

Contents

Overview of the Clinical Utility of Next Generation Sequencing in Molecular Diagnoses of Human Genetic Disorders.	1
Lee-Jun C. Wong	
Detection of Copy Number Variations (CNVs) Based on the Coverage Depth from the Next Generation Sequencing Data.	13
Yanming Feng, David Chen, and Lee-Jun C. Wong	
Next Generation Sequencing (NGS) Based Panel Analysis of Metabolic Pathways	23
Elizabeth B. Gorman and Lee-Jun C. Wong	
The Next Generation Sequencing Based Molecular Diagnosis of Visual Diseases	51
Xia Wang and Richard Alan Lewis	
Application of Next-Generation Sequencing to Hearing Loss	71
Lisa Dyer, Xinjian Wang, Kejian Zhang, John Greinwald, and C. Alexander Valencia	
Next-Generation Sequencing Based Clinical Molecular Diagnosis of Primary Immunodeficiency Diseases	89
Lijun Wang, C. Alexander Valencia, Rebecca A. Marsh, and Kejian Zhang	
Next-Generation Sequencing Based Testing for Disorders of the Skeleton	113
Allison Tam, Victor Wei Zhang, Lee Jun Wong, and Sandesh C.S. Nagamani	
Diagnosing Hereditary Cancer Susceptibility Through Multigene Panel Testing	123
Holly LaDuka, Shuwei Li, A.J. Stuenkel, Virginia Speare, Jill S. Dolinsky, and Elizabeth C. Chao	

Application of Next-Generation Sequencing in Noonan Spectrum Disorders	155
Hui Mei, Hongzheng Dai, Jinglan Zhang, Lee-Jun Wong, and Pilar L. Magoulas	
The Applications and Challenges of Next-Generation Sequencing in Diagnosing Neuromuscular Disorders	177
Lijun Wang, Arunkanth Ankala, Hamoud Al Khallaf, Xinjian Wang, Mikhail Martchenko, Biao Dong, Ammar Husami, Kejian Zhang, and C. Alexander Valencia	
Next-Generation Sequencing for the Diagnosis of Monogenic Disorders of Insulin Secretion	201
Amy E. Knight Johnson, Siri Atma W. Greeley, and Daniela del Gaudio	
Application of NGS in the Diagnosis of Cardiovascular Genetic Diseases	243
Guoliang Wang, Vivan Niewiadonski, Jianping Li, Ruirui Ji, Wenxin Zou, Daniel J. Penny, and Yuxin Fan	
Comprehensive Analyses of the Mitochondrial Genome	287
Victor Wei Zhang and Lee-Jun C. Wong	
Exome Sequencing in the Clinical Setting	305
Theodore Chiang, Magalie Leduc, Mari Tokita, Teresa Santiago-Sim, and Yaping Yang	
Family-Based Next-Generation Sequencing Analysis	321
Xia Wang, Linyan Meng, and Magalie S. Leduc	
Next Generation of Carrier Screening	339
Anastasia Fedick and Jinglan Zhang	
Erratum to:	E1
Index	355