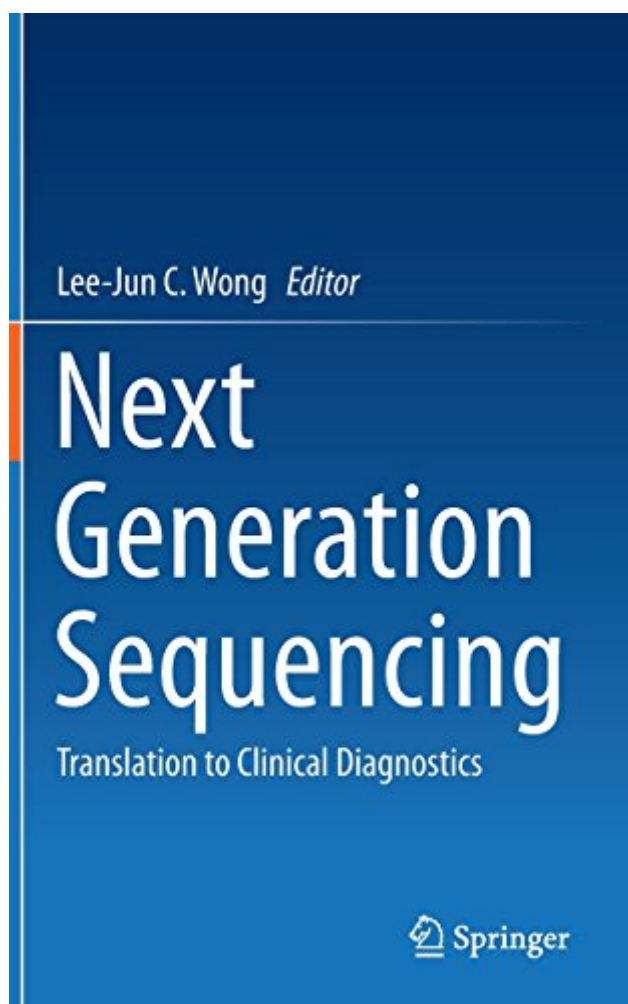


Next Generation Sequencing: Translation to Clinical Diagnostics PDF



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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. ?

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