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Clinical Applications of Next Generation Sequencing

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The appearance of next-generation sequencing (NGS) has significantly changed the way we think about scientific approaches in basic research and clinical application. The RNA sequencing method can provide information for alternative splicing and sequence variation, and quantify novel transcripts even without prior knowledge of them. The DNA sequencing method helps us to perform large-scale comparative and evolutionary studies in whole genome which were unimaginable just a few years ago. And more importantly, NGS not only increased the experiments throughput and extended the results, but also highly decreased the sequencing costs. These features make NGS an effective and promising tool in clinical applications. And in fact NGS is now widely applied to the clinic in many areas such asmolecular diagnostics, pathogen detection, and genetic mutations.

Based on the high sensitivity and high throughput, NGS provides a numbers of novel diagnostic methods in molecular diagnostics. For example, the development of NGS technologies made it possible to sequence a large number of entire cancer genomes and allowed researchers to systematically characterize human cancer genomes at the genomic, transcriptomic and epigenetic levels and discover novel cancer related genes. Furthermore, Individualized therapy can be determined based on the specific somatic mutations of a patient's cancer genome. And the clinicians can design target-specific or patient-specific probes to choose the best personalized treatment and monitor the progress of their patient's treatment.

Pathogen detection is one of the most successful applications for NGS. Most traditional pathogen detection methods (PCR or microarray) rely on prior knowledge of the exact sequence of the potential pathogen, or the ability to cultivate the pathogen. NGS provided an alternativemethod and overcome these limitations. It was used in de novo sequencing of bacterial and viral genomes, discovery and characterization of new viruses, detection of unexpected pathogens in clinical specimen, ultrasensitive monitoring of antiviral drug resistance, and investigation of viral diversity, evolution and spread, and evaluation of the human virome.

In genetic mutations detections, currently most of the genetic mutations were identified by Sanger sequencing, PCR, or microarrays in clinical application, but these methods were limited in single nucleotide variants.And the clinicians usually use phenotype-genotype correlations to perform single gene detection to screen genetic disease. But the genetic diseases may be very complicate and a single phenotype may be caused by various genes. NGS has its particular superiorities in this field. It can not only identify the single nucleotide variants, but also large insertion/deletions and structural recombination, which are common and very important in cancer.At the same time, whole genome sequencing (WGS) or whole exome sequencing (WES) provided a comprehensive view of all mutations in tissues at base-pair resolution. A significant number of new tumor markers and somatic variants have been discovered using NGS systems.

There is an increasingly rapid application of NGS in the clinical field. The fast, affordable, and accurate sequencing data serves as a good diagnostic and prognostic tool which helps clinicians identify specific characteristics in each patient, paving the road towards personalized medicine. But just like other new technologies, there are still many technical, analytical and ethics issues that need further refinement. The NGS technologies provided massive sequencing results and the researcher can identify disease related variants from them. But the abilities of accurate calling the functional variants and comprehensive understanding of disease producing genetic variants are still limited. The further clinical application of NGS requires clinical bioinformatics scientists and clinicians specially trained for complicated sequencing data analysis.And the most important task of them is selecting the meaningful and valuable candidates among the mass of the sequencing results for clinic application.

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