EGFR MUTATION DETECTION

DESCRIPTION

Overexpression and oncogenic mutations in the epidermal growth factor receptor (EGFR) have been identified as oncogenic drivers of various solid tumors, including non-small cell lung cancer (NSCLC). EGFR is a cell surface receptor and plays a central role in transmitting signals that promote cell growth and proliferation through activation of the Ras-Raf-MAPK pathway. Excessive activation of EGFR has been shown to be associated with advanced stages of cancer and poor patient prognosis. The EGFR Mutation Detection assay identifies the presence of 21 mutations found in exons 18, 19, 20, and 21 of EGFR:

GENE	MUTATION			AMINO ACID CHANGE	EXON
EGFR	2156G>C			G719A	18
EGFR	2238_2255del18 2235_2249del15 2236_2250del15 2239_2253del15 2239_2256del18	2240_2254del15 2240_2257del18 2239_2248TTAAGAGAAG>C 2239_2251>C 2237_2255>T	2239_2258>CA 2238_2252>GCA 2238_2248>GC 2235_2252>AAT	Deletions	19
EGFR	2303G>T			S768I	20
EGFR	2319_2320insCAC 2310_2311insGGT			Insertions	20
EGFR	2369C>T			T790M	20
EGFR	2573T>G			L858R	21
EGFR	2582T>A			L861Q	21

CLINICAL UTILITY

Point mutations, insertions and deletions in EGFR identified by the EGFR Mutation Detection assay function as oncogenic drivers in up to 20% of NSCLC. Approximately 85% of patients with EGFR mutations respond to antibody-based therapeutics or tyrosine kinase inhibitors such as cetuximab, panitumumab, gefitinib, and erlotinib.

TEST BENEFITS

New industry standard in accuracy and turnaround time:

- Sensitivity of 100% in formalin fixed paraffin embedded cell lines
- 24-48 hour total time to results

Comprehensive design:

- Can simultaneously detect 21 known variants in the tyrosine kinase region of the EGFR gene including the T790M “gatekeeper” mutation

SPECIFICATIONS

Test Code	719
Turnaround Time	24-48 hours
PCR/Probe Targets	Point mutations, insertions and deletions in exons 18-21 of the EGFR gene and endogenous control

EGFR MUTATION DETECTION

Method Quantitative Real-Time Polymerase Chain Reaction (qPCR)

Specimen Formalin-fixed, paraffin-embedded blocks/slides

- 2-3 10-micron sections preferred
- Storage: Ambient (4°C preferred)
- Sections must be freshly prepared or less than 3 months old.

Causes for Rejection DNA quality can be compromised due to nucleic acid fragmentation, and/or modification by chemical reactions between formaldehyde and nucleic acids which include crosslinking with proteins or other biomolecules. Specimens with control Ct values greater than 31.1 cycles will be deemed as Quantity/Quality Not Sufficient (QNS) and noted as such.

Limit of Detection 17.5% mutant content (validated in formalin-fixed, paraffin-embedded cell line studies)

Detection Range

REACTION	CT VALUE
Endogenous Control	23.7-31.1
EGFR Mutation Detection	ΔCt ≤ 7.4-8.9 (mutation dependent)

Samples with a 40 Ct value are approaching the lower limits of detection for the assay.

How to Order Download and complete the Insight Molecular Labs requisition form at www.insightmdx.com. Fax completed form to (615) 255-1330. For any questions, please call client services at (615) 255-8880.

CPT Code 81235

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