Supplementary Table S1

Filtered variants of Case I.1.

Gene	Chromosome	Position	HGVS Nomenclature	Protein Change	Exon/ Intron	Zygosity	Parental Inheritance	In Silico Prediction (REVEL)	o/e Score (Gnomad)	rsID	MAF (in-house)	Functional Interactions Found with Pyrin Inflammasome Related Genes	Pathogenicity Classification &
ABCA13	7	48349721	ENST00000435803.1:c.9499C>T	p.Arg3167Ter	Exon 24	HET	maternal†	N/A	0,915	N/A	0.001	N/A	LP
ACTN3	11	66327703	ENST00000502692.1:c.1727C>T	p.Ala576Val	Exon 14	HET	maternal†	N/A	N/A	rs372101780	0.001	RHOA (STRING)	VUS
AFDN	6	168370601	ENST00000400822.3:c.5491C>A	p.Leu1831Met	Exon 34	HET	maternal†	0,181	0,8	rs765987505	0.0	YWHAB (GeneMANIA)	VUS
ANKS1A	6	35048916	ENST00000360359.3:c.2690C>T	p.Ala897Val	Exon 17	HET	maternal†	0,017	0,691	rs779234201	0.001	YWHAB (GeneMANIA)	VUS
ARHGAP23	17	36584785	ENST00000431231.2:c.56C>T	p.Pro19Leu	Exon 1	HET	paternal†	0,046	0,785	rs777310873	0.0	RHOA/B/C (STRING)	VUS
ARHGAP32	11	128842491	ENST00000310343.9:c.3868G>C	p.Asp1290His	Exon 21	HET	paternal†	0,071	0,85	rs200698355	0.001	RHOA/B/C (STRING) + RHOA (GeneMANIA)	VUS
CACNA1H	16	1252227	ENST00000348261.5: c.1778_1789delATGCCGCAGCCA	p.His593_Thr597delinsPro	Exon 9	HET	paternal†	N/A	1,174	rs768475346	0.001	PYCARD (GeneMANIA)	VUS
DIAPH1	5	140953556	ENST00000253811.6: c.1852_1860dupCCACCTCCT	p.Pro618_Pro620dup	Exon 16	HET	maternal†	N/A	0,818	N/A	0.0	RHOA/B/C (STRING) + RHOA/C (GeneMANIA)	VUS
EPHA6	3	96533572	ENST00000389672.5:c.105C>G	p.Cys35Trp	Exon 1	HET	paternal†	0,062	0,829	rs759542650	0.0	RHOA/B/C (STRING)	VUS
F10	13	113777176	ENST00000375559.3:c.7C>T	p.Arg3Cys	Exon 1	HET	paternal†	0,117	0,807	rs149972574	0.0	PYCARD (GeneMANIA)	VUS
FGD1	X	54521801	ENST00000375135.3:c.65C>T	p.Pro22Leu	Exon 1	HET	maternal†	0,133	0,515	N/A	0.0	RHOA/B/C (STRING)	VUS
GPALPP1	13	45589160	ENST00000379151.4:c.482C>T	p.Thr161Met	Exon 5	HET	paternal†	0,139	1,006	rs181252018	0.001	RHOA (GeneMANIA)	VUS
HRNR	1	152187936	ENST00000368801.2:c.6169C>T	p.Arg2057Ter	Exon 3	HET	paternal†	N/A	1,86	rs878915709	0.0	PYCARD (STRING)	VUS
MDM4	1	204511936	ENST00000367182.3:c.536C>G	p.Ala179Gly	Exon 8	HET	paternal†	0,02	0,7	rs146492402	0.0	YWHAB (GeneMANIA)	VUS
MEFV	16	3293407	ENST00000219596.1:c.2080A>G	p.Met694Val	Exon 10	HET	maternal†	0,406	1,228	rs61752717	0.094	N/A	P
MEX3B	15	82337998	ENST00000329713.4: c.40_48dupAGCGGCGGC	p.Ser14_Gly16dup	Exon 1	HET	maternal†	N/A	0,842	rs756729330	0.001	YWHAB (GeneMANIA)	VUS
PHKA2	X	18917338	ENST00000379942.4:c.3064T>A	p.Ser1022Thr	Exon 29	HET	paternal†	0,139	0,775	rs773766679	0.001	PYCARD (GeneMANIA)	VUS
PITPNM1	11	67265666	ENST00000534749.1:c.1612C>T	p.Arg538Cys	Exon 10	HET	paternal†	0,211	0,687	rs759052851	0.0	RHOA (GeneMANIA)	VUS
PTBP1	19	805158	ENST00000356948.6:c.863C>T	p.Ser288Leu	Exon 8	HET	paternal†	0,187	0,757	rs147259741	0.001	CASP1 (STRING)	VUS
RIPOR2	6	24848345	ENST00000259698.4:c.985G>A	p.Ala329Thr	Exon 12	HET	paternal†	0,068	0,718	rs35254980	0.0	RHOA (STRING) + RHOA/C (GeneMANIA)	VUS
SHROOM3	4	77660493	ENST00000296043.6:c.1167T>A	p.Ser389Arg	Exon 5	HET	paternal†	0,182	0,897	rs201389959	0.0	YWHAB (GeneMANIA)	VUS
XPA	9	100459431	ENST00000375128.4:c.144C>G	p.Tyr48Ter	Exon 1	HET	maternal†	N/A	0,557	N/A	0.0	N/A	LP

HGVS: 'Human Genome Variation Society', N/A: 'not applicable', HET: Heterozygous, P: Pathogenic, LP: 'likely' pathogenic, VUS: 'variant of uncertain significance', MAF: 'Minor allele frequency. † Since WES was not studied in the father of the case, the parental inheritance of these variants cannot be definitively stated. The possibility of de novo inheritance cannot be excluded! & Pathogenicity assessment and criteria are based on ACMG 2015 guideline.

Filtered variants of Case II.1.

Gene	Chromosome	Position	HGVS Nomenclature	Protein Change	Exon/ Intron	Zygosity	Parental Inheritance	In Silico Prediction (REVEL)	o/e Score (Gnomad)	rsID	MAF (in-house)	Functional Interactions Found with Pyrin Inflammasome Related Genes	Pathogenicity Classification &
ADAM12	10	127843804	ENST00000368679.4:c.331A>G	p.Asn111Asp	Exon 4	HET	maternal	0,055	0,843	rs367875671	0.0	RHOA (GeneMANIA)	VUS
ALB	4	74282037	ENST00000295897.4:c.1256T>A	p.Phe419Tyr	Exon 10	HET	paternal	0,253	0,897	rs1025342545	0.0	PKN1 and YWHAB (STRING)	VUS
ANXA2	15	60674597	ENST00000332680.4:c.146C>G	p.Thr49Ser	Exon 3	HET	paternal	0,066	0,943	rs780461746	0.0	PYCARD (STRING)	VUS
BCL10	1	85736574	ENST00000370580.1:c.73C>T	p.Arg25Cys	Exon 2	HET	de novo	0,680	0,648	N/A	0.0	PYCARD (STRING) - CHUK related interaction	VUS
CHUK	10	101969420	ENST00000370397.7:c.1060C>A	p.Leu354Ile	Exon 10	HET	paternal	0,119	0,608	N/A	0.0	PYCARD (STRING) + 'Nod-like receptor signal pathway' (DAVID)	VUS
CRY1	12	107393742	ENST00000008527.5:c.803A>G	p.Lys268Arg	Exon 6	HET	maternal	0,061	0,748	rs138102124	0.0	YWHAB (GeneMANIA)	VUS
FMN2	1	240371963	ENST00000319653.9:c.3851A>G	p.Lys1284Arg	Exon 5	HET	maternal	0,306	0,970	rs578174204	0.001	CASP1 (GeneMANIA)	VUS
GIGYF2	2	233680423	ENST00000373566.3:c.2250G>C	p.Gln750His	Exon 20	HET	maternal	0,262	0,763	rs895609234	0.001	YWHAB (GeneMANIA)	VUS
IL23R	1	67635297	ENST00000347310.5:c.343T>G	p.Cys115Gly	Exon 3	HET	maternal	0,436	0,829	rs746144971	0.0	IL1B (GeneMANIA)	VUS
IMP4	2	131100663	ENST00000259239.3:c.8G>C	p.Arg3Pro	Exon 2	HET	de novo	0,525	0,927	N/A	0.0	N/A	VUS
LAMB1	7	107616254	ENST00000393561.1:c.1141G>A	p.Val381Ile	Exon 8	HET	paternal	0,034	0,952	rs200303288	0.001	YWHAB (STRING)	VUS
MEFV	16	3293403	ENST00000219596.1:c.2084A>G	p.Lys695Arg	Exon 10	HET	maternal	0,353	1,228	rs104895094	0.005	N/A	P
MYH13	17	10265498	ENST00000252172.4:c.442C>T	p.Arg148Cys	Exon 5	HET	maternal	0,792	0,991	rs367783054	0.0	PSTPIP1 (GeneMANIA)	VUS
NCOA2	8	71068582	ENST00000452400.2:c.2018C>G	p.Pro673Arg	Exon 11	HET	maternal	0,067	0,773	rs61754971	0.001	PKN1 (STRING) + PKN1 (GeneMANIA)	VUS
NLRP12	19	54313570	ENST00000391773.1:c.1343G>C	p.Gly448Ala	Exon 3	HET	paternal	0,240	1,195	rs104895566	0.001	PYCARD (STRING)	VUS
PHLDB2	3	111658386	ENST00000393925.3:c.2195A>G	p.His732Arg	Exon 7	HET	paternal	0,037	0,959	N/A	0.0	YWHAB (GeneMANIA)	VUS
PPM1B	2	44445202	ENST00000282412.4:c.1061G>A	p.Gly354Glu	Exon 4	HET	maternal	0,673	0,896	N/A	0.0	PKN2 (GeneMANIA)	VUS
RPL31	2	101620666	ENST00000409038.1:c.154T>C	p.Phe52Leu	Exon 3	HET	maternal	0,310	0,485	N/A	0.0	PSTPIP1 (GeneMANIA)	VUS
SFN	1	27190265	ENST00000339276.4:c.562G>A	p.Glu188Lys	Exon 1	HET	maternal	0,364	0,694	rs761394832	0.0	YWHAB (STRING)	VUS
TACR3	4	104640580	ENST00000304883.2:c.251_252dupGC	p.Ile85AlafsTer33	Exon 1	HET	paternal	N/A	1,244	N/A	0.001	N/A	LP
TBC1D1	4	38022269	ENST00000261439.4:c.1030G>A	p.Gly344Ser	Exon 5	HET	paternal	0,138	0,918	rs758994457	0.0	YWHAB (STRING) + YWHAB (GeneMANIA)	VUS
TIAM2	6	155469341	ENST00000456144.1:c.1901G>A	p.Arg634Gln	Exon 6	HET	paternal	0,143	0,887	rs138017039	0.001	RHOA/B/C (STRING)	VUS
TTN	2	179602922	ENST00000589042.1:c.14258A>G	p.Tyr4753Cys	Exon 49	HET	maternal	0,345	1,023	rs752678561	0.0	PYCARD (GeneMANIA)	VUS

HGVS: 'Human Genome Variation Society', N/A: 'not applicable', HET: Heterozygous, P: Pathogenic, LP: 'likely' pathogenic, VUS: 'variant of uncertain significance', MAF: 'Minor allele frequency. & Pathogenicity assessment and criteria are based on ACMG 2015 guideline.

Filtered variants of Case III.1.

Gene	Chromosome	Position	HGVS Nomenclature	Protein Change	Exon/ Intron	Zygosity	Parental Inheritance	In Silico Prediction (REVEL)	o/e Score (Gnomad)	rsID	MAF (in-house)	Functional Interactions Found with Pyrin Inflammasome Related Genes	Pathogenicity Classification &
ARHGAP20	11	110450457	ENST00000260283.4:c.3213A>T	p.Leu1071Phe	Exon 16	HET	paternal	0,154	0,870	rs143937766	0.001	RHOA/B/C (STRING)	VUS
ARHGAP27	17	43507530	ENST00000428638.1:c.116G>A	p.Arg39Gln	Exon 1	HET	maternal	0,136	0,756	rs754321879	0.001	RHOA/B/C (STRING)	VUS
ARHGEF7	13	111932966	ENST00000375741.2:c.1730C>T	p.Thr577Met	Exon 16	HET	paternal	0,399	0,666	rs539695846	0.001	RHOA/B/C (STRING) + YWHAB (GeneMANIA)	VUS
C1QA	1	22964189	ENST00000402322.1:c.80G>A	p.Arg27Gln	Exon 1	HET	paternal	0,180	0,703	rs41507347	0.001	PYCARD (GeneMANIA)	VUS
C1QC	1	22973906	ENST00000374640.4:c.368C>T	p.Thr123Met	Exon 3	HET	maternal	0,571	0,900	rs369345026	0.0	PYCARD (GeneMANIA)	VUS
CENPJ	13	25484153	ENST00000381884.4:c.640G>A	p.Ala214Thr	Exon 4	HET	paternal	0,011	0,977	rs769818191	0.0	YWHAB (GeneMANIA)	VUS
CEP131	17	79170833	ENST00000269392.4:c.1679C>T	p.Pro560Leu	Exon 14	HET	paternal	0,024	0,959	rs780199740	0.001	YWHAB (GeneMANIA)	VUS
CORO7	16	4412652	ENST00000251166.4:c.1363G>A	p.Gly455Arg	Exon 15	HET	maternal	0,226	1,023	rs767466448	0.001	RHOA (GeneMANIA)	VUS
EEF1D	8	144671495	ENST00000532741.1:c.907G>A	p.Gly303Ser	Exon 1	HET	paternal	0,015	1,011	rs1399858736	0.0	PKN2 (GeneMANIA)	VUS
ERBIN	5	65350751	ENST00000506030.1:c.3605G>A	p.Arg1202Gln	Exon 21	HET	maternal	0,257	0,905	rs138617538	0.0	Nod-like receptor signal pathway (DAVID)	VUS
FAS	10	90762918	ENST00000355740.2:c.163G>C	p.Asp55His	Exon 2	HET	paternal	0,195	0,731	rs148677058	0.0	RHOA (STRING) + RHOA (GeneMANIA)	VUS
FBN1	15	48704816	ENST00000316623.5:c.8176C>T	p.Arg2726Trp	Exon 65	HET	maternal	0,569	0,645	rs61746008	0.001	RHOB (GeneMANIA)	VUS
GORASP2	2	171811246	ENST00000452526.2:c.689A>G	p.Gln230Arg	Exon 7	HET	maternal	0,139	0,714	N/A	0.0	RHOA (GeneMANIA)	VUS
HSPG2	1	22155422	ENST00000374695.3:c.12143T>C	p.Leu4048Pro	Exon 88	HET	paternal	0,857	0,939	rs1368598061	0.001	PYCARD (GeneMANIA)	VUS
IKZF3	17	37988389	ENST00000346872.3:c.23C>T	p.Ala8Val	Exon 2	HET	paternal	0,020	0,657	rs892775511	0.0	RHOA (GeneMANIA)	VUS
IRAK3	12	66639034	ENST00000261233.4:c.1306A>G	p.Met436Val	Exon 11	HET	maternal	0,231	1,012	rs555279649	0.001	IL1B (STRING) + IL1B (GeneMANIA)	VUS
IRS1	2	227659746	ENST00000305123.5:c.3709C>G	p.Gln1237Glu	Exon 1	HET	maternal	0,113	0,993	rs747703859	0.001	RHOA (STRING) + YWHAB (GeneMANIA)	VUS
ITGAV	2	187455234	ENST00000261023.3:c.169G>C	p.Val57Leu	Exon 1	HET	paternal	0,056	0,895	N/A	0.0	RHOA (STRING) + RHOA (GeneMANIA)	VUS
MAP1A	15	43817076	ENST00000382031.1: c.4120_4122delGAG	p.Glu1374del	Exon 5	HET	paternal	N/A	0,855	rs748717835	0.0	RHOB (GeneMANIA)	VUS
MARS1	12	57910091	ENST00000262027.5:c.2527C>A	p.Gln843Lys	Exon 20	HET	maternal	0,049	0,858	rs561321660	0.0	PKN2 (GeneMANIA)	VUS
MEFV	16	3293407	ENST00000219596.1:c.2080A>G	p.Met694Val	Exon 10	HET	maternal	0,406	1,228	rs61752717	0.094	N/A	P
MEFV	16	3304463	ENST00000219596.1:c.605G>A	p.Arg202Gln	Exon 2	HET	maternal	0,033	1,228	rs224222	0.23	N/A	B
NLRC4	2	32449648	ENST00000402280.1:c.2969T>G	p.Val990Gly	Exon 9	HET	maternal	0,145	0,882	N/A	0.0	IL1B, IL18, PYCARD, CASP1 (STRING) + CASP1 and PYCARD (GeneMANIA) + Nod-like receptor signal pathway (DAVID)	VUS
PELP1	17	4575900	ENST00000301396.4: c.2817_2818insG	p.Pro940AlafsTer35	Exon 16	HET	de novo	N/A	0,050	N/A	0.005	N/A	VUS
PFN1	17	4850009	ENST00000572383.1:c.476A>G	p.Gln159Arg	Exon 3	HET	maternal	0,116	0,407	N/A	0.0	RHOA/C (STRING)	VUS
PPP1R12B	1	202403894	ENST00000336894.4:c.1252A>G	p.Arg418Gly	Exon 9	HET	paternal	0,108	0,882	N/A	0.0	RHOA/B/C (STRING)	VUS
PTBP2	1	97235271	ENST00000394184.3:c.161A>G	p.Asp54Gly	Exon 5	HET	maternal	0,314	0,468	rs765144770	0.0	CASP1 (STRING)	VUS
RAPGEF3	12	48151819	ENST00000389212.3:c.49G>A	p.Val17Met	Exon 3	HET	paternal	0,123	0,897	rs755530927	0.0	YWHAB (STRING)	VUS
REEP4	8	21996564	ENST00000306306.3:c.428C>T	p.Ala143Val	Exon 6	HET	maternal	0,169	0,904	rs756827594	0.0	YWHAB (GeneMANIA)	VUS
SLC19A3	2	228566967	ENST00000258403.3:c.68G>T	p.Gly23Val	Exon 2	HET	maternal	0,906	1,063	rs121917882	0.0	N/A	P
TAB2	6	149700064	ENST00000286332.5:c.1013C>A	p.Ser338Tyr	Exon 2	HET	paternal	0,247	0,760	N/A	0.0	Nod-like receptor signal pathway (DAVID)	VUS
TP53BP2	1	223998183	ENST00000343537.7:c.322A>G	p.Lys108Glu	Exon 4	HET	paternal	0,092	0,788	rs1156872238	0.0	YWHAB (GeneMANIA)	VUS
ZC3HAV1	7	138738829	ENST00000464606.1:c.2566C>T	p.His856Tyr	Exon 11	HET	paternal	0,021	0,807	rs1346684781	0.001	YWHAB (STRING)	VUS

HGVS: 'Human Genome Variation Society', N/A: 'not applicable', HET: Heterozygous, P: Pathogenic, LP: 'likely' pathogenic, VUS: 'variant of uncertain significance', MAF: 'Minor allele frequency. & Pathogenicity assessment and criteria are based on ACMG 2015 guideline.

Filtered variants of Case IV.1.

Gene	Chromosome	Position	HGVS Nomenclature	Protein Change	Exon/ Intron	Zygosity	Parental Inheritance	In Silico Prediction (REVEL)	o/e Score (Gnomad)	rsID	MAF (in-house)	Functional Interactions Found with Pyrin Inflammasome Related Genes	Pathogenicity Classification &
ARHGEF15	17	8216371	ENST00000421050.1:c.733C>T	p.Arg245Trp	Exon 2	HET	maternal	0,181	0,912	rs371870992	0.0	RHOA/B/C (STRING) + RHOA (GeneMANIA)	VUS
ARHGEF5	7	144062733	ENST00000056217.5: c.2971_2972delCCinsGA	p.Pro991Glu	Exon 2	HET	paternal	N/A	0,8	N/A	0.0	RHOA/B/C (STRING) + RHOA (GeneMANIA)	VUS
CHRM5	15	34355796	ENST00000383263.5:c.878A>G	p.Asn293Ser	Exon 3	HET	paternal	0,208	0,802	rs148495102	0.0	PKN1/2 (GeneMANIA)	VUS
CLCN6	1	11887244	ENST00000376496.3:c.806C>T	p.Ser269Leu	Exon 10	HET	maternal	0,915	0,808	rs761839015	0.001	YWHAB (STRING)	VUS
CTSG	14	25043894	ENST00000216336.2: c.320_325delATGACA	p.Asn107_Asp108del	Exon 3	HET	paternal	N/A	0,817	N/A	0.0	PYCARD (STRING)	VUS
IFI16	1	158990150	ENST00000295809.7:c.992C>G	p.Thr331Ser	Exon 6	HET	maternal	0,003	0,919	N/A	0.0	PYCARD (STRING)	VUS
ITGB4	17	73739839	ENST00000200181.3:c.3008G>A	p.Arg1003His	Exon 26	HET	paternal	0,021	0,969	rs1311203130	0.0	YWHAB (STRING) + YWHAB (GeneMANIA)	VUS
LATS2	13	21620032	ENST00000542899.1:c.134C>A	p.Thr45Asn	Exon 2	HET	paternal	0,065	0,757	rs751006272	0.001	YWHAB (STRING)	VUS
LPAR2	19	19735100	ENST00000542587.1:c.1021G>C	p.Glu341Gln	Exon 6	HET	paternal	0,162	0,667	N/A	0.0	RHOA (STRING) + RHOA (GeneMANIA)	VUS
MEFV	16	3293407	ENST00000219596.1:c.2080A>G	p.Met694Val	Exon 10	HET	paternal	0,406	1,228	rs61752717	0.094	N/A	P
MEFV	16	3304463	ENST00000219596.1:c.605G>A	p.Arg202Gln	Exon 2	HET	paternal	0,033	1,228	rs224222	0.23	N/A	B
MYH13	17	10265676	ENST00000252172.4:c.348+1G>A	N/A	Intron 4	HET	paternal	N/A	0,739	rs188631380	0.002	PSTPIP1 (GeneMANIA)	VUS
NGF	1	115829085	ENST00000369512.2:c.332C>T	p.Ala111Val	Exon 3	HET	maternal	0,056	0,798	rs751440257	0.001	RHOA/B/C, YWHAB (STRING) + RHOA, YWHAB, PYCARD (GeneMANIA)	VUS
OBSCN	1	228524825	ENST00000570156.2:c.19529A>G	p.Gln6510Arg	Exon 76	HET	maternal	0,153	1,044	N/A	0.0	RHOA/B/C (STRING) + RHOA/B/C (GeneMANIA)	VUS
PPP3CC	8	22380233	ENST00000397775.3:c.914C>G	p.Pro305Arg	Exon 8	HET	maternal	0,731	0,78	rs750379620	0.0	YWHAB (STRING) + YWHAB (GeneMANIA)	VUS
PRKACB	1	84610087	ENST00000370685.3:c.43A>C	p.Thr15Pro	Exon 1	HET	paternal	0,157	0,445	N/A	0.0	RHOA/B/C (STRING)	VUS
PRR21	2	240982191	ENST00000486799.1:c.181_208delTCCA CGGCTCTTCACCCATGTCCCTTCA	p.Ser61ProfsTer316	Exon 1	HET	de novo	N/A	N/A	N/A	0.0	N/A	VUS
SRC	20	36024610	ENST00000373558.2:c.617T>G	p.Leu206Arg	Exon 6	HET	maternal	0,271	0,396	rs1048616788	0.0	RHOA, YWHAB (STRING) + RHOA (GeneMANIA)	VUS
TBC1D4	13	75880518	ENST00000377636.3:c.2683A>G	p.Lys895Glu	Exon 15	HET	maternal	0,201	0,926	rs369089434	0.001	YWHAB (STRING) + YWHAB (GeneMANIA)	VUS
TTN	2	179650715	ENST00000589042.1:c.2230G>A	p.Ala744Thr	Exon 14	HET	paternal	0,087	1,023	rs144639994	0.001	PYCARD (GeneMANIA)	VUS

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Filtered variants of Case V.1.

Gene	Chromosome	Position	HGVS Nomenclature	Protein Change	Exon/ Intron	Zygosity	Parental Inheritance	In Silico Prediction (REVEL)	o/e Score (Gnomad)	rsID	MAF (in-house)	Functional Interactions Found with Pyrin Inflammasome Related Genes	Pathogenicity Classification &
ATP2B2	3	10392245	ENST00000352432.4:c.2153G>A	p.Arg718His	Exon 14	HET	maternal	0,391	0,545	rs928659205	0.001	PYCARD (GeneMANIA)	VUS
BIRC2	11	102239235	ENST00000227758.2: c.1323_1327delAAGAG	p.Glu443GlyfsTer7	Exon 6	HET	de novo	N/A	0,220	N/A	0.0	CASP1 (GeneMANIA) + Nod-like receptor signal pathway (DAVID)	LP
CACNA1F	X	49082917	ENST00000376265.2:c.1450G>A	p.Asp484Asn	Exon 11	HEMIZYGOUS	maternal	0,268	0,748	rs782709793	0.001	PYCARD (GeneMANIA)	VUS
CACNA1H	16	1258205	ENST00000348261.5:c.3347G>A	p.Gly1116Glu	Exon 16	HET	maternal	0,239	1,174	rs757299200	0.001	PYCARD (GeneMANIA)	VUS
CACNA1I	22	40075377	ENST00000336649.4:c.5339G>A	p.Arg1780Gln	Exon 35	HET	maternal	0,242	0,591	rs925854645	0.001	PYCARD (GeneMANIA)	VUS
CBX4	17	77808287	ENST00000269397.4: c.1139_1153delCCTCACACCACCCGC	p.Pro380_Pro384del	Exon 5	HET	paternal	N/A	0,681	N/A	0.001	YWHAB (GeneMANIA)	VUS
CREBBP	16	3778168	ENST00000262367.5:c.6880C>T	p.Arg2294Trp	Exon 31	HET	maternal	0,533	0,709	rs763223282	0.001	IL1B (STRING)	VUS
DNAJC3	13	96443127	ENST00000602402.1:c.1358A>G	p.Glu453Gly	Exon 12	HET	maternal	0,501	0,703	rs1457856860	0.001	PYCARD (STRING)	VUS
GOLGA2	9	131019771	ENST00000421699.2:c.2671C>T	p.Arg891Trp	Exon 25	HET	maternal	0,085	0,895	rs147923606	0.001	PKN1 (GeneMANIA)	VUS
HRNR	1	152187945	ENST00000368801.2:c.6160G>T	p.Gly2054Cys	Exon 3	HET	maternal	0,058	2,027	rs61814939	0.001	PYCARD (STRING)	VUS
KRIT1	7	91865798	ENST00000340022.2:c.412_413delAT	p.Ile138TyrfsTer5	Exon 7	HET	paternal	N/A	0,319	N/A	0.001	N/A	LP
LZTR1	22	21346593	ENST00000215739.8:c.1084C>T	p.Arg362Ter	Exon 10	HET	paternal	N/A	2,280	rs189150283	0.0	N/A	LP
MCF2L	13	113736779	ENST00000375604.2:c.2085C>A	p.His695Gln	Exon 17	HET	paternal	0,269	0,804	rs201634776	0.001	RHOA (STRING) + RHOA/B/C (GeneMANIA)	VUS
MEFV	16	3293205	ENST00000219596.1:c.2282G>A	p.Arg761His	Exon 10	HET	paternal	0,352	1,228	rs104895097	0.01	N/A	LP
MET	7	116371845	ENST00000318493.6:c.1324A>G	p.Ile442Val	Exon 3	HET	maternal	0,159	0,803	rs532124885	0.001	RHOA (STRING) + RHOA (GeneMANIA)	VUS
PADI2	1	17418911	ENST00000375486.4:c.647A>G	p.Tyr216Cys	Exon 6	HET	paternal	0,142	0,932	rs1161466890	0.001	PYCARD (STRING)	VUS
RPE65	1	68897168	ENST00000262340.5:c.1228delT	p.Ser410GlnfsTer30	Exon 11	HET	paternal	N/A	0,790	N/A	0.0	N/A	LP

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Supplementary Fig. S1. Variant numbers in each of the filtering stage for individual cases.