Contents

Part I   Rare and Orphan Diseases

Why R&D into Rare Diseases Matter .............................. 3
Remco L. A. de Vrueh

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
Laura Montini  
83

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

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

### Social Media and Engaging with Health Providers
Hugh Stephens  
115

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

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

## Part III  Patient Perspectives and Empowerment Issues

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

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

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

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

**Vignette: MPSIIIA (Sanfilippo)**  
Roy and Zezee Zeighami  
173

### The Empowered Patient in the Health System of the Future
Frank Grossmann, Daniela M. Meier and Therese Stutz Steiger  
177
<table>
<thead>
<tr>
<th>Title</th>
<th>Page</th>
</tr>
</thead>
<tbody>
<tr>
<td>Vignette: The Journey of a Lifetime</td>
<td>191</td>
</tr>
<tr>
<td>Deb Purcell</td>
<td></td>
</tr>
<tr>
<td>Personalized Medicine: A Cautionary Tale or Instructional Epic</td>
<td>195</td>
</tr>
<tr>
<td>Dorothy Weinstein</td>
<td></td>
</tr>
</tbody>
</table>

**Part IV  Closing Gaps: Promising Research and Future Considerations**

<table>
<thead>
<tr>
<th>Title</th>
<th>Page</th>
</tr>
</thead>
<tbody>
<tr>
<td>Managing Communication for People with Amyotrophic Lateral Sclerosis: The Role of the Brain-Computer Interface</td>
<td>215</td>
</tr>
<tr>
<td>Gaye Lightbody, Brendan Allison and Paul McCullagh</td>
<td></td>
</tr>
<tr>
<td>Vignette: The Wilderness</td>
<td>237</td>
</tr>
<tr>
<td>Jeneva Stone</td>
<td></td>
</tr>
<tr>
<td>Opportunities and Challenges for Supporting People with Vascular Dementia Through the Use of Common Web 2.0 Services</td>
<td>241</td>
</tr>
<tr>
<td>Kyle Boyd, Chris Nugent, Mark Donnelly, Raymond Bond and Roy Sterritt</td>
<td></td>
</tr>
<tr>
<td>Vignette: Recessive Dystrophic Epidermolysis Bullosa (RDEB):</td>
<td>263</td>
</tr>
<tr>
<td>Sibling Experiences</td>
<td></td>
</tr>
<tr>
<td>Jason Barron</td>
<td></td>
</tr>
<tr>
<td>Health Policies for Orphan Diseases: International Comparison of Regulatory, Reimbursement and Health Services Policies</td>
<td>267</td>
</tr>
<tr>
<td>Durhane Wong-Rieger and Francis Rieger</td>
<td></td>
</tr>
<tr>
<td>Vignette: Route 125 (October 2009)</td>
<td>279</td>
</tr>
<tr>
<td>Jeneva Stone</td>
<td></td>
</tr>
<tr>
<td>Rare Diseases Challenges and Opportunities</td>
<td>283</td>
</tr>
<tr>
<td>Rashmi Gopal-Srivastava and Stephen C. Groft</td>
<td></td>
</tr>
<tr>
<td>Epilogue</td>
<td>291</td>
</tr>
</tbody>
</table>
Rare Diseases in the Age of Health 2.0
2014, XXXIII, 292 p. 14 illus., Hardcover
ISBN: 978-3-642-38642-8