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Altri autori (Persone)	WongLee-Jun C
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Nota di bibliografia	Includes bibliographical references and index.
Nota di contenuto	Part I: Overview.- History of DNA Sequencing Technologies -- Clinical Molecular Diagnostic Techniques: A Brief Review -- Part II: The Technologies and Bioinformatics -- Methods of Gene Enrichment and Massively Parallel Sequencing Technologies -- Sequence Alignment, Analysis, and Bioinformatics Pipelines -- Protein Structural Based Analysis for Variant Interpretation of Missense Variants at the Genomics Era: Using MNGIE Disease as an Example -- Algorithms and Guidelines for Interpretation of DNA Variants -- Part III: Application to Clinical Diagnostics -- NGS-based Clinical Diagnosis of Genetically Heterogeneous Disorders -- Molecular Diagnosis of Congenital Disorders of Glycosylation (CDG) -- NGS Improves the Diagnosis of X-Linked Intellectual Disability (XLID) -- NGS Analysis of Heterogeneous Retinitis Pigmentosa -- Next Generation Sequencing of the Whole Mitochondrial Genome -- Application of Next-Generation Sequencing of Nuclear Genes for Mitochondrial Disorders -- Noninvasive Prenatal Diagnosis Using Next Generation Sequencing -- Part IV: Compliance with CAP/CLIA Regulations -- Guidelines and Approaches to Compliance with Regulatory and Clinical Standards: Quality Control Procedures and Quality Assurance -- Validation of NGS-based Test and Implementation of Quality Control Procedures -- Frequently Asked Questions about the Clinical Utility of Next Generation Sequencing in Molecular Diagnosis of Human Genetic Diseases -- Index.
Sommario/riassunto	In recent years, owing to the fast development of a variety of

sequencing technologies in the post human genome project era, sequencing analysis of a group of target genes, entire protein coding regions of the human genome, and the whole human genome has become a reality. Next Generation Sequencing (NGS) or Massively Parallel Sequencing (MPS) technologies offers a way to screen for mutations in many different genes in a cost and time efficient manner by deep coverage of the target sequences. This novel technology has now been applied to clinical diagnosis of Mendelian disorders of well characterized or undefined diseases, discovery of new disease genes, noninvasive prenatal diagnosis using maternal blood, and population based carrier testing of severe autosomal recessive disorders. This book covers topics of these applications, including potential limitations and expanded application in the future. .