Kyle Boyd completed a B.Sc (Hons) in Interactive Multimedia Design and an MA in Multidisciplinary Design in 2007 and 2009, respectively, from the University of Ulster. He is currently undertaking a Ph.D. in Leveraging Web 2.0 technology for Ambient Assisted Living. His research is to address social isolation in older carers of persons with dementia and chronic diseases. He is addressing both the barriers to use and usability issues of the current technology.

Angela Covato is the Managing Director of the *Canadian Organization for Rare Disorders (CORD)*. Since 2006, she has been actively involved in *CORD*'s advocacy work for a much-needed Canadian orphan drug policy, including an orphan drug regulatory framework. For the past 13 years, she has volunteered with the *Thalassemia Foundation of Canada*. She currently serves as the *Foundation*'s Research Grant Co-ordinator. The *Thalassemia Foundation of Canada*'s grant program provides medical research grants to new investigators in the field of Thalassemia. But most of all, she is the mother of an extraordinary teenager with a rare disorder.

Krissi Danielsson is a student at Lund University in Sweden, where she will complete her medical degree in 2015. During enrolment she has been active in research involving cancer and the PTEN gene and was the primary author of a review article on pancreatic cancer published in *Personalized Medicine* in early 2013. Krissi also holds a Bachelor's degree in Psychology from Excelsior College. Prior to studying medicine, Krissi worked as a freelance Internet content writer covering a range of topics, ranging from software-as-a-service to pregnancy-related issues and has been the author of four published consumer-oriented non-fiction books. After graduation, Krissi plans to pursue a career in oncology or medical genetics.

Dr. Remco de Vrueh works as senior adviser at *Schuttelaar & Partners*, a Dutch communications consultancy firm that strives to promote greater health and sustainability. As a personal initiative, he founded *Rare Disease Matters* at the beginning of 2012 to continue his research and teaching in the area of orphan drug development. Prior to this he worked for the *Netherlands Organisation for Health Research and Development* (*ZonMw*) as adviser to various (orphan) drug innovation programs. Between 2006 and 2011 he was active as Orphan Product Developer for the *Dutch Steering Committee on Orphan Drugs*. He was responsible for stimulating development of orphan drugs by the Dutch pharmaceutical industry. Apart from industry, he also actively interacted with academia and patient organizations. Remco has (co-)organized meetings, given presentations, provided training and teaching, and is the (co-)author of several scientific publications. In the same period he also fulfilled interim project management positions for the *European and Developing Countries Clinical Trials*

Partnership and the *Dutch Medicines Evaluation Board*. Before joining *ZonMw* he worked for seven years at *OctoPlus*, a Dutch pharmaceutical company. Remco de Vrueh holds a Ph. D. degree in Biopharmaceutical Sciences from the *Leiden-Amsterdam Center for Drug Research* in the Netherlands.

Dr. Mark P. Donnelly received both his Bachelor's degree (2004) and Ph.D. in Computer Science (2008) from the University of Ulster where he is currently a Lecturer with the School of Computing and Mathematics. He is a member of the institution's Smart Environments Research Group. His research interests include the application of connected health solutions to support independent living and remote monitoring of people with chronic conditions and, more recently, investigating the application of mobile technologies to support children with autism and their caregivers.

Jocelyn Gardner is a grandparent of a patient with Cryopyrin-Associated Periodic Syndromes. She is the President of the Canadian CAPS Network (CCN), whose mission is to improve the lives of all those affected by Cryopyrin-Associated Periodic Syndromes (CAPS) and related disorders. Amongst several objectives, CCN serves as a forum for bringing together patients and families, healthcare professionals, researchers, industry, funders, and policymakers to raise funds and promote health policies that improve the lives of those affected by CAPS and related disorders.

Dr. Rashmi Gopal-Srivastava is Director of Extramural Research Program in the Office of Rare Diseases Research (National Center for Advancing Translational Science) at the National Institutes of Health (NIH), Bethesda MD, USA. She oversees the national program on Rare Diseases Clinical Research Network (RDCRN). The RDCRN consists of 19 consortia and one Data Management Coordinating Center. She is responsible for developing and expanding the extramural research program to coordinate research activities on rare diseases across NIH in collaboration with other Federal Agencies, patient advocates, and other organizations. Prior to her current position she served as the program director for breast cancer SPOREs (Specialized program of Research Excellence) in the Office of Director, National Cancer Institute (NCI). Dr. Gopal-Srivastava received her Ph.D. in Microbiology and Immunology from the Medical College of Virginia, Virginia Commonwealth University, Richmond, Virginia in 1989 and received a Virginia Commonwealth fellowship. The same year she was selected for and awarded a Research Associateship from the US National Research Council of the National Academies of Science and joined the Laboratory of Molecular and Developmental Biology at the National Eye Institute, NIH and conducted research on regulation of gene expression for alphaB-cystallin (small heat shock protein). She has published several papers in peer-reviewed journals, written book chapters, and delivered invited oral presentations nationally and internationally. Dr. Gopal-Srivastava has received several honors and awards and is a member of a number of scientific committees.

Dr. Sylvie Grégoire is the former President of Shire Human Genetic Therapies (HGT), a division of Shire plc, the global specialty biopharmaceutical company. Headquartered in Lexington, Massachusetts, Shire HGT specializes in discovering, developing, manufacturing, and commercializing protein therapeutics primarily for the treatment of rare genetic diseases. Shire HGT employs 1,300 people in Massachusetts, and an additional 250 worldwide. Prior to joining Shire HGT, Dr. Grégoire served as Executive Chairwoman of the Board of IDM Pharma, a biotechnology company in California. Over the last 20 years, Dr. Grégoire has been the CEO of GlycoFi, held executive positions at Biogen Inc., and worked at Merck & Co. in the US and abroad. Dr. Grégoire received her Doctor of Pharmacy degree from the State University of New York at Buffalo, and her pharmacy degree from Université Laval, Québec City, Canada.

Stephen C. Groft, Pharm.D. is the Director of the Office of Rare Diseases Research (ORDR) in the National Center for Advancing Translational Sciences at the National Institutes of Health (NIH). His major focus is on stimulating research with rare diseases and developing information about rare diseases and conditions for healthcare providers and the public. To help identify research opportunities and establish research priorities, the office has co-sponsored over 1,200 rare diseases-related scientific conferences with the NIH research Institutes and Centers. Current activities include establishing patient registries for rare diseases, developing an inventory of available bio-specimens from existing bio-repositories, establishing a public information center on genetic and rare diseases, developing an international rare diseases research consortium, maintaining the Rare Diseases Clinical Research Network, and providing a special emphasis clinic with senior clinical staff for patients with undiagnosed diseases at NIH's Clinical Research Center Hospital. Steve received the B.S. degree in Pharmacy in 1968 and the Doctor of Pharmacy degree from Duquesne University in 1979.

Frank Grossmann, Dr. Med.vet. studied veterinarian medicine in Germany and conducted research at the ETH (Swiss Federal Institute of Technology) in Zurich. After supporting a start-up practice, he worked in management within the pharmaceutical industry. In addition to experience in dermatology, infections disease, nutrition, and orphan drugs, he is a recognized expert in sustainable businesses that deliver social impact. He is founder of a consulting company in pharmaceutical science, has been working as a guest lecturer in pharmaceutical science at the ETH/Zurich for many years and acts as a member of various organizations and boards. He is co-founder of Orphanbiotec, a social entity and is founder of Foundation Orphanbiotec, a Competence Center for Orphan Disease and Patient Empowerment.

Marie Ibell is the proud mother of Simon Ibell. Marie possesses over 30 years' experience in international business. She demonstrates strategic vision and possesses a portfolio of skills including governance, financial acumen, performance management, relationship-building, and the ability to champion change. She has 20 years' Board-level experience in organizations with significant

budgets and complexity, a considerable reputation within the private and financial sectors and has experience of building alliances and working relationships with a range of stakeholders.

Adrian Francis (Ed) Koning, P. Eng is married to Marlene and together they have three young adult sons and one granddaughter. In early 2001, at the age of 43—and as a result of kidney failure—Adrian was diagnosed with Fabry disease, a life-threatening orphan disease. His life expectancy was reduced to between 45 and 50. A few months later, he was on dialysis, began enzyme replacement therapy on compassionate use, and was blessed with a live donor kidney transplant. From 2003 to 2010, he worked with others to secure access and funding of Enzyme Replacement Therapy (ERT) for all Fabry patients in Canada and was the first President of the *Canadian Fabry Association*. He also served as the Vice President of the *Canadian Organization for Rare Disorders* and the Secretary of the *Fabry International Network*. He continues to utilize his skills and abilities as a professional engineer to help improve the lives of those suffering from Fabry disease as well as support their families and caregivers. He continues to educate society and raise awareness about rare disorders and the need to ensure Canadians have access and funding to "orphan" drugs and therapies that are available in other parts of the world. He believes that Canada must not only adopt an orphan drug policy (ODP), but must also provide a comprehensive care program that includes the medical, psychological, and social needs of patients, families, and care givers who suffer the impact of living with a life-threatening rare disorder.

Caren Kunst is a healthcare consultant with a growing interest in digital healthcare. She is also a nurse and an experienced (homecare) mother of a 21-year-old son with the orphan disease Tracheoesophageal fistula (TOF)/ Esophageal atresia (OA). She supports parents globally using such social media as *Facebook*, *Hyves* (a Dutch social network) and *NING* (an online platform for people to create their own social networks). Her aim is to organize effective and safe self-management in home and hospital situations.

Prof. Paul Lasko received his Ph.D. from the Massachusetts Institute of Technology in 1986 and joined McGill in 1990 after a postdoctoral period at the University of Cambridge. Using the Drosophila system, Dr. Lasko's research concerns regulatory processes that control gene expression at the levels of mRNA stability or translation, and that underlie germ cell or early embryonic development. He received the Award of Excellence from the *Canadian Society of Genetics* in 2004. At McGill he served as Chair of the Department of Biology from 2000 to 2011. He assumed his position at CIHR in May 2010 but maintains his research lab at McGill. Dr. Lasko has been highly active in research grant adjudication and served on CIHR or *Canadian Cancer Society* grant panels continuously since 1995. He has also worked extensively for the *Human Frontiers of Science Program Organization (HFSPO)* over the past 10 years, serving on its program grant panel from 2001 to 2005, and then as one of two Canadian representatives on the Council of Scientists. He chaired the HFSP Council of

Scientists from 2007 to 2010. Dr. Lasko also served as President of the *Genetics Society of Canada* from 2007 to 2010. As Scientific Director of the CIHR Institute of Genetics, Dr. Lasko oversees the Institute's strategic research funding initiatives, many of which involve fostering international partnerships. He is the incoming chair of the Executive Committee of the *International Rare Diseases Research Consortium*.

Dr. Gaye Lightbody received an M.Eng. (1995) and a Ph.D. (2000) in Electrical and Electronic Engineering from *Queen's University Belfast*. She worked in industry for *Amphion Semiconductor Limited* developing high performance FPGA and ASIC cores for image and audio processing before returning to academia in 2005. Since then she has been a Lecturer with the School of Computing and Mathematics in the *University of Ulster*. Her early research interests included high performance VLSI hardware design, FPGAs, and Adaptive Filtering. Progression into Biomedical Signal Processing followed, working in the area of automatic detection of Auditory Brainstem Responses for determining hearing threshold. Recent involvement in a project for development of Brain–Computer Interfaces has expanded her work in the medical computing domain. More recent activities have involved computer systems for supporting remote management and rehabilitation of children with autism. Dr. Lightbody has undertaken a range of teaching responsibilities from Advanced Computer Networks, Mathematics, Web Design, and Health Informatics. She received her PG Certificate in Higher Education in December 2012 and continues to perform a small level of pedagogic research activities alongside her teaching and scientific research activities.

Jimmy Lin, MD, Ph.D., MHS is the president of the *Rare Genomics Institute* (RGI), the world's first platform to enable any community to leverage cutting-edge biotechnology to advance understanding of any rare disease. Partnering with 18 of the top medical institutions, RGI helps custom design personalized research projects for diseases so rare that no organization exists to help. Dr. Lin is also a medical school faculty member at the Washington University in St. Louis and led the computational analysis of the first ever exome sequencing studies for any human disease at *Johns Hopkins*. He has numerous publications in *Science*, *Nature*, *Cell*, *Nature Genetics* and *Nature Biotechnology* and has been featured in *Forbes*, *Bloomberg*, the *Wall Street Journal*, *The Washington Post*, and *The Huffington Post*.

Amanda Lordemann is a senior at Washington University in St. Louis studying Philosophy-Neuroscience-Psychology. She joined the *Rare Genomics Institute* during the summer of 2012 as an intern with the patient advocacy team. She has been contributing to efforts in patient education, social media, marketing, and human resource management. After graduation, Amanda is interested in a career related to healthcare and medical research.

Dr. Alex E. MacKenzie received his MD and Ph.D. at the University of Toronto and specialized in Pediatrics at the University of Ottawa. He is currently a

Pediatrician at the *Children's Hospital of Eastern Ontario (CHEO)*, Senior Scientist at the *CHEO Research Institute*, and a Professor in the Department of Pediatrics at the University of Ottawa. Dr. MacKenzie's research is focused on the identification of treatment modalities for inherited pediatric disease using the neuromuscular disorder spinal muscular atrophy as a model for the repurposing of clinically-approved agents.

Dr. Paul McCullagh received a B.Sc (1979) and a Ph.D. (1983) in Electrical Engineering from *Queen's University of Belfast*. He is a Reader in the *School of Computing and Mathematics* at the the *University of Ulster* and is a member of the *Computer Science Research Institute*. He specializes in the teaching of data communications, computer networking, and health informatics. His research interests include Biomedical Signal and Image Processing, Data Mining, Brain Computer Interface, and Assisted Living applications. He is interested in advancing the education and professionalism of biomedical engineering and health informatics. He has worked on the EU FP7 *BRAIN* project for e-inclusion using the brain–computer interface, EPSRC SMART 2 for self-management of chronic disease, TSB NOCTURNAL for assisting people with dementia and ESRC *New Dynamics of Ageing*, *Design for Ageing Well* funded projects. He is a member of the *British Computer Society*, the *European Society for Engineering and Medicine* and the *UK Council for Health Informatics Professionals*.

Daniela M. Meier, Ph.D., worked in a leading position of a Swiss agency promoting innovation and entrepreneurship before she started her own business *Manda Idea Management*. She studied Modern History, Political Sciences, and Iranian studies at universities in Bern (Switzerland) and Oxford (UK).

Dr. Pierre Meulien was appointed President and CEO of *Genome Canada* in October 2010. Prior to this appointment, he served as Chief Scientific Officer for *Genome British Columbia* from 2007 to 2010. From 2002 to 2007, Dr. Meulien served as the founding CEO of the *Dublin Molecular Medicine Centre* (now *Molecular Medicine Ireland*) which linked the three medical schools and six teaching hospitals in Dublin to build a critical mass in molecular medicine and translational research. The Centre managed the Euro 45 Million "Program for Human Genomics" financed by the Irish government and was responsible for coordinating the successful application for the first *Wellcome Trust* funded Clinical Research Centre to be set up in Ireland. For over 20 years, Dr. Meulien has managed expert research teams with a number of organizations, including *Aventis Pasteur* in Toronto (Senior Vice President of R&D), and in Lyon, France (Director of Research). He also spent 7 years with the French biotechnology company *Transgene* in Strasbourg, France as a research scientist and part of the management team. Dr. Meulien's academic credentials include a Ph.D. from the University of Edinburgh and a post-doctoral appointment at the Institut Pasteur in Paris.

Laura Montini is a blogger and editor for *Health 2.0 News*. She received her B.A. in Journalism from the University of North Carolina at Chapel Hill. As a

student, her reporting focused on several of North Carolina's large health systems as well as the state's internal debate on United States healthcare reform. She has spent much of the past 2 years in San Francisco, California editing *The Health Care Blog* and writing about technology for *Health 2.0 News*. She is passionate about good healthcare reporting and believes that healthcare is too important of an issue for the public not to understand it. Laura currently lives in San Francisco but is a New Jersey girl through and through.

Prof. Sara Newman holds a Ph.D. in Rhetoric and is a Professor in the Department of English at Kent State University, and a member of its Ph.D. program in Literacy, Rhetoric, and Social Practice. She is author of *Aristotle and Style and Disability and Life Writing: A Critical* History as well as articles in such journals as *History of Psychiatry*, *Rhetorica*, *Disability Studies Quarterly* and *Written Communication*. Her current research deals with the rhetoric of disability and medicine.

Prof. Chris Nugent is Professor of Biomedical Engineering at the University of Ulster. He received a Bachelor of Engineering in Electronic Systems and D.Phil in Biomedical Engineering both from the University of Ulster. His research within biomedical engineering addresses the themes of the development and evaluation of Technologies to support independent living. From a technological perspective his work has focused on the integration of mobile devices within smart environments coupled with the development of activity recognition systems. He has published extensively in these areas with work which spans theoretical, clinical, and biomedical engineering.

Grainne Pierse was born and raised in Edmonton, Alberta in Canada. She grew up swimming competitively and Irish dancing, both at a national level until the end of high school. At this point she had been recruited to swim with the University of British Columbia (UBC) swim team and moved out to Vancouver in September of 2008 to pursue this. She had two sisters compete with the UBC team before her and two more have followed since. In 2010, she was forced to take a year off from training and racing due to her myasthenia gravis but 3 months after her surgery she was back at it. Since then, she has won two national championships with her team and is a current Canadian record holder as a part of a 4 x 100 medley relay team. She is currently completing an undergraduate degree in Psychology with hopes of going on to do veterinary medicine.

Deb Purcell is a home-schooling mother to three awesome kids (Trey, Avery, and Sadie) and partner to an amazingly supportive and wonderful man, Ryan. She is an advocate for MPS and all rare diseases in Canada and around the world. When she is not scouring the Internet for the latest research, updates, and journal articles in MPS, or raising funds for research, you can find her reading, doing puzzles, and playing street hockey with her kids or watching them at soccer, hockey, gymnastics, and baseball. You can also find her on her yoga mat all over Vancouver and North America, at home, in hospital, at studios, or out in the sunshine. (www.treypurcell.com).

Dr. Francis (Fritz) Paul Rieger is Associate Professor in the Odette School of Business at the University of Windsor in Ontario, Canada. He has been a regular lecturer with the College of Business, Department of Management Studies at the University of Michigan-Dearborn, USA. Dr. Rieger began his academic teaching career as an Assistant Professor at Oklahoma State University in 1982. He came to the University of Windsor in 1984, where he has served as Director of the *Business Resource Centre* (1995–2000) and Director of the MBA Program (1997–2000). He has served as a Visiting Professor at Université du Québec à Trois-Rivières; Queen's University, Lyon Graduate School of Management, France, the University of Michigan, and the University of Electronic Science and Engineering China (People's Republic). Prior to his academic career, Dr. Rieger worked in the private sector in the Finance area as a Field Engineer for IBM, Stockbroker and Financial Analyst in Anchorage, Alaska, and for the Cree Indian School Board in Val D'Or, Quebec. Dr. Rieger carries out research and publishes on topics of business ethics, global business environment, strategic management and healthcare. Dr. Rieger has a B.Sc. (Physics Education) from Manhattan College, USA, an MBA from Columbia University USA, and Ph.D. (Management) McGill University QC, Canada. He has many published articles and a book.

Michael Seres was diagnosed aged 12 with the incurable bowel condition Crohn's Disease. After over 25 surgeries, and intestinal failure, he became only the 11th person to undergo a small bowel transplant in the UK at *The Churchill Hospital* in Oxford. Michael started blogging to chart his journey from Crohn's to a Bowel Transplant. http://beingapatient.blogspot.com is now syndicated on over 20 websites and has had received 55,000 views from transplant teams, medical students, and patients. He has developed an understanding as to how patient-to-patient interaction can be a powerful tool to assist in recovery. Michael mentors many patients and their families and is a published author and professional speaker. He uses social media to develop global online communities and devises social media strategies around patient engagement. He is the patient lead for #NHSSM, a facilitator for the *Centre for Patient Leadership* and is Digital Strategy Advisor to *The Patients Association* and the *Oxford Transplant Centre*.

Dr. Hannah Spring is a Senior Lecturer in Research and Evidence-Based Practice within the Faculty of Health and Life Sciences at *York St John University* in the UK. Prior to working at York St John, she held a variety of posts in Department of Health and NHS settings. By background, she is a clinical information specialist and is particularly experienced in working with health professionals in the primary and secondary healthcare, and academic sectors. As well as 10 years' teaching experience, she has worked in a variety of independent consultancy roles including working with *Nottingham University* as an independent reviewer and content provider for the *Intute Health and Life Sciences* database, and as a consultant information specialist for general practitioners. She has significant experience in research and information and knowledge management in the health and academic sectors, and have published widely in my

specialist areas. She is currently editor for the *Learning and Teaching in Action* regular feature of the *Health Information and Libraries Journal*, and a member of the editorial board. Her specialist interest areas include systematic reviews and associated research methodologies, research development in health LIS professions, clinical librarianship in primary care and the allied health professions, evidence-based health practice, the impact of Internet and web 2.0 technologies on learning and information behavior, and information literacy and health.

Therese Stutz Steiger, Dr. med., is a physician specializing in prevention and public health. She has worked at the Swiss Federal Office of Public Health (BAG) dealing with new issues, health expertise, and non-contagious diseases (specifically cancer). She co-managed Online Services and Empowerment and implemented the eHealth Switzerland strategy initiated by the federal government. She works as an private consultant in public health, as a lecturer, teacher, and publisher on healthcare issues and is a member of several committees and advisory councils in the public and private sectors.

Hugh Stephens has been interested in the intersection between healthcare and social media for some years now both as a researcher and in its ability to change the way that we provide healthcare. Hugh is an MBBS/Ph.D. candidate studying at *Monash University* in Melbourne, Australia. Hugh is completing his Ph.D. at *The Alfred Hospital*, conducting research into resource allocation issues for deceased organ donation across Australia and is also conducting additional research into the use of social media within the health sector in Australia. For the last 3 years, Hugh has worked on the External Advisory Board of the *Mayo Clinic's Center for Social Media* based in the USA, encouraging health organizations and professionals worldwide to embrace social media technologies. He also works closely planning part of the program of the *International AIDS conference*, the largest health-related conference in the world, attracting 27,000 people in 2012 (Washington, DC).

Roy Sterritt is a faculty member in the School of Computing and Mathematics at the University of Ulster and a member of the University's Computer Science Research Institute. Internationally recognised as a leading innovator in Autonomic Computing and Communications (self-managing and self-healing computer-based systems) with currently 13 patents with NASA and 175 + publications in the field. He was the founding chair of the *IEEE Technical Committee on Autonomous and Autonomic Systems (TCAAS).* Roy has also served on the *IEEE CS Publications board* and chaired the *Conference Publications Operations Committee (CPOC)*, the *Conference Publications Committee (CPC)*, the *Conference Advisory Committee (CAC)* and also served on the *IEEE CS Technical and Conferences Activities Board (T&C Excom)*.

Jeneva Ellen Stone is a writer and editor who lives with her family in the Washington DC area (USA). She is the recipient of fellowships from the MacDowell and Millay Colonies for her work in nonfiction. Her poetry and non-

fiction have appeared in many literary journals, including the *Colorado Review*, *Poetry International*, *Pleiades* and *The Collagist*. Jeneva is currently working on a memoir about her son's rare disease.

Tracy VanHoutan was raised in Marshalltown Iowa, attended University of Iowa in Iowa City, and now lives in the Chicago-land area with his wife Jennifer and their three children, Noah, Laine, and Emily. Two of his three children (Noah and Laine) are affected by the Late Infantile form of a devastating childhood disease known commonly as Batten disease. Tracy has worked in the proprietary trading field trading various financial products for the past 16 years. Since his children's diagnosis, Tracy and his wife started a local organization called *Noah's Hope* to help raise awareness and funds for Batten disease. Tracy currently serves on the *Batten Disease Support and Research Association* board of Directors. Tracy regularly attends research conferences related to Batten disease and has been part of a small group of parents that actively fund peer reviewed projects on a regular basis. In addition to his Batten-related work, Tracy has become very active in the rare disease community within the United States. These efforts include testimony before the FDA on two occasions as well as speaking to a panel at the NIH on cell-based therapies for rare diseases. Tracy has also been very active in speaking to members of the United States Congress and organizing grassroots efforts to further common causes between all rare disease.

Dorothy Weinstein has a long tenure working in Washington DC on national, state, and local health policy. She has been employed at *Georgetown University*'s *Institute of Health Policy*, the *Association of American Medical Colleges*, the *American Diabetes Association*, the *Endocrine Society*, the *American College of Cardiology*, and the *National Health Council*. Her various positions in health policy have been broad based including research and writing, crafting legislation, and directing government relations departments at leading major non-profit health organizations. Her most recent activities are working on designing and now implementing healthcare reform legislation in the United States. The focus of her efforts is on patient engagement in the health care delivery process. Dorothy has a background in healthcare volunteer and philanthropic work at the *Children's National Medical Center* (CNMC) and at the *Prevention of Blindness Society* in Washington DC. Dorothy has a B.A. degree magna cum laude from the honors program in philosophy from the University of Maryland and an M.A. degree from Duke University in Public Policy. She is a member of Phi Beta Kappa and is a published author in the area of fetal tissue/stem cell research and environmental policy on sound and noise abatement.

Dr. David Whiteman joined Shire Pharmaceuticals 8 years ago as medical director of the Elaprase program, and principal medical director in the (then) newly developing department of Global Medical Affairs at Shire HGT. He provided medical leadership and expert input to the pivotal clinical trial and eventual approval of Elaprase for the treatment of Hunter Syndrome. Dr. Whiteman

obtained his undergraduate degree (in Experimental Psychology and Physiology) and his medical degree, from Oxford University in the United Kingdom. Following general postgraduate medical training in Britain, he undertook a residency in Pediatrics in the USA (University of Connecticut Hospitals) and then a clinical and research fellowship in Medical Genetics and Metabolic Diseases at the University of Pennsylvania/Children's Hospital of Philadelphia in the early 1980s. Subsequently, he held a variety of positions in clinical and laboratory genetics and metabolism in medical schools and university hospitals throughout the United States. Dr. Whiteman is a Fellow of the American Academy of Pediatrics and a Fellow of the American College of Medical Genetics. He has served on numerous state and U.S. national committees related to metabolic disease management and newborn screening. Currently he is Senior Medical Director, Clinical Sciences in the Research and Development department at Shire Human Genetic Therapies in Lexington, Massachusetts, USA.

Dr. Durhane Wong-Rieger is President and CEO of the *Institute for Optimizing Health Outcomes*. She is also President of the *Canadian Organization for Rare Disorders (CORD)* and head of the *Consumer Advocare Network*, a national network to promote patient engagement in healthcare policy and advocacy. Internationally, Durhane serves as Chair of the Board of the *International Alliance of Patient Organizations*, Co-Chair of the *Health Technology Assessment International Patient/Citizen Involvement Interest Group* and on the Board of Directors of *DIA International*. She is a certified Health Coach and licensed T-Trainer with the Stanford-based *Living A Healthy Life with Chronic Conditions*. Dr. Wong-Rieger has conducted training, workshops, and evaluation for patient groups in Canada and internationally on all aspects of patient engagement and advocacy. She has served on numerous health policy advisory committees and panels, including the *Policy Dialogues for the Commission on the Future of Healthcare in Canada*, *Ontario Premier's Advisory Board on Organ Donation*, Health Canada's *Expert Advisory Committee on Vigilance of Health Products* and *Expert Advisory Panel on Special Access Programme*, and *Association of Family Health Teams of Ontario*. From 1984 to 1999, Durhane was professor of Psychology at the University of Windsor in Ontario, Canada. Durhane has a BA in Psychology from Barnard College in New York City and an MA and Ph.D. in Social Psychology from McGill University in Montreal. She is author of two books and many articles and is a frequent lecturer and workshop leader.

Roy and Zezee Zeighami are parent advocates for children suffering from rare disease. Their personal connection to the fight is their son Reed who suffers from MPS IIIA, *Sanfilippo Syndrome*. They have been active in the fight to speed treatment and improve upon the incentives provided by the Orphan Drug Act to treat rare diseases such as *Sanfilippo*. They live in Dallas, TX, USA with Reed and his older sister Aziza.

Contents

Part II Health 2.0

Part III Patient Perspectives and Empowerment Issues