

Supplementary Table 2: Variants in Known PID Genes

Proband Number	Age (years)	Sex	Country	Consang.	NGS Technology	Gene	cDNA	AA	Zygoty	Mutation Type	CADD phred	Variant Classification*
1	5	M	Egypt	Yes	Panel	<i>ACPS</i>	c.800C>A	p.Ser267Ter	homozygous	nonsense	.	Pathogenic
2	0.6	M	Algeria	Yes	Panel	<i>ADA</i>	c.965T>C	p.Phe322Ser	homozygous	missense	32.0	VUS
3	0.3	F	Egypt	Yes	Panel	<i>ADA</i>	c.956_960delAAGAG	p.Glu319Glyfs	homozygous	nonsense	.	Pathogenic
4	0.3	M	Egypt	Yes	Panel	<i>ADA</i>	c.956_960delAAGAG	p.Glu319Glyfs	homozygous	frameshift	.	Pathogenic
5	1	F	Egypt	Yes	Panel	<i>ADA</i>	c.467G>A	p.Arg156His	homozygous	missense	32.0	Pathogenic
6	1	M	Egypt	Yes	Panel	<i>ADA</i>	c.845G>A	p.Arg282Gln	homozygous	missense	33.0	Pathogenic
7	2	F	Egypt	Yes	Panel	<i>ADA</i>	c.467G>A	p.Arg156His	homozygous	missense	32.0	Pathogenic
8	0.3	M	Egypt	Yes	Panel	<i>ADA</i>	c.466C>T	p.Arg156Cys	homozygous	missense	32.0	Pathogenic
9	0.3	M	Iran	Yes	Panel	<i>ADA</i>	c.415G>T	p.Glu139Ter	homozygous	nonsense	.	Pathogenic
10	0.3	M	Iran	Yes	Panel	<i>ADA</i>	c.704G>A	p.Arg235Gln	homozygous	missense	31.0	Pathogenic
11	4	F	Iran	Yes	Panel	<i>ADA</i>	c.845G>A	p.Arg282Gln	homozygous	missense	33.0	Pathogenic
12	0.3	M	Jordan	Yes	Panel	<i>ADA</i>	c.956_960delAAGAG	p.Glu319Glyfs	homozygous	frameshift	.	Pathogenic
13	0.3	M	Lebanon	Yes	Panel	<i>ADA</i>	c.996_997del	p.Ser333Phefs	homozygous	frameshift	.	Pathogenic
14	0.2	M	Pakistan	Yes	Panel	<i>ADA</i>	c.716G>A	p.Gly239Asp	homozygous	missense	28.2	Pathogenic
15	12	M	Turkey	Yes	Panel	<i>ADA</i>	c.385G>A	p.Val129Met	homozygous	missense	27.9	Pathogenic
16	1	F	Iran	Yes	Panel	<i>ADA</i>	c.445C>T	p.Arg149Trp	homozygous	missense	28.3	Pathogenic
17	0.5	F	Iran	Yes	Panel	<i>ADA</i>	c.821C>T	p.Pro274Leu	homozygous	missense	17.0	Pathogenic
18	2	M	Egypt	Yes	Panel	<i>ADA2</i>	c.533T>C	p.Phe178Ser	homozygous	missense	25.9	Pathogenic
19	1	M	Egypt	Yes	Panel	<i>ADA2</i>	c.1514T>C	p.Ile505Thr	homozygous	missense	22.6	VUS
20	17	F	Lebanon	Yes	Panel	<i>ADA2</i>	c.1271_1277delAGGCTGA	p.Lys424fs	homozygous	frameshift	.	Pathogenic
22	16	F	Turkey	Yes	Panel	<i>ADA2</i>	c.137dupT	p.Arg49Alafs	homozygous	frameshift	.	Pathogenic
23	15	M	Turkey	Yes	Panel	<i>ADA2</i>	c.139G>A	p.Gly47Arg	homozygous	missense	23.6	Pathogenic
24	14	M	Lebanon	Yes	Panel	<i>ADA2</i>	c.962G>A	p.Gly321Glu	homozygous	missense	21.5	Pathogenic
25	4	M	Egypt	Yes	Panel	<i>AICDA</i>	c.331G>A	p.Ala111Thr	homozygous	missense	25.9	Pathogenic
26	12	M	Iran	Yes	Panel	<i>AICDA</i>	c.428-1G>T	N/A	homozygous	splice site	.	Pathogenic
27	6	M	Iran	Yes	Panel	<i>AICDA</i>	c.334C>T	p.Arg112Cys	homozygous	missense	31.0	Pathogenic
28	3	F	Egypt	Yes	Panel	<i>AIRE</i>	c.47C>T	p.Thr16Met	homozygous	missense	25.8	Pathogenic
29	3	F	Palestinian Territories	Yes	Panel	<i>AIRE</i>	c.274C>T	p.Arg92Trp	homozygous	missense	25.6	Pathogenic
30	16	M	Lebanon	Yes	WES	<i>AK2</i>	c.298G>A	p.Gly100Ser	homozygous	missense	31.0	Pathogenic
31	0.8	M	Lebanon	Yes	Panel	<i>AK2</i>	c.298G>A	p.Gly100Ser	homozygous	missense	31.0	Pathogenic
32	3	F	Kazakhstan	Yes	Panel	<i>AP3B1</i>	c.3158G>A	p.Gly1053Glu	homozygous	missense	23.1	VUS
33	16	F	Turkey	Yes	Panel	<i>AP3B1</i>	c.469_472delGAAG	p.Glu157fs	homozygous	frameshift	.	Pathogenic
34	1	F	Egypt	Yes	WES	<i>ARPC1B</i>	c.91G>T	p.Glu31Ter	homozygous	nonsense	.	Pathogenic
35	4	M	Egypt	Yes	Panel	<i>ATM</i>	c.4864G>T	p.Glu1622Ter	homozygous	nonsense	.	Pathogenic
36	4	M	Egypt	Yes	Panel	<i>ATM</i>	c.1235+1G>A	N/A	homozygous	splice site	.	Pathogenic
37	5	F	Egypt	No	Panel	<i>ATM</i>	c.6259_6259delG	p.Ser2116fs	compound het	frameshift	.	Pathogenic
						<i>ATM</i>	c.1388C>A	p.Ala463Glu		missense	24.6	Pathogenic
38	4	M	Iran	Yes	Panel	<i>ATM</i>	c.3895delG	p.Ala1299fs	homozygous	frameshift	.	Pathogenic
39	5	F	Lebanon	Yes	Panel	<i>ATM</i>	c.8385_8394delTTTCAGTGCC	p.Phe2799fs	homozygous	frameshift	.	Pathogenic
40	5	M	Peru	No	Panel	<i>ATM</i>	del entre gene	del entre gene	homozygous	deletion	.	Pathogenic
41	4	F	Qatar	Yes	Panel	<i>ATM</i>	c.5243_5244insT	p.Trp1750fs	homozygous	frameshift	.	Pathogenic
42	0.7	M	Turkey	Yes	Panel	<i>ATM</i>	c.67C>T	p.Arg23Ter	homozygous	nonsense	.	Pathogenic
43	4	F	United Arab Emirates	Yes	Panel	<i>ATM</i>	c.3104_3107delTATT	p.Ile1035fs	homozygous	frameshift	.	Pathogenic
44	1	M	Iran	No	Panel	<i>BTK</i>	c.773delA	p.Asn258Metfs	hemizygous	frameshift	.	Pathogenic
45	3	M	Kazakhstan	No	Panel	<i>BTK</i>	c.1632-1G>C	N/A	hemizygous	splice site	.	Pathogenic
46	13	M	Peru	No	Panel	<i>BTK</i>	c.894+2dupT	N/A	hemizygous	splice site	.	Pathogenic
47	11	M	United States	No	Panel	<i>BTK</i>	c.1684C>T	p.Arg562Trp	hemizygous	missense	28.0	Pathogenic
48	5	M	Egypt	Yes	Panel	<i>BTK</i>	c.906_908delIAGG	p.Gly303del	hemizygous	frameshift	.	Pathogenic
49	11	M	Egypt	Yes	Panel	<i>BTK</i>	c.82C>T	p.Arg28Cys	hemizygous	missense	28.5	Pathogenic
50	10	M	Egypt	Yes	Panel	<i>BTK</i>	c.763C>T	p.Arg255Ter	hemizygous	nonsense	.	Pathogenic
51	1	M	Egypt	Yes	Panel	<i>BTK</i>	c.494-2A>G	N/A	hemizygous	splice site	.	Pathogenic
52	4	M	Egypt	No	Panel	<i>BTK</i>	c.1085A>T	p.His362Leu	hemizygous	missense	28.6	Pathogenic
53	12	M	Iran	No	Panel	<i>BTK</i>	c.1631G>T	p.Arg544Met	hemizygous	missense	34.0	Pathogenic

54	15	M	Iran	No	Panel	<i>BTK</i>	c.1574G>A	p.Arg525Gln	hemizygous	missense	32.0	Pathogenic
55	8	M	Iran	Yes	Panel	<i>BTK</i>	c.1669-2A>G	N/A	hemizygous	splice site	.	Pathogenic
56	5	M	Iran	No	Panel	<i>BTK</i>	c.1543A>T	p.Lys515Ter	hemizygous	nonsense	.	Pathogenic
57	3	M	Iran	No	Panel	<i>BTK</i>	c.1526T>A	p.Met509Lys	hemizygous	missense	27.1	Pathogenic
58	2	M	Iran	No	Panel	<i>BTK</i>	c.1522G>A	p.Ala508Thr	hemizygous	missense	28.2	Pathogenic
59	6	M	Iran	No	Panel	<i>BTK</i>	c.1475delG	p.Arg492fs	hemizygous	frameshift	.	Pathogenic
60	2	M	Iran	Yes	Panel	<i>BTK</i>	c.1697C>A	p.Pro566Gln	hemizygous	missense	28.3	Pathogenic
61	1	M	Iran	No	Panel	<i>BTK</i>	c.1184G>A	p.Trp395Ter	hemizygous	nonsense	.	Pathogenic
62	1	M	Iran	Yes	Panel	<i>BTK</i>	c.773delA	p.Asn258Metfs	hemizygous	frameshift	.	Pathogenic
63	2	M	Iran	No	Panel	<i>BTK</i>	c.810delT	p.Glu271Lysfs	hemizygous	frameshift	.	Pathogenic
64	1	M	Iran	Yes	Panel	<i>BTK</i>	c.1239T>A	p.Phe413Leu	hemizygous	missense	24.3	Pathogenic
65	27	M	Kuwait	Yes	Panel	<i>BTK</i>	del exons 15-16	del exons 15-16	hemizygous	deletion	.	Pathogenic
66	7	M	Lebanon	No	Panel	<i>BTK</i>	c.763C>T	p.Arg255Ter	hemizygous	nonsense	.	Pathogenic
67	7	M	Pakistan	Yes	Panel	<i>BTK</i>	c.240+1G>T	N/A	hemizygous	splice site	.	Pathogenic
68	1	M	Turkey	No	Panel	<i>BTK</i>	c.1240G>T	p.Gly414Trp	hemizygous	missense	31.0	Pathogenic
69	1	M	Pakistan	Yes	WES	<i>BTK</i>	c.1883delC	p.Thr628fs	hemizygous	frameshift	.	Pathogenic
70	10	M	Egypt	No	Panel	<i>BTK</i>	c.1684C>T	p.Arg562Trp	hemizygous	missense	28.0	Pathogenic
71	9	M	Kazakhstan	No	Panel	<i>BTK</i>	c.83G>A	p.Arg28His	hemizygous	missense	28.6	Pathogenic
72	5	M	Pakistan	Yes	Panel	<i>C3</i>	c.1270-1G>T	N/A	homozygous	splice site	.	Pathogenic
73	1	F	Algeria	Yes	Panel	<i>CARD11</i>	c.1682_1683insC	p.Met561Ilefs	homozygous	frameshift	.	Pathogenic
74	0.7	F	Turkey	Yes	Panel	<i>CARD11</i>	c.1682_1683insC	p.Met561fs	homozygous	frameshift	.	Pathogenic
75	9	M	Italy	No	Panel	<i>CARD11</i>	c.169G>A	p.Glu57Lys	heterozygous	missense	29.1	Pathogenic
76	4	F	Turkey	Yes	Panel	<i>CARD11</i>	c.1682_1683insC	p.Met561Ilefs	homozygous	frameshift	.	Pathogenic
77	17	M	Turkey	No	Panel	<i>CARD9</i>	c.323-1G>A	N/A	compound het	splice site	.	Pathogenic
78	12	M	Egypt	Yes	Panel	<i>CARD9</i>	c.208C>T	p.Arg70Trp	homozygous	missense	23.5	Pathogenic
79	0.6	F	Peru	Yes	Panel	<i>CARMIL2</i>	c.865C>T	p.Gln289Ter	homozygous	nonsense	.	Pathogenic
80	3	M	Lebanon	Yes	Panel	<i>CASP8</i>	c.2509_2510insA	p.Val839Glyfs	homozygous	frameshift	.	Pathogenic
81	12	M	Italy	Yes	Panel	<i>CCBE1</i>	c.710A>G	p.Gln237Arg	homozygous	missense	23.1	VUS
82	10	M	Lebanon	Yes	Panel	<i>CD27</i>	c.767G>A	p.Gly256Asp	homozygous	missense	26.2	VUS
83	0.3	M	Iran	Yes	Panel	<i>CD27</i>	c.158G>A	p.Cys53Tyr	homozygous	missense	26.4	Pathogenic
84	0.4	F	Lebanon	Yes	Panel	<i>CD3D</i>	c.247G>T	p.Glu83Ter	homozygous	nonsense	.	Pathogenic
85	0.4	F	Lebanon	Yes	Panel	<i>CD3E</i>	c.173_173delT	p.Leu58fs	homozygous	frameshift	.	Pathogenic
86	0.9	M	Lebanon	Yes	Panel	<i>CD3E</i>	c.173_173delT	p.Leu58fs	homozygous	frameshift	.	Pathogenic
87	0.7	M	Lebanon	Yes	Panel	<i>CD3E</i>	c.173delT	p.Leu58fs	homozygous	frameshift	.	Pathogenic
88	1	F	Lebanon	Yes	Panel	<i>CD3E</i>	c.173delT	p.Leu58Hisfs	homozygous	frameshift	.	Pathogenic
89	0.5	F	Greece	Yes	WES	<i>CD3E</i>	c.104-1G>A	N/A	homozygous	splice site	.	Pathogenic
90	3	M	Algeria	No	Panel	<i>CD3G</i>	c.18delC	p.Leu7fs	homozygous	frameshift	.	Pathogenic
91	5	M	Iran	No	Panel	<i>CD40LG</i>	c.251G>C	p.Cys84Ser	hemizygous	missense	25.3	Pathogenic
92	8	M	Egypt	Yes	Panel	<i>CD40LG</i>	c.346G>T	p.Gly116Cys	hemizygous	missense	35.0	Pathogenic
93	3	M	Iran	Yes	Panel	<i>CD40LG</i>	c.431G>T	p.Gly144Val	hemizygous	missense	25.1	Pathogenic
94	3	M	Iran	Yes	Panel	<i>CD40LG</i>	c.266delG	p.Ser89fs	hemizygous	frameshift	.	Pathogenic
95	12	M	Peru	No	Panel	<i>CD40LG</i>	c.361C>T	p.Gln121Ter	hemizygous	nonsense	.	Pathogenic
96	0.8	M	Peru	No	Panel	<i>CD40LG</i>	c.770G>T	p.Gly257Val	hemizygous	missense	26.6	Pathogenic
97	2	M	Peru	No	Panel	<i>CD40LG</i>	c.770G>T	p.Gly257Val	hemizygous	missense	26.6	Pathogenic
98	1	M	Greece	Yes	Panel	<i>CD40LG</i>	c.334G>T	p.Glu112Ter	hemizygous	nonsense	.	Pathogenic
99	2	M	Egypt	Yes	Panel	<i>CD40LG</i>	c.208C>T	p.Gln70Ter	hemizygous	nonsense	.	Pathogenic
100	4	M	Turkey	Yes	Panel	<i>CD40LG</i>	c.761C>T	p.Thr254Met	hemizygous	missense	26.3	Pathogenic
101	0.3	M	Turkey	No	Panel	<i>CD40LG</i>	c.155A>C	p.Lys52Trp	hemizygous	missense	29.6	Pathogenic
102	11	F	United Arab Emirates	Yes	Panel	<i>CD79A</i>	c.442G>A	p.Glu148Lys	homozygous	missense	26.3	Pathogenic
103	0.3	M	Lebanon	Yes	Panel	<i>CIITA</i>	c.1648_1649delCG	p.Arg550fs	homozygous	frameshift	.	Pathogenic
104	0.5	M	Egypt	Yes	Panel	<i>CIITA</i>	c.3377G>A	p.Arg1126Gln	homozygous	missense	26.7	VUS
105	0.1	F	Iran	Yes	Panel	<i>CIITA</i>	c.3242_3244delACA	p.Asn1082del	homozygous	in frame deletion	.	Pathogenic
106	13	F	Egypt	Yes	Panel	<i>CIITA</i>	c.1648_1649delCG	p.Arg550Glyfs	homozygous	frameshift	.	Pathogenic
107	22	F	Turkey	Yes	WES	<i>CORO1A</i>	1191_1192insC	p.Ser401fs	homozygous	frameshift	.	Pathogenic
108	1	M	Egypt	Yes	Panel	<i>CR2</i>	c.3033G>C	p.Gln1011His	homozygous	missense	24.3	Pathogenic
109	16	M	ArgentiN/A	No	Panel	<i>CTLA4</i>	c.76delT	p.Leu28Phefs	heterozygous	frameshift	.	Pathogenic
109	17	M	United States	No	Panel	<i>CTLA4</i>	c.436_437delGGinsCT	p.Gly146Leu	heterozygous	missense	24.2	Pathogenic

110	16	F	United States	No	Panel	<i>CTLA4</i>	c.347T>A	p.Ile116Asn	heterozygous	missense	26.2	Pathogenic
111	20	F	Iran	No	Panel	<i>CTLA4</i>	del entire gene	del entire gene	heterozygous	deletion	.	Pathogenic
112	9	F	Pakistan	Yes	Panel	<i>CTLA4</i>	c.545C>G	p.Thr182Arg	homozygous	missense	25.8	VUS
113	2	M	Greece	No	Panel	<i>CXCR4</i>	c.157A>C	p.Ile53Leu	heterozygous	missense	22.0	VUS
114	7	F	Lebanon	Yes	Panel	<i>CYBA</i>	c.539_539delC	p.Pro180fs	homozygous	frameshift	.	Pathogenic
115	0.8	M	Pakistan	Yes	Panel	<i>CYBB</i>	c.1222G>A	p.Gly408Arg	homozygous	missense	26.8	Pathogenic
116	0.3	F	Egypt	Yes	Panel	<i>DCLRE1C</i>	c.2041_2042insA	p.Ala681Aspfs	homozygous	nonsense	.	Pathogenic
117	0.5	F	Egypt	Yes	Panel	<i>DCLRE1C</i>	c.363-1G>C	N/A	homozygous	splice site	.	Pathogenic
118	0.5	F	Iran	Yes	Panel	<i>DCLRE1C</i>	c.632G>T	p.Gly211Val	homozygous	missense	28.6	VUS
119	0.3	M	Iran	Yes	Panel	<i>DCLRE1C</i>	c.1162G>T	p.Glu388Ter	homozygous	nonsense	.	Pathogenic
120	9	M	Jordan	Yes	Panel	<i>DCLRE1C</i>	c.1306_1307insAGGATGCT	p.Cys436Ter	homozygous	nonsense	.	Pathogenic
121	1	F	Jordan	Yes	Panel	<i>DCLRE1C</i>	c.1299_1306dupAGGAT	p.Cys436Ter	homozygous	nonsense	.	Pathogenic
122	0.5	M	Jordan	Yes	Panel	<i>DCLRE1C</i>	del exons 1-4	del exons 1-4	homozygous	deletion	.	Pathogenic
123	0.3	F	Lebanon	Yes	Panel	<i>DCLRE1C</i>	del exons 1-4	del exons 1-4	homozygous	deletion	.	Pathogenic
124	0.3	F	Pakistan	Yes	Panel	<i>DCLRE1C</i>	del exons 1-14	del exons 1-14	homozygous	deletion	.	Pathogenic
125	0.4	M	Pakistan	Yes	Panel	<i>DCLRE1C</i>	del exons 1-3	del exons 1-3	homozygous	deletion	.	Pathogenic
126	1	F	Turkey	Yes	Panel	<i>DCLRE1C</i>	del exons 1-4	del exons 1-4	homozygous	deletion	.	Pathogenic
127	5	M	Turkey	No	Panel	<i>DCLRE1C</i>	c.1350_1356delAGATTG	p.Asp451fs	heterozygous	frameshift	.	Pathogenic
128	0.6	F	Egypt	Yes	WES	<i>DCLRE1C</i>	c.787C>T	p.Arg263Ter	homozygous	nonsense	.	Pathogenic
129	11	M	Iran	Yes	Panel	<i>DCLRE1C</i> <i>INO80</i>	c.1250_1260delCAATGACTGCA c.3160delG	p.Ser417fs p.Ala1054fs	homozygous	frameshift	.	Pathogenic
130	0.3	M	Iran	Yes	Panel	<i>DCLRE1C</i>	del exon 1-4	del exon 1-4	homozygous	deletion	.	Pathogenic
131	0.2	F	Saudi Arabia	No	Panel	<i>DCLRE1C</i>	del exons 1-3	del exons 1-3	homozygous	deletion	.	Pathogenic
132	0.6	F	United Arab Emirates	Yes	Panel	<i>DCLRE1C</i>	c.571C>T	p.Arg191Ter	homozygous	nonsense	.	Pathogenic
133	0.2	M	Turkey	Yes	Panel	<i>DCLRE1C</i>	del exons 1-4	del exons 1-4	homozygous	deletion	.	Pathogenic
134	2	M	Kuwait	Yes	WES	<i>DNMT3B</i>	c.2138G>A	p.Arg713Gln	homozygous	missense	32.0	Pathogenic
135	6	M	Egypt	Yes	Panel	<i>DOCK2</i>	c.2541delC	p.Phe848fs	homozygous	nonsense	.	Pathogenic
136	0.4	F	United Arab Emirates	Yes	Panel	<i>DOCK2</i>	del exons 28-32	del exons 28-32	homozygous	deletion	.	Pathogenic
137	1	M	Iran	Yes	Panel	<i>DOCK8</i>	c.3577C>G	p.Leu1193Val	homozygous	missense	25.1	VUS
138	20	M	Iran	Yes	Panel	<i>DOCK8</i>	c.4201G>T	p.Glu1401Ter	heterozygous	nonsense	.	Pathogenic
139	9	M	Egypt	Yes	Panel	<i>DOCK8</i>	c.1422G>C c.4346C>T	p.Gln474His p.Ser1449Leu	compound het	missense	35.0 19.7	VUS VUS
140	5	M	Egypt	Yes	Panel	<i>DOCK8</i>	c.949C>T	p.Arg317Ter	homozygous	nonsense	.	Pathogenic
141	4	M	Egypt	Yes	Panel	<i>DOCK8</i>	c.3191delA	p.Asp1064Valfs	homozygous	frameshift	.	Pathogenic
142	6	F	Egypt	Yes	Panel	<i>DOCK8</i>	c.3823delA	p.Ile1275Ter	homozygous	nonsense	.	Pathogenic
143	11	F	Egypt	Yes	Panel	<i>DOCK8</i>	del exons 1-4	del exons 1-4	homozygous	deletion	.	Pathogenic
144	2	M	Egypt	Yes	Panel	<i>DOCK8</i>	c.3372_3375delTCCT	p.Ser1126fs	homozygous	frameshift	.	Pathogenic
145	4	F	Egypt	Yes	Panel	<i>DOCK8</i>	c.663delC	p.Phe222fs	homozygous	frameshift	.	Pathogenic
146	6	M	Iran	Yes	Panel	<i>DOCK8</i>	c.5132C>A	p.Ser1711Ter	homozygous	nonsense	.	Pathogenic
147	1	F	Iran	Yes	Panel	<i>DOCK8</i>	c.4106_4107insG	p.Arg1370fs	homozygous	frameshift	.	Pathogenic
148	9	M	Lebanon	Yes	Panel	<i>DOCK8</i>	del entire gene	del entire gene	homozygous	deletion	.	Pathogenic
149	3	M	Oman	Yes	Panel	<i>DOCK8</i>	c.3787delA	p.Ile1263Ter	homozygous	nonsense	.	Pathogenic
150	5	F	Pakistan	Yes	Panel	<i>DOCK8</i>	c.1529_1530delTG	p.Leu510Argfs	homozygous	frameshift	.	Pathogenic
151	0.3	F	Turkey	Yes	Panel	<i>DOCK8</i>	del exons 1-26	del exons 1-26	homozygous	deletion	.	Pathogenic
152	3	M	Saudi Arabia	Yes	WES	<i>DOCK8</i>	c.1517-1G>C	N/A	homozygous	splice site	.	Pathogenic
153	6	M	Lebanon	Yes	Panel	<i>DOCK8</i>	c.3130C>T	p.Gln1044Ter	homozygous	nonsense	.	Pathogenic
154	6	M	Iran	No	Panel	<i>DOCK8</i>	del exons 1,27 and 28	del exons 1,27 and 28	homozygous	deletion	.	Pathogenic
155	5	F	Turkey	Yes	Panel	<i>DOCK8</i>	del exons 1-26	del exons 1-26	homozygous	deletion	.	Pathogenic
156	7	F	United Arab Emirates	Yes	Panel	<i>DOCK8</i>	del exon 11	del exon 11	homozygous	deletion	.	Pathogenic
157	1	M	Egypt	Yes	Panel	<i>DOCK8</i>	c.1517-1G>C	N/A	homozygous	splice site	.	Pathogenic
158	2	M	Iran	Yes	Panel	<i>DOCK8</i>	del exons 9-12	del exons 9-12	homozygous	deletion	.	Pathogenic
159	1	F	Jordan	Yes	Panel	<i>DOCK8</i>	del exons 1-2	del exons 1-2	homozygous	deletion	.	Pathogenic
160	7	M	Egypt	Yes	Panel	<i>DOCK8</i>	c.4826_4827 delAC	p.Thr1610fs	homozygous	frameshift	.	Pathogenic
161	1	M	Lebanon	No	Panel	<i>ELANE</i>	c.416C>T	p.Pro139Leu	Heterozygous	missense	23.9	Pathogenic
162	1	F	Egypt	No	Panel	<i>EPG5</i> <i>EPG5</i>	c.3976C>T c.1261C>T	p.Gln1326Ter p.Gln421Ter	compound het	nonsense nonsense	.	Pathogenic
163	3	M	Algeria	Yes	Panel	<i>FAS</i>	c.397T>G	p.Phe133Val	homozygous	missense	23.8	VUS

164	14	M	United States	No	Panel	FAS	c.185C>G	p.Pro62Arg	heterozygous	missense	13.2	VUS
165	0.3	F	Pakistan	Yes	Panel	FERMT3	c.126delC	p.Gln367fs	homozygous	frameshift	.	Pathogenic
166	3	F	United Arab Emirates	Yes	Panel	FERMT3	c.48G>A	p.Trp16Ter	homozygous	nonsense	.	Pathogenic
167	0.2	F	Lebanon	Yes	WES	FOXP1	c.562delA	p.S188fs	homozygous	frameshift	.	Pathogenic
168	5	M	Egypt	Yes	Panel	FOXP3	c.1190G>A	p.Arg397Gln	heterozygous	missense	32.0	Pathogenic
169	0.3	M	Lebanon	No	Panel	FOXP3	c.1150G>A	p.Ala384Thr	hemizygous	missense	25.0	Pathogenic
170	4	F	Lebanon	No	Panel	GATA2	c.1243G>A	p.Glu415Lys	heterozygous	missense	23.2	Pathogenic
171	21	M	United States	No	WES	GATA2	c.1061C>T	p.Thr354Met	heterozygous	missense	31.0	Pathogenic
172	56	F	Chile	No	Panel	GATA2	c.942_943insCTGT	p.Cys316Serfs	heterozygous	frameshift	.	Pathogenic
173	2	F	Oman	Yes	Panel	GATA2	del exons 1-3	del exons 1-3	homozygous	deletion	.	Pathogenic
174	20	F	United States	No	Panel	GATA2	c.1109G>C	p.Cys370Ser	heterozygous	missense	29.5	Pathogenic
175	7	F	Egypt	Yes	Panel	HAX1	c.355delC	p.Leu119fs	homozygous	frameshift	.	Pathogenic
176	2	F	Iran	Yes	Panel	HAX1	c.130_131insA	p.Trp44Ter	homozygous	nonsense	.	Pathogenic
177	6	M	Lebanon	Yes	Panel	HAX1	c.130_131insT	p.Trp44fs	homozygous	frameshift	.	Pathogenic
178	0.7	M	Iran	Yes	Panel + WES	HYOU1	c.1331C>A	p.Pro444His	homozygous	missense	29.8	VUS
179	0.1	M	Kuwait	Yes	WES	ICOS	c.90delG	p.M30fs	homozygous	frameshift	.	Pathogenic
180	1	F	Jordan	No	Panel	IFNGR1	c.817_820delATTA	p.Asn274Hisfs	heterozygous	frameshift	.	Pathogenic
181	3	M	Peru	No	Panel	IFNGR1	c.816_822delAATTAAT	p.Asn274Hisfs	heterozygous	frameshift	.	Pathogenic
182	12	F	Egypt	Yes	Panel	IFNGR1	c.589G>A	p.Glu197Lys	heterozygous	missense	33.0	Pathogenic
183	1	F	Saudi Arabia	Yes	Panel	IFNGR2	c.537C>A	p.Tyr179Ter	homozygous	nonsense	.	Pathogenic
184**	0.3	M	Saudi Arabia	Yes	WES	IFNGR2	c.798delT	p.Cys266fs	homozygous	frameshift	.	Pathogenic
185	3	M	Turkey	Yes	Panel	IGHM	del exons 1-4	del exons 1-4	homozygous	deletion	.	Pathogenic
186	5	F	Turkey	Yes	Panel	IGHM	del exons 1-4	del exons 1-4	homozygous	deletion	.	Pathogenic
187	12	F	Argentina/A	Yes	Panel	IGLL1	c.258delG	p.Gln88Asnfs	homozygous	frameshift	.	Pathogenic
188	0.6	M	Egypt	Yes	Panel	TNFRSF13B	c.62-2A>G	N/A	homozygous	splice site	.	Pathogenic
189	1	M	Egypt	Yes	Panel	IKBK	del exon 9	del exon 9	homozygous	deletion	.	Pathogenic
190	28	M	Turkey	No	Panel	IKZF1	c.500A>G	p.His167Arg	heterozygous	missense	25.9	Pathogenic
191	0.9	M	Egypt	Yes	Panel	IL10RA	c.499T>C	p.Tyr167His	homozygous	missense	25.2	VUS
192	0.8	M	Iran	Yes	Panel	IL10RB	c.92C>T	p.Ser31Phe	homozygous	missense	29.9	VUS
193	0.9	F	Turkey	Yes	Panel	IL10RB	del exons 3-7	del exons 3-7	homozygous	deletion	.	Pathogenic
194	4	M	Iran	Yes	Panel	IL12B	c.527_528delCT	p.Ser176Cysfs	homozygous	nonsense	.	Pathogenic
195	1	M	Iran	Yes	Panel	IL12B	c.527_528delCT	p.Ser176Cysfs	homozygous	nonsense	.	Pathogenic
196	5	M	Egypt	Yes	Panel	IL12B	c.191delA	p.Gln64fs	homozygous	frameshift	.	Pathogenic
197	3	M	Iran	Yes	Panel	IL12B1	c.684_685insT	p.Val229fs	homozygous	frameshift	.	Pathogenic
198	7	F	Algeria	No	Panel	IL12RB1	c.1791+2T>G	N/A	compound het	splice site	.	Pathogenic
199	6	F	Algeria	Yes	Panel	IL12RB1	c.637C>T	p.Arg213Trp	homozygous	missense	18.3	Pathogenic
200	2	F	Egypt	Yes	Panel	IL12RB1	c.1386_1387delGT	p.Ser463fs	homozygous	frameshift	.	Pathogenic
201	3	F	Egypt	Yes	Panel	IL12RB1	c.184+2T>G	N/A	homozygous	splice site	.	Pathogenic
202	0.5	F	Iran	Yes	Panel	IL12RB1	c.1911+2T>G	N/A	homozygous	splice site	.	Pathogenic
203	0.5	F	Iran	Yes	Panel	IL12RB1	c.509T>A	p.Val170Glu	homozygous	missense	16.0	VUS
204	2	M	Iran	Yes	Panel	IL12RB1	c.440_441delTG	p.Val147Alafs	homozygous	frameshift	.	Pathogenic
205	13	F	Oman	Yes	Panel	IL12RB1	c.804_805insT	p.Val269fs	homozygous	frameshift	.	Pathogenic
206	13	F	Peru	Yes	Panel	IL12RB1	c.1911+2T>G	N/A	homozygous	splice site	.	Pathogenic
207	3	M	Turkey	Yes	Panel	IL12RB1	c.684_685insT	p.Val229fs	homozygous	frameshift	.	Pathogenic
208	0.8	F	Iran	Yes	Panel	IL17RA	c.1867delC	p.Leu623fs	homozygous	frameshift	.	Pathogenic
209	1	M	Egypt	Yes	Panel	IL1RN	c.160C>T	p.Gln54Ter	homozygous	nonsense	.	Pathogenic
210	2	M	Egypt	Yes	Panel	IL1RN	c.169C>T	p.Gln57Ter	homozygous	nonsense	.	Pathogenic
210	1	M	Peru	No	Panel	IL2RG	c.738G>A	p.Trp246Ter	homozygous	nonsense	.	Pathogenic
211	0.5	M	Jordan	No	Panel	IL2RG	c.811G>C	p.Gly271Arg	hemizygous	missense	22.8	Pathogenic
212	0.4	M	United States	No	Panel	IL2RG	c.703C>T	p.Gln235Ter	hemizygous	nonsense	.	Pathogenic
213	0.7	M	Egypt	Yes	Panel	IL2RG	c.436C>G	p.Leu146Val	hemizygous	missense	22.0	VUS
214	2	M	Egypt	Yes	Panel	IL2RG	c.670C>T	p.Arg224Trp	hemizygous	missense	32.0	Pathogenic
215	0.7	M	Egypt	Yes	Panel	IL2RG	c.676C>T	p.Arg226Cys	hemizygous	missense	29.2	Pathogenic
216	0.8	M	Egypt	Yes	Panel	IL2RG	c.670C>T	p.Arg224Trp	hemizygous	missense	32.0	Pathogenic
217	0.3	M	Iran	Yes	Panel	IL2RG	c.850G>T	p.Glu284Ter	hemizygous	nonsense	.	Pathogenic
218	0.3	M	Iran	Yes	Panel	IL2RG	c.181_182delCA	p.Gln61fs	hemizygous	frameshift	.	Pathogenic

219	0.7	M	Lebanon	Yes	Panel	<i>IL2RG</i>	c.437T>C	p.Leu146Pro	hemizygous	missense	23.5	VUS
220	0.7	M	Lebanon	No	Panel	<i>IL2RG</i>	c.982C>T	p.Arg328Ter	hemizygous	nonsense	.	Pathogenic
221	0.7	M	Egypt	No	Panel	<i>IL2RG</i>	c.758-1G>C	N/A	hemizygous	splice site	.	Pathogenic
222	0.4	F	Chile	No	Panel	<i>IL7R</i>	c.83-2A>T	N/A	heterozygous	splice site	.	Pathogenic
223	1	M	Egypt	Yes	Panel	<i>IL7R</i>	del exon 1	del exon 1	homozygous	deletion	.	Pathogenic
224	0.3	M	Pakistan	Yes	Panel	<i>IL7R</i>	c.83-1G>T	N/A	homozygous	splice site	.	Pathogenic
225	0.3	M	Turkey	Yes	Panel	<i>IL7R</i>	c.696dupT	p.Asn233Ter	homozygous	nonsense	.	Pathogenic
226	6	M	Egypt	Yes	Panel	<i>INO80</i>	c.2374A>T	p.Met792Leu	homozygous	missense	21.5	VUS
227	5	M	Tunisia	Yes	Panel	<i>IRAK4</i>	c.609_622delCTACGTAAATAACA	p.Tyr204Asnfs	homozygous	frameshift	.	Pathogenic
228	5	M	United States	No	Panel	<i>IRAK4</i>	c.88G>T	p.Glu30Ter	compound het	nonsense	.	Pathogenic
						<i>IRAK4</i>	c.877C>T	p.Gln293Ter		nonsense	.	Pathogenic
229	0.7	F	Kuwait	Yes	Panel	<i>ITGB2</i>	c.821dupA	p.Leu275Alafs	homozygous	frameshift	.	Pathogenic
230	0.3	F	Egypt	Yes	Panel	<i>ITGB2</i>	c.850G>A	p.Gly284Ser	homozygous	missense	25.7	Pathogenic
						<i>ITGB2</i>	c.821_822insA	p.Leu275fs	homozygous	frameshift	.	Pathogenic
231	1	M	Egypt	Yes	Panel	<i>ITGB2</i>	c.59-1G>A	N/A	homozygous	splice site	.	Pathogenic
232	0.2	F	Pakistan	Yes	Panel	<i>ITGB2</i>	c.134G>A	p.Trp45Ter	homozygous	nonsense	.	Pathogenic
233	10	M	Egypt	Yes	Panel	<i>ITK</i>	c.236C>T	p.Pro79Leu	homozygous	missense	27.3	Pathogenic
234	8	F	Iran	Yes	Panel	<i>ITK</i>	c.1061-1G>C	N/A	homozygous	splice site	.	Pathogenic
235	13	F	Lebanon	Yes	Panel	<i>ITK</i>	c.1562C>T	p.Pro521Leu	homozygous	missense	29.6	VUS
236	7	M	Peru	No	Panel	<i>JAGN1</i>	c.3G>A	p.Met11Ile	compound het	missense (alt start codon)	31.0	Pathogenic
						<i>JAGN1</i>	c.27delG	p.Ala10fs		frameshift	.	Pathogenic
237	1	M	Lebanon	No	Panel	<i>JAK3</i>	c.1744C>T	p.Arg582Trp	heterozygous	missense	27.5	Pathogenic
238	1	F	Lebanon	Yes	Panel	<i>JAK3</i>	c.2351-14T>G	N/A	homozygous	splice site	.	VUS
239	7	F	Egypt	Yes	Panel	<i>JAK3</i>	c.2492G>T	p.Gly831Val	homozygous	missense	32.0	VUS
240	0.4	F	Iran	Yes	Panel	<i>JAK3</i>	c.2125T>A	p.Trp709Arg	homozygous	missense	27.7	Pathogenic
241	0.7	F	Jordan	Yes	Panel	<i>JAK3</i>	c.1766G>T	p.Gly589Val	homozygous	missense	26.3	Pathogenic
242	0.8	M	Pakistan	Yes	Panel	<i>JAK3</i>	del exon 10	del exon 10	homozygous	deletion	.	Pathogenic
243	0.7	M	Qatar	Yes	Panel	<i>JAK3</i>	c.1900G>C	p.Ala634Pro	homozygous	missense	29.6	Pathogenic
244	0.9	F	United Arab Emirates	Yes	Panel	<i>JAK3</i>	c.172G>A	p.Ala58Thr	homozygous	missense	27.7	VUS
245	0.1	F	United States	Yes	WES	<i>JAK3</i>	c.602T>C	p.Leu201Pro	homozygous	missense	22.0	VUS
246	8	F	United States	No	WES	<i>KMT2D</i>	c.16267G>T	p.Val5423Phe	heterozygous	missense	28.8	VUS
247	0.4	M	United Arab Emirates	Yes	Panel	<i>LIG1</i>	c.2014C>T	p.Arg672Cys	homozygous	missense	31.0	Pathogenic
						<i>SP110</i>	c.691C>T	p.Gln231Ter	homozygous	nonsense	.	Pathogenic
248	10	M	Peru	No	Panel	<i>LRBA</i>	del exon 5-6	del exon 5-6	homozygous	deletion	.	Pathogenic
249	13	F	Algeria	Yes	Panel	<i>LRBA</i>	c.7408C>T	p.Gln2470Ter	homozygous	nonsense	.	Pathogenic
250	16	F	ArgentiN/A	Yes	Panel	<i>LRBA</i>	c.2735_2738delGGGT	p.Trp912Ter	homozygous	nonsense	.	Pathogenic
251	5	M	Egypt	Yes	Panel	<i>LRBA</i>	c.491dupT	p.Leu164fs	homozygous	frameshift	.	Pathogenic
252	6	M	Egypt	Yes	Panel	<i>LRBA</i>	c.6587delG	p.Arg2196Leufs	homozygous	frameshift	.	Pathogenic
253	11	F	Egypt	Yes	Panel	<i>LRBA</i>	c.534delA	p.Asp1791Ilefs	homozygous	frameshift	.	Pathogenic
254	7	M	Egypt	Yes	Panel	<i>LRBA</i>	c.8335_8336delGA	p.Asp2779Ter	homozygous	nonsense	.	Pathogenic
255	5	M	Egypt	Yes	Panel	<i>LRBA</i>	c.3229G>T	p.Glu1077Ter	homozygous	nonsense	.	Pathogenic
256	6	M	Egypt	Yes	Panel	<i>LRBA</i>	del exons 1-22	del exons 1-22	homozygous	deletion	.	Pathogenic
257	4	M	Egypt	Yes	Panel	<i>LRBA</i>	c.2368-2A>G	N/A	homozygous	splice site	.	Pathogenic
258	13	F	Peru	Yes	Panel	<i>LRBA</i>	c.949C>T	p.Arg317Ter	homozygous	nonsense	.	Pathogenic
259	13	M	Lebanon	Yes	WES	<i>LRBA</i>	c.865_866del	p.Cys289fs	homozygous	nonsense	.	Pathogenic
260	8	F	Egypt	Yes	Panel	<i>LRBA</i>	c.6415C>T	p.Arg2139Ter	homozygous	nonsense	.	Pathogenic
261	6	M	Egypt	Yes	Panel	<i>LRBA</i>	c.3229G>T	p.Glu1077Ter	homozygous	nonsense	.	Pathogenic
262	6	F	Egypt	Yes	Panel	<i>LRBA</i>	c.8335_8336delGA	p.Asp2779Ter	homozygous	nonsense	.	Pathogenic
263	0.3	M	Saudi Arabia	Yes	WES	<i>LRBA</i>	c.2032C>T	p.Gln678Ter	homozygous	nonsense	.	Pathogenic
264	1	M	Kuwait	Yes	WES	<i>LRBA</i>	dup exons 49-53	N/A	homozygous	duplication	.	Pathogenic
265	4	F	Saudi Arabia	Yes	Panel	<i>MALT1</i>	del exon 11	del exon 11	homozygous	deletion	.	Pathogenic
266	0.8	M	Oman	Yes	Panel + WES	<i>MAP3K14</i>	c.1693C>T	p.Arg565Trp	homozygous	missense	19.3	VUS
267	10	F	Egypt	No	Panel	<i>MEFV</i>	c.2177T>C	p.Val726Ala	compound het	missense	0.0	VUS
						<i>MEFV</i>	c.2076_2078delAAT	p.Ile692del		frameshift	.	Pathogenic
268	6	F	Egypt	Yes	Panel	<i>MEFV</i>	c.2076_2078delAAT	p.Ile692del	heterozygous	nonsense	.	Pathogenic
269	2	M	Lebanon	Yes	WES	<i>MEFV</i>	c.2080A>G	p.Met694Val	homozygous	missense	10.6	Pathogenic
270	1	M	Egypt	Yes	Panel	<i>MRE11A</i>	c.1105T>C	p.Tyr369His	homozygous	missense	24.7	VUS

271	0.4	M	Greece	Yes	Panel	<i>MRE11A</i>	c.318delT	p.Pro107fs	homozygous	frameshift	.	Pathogenic
272	12	M	Egypt	Yes	Panel	<i>MSH6</i>	c.453dupT	p.Thr152Yrfs	homozygous	frameshift	.	Pathogenic
273	1	M	Egypt	Yes	Panel	<i>MVK</i>	c.976G>A	p.Gly326Arg	homozygous	missense	24.8	Pathogenic
274	14	F	Turkey	No	Panel	<i>MVK</i>	c.52G>A	p.Gly18Arg	compound het	missense	31.0	Pathogenic
						<i>MVK</i>	c.1129G>A	p.Val377Ile		missense	14.4	Pathogenic
275	0.3	F	Egypt	Yes	WES	<i>MVK</i>	c.C1162T	p.Arg388Ter	homozygous	nonsense	.	Pathogenic
276	15	M	Palestinian Territories	No	Panel	<i>MVK</i>	c.412delC	p.Ala141fs	heterozygous	frameshift	.	Pathogenic
277	0.8	M	Oman	Yes	Panel	<i>MYD88</i>	c.814C>T	p.Arg272Ter	homozygous	nonsense	.	Pathogenic
278	0.3	M	Egypt	Yes	WES	<i>MYO5B</i>	c.1966C>T	p.Arg656Cys	homozygous	missense	29.8	Pathogenic
279	18	M	Turkey	Yes	WES	<i>MYSM1</i>	c.103_109del	p.Asp35Ilefs	homozygous	frameshift	.	Pathogenic
280	0.8	M	Kuwait	Yes	WES	<i>NBEAL2</i>	c.3908_3909insT	p.P1303fs	homozygous	frameshift	.	Pathogenic
281	1	M	Kuwait	Yes	Panel	<i>NBN</i>	c.900_924del	p.Gly301fs	homozygous	frameshift	.	Pathogenic
282	5	M	Egypt	Yes	Panel	<i>NCF1</i>	del exon 10	del exon 10	homozygous	deletion	.	Pathogenic
283	3	M	Pakistan	Yes	Panel	<i>NCF1</i>	c.124C>T	p.Arg42Trp	homozygous	missense	25.5	Pathogenic
284	2	M	Pakistan	Yes	Panel	<i>NCF1</i>	del exons 2 and 10	del exons 2 and 10	homozygous	deletion	.	Pathogenic
285	0.1	M	Pakistan	Yes	Panel	<i>NCF1</i>	c.294_295insA	p.Gly99fs	homozygous	frameshift	.	Pathogenic
286	1	F	Pakistan	Yes	Panel	<i>NCF2</i>	c.1098_1099delTC	p.Gln367fs	homozygous	frameshift	.	Pathogenic
287	12	F	Peru	No	Panel	<i>NCF2</i>	del exons 1-2	del exons 1-2	homozygous	deletion	.	Pathogenic
288	8	F	Kuwait	Yes	WES	<i>NCF2</i>	c.865+1G>A	N/A	homozygous	splice site	.	Pathogenic
289	7	M	Iran	No	Panel	<i>NFKB2</i>	c.455delA	p.Lys153Asnfs	heterozygous	frameshift	.	Pathogenic
290	20	F	United States	No	Panel	<i>NFKB2</i>	c.2600C>T	p.Ala867Val	heterozygous	missense	25.6	Pathogenic
291	14	F	India	No	Panel	<i>NFKB2</i>	c.1588delC	p.Leu531Cysfs	heterozygous	frameshift	.	Pathogenic
292	40	M	Italy	No	Panel	<i>NFKB2</i>	c.2557C>T	p.Arg853Ter	heterozygous	nonsense	.	Pathogenic
293	12	F	Kuwait	No	Panel	<i>NFKB2</i>	c.2596_2597delAG	p.Ser866fs	heterozygous	frameshift	.	Pathogenic
294	15	M	Egypt	Yes	Panel	<i>NFKB2</i>	c.2557C>T	p.Arg853Ter	heterozygous	nonsense	.	Pathogenic
295	15	F	Egypt	Yes	Panel	<i>NFKB2</i>	c.2557C>T	p.Arg853Ter	heterozygous	nonsense	.	Pathogenic
296	7	M	Egypt	Yes	Panel	<i>NHEJ1</i>	c.532C>T	p.Arg178Ter	homozygous	nonsense	.	Pathogenic
297	2	F	Iran	Yes	Panel	<i>NHEJ1</i>	c.526C>T	p.Arg176Ter	homozygous	nonsense	.	Pathogenic
298	2	M	Egypt	No	Panel	<i>NLRCA</i>	c.1540C>T	p.Gln514Ter	heterozygous	nonsense	.	Pathogenic
299	6	M	ArgentiN/A	No	Panel	<i>NLRP12</i>	c.3068T>C	p.Leu1023Pro	heterozygous	missense	23.1	VUS
300	13	M	United States	Yes	Panel	<i>NLRP12</i>	c.753C>G	p.Asn251Lys	homozygous	missense	22.8	VUS
301	2	M	Egypt	No	Panel	<i>NLRP3</i>	c.932T>C	p.Phe311Ser	heterozygous	missense	22.8	VUS
302	2	M	Lebanon	No	Panel	<i>NOD2</i>	c.1070A>C	p.Asp357Ala	heterozygous	missense	22.1	Pathogenic
303	3	F	ArgentiN/A	No	Panel	<i>NRAS</i>	c.35G>C	p.Gly12Ala	heterozygous	missense	27.3	Pathogenic
304	0.3	M	United Arab Emirates	Yes	WES	<i>ORAI1</i>	c.493_494insC	p.His165Profs	homozygous	frameshift	.	Pathogenic
305	0.6	F	Oman	Yes	WES	<i>ORAI1</i>	c.808C>T	p.Arg270Ter	homozygous	nonsense	.	Pathogenic
306	3	F	Turkey	Yes	Panel + WES	<i>PEPD</i>	c.611_623dupAGGCCACCGTGA	p.Val209Glyfs	homozygous	frameshift	.	Pathogenic
307	1	M	Egypt	Yes	Panel	<i>PGM3</i>	c.975T>G	p.Asp325Glu	homozygous	missense	23.1	Pathogenic
308	3	M	Egypt	No	Panel	<i>PGM3</i>	c.1139G>C	p.Gly380Ala	compound het	missense	25.1	VUS
						<i>PGM3</i>	c.545A>G	p.Tyr182Cys		missense	27.3	VUS
309		M	Lebanon	No	Panel	<i>PIK3CD (GOF)</i>	c.3061G>A	p.Glu1021Lys	heterozygous	missense	24.7	Pathogenic
310	10	M	Lebanon	No	Panel	<i>PIK3CD (GOF)</i>	c.3061G>A	p.Glu1021Lys	heterozygous	missense	24.7	Pathogenic
						<i>WRAP53</i>	c.1565delC	p.Ala522Glyfs*26	homozygous	frameshift	.	Pathogenic
311	4	F	Algeria	No	Panel	<i>PIK3CD (GOF)</i>	c.371G>A	p.Gly124Asp	heterozygous	missense	25.9	Pathogenic
312	9	F	Argentina	No	Panel	<i>PIK3CD (GOF)</i>	c.3061G>A	p.Glu1021Lys	heterozygous	missense	24.7	Pathogenic
313	3	F	Chile	No	Panel	<i>PIK3CD (GOF)</i>	c.1002C>G	p.Asn334Lys	heterozygous	missense	21.5	Pathogenic
314	4	F	Peru	No	Panel	<i>PIK3CD (GOF)</i>	c.3061G>A	p.Glu1021Lys	heterozygous	missense	24.7	Pathogenic
315	3	F	Peru	No	Panel	<i>PIK3CD (GOF)</i>	c.3061G>A	p.Glu1021Lys	heterozygous	missense	24.7	Pathogenic
316	7	F	Peru	No	Panel	<i>PIK3CD (GOF)</i>	c.3061G>A	p.Glu1021Lys	heterozygous	missense	24.7	Pathogenic
317		F	Peru	No	WES	<i>PIK3CD (GOF)</i>	c.3061G>A	p.Glu1021Lys	heterozygous	missense	24.7	Pathogenic
318	4	F	Peru	No	Panel	<i>PIK3CD (GOF)</i>	c.3061G>A	p.Glu1021Lys	heterozygous	missense	24.7	Pathogenic
319	11	M	Egypt	No	Panel	<i>PIK3CD (GOF)</i>	c.3061G>A	p.Glu1021Lys	heterozygous	missense	24.7	Pathogenic
320	6	M	Peru	No	Panel	<i>PIK3CD (GOF)</i>	c.3061G>A	p.Glu1021Lys	heterozygous	missense	24.7	Pathogenic
321	5	F	Iran	Yes	Panel	<i>PIK3R1 (APDS2)</i>	c.1425+1G>A	N/A	heterozygous	splice site	.	Pathogenic
322	2	M	Egypt	Yes	Panel	<i>PNP</i>	c.88delA	p.Ile30fs	homozygous	frameshift	.	Pathogenic
323	1	F	Egypt	Yes	Panel	<i>PNP</i>	c.349G>A	p.Ala117Thr	homozygous	missense	29.7	Pathogenic
324	4	M	Turkey	Yes	Panel	<i>PNP</i>	c.200G>C	p.Arg67Pro	homozygous	missense	25.0	Pathogenic

325	10	M	Iran	Yes	Panel	<i>PRKCD</i>	c.1293_1294insA	p.Gly432fs	homozygous	frameshift	.	Pathogenic
326	16	F	Egypt	No	Panel	<i>PSTPIP1</i>	c.850C>T	p.Gln284Ter	heterozygous	nonsense	.	Pathogenic
327	0.3	F	Iran	Yes	Panel	<i>PTPRC</i>	c.2848_2849insT	p.Arg950Metfs	homozygous	frameshift	.	Pathogenic
328	23	M	Lebanon	Yes	Panel	<i>RAB27A</i>	c.244C>T	p.Arg82Cys	homozygous	missense	32.0	Pathogenic
329	0.8	M	Kazakhstan	Yes	Panel	<i>RAG1</i>	c.2210G>A	p.Arg737His	heterozygous	missense	31.0	Pathogenic
330	2	F	Egypt	Yes	Panel	<i>RAG1</i>	c.2918G>A	p.Arg973His	homozygous	missense	29.6	VUS
331	2	F	Egypt	Yes	Panel	<i>RAG1</i>	c.1277_1279delAAG	p.Glu425del	homozygous	nonsense	.	Pathogenic
332	1	F	Egypt	Yes	Panel	<i>RAG1</i>	c.2867T>C	p.Ile956Thr	homozygous	missense	25.1	VUS
333	1	M	Egypt	Yes	Panel	<i>RAG1</i>	c.2521C>T	p.Arg841Trp	homozygous	missense	28.7	Pathogenic
334	0.5	F	Egypt	Yes	Panel	<i>RAG1</i>	c.2923C>T	p.Arg975Trp	homozygous	missense	28.2	Pathogenic
335	1	F	Iran	Yes	Panel	<i>RAG1</i>	c.1180C>T	p.Arg394Trp	homozygous	missense	28.5	Pathogenic
336	0.5	M	Iran	Yes	Panel	<i>RAG1</i>	c.2570C>A	p.Ala857Asp	homozygous	missense	28.3	Pathogenic
337	0.6	F	Iran	Yes	Panel	<i>RAG1</i>	c.2985G>A	p.Trp995Ter	homozygous	nonsense	.	Pathogenic
338	1	F	Iran	Yes	Panel	<i>RAG1</i>	c.322C>T	p.Arg108Ter	compound het	nonsense	.	Pathogenic
							c.1405G>C	p.Val469Leu		missense	25.8	Pathogenic
339	0.4	F	Iran	Yes	Panel	<i>RAG1</i>	c.1180C>T	p.Arg394Trp	homozygous	missense	28.5	Pathogenic
340	0.4	M	Iran	Yes	Panel	<i>RAG1</i>	c.2917C>T	p.Arg973Cys	homozygous	missense	31.0	VUS
341	0.3	F	Lebanon	No	Panel	<i>RAG1</i>	c.1210C>T	p.Arg404Trp	compound het	missense	26.9	Pathogenic
							c.2210G>A	p.Arg737His		missense	31.0	Pathogenic
342	0.6	M	Pakistan	Yes	Panel	<i>RAG1</i>	c.2275C>T	p.Arg759Cys	homozygous	missense	32.0	VUS
343	0.2	F	Pakistan	Yes	Panel	<i>RAG1</i>	c.2210G>A	p.Arg737His	homozygous	missense	31.0	Pathogenic
344	9	M	Peru	No	Panel	<i>RAG1</i>	c.1685A>G	p.Tyr562Cys	compound het	missense	28.7	VUS
							c.2522G>A	p.Arg841Gln		missense	33.0	VUS
345	0.6	F	Turkey	Yes	Panel	<i>RAG1</i>	c.937T>A	p.Cys313Ser	homozygous	missense	24.2	VUS
346	0.3	F	Turkey	Yes	Panel	<i>RAG1</i>	c.1767C>G	p.Tyr589Ter	homozygous	nonsense	.	Pathogenic
347	0.3	M	Turkey	Yes	Panel	<i>RAG1</i>	c.1378G>T	p.Gly460Ter	homozygous	nonsense	.	Pathogenic
348	10	M	Kuwait	Yes	WES	<i>RAG1</i>	c.1211G>A	p.Arg404Gln	homozygous	missense	29.2	VUS
349	0.5	M	Lebanon	Yes	Panel	<i>RAG1</i>	c.987delC	p.Ser330fs	homozygous	frameshift	.	Pathogenic
350	10	F	United States	No	WES	<i>RAG1</i>	c.1420C>T	p.Arg474Cys	compound het	missense	28.7	Pathogenic
							c.2521C>T	p.Arg841Trp		missense	28.1	Pathogenic
351	6	M	Algeria	Yes	Panel	<i>RAG2</i>	c.1340C>G	p.Ser447Cys	homozygous	missense	27.4	VUS
352	0.8	F	Algeria	Yes	Panel	<i>RAG2</i>	c.104G>T	p.Gly35Val	homozygous	missense	24.4	Pathogenic
353	0.2	M	Egypt	Yes	Panel	<i>RAG2</i>	c.650A>G	p.Tyr217Cys	homozygous	missense	25.8	VUS
354	9	M	Egypt	Yes	Panel	<i>RAG2</i>	c.104G>T	p.Gly35Val	homozygous	missense	24.4	Pathogenic
355	2	F	Iran	Yes	Panel	<i>RAG2</i>	c.734C>T	p.Pro245Leu	homozygous	missense	27.6	VUS
356	0.5	M	Iran	Yes	Panel	<i>RAG2</i>	c.685C>T	p.Arg229Trp	homozygous	missense	28.2	VUS
357	0.4	F	Iran	Yes	Panel	<i>RAG2</i>	c.130G>A	p.Gly44Arg	homozygous	missense	25.7	Pathogenic
358	0.3	M	Iran	Yes	Panel	<i>RAG2</i>	c.130G>A	p.Gly44Arg	homozygous	missense	25.7	Pathogenic
359	0.5	F	Jordan	Yes	Panel	<i>RAG2</i>	c.104G>T	p.Gly35Val	homozygous	missense	24.4	Pathogenic
360	0.8	M	Jordan	Yes	Panel	<i>RAG2</i>	c.104G>T	p.Gly35Val	homozygous	missense	24.4	Pathogenic
361	12	F	Kuwait	Yes	Panel	<i>RAG2</i>	c.104G>C	p.Gly35Ala	homozygous	missense	24.1	VUS
362	0.3	M	Pakistan	Yes	Panel	<i>RAG2</i>	c.1247G>T	p.Trp416Leu	homozygous	missense	29.1	Pathogenic
363	0.3	F	Egypt	Yes	Panel	<i>RAG2</i>	c.644C>T	p.Thr215Ile	homozygous	missense	22.8	Pathogenic
364	0.5	M	Iran	Yes	Panel	<i>RAG2</i>	c.358delG	p.Val120Leufs	homozygous	frameshift	.	Pathogenic
365	0.3	M	Jordan	Yes	Panel	<i>RAG2</i>	c.104G>T	p.Gly35Val	homozygous	missense	24.4	Pathogenic
366	46	F	Algeria	Yes	Panel	<i>RECQL4</i>	c.2269C>T	p.Gln757Ter	compound het	nonsense	.	Pathogenic
							c.3268G>A	p.Glu1090Lys		missense	26.8	Pathogenic
367	1	M	Egypt	Yes	Panel	<i>RFX5</i>	c.715C>T	p.Arg239Ter	homozygous	nonsense	.	Pathogenic
368	0.4	F	Jordan	Yes	Panel	<i>RFX5</i>	c.445C>T	p.Arg149Ter	homozygous	nonsense	.	Pathogenic
369	0.3	M	Jordan	Yes	Panel	<i>RFX5</i>	c.445C>T	p.Arg149Ter	homozygous	nonsense	.	Pathogenic
370	0.6	F	Jordan	Yes	Panel	<i>RFX5</i>	c.445C>T	p.Arg149Ter	homozygous	nonsense	.	Pathogenic
371	0.2	M	Jordan	Yes	Panel	<i>RFX5</i>	c.445C>T	p.Arg149Ter	homozygous	nonsense	.	Pathogenic
372	0.4	F	Jordan	Yes	Panel	<i>RFX5</i>	c.445C>T	p.Arg149Ter	homozygous	nonsense	.	Pathogenic
373	17	M	Lebanon	Yes	Panel	<i>RFX5</i>	c.1801dupC	p.Gln601fs	homozygous	frameshift	.	Pathogenic
374	0.9	M	Jordan	Yes	Panel	<i>RFX5</i>	c.715C>T	p.Arg239Ter	homozygous	nonsense	.	Pathogenic
375	2	F	Egypt	Yes	Panel	<i>RFXANK</i>	c.713-2A>G	N/A	homozygous	splice site	.	Pathogenic
376	0.3	M	Iran	Yes	Panel	<i>RFXANK</i>	c.162delG	p.Asp55fs	homozygous	frameshift	.	Pathogenic

377	1	M	Algeria	Yes	Panel	<i>RFXANK</i>	c.673insC	p.Met227Aspfs	homozygous	frameshift	.	Pathogenic
378	0.6	M	Iran	Yes	Panel	<i>RFXANK</i>	c.634C>T	p.Arg212Ter	homozygous	nonsense	.	Pathogenic
379	9	F	Iran	Yes	Panel	<i>RFXANK</i>	c.163delA	p.Asp55fs	homozygous	frameshift	.	Pathogenic
380	0.6	F	Iran	Yes	Panel	<i>RFXAP</i>	c.467delA	p.Gln156Argfs	homozygous	frameshift	.	Pathogenic
381	5	M	Algeria	Yes	Panel + WES	<i>RIPK1</i>	c.1835T>C	p.Ile612Thr	homozygous	missense	27.4	VUS
382	4	M	Egypt	Yes	Panel	<i>RMRP</i>	g.9:35657888C>G	N/A	homozygous	missense	11.1	Pathogenic
383	1	M	Algeria	Yes	Panel	<i>RMRP</i>	g.9:35657917G>T	N/A	homozygous	missense	17.9	Pathogenic
384	8	M	Argentina	Yes	Panel	<i>SBDS</i>	c.258+2T>C	N/A	homozygous	splice site	.	Pathogenic
385	26	F	Argentina	Yes	Panel	<i>SBDS</i>	c.258+2T>C	N/A	homozygous	splice site	.	Pathogenic
386	6	M	Egypt	No	Panel	<i>SH2D1A</i>	c.32T>G	p.Ile11Ser	hemizygous	missense	24.6	VUS
387	1	M	Egypt	No	Panel	<i>SH2D1A</i>	c.295C>T	p.Gln99Ter	hemizygous	nonsense	.	Pathogenic
388	7	M	Peru	No	Panel	<i>SH2D1A</i>	c.251T>G	p.Ile84Ser	hemizygous	missense	32.0	Pathogenic
389	6	F	Lebanon	Yes	Panel + WES	<i>SLC39A7</i>	c.469C>G	p.Pro157Ala	homozygous	missense	25.5	VUS
390	0.5	F	Pakistan	Yes	Panel + WES	<i>SLC7A7</i>	del exons 6-11	del exons 6-11	homozygous	deletion	.	Pathogenic
391	5	M	Egypt	Yes	WES	<i>SLC7A7</i>	c.404delT	p.I135fs	homozygous	frameshift	.	Pathogenic
392	5	M	Iran	Yes	Panel	<i>SMARCAL1</i>	del exons 1-2	del exons 1-2	homozygous	deletion	.	Pathogenic
393	0.5	F	Lebanon	Yes	Panel	<i>SP110</i>	c.642delC	p.Ser215Alafs	homozygous	frameshift	.	Pathogenic
394	1	M	Lebanon	Yes	Panel	<i>SP110</i>	c.642_642delC	p.Ser215fs	homozygous	frameshift	.	Pathogenic
395	5	M	Oman	Yes	Panel	<i>SP110</i>	c.55C>T	p.Gln19Ter	homozygous	nonsense	.	Pathogenic
396	7	M	Saudi Arabia	Yes	Panel	<i>STAT1 (GOF)</i>	c.1066T>G	p.Tyr356Asp	heterozygous	missense	23.3	VUS
397	10	F	Peru	No	Panel	<i>STAT1 (GOF)</i>	c.1154C>T	p.Thr385Met	heterozygous	missense	26.5	Pathogenic
398	24	M	Italy	No	Panel	<i>STAT1 (GOF)</i>	c.890delA	p.Asn297fs	heterozygous	frameshift	.	Pathogenic
399	2	F	Algeria	No	Panel	<i>STAT1 (GOF)</i>	c.1154C>T	p.Thr385Met	heterozygous	missense	26.5	Pathogenic
400	15	F	United States	No	Panel	<i>STAT1 (GOF)</i>	c.1213C>T	p.Arg405Trp	heterozygous	missense	32.0	Pathogenic
401	1	M	Egypt	Yes	Panel	<i>STAT1 (GOF)</i>	c.537C>A	p.Asn179Lys	heterozygous	missense	22.0	Pathogenic
402	0.7	M	Egypt	No	Panel	<i>STAT1 (GOF)</i>	c.1031A>C	p.Lys344Thr	heterozygous	missense	27.4	Pathogenic
403	1	M	Egypt	Yes	Panel	<i>STAT1 (LOF)</i>	c.1652dupA	p.Asn551Lysfs	homozygous	frameshift	.	Pathogenic
404	1	F	Egypt	No	Panel	<i>STAT1 (GOF)</i>	c.1154C>T	p.Thr385Met	heterozygous	missense	26.5	Pathogenic
405	3	M	Mexico	No	Panel	<i>STAT1 (GOF)</i>	c.972C>G	p.Cys324Trp	heterozygous	missense	28.3	Pathogenic
406	39	M	Oman	No	Panel	<i>STAT1 (GOF)</i>	c.820C>T	p.Arg274Trp	heterozygous	missense	32.0	Pathogenic
407	10	F	Peru	No	Panel	<i>STAT1 (GOF)</i>	c.1154C>T	p.Thr385Met	heterozygous	missense	26.5	Pathogenic
408	9	M	Peru	Yes	Panel	<i>STAT1 (GOF)</i>	c.511G>A	p.Asp171Asn	heterozygous	missense	31.0	Pathogenic
409	4	F	Peru	No	Panel	<i>STAT1 (GOF)</i>	c.1154C>T	p.Thr385Met	heterozygous	missense	26.5	Pathogenic
410	12	M	Peru	No	Panel	<i>STAT1 (GOF)</i>	c.1219C>G	p.Leu407Val	heterozygous	missense	25.4	VUS
411	0.9	F	Peru	No	Panel	<i>TNFRSF13B</i>	c.832delG	p.Gly278Alafs	homozygous	frameshift	.	Pathogenic
						<i>STAT1 (GOF)</i>	c.1154C>T	p.Thr385Met	heterozygous	missense	26.5	Pathogenic
412	15	F	Lebanon	No	Panel	<i>STAT1 (GOF)</i>	c.821G>A	p.Arg274Gln	heterozygous	missense	28.5	Pathogenic
413	2	M	Saudi Arabia	No	Panel	<i>STAT1 (LOF)</i>	del exons 3-4	del exons 3-4	homozygous	deletion	.	Pathogenic
414	4	F	Greece	No	Panel	<i>STAT1 (GOF)</i>	c.800C>T	p.Ala267Val	heterozygous	missense	24.7	Pathogenic
415	11	F	United States	No	Panel	<i>STAT3 (GOF)</i>	c.519T>G	p.Asp173Glu	heterozygous	missense	23.4	VUS
416	7	M	Chile	No	Panel	<i>STAT3 (LOF)</i>	c.1771A>G	p.Lys591Glu	heterozygous	missense	25.0	Pathogenic
417	7	M	Egypt	No	Panel	<i>STAT3 (GOF)</i>	c.2117T>C	p.Leu706Pro	heterozygous	missense	32.0	Pathogenic
418	5	F	Egypt	Yes	Panel	<i>STAT3 (LOF)</i>	c.1268G>A	p.Arg423Gln	heterozygous	missense	33.0	Pathogenic
419	1	M	Egypt	No	Panel	<i>STAT3 (LOF)</i>	c.1909G>A	p.Val637Met	heterozygous	missense	23.6	Pathogenic
420	11	F	Iran	No	Panel	<i>STAT3 (LOF)</i>	c.1144C>T	p.Arg382Trp	heterozygous	missense	32.0	Pathogenic
421	8	F	Kazakhstan	No	Panel	<i>STAT3 (GOF)</i>	c.2144C>T	p.Pro715Leu	heterozygous	missense	33.0	Pathogenic
422	2	F	Oman	Yes	Panel	<i>STAT3 (LOF)</i>	c.1144C>T	p.Arg382Trp	heterozygous	missense	32.0	Pathogenic
423	3	M	Oman	Yes	Panel	<i>STAT3 (LOF)</i>	c.2132T>C	p.Ile711Thr	heterozygous	missense	28.6	Pathogenic
424	9	F	Peru	No	Panel	<i>STAT3 (LOF)</i>	c.1144C>T	p.Arg382Trp	heterozygous	missense	32.0	Pathogenic
425	3	F	Peru	Yes	Panel	<i>STAT3 (LOF)</i>	c.1145G>A	p.Arg382Gln	heterozygous	missense	33.0	Pathogenic
426	24	M	Peru	No	Panel	<i>STAT3 (LOF)</i>	c.1913A>G	p.Glu638Gly	heterozygous	missense	27.6	VUS
427	13	M	Peru	No	Panel	<i>STAT3 (LOF)</i>	c.1144C>T	p.Arg382Trp	heterozygous	missense	32.0	Pathogenic
428	8	F	Turkey	No	WES	<i>STAT3 (LOF)</i>	c.992T>G	p.Met331Arg	heterozygous	missense	26.3	Pathogenic
429	13	M	Argentina	No	Panel	<i>STAT3 (LOF)</i>	c.551-1G>C	N/A	heterozygous	splice site	.	Pathogenic
430	0.3	F	Lebanon	Yes	Panel	<i>STAT5B</i>	c.1101_1102insA	p.Gln368Thrfs	homozygous	frameshift	.	Pathogenic
432	7	M	Egypt	No	Panel	<i>STK4</i>	del exons 4-8	del exons 4-8	homozygous	deletion	.	Pathogenic
431	2	M	Egypt	Yes	Panel	<i>STK4</i>	c.442C>T	p.Arg148Ter	homozygous	nonsense	.	Pathogenic



433	14	F	Egypt	Yes	Panel	<i>STK4</i>	c.1A>T	p. Met1Leu	homozygous	missense	23.7	Pathogenic
434	14	M	Egypt	Yes	Panel	<i>STK4</i>	c.722delC	p.Pro241Leufs	homozygous	frameshift	.	Pathogenic
435	10	F	Turkey	Yes	Panel	<i>TAP1</i>	c.2143C>T	p.Arg715Ter	homozygous	nonsense	.	Pathogenic
436	8	F	Pakistan	Yes	Panel	<i>TCF3</i>	del exons 5-11	del exons 5-11	homozygous	deletion	.	Pathogenic
437	0.3	M	Oman	Yes	Panel + WES	<i>TCN2</i>	c.924_927delCTGT	p.Cys309Trpfs	homozygous	frameshift	.	Pathogenic
438	3	F	Egypt	No	Panel	<i>TERT</i>	del exon 5	del exon 5	homozygous	deletion	.	Pathogenic
439	33	M	Algeria	No	Panel	<i>TNFRSF13B</i>	c.204dupA	p.Leu69Thrfs	heterozygous	frameshift	.	Pathogenic
440	16	M	Turkey	Yes	Panel	<i>TNFRSF13B</i> <i>TNFRSF13B</i>	c.204dupA c.834delC	p.Leu69Thrfs p.Gly280Alafs	heterozygous heterozygous	frameshift frameshift	.	Pathogenic
441	2	M	Egypt	Yes	Panel	<i>TRNT1</i>	c.947_948insG	p.Glu318fs	homozygous	frameshift	.	Pathogenic
442	0.6	F	United States	No	Panel	<i>TTC7A</i> <i>TTC7A</i>	c.211G>A c.911delT	p.Glu71Lys p.Leu304Argfs	compound het	missense frameshift	29.8	Pathogenic
443	8	F	Iran	Yes	Panel	<i>TYK2</i>	del exons 8-25	del exons 8-25	homozygous	deletion	.	Pathogenic
444	0.6	M	Iran	Yes	Panel	<i>UNC13D</i>	c.1208T>C	p.Leu403Pro	homozygous	missense	25.1	Pathogenic
445	21	M	Egypt	No	Panel	<i>UNC13D</i> <i>UNC13D</i>	c.2942_2945dupAGAC c.1193C>T	p.Phe983Aspfs p.Ser398Leu	compound het	frameshift missense	.	Pathogenic
446	8	M	Egypt	Yes	Panel	<i>UNC13D</i>	c.2215_2239del	p.Asn739_Alafs	homozygous	frameshift	.	Pathogenic
447	0.5	M	Lebanon	Yes	Panel	<i>UNC13D</i>	c.627delT	p.Val210Trpfs	homozygous	frameshift	.	Pathogenic
448	6	M	Egypt	No	Panel	<i>WAS</i>	c.274-2A>G	N/A	hemizygous	splice site	.	Pathogenic
449	0.2	M	Peru	No	Panel	<i>WAS</i>	c.172delC	p.Pro59fs	hemizygous	frameshift	.	Pathogenic
450	7	M	Egypt	Yes	Panel	<i>WAS</i>	c.289T>A	p.Trp97Arg	hemizygous	missense	29.7	Pathogenic
451	1	M	Egypt	No	Panel	<i>WAS</i>	del exons 10-12	del exons 10-12	hemizygous	deletion	.	Pathogenic
452	4	M	Egypt	No	Panel	<i>WAS</i>	c.91G>A	p.Glu31Lys	hemizygous	missense	25.9	Pathogenic
453	1	M	Egypt	Yes	Panel	<i>WAS</i>	c.176_188delCTGGAGCTGAGCA	p.Ala61fs	hemizygous	frameshift	.	Pathogenic
454	1	M	Iran	Yes	Panel	<i>WAS</i>	c.687delG	p.Lys230fs	hemizygous	frameshift	.	Pathogenic
455	0.8	M	Iran	Yes	Panel	<i>WAS</i>	c.777+1G>C	N/A	hemizygous	splice site	.	Pathogenic
456	0.5	M	Iran	No	Panel	<i>WAS</i>	c.397G>A	p.Glu133Lys	hemizygous	missense	27.7	Pathogenic
457	4	M	Jordan	No	Panel	<i>WAS</i>	c.121C>T	p.Arg41Ter	hemizygous	nonsense	.	Pathogenic
458	4	M	Jordan	No	Panel	<i>WAS</i>	c.121C>T	p.Arg41Ter	hemizygous	nonsense	.	Pathogenic
459	2	M	Pakistan	No	Panel	<i>WAS</i>	c.280_280delC	p.Arg94fs	hemizygous	frameshift	.	Pathogenic
460	1	M	Egypt	No	Panel	<i>WAS</i>	c.1210_1211insCG	p.Ser405Profs	hemizygous	frameshift	.	Pathogenic
461	1	M	Greece	Yes	Panel	<i>WAS</i>	c.764delA	p. Gln255fs	hemizygous	frameshift	.	Pathogenic
462	0.4	M	Peru	Yes	Panel	<i>WAS</i>	c.361_361+1insG	N/A	hemizygous	splice site	.	Pathogenic
463	4	M	Egypt	No	Panel	<i>WAS</i>	c.177delT	p.Gly60fs	hemizygous	frameshift	.	Pathogenic
464	0.5	M	Greece	No	Panel	<i>WAS</i>	c.70T>C	p.Ser24Pro	hemizygous	missense	24.3	Pathogenic
465	8	M	Greece	No	Panel	<i>XIAP</i>	c.1336G>T	p.Glu446Ter	hemizygous	nonsense	.	Pathogenic
466	10	M	Egypt	Yes	Panel	<i>XIAP</i>	del exons 3-4	del exons 3-4	hemizygous	deletion	.	Pathogenic
467	6	M	Peru	No	Panel	<i>XIAP</i>	del exons 2-3	del exons 2-3	hemizygous	deletion	.	Pathogenic
468	1	F	Egypt	Yes	Panel	<i>ZAP70</i>	c.1799delT	p.Leu600Argfs	homozygous	nonsense	.	Pathogenic
469	1	F	Egypt	Yes	Panel	<i>ZAP70</i>	c.261C>G	p.Tyr87Ter	homozygous	nonsense	.	Pathogenic
470	1	M	Egypt	Yes	Panel	<i>ZAP70</i>	c.1780C>T	p.Arg594Ter	homozygous	nonsense	.	Pathogenic
471	1	F	Pakistan	Yes	Panel	<i>ZBTB24</i>	c.396_397delTA	p.His132fs	homozygous	frameshift	.	Pathogenic

\*Pathogenic = Pathogenic or Likely Pathogenic by Pathogenic criteria, VUS = variant of uncertain significance with features concordant with disease

\*\*patient had digenic mutations in IFNAR1 and IFNGR2. Details of IFNAR1 mutation in Supplementary Table 3