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Autore	Valencia C. Alexander
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Nota di contenuto	Sanger sequencing principles, history and landmarks -- A survey of next-generation sequencing technologies -- A review of DNA enrichment technologies -- Application of next-generation sequencing to the diagnosis of genetic disorders a brief overview -- Next-generation sequencing-based noninvasive prenatal diagnosis -- Diagnosis of inherited neuromuscular disorders by next-generation sequencing -- Application of next-generation sequencing in hearing loss diagnosis -- Exome sequencing as a discovery and a diagnostic tool -- Challenges of next-generation sequencing-based molecular diagnostics.
Sommario/riassunto	This book introduces readers to Next Generation Sequencing applications in medical genetics. The authors discuss the direct application of next-generation sequencing to medicine, specifically, laboratory medicine or molecular diagnostics. The first part of the book contains chapters on sanger sequencing, NGS technologies, targeted-amplification and capture, and exome sequencing. The second part of the book focuses on genetic disorders diagnoses by NGS, prenatal diagnosis, muscular dystrophies, mitochondrial disorders diagnosis, and challenges in molecular diagnosis. Recent developments and potential future trends in NGS sequencing applications are highlighted, as well.