Supplementary Table 1. Data represents the mean of the NGS Quality Control Metrics of all runs.

Miseq platform (3 samples per run)

Number of runs = 20			
Cluster PF (%)	**Reads PF**	**Density (K/mm^2)**	**%≥Q30**
88.58	23,675,292	1,183	89.68

Nextseq platform (36 samples per run)

Number of runs = 2			
Cluster PF (%)	**Reads PF**	**Density (K/mm^2)**	**%≥Q30**
86.53	96,727,198	151	80.55

Supplementary Table 2. Individual sample general sequencing data.

Patient	Aligned Reads (%)	Bases Q-score>30 (%)	Target Coverage at 20X (%)	Uniformity of Coverage (%)	Mean Region Coverage Depth (X)
P1	95.3	91.3	87.4	90.8	81.1
P2	99.8	94.9	91.7	95.5	64.1
P3	53	51.7	85.3	95.8	53.8
P4	98.4	92.8	89.7	93.2	78.6
P5	94.9	87.9	77.2	88.6	62.9
P6	95.8	94.7	88.9	91.9	81.9
P7	88.7	93.4	83.9	88.9	75.5
P8	83.8	94.7	92.1	96.3	67.2
P9	89.1	90.2	80.1	87	72.5
P10	95.6	94.8	85.6	91.7	71.1
P11	95.8	95.4	92	94.1	80.9
P12	99.1	86.4	97.4	96.7	111.8
P13	98.6	94	85	91	70
P14	90.1	93.2	79.8	94.4	48.4
P15	99.7	85	97.1	96.4	110.8
P16	98	94.5	78.1	91.1	55.2
P17	95.3	95.6	71.6	95.1	38.4
P18	86.6	89.1	93.3	95.8	78.7
P19	96.5	95	77.4	95.6	44.8
P20	98.9	96.8	84.9	95.4	51.9
P21	95.7	88.3	88.8	90.9	85.8
P22	87	95.2	93.9	96.1	77.5
P23	87.1	91	94.2	94.8	93.4
P24	99.7	83	97.2	96.6	105.5
P25	98.8	95.2	93	95.5	78.3
P26	98.4	95.5	93.6	95.9	79.5
P27	98	94.6	64.8	91.4	38.7
P28	86.7	93.8	86.9	94.3	62.2
P29	96.5	95	90	93.5	77.1
P30	86.4	92.4	94.8	95.4	92.2
P31	98.6	93.4	95.4	94.9	100.6
P32	99	96.7	92.4	95.1	78.1
P33	94.9	95.7	83.2	94.7	53
P34	95.5	94.5	54.9	88.2	32
P35	95.5	88.8	85	90.2	75.3
P36	52.7	51.9	90.1	94.7	72

P37	88.1	91.5	88.6	92.5	77.8
P38	86.5	88.7	97.1	96.5	107.5
P39	87	88.9	96.6	95.9	106.9
P40	86.4	90.8	69.9	82.3	61
P41	85.2	88.7	92.6	96	70.3
P42	86.2	88.9	90.2	95.5	69.7
P43	81.9	87.7	97.4	97.1	105.2
P44	85	89.2	95.4	97.4	75
P45	83.4	90.1	96.3	97	87.8
P46	99.6	77.1	96.8	97.1	90.2
P47	99.6	82.4	89.8	91.6	89.1
P48	99.7	82.6	95.3	96.1	86.1
P49	99.5	87.9	97.6	97.1	108.7
P50	99.7	84.8	94.2	96.2	77.6
P51	99.4	89.4	92	85.2	184.3
P52	99.6	77.7	96.4	96.4	97.4
P53	99.3	85.6	96.6	96.1	106.6
P54	99.7	84.2	97.3	96.4	119.4
P55	99.6	82.4	88.2	86.4	107.7
P56	99.7	83.9	93.9	95.6	83.1
P57	99.7	82.9	93.9	93.4	102.5
P58	99.9	93.6	84.8	90	62
P59	99.4	89.6	92	84.9	193.9
P60	99.9	94.2	89.4	95.7	52.1
P61	95.2	90.9	76.2	86.1	65.3
Mean	**93.2**	**89.0**	**88.8**	**93.5**	**81.8**
SD	**9.4**	**8.4**	**8.6**	**3.6**	**28.4**
Max	**99.9**	**96.8**	**97.6**	**97.4**	**193.9**
Min	**52.7**	**51.7**	**54.9**	**82.3**	**32**

Supplementary Table 3. Genes included in the virtual primary immunodeficiencies panel.

ACP5	CARD14	CFP	FOXP3	IL7R	NCF2	RAB27A	STAT5B	TNFSF11
ACTB	CARD9	CFTR	FPR1	IRAK4	NCF4	RAC2	STIM1	TPP1
ADA	CASP10	CHD7	G6PC3	IRF7	NCSTN	RAG1	STK4	TPP2
ADAM17	CASP8	CIITA	G6PD	IRF8	NFKB1	RAG2	STX11	TRAC
AICDA	CCBE1	CLCN7	GATA2	ITCH	NFKBIA	RANBP2	STXBP2	TRAF3
AIRE	CD19	CORO1A	GFI1	ITGB2	NHEJ1	RFX5	TAP1	TRAF3IP2
AK2	CD247	CR2	HAX1	ITK	NHP2	RFXANK	TAP2	TREX1
AP3B1	CD27	CSF2RA	HMOX1	JAK3	NLRP12	RFXAP	TAPBP	TTC37
APOL1	CD3D	CSF2RB	ICOS	KDM6A	NLRP3	RMRP	TAZ	TYK2
ATM	CD3E	CSF3R	IFIH1	KMT2D	NOD2	RNASEH2A	TBK1	UNC13D
B2M	CD3G	CTC1	IFNAR2	KRAS	NOP10	RNASEH2B	TBX1	UNC93B1
BCL10	CD40	CTLA4	IFNGR1	LAMTOR2	NRAS	RNASEH2C	TCIRG1	UNG
BLM	CD40LG	CTSC	IFNGR2	LIG1	ORAI1	RNF168	TCN2	USB1
BLNK	CD46	CXCR4	IGHM	LIG4	OSTM1	RNU4ATAC	TERC	VPS13B
BTK	CD55	CYBA	IGLL1	LPIN2	PEPD	SAMD9	TERT	WAS
C1QA	CD59	CYBB	IKBKG	LYST	PIK3CD	SAMHD1	TFRC	WIPF1
C1QB	CD79A	DCLRE1C	IL10	MAGT1	PIK3R1	SBDS	THBD	WRAP53
C1QC	CD79B	DDX58	IL10RA	MASP2	PLCG2	SEMA3E	TICAM1	XIAP
C1R	CD81	DKC1	IL10RB	MCM4	PLEKHM1	SERPING1	TINF2	ZAP70
C1S	CD8A	DNMT3B	IL12B	MEFV	PMS2	SH2D1A	TIRAP	ZBTB24
C2	CEBPE	DOCK8	IL12RB1	MKL1	PNP	SH3BP2	TLR3	
C3	CFB	ELANE	IL17F	NFKB1	POLE2	SLC29A3	TMC6	
C4A	CFD	EXTL3	IL17RA	MOGS	PRF1	SLC35C1	TMC8	
C4B	CFH	FADD	IL1RN	MS4A1	PRKDC	SLC46A1	TMEM173	
C5	CFHR1	FASLG	IL21	MSH6	PSEN1	SMARCAL1	TNFAIP3	
C6	CFHR2	FCGR3A	IL21R	MTHFD1	PSENEN	SNX10	TNFRSF11A	
C7	CFHR3	FCN3	IL2RA	MVK	PSMB8	SP110	TNFRSF13B	
C8A	CFHR4	FERMT3	IL2RG	MYD88	PSTPIP1	SPINK5	TNFRSF13C	
C8B	CFHR5	FAS	IRAK1	NBAS	PTEN	STAT1	TNFRSF1A	
C9	CFI	FOXN1	IL36RN	NCF1	PTPRC	STAT3	TNFRSF4	

Supplementary Table 4. Main clinical features of patients included in this study.

ID	Sex	Age at study (y)	Gene	Family History	Consanguinity	Age of onset (y)	Syndromic features	Infections	Autoimmunity	Lymphoproliferation	Inflammation	Main clinical features, key words
P1	M	15	PIK3R1	-	-	7	-	V	-	Malign	Gut	IgA deficiency, immune dysregulation, growth delay, enteropathy, intestinal large bowel lymphoma
P2	F	57	TNFRSF13B	-	-	14	-	B	-	Benign	Gut, lung	CVID, infections, GLILD, granulomas, adenopathies, pancytopenia
P3	F	8	TNFRSF13B	√	-	3	-	B, V	√	Benign	-	EBV, pancytopenia, hepatosplenomegaly, hypogammaglobulinemia, ALPS-like, CVID-like
P4	M	18	IKBKG	√	-	14	-	-	-	-	Gut, joints	Crohn's disease, joint affectation, CGD, NEMO deficiency
P5	F	10	STAT3	-	-	4	√	B, F, Myc	-	-	-	Hyper IgE, atypical mycobacteria
P6	M	2	XIAP	-	√	0,8	√	B	-	-	Gut	Enteropathy, bacterial infection, growth delay
P7	M	1	G6PD	-	-	0,3	-	B	-	-	Bone	Osteomyelitis, *Salmonella* spp.
P8	M	30	STAT1	-	-	0,0	-	F	√	-	Skin, mucosa	Dermatophytosis, oral candidiasis
P9	F	4	STAT1	-	-	0,8	-	F	-	-	Joints	CMC, polyarthritis, episcleritis, bronchiectasis
P10	M	6	STAT1	√	√	4	-	F	-	-		Familiar CMC
P11	F	12	PLCG2	-	-	0,8	√	B	-	-	Skin, lungs	Agammaglobulinemia, severe cutaneous inflammation, bronchiectasis, B cell lymphopenia, growth delay
P12	M	15	ADA	-	-	8	-	B	-	Malign		Hodgkin lymphoma, B cell deficiency
P13	M	5	SKIV2L	-	-	0,8	√	B	-	-	Gut	Inflammatory enteropathy, growth delay
P14	M	0,25	MMACHC	√	-	0,1	-	-	-	-	-	fHLH, XLP
P15	F	44	SLC27A4	-	√	n.a.	-	-	-	-	Skin	Netherton syndrome
P16	F	0,3	DSG1	-	-	0,3	-	B	-	-	-	Erythroderma, Netherton syndrome, hyper IgE, eosinophilia
P17	F	1	DNAI2	√	√	0,1	-	V, F	√	Malign	-	Recurrent bronchitis, biphenotypic leukemia, growth delay
P18	M	38	SIX6	√	√	n.a.	-	B	-	-	-	CID, Low IgA
P19	M	5	RECQL4	-	√	1	√	V, F	-	-	-	CVID, growth delay
P20	M	11	UNC13D	√	-	8	-	V	-	Malign	Gut	Pancytopenia, hepatosplenomegaly, hemophagocytosis, panniculitic T cell lymphoma
P21	M	13	RAG2	√	-	6	√	-	-	Benign	Systemic	Persistent fever, intermittent abdominal pain, granulomatous hepatitis
P22	M	4	PLCG2	√	√	1	-	V	-	-	Skin	Periodic fever, skin rash
P23	F	11	TRAF3	-	-	11	-	V	-	-	-	Herpes Zoster, VZV meningoencephalitis
P24	M	2	NOD2	-	-	0,6	-	-	-	-	Gut	Early onset colitis
P25	F	8	LRBA	√		0,7		B, V, F	√	Malign	Gut	EBV, lymphoproliferation, autoimmunity, infections, dysregulation, enteropathy, autoimmune cytopenia
P26	F	11	LRBA	-	-	2	-	V	√	Benign	-	ALPS
P27	F	7	LRBA	-	-	7	-	B	√	Benign	-	Antibody deficiency, autoimmunity, lymphoproliferative syndrome
P28	F	14	IKZF1	√	-	5	√	B	-	-	-	Agammaglobulinemia, neurological delay
P29	M	9	13 Mb del cr.6	√	√	0,0	√	B, V	-	-	-	Neutrophilic dermatosis, oral and genital aphthae, growth delay
P30	M	6	BTK	-	-	4	-	B	-	-	-	Pneumonia, hypogammaglobulinemia, absence of B cells
P31	M	25	Gorham-Staut disease	-	-	21	√	B	-	-	Joints	Osteopenia, chylothorax, lymphopenia, *S.Aureus* bacteremia, septic arthritis
P32	M	11	inconclusive	√	-	14	-	V	-	Malign	Skin	CAEBV, T-lymphoproliferative syndrome associated with EBV, hemophagocytic syndrome
P33	M	0.17	inconclusive	n.a.	n.a.	0.17	n.a.	-	-	-	Gut	Syndromic enteropathy, chronic diarrhea
P34	F	1	inconclusive	-	-	0.17	-	B	-	-	-	Bacteremia due to *S.pneumoniae,* low acute phase reactants, neutropenia
P35	F	2	inconclusive	-	-	2	-	Myc	-	-	-	Previously healthy
P36	F	2	inconclusive	√	-	0.58		Myc	-	-		BCGitis
P37	F	3	inconclusive	-	-	0	-	V	-	-	-	HSV2 meningoencephalitis
P38	M	4	inconclusive	-	-	0.04	√	V	√	Benign	-	Eczema, thrombocytopenia
P39	M	2	inconclusive	-	-		-	B,V	√	-	-	Thrombocytopenia
P40	F	11	inconclusive	-	-	12	-	B	-	-	-	Meningitis caused by *Listeria monocytogenes*
P41	M	0.7	inconclusive	√	√	0.02	√	B, V	-	-	Gut	Coloboma in both eyes, chronic diarrhea, febrile peaks with increased acute phase reactants without infection
P42	F	0	inconclusive	-	-	0.16	-	B	-	-	-	Necrotizing pneumonia due to *S.aureus*
P43	F	1	inconclusive	-	-	0.08	-	V	-	Benign	Systemic	Hemophagocytosis, enterovirus encephalitis
P44	M	15	inconclusive	-	-	5	√	B,V	-	-	CNS, Gut	Behavior alterations, self-injury
P45	M	18	inconclusive	-	-	5	-	-	√	Benign	-	Adenopathies, mild hemolytic anemia, neutropenia and trombocytopenia

P46	M	5	inconclusive	-	√	0.16	√	-	-	Benign	Skin	Erythroderma, SCID suspicion, hepato-dermal GVHD confirmed with biopsy, pancytopenia
P47	M	9	inconclusive	-	-	7	-	-	√	Benign	Skin, Joint, Gut, Kidney	Leukocytoclastic vasculitis, colitis, glomerulonephritis
P48	F	0.1	inconclusive	-	√	0	√	-	-	Benign	-	Polymalformative syndrome, PID secondary to thymic aplasia, cardiopathy
P49	M	22	inconclusive	-	-	15	-	B	-	-	Gut	Diarrhea
P50	M	15	inconclusive	-	-	9	-	B	√	Benign	-	Kikuchi disease, nephrotic syndrome, pneumonia, osteomyelitis, chronic otitis media, adenopathies, hypogammaglobulinemia
P51	M	17	inconclusive	-	-	15	-	B	-	Malign	-	Liver transplantation, hyper IgM, absent response to polysaccharide vaccines
P52	M	18	inconclusive	√	-	6	-	B	-	-	-	Bronchiectasis
P53	F	0	inconclusive	-	√	0.33	-	V	-	Benign	Systemic	SCID phenotype, CMV encephalitis
P54	F	9	inconclusive	-	-	7	√	B	√	Benign	Skin	Atopic dermatitis, absent response to polysaccharide vaccines, bronchiectasis
P55	M	8	inconclusive	√	-	4	-	B	-	-	-	Thrombocytopenia
P56	M	13	inconclusive	√	-	5	-	B,V	-	Benign	-	Hypogammaglobulinemia, CVID like, splenomegaly
P57	M	23	inconclusive	-	-	infancy	√	-	√	-	-	Alopecia universalis
P58	F	4	inconclusive	√	-	2.4	-	B	-	-	-	*Exitus*
P59	F	40	inconclusive	-	-	5	-	B, F	-	Benign	Skin, joints	CID, T-lymphoproliferative syndrome associated with EBV in skin, ENT mucosa and bone. Infections, bronchiectasis
P60	M	11	inconclusive	-	-	4	-	B	-	-	-	-
P61	M	0.17	inconclusive	√	√	0,08	√	V,F	-	-	-	Absence of thymic shadow. SCID phenotype, Pneumocystis pneumonia

n.a.: not available.

V: Virus, B: Bacteria, F: Fungi, Myc: Mycobacteria

ALPS: Autoimmune Lymphoproliferative Syndrome, BCG: Bacille Calmette-Guérin, CAEBV: Chronic active Epstein-Barr virus, CID: Combined Immunodeficiency, CGD: Chronic Granulomatous Disease, CMC: Chronic Mucocutaneous Candidiasis, CMV: cytomegalovirus, CNS: Central Nervous System, CVID: Common Variable Immunodeficiency, ENT: ear-nose-throat, EBV: Epstein-Barr Virus, fHLH: familial Hemophagocytic Lymphohistiocytosis, GLILD: granulomatous and lymphocytic interstitial lung disease, GVHD: Graft Versus Host Disease, HSV2: Herpes Simplex Virus type 2, SCID: Severe Combined Immunodeficiency, VZV: Varizella Zoster Virus, XLP: X-linked Lymphoproliferation

Supplementary Table 5. Laboratory data of patients included in this study.

ID	Sex	Age at study (y)	Gene	Hypogammaglobulinemia	Neutropenia	Lymphopenia	Thrombocytopenia	Immunophenotype	Functional tests
P1	M	15	PIK3R1	-	-	-	-	Inverse CD4/CD8. No switched memory B cells	Low proliferation with anti-CD3, altered *in vivo* response to *Haemophilus influenzae*
P2	F	57	TNFRSF13B	√	√	√	√	Low CD4 naïve T cells, low Tregs, low pre-switched and switched B cells	n.a.
P3	F	8	TNFRSF13B	√	√	√	√	Low CD4 naïve T cells, low pre-switched and switched B cells	Normal proliferation, cytotoxicity and degranulation.
P4	M	18	IKBKG	-	-	-	-	Increased DN gamma delta T cells (22%)	Normal respiratory burst test and cytotoxicity/degranulation assays
P5	F	10	STAT3	-	-	-	-	Low Tregs, low Th17	Very low proliferation with anti-CD3
P6	M	2	XIAP	-	-	-	-	n.a.	n.a.
P7	M	1	G6PD	-	-	-	-	Inverse CD4/CD8 ratio. Increased effector HLA-DR+ CD8+ T cells, low switched B cells	Very low proliferation with anti-CD3 and ConA, very low IFNγ and IL12 production
P8	M	30	STAT1	-	-	-	-	Myeloid DC >>> plasmacytoid DC	Negative *in vivo* response to Candidin.
P9	F	4	STAT1	-	-	-	-	Low CD4 naïve T cells, low Th17, low pre-switched B cells	Low proliferation with PHA
P10	M	6	STAT1	-	-	-	-	3,4% DN TCR αβ T cells, increased Th1, low Th17, low Tregs	Low IL12 and IFNγ production
P11	F	12	PLCG2	-	-	-	-	T+ B- NK+	Normal proliferation, normal respiratory burst test
P12	M	15	ADA	√	-	-	-	T+ B- NK-	Normal respiratory burst test
P13	M	5	SKIV2L	√	-	-	-	Normal	Low proliferation with PHA (normal to anti-CD3 and ConA), normal respiratory burst test
P14	M	0,25	MMACHC	-	√	-	√	n.a.	Absent degranulation and cytotoxicity
P15	F	44	SLC27A4	-	-	-	-	Normal	Normal
P16	F	0,3	DSG1	-	-	-	-	Very low effector and memory T cells, low Th2, low Th1/Th17	n.a.
P17	F	1	DNAI2	-	-	-	-	Normal	Low proliferation with PWM and PHA
P18	M	38	SIX6	-	-	-	√	Low switched memory B cells	Low proliferation with anti-CD3
P19	M	5	RECQL4	√	-	√	√	Increased CD4/CD8 ration	Low proliferation with PWM,antiI-CD3 and ConA.
P20	M	11	UNC13D	-	√	√	-	Normal	Alternate low/normal degranulation and cytotoxicity
P21	M	13	RAG2	-	-	√	√	Low T cells, low NK cells.	Normal proliferation, normal respiratory burst test
P22	M	4	PLCG2	-	√	-	-	Normal	n.a.
P23	F	11	TRAF3	-	-	-	-	Normal	Absent IL-12 production, normal IFNγ production
P24	M	2,0	NOD2	-	-	-	-	Normal	Normal proliferation, normal respiratory burst test
P25	F	8	LRBA	√	√	√	-	T+ B- NK-	Low/absent degranulation and cytotoxicity
P26	F	11	LRBA	-	-	-	-	4.2% DN TCR αβ T cells, low effector and memory T cells, low Tregs	Low CD69 and CD40L expression
P27	F	7	LRBA	√	-	-	-	Low pre-switched and switched B cells	Low pneumococcal response
P28	F	14	IKZF1	√	-	-	-	T+ B- NK+, low pre-switched and switched B cells	Negative ASLO
P29	M	9	13 Mb del cr.6	-	-	-	-	Normal	Low proliferation with anti-CD3, normal TNFα production in response to LPS, normal degranulation and cytotoxicity
P30	M	6	BTK	√	-	-	-	T+ B- NK+	n.a.
P31	M	25	Gorham-Staut	-	√	-	-	Low CD4 T cells, increased NK cells	Severe defect in IL12 production, altered respiratory burst test

			disease						
P32	M	11	inconclusive	-	√	-	√	Low pre-switched and switched B cells	Absent degranulation and cytotoxicity
P33	M	0.17	inconclusive	n.d.	-	-	√	Low Th1/Th17. Low pre-switched and switched B cells	Low proliferation with anti-CD3. Normal respiratory burst test
P34	F	1	inconclusive	-	√	-	-	Normal	Normal respiratory burst test. Low IFNγ production in response to LPS
P35	F	2	inconclusive	-	√	-	-	Normal	Absent IFNγ and IL-12 production
P36	F	2	inconclusive	n.d.	√	-	-	Normal	Absent IFNγ and IL-12 production
P37	F	3	inconclusive	-	n.a.	n.a.	n.a.	Normal	Low response to TLR3
P38	M	4	inconclusive	√	-	-	√	Increased DN gamma delta T cells (7.3%)	Low proliferation with anti-CD3+IL-2 and with ConA.
P39	M	2	inconclusive	-	-	√	√	Low B cells, low NK cells.	Very low proliferation with anti-CD3
P40	F	11	inconclusive	√	n.a.	n.a.	n.a.	Increased effector and memory CD4+ T cells. Increased pre-switch B cells	Absent IL-12 and low IFNγ production. Low CD40L up-regulation after PMA+ionomycin stimulation
P41	M	0.7	inconclusive	-	n.a.	n.a.	n.a.	Normal	Absent proliferation with anti-CD3 and with anti-CD3+IL-2. Very low CD40L up-regulation after PMA+ionomycin stimulation
P42	F	0	inconclusive	-	n.a.	n.a.	n.a.	Very low Th1. Th17 in lower limit	Normal proliferation, normal respiratory burst test. Absent IFNγ production. Low IL-17 production.
P43	F	1	inconclusive	-	n.a.	n.a.	n.a.	Normal	Normal proliferation, cytotoxicity and degranulation.
P44	M	15	inconclusive	-	-	√	-	Low T cells, inverse CD4/CD8 ratio	n.a.
P45	M	18	inconclusive	√	√	-	√	Increased CD4+ effector memory T cells. Increased Th1 cells. Low Th2 and Th17. Low switched B cells	n.a.
P46	M	5	inconclusive	n.d.	√	-	√	T-B-NK-	Absent proliferation with PMA+Ionomycin, anti-CD3 and anti-CD3+IL2
P47	M	9	inconclusive	-	n.a.	n.a.	n.a.	Increased CD4/CD8 ratio, increased HLA-DR expression	n.a.
P48	F	0.1	inconclusive	-	-	√	-	Low T and B cells. Increased CD4+ effector memory T cells	Absent proliferation, cytotoxicity and degranulation.
P49	M	22	inconclusive	√	-	√	√	Low T and B cells. Very low naïve T cells. Low switched B cells. Increased transitional B cells.	Very low proliferation
P50	M	15	inconclusive	√	-	√	-	n.a.	n.a.
P51	M	17	inconclusive	√	-	-	-	Increased CD4+ and CD8+ effector memory T cells. Low pre-switched and switched B cells	Low cytotoxicity and degranulation.
P52	M	18	inconclusive	√	n.a.	n.a.	n.a.	Increased CD4+ effector memory T cells. Low pre-switched and switched B cells, increased Tregs	n.a.
P53	F	0	inconclusive	√	√	√	√	T-B-NK-	Very low proliferation with anti-CD3. Absent cytoxicity and degranulation.
P54	F	9	inconclusive	√	-	-	-	T+B+NKlow	Low proliferation with anti-CD3 and anti-CD3+IL-2. Normal cytoxicity and degranulation. Normal IFNγ production.
P55	M	8	inconclusive	√	-	-	√	Low CD8 and increased ratio CD4/CD8.	n.a.
P56	M	13	inconclusive	√	√	√	√	Low B and NK cells	Low proliferation with anti-CD3. Slightly reduced with anti-CD3+IL2
P57	M	23	inconclusive	√	n.a.	n.a.	n.a.	Normal. Increased DN T cells (21%). Increased plasmablasts.	n.a.
P58	F	4	inconclusive	√	-	-	-	Normal	n.a.
P59	F	40	inconclusive	√	n.a.	n.a.	n.a.	Absent pre-switched and switched B cells. Low CD4 naive T cells, increased Th1 T cells.	Very low proliferation with PMA+ionomycin. Normal cytotoxicity
P60	M	11	inconclusive	√	-	-	-	Absent pre-switched and switched B cells	n.a.
P61	M	0.17	inconclusive	√	-	√	-	Normal	Very low proliferation with PMA+ionomycin and with ConA. Low proliferation with PHA. Absent proliferation with anti-CD3. Normal cytotoxicity

n.a.: not available. n.d.: not determined

ASLO: Anti-streptolysin O, ConA: Concanavalin A, DC: Dendritic cell, DN: Double negative, PHA: Phytohemagglutinin, PMA: Phorbol 12-myristate 13-acetate, PWM: Pokeweed, TLR3: Toll-like receptor type 3

Supplementary Table 6. Low-frequency variants of genes included in the virtual PID panel.

Patient	Gene	Chr	Genotype	Consequence	cDNA	Protein	dbSNP ID	ExAC freq	Sift	PolyPhen
P1	MAN2B1	19	het	missense	c.1444C>T (NM_000528.3)	p.Arg482Trp (NP_000519.2)	rs533428309	0.000029	deleterious (0.01)	benign (0.254)
P1	JAK3	19	het	missense	c.997C>T (NM_000215.3)	p.Pro333Ser (NP_000206.2)	rs199908476	.	tolerated (0.47)	benign (0.062)
P1	IL7R	5	het	missense	c.214G>C (NM_002185.3)	p.Glu72Gln (NP_002176.2)	rs148001159	0.000264	tolerated (0.35)	benign (0.02)
P1	CR2	1	het	missense	c.1676G>A (NM_001006658.2)	p.Gly559Glu (NP_001006659.1)	rs143614333	0.000775	deleterious (0.01)	probably_damaging (0.954)
Patient	**Gene**	**Chr**	**Genotype**	**Consequence**	**cDNA**	**Protein**	**dbSNP ID**	**ExAC freq**	**Sift**	**PolyPhen**
P2	LRRC8A	9	het	missense	c.1111G>A (NM_019594.3)	p.Asp371Asn (NP_062540.2)	.	.	deleterious (0)	probably damaging (0.985)
P2	DOCK8	9	het	missense	c.950G>A (NM_203447.3)	p.Arg317Gln (NP_982272.2)	rs191413750	0.000074	tolerated (0.18)	possibly damaging (0.781)
P2	CD3G	11	het	missense	c.56G>A (NM_000073.2)	p.Gly19Asp (NP_000064.1)	rs146393315	0.000634	tolerated (0.11)	possibly damaging (0.462)
Patient	**Gene**	**Chr**	**Genotype**	**Consequence**	**cDNA**	**Protein**	**dbSNP ID**	**ExAC freq**	**Sift**	**PolyPhen**
P3	PLCG2	16	het	missense	c.3493G>A (NM_002661.3)	p.Val1165Ile (NP_002652.2)	rs372557475	0.000025	tolerated (0.05)	probably_damaging (0.988)
P3	SKIV2L	6	het	missense	c.3334C>T (NM_006929.4)	p.Leu1112Phe (NP_008860.4)	.	.	deleterious (0)	probably_damaging (0.942)
P3	DOCK8	9	het	missense	c.950G>A (NM_203447.3)	p.Arg317Gln (NP_982272.2)	rs191413750	0.000074	tolerated (0.11)	benign (0.347)
P3	PEPD	19	het	missense	c.751T>A (NM_000285.3)	p.Ser251Thr (NP_000276.2)	rs201572375	0.000085	tolerated (0.08)	benign (0.184)
P3	VPS13B	8	het	missense	c.8645C>T (NM_017890.4)	p.Pro2882Leu (NP_060360.3)	rs145890213	0.000955	tolerated (0.11)	benign (0.043)
P3	IL12B	5	het	missense	c.823G>A (NM_002187.2)	p.Val275Ile (NP_002178.2)	rs189324104	0.000173	tolerated (0.4)	benign (0.014)
Patient	**Gene**	**Chr**	**Genotype**	**Consequence**	**cDNA**	**Protein**	**dbSNP ID**	**ExAC freq**	**Sift**	**PolyPhen**

P4	GFI1	1	het	missense	c.319C>G (NM_001127216.1)	p.Pro107Ala (NP_001120688.1)	rs149914857	0.002583	deleterious (0.01)	benign (0.239)
P4	NLRP3	1	het	missense	c.1312A>T (NM_001079821.2)	p.Thr438Ser (NP_001073289.1)	rs180177465	.	tolerated (0.05)	possibly_damaging (0.864)
P4	IFNGR2	21	het	missense	c.940G>A (NM_005534.3)	p.Asp314Asn (NP_005525.2)	rs369529404	0.000041	deleterious (0.01)	possibly_damaging (0.683)
P4	NHP2	5	het	missense	c.190G>A (NM_017838.3)	p.Val64Met (NP_060308.1)	rs79031130	0.000428	tolerated (0.05)	probably_damaging (0.973)
P4	TAP2	6	het	missense	c.1655A>G (NM_018833.2)	p.Glu552Gly (NP_061313.2)	rs151064661	0.000206	deleterious (0.01)	probably_damaging (0.988)
P4	TLR3	4	het	missense	c.2486G>A (NM_003265.2)	p.Arg829Lys (NP_003256.1)	.	.	tolerated (0.08)	benign (0.356)
P4	UNC13D	17	het	missense	c.2180G>A (NM_199242.2)	p.Arg727Gln (NP_954712.1)	rs747390615	0.000635	tolerated (0.47)	benign (0.064)
Patient	**Gene**	**Chr**	**Genotype**	**Consequence**	**cDNA**	**Protein**	**dbSNP ID**	**ExAC freq**	**Sift**	**PolyPhen**
P5	C3	19	het	missense	c.2027C>T (NM_000064.2)	p.Thr676Met (NP_000055.2)	rs139945572	0.000016	tolerated (1)	benign (0.007)
P5	KMT2D	12	het	missense	c.11006G>A (NM_003482.3)	p.Gly3669Asp (NP_003473.3)	rs760279764	0.000017	.	possibly_damaging (0.949)
P5	KMT2D	12	het	missense	c.1967T>C (NM_003482.3)	p.Leu656Pro (NP_003473.3)	rs1169567206	.	.	benign (0)
Patient	**Gene**	**Chr**	**Genotype**	**Consequence**	**cDNA**	**Protein**	**dbSNP ID**	**ExAC freq**	**Sift**	**PolyPhen**
P6	ADA	20	het	missense	c.454C>A (NM_000022.2)	p.Leu152Met (NP_000013.2)	rs121908728	0.000398	deleterious (0.01)	probably_damaging (0.999)
P6	DCLRE1C	10	het	missense	c.1560G>C (NM_001033855.1)	p.Lys520Asn (NP_001029027.1)	rs753955796	0.000016	tolerated (0.19)	benign (0.067)
P6	PIK3CD	1	het	missense	c.1394C>T (NM_005026.3)	p.Thr465Met (NP_005017.3)	rs368722127	0.001058	tolerated (0.23)	possibly_damaging (0.741)
P6	TYK2	19	het	missense	c.1807G>A (NM_003331.4)	p.Val603Met (NP_003322.3)	rs140594440	0.000281	deleterious (0)	possibly_damaging (0.885)
P6	UNC13D	17	het	missense	c.1616A>G (NM_199242.2)	p.Asp539Gly (NP_954712.1)	rs780794721	0.000008	tolerated (0.37)	benign (0.06)
Patient	**Gene**	**Chr**	**Genotype**	**Consequence**	**cDNA**	**Protein**	**dbSNP ID**	**ExAC freq**	**Sift**	**PolyPhen**
P7	CD247	1	het	stop gained	c.301C>T (NM_198053.2)	p.Gln101Ter (NP_932170.1)	rs55729925	0.000224	.	.
Patient	**Gene**	**Chr**	**Genotype**	**Consequence**	**cDNA**	**Protein**	**dbSNP ID**	**ExAC freq**	**Sift**	**PolyPhen**
P08	BLM	15	het	missense	c.43C>T (NM_000057.2)	p.Arg15Cys (NP_000048.1)	rs148545569	0.000229	deleterious (0.02)	possibly_damaging (0.853)

P8	C7	5	het	stop gained	c.2188C>T (NM_000587.2)	p.Gln730Ter (NP_000578.2)	rs768866511	0.000008	.	.
P08	IL12RB2	1	het	missense	c.742G>C (NM_001559.2)	p.Asp248His (NP_001550.1)	rs545150086	0.000016	tolerated (0.14)	probably_damaging (0.991)
P8	LIG4	13	het	inframe deletion	c.1172_1174del (NM_206937.1)	p.Ser391del (NP_996820.1)	rs775835184	0.000025	.	.
P8	PSTPIP1	15	het	missense	c.1213C>T (NM_003978.3)	p.Arg405Cys (NP_003969.2)	rs201253322	0.000469	tolerated (0.11)	benign (0.105)
P08	WIPF1	2	het	frameshift	c.768dupC (NM_003387.4)	p.Ser257GlnfsTer6 (NP_003378.3)	.	.	.	.
Patient	**Gene**	**Chr**	**Genotype**	**Consequence**	**cDNA**	**Protein**	**dbSNP ID**	**ExAC freq**	**Sift**	**PolyPhen**
P9	C1R	12	het	missense	c.647C>G (NM_001733.4)	p.Pro216Arg (NP_001724.3)	rs189155429	0.000627	tolerated (0.3)	possibly_damaging (0.58)
P9	CD79A	19	het	missense	c.655G>C (NM_001783.3)	p.Gly219Arg (NP_001774.1)	rs1478564368	.	tolerated (0.29)	probably_damaging (0.991)
P9	MEFV	16	het	missense	c.74A>G (NM_000243.2)	p.Lys25Arg (NP_000234.1)	rs924530771	.	deleterious (0)	probably_damaging (0.999)
Patient	**Gene**	**Chr**	**Genotype**	**Consequence**	**cDNA**	**Protein**	**dbSNP ID**	**ExAC freq**	**Sift**	**PolyPhen**
P10	UNC13D	17	het	missense	c.2341G>A (NM_199242.2)	p.Val781Ile (NP_954712.1)	rs149871493	0.001269	tolerated(0.47)	benign(0.001)
P10	LPIN2	18	het	missense	c.1159A>G (NM_014646.2)	p.Lys387Glu (NP_055461.1)	rs104895501	0.00433	tolerated(0.07)	benign(0.414)
P10	KMT2D	12	het	missense	c.8774C>T (NM_003482.3)	p.Ala2925Val (NP_003473.3)	rs199547661	0.001731	.	benign(0.004)
P10	KMT2D	12	het	missense	c.13274C>T (NM_003482.3)	p.Ala4425Val (NP_003473.3)	rs752531267	.	.	benign(0.067)
P10	DKC1	X	hom	missense	c.838A>C (NM_001363.3)	p.Ser280Arg (NP_001354.1)	rs146700772	0.000319	deleterious(0.03)	benign(0.008)
P10	CSF3R	1	het	missense	c.1748G>A (NM_156039.3)	p.Arg583His (NP_724781.1)	rs148104401	0.000594	tolerated(0.88)	benign(0.003)
P10	CSF3R	1	het	missense	c.1919C>T (NM_156039.3)	p.Thr640Ile (NP_724781.1)	rs121918426	0.000541	tolerated(0.99)	benign(0)
P10	ATM	11	het	missense	c.3175G>A (NM_000051.3)	p.Ala1059Thr (NP_000042.3)	rs370282831	0.000008	tolerated(0.28)	benign(0.024)
Patient	**Gene**	**Chr**	**Genotype**	**Consequence**	**cDNA**	**Protein**	**dbSNP ID**	**ExAC freq**	**Sift**	**PolyPhen**
P11	LYST	1	het	missense	c.1727G>A (NM_000081.2)	p.Gly576Asp (NP_000072.2)	rs1240621396	.	deleterious (0.04)	probably_damaging (0.999)
P11	RAG1	11	het	missense	c.1864G>A (NM_000448.2)	p.Ala622Thr (NP_000439.1)	rs148380512	0.000141	deleterious (0)	probably_damaging (0.956)

P11	TAP1	6	het	missense	c.572T>C (NM_000593.5)	p.Leu191Pro (NP_000584.2)	rs142907576	0.001506	deleterious (0)	probably_damaging (0.999)
P11	VPS13B	8	het	splice (donor)	c.580+1G>A (NM_017890.4)	.	.	.	.	.
Patient	**Gene**	**Chr**	**Genotype**	**Consequence**	**cDNA**	**Protein**	**dbSNP ID**	**ExAC freq**	**Sift**	**PolyPhen**
P12	CHD7	8	het	inframe insertion	c.8451_8452ins (NM_017780.3)	p.Asn2817dup (NP_060250.2)	rs1443281585	.	.	.
P12	DNMT3B	20	het	missense	c.73G>A (NM_006892.3)	p.Gly25Arg (NP_008823.1)	rs151128145	0.000762	tolerated (0.09)	probably_damaging (0.992)
P12	CFI	4	het	missense	c.253T>A (NM_000204.3)	p.Tyr85Asn (NP_000195.2)	.	.	deleterious (0)	probably_damaging (0.999)
P12	CSF3R	1	het	missense	c.2480T>C (NM_156039.3)	p.Leu827Pro (NP_724781.1)	.	.	tolerated (0.32)	benign (0.002)
Patient	**Gene**	**Chr**	**Genotype**	**Consequence**	**cDNA**	**Protein**	**dbSNP ID**	**ExAC freq**	**Sift**	**PolyPhen**
P13	RECQL4	8	het	missense	c.1954G>A (NM_004260.3)	p.Val652Met (NP_004251.3)	rs61754061	0.005414	.	probably_damaging (0.995)
P13	SLC46A1	17	het	missense	c.189G>C (NM_080669.4)	p.Arg63Ser (NP_542400.2)	rs41297071	0.002711	.	possibly_damaging (0.514)
Patient	**Gene**	**Chr**	**Genotype**	**Consequence**	**cDNA**	**Protein**	**dbSNP ID**	**ExAC freq**	**Sift**	**PolyPhen**
P14	LIG1	19	het	missense	c.2216C>G (NM_000234.1)	p.Ser739Trp (NP_000225.1)	.	.	deleterious (0)	probably_damaging (1)
P14	USB1	16	het	missense	c.74G>C (NM_024598.3)	p.Arg25Thr (NP_078874.2)	rs772792606	0.000107	tolerated (0.43)	benign (0.008)
Patient	**Gene**	**Chr**	**Genotype**	**Consequence**	**cDNA**	**Protein**	**dbSNP ID**	**ExAC freq**	**Sift**	**PolyPhen**
P15	C8A	1	het	missense	c.1454G>A (NM_000562.2)	p.Arg485His (NP_000553.1)	rs1620075	0.00183	deleterious (0.01)	benign (0.073)
P15	ITGB2	21	het	missense	c.1117G>A (NM_000211.3)	p.Ala373Thr (NP_000202.2)	rs752903416	0.000025	tolerated (0.7)	benign (0.002)
P15	CD3G	11	het	missense	c.56G>A (NM_000073.2)	p.Gly19Asp (NP_000064.1)	rs146393315	0.000634	deleterious (0.04)	probably_damaging (0.987)
P15	PRKDC	8	het	missense	c.3743C>T (NM_006904.6)	p.Ser1248Leu (NP_008835.5)	rs200729621	0.000142	.	.
Patient	**Gene**	**Chr**	**Genotype**	**Consequence**	**cDNA**	**Protein**	**dbSNP ID**	**ExAC freq**	**Sift**	**PolyPhen**
P16	ADA	20	het	missense	c.395T>C (NM_000022.2)	p.Leu132Pro (NP_000013.2)	.	.	deleterious (0.03)	probably_damaging (0.987)
P16	ITGB2	21	het	missense	c.1172C>T (NM_000211.3)	p.Thr391Met (NP_000202.2)	rs141201564	0.000874	tolerated (0.09)	possibly_damaging (0.639)

Patient	Gene	Chr	Genotype	Consequence	cDNA	Protein	dbSNP ID	ExAC freq	Sift	PolyPhen
P17	ITGB2	21	het	missense	c.28G>A (NM_000211.3)	p.Ala10Thr (NP_000202.2)	rs201802601	0.005286	tolerated (0.57)	benign (0.02)
P17	SKIV2L	6	het	missense	c.3409C>T (NM_006929.4)	p.Arg1137Cys (NP_008860.4)	rs553733225	0.000379	deleterious (0.05)	probably_damaging (0.952)
P17	TYK2	19	het	missense	c.1534G>A (NM_003331.4)	p.Gly512Arg (NP_003322.3)	rs146786766	0.000689	tolerated (0.16)	benign (0.02)
Patient	**Gene**	**Chr**	**Genotype**	**Consequence**	**cDNA**	**Protein**	**dbSNP ID**	**ExAC freq**	**Sift**	**PolyPhen**
P18	CIITA	16	het	missense	c.2924A>G (NM_000246.3)	p.Lys975Arg (NP_000237.2)	rs140103491	0.007423	tolerated (0.26)	benign (0.004)
P18	DOCK8	9	het	missense	c.3044G>A (NM_203447.3)	p.Arg1015His (NP_982272.2)	rs200494857	0.000033	deleterious (0)	possibly_damaging (0.875)
P18	TICAM1	19	hom	missense	c.212G>A (NM_182919.3)	p.Arg71Gln (NP_891549.1)	rs372818181	0.000504	tolerated (0.38)	benign (0.001)
P18	ZAP70	2	het	missense	c.939C>G (NM_001079.3)	p.Ser313Arg (NP_001070.2)	rs145218891	0.000537	tolerated (0.44)	possibly_damaging (0.847)
Patient	**Gene**	**Chr**	**Genotype**	**Consequence**	**cDNA**	**Protein**	**dbSNP ID**	**ExAC freq**	**Sift**	**PolyPhen**
P19	C5	9	het	missense	c.64A>G (NM_001735.2)	p.Thr22Ala (NP_001726.2)	rs564964646	0.000774	tolerated (0.08)	benign (0.411)
P19	CIITA	16	het	missense	c.2384G>A (NM_000246.3)	p.Arg795Gln (NP_000237.2)	rs553503699	0.000079	tolerated (0.11)	benign (0.291)
P19	TAZ	X	hom	missense	c.606G>C (NM_000116.3)	p.Glu202Asp (NP_000107.1)	rs781893396	0.000023	deleterious (0.04)	possibly_damaging (0.695)
Patient	**Gene**	**Chr**	**Genotype**	**Consequence**	**cDNA**	**Protein**	**dbSNP ID**	**ExAC freq**	**Sift**	**PolyPhen**
P20	CYBB	X	hom	missense	c.1551T>A (NM_000397.3)	p.Asp517Glu (NP_000388.2)	rs151344452	0.001586	tolerated (0.27)	benign (0.031)
P20	NCF2	1	het	missense	c.1184G>A (NM_000433.3)	p.Arg395Gln (NP_000424.2)	rs145229115	0.001063	tolerated (0.2)	benign (0.023)
Patient	**Gene**	**Chr**	**Genotype**	**Consequence**	**cDNA**	**Protein**	**dbSNP ID**	**ExAC freq**	**Sift**	**PolyPhen**
P21	UNC13D	17	het	missense	c.2341G>A (NM_199242.2)	p.Val781Ile (NP_954712.1)	rs149871493	0.001269	tolerated (0.47)	benign (0.001)
P21	NFKBIA	14	het	missense	c.220G>C (NM_020529.2)	p.Gly74Arg (NP_065390.1)	rs766345843	0.000013	deleterious (0.02)	possibly_damaging (0.79)
P21	MBL2	10	het	missense	c.727G>A (NM_000242.2)	p.Val243Ile (NP_000233.1)	rs185230071	0.000061	tolerated (0.2)	benign (0.03)
P21	LYST	1	het	missense	c.2258G>A (NM_000081.2)	p.Ser753Asn (NP_000072.2)	rs746829669	0.000058	tolerated (0.58)	benign (0.039)

P21	ATM	11	het	missense	c.4388T>G (NM_000051.3)	p.Phe1463Cys (NP_000042.3)	rs138327406	0.001378	deleterious (0)	probably_damaging (0.946)
P21	AIRE	21	het	missense	c.1084G>T (NM_000383.3)	p.Val362Leu (NP_000374.1)	rs763954225	0.000107	tolerated (0.48)	benign (0.047)
Patient	**Gene**	**Chr**	**Genotype**	**Consequence**	**cDNA**	**Protein**	**dbSNP ID**	**ExAC freq**	**Sift**	**PolyPhen**
P22	C5	9	het	missense	c.3029C>T (NM_001735.2)	p.Ala1010Val (NP_001726.2)	rs34362143	0.000659	tolerated (0.22)	benign (0.208)
P22	TICAM1	19	het	missense	c.479C>T (NM_182919.3)	p.Ser160Phe (NP_891549.1)	rs145148929	0.002425	deleterious (0)	probably_damaging (0.951)
Patient	**Gene**	**Chr**	**Genotype**	**Consequence**	**cDNA**	**Protein**	**dbSNP ID**	**ExAC freq**	**Sift**	**PolyPhen**
P23	CR2	1	het	missense	c.1537A>G (NM_001006658.2)	p.Ile513Val (NP_001006659.1)	rs1210123747	.	tolerated (0.69)	benign (0.019)
P23	TERT	5	het	missense	c.193C>A (NM_198253.2)	p.Pro65Thr (NP_937983.2)	rs544215765	.	tolerated (0.4)	benign (0.255)
P23	PIK3R1	5	het	splice region	c.1020-3C>T (NM_181523.2)	.	rs200653607	0.000266	.	.
P23	AP3B1	5	het	missense	c.1069A>G (NM_003664.3)	p.Ile357Val (NP_003655.3)	rs142025324	0.001484	tolerated (0.22)	possibly_damaging (0.447)
P23	PSMB8	6	het	missense	c.371G>C (NM_148919.3)	p.Cys124Ser (NP_683720.2)	rs139299166	0.000008	deleterious (0)	probably_damaging (0.972)
P23	CIITA	16	het	missense	c.326C>G (NM_000246.3)	p.Ser109Cys (NP_000237.2)	rs375019139	0.000083	deleterious (0.02)	possibly_damaging (0.705)
Patient	**Gene**	**Chr**	**Genotype**	**Consequence**	**cDNA**	**Protein**	**dbSNP ID**	**ExAC freq**	**Sift**	**PolyPhen**
P24	DNMT3B	20	het	missense	c.1079C>T (NM_006892.3)	p.Ser360Leu (NP_008823.1)	rs766544393	0.000025	tolerated (0.23)	benign (0.005)
P24	ITCH	20	het	missense	c.1154C>T (NM_001257137.1)	p.Thr385Met (NP_001244066.1)	.	.	deleterious (0)	probably_damaging (0.984)
Patient	**Gene**	**Chr**	**Genotype**	**Consequence**	**cDNA**	**Protein**	**dbSNP ID**	**ExAC freq**	**Sift**	**PolyPhen**
P25	CIITA	16	het	missense	c.2536C>T (NM_000246.3)	p.His846Tyr (NP_000237.2)	rs769472311	0.000035	tolerated (0.62)	benign (0.003)
P25	JAK2	9	het	missense	c.337C>G (NM_004972.3)	p.Leu113Val (NP_004963.1)	rs143103233	0.000636	tolerated (0.51)	benign (0.009)
P25	LIG1	19	het	missense	c.2535G>T (NM_000234.1)	p.Lys845Asn (NP_000225.1)	rs145821638	0.000895	deleterious (0.01)	probably_damaging (0.998)
Patient	**Gene**	**Chr**	**Genotype**	**Consequence**	**cDNA**	**Protein**	**dbSNP ID**	**ExAC freq**	**Sift**	**PolyPhen**
P26	CD8A	2	het	missense	c.425C>T (NM_001145873.1)	p.Ala142Val (NP_001139345.1)	rs1218870889	.	tolerated (0.29)	benign (0.109)

P26	CFB	6	het	missense	c.1106C>T (NM_001710.5)	p.Pro369Leu (NP_001701.2)	rs200890358	0.000491	tolerated (0.28)	benign (0.004)
P26	CIITA	16	het	missense	c.2924A>G (NM_000246.3)	p.Lys975Arg (NP_000237.2)	rs140103491	0.007423	tolerated (0.26)	benign (0.004)
P26	CORO1A	16	het	missense	c.520G>A (NM_001193333.2)	p.Val174Met (NP_001180262.1)	rs769707848	0.000041	tolerated (0.49)	benign (0.022)
P26	CSF3R	1	het	splice (donor)	c.2040+2T>C (NM_156039.3)	.	.	.	.	.
P26	ZAP70	2	het	missense	c.939C>G (NM_001079.3)	p.Ser313Arg (NP_001070.2)	rs145218891	0.000537	tolerated (0.44)	possibly_damaging (0.847)
Patient	**Gene**	**Chr**	**Genotype**	**Consequence**	**cDNA**	**Protein**	**dbSNP ID**	**ExAC freq**	**Sift**	**PolyPhen**
P27	CARD9	9	het	stop gained	c.1228G>T (NM_052813.4)	p.Glu410Ter (NP_434700.2)	.	.	.	.
P27	CSF3R	1	het	missense	c.2153C>T (NM_156039.3)	p.Pro718Leu (NP_724781.1)	rs183614500	0.000158	tolerated (0.7)	benign (0.042)
P27	IL17RA	22	het	missense	c.152C>T (NM_014339.5)	p.Thr51Met (NP_055154.3)	rs143008696	0.001738	tolerated (0.19)	possibly_damaging (0.588)
P27	IL17RA	22	het	missense	c.1604G>A (NM_014339.5)	p.Arg535His (NP_055154.3)	rs1415141558	.	tolerated (0.07)	probably_damaging (0.999)
P27	LRRC8A	9	het	missense	c.779T>C (NM_019594.3)	p.Val260Ala (NP_062540.2)	.	.	deleterious (0.01)	benign (0.406)
Patient	**Gene**	**Chr**	**Genotype**	**Consequence**	**cDNA**	**Protein**	**dbSNP ID**	**ExAC freq**	**Sift**	**PolyPhen**
P28	C6	5	het	missense	c.1471G>A (NM_000065.2)	p.Val491Met (NP_000056.2)	rs200823179	0.000198	tolerated (0.09)	benign (0.143)
P28	LIG4	13	het	missense	c.1208T>C (NM_206937.1)	p.Val403Ala (NP_996820.1)	rs779305286	0.000033	tolerated (0.25)	benign (0.032)
P28	CHD7	8	het	missense	c.7538G>A (NM_017780.3)	p.Arg2513Gln (NP_060250.2)	rs1064794649	.	deleterious (0)	possibly_damaging (0.902)
Patient	**Gene**	**Chr**	**Genotype**	**Consequence**	**cDNA**	**Protein**	**dbSNP ID**	**ExAC freq**	**Sift**	**PolyPhen**
P29	C7	5	het	missense	c.1852C>T (NM_000587.2)	p.Arg618Trp (NP_000578.2)	rs202140226	0.000803	deleterious (0.02)	possibly_damaging (0.549)
P29	CD55	1	het	missense	c.155G>C (NM_001114752.1)	p.Arg52Pro (NP_001108224.1)	rs28371588	0.000272	tolerated (0.16)	possibly_damaging (0.563)
P29	KMT2D	12	het	missense	c.2992C>A (NM_003482.3)	p.Pro998Thr (NP_003473.3)	rs143711798	0.000472	deleterious (0)	.
P29	LYST	1	het	missense	c.9243A>C (NM_000081.2)	p.Gln3081His (NP_000072.2)	rs757239793	0.000025	deleterious (0)	probably_damaging (0.999)
Patient	**Gene**	**Chr**	**Genotype**	**Consequence**	**cDNA**	**Protein**	**dbSNP ID**	**ExAC freq**	**Sift**	**PolyPhen**

P30	VPS13B	8	het	missense	c.11263C>G (NM_017890.4)	p.Leu3755Val (NP_060360.3)	rs201448515	0.000021	deleterious (0)	probably_damaging (0.992)
P30	DOCK8	9	het	missense	c.663C>A (NM_203447.3)	p.Asp221Glu (NP_982272.2)	rs139391329	0.001731	tolerated (1)	benign (0.001)
P30	NOD2	16	het	missense	c.2264C>T (NM_022162.1)	p.Ala755Val (NP_071445.1)	rs61747625	0.002318	deleterious (0.01)	probably_damaging (0.999)
P30	LPIN2	18	het	missense	c.2161C>T (NM_014646.2)	p.His721Tyr (NP_055461.1)	rs200256485	0.000165	deleterious (0)	probably_damaging (1)
Patient	**Gene**	**Chr**	**Genotype**	**Consequence**	**cDNA**	**Protein**	**dbSNP ID**	**ExAC freq**	**Sift**	**PolyPhen**
P31	C1R	12	het	missense	c.446G>A (NM_001733.4)	p.Arg149Gln (NP_001724.3)	rs73046147	0.000025	tolerated (0.95)	benign (0.001)
P31	C2	6	het	missense	c.386G>A (NM_000063.4)	p.Arg129His (NP_000054.2)	rs367996721	0.000083	deleterious (0)	probably_damaging (0.999)
P31	CFP	X	hom	missense	c.151G>T (NM_002621.2)	p.Val51Phe (NP_002612.1)	rs751421714	.	deleterious (0)	possibly_damaging (0.72)
P31	CIITA	16	het	missense	c.835A>G (NM_000246.3)	p.Thr279Ala (NP_000237.2)	rs145961289	0.000132	tolerated (0.89)	benign (0.016)
P31	DOCK8	9	het	missense	c.6064A>G (NM_203447.3)	p.Met2022Val (NP_982272.2)	rs143458628	0.000091	tolerated (0.17)	benign (0.03)
P31	IRAK4	12	het	missense	c.1169A>G (NM_016123.3)	p.His390Arg (NP_057207.2)	rs4251583	0.000593	tolerated (0.09)	benign (0.033)
P31	PMM2	16	het	splice (donor)	c.255+2T>C (NM_000303.2)	.	rs139716296	0.000082	.	.
P31	STAT1	2	het	missense	c.722G>A (NM_007315.3)	p.Arg241Gln (NP_009330.1)	rs146273341	0.000222	tolerated (0.19)	benign (0.002)
P31	TICAM1	19	het	missense	c.479C>T (NM_182919.3)	p.Ser160Phe (NP_891549.1)	rs145148929	0.002425	deleterious (0)	probably_damaging (0.951)
Patient	**Gene**	**Chr**	**Genotype**	**Consequence**	**cDNA**	**Protein**	**dbSNP ID**	**ExAC freq**	**Sift**	**PolyPhen**
P32	C8A	1	het	missense	c.89G>A (NM_000562.2)	p.Arg30Gln (NP_000553.1)	rs201955480	0.000058	tolerated (0.58)	probably_damaging (0.981)
P32	DNMT3B	20	het	missense	c.73G>A (NM_006892.3)	p.Gly25Arg (NP_008823.1)	rs151128145	0.000761	tolerated (0.09)	probably_damaging (0.992)
P32	HAX1	1	het	missense	c.137G>A (NM_006118.3)	p.Arg46His (NP_006109.2)	.	.	tolerated (0.55)	benign (0.004)
P32	KMT2D	12	het	missense	c.1955G>C (NM_003482.3)	p.Arg652Pro (NP_003473.3)	.	.	tolerated (0.123)	benign (0.000)
P32	MPO	17	het	splice (acceptor)	c.2031-2A>C (NM_000250.1)	.	rs35897051	0.004257	.	.
P32	TAP1	6	het	stop gained	c.856C>T (NM_000593.5)	p.Arg286Ter (NP_000584.2)	.	.	.	.

P32	VPS13B	8	het	missense	c.2880A>T (NM_017890.4)	p.Leu960Phe (NP_060360.3)	.	0.000149	tolerated (0.32)	probably_damaging (0.975)
Patient	**Gene**	**Chr**	**Genotype**	**Consequence**	**cDNA**	**Protein**	**dbSNP ID**	**ExAC freq**	**Sift**	**PolyPhen**
P33	ERCC3	2	het	missense	c.144G>C (NM_000122.1)	p.Glu48Asp (NP_000113.1)	rs149309991	0.000198	tolerated (0.53)	benign (0.002)
P33	PLCG2	16	het	missense	c.1475C>T (NM_002661.3)	p.Thr492Ile (NP_002652.2)	rs1368273226	.	tolerated (0.21)	benign (0.03)
P33	RAG1	11	het	missense	c.101G>A (NM_000448.2)	p.Arg34Gln (NP_000439.1)	rs377307948	0.000115	deleterious (0)	possibly_damaging (0.643)
Patient	**Gene**	**Chr**	**Genotype**	**Consequence**	**cDNA**	**Protein**	**dbSNP ID**	**ExAC freq**	**Sift**	**PolyPhen**
P34	LRRC8A	9	het	missense	c.1250G>A (NM_019594.3)	p.Arg417Gln (NP_062540.2)	rs746556593	0.000017	tolerated (0.08)	probably_damaging (0.913)
P34	CHD7	8	het	missense	c.8672A>G (NM_017780.3)	p.Asn2891Ser (NP_060250.2)	rs202039728	0.000216	tolerated (0.09)	benign (0.01)
P34	C4B	6	het	missense	c.4072C>G (NM_001002029.3)	p.Leu1358Val (NP_001002029.3)	rs747337926	0.002556	deleterious (0.01)	possibly_damaging (0.677)
Patient	**Gene**	**Chr**	**Genotype**	**Consequence**	**cDNA**	**Protein**	**dbSNP ID**	**ExAC freq**	**Sift**	**PolyPhen**
P35	IL21R	16	het	missense	c.681G>C (NM_181079.4)	p.Met227Ile (NP_851565.4)	rs201566028	0.0002672	.	.
P35	KRAS	12	het	missense	c.535G>A (NM_033360.2)	p.Gly179Ser (NP_203524.1)	rs200970347	0.0003626	tolerated (0.14)	benign (0.008)
Patient	**Gene**	**Chr**	**Genotype**	**Consequence**	**cDNA**	**Protein**	**dbSNP ID**	**ExAC freq**	**Sift**	**PolyPhen**
P37	CD19	16	het	missense	c.395T>G (NM_001178098.1)	p.Leu132Arg (NP_001171569.1)	rs146795664	0.002125	tolerated (0.63)	benign (0.015)
P37	CTLA4	2	het	missense	c.326G>A (NM_005214.4)	p.Gly109Glu (NP_005205.2)	rs144988077	0.000239	tolerated (0.56)	benign (0.05)
P37	LIG4	13	het	missense	c.2425C>G (NM_206937.1)	p.Pro809Ala (NP_996820.1)	rs137899041	0.000082	tolerated (0.19)	benign (0.003)
P37	TICAM1	19	het	missense	c.238G>A (NM_182919.3)	p.Val80Met (NP_891549.1)	rs199816697	0.000714	tolerated (0.35)	benign (0.005)
Patient	**Gene**	**Chr**	**Genotype**	**Consequence**	**cDNA**	**Protein**	**dbSNP ID**	**ExAC freq**	**Sift**	**PolyPhen**
P38	PSTPIP1	15	het	missense	c.525C>G (NM_003978.3)	p.Asn175Lys (NP_003969.2)	.	.	tolerated (0.48)	benign (0.049)
P38	VPS13B	8	het	missense	c.5957G>A (NM_017890.4)	p.Gly1986Glu (NP_060360.3)	rs1440579372	.	deleterious (0)	probably_damaging (0.997)
Patient	**Gene**	**Chr**	**Genotype**	**Consequence**	**cDNA**	**Protein**	**dbSNP ID**	**ExAC freq**	**Sift**	**PolyPhen**

P39	TICAM1	19	het	missense	c.479C>T (NM_182919.3)	p.Ser160Phe (NP_891549.1)	rs145148929	0.002425	deleterious (0)	probably_damaging (0.951)
Patient	**Gene**	**Chr**	**Genotype**	**Consequence**	**cDNA**	**Protein**	**dbSNP ID**	**ExAC freq**	**Sift**	**PolyPhen**
P40	KMT2D	12	het	missense	c.14438A>G (NM_003482.3)	p.Asn4813Ser (NP_003473.3)	rs756696295	0.000008	tolerated (0.39)	benign (0.004)
P40	ALG13	X	het	missense	c.3249C>A (NM_001099922.2)	p.Asp1083Glu (NP_001093392.1)	.	.	deleterious (0.04)	probably_damaging (0.946)
Patient	**Gene**	**Chr**	**Genotype**	**Consequence**	**cDNA**	**Protein**	**dbSNP ID**	**ExAC freq**	**Sift**	**PolyPhen**
P41	FOXN1	17	het	missense	c.8C>T (NM_003593.2)	p.Ser3Leu (NP_003584.2)	rs146091703	0.000026	deleterious (0)	probably_damaging (0.975)
P41	C2	6	het	missense	c.614G>A (NM_000063.4)	p.Arg205His (NP_000054.2)	rs147186833	0.000202	deleterious (0.01)	probably_damaging (0.959)
P41	ITGB2	21	het	missense	c.1172C>T (NM_000211.3)	p.Thr391Met (NP_000202.2)	rs141201564	0.000874	tolerated (0.09)	possibly_damaging (0.639)
P41	CTSC	11	het	missense	c.1201G>A (NM_001814.4)	p.Glu401Lys (NP_001805.3)	rs200627023	0.000107	tolerated (0.09)	possibly_damaging (0.452)
P41	TCIRG1	11	het	missense	c.1615G>A (NM_006019.3)	p.Val539Ile (NP_006010.2)	rs142539969	0.000157	tolerated (0.5)	benign (0.099)
P41	UNC13D	17	het	missense	c.2341G>A (NM_199242.2)	p.Val781Ile (NP_954712.1)	rs149871493	0.001269	tolerated (0.47)	benign (0.001)
P41	PRKDC	8	het	missense	c.1338C>A (NM_006904.6)	p.Phe446Leu (NP_008835.5)	rs61729514	0.000879	.	probably_damaging (0.999)
Patient	**Gene**	**Chr**	**Genotype**	**Consequence**	**cDNA**	**Protein**	**dbSNP ID**	**ExAC freq**	**Sift**	**PolyPhen**
P42	CEBPE	14	het	missense	c.410G>A (NM_001805.3)	p.Ser137Asn (NP_001796.2)	rs140606768	0.000346	tolerated (0.52)	benign (0.001)
P42	VPS13B	8	het	missense	c.4774G>A (NM_017890.4)	p.Ala1592Thr (NP_060360.3)	.	.	deleterious (0.03)	probably_damaging (0.966)
P42	C7	5	het	missense	c.659G>A (NM_000587.2)	p.Arg220Gln (NP_000578.2)	rs369349760	0.000167	tolerated (0.58)	benign (0.024)
P42	LYST	1	het	missense	c.8214G>C (NM_000081.2)	p.Glu2738Asp (NP_000072.2)	rs140944484	0.000207	tolerated (1)	benign (0.002)
Patient	**Gene**	**Chr**	**Genotype**	**Consequence**	**cDNA**	**Protein**	**dbSNP ID**	**ExAC freq**	**Sift**	**PolyPhen**
P43	KMT2D	12	het	missense	c.1967T>C (NM_003482.3)	p.Leu656Pro (NP_003473.3)	rs1169567206	.	tolerated (0.06)	benign (0)
Patient	**Gene**	**Chr**	**Genotype**	**Consequence**	**cDNA**	**Protein**	**dbSNP ID**	**ExAC freq**	**Sift**	**PolyPhen**
P44	LYST	1	het	missense	c.5676G>A (NM_000081.2)	p.Met1892Ile (NP_000072.2)	rs143857674	0.000041	tolerated (0.98)	benign (0)

P44	TMC8	17	het	missense	c.1168G>A (NM_152468.4)	p.Val390Ile (NP_689681.2)	rs150546646	0.001003	tolerated (0.1)	benign (0.009)
P44	DNMT3B	20	het	missense	c.1345C>G (NM_006892.3)	p.Leu449Val (NP_008823.1)	rs757117952	0.000008	deleterious (0)	probably_damaging (0.964)
P44	DNMT3B	20	het	missense	c.1352A>T (NM_006892.3)	p.Glu451Val (NP_008823.1)	.	.	tolerated (0.19)	benign (0.02)
Patient	**Gene**	**Chr**	**Genotype**	**Consequence**	**cDNA**	**Protein**	**dbSNP ID**	**ExAC freq**	**Sift**	**PolyPhen**
P45	ZBTB24	6	het	missense	c.855G>T (NM_014797.2)	p.Arg285Ser (NP_055612.2)	rs143216162	0.0000494	deleterious (0.03)	benign (0.039)
P45	NCF1	7	het	missense	c.292T>G (NM_000265.4)	p.Cys98Gly (NP_000256.3)	rs144018361	0.0007298	deleterious (0.02)	possibly_damaging (0.849)
P45	CHD7	8	het	missense	c.5848G>A (NM_017780.3)	p.Ala1950Thr (NP_060250.2)	rs201423234	0.0003115	tolerated (0.64)	benign (0.032)
P45	TNFRSF1A	12	het	missense	c.1354A>G (NM_001065.3)	p.Ser452Gly (NP_001056.1)	rs200346150	0.000012	tolerated (0.14)	benign (0)
P45	AIRE	21	het	missense	c.1249C>A (NM_000383.3)	p.Leu417Ile (NP_000374.1)	.	.	tolerated (0.06)	probably_damaging (0.971)
Patient	**Gene**	**Chr**	**Genotype**	**Consequence**	**cDNA**	**Protein**	**dbSNP ID**	**ExAC freq**	**Sift**	**PolyPhen**
P46	FOXN1	17	het	missense	c.1727C>T (NM_003593.2)	p.Pro576Leu (NP_003584.2)	rs756165992	0.000025	tolerated (0.15)	probably_damaging (0.987)
P46	PEPD	19	het	missense	c.794G>A (NM_000285.3)	p.Arg265Gln (NP_000276.2)	rs200351927	0.000203	tolerated (0.11)	benign (0.148)
P46	TAP1	6	het	missense	c.919G>A (NM_000593.5)	p.Gly307Arg (NP_000584.2)	rs59328013	0.000121	deleterious (0.03)	probably_damaging (0.949)
P46	SKIV2L	6	het	missense	c.2564C>A (NM_006929.4)	p.Ala855Glu (NP_008860.4)	rs561701823	0.000034	tolerated (0.48)	benign (0.076)
P46	CTSC	11	het	missense	c.113C>G (NM_001814.4)	p.Thr38Ser (NP_001805.3)	rs758568173	0.000075	tolerated (0.31)	benign (0.057)
P46	TBK1	12	het	missense	c.1522C>A (NM_013254.3)	p.Leu508Ile (NP_037386.1)	rs144424516	0.000776	tolerated (0.46)	benign (0.033)
P46	STK4	20	het	missense	c.47A>G (NM_006282.2)	p.Lys16Arg (NP_006273.1)	rs142594802	0.000206	tolerated (0.11)	benign (0.402)
Patient	**Gene**	**Chr**	**Genotype**	**Consequence**	**cDNA**	**Protein**	**dbSNP ID**	**ExAC freq**	**Sift**	**PolyPhen**
P47	CHD7	8	het	missense	c.602A>G (NM_017780.3)	p.Gln201Arg (NP_060250.2)	rs764496155	0.000092	tolerated (0.27)	benign (0)
P47	UNC13D	17	het	missense	c.869C>T (NM_199242.2)	p.Ser290Leu (NP_954712.1)	rs202020396	0.000079	tolerated (0.22)	benign (0.01)
P47	VPS13B	8	het	missense	c.1559A>G (NM_017890.4)	p.His520Arg (NP_060360.3)	rs143205296	0.000538	deleterious (0)	possibly_damaging (0.897)

Patient	Gene	Chr	Genotype	Consequence	cDNA	Protein	dbSNP ID	ExAC freq	Sift	PolyPhen
P48	PLCG2	16	het	missense	c.1258G>A (NM_002661.3)	p.Ala420Thr (NP_002652.2)	rs201490178	0.00431	tolerated (0.39)	benign (0.004)
P48	C5	9	het	missense	c.3463G>A (NM_001735.2)	p.Ala1155Thr (NP_001726.2)	rs200624729	0.000382	deleterious (0.01)	possibly_damaging (0.844)
P48	C6	5	het	missense	c.1355G>A (NM_000065.2)	p.Gly452Glu (NP_000056.2)	rs142896559	0.004985	deleterious (0.01)	benign (0.003)
P48	ZAP70	2	het	missense	c.939C>G (NM_001079.3)	p.Ser313Arg (NP_001070.2)	rs145218891	0.000537	tolerated (0.44)	possibly_damaging (0.847)
P48	KMT2D	12	het	missense	c.6844C>T (NM_003482.3)	p.Arg2282Trp (NP_003473.3)	rs587783726	0.000042		possibly_damaging
Patient	**Gene**	**Chr**	**Genotype**	**Consequence**	**cDNA**	**Protein**	**dbSNP ID**	**ExAC freq**	**Sift**	**PolyPhen**
P49	UNC13D	17	het	missense	c.2341G>A (NM_199242.2)	p.Val781Ile (NP_954712.1)	rs149871493	0.001269	tolerated (0.47)	benign (0.001)
P49	CFH	1	het	missense	c.1548T>A (NM_000186.3)	p.Asn516Lys (NP_000177.2)	rs147403664	0.000405	tolerated (0.11)	probably_damaging (0.977)
P49	TAP1	6	het	splice region	c.1231-3C>T (NM_000593.5)	.	rs56366814	0.003445	.	.
P49	RAG2	11	het	missense	c.22G>A (NM_000536.3)	p.Val8Ile (NP_000527.2)	rs150762709	0.003189	tolerated (0.41)	possibly_damaging (0.842)
Patient	**Gene**	**Chr**	**Genotype**	**Consequence**	**cDNA**	**Protein**	**dbSNP ID**	**ExAC freq**	**Sift**	**PolyPhen**
P50	SKIV2L	6	het	missense	c.797C>G (NM_000063.4)	p.Ser266Trp (NP_000054.2)	rs116568722	0.000214	deleterious (0)	probably_damaging (0.962)
P50	C3	19	het	missense	c.2203C>T (NM_000064.2)	p.Arg735Trp (NP_000055.2)	rs117793540	0.002086	deleterious (0)	probably_damaging (0.99)
Patient	**Gene**	**Chr**	**Genotype**	**Consequence**	**cDNA**	**Protein**	**dbSNP ID**	**ExAC freq**	**Sift**	**PolyPhen**
P51	KMT2D	12	het	missense	c.7670C>T (NM_003482.3)	p.Pro2557Leu (NP_003473.3)	rs189888707	0.008494		probably damaging (0.998)
P51	TCIRG1	11	het	missense	c.479G>A (NM_006019.3)	p.Gly160Glu (NP_006010.2)	rs186758849	0.005491	tolerated (0.61)	benign (0.076)
Patient	**Gene**	**Chr**	**Genotype**	**Consequence**	**cDNA**	**Protein**	**dbSNP ID**	**ExAC freq**	**Sift**	**PolyPhen**
P52	TNFRSF11A	18	het	missense	c.1279G>A (NM_003839.2)	p.Asp427Asn (NP_003830.1)	rs201402594	0.000796	tolerated (0.06)	benign (0.238)
P52	LPIN2	18	het	missense	c.1876C>T (NM_014646.2)	p.Pro626Ser (NP_055461.1)	rs150806357	0.002973	tolerated (0.69)	benign (0.003)
Patient	**Gene**	**Chr**	**Genotype**	**Consequence**	**cDNA**	**Protein**	**dbSNP ID**	**ExAC freq**	**Sift**	**PolyPhen**

P53	C1QA	1	het	missense	c.295A>C (NM_015991.2)	p.Ile99Leu (NP_057075.1)	rs180679721	0.000304	tolerated (0.35)	benign (0.001)
P53	PTPRC	1	het	missense	c.260C>T (NM_002838.4)	p.Pro87Leu (NP_002829.3)	rs149798940	0.000074	tolerated (0.71)	benign (0)
P53	TERT	5	het	missense	c.1234C>T (NM_198253.2)	p.His412Tyr (NP_937983.2)	rs34094720	0.006342	tolerated (0.21)	possibly_damaging (0.885)
P53	SKIV2L	6	het	missense	c.1705G>A (NM_006929.4)	p.Val569Met (NP_008860.4)	rs144147284	0.002596	tolerated (0.07)	possibly_damaging (0.581)
P53	RFXANK	19	het	missense	c.187G>A (NM_003721.2)	p.Ala63Thr (NP_003712.1)	rs150525759	0.000074	tolerated (0.58)	benign (0.001)
Patient	**Gene**	**Chr**	**Genotype**	**Consequence**	**cDNA**	**Protein**	**dbSNP ID**	**ExAC freq**	**Sift**	**PolyPhen**
P54	TRAF3	14	het	missense	c.74G>T (NM_003300.3)	p.Arg25Leu (NP_003291.2)	rs370955205	0.000189	tolerated (0.17)	possibly_damaging (0.822)
P54	NOD2	16	het	missense	c.1627C>T (NM_022162.1)	p.Arg543Cys (NP_071445.1)	rs545580252	0.000041	tolerated (0.09)	benign (0.008)
P54	KMT2D	12	het	missense	c.10469C>T (NM_003482.3)	p.Pro3490Leu (NP_003473.3)	rs376980951	0.000126	deleterious (0.01)	possibly_damaging (0.483)
P54	ITK	5	het	missense	c.1510A>T (NM_005546.3)	p.Thr504Ser (NP_005537.3)	rs151046132	0.000824	tolerated (0.48)	benign (0.037)
P54	MTHFD1	14	het	missense	c.920A>G (NM_005956.3)	p.Tyr307Cys (NP_005947.3)	rs577555809	0.001188	tolerated (0.19)	possibly_damaging (0.871)
P54	PRKDC	8	het	missense	c.9445G>A (NM_006904.6)	p.Ala3149Thr (NP_008835.5)	rs8178208	0.008821	.	.
P54	ATM	11	het	missense	c.1274C>A (NM_000051.3)	p.Ala425Glu (NP_000042.3)	.	.	tolerated (1)	benign (0.001)
Patient	**Gene**	**Chr**	**Genotype**	**Consequence**	**cDNA**	**Protein**	**dbSNP ID**	**ExAC freq**	**Sift**	**PolyPhen**
P55	FAS	10	het	missense	c.580G>A (NM_000043.4)	p.Glu194Lys (NP_000034.1)	rs56006128	0.001801	tolerated (0.95)	benign (0)
P55	DOCK8	9	het	missense	c.287C>T (NM_203447.3)	p.Thr96Met (NP_982272.2)	.	.	tolerated (0.1)	possibly_damaging (0.793)
Patient	**Gene**	**Chr**	**Genotype**	**Consequence**	**cDNA**	**Protein**	**dbSNP ID**	**ExAC freq**	**Sift**	**PolyPhen**
P56	IFNGR2	21	het	missense	c.439G>A (NM_005534.3)	p.Glu147Lys (NP_005525.2)	rs17878639	0.00152	tolerated (0.93)	benign (0.156)
Patient	**Gene**	**Chr**	**Genotype**	**Consequence**	**cDNA**	**Protein**	**dbSNP ID**	**ExAC freq**	**Sift**	**PolyPhen**
P57	C8A	1	het	missense	c.385G>A (NM_000562.2)	p.Asp129Asn (NP_000553.1)	rs150404785	0.00223	tolerated (0.39)	benign (0.059)
Patient	**Gene**	**Chr**	**Genotype**	**Consequence**	**cDNA**	**Protein**	**dbSNP ID**	**ExAC freq**	**Sift**	**PolyPhen**

P58	C1S	12	het	missense	c.943G>A (NM_001734.3)	p.Asp315Asn (NP_001725.1)	rs117907409	0.003419	deleterious (0)	probably damaging (1)
P58	DNMT3B	20	het	missense	c.1150G>A (NM_006892.3)	p.Ala384Thr (NP_008823.1)	rs150682895	0.007936	tolerated (1)	benign (0.007)
P58	KMT2D	12	het	missense	c.13258C>T (NM_003482.3)	p.Arg4420Trp (NP_003473.3)	rs199797812	0.000059	deleterious (0.00)	probably damaging (1)
P58	CFH	1	het	missense	c.2850G>T (NM_000186.3)	p.Gln950His (NP_000177.2)	rs149474608	0.003583	deleterious (0.04)	probably damaging (0.962)
P58	STX11	6	het	missense	c.799G>A (NM_003764.3)	p.Val267Met (NP_003755.2)	rs45574234	0.00562	tolerated (0.08)	possibly damaging (0.847)
P58	PRKDC	8	het	missense	c.6479C>T (NM_006904.6)	p.Thr2160Met (NP_008835.5)	rs55991828	0.004512	.	.
P58	C7	5	het	missense	c.2008G>A (NM_000587.2)	p.Ala670Thr (NP_000578.2)	rs200737768	0.000725	tolerated (0.79)	benign (0.004)
P58	C9	5	het	missense	c.499C>T (NM_001737.3)	p.Pro167Ser (NP_001728.1)	rs34882957	0.004711	deleterious (0)	probably damaging (0.971)
Patient	**Gene**	**Chr**	**Genotype**	**Consequence**	**cDNA**	**Protein**	**dbSNP ID**	**ExAC freq**	**Sift**	**PolyPhen**
P59	STAT1	2	het	missense	c.722G>A (NM_007315.3)	p.Arg241Gln (NP_009330.1)	rs146273341	0.000222	tolerated (0.18)	benign (0.182)
P59	PRKDC	8	het	missense	c.16G>T (NM_006904.6)	p.Ala6Ser (NP_008835.5)	rs8177999	0.006118	.	benign (0.003)
P59	JAK2	9	het	missense	c.1711G>A (NM_004972.3)	p.Gly571Ser (NP_004963.1)	rs139504737	0.000481	tolerated (0.15)	probably damaging (0.974)
P59	PLCG2	16	het	missense	c.2011A>G (NM_002661.3)	p.Ile671Val (NP_002652.2)	rs150833842	0.006124	tolerated (1)	benign (0.016)
P59	CIITA	16	het	missense	c.3317C>T (NM_000246.3)	p.Ala1106Val (NP_000237.2)	rs1231888573	.	tolerated (0.17)	possibly damaging (0.611)
Patient	**Gene**	**Chr**	**Genotype**	**Consequence**	**cDNA**	**Protein**	**dbSNP ID**	**ExAC freq**	**Sift**	**PolyPhen**
P60	PSTPIP1	15	het	missense	c.657A>C (NM_003978.3)	p.Gln219His (NP_003969.2)	rs139362350	0.000702	deleterious (0)	probably damaging (0.94)
P60	CHD7	8	het	missense	c.8740G>A (NM_017780.3)	p.Gly2914Arg (NP_060250.2)	rs187751757	0.000352	.	probably damaging (0.998)
P60	VPS13B	8	het	missense	c.5350A>T (NM_017890.4)	p.Thr1784Ser (NP_060360.3)	.	.	tolerated (0.61)	possibly damaging (0.899)
Patient	**Gene**	**Chr**	**Genotype**	**Consequence**	**cDNA**	**Protein**	**dbSNP ID**	**ExAC freq**	**Sift**	**PolyPhen**
P61	PLCG2	16	het	missense	c.3786G>C (NM_002661.3)	p.Lys1262Asn (NP_002652.2)	rs374430619	0.000008	deleterious (0.01)	benign (0.045)
P61	NLRP12	19	het	missense	c.857C>T (NM_144687.2)	p.Pro286Leu (NP_653288.1)	rs201940393	0.000099	deleterious (0)	probably_damaging (0.971)

P61	GFI1	1	het	missense	c.319C>G (NM_001127216.1)	p.Pro107Ala (NP_001120688.1)	rs149914857	0.002583	deleterious (0.01)	benign (0.239)
P61	G6PC3	17	het	missense	c.403A>G (NM_138387.3)	p.Thr135Ala (NP_612396.1)	.	.	tolerated (0.77)	benign (0.001)

Predicted loss-of-function variants are in blue.