

Next Generation Sequencing Technologies in Medical Genetics (SpringerBriefs in Genetics)

C. Alexander Valencia, M. Ali Pervaiz, Ammar Husami, Yaping Qian, Kejian Zhang



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Alexander Valencia, M. Ali Pervaiz, Ammar Husami, Yaping Qian, Kejian Zhang 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.?

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