Taipei Veterans General Hospital

Department of Pathology and Laboratory Medicine
No.201, Sec. 2, Shipai Rd., Beitou District, Taipei City, Taiwan
11217, R.O.C.
Tel: 02-2875-7449

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

Report Highlights
0 Relevant Biomarkers
0 Therapies Available
0 Clinical Trials

Relevant Biomarkers

No relevant biomarkers found in this sample.

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

Variant Details

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
ALK	p.(D1529E)	c.4587C>G	.	chr2:29416366	100.00%	NM_004304.4	missense	2000
ALK	p.(I1461V)	c.4381A>G	.	chr2:29416572	99.95%	NM_004304.4	missense	1999
ALK	p.(=)	c.3375C>A	.	chr2:29445458	100.00%	NM_004304.4	synonymous	1993
FGFR3	p.(=)	c.1953G>A	.	chr4:1807894	100.00%	NM_000142.4	synonymous	1011

Variant Details (continued)

DNA Sequence Variants (continued)

Gene	Amino Acid Change	Coding	Variant ID	Locus	Allele Frequency	Transcript	Variant Effect	Coverage
PDGFRA	p.(=)	c.1701A>G	.	chr4:55141055	100.00%	NM_006206.5	synonymous	1998
FGFR4	p.(P136L)	c.407C>T	.	chr5:176517797	99.60%	NM_213647.2	missense	2000
RET	p.(=)	c.2307G>T	.	chr10:43613843	100.00%	NM_020975.4	synonymous	1993

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
15:55:29 +08'00'

Laboratory Supervisor:

2020.10.08
15:55:44 +08'00'

Pathologist:

2020.10.08
15:56:02 +08'00'

References

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