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: 郭照娟
Gender: Female
ID No.: P220945649
History No.: 48430871
Age: 48

Ordering Doctor: DOC8919E 周逸峰
Ordering REQ.: 0BXEGXR
Signing in Date: 2022/07/12

Path No.: S111-99746
MP No.: MY22018
Assay: Oncomine Myeloid Assay
Sample Type: Bone Marrow
Bone Marrow Aspirating Date: 2022/07/04

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

Note:

Sample Cancer Type: Acute Myeloid Leukemia

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Relevant Acute Myeloid Leukemia Variants

Gene	Finding	Gene	Finding
ABL1	None detected	MECOM	None detected
ASXL1	None detected	MLLT3	None detected
CEBPA	None detected	MYH11	None detected
CREBBP	None detected	NPM1	None detected
FLT3	None detected	NUP214	None detected
IDH1	None detected	RARA	None detected
IDH2	None detected	RUNX1	None detected
KMT2A	None detected	TP53	None detected

Relevant Biomarkers

No clinically significant biomarkers found in this sample.

Prevalent cancer biomarkers without relevant evidence based on included data sources
STAG2 c.2096+1G>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
STAG2	p.(?)	c.2096+1G>A	.	chrX:123199797	42.30%	NM_001042749.2	unknown	1986
JAK2	p.(W659R)	c.1975T>C	.	chr9:5077563	52.05%	NM_004972.4	missense	2000

Biomarker Descriptions

STAG2 (stromal antigen 2)

Background: The STAG2 gene encodes the stromal antigen 2 protein, one of the core proteins in the cohesin complex, which regulates the separation of sister chromatids during cell division[1,2]. Components of the cohesion complex include SMC1A, SMC3, and RAD21, which bind to STAG1/STAG2 paralogs[3,4]. Inactivating mutations in STAG2 contribute to X-linked neurodevelopmental disorders, aneuploidy, and chromosomal instability in cancer[3,5].

Alterations and prevalence: Somatic mutations in STAG2 include nonsense, frameshift, splice site variants[6]. Somatic mutations in STAG2 are observed in various solid tumors including 14% of bladder cancer, 10% of uterine cancer, 3% of stomach cancer, and 4% of lung adenocarcinoma[7]. In addition, mutations in STAG2 are observed in 5-10% of myelodysplastic syndrome(MDS), 3% of acute myeloid leukemia, and 2% of diffuse large B-cell lymphoma[6,7].

Potential relevance: Nonsense, frameshift, and splice site STAG2 mutations are associated with poor prognosis in MDS[6]. Truncating mutations in STAG2 lead to a loss of function in bladder cancer and are often identified as an early event associated with low grade and stage tumors[8].

Signatures

Testing Personnel:

2022.07.15
14:05:01
+08'00'

Laboratory Supervisor:

2022.07.15
14:05:15 +08'00'

Pathologist:

2022.07.15
14:05:35
+08'00'

References

1. Mehta et al. Cohesin: functions beyond sister chromatid cohesion. FEBS Lett. 2013 Aug 2;587(15):2299-312. PMID: 23831059
2. Aquila et al. The role of STAG2 in bladder cancer. Pharmacol. Res. 2018 May;131:143-149. PMID: 29501732
3. Mullegama et al. De novo loss-of-function variants in STAG2 are associated with developmental delay, microcephaly, and congenital anomalies. Am. J. Med. Genet. A. 2017 May;173(5):1319-1327. PMID: 28296084
4. van et al. Synthetic lethality between the cohesin subunits STAG1 and STAG2 in diverse cancer contexts. Elife. 2017 Jul 10;6. PMID: 28691904
5. Solomon et al. Mutational inactivation of STAG2 causes aneuploidy in human cancer. Science. 2011 Aug 19;333(6045):1039-43. PMID: 21852505
6. NCCN Guidelines® - NCCN-Myelodysplastic Syndromes [Version 3.2022]
7. Cerami et al. The cBio cancer genomics portal: an open platform for exploring multidimensional cancer genomics data. Cancer Discov. 2012 May;2(5):401-4. PMID: 22588877
8. Solomon et al. Frequent truncating mutations of STAG2 in bladder cancer. Nat. Genet. 2013 Dec;45(12):1428-30. PMID: 24121789