

Test Description

NRAS mutation test is an in vitro diagnostic test for the qualitative detection of mutations in codons 12, 13, 59, 61, 117 and 146 of NRAS oncogene.

Patient Demographic

Name: Naresh Malhotra
Sex: Male
Date of Birth/Age: 61 Years
Disease: Colorectal liver metastasis

Clinician

Clinician Name: Dr Bhuvan Chugh
Medical Facility: Medanta Hospital
Pathologist: Not Provided

Specimen

Site: Gall Bladder
Sample Type: FFPE block B 11630/19
Date of Collection: 05-03-2020
Date of Booking: 05-03-2020

NRAS Mutation Analysis

Result

Mutation Detected in NRAS codon 61

GENOMIC FINDINGS

Mutation: Q61H
Protein: p.Gln61His
Nucleotide Change: c.183A>C ; c.183A>T

INTERPRETATION

Mutation detected

Q61H is a hotspot mutation that lies within a GTP-binding region of the Nras protein (UniProt.org). *Q61H* confers a loss of function to Nras protein as indicated by increased GTP-bound Nras, which leads to increased downstream pathway activation and cell proliferation in cell culture.

METHODOLOGY

The NRAS Mutation Test, performed on the Biocartis Idylla™ system, is an *in vitro* diagnostic test for the qualitative detection of 18 mutations (G12C, G12S, G12D, G12A, G12V, G13D, G13V, G13R, A59T, Q61H/Q61H, Q61K/R/L, K117N/K117N and A146T/V) in codons 12, 13, 59, 61, 117 and 146 of the NRAS gene. Formalin-fixed paraffin-embedded (FFPE) human cancer tissue is lysed liberate DNA for subsequent real-time PCR amplification using allele specific primers. Two sample processing controls (SPC) are amplified simultaneously i.e. (1) a conserved region of the NRAS gene (referred to as NRAS-Total) and (2) a conserved region of the BRAF gene. The presence of a mutant genotype is determined by calculating the difference between the NRAS Sample Processing Control Cq and the Cq obtained for the NRAS mutant signal(s).

The analytic sensitivity of this assay has been determined at < or = 5%

REFERENCES

1. Allegra et al. J Clin Oncol (2016) 34:179-85
2. Boleij et al. BMC Cancer (2016) 16:825.



March 06, 2019

Dr Gulshan Yadav, MD, Consultant Pathology

Date