

# Genetic and clinical characteristics of 54 pediatric lymphoma patients with variant mutation sites of *UNC13D*

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<https://doi.org/10.3324/haematol.2025.287782>

Received: March 17, 2025.

Accepted: February 20, 2026.

Early view: March 5, 2026.

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**Suppl.Table 1** Clinical characteristics and Variant details of patients

Patient	Age of onset	Mutant Allele 1	Mutant Allele 2	Mutation type	Gene region	REVEL prediction	ACMG interpretation	Clinical diagnosis	Severe features	Complication with HLH	Stage	Regimen of therapy	Outcome
P1	8y	c.2588G>A (p.G863D)	c.2588G>A (p.G863D)	hom	exonic/exonic	D	Uncertain/ Uncertain	T- LBL	Y	Y	III	modified BFM-95 regimen	alive
P2	1y	c.118-308C>T (splicing)	c.118-308C>T (splicing)	hom	UTR/UTR	D	Pathogenic/ Pathogenic	DLBCL;	Y	Y	IV	CNCL-B-NHL-2017 regimen; HLH-04 protocol	dead
P3	8y	c.2553+5C>G (splicing)	c.2553+5C>G (splicing)	hom	intron/intron	-	Uncertain/ Uncertain	cHL	N	N	II	BV+AVD+CD20	alive
		c.2440G>A (p.S747N)	c.3134C>T (p.T1045M)	Compound het	exonic/exonic	LB/LB	Uncertain/ Uncertain						
P4	8y	c.1228A>C (p.I410L)	c.11743-C>G (splicing)	Compound het	exonic/intron	LD/-	Uncertain/ Uncertain	cHL	Y	Y	III	BV+AVD+CD20; HLH-04 protocol	dead
P5	6y	c.1786T>C (p.T596A)	c.1596+1GC (splicing)	Compound het	exonic/intron	D/T	Likely Pathogenic /Pathogenic	cHL;progression	Y	Y	III	BV+AVD+CD20; HLH-04 protocol	alive
P6	7y	c.3131T>G (p.L1044R)	c.2588G>A (p.G863D)	Compound het	exonic/exonic	LD/D	Uncertain/ Uncertain	cHL	N	N	II	BV+AVD+CD20	alive
P7	3y	c.3134C>T (p.T1045M)	c.2553+5C>G (splicing)	Compound het	exonic/intron	F	Uncertain/ Uncertain	cHL	N	N	II	BV+AVD+CD20	alive
P8	12y	c.2270G>A (p.R757H)	c.888G>A (p.P296P)	Compound het	exonic/exonic	U/U	Uncertain/ Uncertain	BL-11q	N	N	II	CNCL-B-NHL-2017 regimen	alive
P9	4y	c.3270G>A (p.P1090P)	c.754-8C>T (splicing)	Compound het	exonic/intron	D/-	Uncertain/ Uncertain	DLBCL; Glioblastoma	Y	Y	III	CNCL-B-NHL-2017 regimen	alive
P10	14y	c.683+1G>T	c.544C>A	Compound	intron/exonic	T/D	Likely	cHL; XMEN	N	N	II	BV+AVD+CD20	alive

		(splicing)	(p.P182T)	het			Pathogenic/ Uncertain							
P11	14y	c.1607G>T (p.R536L)	-	het	exonic/-	LD	Likely Pathogenic	cHL	N	N	III	BV+AVD+CD20	alive	
P12	13y	c.1607G>T (p.R536L)	-	het	exonic/-	LD	Likely Pathogenic	cHL	N	N	II	BV+AVD+CD20	alive	
P13	12y	c.3067C>T (p.R1023C)	-	het	exonic/-	B	Uncertain	cHL	N	N	IV	BV+AVD+CD20	alive	
P14	9y	c.3067C>T (p.R1023C)	-	het	exonic/-	B	Uncertain	cHL	N	N	IV	BV+AVD+CD20	alive	
P15	5y	c.3229_3235del (p.R1077Sfs*48)	-	het	exonic/-	-	Likely Pathogenic	cHL; CAEBV; recurrence	Y	Y	III	BV+AVD+CD20; HLH-04 protocol	alive	
P16	13y	c.3229_3235del (p.R1077Sfs*48)	-	het	exonic/-	-	Likely Pathogenic	DLBCL; XMEN; recurrence	Y	Y	IV	CNCL-B-NHL-2017 regimen; HLH-04 protocol	HSCT, alive	
P17	7y	c.3229_3235del (p.R1077Sfs*48)	-	het	exonic/-	-	Likely Pathogenic	LBCL	Y	Y	III	CNCL-B-NHL-2017 regimen	alive	
P18	10y	c.2588G>A (p.G863D)	-	het	exonic/-	D	Uncertain	cHL	N	N	III	BV+AVD+CD20	alive	
P19	9y	c.2588G>A (p.G863D)	-	het	exonic/-	D	Uncertain	BL-11q	N	N	II	CNCL-B-NHL-2017 regimen	alive	
P20	9y	c.2588G>A (p.G863D)	-	het	exonic/-	LD	Likely Pathogenic	ENKTL	N	N	III	CNCL-B-NHL-2017 regimen	alive	
P21	11y	c.2588G>A (p.G863D)	-	het	exonic/-	D	Uncertain	SPTCL	N	N	II	ALL-like regimen	alive	
P22	10y	c.2588G>A	-	het	exonic/-	D	Uncertain	BL	N	N	I	CNCL-B-NHL-2017	alive	

		(p.G863D)											regimen		
P23	11y	c.2588G>A	-	het	exonic/-	D	Uncertain	T-LBL	N	N	III	modified	BFM-95	alive	
		(p.G863D)											regimen		
P24	12y	c.1228A>C	-	het	exonic/-	LD	Uncertain	BL-11q	N	N	II	CNCL-B-NHL-2017		alive	
		(p.I410L)											regimen		
P25	4y	c.1228A>C	-	het	exonic/-	LD	Uncertain	B-LBL	N	N	IV	modified	BFM-95	alive	
		(p.I410L)											regimen		
P26	10y	c.1228A>C	-	het	exonic/-	LD	Uncertain	SPTCL	N	N	I	ALL-like regimen		alive	
		(p.I410L)													
P27	13y	c.1228A>C	-	het	exonic/-	LD	Uncertain	B-LBL	N	N	III	modified	BFM-95	alive	
		(p.I410L)											regimen		
P28	11y	c.1228A>C	-	het	exonic/-	LD	Uncertain	BL	N	N	IV	CNCL-B-NHL-2017		alive	
		(p.I410L)											regimen		
P29	5y	c.1228A>C	-	het	exonic/-	LD	Uncertain	BL	N	N	IV	CNCL-B-NHL-2017		alive	
		(p.I410L)											regimen		
P30	2y	c.1228A>C	-	het	exonic/-	LD	Uncertain	B-LBL	N	N	IV	modified	BFM-95	alive	
		(p.I410L)											regimen		
P31	10y	c.1228A>C	-	het	exonic/-	LD	Uncertain	PMBCL	N	N	III	CNCL-B-NHL-2017		alive	
		(p.I410L)											regimen		
P32	7y	c.1228A>C	-	het	exonic/-	LD	Uncertain	BL	N	N	III	CNCL-B-NHL-2017		alive	
		(p.I410L)											regimen		
P33	14y	c.1228A>C	-	het	exonic/-	LD	Uncertain	ALK+ ALCL	N	N	II	CNCL-B-NHL-2017		alive	
		(p.I410L)											regimen		
P34	12y	c.1228A>C	-	het	exonic/-	LD	Uncertain	ALK+ ALCL	N	N	III	CNCL-B-NHL-2017		alive	
		(p.I410L)											regimen		
P35	12y	c.2553+5C>G	-	het	intron/-	D	Uncertain	ALK+ALCL;	Y	N	IV	CNCL-B-NHL-2017		alive	

P36	12y	(splicing) c.2553+5C>G	-	het	intron/-	D	Uncertain	DLBCL	Y	N	III	CNCL-B-NHL-2017	alive	
P37	9y	(splicing) c.-28C>T	-	het	unknown	D	Uncertain	LBCL	N	N	I	CNCL-B-NHL-2017	alive	
P38	14y	c.594G>A (p.A198A)	-	het	exonic/-	LD	Uncertain	T-LBL; refractory	N	N	IV	modified regimen	BFM-95	HSCT, alive
P39	4y	c.1818G>A (p.A606A)	-	het	exonic/-	LB	Uncertain	B-LBL	N	N	III	modified regimen	BFM-95	alive
P40	15y	c.2499C>T (p.A833A)	-	het	exonic/-	LD	Uncertain	ALK+ ALCL	N	N	III	CNCL-B-NHL-2017	alive	
P41	2y	c.2499C>T (p.A833A)	-	het	exonic/-	LD	Uncertain	DLBCL	N	N	IV	CNCL-B-NHL-2017	alive	
P42	7y	c.2499C>T (p.A833A)	-	het	exonic /-	LD	Uncertain	B-LBL	N	N	IV	modified regimen	BFM-95	alive
P43	10y	c.1157G>T (p.R386L)	-	het	exonic/-	LB	Uncertain	T-LBL;	Y	N	IV	modified regimen	BFM-95	alive
P44	8y	c.754-8C>T (splicing)	-	het	intron/-	D	Uncertain	ALK+ ALCL; refractory	N	Y	III	CNCL-B-NHL-2017	HSCT, alive	
P45	4y	c.2292C>T (p.A764A)	-	het	exonic/-	LD	Uncertain	BL	N	N	III	CNCL-B-NHL-2017	alive	
P46	6y	c.986C>T (p.P329L)	-	het	exonic/-	LB	Uncertain	BL	N	N	III	CNCL-B-NHL-2017	alive	
P47	5y	c.1188C>T (p.A396A)	-	het	exonic/-	D	Uncertain	ALK+ ALCL	N	N	III	CNCL-B-NHL-2017	alive	
P48	12y	c.2470C>T	-	het	exonic/-	U	Uncertain	T-LBL	N	N	IV	modified	BFM-95	alive

P49	9y	(p.H824Y) c.2892C>T	-	het	exonic/-	LB	Uncertain	T-LBL	N	N	III	modified regimen	BFM-95	alive
P50	7y	(p.A964A) c.261+8C>T	-	het	intron/-	LD	Uncertain	DLBCL	N	N	II	CNCL-B-NHL-2017 regimen		alive
P51	10y	(splicing) c.205C>T	-	het	exonic/-	B	Uncertain	T-LBL	Y	N	III	modified regimen	BFM-95	alive
P52	7y	(p.H69Y) c.2214G>A	-	het	exonic/-	LD	Uncertain	cHL	N	N	IV	BCH-2003 regimen		alive
P53	8y	(p.Q738Q) c.1280G>A	-	het	exonic/-	D	Uncertain	ALK+ ALCL	N	N	IV	CNCL-B-NHL-2017 regimen		alive
P54	12y	(p.R427Q) c.3142G>A	-	het	exonic/-	LD	Uncertain	BL	N	N	III	CNCL-B-NHL-2017 regimen		alive
		(p.A1048T)												

Notes:

compound het: compound heterozygous mutations; hom: homozygous mutations; het: heterozygous mutations; Y: Yes; N: No; REVEL: Rare Exome Variant Ensemble Learner(D:Damaging; LD:Likely Damaging; B:Benign; LB: Likely Benign; U:Uncertain; T:Splice donor variant; F: No impact on splicing); ACMG: American College of Medical Genetics and Genomics; ALCL: Anaplastic Large Cell Lymphoma; T/B-LBL: T/B-cell acute lymphoblastic leukemia; BL: Burkitt Leukemia; DLBCL: Large B-cell lymphoma; SPTCL: Subcutaneous panniculitis-like T-cell lymphoma; LBCL: Large B cell lymphoma with interferon regulatory factor 4(*IRF4*) gene rearrangement; MBCL: mature B-cell lymphoma with chromosome 11 long-arm abnormalities; ENKTL: extranodal NK/T-cell lymphoma of nasal type; PMBCL: Primary Mediastinum Large B cell Lymphoma; BFM-95 regimen: modified Berlin-Frankfurt-Munster-95 regimen; CNCL-B-NHL-2017 regimen: China Net Childhood Lymphoma-mature B-cell lymphoma 2017 regimen; BCH-2003 regimen: Beijing Children's Hospital-2003 regimen. , HLH-04 Hemophagocytic lymphohistiocytosis-04 protocol; CD20 refers to anti-CD20 antibody therapy.

