



Sample Information

Patient Name: 謝國棟
Gender: Male
ID No.: E101307370
History No.: 49519020
Age: 73

Ordering Doctor: DOC6496F 盧佩妤
Ordering REQ.: 0CVRGHC
Signing in Date: 2024/1/9

Path No.: M113-00006
MP No.: F24004
Assay: Oncomine Focus Assay
Sample Type: FFPE
Block No.: S112-23085A
Percentage of tumor cells: 20%

Reporting Doctor: DOC5466K 葉奕成 (Phone: 8#5466)

Note:

Sample Cancer Type: Non-Small Cell Lung Cancer

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Relevant Non-Small Cell Lung Cancer Variants

Gene	Finding	Gene	Finding
ALK	None detected	NTRK1	None detected
BRAF	None detected	NTRK2	None detected
EGFR	None detected	NTRK3	None detected
ERBB2	None detected	RET	None detected
KRAS	None detected	ROS1	None detected
MET	None detected		

Relevant Biomarkers

No relevant biomarkers found in this sample.

Prevalent cancer biomarkers without relevant evidence based on included data sources

CCND1 amplification

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
RET	p.(E632K)	c.1894G>A	.	chr10:43609942	15.81%	NM_020975.6	missense	1929

Copy Number Variations

Gene	Locus	Copy Number
CCND1	chr11:69456942	8.75

Biomarker Descriptions

CCND1 amplification

cyclin D1

Background: The CCND1 gene encodes the cyclin D1 protein, a member of the highly conserved D-cyclin family that also includes CCND2 and CCND3^{1,2,3}. D-type cyclins are known to regulate cell cycle progression by binding to and activating cyclin dependent kinases (CDKs), specifically CDK4 and CDK6, which leads to the phosphorylation and inactivation of the retinoblastoma (RB1) protein^{1,2}. Consequently, RB1 inactivation results in E2F transcription factor activation and cellular G1/S phase transition thereby resulting in cell cycle progression, a common event observed in tumorigenesis^{1,2,4}. Aberrations in the D-type cyclins have been observed to promote tumor progression suggesting an oncogenic role for CCND1^{3,5}.

Alterations and prevalence: Recurrent somatic alterations to CCND1, including mutations, amplifications, and chromosomal translocations, are observed in many cancer types. A common mechanism of these alterations is to increase the expression and nuclear localization of the cyclin D1 protein. Recurrent somatic mutations include missense mutations at codons T286 and P287 and c-terminal truncating mutations that are enriched in about 33% of uterine cancer, and missense mutations at Y44 that are enriched in about 50% of Mantle cell lymphoma (MCL)^{6,7,8,9}. These mutations block phosphorylation-dependent nuclear export and proteolysis^{10,11,12,13}. CCND1 is recurrently amplified in many cancer types, including up to 35% of esophageal cancer, 20-30% of head and neck cancer, and 10-20% of breast, squamous lung, and bladder cancers^{6,8,14}. MCL is genetically characterized by the t(11;14) (q13;q13) translocation, a rearrangement that juxtaposes CCND1 to the immunoglobulin heavy (IgH) chain gene. This rearrangement leads to constitutive expression of cyclin D1 and plays an important role in MCL pathogenesis^{15,16}.

Potential relevance: Currently, no therapies are approved for CCND1 aberrations. The t(11;14) translocation involving CCND1 can be used to help diagnose some lymphoma subtypes including non-gastric MALT lymphoma, splenic marginal cell lymphoma, and mantle cell lymphoma¹⁷.

Clinical Trials in Taiwan region:

References

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