Cancer Diagnostics in the Era of Precision Medicine

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Advanced diagnostic pathology techniques have become increasingly necessary in the treatment of both cancer and non-neoplastic diseases, as targeted therapy has gained importance in recent years. This emphasizes the importance of developing and utilizing precise diagnostic techniques to identify potential targets for targeted therapy. As a result, cutting-edge diagnostic pathology techniques are essential[1]. Significant changes in cancer patient care have occurred over the last decade. Understanding the omics data of disease lesions, particularly cancer, is essential to the development of targeted therapy and precision medicine[2]. Molecular characterization techniques for diseases provide valuable information about disease diagnostic and therapeutic targets. With the introduction of pharmacogenomics, the conventional approach of using a "one size fits all" strategy for disease management has become outdated. Oncologists now prefer targeted therapy and personalized medicine. Precision medicine in cancer therapy entails developing therapies for patients based on the molecular genetic patterns of their specific tumors[3,4].

Recently, cancer researchers have been focusing on non-invasive approaches for detecting tumor molecular profiles. Liquid biopsy is one such diagnostic technique, which allows for the separation of circulating tumor cells and cancer cell-derived circulating components, including ctDNA and exosomes, from peripheral blood for genomic and proteomic analysis[5,6]. Liquid biopsy is a non-invasive, real-time method for detecting biomarkers in body fluids that has potential clinical applications for cancer screening, diagnosis, and prognosis. Among its potential clinical applications are cancer genomic profiling, identifying molecular targets for therapy, monitoring response to therapy, and determining minimal residual disease. Although the role of liquid biopsy in disease management is still being developed, it has the potential to revolutionize cancer diagnosis and treatment.

Advances in sequencing techniques have provided deep insights into the molecular profiles of tumors with the development of targeted therapies. NGS and array technologies can identify genetic variations like copy number variations, chromosomal rearrangements, and single nucleotide polymorphisms, insertions, and deletions. Computational biology and bioinformatics made it possible to analyze large amounts of data generated by OMICS, for cancer diagnostics and prediction of biological behavior and progression with greater accuracy.

RNA sequencing (RNA seq) has transformed differential expression analysis in specific cell types along with single-cell RNA sequencing methods. There is still a knowledge gap despite the availability of advanced technology related to the mutational profiles of cancer patients in our genetically distinct population, which is essential for the development of targeted therapies. Skilled personnel in the molecular genetics subject area are required in this context. Therefore, the best use of rapidly emerging diagnostic technology is based on the identification of biological hotspots and networks in Oncology. The ultimate goal is to use molecular and genetic information to tailor treatment decisions for individual patients, improving patient outcomes and reducing unnecessary treatment-related side effects.

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