

Deficiency	CD3 γ											CD3 ϵ							CD247																				
Family	1		2	3		4		5		6	7	1		2		3	4	5	6	7	1	2	3	4	5														
Ethnic background	Turkey		Turkey	Spain		Turkey		Turkey		Turkey	Taiwan	France		France		Unknown	Turkey	India	India	India	Caribbean	Hawaii	Turkey	India	Turkey														
Patient/sex	1M	2M	3M	4M	5M	6F	7F	8F	9M	10F	11 / NR	12M	1M	2F	3M	4F	5F	6M	7F	8M	9F	1M	2F	3F	4F	5F													
Consanguineous	Yes			No			No			Yes			NR			No			Yes			Yes			Yes														
Mutation	c.205A>T			c.1A>G & c.80-1 G>C			c.80(-1) G>C		c.80(-1) G>C			c.1A>G		c.213 del A		c.520+2T>C , c.176G>A		c.128_129del		c.49+1G>C		c.176G>A		c.46T>C		c.288T>A		c.352+1G>A		c.207C		c.411insC		c.2T>C		c.62C>T		c.43_44delCA	
Predicted effect (<i>leaky</i>) ⁷	p.K69X			p.M1V, p.N28VfsX1			p.N28VfsX1		p.N28VfsX1			p.M1V		p.K71Nfs39X		p.P34_R117del, p.W59X		p.T43fsX56		p.0		p.W59X		p.S16P		p.Y96X		Trunc		p.Q70X		p.D138fsX272		p.M1T		p.A21V		p.Q15VfsTer72	
Diagnosis at (m) ¹	3	7	48	12	48	144	240	84	132	14	15	432	48	5	1	0	2	4	6	8	7	4	10	2	48	4													
% CD3 ⁺ cells ^{2,6}	37	27	30	10	18	40	45	33	32	45	40	78	63	NR ⁵	NR	<1	80 ⁸	0	0.3	0	0.2	21	64	60	2	27													
HSCT ³ at (m,y) ¹	No	ID	No	No	No	No	No	No	No	No	No	No	No	No	No	H	H7m	H	No	NR	NR	NR	H30m	H>12m	No	No	No												
Cause of death ⁴	Sepsis	Pneum	AW	Pneum	AW	AW	AW	AW	AW	AW	AW	AW	AW	Pneum	CMV	ADV	AW	AW	Pneum	NR	NR	AW	AW	Pneum	Pneum	AW													

Deficiency	CD3 δ																				TCR α															
Family	1				2	3	4				5				6				7				8	9		10	11	1	2	3						
Ethnic background	Mennonites (Canada, USA, Germany)										France										Japan				Ecuador		India		India		Pakistan		India			
Patient/sex	1F	2M	3M	4F	5M	6F	7M	8F	9M	10F	11F	12F	13M		14F	15M	16M	17M	18M	19F	20M		1F	2M	3M	5F	6F									
Consanguineous	Yes										Yes										Yes				No		NR		NR		Yes		No			
Mutation (<i>leaky</i>) ⁷	c.202C>T										c.279C>A										c.275-2A>G				c.411+5G>A		c.158C>A		c.275-2A>G		c.*1G>A					
Predicted effect	p.R68X										p.C93X										p.M92_A136del				p.V13_M92del		p.S53X		p.M92_A136del		p.T107LfsX56					
Diagnosis at (m) ¹	0	2	2	9	0	0	0	10	13	0	3	0	2		3	0	0	14	4	5	6	15	6	3	6	2										
% CD3 ⁺ cells ²	0.3	0.1	0.6	2	0	0	1	0	27	1	<1	<1	0		1.7	0.2	0	14	30	1.2	66	21	50	45	43	52										
HSCT ³ at (m,y) ¹	MB4m	No	No	MB16m	MB4m	IDB1m	HP6m	HP12m	HB14m	HB2m	No	H	H		HP4m	MIC1m	MC1m	IDP23m	HP8m	H9D	No	IDB6y	IDB7y	No	No	No										
Cause of death ⁴	AW	ADV	CMV	AW	AW	AW	GvHD	HHV6	CMV	AW	CMV	Asp	EBV		CMV	AW	AW	AW	CMV	LH	NR	AW	AW	Pneum	Pneum	EBV										

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); ADV,CMV,EBV,HHV (viral infection); Pneum (Pneumonia); Asp (Aspergillus); GvHD (Graft vs Host Disease); LH (Lung hemorrhage)

5- NR (Not reported)

6- For TCR α defects mostly TCR $\gamma\delta$ + T cells, few TCR $\alpha\beta$ T cells

7- Leaky refers to wild type protein levels. Trunc (protein truncation)

8- T cells from maternal origin