

Deficiency	CD3δ																		
Family	1			2	3	4				5		6	7			8	9	10	
Ethnic background	Mennonites (Canada, USA, Germany)									France			Japan			Ecuador			
Patient/sex	1F	2M	3M	4F	5M	6F	7M	8F	9M	10F	11F	12F	13M	14F	15M	16M	17M	18M	19F
Consanguineous	Yes									Yes			Yes			No			
Mutation	c.202C>T									c.279C>A			c.275-2A>G			c.27415G>A			
Predicted effect (<i>leaky</i>) ⁷	Truncation									Truncation			Skip3			Skip2			
Diagnosis at (m) ¹	0	2	2	9	0	0	0	10	13	0	3	0	2	3	0	0	14	4	10
% CD3 ⁺ cells ^{2,6}	0.3	0.1	0.6	2	0	0	1	0	27	1	<1		0	1.7	0.2	0	14	30	2
HSC ³ at (m,y) ¹	MB4m	No	No	MB16m	MB4m	IDB1m	HP6m	HP12m	HB14m	HB2m	No	H	H	HP4m	MIC1m	MC1m	IDP23m	HP8m	No
Cause of death ⁴	AW	ADV	CMV	AW	AW	AW	GvHD	HHV6	CMV	AW	CMV	Asp	EBV	CMV	AW	AW	AW	CMV	CMV

Deficiency	CD3γ							CD3ε					ζ			TCRα			
Family	1		2	3		4		1	2		3	4	5	1	2	3	1	2	
Ethnic background	Turkey		Turkey	Spain		Turkey		France	France		Unknown	Turkey		Caribbean	Hawaii	Turkey	Pakistan		
Patient/sex	1M	2M	3M	4M	5M	6F	7F	1M	2F	3M	4F	5F	6F	7M	1M	2F	3F	1F	2M
Consanguineous	Yes		No	No		No		No	Yes		No	Yes		NR ⁵	No	Yes	Yes		
Mutation	c.205A>T		c.1A>G & c.IVS2-1G>C	c.IVS2-1G>C		c.IVS7+2T>C & c.230G>A		c.128_129del		c.IVS2+1G>C	c.173delT	c.207C	c.411insC	c.2T>C	c.*1G>A				
Predicted effect (<i>leaky</i>) ⁷	Truncation		Truncation, Het	Truncation		Skip7, Tr, Het		Truncation		Skip 2	Truncation	Trunc	Insertion	Trunc	Skip3				
Diagnosis at (m) ¹	3	7	48	12	48	144	240	48	5	1	0	2	3	2	4	10	2	15	6
% CD3 ⁺ cells ^{2,6}	37	27	30	10	18	40	45	63	NR ⁵	NR	<1	100 ⁸	0	0	21	64	60	21	50
HSC ³ at (m,y) ¹	No	ID	No	No	No	No	No	No	No	No	H	H7m	No	H>3m	H30m	H>12m	H18m	IDB6y	IDB7y
Cause of death ⁴	Sepsis	Pneum	AW	Pneum	AW	AW	AW	AW	Pneum	CMV	ADV	AW	CMV	AW	AW	AW	RF	AW	AW

1- m (months); y (years).

2- At diagnosis. Normal values 60-85%, includes dull and bright CD3⁺ cells.

3- First hematopoietic stem cell transplantation from related (matched = ID, haploidentical = H) or unrelated (matched = M, mismatched = MI) donor bone marrow (B), peripheral blood (P) or cord blood (C).

4- AW (Alive and Well); A (Alive); ADV, CMV, EBV, HHV (viral infection); Pneum (Pneumonia); Asp (Aspergillus); GvHD (Graft vs Host Disease); RF (respiratory failure)

5- NR (Not reported)

6- For TCRα defects mostly TCRgd⁺ T cells, few TCRab^{low} T cells

7- Leaky refers to wild type protein levels. Skip7 (exon 7 skipping), Tr/Trunc (protein truncation), Het (compound heterozygote)

8- T cells from maternal origin

[For more information read "Human congenital T-cell receptor disorders"](#)