Taipei Veterans General Hospital

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Sample Information

Patient Name: 王玉薇
Gender: Female
ID No.: Y200264631
History No.: 11080629
Age: 68

Ordering Doctor: DOC1483K 王浩元
Ordering REQ.: 0CNJQMC
Signing in Date: 2023/07/18

Path No.: M112-00185
MP No.: MY23045
Assay: Oncomine Myeloid Assay
Sample Type: Bone Marrow
Bone Marrow Aspirating Date: 2023/07/13

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

Note:

Sample Cancer Type: Myelodysplastic Syndrome

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Relevant Myelodysplastic Syndrome Variants

Gene	Finding	Gene	Finding
ASXL1	None detected	NPM1	None detected
BCOR	None detected	NRAS	None detected
CBL	None detected	NUP214	None detected
CREBBP	None detected	RUNX1	None detected
ETV6	None detected	SF3B1	None detected
EZH2	None detected	SRSF2	None detected
FLT3	None detected	STAG2	None detected
GATA2	None detected	TP53	None detected
IDH2	None detected	U2AF1	None detected
KIT	None detected	WT1	None detected
KMT2A	None detected	ZRSR2	None detected
MECOM	None detected		

Relevant Biomarkers

No clinically significant biomarkers found in this sample.

Prevalent cancer biomarkers without relevant evidence based on included data sources
*TET2 p.(E149Nfs*3) c.445delG*

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
TET2	p.(E149Nfs*3)	c.445delG	.	chr4:106155543	4.37%	NM_001127208.2	frameshift Deletion	1993

Biomarker Descriptions

TET2 (tet methylcytosine dioxygenase 2)

Background: TET2 encodes the tet methylcytosine dioxygenase 2 protein and belongs to a family of ten-eleven translocation (TET) proteins that also includes TET1 and TET3[1]. TET2 is involved in DNA methylation, specifically in the conversion of 5-methylcytosine to 5-hydroxymethylcytosine[2,3]. The TET proteins contain a C-terminal core catalytic domain that contains a cysteine-rich domain and a double stranded ß-helix domain (DSBH)[4]. TET2 is a tumor suppressor gene. Loss of function mutations in TET2 are associated with loss of catalytic activity and transformation to hematological malignancies[1,2,3]

Alterations and prevalence: Somatic TET2 mutations, including nonsense, frameshift, splice site, and missense, are observed in 20-25% of myelodysplastic syndrome (MDS) associated diseases, including 40%-60% chronic myelomonocytic leukemia (CMML)[5]. TET2 mutations at H1881 and R1896 are frequently observed in myeloid malignancies[2,6]. TET2 mutations are also observed in 9% of uterine, 8% of melanoma and acute myeloid leukemia (AML), as well as 6% of diffuse large B-cell lymphoma (DLBCL).

Potential relevance: The presence of TET2 mutations may be used as one of the major diagnostic criteria in pre-primary myelofibrosis (pre-PMF) and overt PMF in the absence of JAK2/CALR/MPL mutations[7]. TET2 mutations are associated with poor prognosis in PMF and increased rate of transformation to leukemia[7,8]

References

1. Pan et al. The TET2 interactors and their links to hematological malignancies. IUBMB Life. 2015 Jun;67(6):438-45. PMID: 26099018
2. Ko et al. Impaired hydroxylation of 5-methylcytosine in myeloid cancers with mutant TET2. Nature. 2010 Dec 9;468(7325):839-43. PMID: 21057493
3. Solary et al. The Ten-Eleven Translocation-2 (TET2) gene in hematopoiesis and hematopoietic diseases. Leukemia. 2014 Mar;28(3):485-96. PMID: 24220273
4. An et al. TET family dioxygenases and DNA demethylation in stem cells and cancers. Exp. Mol. Med. 2017 Apr 28;49(4):e323. PMID: 28450733
5. NCCN Guidelines® - NCCN-Myelodysplastic Syndromes [Version 1.2023]
6. Kosmider et al. TET2 mutation is an independent favorable prognostic factor in myelodysplastic syndromes (MDSs). Blood. 2009 Oct 8;114(15):3285-91. PMID: 19666869
7. NCCN Guidelines® - NCCN-Myeloproliferative Neoplasms [Version 3.2022]
8. Lundberg et al. Clonal evolution and clinical correlates of somatic mutations in myeloproliferative neoplasms. Blood. 2014 Apr 3;123(14):2220-8. PMID: 24478400