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Next-Generation Sequencing-Based Profiling of Genetic Alterations in Solid Tumors: Implications for Targeted Therapy

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Introduction

The advent of Next-Generation Sequencing (NGS) has revolutionized the field of oncology by enabling high-throughput, comprehensive profiling of genetic alterations in solid tumors. Traditional diagnostic techniques, such as immunohistochemistry and Sanger sequencing, often lack the sensitivity and breadth needed to detect complex genomic landscapes. In contrast, NGS provides a powerful platform for simultaneously analyzing mutations, copy number variations, gene fusions, and other molecular aberrations at single-base resolution. This technology has accelerated the discovery of driver mutations, such as those in EGFR, BRAF, KRAS, and PIK3CA, which play critical roles in tumorigenesis and response to therapy. Through precise molecular characterization, NGS supports personalized treatment strategies, helping oncologists tailor therapies based on the unique genomic signature of each patient's tumor, thereby improving clinical outcomes and reducing unnecessary toxicity [1].

Description

NGS-based profiling has become a cornerstone in the precision oncology paradigm, transforming both research and clinical practice. Panels such as Foundation One CDx, MSK-IMPACT, and Oncomine Comprehensive Assay allow for the simultaneous interrogation of hundreds of cancer-associated genes from a single biopsy sample. These platforms have revealed that solid tumors whether in the lung, breast, colon, or prostate harbor distinct molecular alterations that can guide targeted therapies. For example, the identification of EGFR mutations in Non-Small Cell Lung Cancer (NSCLC) has led to the successful use of Tyrosine Kinase Inhibitors (TKIs), while BRAF V600E mutations in melanoma are effectively targeted by BRAF inhibitors like vemurafenib [2].

Moreover, BRCA1/2 mutations in ovarian and breast cancers have paved the way for the clinical use of PARP inhibitors, marking a significant milestone in genotype-driven therapy. The integration of NGS data with bioinformatics pipelines enables comprehensive tumor profiling, including the detection of Tumor Mutational Burden (TMB) and Microsatellite Instability (MSI), both of which are predictive biomarkers for immunotherapy response. The continuous evolution of NGS technologies from Whole-Exome Sequencing (WES) to RNA-seq and single-cell sequencing offers deeper insights into tumor heterogeneity and resistance mechanisms [3].

However, challenges remain in translating vast genomic data into actionable clinical decisions. Issues such as data interpretation, assay standardization, and reimbursement barriers must be addressed for widespread clinical adoption. Nevertheless, as computational algorithms and databases like COSMIC, ClinVar, and OncoKB expand, the clinical relevance of NGS-guided decision-making continues to grow [4,5].

Conclusion

Next-Generation Sequencing-based profiling has transformed cancer diagnostics and therapeutics by revealing the molecular underpinnings of solid tumors. Its ability to identify actionable mutations has propelled the advancement of precision oncology, facilitating personalized treatment strategies that improve survival and quality of life. Despite technical and regulatory challenges, ongoing innovations in sequencing technologies, data integration, and clinical validation are expected to further solidify NGS as a standard diagnostic tool in oncology. As targeted therapies and companion diagnostics continue to evolve, NGS will remain indispensable in shaping the future of individualized cancer care.

Acknowledgement

None

Conflicts of Interest

None

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