The American Journal of Human Genetics, *91*

Supplemental Data

A Hypermorphic Missense Mutation in *PLCG2*, Encoding Phospholipase Cγ2, Causes a Dominantly Inherited Autoinflammatory Disease with Immunodeficiency

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Table S1. Quantification of Lymphocyte Subsets in Two Affected Individuals

Cell Type	II-1		III-2	
	Quantity	Normal Range	Quantity	Normal Range
CD3%	79.7	[60.0-83.7 %]	72.0	[60.0-83.7 %]
CD3#	909	[714-2266 /uL]	1087	[714-2266 /uL]
CD5%	81.5	[59.6-83.7 %]	72.3	[59.6-83.7 %]
CD5#	929	[723-2276 /uL]	1092	[723-2276 /uL]
T4/T8 Ratio	1.37	[1.11-5.17	1.92	[1.11-5.17
CD4/CD3%	45.3	[31.9-62.2 %]	45.6	[31.9-62.2 %]
CD4/CD3#	516	[359-1565 /uL]	689	[359-1565 /uL]
CD8/CD3%	33.1	[11.2-34.8 %]	23.7	[11.2-34.8 %]
CD8/CD3#	377	[178-853 /uL]	358	[178-853 /uL]
DNT Cell%	0.6 ↓	[1.3-9.2 %]	1.9	[1.3-9.2 %]
DNT Cell#	7 ↓	[18-185 /uL]	29	[18-185 /uL]
CD4/CD45RO/CD3%	15.8	[13.1-50.1 %]	21.1	[13.1-50.1 %]
CD4/CD45RO/CD3#	180 ↓	[219-1048 /uL]	319	[219-1048 /uL]
CD4/CD45RA/CD3%	15.2	[5.5-27.0 %]	12.5	[5.5-27.0 %]
CD4/CD45RA/CD3#	173	[82-755 /uL]	189	[82-755 /uL]
CD8/CD45RO/CD3%	1.3 ↓	[2.8-15.8 %]	2.1 ↓	[2.8-15.8 %]
CD8/CD45RO/CD3#	15 ↓	[50-352 /uL]	32 ↓	[50-352 /uL]
CD8/CD45RA/CD3%	27.8 ↑	[3.2-26.1 %]	16.8	[3.2-26.1 %]
CD8/CD45RA/CD3#	317	[51-572 /uL]	254	[51-572 /uL]
CD20%	7.1	[3.0-19.0 %]	3.1	[3.0-19.0 %]
CD20#	81	[59-329 /uL]	47 ↓	[59-329 /uL]
CD19%	7.3	[3.3-19.3 %]	3.4	[3.3-19.3 %]
CD19#	83	[61-321 /uL]	51 ↓	[61-321 /uL]
CD20/CD5%	3.8	[0.7-9.7 %]	1.8	[0.7-9.7 %]
CD20/CD5#	43	[14-159 /uL]	27	[14-159 /uL]
CD20/IgG%	0.1 ↓	[0.2-2.2 %]	0.0 ↓	[0.2-2.2 %]
CD20/IgG#	1 ↓	[4-49 /uL]	0 ↓	[4-49 /uL]
CD20/IgA%	0.0 ↓	[0.1-1.3 %]	0.0 ↓	[0.1-1.3 %]
CD20/IgA#	0 ↓	[3-19 /uL]	0 ↓	[3-19 /uL]
CD20/IgM%	6.9	[2.4-16.9 %]	3.0	[2.4-16.9 %]
CD20/IgM#	79	[51-297 /uL]	45 ↓	[51-297 /uL]
CD20/IgD%	6.9	[2.3-17.1 %]	3.0	[2.3-17.1 %]
CD20/IgD#	79	[51-277 /uL]	45 ↓	[51-277 /uL]
CD20/IgD/IgM%	6.9	[2.3-16.9 %]	3.0	[2.3-16.9 %]
CD20/IgD/IgM#	79	[50-276 /uL]	45 ↓	[50-276 /uL]
CD20/CD27%	0.2 ↓	[0.8-3.6 %]	0.3 ↓	[0.8-3.6 %]
CD20/CD27#	2 ↓	[12-68 /uL]	5 ↓	[12-68 /uL]
CD20/CD23%	6.0	[1.2-13.8 %]	2.2	[1.2-13.8 %]
CD20/CD23#	68	[27-229 /uL]	33	[27-229 /uL]
CD20/CD40%	6.8	[2.7-18.6 %]	2.9	[2.7-18.6 %]
CD20/CD40#	78	[52-313 /uL]	44 ↓	[52-313 /uL]
CD20/CD38%	7.0	[1.2-17.6 %]	3.0	[1.2-17.6 %]
CD20/CD38#	80	[30-282 /uL]	45	[30-282 /uL]
NK Cell%	13.2	[6.2-34.6 %]	25.0	[6.2-34.6 %]
NK Cell#	150	[126-729 /uL]	378	[126-729 /uL]
NK TCell%	0.6 ↓	[2.2-12.4 %]	1.9 ↓	[2.2-12.4 %]
NK TCell#	7 ↓	[29-299 /uL]	29	[29-299 /uL]

↓: indicates quantity of lymphocyte subset below the normal range.
↑: indicates quantity of lymphocyte subset above the normal range.

Table S2. List of Eight Candidate Variants from Filtering Exome-Sequencing Data in the Affected Father (II-1) and Daughter (III-2) and Unaffected Mother (II-2)

Chromosome Position	Reference Allele	Variant Allele	Variant Type	Nucleotide Change	Protein Alteration	Gene	GERP Score	Polyphen Prediction
Chr1:42637797	A	G	missense	c.299A>G RefSeq NM_173642.3	p.Asn100Ser	*RIMKLA*	4.34	probably damaging
Chr7:66055736	T	C	missense	c.869T>C RefSeq NM_017994.4	p.Ile290Thr	*C7orf42*	5.22	probably damaging
Chr8: 39208718	C	T	missense	c.1541C>T RefSeq NM_145004.5	p.Pro514Leu	*ADAM32*	5.15	probably damaging
Chr15: 49014175	A	G	missense	c.932A>G RefSeq NM_007347.3	p.Asn311Ser	*AP4E1* [MIM 607244]	5.68	probably damaging
Chr16: 80510655	C	A	missense	c.2120C>A RefSeq NM_002661.3	p.Ser707Tyr	*PLCG2* [MIM 600220]	4.64	probably damaging
Chr2: 162842199-162842199	T	-	frameshift deletion	c.2016delA RefSeq NM_022168.2	p.Asp673Ilefs*5	*IFIH1* [MIM 606951]	/	/
Chr10: 38281403-38281412	TTCAAAGGGT	-	frameshift deletion	c.1020_1029del RefSeq NM_145011.2	p.Lys340Asnfs*39	*ZNF25* [MIM 194528]	/	/
Chr14: 76915028-76915029	GT	-	frameshift deletion	c.1514_1515del RefSeq NM_001010860.1	p.Ser505Argfs*5	*SAMD15*	/	/