

ID	gender	age	age groups (1-5)+1, (6-11)+2, (12-18)+3	zignosity	MAIN GENETIC FINDINGS	Interpretation	ACMG category	zignosity	ADDITIONAL NGS findings	Interpretation	ACMG category	clinical presentation: 0=no, 1=yes	immunological data: normal=0, ↑=1, ↑↑=2	
1	ID 1-3	f	6 y 2 mo	2	het	CARD11(NM_032415):c.2542C>T; p.Arg846>Gys	VUS	PP5, PM2	het	IRAK4(NM_016123):c.431G>C; p.Ser1441Thr	VUS	PM2, BP1	food allergy, allergic rhinitis, asthma	↓ CD3 <sup>DR</sup> , ↓ CD3 <sup>CD4</sup> , ↓ CD45RA <sup>CD62L</sup> , ↓ CD45RA <sup>CD62L</sup> , ↑ CD45RA <sup>CD62L</sup> , ↓ CD4/CD8, ↓ CD19+, ↑ CD3 <sup>CD16</sup> /CD56, ↑ Ige, ↓ Ige4
2	ID 2-3	m	3 y 5 mo	1	het	TNFRSF1A(NM_001065):c.362G>A; p.Arg121Gln (mat)	VUS	BS1, BS2, PM1, PMS, PPS	het	TYK2(NM_003315):c.3310C>G; p.Pro1104Ala	VUS	PM2, PPS	cow's milk protein allergy, atopic dermatitis	↓ CD3 <sup>DR</sup> , ↓ CD45RA <sup>CD62L</sup> , ↑ CD45RA <sup>CD62L</sup> , ↓ CD3 <sup>CD8</sup> , ↑ CD4/CD8, ↑ Ige
					het	ALOXE3(NM_001165960):c.1109G>T; p.Arg366Ile (pat)	Path	PVS1, PM2, PM2	het	RFXAP(NM_000538):c.665C>T; p.Ala122Val	VUS	PM2, BP1		
					het	ALOXE3(NM_001165960):c.1161A>T; p.Cys548Arg (mat)	Likely Path	PM1, PM2, PM3, PP4, PPS	het	THBD(NM_000363):c.1550G>T; p.Pro501Leu	VUS	PM2, PM4		
3	ID 3-4	m	5 y	1	no Path/Likely Path/VUS genetic variants detected				het	DOCK8(NM_203447):c.1453T>C; p.Phe485Leu	VUS	PM2	recurrent wheezing, chronic spontaneous urticaria	↓ CD3 <sup>DR</sup> , ↓ CD3 <sup>DR</sup> , ↑ CD25 <sup>CD127</sup> low, ↓ CD3 <sup>CD8</sup> , ↑ CD45RA <sup>CD62L</sup> , ↓ CD45RA <sup>CD62L</sup> , ↓ CD19 <sup>CD27</sup> IgD, ↑ CD3 <sup>CD16</sup> /CD56
4	ID 4-4	m	6 y 2 mo	2	het	MEFV(NM_000243):c.2177T>C; p.Val726Ala (pat)	Likely Path	PP5, PM1	het	MBL2(NM_000242):c.154C>T; p.Arg52Cys	VUS	PP5	recurrent respiratory infections with wheezing, tic disorder, EEG abnormalities	↓ CD3 <sup>DR</sup> , ↓ CD3 <sup>DR</sup> , ↓ CD3 <sup>CD4</sup> , ↑ CD45RA <sup>CD62L</sup> , ↑ CD45RA <sup>CD62L</sup> , ↑ CD35 <sup>CD127</sup> low, ↓ CD3 <sup>CD8</sup> , ↑ CD4/CD8, ↓ CD19+, ↑ CD3 <sup>CD16</sup> /CD56, ↑ Ige
					het	CHRNA2(NM_000747):c.2172T>G; p.Arg275Leu/ter28 (mat)	Likely Path	PM2, PPS						
					het	ADGRV1(NM_012119):c.2366T>C; p.Arg987Ala (mat)	VUS	PM2						
5	ID 5-3	m	3 y	1	het	NOD2(NM_002162):c.3019G>A; p.Leu1007ProfsTer2 (pat)	Likely Benign	BS1, BS2, PPS	het	KOBQ4(NM_019505):c.6170C>T; p.Arg568Ile (pat)	VUS	PVS1, PPS, BS2	immune thrombocytopenia with leukopenia and neutropenia, allergic rhinitis	↓ CD3 <sup>DR</sup> , ↓ CD3 <sup>DR</sup> , ↓ CD45RA <sup>CD62L</sup> , ↑ CD25 <sup>CD127</sup> low, ↓ CD3 <sup>CD8</sup> , ↑ CD4/CD8, ↑ CA
					het	NLRP12(NM_001277):c.2120G>C; p.Phe402Leu (pat)	Benign	BS1, BP1, BP6						
6	ID 6-3	m	12 y	3	het	HRF2BP2(NM_182972):c.1021G>A; p.Ala311Thr (mat)	VUS	PM2, BP1	het	MBL2(NM_000242):c.161G>A; p.Gly544Asp	VUS	BP6, PPS	asthma, disruptive behavior pattern with features of ADHD	↓ CD3 <sup>DR</sup> , ↓ CD3 <sup>DR</sup> , ↓ CD3 <sup>CD4</sup> , ↓ CD3 <sup>CD8</sup> , ↑ CD45RA <sup>CD62L</sup> , ↑ CD45RA <sup>CD62L</sup> , ↑ CD3 <sup>CD16</sup> /CD56, ↓ CA, ↑ Ige4
7	ID 7-3	m	2 y	1	no Path/Likely Path/VUS genetic variants detected				het	no Path/Likely Path/VUS genetic variants detected			recurrent wheezing	↓ CD3 <sup>DR</sup> , ↓ CD3 <sup>DR</sup> , ↓ CD3 <sup>CD4</sup> , ↓ CD3 <sup>CD8</sup> , ↓ CD45RA <sup>CD62L</sup> , ↓ CD45RA <sup>CD62L</sup> , ↓ CD25 <sup>CD127</sup> low, ↓ CD3 <sup>CD8</sup> , ↑ CD4/CD8, ↓ CD19+, ↑ CD3 <sup>CD16</sup> /CD56, ↓ CA, ↑ Ige3, ↑ Ige4
8	ID 8-3	m	2 y	1	het	TNFRSF1A(NM_001065):c.362G>A; p.Arg121Gln (mat)	VUS	BS1, BS2, PM1, PMS, PPS	het	MBL2(NM_000242):c.161G>A; p.Gly544Asp	VUS	BP6, PPS	recurrent respiratory infections	↓ CD3 <sup>DR</sup> , ↓ CD3 <sup>DR</sup> , ↓ CD3 <sup>CD4</sup> , ↓ CD3 <sup>CD8</sup> , ↑ CD45RA <sup>CD62L</sup> , ↑ CD45RA <sup>CD62L</sup> , ↑ CD3 <sup>CD16</sup> /CD56, ↓ CA, ↑ Ige4
9	ID 9-3	f	12 y	3	no Path/Likely Path/VUS genetic variants detected				homoz	MBL2(NM_000242):c.161G>A; p.Gly544Asp	VUS	BP6, PPS	immune thrombocytopenia	↑ CD3 <sup>CD4</sup> , ↑ CD45RA <sup>CD62L</sup> , ↑ CD3 <sup>CD8</sup> , ↑ CD45RA <sup>CD62L</sup> , ↑ CD45RA <sup>CD62L</sup> , ↑ CD3 <sup>CD16</sup> /CD56
10	ID 10-3	m	8 y 2 mo	2	het	STAT3(NM_139276):c.1385T>F>G (mat)	Likely Path	PVS1, PM2	het	MBL2(NM_000242):c.154C>T; p.Arg52Cys	VUS	PP5	allergic rhinitis, atopic dermatitis, hypopituitarism, recurrent fever and tonsillitis	↑ CD3 <sup>CD4</sup> , ↑ CD45RA <sup>CD62L</sup> , ↑ CD3 <sup>CD8</sup> , ↑ CD45RA <sup>CD62L</sup> , ↑ CD3 <sup>CD16</sup> /CD56
					het	NOD2(NM_002162):c.922C>T; p.Leu388Phe	VUS	PP3, PM2	het	MBL2(NM_000242):c.161G>A; p.Gly544Asp	VUS	BP6, PPS		
					het	IGS5(NM_002184):c.573G>C; p.Pro191Ile	VUS	PM2, BP1						
11	ID 11-3	m	5 y 10 mo	1	no Path/Likely Path/VUS genetic variants detected				het	no Path/Likely Path/VUS genetic variants detected			congenital megarureter and vesicoureteral reflux, recurrent hiccups, iridocyclitis, and keratitis	↑ CD3 <sup>DR</sup> , ↑ CD3 <sup>DR</sup> , ↑ CD45RA <sup>CD62L</sup> , ↑ CD45RA <sup>CD62L</sup> , ↑ Ige
12	ID 12-3	m	5 y 6 mo	1	het	MEFV(NM_000243):c.2080A>G; p.Met694Val (mat)	Path	PP5, PM1, PM2, PMS	het	HBB(NM_000515):c.118C>T; p.Gln40Ile (mat)	Path	PVS1, PPS, PM2	immune thrombocytopenia	↓ CD3 <sup>DR</sup> , ↓ CD3 <sup>DR</sup> , ↓ CD3 <sup>CD4</sup> , ↓ CD3 <sup>CD8</sup> , ↑ CD45RA <sup>CD62L</sup> , ↓ CD45RA <sup>CD62L</sup> , ↓ CD25 <sup>CD127</sup> low, ↓ CD3 <sup>CD8</sup> , ↑ CD4/CD8, ↓ CD19+, ↑ CD3 <sup>CD16</sup> /CD56, ↑ IgeM, ↓ CA
13	ID 13-3	m	8 y 2 mo	2	het	MEFV(NM_000243):c.605G>A; p.Arg202Gln (mat)	Benign	BS1, BP4, BP6, BS3	het	HBB(NM_000515):c.82G>T; p.Ala28Ser (pat)	VUS	PP5, BP1	asthma, thalassemia minor	↓ CD3 <sup>DR</sup> , ↓ CD3 <sup>DR</sup> , ↓ CD3 <sup>CD4</sup> , ↓ CD3 <sup>CD8</sup> , ↑ CD45RA <sup>CD62L</sup> , ↑ CD45RA <sup>CD62L</sup> , ↑ CD25 <sup>CD127</sup> low, ↓ CD3 <sup>CD8</sup> , ↑ CD4/CD8, ↓ CD19+, ↑ CD3 <sup>CD16</sup> /CD56, ↑ IgeM, ↓ CA
					het	NLRP2(NM_001277):c.2120G>C; p.Phe402Leu (pat)	Likely Benign	BS2, BP1, BP4	het	MBL2(NM_000514):c.82G>T; p.Ala28Ser (pat)	VUS	PP5, BP1		
					het	RELN(NM_002375):c.1168G>A; p.Ala330Thr (pat)	VUS	PM2, BP1	het	MBL2(NM_000242):c.161G>A; p.Gly544Asp	VUS	BP6, PPS		
14	ID 14-3	f	4 y	1	het	PKnox1(NM_005026):c.521G>C; p.Val212Ala (mat)	VUS	PP5	het	no Path/Likely Path/VUS genetic variants detected			asthma	↑ CD3 <sup>DR</sup> , ↑ CD45RA <sup>CD62L</sup> , ↑ CD45RA <sup>CD62L</sup> , ↑ CD3 <sup>CD8</sup> , ↑ CD19+, ↑ CD19 <sup>CD27</sup> IgD, ↑ CD19 <sup>CD27</sup> IgD, ↑ CD3 <sup>CD16</sup> /CD56, ↑ CA
15	ID 15-3	m	9 y 8 mo	2	het	PKnox1(NM_005026):c.521G>C; p.Val212Ala (mat)	VUS	PP5	het	no Path/Likely Path/VUS genetic variants detected			asthma, cow's milk protein allergy, neurodevelopmental delay, ataxic cerebral palsy, expressive speech disorder	↑ CD3 <sup>DR</sup> , ↑ CD3 <sup>DR</sup> , ↑ CD45RA <sup>CD62L</sup> , ↑ CD25 <sup>CD127</sup> low, ↑ CD45RA <sup>CD62L</sup> , ↑ CD19+, ↑ CD19 <sup>CD27</sup> IgD, ↑ CD19 <sup>CD27</sup> IgD, ↑ Ige
					het	LRBA(NM_006726):c.2674G>A; p.Ala892Ile	VUS	PP5	het	no Path/Likely Path/VUS genetic variants detected				
					het	TTC7A(NM_020458):c.2564C>G; p.Pro855Arg	VUS	PP5	het	no Path/Likely Path/VUS genetic variants detected				
					het	L10RA(NM_001558):c.4188C>G; p.Leu298Val	VUS	PP5	het	no Path/Likely Path/VUS genetic variants detected				
16	ID 16-3	f	8 y 9 mo	2	het	MEFV(NM_000243):c.442G>C; p.Pro138Gln	VUS	PM5, PPS	het	MBL2(NM_000242):c.161G>A; p.Gly544Asp	VUS	BP6, PPS	asthma, atopic dermatitis, food allergy	↓ CD3 <sup>DR</sup> , ↓ CD3 <sup>DR</sup> , ↓ CD3 <sup>CD8</sup> , ↑ CD45RA <sup>CD62L</sup> , ↑ CD45RA <sup>CD62L</sup> , ↑ CD19 <sup>CD27</sup> IgD, ↑ CD3 <sup>CD16</sup> /CD56, ↑ Ige
					het	TNFRSF13B(NM_012452):c.310T>C; p.Cys104Arg (mat)	Path	PMS1, PPS	het	no Path/Likely Path/VUS genetic variants detected				
					het	IFIH1(NM_022168):c.432C>T; p.Pro141Gln	VUS	PVS1, BS2	het	no Path/Likely Path/VUS genetic variants detected				
					het	IFIH1(NM_022168):c.432C>T; p.Pro141Gln	Benign	PM2, BP6	het	no Path/Likely Path/VUS genetic variants detected				
17	ID 17-3	m	2 y 10 mo	1	het	MEFV(NM_000243):c.2080A>G; p.Met694Val (mat)	Path	PM1, PM5, PP5	het	no Path/Likely Path/VUS genetic variants detected			asthma, atopic dermatitis, chronic spontaneous urticaria	↓ CD3 <sup>DR</sup> , ↓ CD3 <sup>DR</sup> , ↓ CD45RA <sup>CD62L</sup> , ↑ CD45RA <sup>CD62L</sup> , ↑ CD45RA <sup>CD62L</sup> , ↑ CD45RA <sup>CD62L</sup> , ↑ CD25 <sup>CD127</sup> low, ↓ CD3 <sup>CD8</sup> , ↑ CD4/CD8, ↑ CD3 <sup>CD16</sup> /CD56, ↑ Ige
					het	UGS1(NM_000238):c.296C>T; p.Pro99Leu	VUS	PP5	het	no Path/Likely Path/VUS genetic variants detected				
					het	NCFE1(NM_015331):c.896T>G>A	VUS	PP5	het	no Path/Likely Path/VUS genetic variants detected				
18	ID 18-3	f	6 y 2 mo	2	het	PS1P1P(NM_003978):c.1150G>A; p.Asp384Asn (mat)	Likely Path	PP3, PM5, PM2	het	no Path/Likely Path/VUS genetic variants detected			asthma, craniosynostosis, neurodevelopmental delay, febrile seizure	↑ CD3 <sup>DR</sup> , ↑ CD3 <sup>CD8</sup>
					het	NOD2(NM_002162):c.3019G>A; p.Leu1007ProfsTer2	Likely Benign	BS1, BS2, PPS	het	no Path/Likely Path/VUS genetic variants detected				
19	ID 20-3	f	8 y 4 mo	2	het	NOD2(NM_002162):c.3019G>A; p.Leu1007ProfsTer2	Likely Benign	BS1, BS2, PPS	het	ONTNAP1(NM_002162):c.1062G>T; p.Lys135Asn (de novo)	VUS	PM2, BP1	developmental delay and behavioral characteristics/stereotypies/	↓ CD3 <sup>DR</sup> , ↓ CD3 <sup>DR</sup> , ↓ CD3 <sup>CD4</sup> , ↑ CD45RA <sup>CD62L</sup> , ↑ CD25 <sup>CD127</sup> low, ↓ CD3 <sup>CD8</sup> , ↑ CD4/CD8, ↓ CD19 <sup>CD27</sup> IgD, ↑ CD3 <sup>CD16</sup> /CD56, ↑ Ige, ↑ CA
					het	OPN1(NM_001040):c.1705G>A	VUS	PP5	het	no Path/Likely Path/VUS genetic variants detected				
20	ID 21-3	m	10 y 1 mo	2	het	STAT1(NM_007315):c.1341C>A; p.Arg447Glu (pat)	VUS	PM1, PM2	het	no Path/Likely Path/VUS genetic variants detected			asthma, allergic rhinitis	↑ CD3 <sup>DR</sup> , ↓ CD3 <sup>CD4</sup> , ↓ CD3 <sup>CD8</sup> , ↑ CD3 <sup>CD16</sup> /CD56, ↑ Ige
					hemi	DKC1(NM_001363):c.244G>C; p.Glu82Gln (mat)	VUS	PM1, PM2	het	no Path/Likely Path/VUS genetic variants detected				
21	ID 22-3	m	15 y	3	het	TNFR2(NM_001092):c.282C>G; p.Pro142Ala (mat)	VUS	PM2	het	MBL2(NM_000242):c.161G>A; p.Gly544Asp	VUS	BP6, PPS	anaemia	n.a.
					het	KIF23(NM_138555):c.2867T>C (mat)	Likely Path	PVS1, PM2	het	no Path/Likely Path/VUS genetic variants detected				
					het	THBD(NM_000363):c.836G>C; p.Ser287Ile	VUS	PM2	het	no Path/Likely Path/VUS genetic variants detected				
					het	NOD2(NM_002162):c.3019G>A; p.Leu1007ProfsTer2	Benign	BS1, BS2, BS3, BP4, PPS	het	no Path/Likely Path/VUS genetic variants detected				
22	ID 23-3	f	3 y	1	het	FOXN1(NM_003593):c.497C>T; p.Ala166Val (mat)	VUS	PM2	het	MBL2(NM_000242):c.154C>T; p.Arg52Cys	VUS	PP5	recurrent wheezing	↓ CD3 <sup>DR</sup> , ↓ CD3 <sup>CD4</sup> , ↓ CD4/CD8, ↑ CD3 <sup>CD16</sup> /CD56
					het	GGPD(NM_001423):c.1360C>T; p.Arg454Cys	Path	PP3, PPS, PM1, PMS	het	DOCK2(NM_004946):c.488G>A; p.Arg1629Ile	VUS	PM2, BP1		
23	ID 24-3	f	10 y 6 mo	2	het	NOD2(NM_002162):c.3105C>T; p.Arg102Ile (pat)	VUS	BS1, BS2, BS3, BP1, BP4, BP6, PPS	het	ACAD8(NM_014384):c.455T>C; p.Met152Ile	Likely Path	PS3, PPS, PM2	atopic dermatitis	↓ CD3 <sup>DR</sup> , ↓ CD3 <sup>DR</sup> , ↓ CD3 <sup>CD8</sup>
					het	FLG(NM_002016):c.7398C>T; p.Arg2447Ile (pat)	Path	PP5, PM2, PM2	het	TSHB(NM_000369):c.484C>G; p.Pro162Ala	Path	PP5, PS2, PM2, PMS		
24	ID 25-3	f	2 y	1	het	CARD11(NM_032415):c.6558C>G; p.Glu188Asp (mat)	VUS	PM2	het	RANBP2(NM_006267):c.8395A>G; p.Ile2799Val	VUS	PM2, BP1	periodic fevers with aphthous stomatitis, pharyngitis, and adenitis	↑ CD3 <sup>DR</sup> , ↓ CD3 <sup>CD8</sup> , ↑ CD45RA <sup>CD62L</sup> , ↓ CD19+, ↑ CD3 <sup>CD16</sup> /CD56, ↑ CA, ↑ Ige4
25	ID 26-3	m	3 y	1	no Path/Likely Path/VUS genetic variants detected				het	DOCK8(NM_203447):c.1178G>A; p.Arg393His	VUS	PM2, BP1	atopic dermatitis, food allergy, chronic spontaneous urticaria, febrile seizures	↓ CD3 <sup>DR</sup> , ↓ CD3 <sup>CD4</sup> , ↓ CD45RA <sup>CD62L</sup> , ↑ CD45RA <sup>CD62L</sup> , ↑ CD45RA <sup>CD62L</sup> , ↑ CD25 <sup>CD127</sup> low, ↓ CD3 <sup>CD8</sup> , ↑ CD4/CD8, ↑ CD3 <sup>CD16</sup> /CD56, ↑ IgeM, ↑ Ige, ↑ Ige4
					het	HRF(NM_000410):c.187C>G; p.His64Asp (pat)	Likely Path	PS1, PS3, PPS, PM1	het	no Path/Likely Path/VUS genetic variants detected				
					het	HRF(NM_000410):c.845G>A; p.Cys282Ile	Likely Path	PS1, PS3, PPS	het	no Path/Likely Path/VUS genetic variants detected				
26	ID 27-3	f	16 y 11 mo	3	het	DNMT3B(NM_000897):c.181T>A; p.Gln66Glu	Likely Path	PM1, PM2, PP3, PPS	het	no Path/Likely Path/VUS genetic variants detected			JA pneumococcal meningitis	↓ CD3 <sup>DR</sup> , ↓ CD3 <sup>DR</sup> , ↓ CD3 <sup>CD4</sup> , ↓ CD45RA <sup>CD62L</sup> , ↑ CD3 <sup>CD8</sup> , ↑ CD4/CD8, ↓ CD19+, ↑ CD19 <sup>CD27</sup> IgD, ↑ CD19 <sup>CD27</sup> IgD, ↓ CD3 <sup>CD16</sup> /CD56, ↑ IgeM, ↓ IgeA, ↓ IgeB, ↓ Ige4
					het	DNMT3B(NM_000897):c.181T>A; p.Gln66Glu	Likely Path	PM1, PM2, PP3, PPS	het	no Path/Likely Path/VUS genetic variants detected				
27	ID 28-3	m	16 y	3	het	CARD11(NM_032415):c.6558C>G; p.Glu188Asp (mat)	VUS	PM2	het	MBL2(NM_000242):c.161G>A; p.Gly544Asp	VUS	BP6, PPS	immune thrombocytopenia	↓ CD3 <sup>DR</sup> , ↓ CD3 <sup>CD4</sup> , ↓ CD45RA <sup>CD62L</sup> , ↑ CD25 <sup>CD127</sup> low, ↓ CD3 <sup>CD8</sup> , ↑ CD45RA <sup>CD62L</sup> , ↑ CD19+, ↑ CD19 <sup>CD27</sup> IgD, ↑ Ige
					het	TUBB1(NM_030773):c.326G>A; p.Gly109Glu (pat)	VUS	PP4, PPS	het	DOCK8(NM_203447):c.2428G>A; p.Ala810Ile (pat)	VUS	PM2, BP1	recurrent tonsillitis	↓ CD3 <sup>DR</sup> , ↓ CD3 <sup>DR</sup> , ↓ CD3 <sup>CD4</sup> , ↑ CD45RA <sup>CD62L</sup> , ↑ CD45RA <sup>CD62L</sup> , ↑ CD3 <sup>CD8</sup> , ↑ CD45RA <sup>CD62L</sup> , ↑ CD4/CD8, ↑ CD19+
28	ID 29-3	m	2 y 6 mo	1	no Path/Likely Path/VUS genetic variants detected				het	DOCK8(NM_203447):c.488G>A; p.Arg1629Ile	VUS	PM2, BP1		
					het	DOCK8(NM_203447):c.488G>A; p.Arg1629Ile	VUS	PM2, BP1	het	MBL2(NM_000242):c.154C>T; p.Arg52Cys	VUS	PP5		
					het	RANBP2(NM_006267):c.8395A>G; p.Ile2799Val	VUS	PM2, BP1	het	no Path/Likely Path				

