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Mut-map, a high-throughput micro fluidics chip-based mutation detection panel

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Abstract

With the advent of personalized medicine, the need for molecular profiling of the tumor samples - such as detecting oncogenic mutations or profiling gene expression profiling - has taken center stage in the treatment of cancer. Commercially available tests are usually designed for detecting hotspot mutations detect specific mutations in a single targeted gene such as BRAF, KRAS or EGFR and require relatively large amount of DNA for testing. On the other hand, although Next-Generation Sequencing (NGS) can profile entire genomes, high costs, and complex data analysis requirements and, high turnaround times render the technology not being suited for routine analysis. We have developed a micro fluidic chip based high-throughput platform for detecting over 120 hotspot mutations across 11 genes using 2-100 ng of DNA extracted from formalin-fixed paraffin-embedded tissues. The genes targeted on this panel include *AKT1*, *BRAF*, *EGFR*, *FGFR3*, *FLT3*, *HRAS*, *KIT*, *KRAS*, *MET*, *NRAS*, and *PIK3CA*. Allele-specific PCR (AS-PCR) has been optimized to detect the hotspot mutations using the Fluidigm micro fluidic chip.

Automated data analysis has been integrated to the mutation detection workflow. This MUT-MAP assay is designed to analyze mutation status in 88-132 samples per day. Platform validation including assay cross-reactivity studies were performed using synthetic oligomers as well as cell line DNA with known mutations reported in the COSMIC database. The assay sensitivity for the above assays range from 1-4%. This platform is routinely used in our lab to detect mutations in clinical samples and provide a valuable platform for exploratory biomarker assessments during drug development.

Biography

Rajesh D. Patel, PhD is a Scientific Manager in the Oncology Biomarker Development group at Genentech, in South San Francisco, USA. He has been associated with a number of Pharmaceutical as well as diagnostic companies and has been a consultant in the field of Immuno/Molecular Diagnostics for the past 25 + years.

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