Taipei Veterans General Hospital

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Sample Information

Patient Name: 王文賢
Gender: Male
ID No.: F120553642
History No.: 39779550
Age: 53

Ordering Doctor: DOC1373L 陳三奇
Ordering REQ.: 0AWYGSE
Signing in Date: 2020/10/07

Path No.: S109-89712
MP No.: F20084
Assay: Oncomine Focus Assay
Sample Type: FFPE
Block No.: S109-28920A
Percentage of tumor cells: 90%
Note:

Sample Cancer Type: Liver Cancer

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Report Highlights

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Relevant Biomarkers

No clinically significant biomarkers found in this sample.

Prevalent cancer biomarkers without relevant evidence based on included data sources
CTNNB1 p.(D32G) c.95A>G

Variants (Exclude variant in Taiwan BioBank with >1% allele frequency)

DNA Sequence Variants

Gene	Amino Acid Change	Coding	Variant ID	Locus	Allele Frequency	Transcript	Variant Effect	Coverage
CTNNB1	p.(D32G)	c.95A>G	COSM5681	chr3:41266098	17.76%	NM_001904.3	missense	1999

Biomarker Descriptions

CTNNB1 (catenin beta 1)

Background: The CTNNB1 gene encodes catenin beta-1 (β-catenin), an integral component of cadherin-based adherens junctions involved in maintaining adhesion and regulating the growth of epithelial cell layers[1]. CTNNB1 binds to the APC protein in the cytoplasm and also interacts with TCF and LEF transcription factors in the nucleus to regulate WNT signaling[2]. Steady state levels of CTNNB1 are regulated by ubiquitin-dependent proteolysis[3,4,5].

Alterations and prevalence: Recurrent somatic mutations leading to CTNNB1 activation are common in cancer. The most prevalent alterations include missense mutations in exon 3 at codons S33, S37, T41, and S45 that block phosphorylation by GSK-β and inhibit CTNNB1 degradation[6,7,8,9]. These activating mutations are observed in diverse solid tumors and have a prevalence of 20-30% in hepatocellular carcinoma, 20% of uterine carcinoma, and 15% of adrenocortical carcinoma[10,11,12,13,14,15,16].

Potential relevance: Currently, no therapies have been approved for CTNNB1 aberrations. CTNNB1 alterations in EGFR positive lung cancer have been proposed to promote cancer progression and limit the response to EGFR tyrosine kinase inhibitors[17].

Signatures

Testing Personnel:

2020.10.08
16:08:38 +08'00'

Laboratory Supervisor:

2020.10.08
16:08:52 +08'00'

Pathologist:

2020.10.08
16:09:06 +08'00'

References

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