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: 陳詮鈞
Gender: Male
ID No.: F121825665
History No.: 43467909
Age: 50

Ordering Doctor: DOC6258D 林益庭
Ordering REQ.: 0CLNRCZ
Signing in Date: 2023/06/07

Path No.: M112-00131
MP No.: MY23031
Assay: Oncomine Myeloid Assay
Sample Type: Bone Marrow
Bone Marrow Aspirating Date: 2023/05/29

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

Note:

Sample Cancer Type: Acute Myeloid Leukemia

Table of Contents

Report Highlights

1 Relevant Biomarkers
12 Therapies Available
0 Clinical Trials

Relevant Acute Myeloid Leukemia Variants

Gene	Finding	Gene	Finding
ABL1	None detected	MECOM	None detected
ASXL1	***ASXL1 p.(A640Gfs*13) c.1919_1932delCCATCGGAGGGGGG***	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

Tier	Genomic Alteration	Relevant Therapies (In this cancer type)	Relevant Therapies (In other cancer type)	Clinical Trials
IA	*ASXL1 p.(A640Gfs*13) c.1919_1932delCCATCGGAGGGGGG* ASXL transcriptional regulator 1 Allele Frequency: 62.72%	allogeneic stem cells azacitidine cytarabine cytarabine + daunorubicin cytarabine + daunorubicin + etoposide cytarabine + etoposide + idarubicin cytarabine + fludarabine + idarubicin + filgrastim cytarabine + idarubicin cytarabine + mitoxantrone decitabine liposomal cytarabine-daunorubicin CPX-351 venetoclax + chemotherapy	None	0

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

Prevalent cancer biomarkers without relevant evidence based on included data sources
*ETV6 p.(S26Lfs*41) c.74_75insCTTA*

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
ETV6	p.(S26Lfs*41)	c.74_75insCTTA	.	chr12:11905424	3.27%	NM_001987.5	frameshift Insertion	1987
ASXL1	p.(A640Gfs*13)	c.1919_1932delCCATCGGAGGGGGG	.	chr20:31022433	62.72%	NM_015338.6	frameshift Deletion	1894
ETV6	p.(D65_V66insGS)	c.196_197insGGTCCG	.	chr12:11992105	12.42%	NM_001987.5	nonframeshift Insertion	1980
FLT3	p.(S543P)	c.1627T>C	.	chr13:28608515	11.65%	NM_004119.3	missense	2000
CEBPA	p.(H195_P196dup)	c.589_590insACCCGC	.	chr19:33792731	36.25%	NM_004364.4	nonframeshift Insertion	549

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].

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

Biomarker Descriptions (continued)

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].

ETV6 (ETS variant transcription factor 6)

Background: The ETV6 gene encodes the E twenty-six (ETS) variant 1 transcription factor. ETV6 contains an N-terminal pointed (PNT) domain responsible for protein-protein interactions and a C-terminal ETS domain involved in DNA binding[25]. ETV6 plays a critical role in embryonic development as well as hematopoiesis and is the target of chromosomal rearrangement and missense mutations in hematological malignancies as well as solid tumors[26,27]. Hereditary mutations in ETV6 are associated with a predisposition to hematological cancers, including acute lymphoblastic leukemia (ALL), acute myeloid leukemia (AML), and myelodysplastic syndromes (MDS)[22,28,29].

Alterations and prevalence: ETV6 translocations are prevalent in hematological malignancies and have been observed with numerous fusion partners[30]. The most recurrent translocation is t(12;21)(p13;q22) which results in ETV6-RUNX1 fusion and is observed in 20-25% childhood acute lymphoblastic leukemia (ALL)[30,31,32]. ETV6-RUNX1 fusions are also observed in adult ALL (2%)[31,32]. The t(5;12)(q33;p13) translocation which results in the ETV6-PDGFRB fusion is recurrent in chronic myelomonocytic leukemia (CMML)[30,33]. Other ETV6 fusions including ETV6-PDGFRA, ETV6-NTRK2, ETV6-NTRK3, and ETV6-ABL1 are reported in hematological malignancies as well as solid tumors[27,30,34]. ETV6 fusions involving a receptor tyrosine kinase (RTK) fusion partner retains the ETV6 PNT domain and the tyrosine kinase domain of the RTK, leading to constitutive kinase activation[30,34]. Mutations in ETV6 are primarily missense, nonsense, or frameshift and are observed in about 1-5% of select myeloid malignancies and solid tumors, including chronic lymphocytic leukemia (CLL), chronic myeloid leukemia (CML), diffuse large B-cell lymphoma (DLBCL), MDS, AML, ALL, melanoma, lung, bladder, stomach, colorectal, and uterine cancers[12,25,35]. ETV6 mutations occur in the PNT and ETS domain of ETV6 and may impair ETV6 oligomerization or DNA-binding, respectively[25].

Potential relevance: ETV6-NTRK3 fusion is useful as an ancillary diagnostic marker in congenital/infantile fibrosarcoma and inflammatory myofibroblastic tumors[36]. Additionally, ETV6-RUNX1 fusion is diagnostic of the B-cell Lymphoblastic Leukemia/Lymphoma subtype of ALL[37]. ETV6 fusion is Nonsense or frameshift mutations in ETV6 are independently associated with poor prognosis in MDS[22]. However, ETV6-RUNX1 fusions are associated with favorable outcomes in ALL and good risk in B-cell ALL (B-ALL)[32]. ETV6 fusions that partner with a RTKs demonstrate response to various tyrosine kinase inhibitors such as imatinib, nilotinib, and entrectinib. Specifically, individual case reports of an ETV6-PDGFRA fusion chronic eosinophilic leukemia patient and an ETV6-PDGFRB fusion CMML patient treated with imatinib demonstrated complete cytogenetic response (CCyR) and complete hematological responses, respectively[38,39]. Additionally, an ETV6-ABL1 fusion Ph-negative CML patient treated with nilotinib demonstrated CCyR and major molecular response (MMR) at 22 months from diagnosis[40]. In another case report, an ETV6-NTRK3 fusion mammary analogue secretory carcinoma (MASC) patient demonstrated partial response to entrectinib with 89% reduction in tumor burden[41].

Relevant Therapy Summary

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

ASXL1 p.(A640Gfs*13) c.1919_1932delCCATCGGAGGGGGG

Relevant Therapy	FDA	NCCN	EMA	ESMO	Clinical Trials*
Allogeneic hematopoietic stem cell transplantation	✖	●	✖	✖	✖
azacitidine	✖	●	✖	✖	✖

Relevant Therapy Summary (continued)

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

ASXL1 p.(A640Gfs*13) c.1919_1932delCCATCGGAGGGGGG (continued)

Relevant Therapy	FDA	NCCN	EMA	ESMO	Clinical Trials*
cytarabine	✖	●	✖	✖	✖
cytarabine + daunorubicin	✖	●	✖	✖	✖
cytarabine + daunorubicin + etoposide	✖	●	✖	✖	✖
cytarabine + etoposide + idarubicin	✖	●	✖	✖	✖
cytarabine + fludarabine + idarubicin + filgrastim	✖	●	✖	✖	✖
cytarabine + idarubicin	✖	●	✖	✖	✖
cytarabine + mitoxantrone	✖	●	✖	✖	✖
decitabine	✖	●	✖	✖	✖
liposomal cytarabine-daunorubicin CPX-351	✖	●	✖	✖	✖
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 2023-03-01. 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.(A640Gfs*13) c.1919_1932delCCATCGGAGGGGGG

● 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 3.2022]

ASXL1 p.(A640Gfs*13) c.1919_1932delCCATCGGAGGGGGG (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 3.2022]

---

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 3.2022]

---

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 3.2022]

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); Preferred intervention

Reference: NCCN Guidelines® - NCCN-Acute Myeloid Leukemia [Version 3.2022]

---

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 3.2022]

ASXL1 p.(A640Gfs*13) c.1919_1932delCCATCGGAGGGGGG (continued)

cytarabine + daunorubicin

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

NCCN Recommendation category: 2A

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

Reference: NCCN Guidelines® - NCCN-Acute Myeloid Leukemia [Version 3.2022]

---

cytarabine + idarubicin

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

NCCN Recommendation category: 2A

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

Reference: NCCN Guidelines® - NCCN-Acute Myeloid Leukemia [Version 3.2022]

---

cytarabine + mitoxantrone

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

NCCN Recommendation category: 2A

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

Reference: NCCN Guidelines® - NCCN-Acute Myeloid Leukemia [Version 3.2022]

---

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 3.2022]

---

liposomal cytarabine-daunorubicin CPX-351

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 3.2022]

ASXL1 p.(A640Gfs*13) c.1919_1932delCCATCGGAGGGGGG (continued)

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 3.2022]

---

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 3.2022]

---

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 3.2022]

---

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 3.2022]

---

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 3.2022]

ASXL1 p.(A640Gfs*13) c.1919_1932delCCATCGGAGGGGGG (continued)

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 3.2022]

---

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 3.2022]

References

1. 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
2. Katoh. Functional and cancer genomics of ASXL family members. Br. J. Cancer. 2013 Jul 23;109(2):299-306. PMID: 23736028
3. Gelsi-Boyer et al. Mutations of polycomb-associated gene ASXL1 in myelodysplastic syndromes and chronic myelomonocytic leukaemia. Br. J. Haematol. 2009 Jun;145(6):788-800. PMID: 19388938
4. Gelsi-Boyer et al. Mutations in ASXL1 are associated with poor prognosis across the spectrum of malignant myeloid diseases. J Hematol Oncol. 2012 Mar 21;5:12. doi: 10.1186/1756-8722-5-12. PMID: 22436456
5. Larsson et al. The changing mutational landscape of acute myeloid leukemia and myelodysplastic syndrome. Mol. Cancer Res. 2013 Aug;11(8):815-27. PMID: 23645565
6. Alvarez et al. ASXL1 mutations in myeloid neoplasms: pathogenetic considerations, impact on clinical outcomes and survival. Curr Med Res Opin. 2018 May;34(5):757-763. PMID: 28027687
7. Yang et al. Gain of function of ASXL1 truncating protein in the pathogenesis of myeloid malignancies. Blood. 2018 Jan 18;131(3):328-341. PMID: 29113963
8. Abdel-Wahab et al. ASXL1 mutations promote myeloid transformation through loss of PRC2-mediated gene repression. Cancer Cell. 2012 Aug 14;22(2):180-93. PMID: 22897849
9. Alberti et al. Discriminating a common somatic ASXL1 mutation (c.1934dup; p.G646Wfs*12) from artifact in myeloid malignancies using NGS. Leukemia. 2018 Aug;32(8):1874-1878. PMID: 29959414
10. Kakosaiou et al. ASXL1 mutations in AML are associated with specific clinical and cytogenetic characteristics. Leuk. Lymphoma. 2018 Oct;59(10):2439-2446. PMID: 29411666
11. Paschka et al. ASXL1 mutations in younger adult patients with acute myeloid leukemia: a study by the German-Austrian Acute Myeloid Leukemia Study Group. Haematologica. 2015 Mar;100(3):324-30. PMID: 25596267
12. Weinstein et al. The Cancer Genome Atlas Pan-Cancer analysis project. Nat. Genet. 2013 Oct;45(10):1113-20. PMID: 24071849
13. Jawhar et al. The clinical and molecular diversity of mast cell leukemia with or without associated hematologic neoplasm. Haematologica. 2017 Jun;102(6):1035-1043. PMID: 28255023
14. Jawhar et al. KIT D816 mutated/CBF-negative acute myeloid leukemia: a poor-risk subtype associated with systemic mastocytosis. Leukemia. 2019 May;33(5):1124-1134. PMID: 30635631
15. Damaj et al. ASXL1 but not TET2 mutations adversely impact overall survival of patients suffering systemic mastocytosis with associated clonal hematologic non-mast-cell diseases. PLoS ONE. 2014;9(1):e85362. PMID: 24465546
16. NCCN Guidelines® - NCCN-Acute Myeloid Leukemia [Version 3.2022]
17. Boultwood et al. Frequent mutation of the polycomb-associated gene ASXL1 in the myelodysplastic syndromes and in acute myeloid leukemia. Leukemia. 2010 May;24(5):1062-5. doi: 10.1038/leu.2010.20. Epub 2010 Feb 25. PMID: 20182461
18. Yannakou et al. ASXL1 c.1934dup;p.Gly646Trpfs*12-a true somatic alteration requiring a new approach. Blood Cancer J. 2017 Dec 15;7(12):656. doi: 10.1038/s41408-017-0025-8. PMID: 29242575
19. Abdel-Wahab et al. The most commonly reported variant in ASXL1 (c.1934dupG;p.Gly646TrpfsX12) is not a somatic alteration. Leukemia. 2010 Sep;24(9):1656-7. doi: 10.1038/leu.2010.144. Epub 2010 Jul 1. PMID: 20596031
20. Montes-Moreno et al. Clinical molecular testing for ASXL1 c.1934dupG p.Gly646fs mutation in hematologic neoplasms in the NGS era. PLoS ONE. 2018;13(9):e0204218. PMID: 30222780
21. Landrum et al. ClinVar: improving access to variant interpretations and supporting evidence. Nucleic Acids Res. 2018 Jan 4;46(D1):D1062-D1067. PMID: 29165669
22. NCCN Guidelines® - NCCN-Myelodysplastic Syndromes [Version 1.2023]
23. NCCN Guidelines® - NCCN-Myeloproliferative Neoplasms [Version 3.2022]
24. NCCN Guidelines® - NCCN-Systemic Mastocytosis [Version 1.2020]
25. Wang et al. ETV6 mutation in a cohort of 970 patients with hematologic malignancies. Haematologica. 2014 Oct;99(10):e176-8. PMID: 24997145
26. Wang et al. The TEL/ETV6 gene is required specifically for hematopoiesis in the bone marrow. Genes Dev. 1998 Aug 1;12(15):2392-402. PMID: 9694803
27. Huret et al. Atlas of genetics and cytogenetics in oncology and haematology in 2013. Nucleic Acids Res. 2013 Jan;41(Database issue):D920-4. PMID: 23161685
28. Feurstein et al. Germline ETV6 mutations and predisposition to hematological malignancies. Int. J. Hematol. 2017 Aug;106(2):189-195. PMID: 28555414

References (continued)

29. Melazzini et al. Clinical and pathogenic features of ETV6-related thrombocytopenia with predisposition to acute lymphoblastic leukemia. Haematologica. 2016 Nov;101(11):1333-1342. PMID: 27365488
30. De et al. ETV6 fusion genes in hematological malignancies: a review. Leuk. Res. 2012 Aug;36(8):945-61. PMID: 22578774
31. Pui et al. Acute lymphoblastic leukemia. N. Engl. J. Med. 2004 Apr 8;350(15):1535-48. PMID: 15071128
32. NCCN Guidelines® - Acute Lymphoblastic Leukemia [Version 2.2019]. 2019 May 15
33. Golub et al. Fusion of PDGF receptor beta to a novel ets-like gene, tel, in chronic myelomonocytic leukemia with t(5;12) chromosomal translocation. Cell. 1994 Apr 22;77(2):307-16. PMID: 8168137
34. Taylor et al. Oncogenic TRK fusions are amenable to inhibition in hematologic malignancies. J. Clin. Invest. 2018 Aug 31;128(9):3819-3825. PMID: 29920189
35. Bejar et al. Clinical effect of point mutations in myelodysplastic syndromes. N. Engl. J. Med. 2011 Jun 30;364(26):2496-506. PMID: 21714648
36. NCCN Guidelines® - NCCN-Soft Tissue Sarcoma [Version 2.2022]
37. NCCN Guidelines® - NCCN-Acute Lymphoblastic Leukemia [Version 1.2022]
38. Curtis et al. Two novel imatinib-responsive PDGFRA fusion genes in chronic eosinophilic leukaemia. Br. J. Haematol. 2007 Jul;138(1):77-81. PMID: 17555450
39. Curtis et al. A novel ETV6-PDGFRB fusion transcript missed by standard screening in a patient with an imatinib responsive chronic myeloproliferative disease. Leukemia. 2007 Aug;21(8):1839-41. Epub 2007 May 17. PMID: 17508004
40. Gancheva et al. Myeloproliferative neoplasm with ETV6-ABL1 fusion: a case report and literature review. Mol Cytogenet. 2013 Sep 20;6(1):39. doi: 10.1186/1755-8166-6-39. PMID: 24053143
41. Drilon et al. What hides behind the MASC: clinical response and acquired resistance to entrectinib after ETV6-NTRK3 identification in a mammary analogue secretory carcinoma (MASC). Ann Oncol. 2016 May;27(5):920-6. doi: 10.1093/annonc/mdw042. Epub 2016 Feb 15. PMID: 26884591