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Nota di bibliografia	Includes bibliographical references at the end of each chapters and index.
Nota di contenuto	Part I: overview -- 1. NGS, The new gold standard of identification of defective genes -- 2. Principles of target gene enrichments, pros and cons -- 3. Criteria for clinical application: Full validation and performance characteristics -- 4. Clinical requirements: variant interpretation, confirmation, and turnaround time -- Part II: Experiences in various applications -- 5. The metabolic pathways: GSD, CDG, cobalamin metabolism, and others -- 6. The eye gene panels -- 7. The otogenes -- 8. The immunodeficiency disorders -- 9. The bone density and skeletal related disorders -- 10.The hereditary cancer genes -- 11.The molecular diagnosis of cancers and implications in treatment: Marilyn Li -- 12. Neuromuscular disorders -- 13. The cardiac panel: YuXin Fan, GeneDx or Harvard Partner -- 14. The mitochondrial genome -- 15. The Nuclear Mitomes.
Sommario/riassunto	Next Generation Sequencing technology has been applied to clinical diagnoses in the past three to five years using various approaches, including target gene panels and whole exomes. The purpose of this

book is to summarize the experiences, the results, advantages and disadvantages, along with future development in the area of NGS-based molecular diagnosis. This up-to-date volume will not only provide the readers working with Next Generation Sequencing the basics on how to apply the technology to molecular diagnosis, but will present the results and experience of practical application.