Taipei Veterans General Hospital

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Sample Information

Patient Name: 胡慧貞
Gender: Female
ID No.: A221321454
History No.: 22731960
Age: 55

Ordering Doctor: DOC1686E 陳玟均
Ordering REQ.: 0CPMBSY
Signing in Date: 2023/08/09

Path No.: M112-00211
MP No.: MY23054
Assay: Oncomine Myeloid Assay
Sample Type: Bone Marrow
Bone Marrow Aspirating Date: 2023/08/08

Reporting Doctor: DOC5444B 楊靜芬 (Phone: 8#5444)

Note:

Sample Cancer Type: Myelodysplastic Syndrome

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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
RB1 p.(R661Q) c.1982G>A

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
RB1	p.(R661Q)	c.1982G>A	COSM4428412	chr13:49033845	49.75%	NM_000321.2	missense	2000
CALR	p.(?)	c.-68C>T	.	chr19:13049426	49.52%	NM_004343.4	unknown	1999

Biomarker Descriptions

RB1 (RB transcriptional corepressor 1)

Background: The RB1 gene encodes the retinoblastoma protein (pRB), and is an early molecular hallmark of cancer. RB1 belongs to the family of pocket proteins that also includes p107 and p130, which play a crucial role in the cell proliferation, apoptosis, and differentiation[1,2]. RB1 is well characterized as a tumor suppressor gene that restrains cell cycle progression from G1 phase to S phase[3]. Specifically, RB1 binds and represses the E2F family of transcription factors that regulate the expression of genes involved in the G1/S cell cycle regulation[1,2,4]. Germline mutations in RB1 are associated with retinoblastoma (a rare childhood tumor) as well as other cancer types such as osteosarcoma, soft tissue sarcoma, and melanoma[5].

Alterations and prevalence: Recurrent somatic alterations in RB1, including mutations and biallelic loss, lead to the inactivation of the RB1 protein. RB1 mutations are observed in urothelial carcinoma (approximately 16%), endometrial cancer (approximately 12%), and sarcomas (approximately 9%)[6]. Similarly, biallelic loss of RB1 is observed in sarcomas (approximately 13%), urothelial carcinoma (approximately 6%), and endometrial cancer (approximately 1%)[6]. Biallelic loss of the RB1 gene is also linked to the activation of chemotherapy-induced acute myeloid leukemia (AML) and acute lymphoblastic leukemia (ALL)[7,8,9].

Potential relevance: Currently, there are no therapies approved for RB1 aberrations.

References

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