

Contents

Part I Rare and Orphan Diseases

Why R&D into Rare Diseases Matter	3
Remco L. A. de Vrueth	
Vignette: Autoimmune Polyendocrine Syndrome Type I (APS 1)	21
Patrice F. Band	
Rare Diseases: How Genomics has Transformed Thinking, Diagnoses and Hope for Affected Families	27
Pierre Meulien, Paul Lasko, Alex MacKenzie, Cindy Bell and Kym Boycott	
Vignette: A Giant of a Man: Simon Ibell (MPS II)	39
Marie Ibell	
Innovative Funding Models for Rare Diseases	43
Amanda Lordemann, Krissi Danielsson and Jimmy Cheng-Ho Lin	
Rare Diseases: The Medical and the Disability Perspectives in the Age of 2.0	51
Sara Newman	
Vignette: Taking Control of Thalassemia	67
Angela Covato	
Industry Perspectives on Orphan Drug Development	71
Sylvie Grégoire, Norman Barton and David Whiteman	

Part II Health 2.0

Health 2.0: The Power of the Internet to Raise Awareness of Rare Diseases. 83
 Laura Montini

Vignette: Living with NOMID: Michael’s Story 97
 Jocelyn Gardner

Health 2.0 and Information Literacy for Rare and Orphan Diseases 101
 Hannah Spring

Social Media and Engaging with Health Providers. 115
 Hugh Stephens

Vignette: Hope–Overcoming Fabry 123
 Adrian (Ed) Koning

Empowering the Rare Disease Community: Thirty Years of Progress . . . 127
 Jason R. Barron

Part III Patient Perspectives and Empowerment Issues

The Role of Social Media in Healthcare: Experiences of a Crohn’s Disease Patient. 139
 Michael Seres

Vignette: The Blessings and Curse of Diagnosis: Myasthenia Gravis 145
 Grainne Pierse

Noah’s Hope: Family Experiences of Batten Disease 149
 Tracy VanHoutan

Using Technology to Share Information: Experiences of Oesophagus Atresia (OA) and Tracheoesophageal Fistel (TOF) 163
 Caren Kunst

Vignette: MPSIIIA (Sanfilippo) 173
 Roy and Zezee Zeighami

The Empowered Patient in the Health System of the Future 177
 Frank Grossmann, Daniela M. Meier and Therese Stutz Steiger

Vignette: The Journey of a Lifetime 191
 Deb Purcell

Personalized Medicine: A Cautionary Tale or Instructional Epic 195
 Dorothy Weinstein

Part IV Closing Gaps: Promising Research and Future Considerations

Managing Communication for People with Amyotrophic Lateral Sclerosis: The Role of the Brain-Computer Interface 215
 Gaye Lightbody, Brendan Allison and Paul McCullagh

Vignette: The Wilderness 237
 Jeneva Stone

Opportunities and Challenges for Supporting People with Vascular Dementia Through the Use of Common Web 2.0 Services 241
 Kyle Boyd, Chris Nugent, Mark Donnelly, Raymond Bond and Roy Sterritt

Vignette: Recessive Dystrophic Epidermolysis Bullosa (RDEB): Sibling Experiences 263
 Jason Barron

Health Policies for Orphan Diseases: International Comparison of Regulatory, Reimbursement and Health Services Policies 267
 Durhane Wong-Rieger and Francis Rieger

Vignette: Route 125 (October 2009) 279
 Jeneva Stone

Rare Diseases Challenges and Opportunities 283
 Rashmi Gopal-Srivastava and Stephen C. Groft

Epilogue 291



<http://www.springer.com/978-3-642-38642-8>

Rare Diseases in the Age of Health 2.0

Bali, R.K.; Bos, L.; Gibbons, M.C.; Ibell, S.R. (Eds.)

2014, XXXIII, 292 p. 14 illus., Hardcover

ISBN: 978-3-642-38642-8