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Transforming routine solid tumor profiling with automated next-generation sequencing: experience from a reference general hospital

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Abstract

The growing use of targeted therapies highlights the need for integrated DNA- and RNA-based next-generation sequencing (NGS) to comprehensively profile tumors. A single-center study was conducted to screen tumor types for which a first-line, integrated DNA-RNA NGS strategy provides a valuable advantage for rapid therapeutic decisions. A total of 546 tumor samples, including early- and late-stage lung adenocarcinoma (LUAD), intrahepatic and extrahepatic cholangiocarcinoma (iCCA/eCCA), and glioblastoma were analyzed using an automated, in-house NGS platform. Sequencing success rates ranged from 98.7% to 100%, with a median turnaround time of eight days from sample collection or histological diagnosis to report delivery. The assay simultaneously detected point mutations, gene fusions, copy number variants, and other clinically relevant alterations, even from small tissue samples. In LUAD, EGFR exon 19 deletions and KRAS p.G12C were the most frequent actionable mutations in early- and late-stage disease, respectively. A significant association was observed between EGFR amplification and the presence of actionable EGFR mutations in late-stage tumors. Actionable co-mutations of EGFR or KRAS with TP53 occurred at distinct frequencies, suggesting relevant clinical implications for therapy selection. Mutation profiles in cholangiocarcinoma and glioblastoma were consistent with published data, reinforcing the robustness of the approach. The results demonstrate that integrated DNA-RNA high-throughput NGS enables timely, precise molecular profiling for personalized therapy in solid tumors.

Keywords: Automated next-generation sequencing (NGS); Precision oncology; Routine molecular profiling; Solid tumor.

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