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: 楊桑滿
Gender: Female
ID No.: R222228590
History No.: 17606198
Age: 63

Ordering Doctor: DOC1901H 高志平
Ordering REQ.: 0BRWUBN
Signing in Date: 2022/02/11

Path No.: S111-98363
MP No.: MY22005
Assay: Oncomine Myeloid Assay
Sample Type: Bone Marrow
Bone Marrow Aspirating Date: 2022/02/07

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	***ASXL1 p.(R693*) c.2077C>T, ASXL1 p.(E635Rfs*15) c.1900_1922delAGAGAGGCGGCCACCACT GCCAT***	MLLT3	None detected
CEBPA	None detected	MYH11	None detected
CREBBP	None detected	NPM1	None detected
FLT3	None detected	NUP214	None detected
IDH1	***IDH1 p.(R132C) c.394C>T***	RARA	None detected
IDH2	None detected	RUNX1	None detected
KMT2A	None detected	TP53	None detected

Relevant Biomarkers

Genomic Alteration	Relevant Therapies (In this cancer type)	Relevant Therapies (In other cancer type)	Clinical Trials
IDH1 p.(R132C) c.394C>T isocitrate dehydrogenase (NADP(+)) 1 Allele Frequency: 30.83% **Prognostic significance:** None **Diagnostic significance:** None	**ivosidenib** [1] azacitidine decitabine venetoclax + chemotherapy	None	1
ASXL1 p.(R693) c.2077C>T, ASXL1 p.(E635Rfs*15) c.1900_1922delAGAGAGGCGGC CACCACTGCCAT* ASXL transcriptional regulator 1 Allele Frequency: 14.85%, 10.48% (2 variants) **Prognostic significance:** ELN 2017: Adverse **Diagnostic significance:** None	None	None	0

Public data sources included in relevant therapies: FDA[1], NCCN, **EMA**[2], ESMO
Public data sources included in prognostic and diagnostic significance: NCCN, ESMO

Prevalent cancer biomarkers without relevant evidence based on included data sources
PHF6 p.(Y103) c.308_309insA, PHF6 p.(R274*) c.820C>T*

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
IDH1	p.(R132C)	c.394C>T	COSM28747	chr2:209113113	30.83%	NM_005896.3	missense	1998
ASXL1	p.(E635Rfs*15)	c.1900_1922delAGAGAGGCGGCCACCACTGCCAT	.	chr20:31022403	10.48%	NM_015338.6	frameshift Deletion	1889
ASXL1	p.(R693*)	c.2077C>T	COSM51388	chr20:31022592	14.85%	NM_015338.6	nonsense	2000
PHF6	p.(Y103*)	c.308_309insA	.	chrX:133527597	16.94%	NM_032458.3	nonsense	1995
PHF6	p.(R274*)	c.820C>T	COSM144567	chrX:133549136	12.56%	NM_032458.3	nonsense	1999
BCOR	p.(A1527=)	c.4581C>T	.	chrX:39916422	49.90%	NM_001123385.2	synonymous	2000

Biomarker Descriptions

ASXL1 (ASXL transcriptional regulator 1)

Background: The ASXL1 gene encodes the ASXL transcriptional regulator 1 protein, a ligand-dependent co-activator and epigenetic scaffolding protein involved in transcriptional regulation[1,2]. ASXL1 belongs to the ASXL gene family, which also includes ASXL2 and ASXL3[2]. ASXL proteins contain a conserved c-terminal plant homeodomain (PHD) which facilitates interaction with DNA and histones[2,3]. ASXL1 influences chromatin remodeling and transcription through interaction with BAP1 and polycomb repressive complex (PRC) proteins, as well as other transcriptional activators and repressors[2,4]. In cancer, ASXL1 is the target of somatic mutations which often result in a truncated ASXL1 protein and loss of its PHD[5,6,7]. Such mutations can lead to impaired protein function and consequent upregulation of HOXA gene expression, supporting a tumor suppressor role for ASXL1[8].

Biomarker Descriptions (continued)

Alterations and prevalence: Missense, nonsense, and frameshift mutations in ASXL1 are reported in 3-6% of de novo acute myeloid leukemia (AML), up to 36% of secondary AML, approximately 15% of myelodysplastic syndromes (MDS), up to 23% of myeloproliferative neoplasms (MPN), up to 30% of systemic mastocytosis (SM), and approximately 45% of chronic myelomonocytic leukemia (CMML)[4,9,10,11,12,13,14,15,16]. The ASXL1 G646Wfs*12 mutation accounts for over 50% of ASXL1 mutated cases in myeloid malignancies[6,11,17]. This mutation results from a single nucleotide expansion that occurs within an eight base pair guanine repeat that extends from c.1927 to c.1934. It is proposed that the high prevalence of the G646Wfs*12 variant is due to replication slippage which can occur in areas of repetitive sequence[18]. As a consequence, detection of G646Wfs*12 may result as an artifact of PCR and/or sequencing[19]. However, multiple studies observe an increase in the frequency of G646Wfs*12 in myeloid cancer relative to normal suggesting that G646Wfs*12 is a bona fide somatic mutation[9,18,20].

Potential relevance: The majority of frameshift and nonsense mutations in ASXL1 that result in protein truncation and removal of the PHD domain are considered pathogenic[21]. Mutations in ASXL1 confer poor/adverse risk in AML and are associated with poor outcomes and adverse risk[16]. Additionally, ASXL1 nonsense or frameshift mutations are independently associated with poor prognosis in MDS and CMML[22]. Moreover, ASXL1 mutations are independently associated with inferior overall survival (OS) in patients with MPN or SM[23,24].

IDH1 (isocitrate dehydrogenase (NADP(+)) 1)

Background: The IDH1 and IDH2 genes encode homologous isocitrate dehydrogenase enzymes that catalyze the conversion of isocitrate to α-ketoglutarate (α-KG)[25]. The IDH1 gene encodes the NADP+ dependent cytoplasmic isocitrate dehydrogenase enzyme; IDH2 encodes the mitochondrial isoform.

Alterations and prevalence: Recurrent somatic mutations in IDH1 and IDH2 are mutually exclusive and observed in several malignancies including glioma, chondrosarcoma, intrahepatic cholangiocarcinoma, acute myeloid leukemia (AML), and myelodysplastic syndrome (MDS)[26]. Recurrent IDH1 variants include predominately R132H/C plus other substitutions at lower frequencies. These gain of function variants confer neomorphic enzyme activity[27]. Although wild-type enzymatic activity is ablated, recurrent IDH1 variants catalyze the conversion of α-KG to D-2-hydroxyglutarate, an oncometabolite with diverse effects on cellular metabolism, epigenetic regulation, redox states, and DNA repair[25,28]. Recurrent IDH1 mutations are present in 5-10% of patients with AML and 5% of patients with MDS[29,30,31]. Recurrent IDH1 mutations are present in nearly 80% of lower grade diffuse gliomas[12,32].

Potential relevance: Ivosidenib[33] is FDA approved (2018) for the treatment of AML patients with IDH1 R132C/G/H/L/S variants[34]. Ivosidenib has also been granted breakthrough designation (2020) for IDH1 mutated relapsed or refractory myelodysplastic syndrome (MDS)[35]. IDH1 mutations are associated with inferior leukemia-free survival in primary myelofibrosis (PMF) and inferior overall survival in polycythemia vera (PV) but have been shown to confer improved prognosis in lower grade gliomas[23,36,37].

PHF6 (PHD finger protein 6)

Background: The PHF6 gene encodes the plant homeodomain (PHD) finger protein 6 which contains four nuclear localization signals and two imperfect PHD zinc finger domains. PHF6 is a tumor suppressor that interacts with the nucleosome remodeling deacetylase (NuRD) complex, which regulates nucleosome positioning and transcription of genes involved in development and cell-cycle progression[38,39].

Alterations and prevalence: The majority of PHF6 aberrations are nonsense, frameshift (70%), or missense (30%) mutations, which result in complete loss of protein expression[38,40,41,42]. Truncating or missense mutations in PHF6 are observed in 38% of adult and 16% of pediatric T-cell acute lymphoblastic leukemia (T-ALL), 20-25% of mixed phenotype acute leukemias (MPAL), and 3% of AML, and 2.6% of hepatocellular carcinoma (HCC)[40,42]. Missense mutations recurrently involve codon C215 and the second zinc finger domain of PHF6[40]. PHF6 mutations are frequently observed in hematologic malignancies from male patients[38,40].

Potential relevance: Somatic mutations in PHF6 are associated with reduced overall survival in AML patients treated with high-dose induction chemotherapy[43].

Relevant Therapy Summary

● In this cancer type ○ In other cancer type ◑ In this cancer type and other cancer types ✖ No evidence

IDH1 p.(R132C) c.394C>T

Relevant Therapy	FDA	NCCN	EMA	ESMO	Clinical Trials*
ivosidenib	●	●	✖	●	✖
azacitidine	✖	●	✖	✖	✖
decitabine	✖	●	✖	✖	✖
venetoclax + azacitidine	✖	●	✖	✖	✖
venetoclax + cytarabine	✖	●	✖	✖	✖
venetoclax + decitabine	✖	●	✖	✖	✖
LY-3410738	✖	✖	✖	✖	● (I)

* Most advanced phase (IV, III, II/III, II, I/II, I) is shown and multiple clinical trials may be available.

Relevant Therapy Details

Current FDA Information

● In this cancer type ○ In other cancer type ◑ In this cancer type and other cancer types

FDA information is current as of 2021-11-17. For the most up-to-date information, search www.fda.gov.

IDH1 p.(R132C) c.394C>T

● ivosidenib

Cancer type: Acute Myeloid Leukemia **Label as of:** 2021-08-25 **Variant class:** IDH1 R132C mutation

Indications and usage:

TIBSOVO® is an isocitrate dehydrogenase-1 (IDH1) inhibitor indicated for the treatment of adult patients with a susceptible IDH1 mutation as detected by an FDA-approved test with:

Acute Myeloid Leukemia (AML)

- Newly-diagnosed AML who are ≥ 75 years old or who have comorbidities that preclude use of intensive induction chemotherapy.
- Relapsed or refractory AML.

Locally Advanced or Metastatic Cholangiocarcinoma

- Locally advanced or metastatic cholangiocarcinoma who have been previously treated.

Reference:

https://www.accessdata.fda.gov/drugsatfda_docs/label/2021/211192_s008lbl.pdf

Current NCCN Information

● In this cancer type ○ In other cancer type ◑ In this cancer type and other cancer types

NCCN information is current as of 2021-11-01. For the most up-to-date information, search www.nccn.org.
For NCCN International Adaptations & Translations, search www.nccn.org/global/international_adaptations.aspx.

IDH1 p.(R132C) c.394C>T

● venetoclax + azacitidine

Cancer type: Acute Myeloid Leukemia **Variant class:** IDH1 mutation

NCCN Recommendation category: 1

Population segment (Line of therapy):
- (Induction therapy); Preferred intervention

Reference: NCCN Guidelines® - NCCN-Acute Myeloid Leukemia [Version 3.2021]

● azacitidine

Cancer type: Acute Myeloid Leukemia **Variant class:** IDH1 mutation

NCCN Recommendation category: 2A

Population segment (Line of therapy):
- (Induction therapy); Other recommended intervention

Reference: NCCN Guidelines® - NCCN-Acute Myeloid Leukemia [Version 3.2021]

● decitabine

Cancer type: Acute Myeloid Leukemia **Variant class:** IDH1 mutation

NCCN Recommendation category: 2A

Population segment (Line of therapy):
- (Induction therapy); Other recommended intervention

Reference: NCCN Guidelines® - NCCN-Acute Myeloid Leukemia [Version 3.2021]

● ivosidenib

Cancer type: Acute Myeloid Leukemia **Variant class:** IDH1 mutation

NCCN Recommendation category: 2A

Population segment (Line of therapy):
- (Induction therapy); Preferred intervention
- Relapsed, Refractory (Line of therapy not specified)

Reference: NCCN Guidelines® - NCCN-Acute Myeloid Leukemia [Version 3.2021]

IDH1 p.(R132C) c.394C>T (continued)

venetoclax + cytarabine

Cancer type: Acute Myeloid Leukemia **Variant class:** IDH1 mutation

NCCN Recommendation category: 2A

Population segment (Line of therapy):
- (Induction therapy); Other recommended intervention

Reference: NCCN Guidelines® - NCCN-Acute Myeloid Leukemia [Version 3.2021]

venetoclax + decitabine

Cancer type: Acute Myeloid Leukemia **Variant class:** IDH1 mutation

NCCN Recommendation category: 2A

Population segment (Line of therapy):
- (Induction therapy); Preferred intervention

Reference: NCCN Guidelines® - NCCN-Acute Myeloid Leukemia [Version 3.2021]

Current ESMO Information

● In this cancer type ○ In other cancer type ◑ In this cancer type and other cancer types

ESMO information is current as of 2021-11-01. For the most up-to-date information, search www.esmo.org.

IDH1 p.(R132C) c.394C>T

● ivosidenib

Cancer type: Acute Myeloid Leukemia **Variant class:** IDH1 mutation

ESMO Level of Evidence/Grade of Recommendation: IV / B

Population segment (Line of therapy):
- Relapsed, Refractory (Second-line therapy)

Reference: ESMO Clinical Practice Guidelines - ESMO-Acute Myeloblastic Leukaemia in Adult Patients [Ann Oncol (2020); 31(6): 697-712.]

Prognostic Details

Current NCCN Information

NCCN information is current as of 2021-11-01. For the most up-to-date information, search www.nccn.org.
For NCCN International Adaptations & Translations, search www.nccn.org/global/international_adaptations.aspx.

ASXL1 p.(R693*) c.2077C>T, ASXL1 p.(E635Rfs*15) c.1900_1922delAGAGAGGCGGCCACCACTGCCAT

Prognostic significance: ELN 2017: Adverse

Cancer type: Acute Myeloid Leukemia **Variant class:** ASXL1 mutation

NCCN Recommendation category: 2A

Summary:
- Do not use as an adverse prognostic marker if it co-occurs with favorable-risk AML subtypes.

Reference: NCCN Guidelines® - NCCN-Acute Myeloid Leukemia [Version 3.2021]

Current ESMO Information

ESMO information is current as of 2021-11-01. For the most up-to-date information, search www.esmo.org.

ASXL1 p.(R693*) c.2077C>T, ASXL1 p.(E635Rfs*15) c.1900_1922delAGAGAGGCGGCCACCACTGCCAT

Prognostic significance: ELN 2017: Adverse

Cancer type: Acute Myeloid Leukemia **Variant class:** ASXL1 mutation

Reference: ESMO Clinical Practice Guidelines - ESMO-Acute Myeloblastic Leukaemia in Adult Patients [Ann Oncol (2020); 31(6): 697-712.]

Clinical Trials Summary

IDH1 p.(R132C) c.394C>T

NCT ID	Title	Phase
NCT04603001	A Phase I Study of Oral LY3410738 in Patients With Advanced Hematologic Malignancies With IDH1 or IDH2 Mutations.	I

Alerts Informed By Public Data Sources

Current FDA Information

Contraindicated Not recommended Resistance Breakthrough Fast Track

FDA information is current as of 2021-11-17. For the most up-to-date information, search www.fda.gov.

IDH1 p.(R132C) c.394C>T

ivosidenib

Cancer type: Myelodysplastic Syndrome

Variant class: IDH1 mutation

Supporting Statement:
The FDA has granted Breakthrough Designation to the isocitrate dehydrogenase-1 inhibitor, ivosidenib, for the treatment of adult patients with relapsed or refractory myelodysplastic syndrome (MDS) with a susceptible IDH1 mutation as detected by an FDA-approved test.

Reference:

https://investor.agios.com/news-releases/news-release-details/agios-receives-fda-breakthrough-therapy-designation-tibsovor-0

Signatures

Testing Personnel: Shu-Ying Wang 2022.02.14 08:37:36 +08'00'

Laboratory Supervisor: 2022.02.14 08:37:50 +08'00'

Pathologist: Yi-Chu Yeh 2022.02.14 08:38:03 +08'00'

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