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Patient Name: 유경자
Gender: F
Sample ID: N25-39

Primary Tumor Site: lymph node
Collection Date: 2025.05.21

Sample Cancer Type: Large B-Cell Lymphoma

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Relevant Biomarkers

Tier	Genomic Alteration	Relevant Therapies (In this cancer type)	Relevant Therapies (In other cancer type)	Clinical Trials
IIC	*CD79B p.(Y197H) c.589T>C* CD79b molecule Allele Frequency: 46.45% Locus: chr17:62006799 Transcript: NM_001039933.3 **Diagnostic significance:** Diffuse Large B-Cell Lymphoma	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
Line of therapy: I: First-line therapy, II+: Other line of therapy
Tier Reference: Li et al. *Standards and Guidelines for the Interpretation and Reporting of Sequence Variants in Cancer: A Joint Consensus Recommendation of the Association for Molecular Pathology, American Society of Clinical Oncology, and College of American Pathologists.* J Mol Diagn. 2017 Jan;19(1):4-23.

Prevalent cancer biomarkers without relevant evidence based on included data sources
ARID1A p.(Q1334del) c.3999_4001delGCA

Variant Details

DNA Sequence Variants

Gene	Amino Acid Change	Coding	Variant ID	Locus	Allele Frequency	Transcript	Variant Effect
ARID1A	p.(Q1334del)	c.3999_4001delGCA	COSM298325	chr1:27100181	3.98%	NM_006015.6	nonframeshift Deletion
CD79B	p.(Y197H)	c.589T>C	COSM220734	chr17:62006799	46.45%	NM_001039933.3	missense
TNFRSF14	p.(K17R)	c.50A>G	.	chr1:2488153	49.60%	NM_003820.3	missense
ID3	p.(T105A)	c.313A>G	.	chr1:23885498	99.75%	NM_002167.5	missense
ARID1A	p.(E28G)	c.83A>G	.	chr1:27022977	4.10%	NM_006015.6	missense
ARID1A	p.(G864C)	c.2590G>T	.	chr1:27089634	5.66%	NM_006015.6	missense
NFKBIE	p.(V55A)	c.164T>C	.	chr6:44232920	48.37%	NM_004556.3	missense
NFKBIE	p.(P36L)	c.107C>T	.	chr6:44232977	50.63%	NM_004556.3	missense

Variant Details (continued)

DNA Sequence Variants (continued)

Gene	Amino Acid Change	Coding	Variant ID	Locus	Allele Frequency	Transcript	Variant Effect
KLLN	p.(?)	c.-1616AG>G	.	chr10:89623860	100.00%	NM_001126049.2	unknown
PTEN	p.(?)	c.-367CT>C	.	chr10:89623860	100.00%	NM_000314.8	unknown
ATM	p.(N1983S)	c.5948A>G	.	chr11:108183167	99.80%	NM_000051.4	missense
KMT2D	p.(R4162Q)	c.12485G>A	.	chr12:49426003	64.70%	NM_003482.4	missense
KMT2D	p.(A476T)	c.1426G>A	.	chr12:49446040	6.65%	NM_003482.4	missense
KMT2D	p.(S456L)	c.1367C>T	.	chr12:49446099	3.30%	NM_003482.4	missense
FOXO1	p.(A102dup)	c.292_294dup	.	chr13:41240055	67.32%	NM_002015.4	nonframeshift Insertion
FOXO1	p.(D82N)	c.244G>A	.	chr13:41240106	37.50%	NM_002015.4	missense
DIS3	p.(N269S)	c.806A>G	.	chr13:73350079	99.70%	NM_014953.5	missense
TP53	p.(P72R)	c.215C>G	.	chr17:7579472	80.81%	NM_000546.6	missense
BCL2	p.(P40Rfs*112)	c.119_120delCG	.	chr18:60985779	96.10%	NM_000633.3	frameshift Deletion
MEF2B	p.(G328Afs*75)	c.981_984delCGGG	.	chr19:19256728	10.26%	NM_001145785.2	frameshift Deletion

Biomarker Descriptions

ARID1A p.(Q1334del) c.3999_4001delGCA

AT-rich interaction domain 1A

Background: The ARID1A gene encodes the AT-rich interaction domain 1A tumor suppressor protein[1]. ARID1A, also known as BAF250A, belongs to the ARID1 subfamily that also includes AR1D1B[1,2]. ARID1A and ARID1B are mutually exclusive subunits of the BAF variant of the SWI/SNF chromatin-remodeling complex[2,3]. The BAF complex is a multisubunit protein that consists of SMARCB1/IN1, SMARCC1/BAF155, SMARCC2/BAF170, SMARCA4/BRG1 or SMARCA2/BRM, and ARID1A or ARID1B[3]. The BAF complex remodels chromatin at promoter and enhancer elements to alter and regulate gene expression[3,4]. ARID1A binds to transcription factors and coactivator/corepressor complexes to alter transcription[2]. Recurrent inactivating mutations in BAF complex subunits, including ARID1A, lead to transcriptional dysfunction thereby, altering its tumor suppressor function[2].

Alterations and prevalence: Mutations in SWI/SNF complex subunits are the most commonly mutated chromatin modulators in cancer and have been observed in 20% of all tumors[4]. The majority of ARID1A inactivating mutations are nonsense or frameshift mutations[2]. Somatic mutations in ARID1A have been identified in 50% of ovarian clear cell carcinoma, 30% of endometrioid carcinoma, and 24-43% of uterine corpus endometrial carcinoma, bladder urothelial carcinoma, and stomach adenocarcinoma[3,5,6]. In microsatellite stable (MSS) colorectal cancer, mutations in ARID1A have been observed to correlate with increased tumor mutational burden (TMB) and expression of genes involved in the immune response[7].

Potential relevance: Currently, no therapies are approved for ARID1A aberrations. However, the FDA has granted fast track designation (2022) to HSF1 pathway inhibitor, NXP-800[8], for the treatment of platinum resistant ARID1A-mutated ovarian carcinoma. Tulmimetostat[9], dual inhibitor of EZH2 and EZH1, was also granted a fast track designation (2023) for the treatment of patients with advanced, recurrent or metastatic endometrial cancer harboring ARID1A mutations and who have progressed on at least one prior line of treatment.

Biomarker Descriptions (continued)

CD79B p.(Y197H) c.589T>C

CD79b molecule

Background: The CD79B gene encodes the cluster of differentiation 79b molecule, also known as B29[10]. CD79B, along with CD79A, are two distinct proteins that heterodimerize to form the CD79 transmembrane protein complex[11]. CD79 is a member of the immunoglobulin superfamily of cell surface and soluble proteins, and is physically linked to membrane immunoglobulin to form the B-cell antigen receptor complex present on the surface of B-cells[10]. Specifically, CD79B is essential for B-cell development and acts as a signal transducer after antigen recognition by surface immunoglobulin[12]. Due to its exclusivity to B-cells, CD79 is considered a useful marker for the detection of various B-cell neoplasms[10]. Mutations in CD79B or CD79A lead to chronic activation of B-cell receptor (BCR) signaling, supporting an oncogenic role for CD79B[10].

Alterations and prevalence: Somatic mutations in CD79B are observed in 12% of diffuse large B-cell lymphoma (DLBCL)[5,6]. Oncogenic missense mutations at Y196 located in the ITAM domain is shown to result in constitutive BCR signaling[13]. Amplifications in CD79B are observed in 6% of breast invasive carcinoma, 5% of mesothelioma, 3% of liver cancer, and 2% of bladder cancer[5,6]. CD79B is expressed in 80-90% of mature B-cell neoplasms, and is also observed in 96% of mantle cell lymphoma, 83% of follicular lymphoma, and 80% of DLBCL[10].

Potential relevance: Currently, no therapies are approved for CD79B aberrations. Polatuzumab vedotin is a CD79B targeting antibody-drug conjugate with a payload of microtubule inhibitor monomethyl auristatin E (MMAE) approved by the FDA in 2019 for the treatment of releapsed/refractory DLBCL in combination with bendamustine and rituximab[14].

Genes Assayed

Genes Assayed for the Detection of DNA Sequence Variants

BRAF, BTK, CARD11, GNA13, HIST1H1E, MTOR, SF3B1, XPO1

Genes Assayed for the Detection of Copy Number Variations

BRAF, CARD11, GNA13, HIST1H1E, MTOR, SF3B1, XPO1

Genes Assayed with Full Exon Coverage

ARID1A, ATM, B2M, BCL2, BCL6, CD79B, CDKN2A, CREBBP, EZH2, KMT2D, MYC, MYD88, PIM1, SOCS1, TNFAIP3, TNFRSF14, TP53

Thermo Fisher Scientific's Ion Torrent Oncomine Reporter software was used in generation of this report. Software was developed and designed internally by Thermo Fisher Scientific. The analysis was based on Oncomine Reporter (6.0.2 data version 2025.04(004)). The data presented here are from a curated knowledge base of publicly available information, but may not be exhaustive. FDA information was sourced from www.fda.gov and is current as of 2025-03-19. NCCN information was sourced from www.nccn.org and is current as of 2025-03-03. EMA information was sourced from www.ema.europa.eu and is current as of 2025-03-19. ESMO information was sourced from www.esmo.org and is current as of 2025-03-03. Clinical Trials information is current as of 2025-03-03. For the most up-to-date information regarding a particular trial, search www.clinicaltrials.gov by NCT ID or search local clinical trials authority website by local identifier listed in 'Other identifiers.' Variants are reported according to HGVS nomenclature and classified following AMP/ASCO/CAP guidelines (Li et al. 2017). Based on the data sources selected, variants, therapies, and trials listed in this report are listed in order of potential clinical significance but not for predicted efficacy of the therapies.

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. Wu et al. ARID1A mutations in cancer: another epigenetic tumor suppressor?. Cancer Discov. 2013 Jan;3(1):35-43. PMID: 23208470
3. Wilson et al. SWI/SNF nucleosome remodellers and cancer. Nat. Rev. Cancer. 2011 Jun 9;11(7):481-92. PMID: 21654818
4. Alver et al. The SWI/SNF Chromatin Remodelling Complex Is Required for Maintenance of Lineage Specific Enhancers. Nat Commun. 8;14648. PMID: 28262751
5. Weinstein et al. The Cancer Genome Atlas Pan-Cancer analysis project. Nat. Genet. 2013 Oct;45(10):1113-20. PMID: 24071849
6. 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
7. Mehrvarz et al. ARID1A Mutation May Define an Immunologically Active Subgroup in Patients with Microsatellite Stable Colorectal Cancer. Clin Cancer Res. 2021 Mar 15;27(6):1663-1670. PMID: 33414133
8. https://nuvectis.com/press-release-view/?i=114174
9. https://www.morphosys.com/en/news/morphosys-receives-us-fda-fast-track-designation-tulmimetostat-endometrial-cancer
10. Chu et al. CD79: a review. Appl Immunohistochem Mol Morphol. 2001 Jun;9(2):97-106. PMID: 11396639
11. Astsaturov et al. Differential expression of B29 (CD79b) and mb-1 (CD79a) proteins in acute lymphoblastic leukaemia. Leukemia. 1996 May;10(5):769-73. PMID: 8656670
12. Akerblad et al. The B29 (immunoglobulin beta-chain) gene is a genetic target for early B-cell factor. Mol Cell Biol. 1999 Jan;19(1):392-401. PMID: 9858563
13. Nakamura et al. Recurrent mutations of CD79B and MYD88 are the hallmark of primary central nervous system lymphomas. Neuropathol Appl Neurobiol. 2016 Apr;42(3):279-90. PMID: 26111727
14. Bourbon et al. Polatuzumab vedotin: an investigational anti-CD79b antibody drug conjugate for the treatment of diffuse large B-cell lymphoma. Expert Opin Investig Drugs. 2020 Oct;29(10):1079-1088. PMID: 32700972