

Table E1: Clinical and genotypic features of DOCK8-deficient subjects.

Patients	Age (year)	Clinical findings		Viral infections	Genotyping (Reference Sequence NM_203447.3)	References
		Gender				
1*	5	F		HSV	Homozygous IVS16 splice acceptor site mutation: c.[1869 -1 G>C]; [1869 -1 G>C]	This report
2*	4	F		HSV	Homozygous deletion encompassing exons 26-48: c.[3121 -?_7340 +?del]; [3121 -?_7340 +?del]	This report
3	1	F			Homozygous deletion encompassing exons 1-23: c.[1 -?_2874 +?del]; [1 -?_2874 +?del]	This report
4	9	F		HPV	Homozygous deletion encompassing exons 26-48: c.[3121 -?_7340 +?del]; [3121 -?_7340 +?del]	This report
5	4	M		MCV	Homozygous deletion encompassing exon 27: c.[3235 -?_3390+?del]; [3235 -?_3390 +?del]	(Reference 5)
6	6	M		MCV	Homozygous deletion encompassing exons 1 and 2: c.[1 -?_156 +?del]; [1 -?_156 +?del]	(Reference 5)
7	8	F		HSV, HPV, MCV	Homozygous deletion encompassing exons 28-35: c.[3391 -?_4473 +?del]; [3391 -?_4473 +?del]	(Reference 5)
8	10	F		HSV, VZV	Homozygous nonsense mutation: c.[2248G>T]; [2248G>T]	(Reference 2)
9	7	M			Homozygous IVS44 splice donor site mutation: c.[5817 +1 G>A]; [5817 +1 G>A]	(Reference E2)
10†	8	M		HSV	Homozygous IVS44 splice donor site mutation: c.[5817+1 G>A]; [5817 +1 G>A]	This report

*Patients treated with IFN- α 2b

†Deceased

Table E2: Hematological and Immunological findings in DOCK8-deficient patients treated with IFN- α 2b.

Peripheral Blood Cells	Pt1 at diagnosis	Pt1 before IFN- α 2b Therapy	Pt1 2 months into IFN- α 2b Therapy	Pt2 at diagnosis	Pt2 before IFN- α 2b Therapy	Pt2 2 months into IFN- α 2b Therapy
White Blood Cells (mm ³)	11500	15400	10700	17100	25000	11900
Neutrophils (mm ³)	6040	12800	8999	6480	9500	8150
Lymphocytes (mm ³)	2290	1810	1490	2400	1999	1680
Eosinophils (mm ³)	3500	872	7	8000	806	1100
Hemoglobin (g/dl)	9.9	11.3	9.79	10.1	13.1	13.8
Platelets (mm ³)	719,000	435,000	847,000	825,000	374,000	392,000
Serum Immunoglobulins						
IgE (IU/ml)	334	1330		14000	2450	
IgA (mg/dl)	94	94.1		176	199	
IgM (mg/dl)	36.5	63.1		84.5	60.9	
IgG (mg/dl)	1280	994		2100	1230	
Lymphocyte subset analysis						
CD3 ⁺ (%)	40	54	54	58	51	53
CD3 ⁺ 4 ⁺ (%)	29	24	28	27	28	33
CD3 ⁺ 8 ⁺ (%)	12	34	33	30	24	19
CD19 ⁺ (%)	47	19	20	15	22	24
CD3 ⁻ CD16 ⁺ CD56 ⁺ (%)	10	24	18	27	24	20