

Contents

National Plans on Rare Diseases	1
Domenica Taruscio, Amalia Egle Gentile, Marta De Santis, Rita Ferrelli, Rosa Giuseppa Frazzica, Georgi Iskrov, and Rumen Stefanov	
Biobanking for Rare Diseases – Impact on Personalised Medicine	23
Jeanne-Hélène di Donato	
Emerging Technologies for Gene Identification in Rare Diseases	33
Filippo Beleggia and Bernd Wollnik	
Personalized Medicine for Hereditary Deafness	47
Jessica Ordóñez, Oscar Diaz-Horta, and Mustafa Tekin	
Mitochondrial Diseases	61
Maria Judit Molnar and Klara Pentelenyi	
Complexity of Genotype-Phenotype Correlations in Mendelian Disorders: Lessons from Gaucher Disease	69
Nima Moaven, Nahid Tayebi, Ehud Goldin, and Ellen Sidransky	
Enzyme Replacement Therapy in Lysosomal Storage Diseases	91
Vassili Valayannopoulos	
Rare Cancers	109
Nikolajs Zeps and Chris Hemmings	
Adeno-Associated Virus Gene Therapy and Its Application to the Prevention and Personalised Treatment of Rare Diseases	131
Konstantina Grosios, Harald Petry, and Jacek Lubelski	

Induced Pluripotency for the Study of Disease Mechanisms and Cell Therapy	159
Toivo Maimets	
Author Index	175
Subject Index	205