Appendix 3. List of variants identified after filtering the exome sequencing data from patient 863.

Chr	Position	Gene	Coding Effect	Transcript Accession Code: Exon: cDNA change: Protein change	Minor Allele Frequency	
					Exome variant server	1000 Genomes database
1	18809351	*KLHDC7A*	Missense	NM_152375:exon1:c.G1876C:p.A626P	0.000231	0
1	152195729	*HRNR*	Frameshift	NM_001009931:exon2:c.1delA:p.M1fs	0	0
2	55523008	*CCDC88A*	Missense	NM_018084:exon30:c.G5192A:p.R1731Q	0.000154	0
2	168099316	*XIRP2*	Missense	NM_001199144:exon7:c.T748C:p.S250P	0.002507	0.0023
2	220032934	*SLC23A3*	Missense	NM_001144889:exon6:c.G781C:p.V261L	0.000239	0
2	231256829	*SP140L*	Missense	NM_138402:exon12:c.A992G:p.E331G	0	0
4	140811064	*MAML3*	Frameshift	NM_018717:exon3:c.1513_1514del:p.505_505del	0	0
5	38876329	*OSMR*	Missense	NM_001168355:exon3:c.C100T:p.P34S	0	0
5	40843493	*CARD6*	Missense	NM_032587:exon2:c.A523T:p.T175S	0.007074	0.0037
5	50045986	*PARP8*	Missense	NM_001178056:exon3:c.G148T:p.V50L	0	0
5	56777892	*ACTBL2*	Missense	NM_001017992:exon1:c.G643C:p.E215Q	0	0
5	139931629	*SRA1*	Frameshift	NM_001035235:exon3:c.327_328insC:p.V110fs	0	0
5	175811094	*NOP16*	Frameshift	NM_001256539:exon5:c.586_587insAC:p.R196fs	0	0
5	176931108	*DOK3*	Missense	NM_024872:exon6:c.T1367C:p.L456P	0	0
5	180622354	*TRIM7*	Stopgain	NM_203293:exon7:c.C1348T:p.Q450X	0.003597	0.01
6	139189275	*ECT2L*	Missense	NM_001195037:exon12:c.A1510G:p.I504V	0	0
6	150209805	*RAET1E*	Splicing	NM_001243325:exon5:c.515-2->TTTT	0	0
6	152725361	*SYNE1*	Missense	NM_033071:exon46:c.C6833T:p.P2278L	0	0
7	130418720	*KLF14*	Missense	NM_138693:exon1:c.C141G:p.S47R	0	0
7	150783922	*AGAP3*	Missense	NM_001042535:exon1:c.C94G:p.P32A	0	0
8	70980758	*PRDM14*	Missense	NM_024504:exon3:c.C710T:p.S237F	0	0
8	71495918	*TRAM1*	Missense	NM_014294:exon9:c.T857G:p.F286C	0.000077	0.0005
8	87560604	*CPNE3*	Missense	NM_003909:exon12:c.G955A:p.V319I	0	0.0005

8	**96259914**	***C8orf37***	**Stopgain**	**NM_177965:exon6:c.G555A:p.W185X**	**0**	**0**
8	101733670	*PABPC1*	Missense	NM_002568:exon1:c.A142C:p.T48P	0	0
8	125464321	*TRMT12*	Missense	NM_017956:exon1:c.T1153C:p.S385P	0	0
9	33798017	*PRSS3*	Missense	NM_001197098:exon3:c.A370G:p.T124A	0	0
9	74300311	*TMEM2*	Splicing	NM_013390:exon25:c.3956-2->T	0	0
9	111929421	*FRRS1L*	Missense	NM_014334:exon1:c.C151G:p.R51G	0	0
10	82187106	*FAM213A*	Missense	NM_001243778:exon5:c.C430G:p.Q144E	0	0
10	88441223	*LDB3*	Missense	NM_001080115:exon4:c.G352A:p.V118M	0.005155	0.0018
11	1017981	*MUC6*	Missense	NM_005961:exon31:c.A4820T:p.H1607L	0	0
11	1093057	*MUC2*	Missense	NM_002457:exon30:c.A4876G:p.T1626A	0	0
11	7717219	*OVCH2*	Splicing	NM_198185:exon12:c.1179+1T>A	0	0
11	46342259	*CREB3L1*	Splicing	NM_052854:exon12:c.1524-1->G	0	0
11	64112019	*CCDC88B*	Missense	NM_032251:exon14:c.G2006A:p.G669D	0.000077	0
12	7080212	*EMG1*	Splicing	NM_006331:exon1:c.125+1T>G	0	0
12	51740409	*CELA1*	Missense	NM_001971:exon1:c.A14T:p.Y5F	0	0
12	51740410	*CELA1*	Missense	NM_001971:exon1:c.T13G:p.Y5D	0	0
12	132269646	*SFSWAP*	Missense	NM_001261411:exon15:c.G2437A:p.A813T	0	0.0014
12	133198187	*P2RX2*	Missense	NM_170683:exon10:c.G1123A:p.G375R	0	0
13	21729952	*SKA3*	Splicing	NM_145061:exon9:c.1120-2->TT	0	0
13	31287979	*ALOX5AP*	Splicing	NM_001204406:exon1:c.116+1->TA	0	0
13	50100538	*PHF11*	Missense	NM_001040443:exon9:c.G785C:p.G262A	0	0
15	23891604	*MAGEL2*	Missense	NM_019066:exon1:c.C1286T:p.P429L	0	0.0037
15	42139703	*PLA2G4B*	Missense	NM_001114633:exon19:c.C2116T:p.R706W	0.000308	0.0027
15	43927973	*CATSPER2*	Missense	NM_054020:exon9:c.G1073A:p.R358Q	0.006542	0.0041
15	44889091	*SPG11*	Missense	NM_001160227:exon24:c.G4052A:p.R1351K	0	0
15	45399075	*DUOX2*	Missense	NM_014080:exon15:c.C1786T:p.P596S	0.000154	0
15	51790979	*DMXL2*	Missense	NM_001174116:exon18:c.A4442G:p.D1481G	0.009787	0.01
16	2059708	*ZNF598*	Missense	NM_178167:exon2:c.A40C:p.M14L	0	0
16	88599701	*ZFPM1*	Frameshift	NM_153813:exon10:c.1335_1338del:p.445_446del	0	0
16	89291210	*ZNF778*	Frameshift	NM_001201407:exon5:c.328_329insGTGA:p.E110fs	0	0

17	21319543	*KCNJ12*	Missense	NM_001194958:exon3:c.G889A:p.V297I	0	0
17	73626918	*RECQL5*	Splicing	NM_004259:exon13:c.1586-1->CA	0	0
19	16268208	*HSH2D*	Splicing	NM_032855:exon8:c.662+1A>-	0	0
19	50832152	*KCNC3*	Missense	NM_004977:exon1:c.A188G:p.D63G	0	0
20	61050082	*GATA5*	Missense	NM_080473:exon2:c.G496A:p.G166S	0.000084	0.0005
21	33686957	*MRAP*	Missense	NM_206898:exon5:c.C302T:p.A101V	0.000846	0.0009
21	33735605	*URB1*	Missense	NM_014825:exon11:c.T1369A:p.S457T	0.001314	0.0009
21	34166190	*C21orf62*	Missense	NM_001162496:exon2:c.T543A:p.F181L	0	0
22	19189003	*CLTCL1*	Frameshift	NM_001835:exon23:c.3601dupG:p.V1201fs	0	0
22	35743083	*TOM1*	Missense	NM_001135730:exon14:c.G1225A:p.A409T	0.000154	0
22	37331676	*CSF2RB*	Missense	NM_000395:exon12:c.C1411T:p.R471C	0	0
22	42609195	*TCF20*	Missense	NM_005650:exon1:c.C2117A:p.P706Q	0	0
X	12734899	*FRMPD4*	Missense	NM_014728:exon15:c.C2321A:p.S774Y	0	0
X	47307623	*ZNF41*	Missense	NM_007130:exon5:c.A1546G:p.T516A	0	0
X	54949462	*TRO*	Missense	NM_001039705:exon3:c.T497C:p.L166P	0	0
X	83128470	*CYLC1*	Missense	NM_021118:exon4:c.G754A:p.G252R	0.000664	0
X	83129189	*CYLC1*	Missense	NM_021118:exon4:c.G1473T:p.E491D	0.000663	0
X	100747434	*ARMCX4*	Missense	NM_001256155:exon2:c.G3858C:p.M1286I	0	0
X	102979522	*GLRA4*	Missense	NM_001024452:exon3:c.G217A:p.V73M	0	0
X	118250609	*KIAA1210*	Missense	NM_020721:exon4:c.T500G:p.F167C	0	0

The chromosome, position, gene, coding effect, transcript accession number, exon, cDNA and protein changes as well as the minor allele frequencies in the exome variant server and the 1000 Genomes database are shown for the 73 homozygous variants. The *C8orf37* mutation is highlighted in bold.