Prima Cancer Genetics

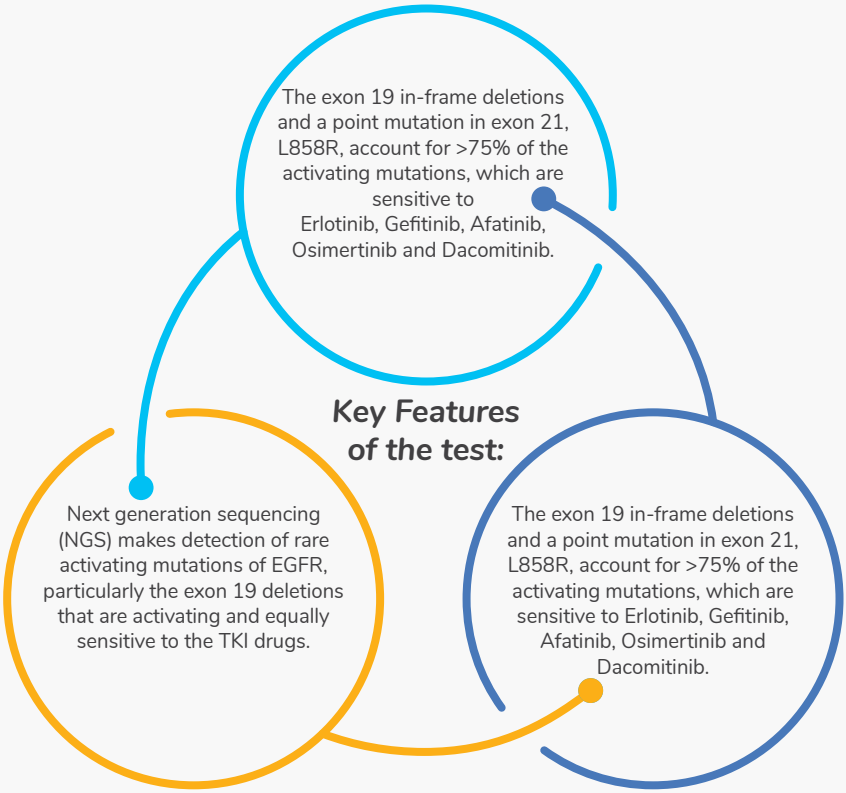
MEDGENOME

EGFR Gene Testing

Routinizing genetic testing in clinical practice

Back in 2003, based on retrospective analysis on efficacy of gefitinib/erlotinib, in a single arm study of non-small cell lung cancer (NSCLC) patients, it was discovered that patients who had activating mutations in the kinase domain of EGFR responded well to targeted therapy with tyrosine kinase inhibitors as compared to those patients who did not have EGFR mutations. Also, the study noted that these mutations were common in never-smokers with adenocarcinoma histology. The most common mutations were exon 19 deletions and exon 21 L858R.

MedGenome testing for EGFR gene involves the analysis of tumor specimen to detect mutations in EGFR gene region of tumor DNA. It has become a gold standard technique for stratifying NSCLC patient to tyrosine kinase inhibitor.



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Comparison of testing techniques:

Particulars	Sanger Sequencing	Real-Time PCR	NGS
Tumor content required in the tumor biopsy sample for accurate detection of mutations	>30%	5%	5%
Sensitivity (% of mutant gene required for reliable detection against the wild type background)	Low (10-20%)	High (1-2%)	High (1-2%)
Accuracy	Low	High	Very High
Mutation types tested	All mutations present in the exons	Only specific hotspot mutations (covers 95% of all mutations)	All mutations present in the entire gene

Therapeutic Implications:

- Activation of EGFR signaling by mutations in the receptor drives tumor growth by promoting cell proliferation, invasion, angiogenesis, metastasis and inhibition of cell death.
- EGFR Mutation status can predict response to Tyrosine Kinase Inhibitors (TKIs)
- Activating EGFR mutations are more sensitive to TK inhibitors Gefitinib, Erlotinib, Afatinib, Osimertinib and Dacomitinib than wild-type receptor
- TKIs are transforming the treatment of non-small cell lung cancer by increasing survival of patients who carry mutant EGFR gene, an example of widely accepted application of genetic testing and personalized medicine in NSCLC for the last decade.

Reference:

1. https://science.sciencemag.org/content/304/5676/1497?ijkey=81d79adf5d70bab1c500fab66a7fbb095cfbf794&keytype2=tf_ipsecsha
2. https://www.pnas.org/content/101/36/13306
3. https://www.ncbi.nlm.nih.gov/pubmed/15118073?dopt=Abstract

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