



Sample Information

Patient Name: 陳津
Gender: Female
ID No.: F201620844
History No.: 37818545
Age: 76

Ordering Doctor: DOC1686E 陳玟均
Ordering REQ.: G1F87JM
Signing in Date: 2022/03/18

Path No.: S111-98725
MP No.: MY22010
Assay: Oncomine Myeloid Assay
Sample Type: Bone Marrow
Bone Marrow Aspirating Date: 2022/03/15

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

Note:

Sample Cancer Type: Myelodysplastic Syndrome

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Report Highlights

2 Relevant Biomarkers
12 Therapies Available
0 Clinical Trials

Relevant Myelodysplastic Syndrome Variants

Gene	Finding	Gene	Finding
ASXL1	ASXL1 p.(R693*) c.2077C>T	NPM1	None detected
BCOR	None detected	NRAS	None detected
CBL	None detected	NUP214	None detected
CREBBP	None detected	RUNX1	RUNX1 p.(F40Wfs*14) c.119_173delTCACGCCGCCTTCCACCGCG CTGAGCCCAGGCAAGATGAGCGAGGCGTT GCCGCT
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

Relevant Myelodysplastic Syndrome Variants (continued)

Gene	Finding	Gene	Finding
KMT2A	None detected	ZRSR2	None detected
MECOM	None detected		

Relevant Biomarkers

Genomic Alteration	Relevant Therapies (In this cancer type)	Relevant Therapies (In other cancer type)	Clinical Trials
<p><i>RUNX1 p.(F40Wfs*14)</i> <i>c.119_173delTCACGCCGCCTTCC</i> <i>ACCGCGCTGAGCCCAGGCAAGAT</i> <i>GAGCGAGGCGTTGCCGCT</i></p> <p>RUNX family transcription factor 1 Allele Frequency: 38.99%</p> <p>Prognostic significance: NCCN: Poor Diagnostic significance: None</p>	None	allogeneic stem cells azacitidine cytarabine cytarabine + daunorubicin cytarabine + daunorubicin + etoposide cytarabine + etoposide + idarubicin cytarabine + fludarabine + idarubicin + filgrastim cytarabine + idarubicin cytarabine + mitoxantrone decitabine gemtuzumab ozogamicin + chemotherapy venetoclax + chemotherapy	0
<p><i>ASXL1 p.(R693*) c.2077C>T</i></p> <p>ASXL transcriptional regulator 1 Allele Frequency: 23.34%</p> <p>Prognostic significance: NCCN: Poor Diagnostic significance: None</p>	None	allogeneic stem cells azacitidine cytarabine cytarabine + daunorubicin cytarabine + daunorubicin + etoposide cytarabine + etoposide + idarubicin cytarabine + fludarabine + idarubicin + filgrastim cytarabine + idarubicin cytarabine + mitoxantrone decitabine gemtuzumab ozogamicin + chemotherapy venetoclax + chemotherapy	0

Public data sources included in relevant therapies: FDA1, NCCN, EMA2, ESMO

Public data sources included in prognostic and diagnostic significance: NCCN, ESMO

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
ASXL1	p.(R693*)	c.2077C>T	COSM51388	chr20:31022592	23.34%	NM_015338.6	nonsense	921

Variants (Exclude variant in Taiwan BioBank with >1% allele frequency) (continued)

DNA Sequence Variants (continued)

Gene	Amino Acid Change	Coding	Variant ID	Locus	Allele Frequency	Transcript	Variant Effect	Coverage
RUNX1	p.(F40Wfs*14)	c.119_173delTCACG CCGCCTTCCACCGC GCTGAGCCCAGGCA AGATGAGCGAGGCG TTGCCGCT	.	chr21:36259317	38.99%	NM_001754.4	frameshift Deletion	1952
DNMT3A	p.(A741G)	c.2222C>G	.	chr2:25463271	46.24%	NM_022552.4	missense	1994
TET2	p.(P363L)	c.1088C>T	.	chr4:106156187	8.60%	NM_001127208.2	missense	1999
TP53	p.(V143M)	c.427G>A	.	chr17:7578503	85.55%	NM_000546.5	missense	685

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 ASXL3². 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 ASXL1⁸.

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 sequence¹⁸. As a consequence, detection of G646Wfs*12 may result as an artifact of PCR and/or sequencing¹⁹. 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 pathogenic²¹. Mutations in ASXL1 confer poor/adverse risk in AML and are associated with poor outcomes and adverse risk¹⁶. Additionally, ASXL1 nonsense or frameshift mutations are independently associated with poor prognosis in MDS and CMML²². Moreover, ASXL1 mutations are independently associated with inferior overall survival (OS) in patients with MPN or SM^{23,24}.

RUNX1 (RUNX family transcription factor 1)

Background: The RUNX1 gene encodes the runt-related transcription factor (RUNX) 1, part of the RUNX family of transcription factors which also includes RUNX2 and RUNX3²⁵. All RUNX proteins share several conserved regions with similar functionality including a highly conserved N-terminal 'runt' domain responsible for binding DNA, a C-terminal region composed of an activation domain, inhibitory domain, protein interacting motifs, and a nuclear matrix targeting signal²⁶. Each of these proteins are capable of interacting with core binding factor beta (CBFβ) to form the core binding factor (CBF) complex. Consequently, RUNX1, RUNX2, and RUNX3 are collectively known as core binding factor alpha (CBFα) since they can each function as the alpha subunit of CBF. Specifically, CBFβ binds to the 'runt' domain of RUNX1 leading to RUNX1 stabilization and increased affinity of the CBF complex for promoters involved in hematopoietic differentiation and cell cycle regulation^{27,28}. RUNX1 is frequently mutated in various hematological malignancies²⁸. Germline mutations in RUNX1 result in a rare autosomal dominant condition known as familial platelet disorder, with predisposition to acute myeloid leukemia (FPD/AML)^{29,30}. Somatic mutations and chromosomal translocations in RUNX1 are often observed in myelodysplastic syndrome (MDS), acute myeloid leukemia (AML), acute lymphoblastic leukemia (ALL), and chronic myelomonocytic leukemia (CMML)²⁸.

Biomarker Descriptions (continued)

Alterations and prevalence: RUNX1 is frequently rearranged in hematological malignancies with over 50 different observed translocations³¹. The most recurrent translocation, t(12;21)(q34;q11), results in ETV6-RUNX1 fusion and is observed in 20-25% of childhood ALL^{32,33,34}. This translocation is also observed in adult ALL at a lower frequency (2%)^{33,34}. Another recurrent translocation, t(8;21)(q22;q22), results in RUNX1-RUNX1T1 fusion and is observed in 5-10% of AML³⁵. The RUNX1-RUNX1T1 fusion, consists of the RHD domain of RUNX1 and the majority of RUNX1T1, which promotes oncogenesis by altering transcriptional regulation of RUNX1 target genes^{28,35}. Somatic mutations in RUNX1 include missense, nonsense, and frameshift mutations resulting in loss of function or dominant negative effects²⁸. RUNX1 mutations are reported in approximately 10% of de novo AML as well as 10-15% of MDS^{12,16,22,28}.

Potential relevance: The t(8;21)(q22;q22.1)/RUNX1-RUNX1T1 translocation is recognized as a distinct AML disease category by the World Health Organization (WHO)³⁶. Additionally, AML with RUNX1 mutations is a provisional entity in the WHO³⁶. Translocations involving RUNX1, specifically t(8;21)(q22;q22)/RUNX1-RUNX1T1 in AML and t(12;21)(q34;q11)/ETV6-RUNX1 in ALL, are associated with favorable risk^{16,37}. On the other hand, mutations in RUNX1 confer poor prognosis in AML, MDS, and systemic mastocytosis (SM)^{16,22,24}.

Relevant Therapy Summary

☒ In this cancer type
 ☐ In other cancer type
 ☒ In this cancer type and other cancer types
 ☒ No evidence

RUNX1 p.(F40Wfs*14) c.119_173delTCACGCCGCTTCCACCGCGCTGAGCCCAGGCAAGATGAGCGAGGCGTTGCCG CT

Relevant Therapy	FDA	NCCN	EMA	ESMO	Clinical Trials*
Allogeneic hematopoietic stem cell transplantation	×	○	×	×	×
azacitidine	×	○	×	×	×
cytarabine	×	○	×	×	×
cytarabine + daunorubicin	×	○	×	×	×
cytarabine + daunorubicin + etoposide	×	○	×	×	×
cytarabine + etoposide + idarubicin	×	○	×	×	×
cytarabine + fludarabine + idarubicin + filgrastim	×	○	×	×	×
cytarabine + idarubicin	×	○	×	×	×
cytarabine + mitoxantrone	×	○	×	×	×
decitabine	×	○	×	×	×
gemtuzumab ozogamicin + cytarabine + daunorubicin	×	○	×	×	×
venetoclax + azacitidine	×	○	×	×	×
venetoclax + cytarabine	×	○	×	×	×
venetoclax + decitabine	×	○	×	×	×

ASXL1 p.(R693*) c.2077C>T

Relevant Therapy	FDA	NCCN	EMA	ESMO	Clinical Trials*
Allogeneic hematopoietic stem cell transplantation	×	○	×	×	×

Relevant Therapy Summary (continued)

☒ In this cancer type
 ☐ In other cancer type
 ☒ In this cancer type and other cancer types
 ☒ No evidence

ASXL1 p.(R693*) c.2077C>T (continued)

Relevant Therapy	FDA	NCCN	EMA	ESMO	Clinical Trials*
azacitidine	×	○	×	×	×
cytarabine	×	○	×	×	×
cytarabine + daunorubicin	×	○	×	×	×
cytarabine + daunorubicin + etoposide	×	○	×	×	×
cytarabine + etoposide + idarubicin	×	○	×	×	×
cytarabine + fludarabine + idarubicin + filgrastim	×	○	×	×	×
cytarabine + idarubicin	×	○	×	×	×
cytarabine + mitoxantrone	×	○	×	×	×
decitabine	×	○	×	×	×
gemtuzumab ozogamicin + cytarabine + daunorubicin	×	○	×	×	×
venetoclax + azacitidine	×	○	×	×	×
venetoclax + cytarabine	×	○	×	×	×
venetoclax + decitabine	×	○	×	×	×

Relevant Therapy Details

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 2022-01-04. For the most up-to-date information, search www.nccn.org.
For NCCN International Adaptations & Translations, search www.nccn.org/global/international_adaptations.aspx.

RUNX1 p.(F40Wfs*14) c.119_173delTCACGCCGCTTCCACCGCGCTGAGCCCAGGCAAGATGAGCGAGGCGTTGCCG CT

☐ cytarabine + daunorubicin

Cancer type: Acute Myeloid Leukemia

Variant class: RUNX1 mutation

NCCN Recommendation category: 1

Population segment (Line of therapy):

- (Induction therapy); Other recommended intervention

Reference: NCCN Guidelines® - NCCN-Acute Myeloid Leukemia [Version 1.2022]

RUNX1 p.(F40Wfs*14)
c.119_173delTCACGCCGCTTCCACCGCGCTGAGCCCAGGCAAGATGAGCGAGGCGTTGCCG CT
 (continued)

☐ **cytarabine + daunorubicin + etoposide**

Cancer type: Acute Myeloid Leukemia

Variant class: RUNX1 mutation

NCCN Recommendation category: 1

Population segment (Line of therapy):

- (Induction therapy); Other recommended intervention

Reference: NCCN Guidelines® - NCCN-Acute Myeloid Leukemia [Version 1.2022]

☐ **cytarabine + etoposide + idarubicin**

Cancer type: Acute Myeloid Leukemia

Variant class: RUNX1 mutation

NCCN Recommendation category: 1

Population segment (Line of therapy):

- (Induction therapy); Other recommended intervention

Reference: NCCN Guidelines® - NCCN-Acute Myeloid Leukemia [Version 1.2022]

☐ **cytarabine + idarubicin**

Cancer type: Acute Myeloid Leukemia

Variant class: RUNX1 mutation

NCCN Recommendation category: 1

Population segment (Line of therapy):

- (Induction therapy); Other recommended intervention

Reference: NCCN Guidelines® - NCCN-Acute Myeloid Leukemia [Version 1.2022]

☐ **Allogeneic hematopoietic stem cell transplantation**

Cancer type: Acute Myeloid Leukemia

Variant class: RUNX1 mutation

NCCN Recommendation category: 2A

Population segment (Line of therapy):

- (Consolidation therapy)

Reference: NCCN Guidelines® - NCCN-Acute Myeloid Leukemia [Version 1.2022]

☐ **cytarabine**

Cancer type: Acute Myeloid Leukemia

Variant class: RUNX1 mutation

NCCN Recommendation category: 2A

Population segment (Line of therapy):

- (Consolidation therapy)

Reference: NCCN Guidelines® - NCCN-Acute Myeloid Leukemia [Version 1.2022]

RUNX1 p.(F40Wfs*14)
c.119_173delTCACGCCGCTTCCACCGCGCTGAGCCCAGGCAAGATGAGCGAGGCGTTGCCG CT
 (continued)

☐ **cytarabine + daunorubicin**

Cancer type: Acute Myeloid Leukemia

Variant class: RUNX1 mutation

NCCN Recommendation category: 2A

Population segment (Line of therapy):

- (Consolidation therapy)

Reference: NCCN Guidelines® - NCCN-Acute Myeloid Leukemia [Version 1.2022]

☐ **cytarabine + daunorubicin**

Cancer type: Acute Myeloid Leukemia

Variant class: RUNX1 mutation

NCCN Recommendation category: 2A

Population segment (Line of therapy):

- (Induction therapy); Preferred intervention

Reference: NCCN Guidelines® - NCCN-Acute Myeloid Leukemia [Version 1.2022]

☐ **cytarabine + idarubicin**

Cancer type: Acute Myeloid Leukemia

Variant class: RUNX1 mutation

NCCN Recommendation category: 2A

Population segment (Line of therapy):

- (Induction therapy); Preferred intervention

Reference: NCCN Guidelines® - NCCN-Acute Myeloid Leukemia [Version 1.2022]

☐ **cytarabine + mitoxantrone**

Cancer type: Acute Myeloid Leukemia

Variant class: RUNX1 mutation

NCCN Recommendation category: 2A

Population segment (Line of therapy):

- (Induction therapy); Preferred intervention

Reference: NCCN Guidelines® - NCCN-Acute Myeloid Leukemia [Version 1.2022]

☐ **decitabine**

Cancer type: Acute Myeloid Leukemia

Variant class: RUNX1 mutation

NCCN Recommendation category: 2A

Population segment (Line of therapy):

- (Induction therapy)

Reference: NCCN Guidelines® - NCCN-Acute Myeloid Leukemia [Version 1.2022]

RUNX1 p.(F40Wfs*14)
c.119_173delTCACGCCGCTTCCACCGCGCTGAGCCCAGGCAAGATGAGCGAGGCGTTGCCG CT
 (continued)

☐ **gemtuzumab ozogamicin + cytarabine + daunorubicin**

Cancer type: Acute Myeloid Leukemia

Variant class: RUNX1 mutation

NCCN Recommendation category: 2A

Population segment (Line of therapy):

- (Induction therapy); Preferred intervention

Reference: NCCN Guidelines® - NCCN-Acute Myeloid Leukemia [Version 1.2022]

☐ **venetoclax + azacitidine**

Cancer type: Acute Myeloid Leukemia

Variant class: RUNX1 mutation

NCCN Recommendation category: 2A

Population segment (Line of therapy):

- (Induction therapy)

Reference: NCCN Guidelines® - NCCN-Acute Myeloid Leukemia [Version 1.2022]

☐ **venetoclax + cytarabine**

Cancer type: Acute Myeloid Leukemia

Variant class: RUNX1 mutation

NCCN Recommendation category: 2A

Population segment (Line of therapy):

- (Induction therapy)

Reference: NCCN Guidelines® - NCCN-Acute Myeloid Leukemia [Version 1.2022]

☐ **venetoclax + decitabine**

Cancer type: Acute Myeloid Leukemia

Variant class: RUNX1 mutation

NCCN Recommendation category: 2A

Population segment (Line of therapy):

- (Induction therapy)

Reference: NCCN Guidelines® - NCCN-Acute Myeloid Leukemia [Version 1.2022]

☐ **azacitidine**

Cancer type: Acute Myeloid Leukemia

Variant class: RUNX1 mutation

NCCN Recommendation category: 2B

Population segment (Line of therapy):

- (Induction therapy)

Reference: NCCN Guidelines® - NCCN-Acute Myeloid Leukemia [Version 1.2022]

RUNX1 p.(F40Wfs*14)
c.119_173delTCACGCCGCTTCCACCGCGCTGAGCCCAGGCAAGATGAGCGAGGCGTTGCCG CT
 (continued)

☐ **cytarabine + daunorubicin + etoposide**

Cancer type: Acute Myeloid Leukemia

Variant class: RUNX1 mutation

NCCN Recommendation category: 2B

Population segment (Line of therapy):

- (Induction therapy); Other recommended intervention

Reference: NCCN Guidelines® - NCCN-Acute Myeloid Leukemia [Version 1.2022]

☐ **cytarabine + etoposide + idarubicin**

Cancer type: Acute Myeloid Leukemia

Variant class: RUNX1 mutation

NCCN Recommendation category: 2B

Population segment (Line of therapy):

- (Induction therapy); Other recommended intervention

Reference: NCCN Guidelines® - NCCN-Acute Myeloid Leukemia [Version 1.2022]

☐ **cytarabine + fludarabine + idarubicin + filgrastim**

Cancer type: Acute Myeloid Leukemia

Variant class: RUNX1 mutation

NCCN Recommendation category: 2B

Population segment (Line of therapy):

- (Induction therapy); Other recommended intervention

Reference: NCCN Guidelines® - NCCN-Acute Myeloid Leukemia [Version 1.2022]

ASXL1 p.(R693*) c.2077C>T

☐ **cytarabine + daunorubicin**

Cancer type: Acute Myeloid Leukemia

Variant class: ASXL1 mutation

NCCN Recommendation category: 1

Population segment (Line of therapy):

- (Induction therapy); Other recommended intervention

Reference: NCCN Guidelines® - NCCN-Acute Myeloid Leukemia [Version 1.2022]

ASXL1 p.(R693*) c.2077C>T (continued)**○ cytarabine + daunorubicin + etoposide**

Cancer type: Acute Myeloid Leukemia

Variant class: ASXL1 mutation

NCCN Recommendation category: 1

Population segment (Line of therapy):

- (Induction therapy); Other recommended intervention

Reference: NCCN Guidelines® - NCCN-Acute Myeloid Leukemia [Version 1.2022]

○ cytarabine + etoposide + idarubicin

Cancer type: Acute Myeloid Leukemia

Variant class: ASXL1 mutation

NCCN Recommendation category: 1

Population segment (Line of therapy):

- (Induction therapy); Other recommended intervention

Reference: NCCN Guidelines® - NCCN-Acute Myeloid Leukemia [Version 1.2022]

○ cytarabine + idarubicin

Cancer type: Acute Myeloid Leukemia

Variant class: ASXL1 mutation

NCCN Recommendation category: 1

Population segment (Line of therapy):

- (Induction therapy); Other recommended intervention

Reference: NCCN Guidelines® - NCCN-Acute Myeloid Leukemia [Version 1.2022]

○ Allogeneic hematopoietic stem cell transplantation

Cancer type: Acute Myeloid Leukemia

Variant class: ASXL1 mutation

NCCN Recommendation category: 2A

Population segment (Line of therapy):

- (Consolidation therapy)

Reference: NCCN Guidelines® - NCCN-Acute Myeloid Leukemia [Version 1.2022]

○ cytarabine

Cancer type: Acute Myeloid Leukemia

Variant class: ASXL1 mutation

NCCN Recommendation category: 2A

Population segment (Line of therapy):

- (Consolidation therapy)

Reference: NCCN Guidelines® - NCCN-Acute Myeloid Leukemia [Version 1.2022]

ASXL1 p.(R693*) c.2077C>T (continued)**○ cytarabine + daunorubicin**

Cancer type: Acute Myeloid Leukemia

Variant class: ASXL1 mutation

NCCN Recommendation category: 2A

Population segment (Line of therapy):

- (Consolidation therapy)

Reference: NCCN Guidelines® - NCCN-Acute Myeloid Leukemia [Version 1.2022]

○ cytarabine + daunorubicin

Cancer type: Acute Myeloid Leukemia

Variant class: ASXL1 mutation

NCCN Recommendation category: 2A

Population segment (Line of therapy):

- (Induction therapy); Preferred intervention

Reference: NCCN Guidelines® - NCCN-Acute Myeloid Leukemia [Version 1.2022]

○ cytarabine + idarubicin

Cancer type: Acute Myeloid Leukemia

Variant class: ASXL1 mutation

NCCN Recommendation category: 2A

Population segment (Line of therapy):

- (Induction therapy); Preferred intervention

Reference: NCCN Guidelines® - NCCN-Acute Myeloid Leukemia [Version 1.2022]

○ cytarabine + mitoxantrone

Cancer type: Acute Myeloid Leukemia

Variant class: ASXL1 mutation

NCCN Recommendation category: 2A

Population segment (Line of therapy):

- (Induction therapy); Preferred intervention

Reference: NCCN Guidelines® - NCCN-Acute Myeloid Leukemia [Version 1.2022]

○ decitabine

Cancer type: Acute Myeloid Leukemia

Variant class: ASXL1 mutation

NCCN Recommendation category: 2A

Population segment (Line of therapy):

- (Induction therapy)

Reference: NCCN Guidelines® - NCCN-Acute Myeloid Leukemia [Version 1.2022]

ASXL1 p.(R693*) c.2077C>T (continued)**○ gemtuzumab ozogamicin + cytarabine + daunorubicin**

Cancer type: Acute Myeloid Leukemia

Variant class: ASXL1 mutation

NCCN Recommendation category: 2A

Population segment (Line of therapy):

- (Induction therapy); Preferred intervention

Reference: NCCN Guidelines® - NCCN-Acute Myeloid Leukemia [Version 1.2022]

○ venetoclax + azacitidine

Cancer type: Acute Myeloid Leukemia

Variant class: ASXL1 mutation

NCCN Recommendation category: 2A

Population segment (Line of therapy):

- (Induction therapy)

Reference: NCCN Guidelines® - NCCN-Acute Myeloid Leukemia [Version 1.2022]

○ venetoclax + cytarabine

Cancer type: Acute Myeloid Leukemia

Variant class: ASXL1 mutation

NCCN Recommendation category: 2A

Population segment (Line of therapy):

- (Induction therapy)

Reference: NCCN Guidelines® - NCCN-Acute Myeloid Leukemia [Version 1.2022]

○ venetoclax + decitabine

Cancer type: Acute Myeloid Leukemia

Variant class: ASXL1 mutation

NCCN Recommendation category: 2A

Population segment (Line of therapy):

- (Induction therapy)

Reference: NCCN Guidelines® - NCCN-Acute Myeloid Leukemia [Version 1.2022]

○ azacitidine

Cancer type: Acute Myeloid Leukemia

Variant class: ASXL1 mutation

NCCN Recommendation category: 2B

Population segment (Line of therapy):

- (Induction therapy)

Reference: NCCN Guidelines® - NCCN-Acute Myeloid Leukemia [Version 1.2022]

ASXL1 p.(R693*) c.2077C>T (continued)**○ cytarabine + daunorubicin + etoposide**

Cancer type: Acute Myeloid Leukemia

Variant class: ASXL1 mutation

NCCN Recommendation category: 2B

Population segment (Line of therapy):

- (Induction therapy); Other recommended intervention

Reference: NCCN Guidelines® - NCCN-Acute Myeloid Leukemia [Version 1.2022]

○ cytarabine + etoposide + idarubicin

Cancer type: Acute Myeloid Leukemia

Variant class: ASXL1 mutation

NCCN Recommendation category: 2B

Population segment (Line of therapy):

- (Induction therapy); Other recommended intervention

Reference: NCCN Guidelines® - NCCN-Acute Myeloid Leukemia [Version 1.2022]

○ cytarabine + fludarabine + idarubicin + filgrastim

Cancer type: Acute Myeloid Leukemia

Variant class: ASXL1 mutation

NCCN Recommendation category: 2B

Population segment (Line of therapy):

- (Induction therapy); Other recommended intervention

Reference: NCCN Guidelines® - NCCN-Acute Myeloid Leukemia [Version 1.2022]

Prognostic Details

Current NCCN Information

NCCN information is current as of 2022-01-04. For the most up-to-date information, search www.nccn.org.
For NCCN International Adaptations & Translations, search www.nccn.org/global/international_adaptations.aspx.

RUNX1 p.(F40Wfs*14)
c.119_173delTCACGCCGCTTCCACGCGCTGAGCCCAGGCAAGATGAGCGAGGCGTTGCCG CT

Prognostic significance: NCCN: Poor

Cancer type: Myelodysplastic Syndrome

Variant class: RUNX1 truncating mutation

NCCN Recommendation category: 2A

Summary:

- NCCN Guidelines® independently associate the biomarker with poor prognosis

Reference: NCCN Guidelines® - NCCN-Myelodysplastic Syndromes [Version 2.2022]

ASXL1 p.(R693*) c.2077C>T

Prognostic significance: NCCN: Poor

Cancer type: Myelodysplastic Syndrome

Variant class: ASXL1 truncating mutation

NCCN Recommendation category: 2A

Summary:

- NCCN Guidelines® independently associate the biomarker with poor prognosis in MDS and CMML

Reference: NCCN Guidelines® - NCCN-Myelodysplastic Syndromes [Version 2.2022]

Signatures

Testing Personnel:

Laboratory Supervisor:

Pathologist:

References

- O'Leary et al. Reference sequence (RefSeq) database at NCBI: current status, taxonomic expansion, and functional annotation. *Nucleic Acids Res.* 2016 Jan 4;44(D1):D733-45. PMID: 26553804
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