Table S2. List of homozygous rare nonsynonymous variations in the two pairs of patients from kindred A and B, Related to Figure 1.

Patients	Chr	Pos	ID	Ref	Alt	Type	Gene	AAChange	1000Genomes_Freq	ExAC_Freq	CADD_Phred	CADD_MSC	GDI
Common to P1 and P2	3	137890519		A	G	Missense	DBR1	p.Ile120Thr			27.3	4.75	2.47
	3	142402975	rs148154571	T	C	Missense	PLS1	p.Val236Ala	0.0026	0.0028	23.4	1.25	4.39
	11	6411935	rs558809956	TGCTGGC	T	Indel-in-frame	SMPD1	p.Leu47_Ala48del			12.7	2.9	7.42
	11	125830970	rs684535	A	T	Missense	CDON	p.Ile1244Asn			1.592	9.05	8.65
Common to P5 and P6	3	42251577		CGGA	C	Indel-in-frame	TRAK1	p.Glu640del			16.05	2.31	3.44
	3	46751073	rs71619660	TAAG	T	Indel-in-frame	TMIE	p.Lys131del			13.71	25.6	0.96
	3	113376110	rs59601191	TTGCTGCTGC	T	Indel-in-frame	KIAA2018	p.Gln1471_Gln1473del			19.01	2.31	8.22
	3	136047691	rs142403318	C	T	Missense	PCCB	p.Ala497Val	0.001	0.002	34	0.06	2.61
	3	137893589		A	G	Missense	DBR1	p.Tyr17His			25.4	4.75	2.47
	3	184429133	rs397720350	A	ATCC	Indel-in-frame	MAGEF1	p.Glu158dup			4.953	2.31	9.13
	4	140811085	rs58287721	TGCTGCTGCTGC	T	Indel-frameshift	MAML3	p.Gln498fs	0.0092		28.1	2.31	6.56
	4	143003205		C	T	Missense	INPP4B	p.Arg874Gln		0	25.6	2.31	2.17
	4	147560457	rs5862765	T	TGGC	Indel-in-frame	POU4F2	p.Gly67dup			2.45	5.6	6.04
	4	154477046	rs139597924	C	T	Missense	KIAA0922	p.Pro139Leu	0.0012	0.0011	26.2	2.31	8.61
	4	164246489	rs5578	T	G	Missense	NPY1R	p.Lys374Thr	0.003	0.0047	15.51	4.8	0.94
	4	164394697	rs551489665	GGTCT	G	Indel-frameshift	TKTL2	p.Thr62fs	0.0016	0.0022	23.4	2.31	4.39
	6	16327864		G	GTGC	Indel-in-frame	ATXN1	p.Gln225dup			0.013	2.31	3.77
	6	161519350	rs5881391	CCTG	C	Indel-in-frame	MAP3K4	p.Ala1199del			18.68	2.31	4.58
	7	131241029	rs532078953	GGGCGAC	G	Indel-in-frame	PODXL	p.Ser29_Pro30del			7.336	2.31	3.35
	12	76424937	rs71716769	TTGC	T	Indel-in-frame	PHLDA1	p.Gln195del	0.0094		0.372	4.75	5.29
	16	58577315	rs5817153	GA	G	Indel-frameshift	CNOT1	p.Phe1543fs			0.483	2.31	2.12
	17	17039561	rs3833098	CCAG	C	Indel-in-frame	MPRIP	p.Ser189del			17.36	3.14	
	17	17697101	rs34083643	AG	A	Indel-frameshift	RAI1	p.Gln280fs			23.1	0	3.23
	18	30352057	rs200605696	GCGCCGGCC	G	Indel-frameshift	AC012123.1	p.Pro125fs			25.5	6.71	2.13
	19	30500118	rs556897562	GTGA	G	Indel-in-frame	URI1	p.Asp307del			10.73	2.31	2.92

Note: All nonsynonymous or essential-splicing variants revealed by whole-exome sequencing, with a MAF<0.01 in ExAC, in genes with a gene damage index (GDI) below 13.35564 (95% true positive confidence interval GDI cutoff for autosomal recessive mode primary immune deficiency gene prediction), either common to P1 and P2 from kindred A, or common to P5 and P6 in kindred B, are listed in this table.