

Supplementary Table 3: Variants in Novel Genes at the Time of Sequencing

Proband Number	Age (years)	Sex	Country	Consang.	NGS Technology	Gene	Mutation Type	cDNA	AA	Zygoty	Reference
472	0.3	M	Lebanon	Yes	WES	<i>DOCK2</i>	frameshift	c.3725_3726insTA	p.Y1242fs	homozygous	1
473	2	F	Algeria	Yes	Panel + WES	<i>FCHO1</i>	frameshift	c.184_185insC	p.Met64Hisfs	homozygous	2
474	6	F	Turkey	Yes	Panel + WES	<i>FCHO1</i>	splice site	c.27+1G>A	N/A	homozygous	2
184*	0.3	M	Saudi Arabia	Yes	WES	<i>IFNAR1</i>	deletion of stop codon	c.1671_1821del	p.557Gluext*46	homozygous	3
476	6	F	Egypt	Yes	Panel + WES	<i>ITPKB</i>	frameshift	c.2583delT	p.Arg862Glufs	homozygous	4
477	13	F	Lebanon	Yes	WES	<i>MALT1</i>	missense	c.266G>T	p.Ser89Ile	homozygous	5
478	1	M	Kuwait	Yes	WES	<i>NEIL3</i>	insertion	c.395_396AC>TG	p.Asp132Val	homozygous	6
479	6	M	Pakistan	Yes	WES	<i>PIK3CD**</i>	frameshift	c.2558_2559	p.Asp853Glyfs	homozygous	7
480	39	M	Italy	No	Panel + WES	<i>RAC2***</i>	missense	c.101C>A	p.Pro34His	heterozygous	8
481	6	M	Kuwait	Yes	Panel + WES	<i>REL</i>	splice site	c.535+1G>A	N/A	homozygous	9
482	5	F	United States	No	WES	<i>RELA</i>	splice site	c.550+1G>A	N/A	heterozygous	10
483	5	M	Saudi Arabia	Yes	WES	<i>TFRC</i>	missense	c.58T>C	p.Tyr20His	homozygous	11
484	9	M	Kuwait	Yes	WGS	<i>TFRC</i>	missense	c.58T>C	p.Tyr20His	homozygous	11
485	12	M	United Arab Emirates	Yes	WES	<i>TNFSF9</i>	missense	c.466 G>A	p.Glu156Lys	homozygous	12
486	1	F	Turkey	Yes	Panel + WES	<i>WDR1</i>	missense	c.1070A>T	p.Asp357Val	homozygous	#
487	0.3	M	Kuwait	Yes	WES	<i>WDR1</i>	missense	c.491C>T	p.Thr164Met	homozygous	#

\*patient had digenic mutations in *IFNAR1* and *IFNGR2*. Details of *IFNGR2* mutation in Table S2

\*\* loss of function mutation

\*\*\*gain of function mutation

# novel gene at time of sequencing, though subsequently published by other groups