

Supplementary Table 3. Continued

Gene	Reference position	Chromosome	Coding sequence	Coverage	Allele change	Amino acid change	Functional predictions by SIFT	Patient no.
RGPD3	329534289	2	1	37	A < G	S	N	4
LIMS1	331725649	2	1	96	G < A	NS	D	4
10-Sep	332632987	2	6	104	G < A	S	N	4
LOC645529	336733665	2	3	118	T < C	NS	NR	4
POTEF	353150970	2	13	98	T < A	NS	N	4
POTEF	353185302	2	1	71	T < C	NS	N	4
TUBA3E	353260275	2	3	94	C < T	NS	N	2
ACTBL3	354657198	2	1	108	T < G	S	NR	4
THSD7B	360341133	2	11	14	G < C	S	N	2
GALNT5	380364795	2	7	19	T < G	NS	D	3
SCN9A	389348565	2	11	38	T < C	NS	N	3
SCN9A	389352545	2	9	39	T < C	S	N	4
ABCB11	391996481	2	23	27	G < A	NS	NO	3
OLA1	397151333	2	9	14	A < T	S	N	3
TTN	401781960	2	95	24	T < G	NS	D	3
TTN	401781961	2	95	24	C < T	S	N	3
SESTD1	402189020	2	14	28	G < T	NS	D	3
DUSP19	406151282	2	1	41	C < G	NS	D	3
ZNF804A	408011183	2	4	20	A < G	S	N	3
LOC200726	429716908	2	1	28	C < T	NS	NR	3
ERBB4	434456096	2	28	29	C < T	NS	N	3
RNF25	441737431	2	8	18	G < A	S	N	3
C2orf24	442244994	2	8	51	G < A	NS	N	4
C2orf24	442245306	2	8	31	A < G	NS	N	4
TUBA4A	442323330	2	4	103	C < T	NS	D	4
OBSL1	442639564	2	4	59	G < A	S	N	4
SERPINE2	447057057	2	5	24	G < A	S	N	3
DOCK10	447917888	2	20	54	G < A	NS	NO	1
DIS3L2	455310852	2	10	90	G < A	NS	N	1
ALPP	455451136	2	1	40	C < T	NS	D	4
LRRFIP1	460828821	2	11	16	A < G	S	N	1
HDAC4	462131420	2	20	77	G < A	S	N	4
ITPR1	469948734	3	21	68	A < C	S	N	4
WNT7A	479128207	3	3	47	C < T	S	N	4
ZFYVE20	480358270	3	5	13	C < T	NS	N	3
OXNAD1	481544487	3	1	101	C < T	S	N	4
RARB	490854079	3	5	14	T < C	S	N	3
EOMES	492992244	3	4	84	C < T	NS	N	1
SCN10A	504030094	3	9	56	C < T	S	N	4
SCN11A ^a	504168069	3	15	110	T < A	NS	NO	1
CX3CR1	504539250	3	1	36	T < C	NS	N	3
CTNBN1	506498027	3	2	39	G < A	NS	D	1
CCR5	511646366	3	1	20	C < A	NS	NO	3
COL7A1	513857189	3	21	50	T < C	S	N	4
RBM6	515335643	3	16	16	G < A	NS	D	3
RBM5	515386440	3	22	46	G < A	NS	D	3
GLYCTK	517558462	3	4	64	C < A	S	N	2
KBTBD8	532186608	3	2	48	G < T	NS	NO	3
FOXP1	536153715	3	13	189	A < C	NS	D	3
FOXP1	536153718	3	13	190	C < T	S	N	3
LOC100288801	540845478	3	1	220	G < A	NS	N	4
LOC100288801 ^a	540846776	3	2	228	G < A	S	N	2
EPHA3	554308286	3	2	14	T < C	S	N	3
DCBLD2	560650518	3	16	37	G < A	NS	N	3
DCBLD2	560650520	3	16	35	A < C	NS	NO	3
BOC	575123882	3	6	56	C < T	S	N	4
GPR156	582017947	3	9	64	A < T	S	N	2
HEG1	586864008	3	6	41	T < C	NS	N	3
MCM2 ^a	589457039	3	5	53	A < G	NS	N	1
RUVBL1	589951418	3	6	33	T < A	NS	D	3

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Gene	Reference position	Chromosome	Coding sequence	Coverage	Allele change	Amino acid change	Functional predictions by SIFT	Patient no.
RUVBL1	589951420	3	6	30	T < G	NS	D	3
C3orf25	591272422	3	2	44	T < C	NS	D	4
PLXND1	591434995	3	7	116	G < A	S	N	4
COL6A6	592486478	3	27	23	G < T	NS	D	3
SLCO2A1	595793430	3	11	60	A < G	NS	D	1
RYK	596026424	3	13	102	C < T	NS	N	1
ZBTB38 ^a	603296515	3	1	39	A < G	NS	N	1
PLOD2	607970905	3	3	11	T < C	S	N	2
PLSCR2	608303872	3	4	55	A < T	NS	D	3
TMEM183B	611832538	3	1	44	A < C	NS	NR	3
TSC22D2	612260917	3	1	14	T < G	S	N	4
MYNN	631624024	3	1	54	C < T	S	N	4
TNIIK ^a	633027074	3	8	63	C < G	NS	D	1
IL1RAP	652454036	3	2	73	C < A	S	N	2
MUC4	657634114	3	3	76	A < G	NS	N	4
MUC4	657640844	3	2	214	T < C	NS	N	4
FGFRL1	661098114	4	6	16	C < A	NS	N	3
TNIP2	662767650	4	6	33	G < A	NS	N	4
LOC100288212	680710684	4	2	44	G < A	S	N	3
GPR125	682521774	4	1	21	C < A	NS	N	2
TBC1D1	698120919	4	19	29	G < A	NS	N	4
SCFD2	711062849	4	1	22	A < C	NS	D	3
SCFD2	711062850	4	1	24	G < T	NS	N	3
KIAA1211	714010832	4	4	25	A < G	NS	D	3
UGT2B28	726937878	4	5	40	A < G	S	N	3
UGT2B28	726937879	4	5	39	A < T	NS	D	3
SULT1B1	727380561	4	5	28	A < C	NS	D	3
ENAM	728278770	4	2	31	G < T	NS	N	3
ANKRD17	730738780	4	29	56	T < A	S	N	3
ANKRD17	730738782	4	29	58	A < G	NS	N	3
FRAS1	735932986	4	6	28	T < A	NS	NO	3
FRAS1	735932989	4	6	28	C < T	S	N	3
AFF1	744724579	4	3	25	G < A	S	N	3
SPARCL1	745172651	4	2	66	G < T	NS	N	4
HERC6	746068457	4	4	52	T < A	NS	N	4
CXXC4	762168791	4	1	23	C < T	NS	N	3
PDE5A	777231322	4	8	81	A < G	S	N	4
FAT4	783168335	4	17	26	A < T	NS	D	3
FAT4	783168337	4	17	25	G < T	NS	N	3
INPP4B ^a	799948368	4	7	144	C < A	NS	N	1
EDNRA	805163608	4	1	58	C < T	S	N	3
RBM46 ^a	812505537	4	4	102	T < C	NS	N	1
ACCN5	813543803	4	1	41	G < A	S	N	4
1-Mar	821206546	4	4	89	C < T	S	N	4
DDX60	825930266	4	26	15	C < T	NS	N	3
DDX60	825930267	4	26	15	A < T	NS	N	3
MFAP3L	827669812	4	2	33	G < C	NS	D	3
AGA	835115128	4	5	41	T < A	S	N	3
IRF2	842095828	4	4	19	T < C	NS	D	3
SORBS2 ^a	843292510	4	13	232	C < T	NS	D	1
FAM149A	843833669	4	4	50	A < G	NS	D	4
FAT1	844287485	4	14	146	A < G	NS	N	4
FAT1	844298124	4	9	19	G < A	NS	D	3
TRIML2	845769469	4	7	40	C < T	NS	N	2
FRG2	847704735	4	1	72	T < A	NS	N	4
MAFIP	848044252	4	7	56	C < G	S	NR	4
MAFIP	848046049	4	4	33	G < A	S	NR	4
SLC6A18	849416422	5	10	180	C < T	NS	N	4
NDUFS6	849988000	5	4	70	G < T	S	N	3
TRIO	862540906	5	17	113	C < T	S	N	2

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Gene	Reference position	Chromosome	Coding sequence	Coverage	Allele change	Amino acid change	Functional predictions by SIFT	Patient no.
ANKH	862913981	5	8	116	T < C	S	N	4
FBXL7	864100358	5	3	48	A < G	NS	N	2
RNASEN	879698242	5	2	141	G < A	S	N	4
ADAMTS12	881707057	5	23	45	G < A	NS	N	4
EGFLAM	886579974	5	9	30	T < G	NS	NO	3
EGFLAM	886610442	5	17	63	G < A	S	N	2
CD180	911650805	5	3	38	G < A	S	N	3
MARVELD2	913887307	5	1	47	C < T	NS	N	4
WDR41	921906081	5	10	129	C < T	NS	N	4
GDF9	977321994	5	1	37	G < T	NS	D	3
C5orf15	978417392	5	2	18	G < A	S	N	3
KIF20A	982639408	5	3	29	T < C	S	N	3
KDM3B	982876692	5	14	17	T < C	S	N	4
LOC202051	983854576	5	6	61	C < T	S	N	4
PCDHB11	985652769	5	1	24	T < C	S	N	4
HMHB1	988272050	5	2	125	C < T	NS	D	4
ABLIM3 ^a	993692261	5	13	55	A < G	S	N	1
PDGFRB	994581527	5	9	24	A < C	NS	D	1
NDST1	994979638	5	2	57	C < T	S	N	4
NDST1	994984515	5	3	14	C < G	NS	N	3
KIF4B	999468839	5	1	50	C < A	S	N	3
KIF4B	999468844	5	1	48	A < C	NS	N	3
ADAM19	1001937293	5	21	26	C < A	NS	N	3
FBXW11 ^a	1016325320	5	8	98	T < C	NS	D	1
C5orf47	1018438284	5	1	27	T < G	S	N	3
FGFR4	1021542240	5	8	26	G < A	NS	N	4
FLT4	1025068341	5	19	33	G < C	NS	D	4
BTNL3	1025454701	5	8	38	G < T	S	N	3
TUBB2A	1029022083	6	4	28	A < G	S	N	3
LRR16A	1051287607	6	3	142	A < C	S	N	4
SLC17A4	1051637752	6	3	191	C < T	S	N	4
BTN3A2	1052237964	6	3	68	T < C	S	N	4
HLA-G	1055664896	6	5	85	C < T	S	N	4
HLA-A	1055777815	6	2	33	T < A	NS	N	2
HLA-A ^a	1055777819	6	2	85	A < C	S	N	2
C4A	1057829599	6	21	20	T < G	NS	D	2
TNXB	1057902726	6	17	35	G < A	S	N	4
BTNL2 ^a	1058229998	6	6	218	C < T	NS	N	2
BTNL2 ^a	1058230002	6	6	220	G < A	NS	N	2
HLA-DRB1	1058415838	6	4	83	A < G	S	N	4
HLA-DQB1	1058497104	6	3	50	A < G	S	N	2
HLA-DPB1	1058920866	6	4	94	G < A	NS	N	4
GRM4	1059927081	6	2	94	C < T	NS	D	2
C6orf127	1061622915	6	3	78	A < G	S	N	4
SRPK1	1061704552	6	11	58	G < T	S	N	3
SLC26A8	1061790503	6	16	180	T < C	NS	N	4
TREML2	1067029775	6	3	241	T < C	NS	N	4
TTBK1	1069098172	6	12	37	G < C	NS	N	2
HSP90AB1	1070084817	6	3	24	G < A	NS	N	2
GPR116	1072716510	6	7	52	A < C	S	N	3
CD2AP	1073430855	6	12	58	T < A	NS	N	3
PKHD1	1077359171	6	65	80	T < A	NS	N	3
GSTA2	1078484988	6	4	89	C < G	NS	N	4
GFRAL	1081063844	6	2	32	C < T	NS	N	4
PRIM2	1083334432	6	10	62	A < G	S	NR	4
PRIM2	1083379733	6	13	50	G < A	NS	NR	4
PRIM2	1083379822	6	13	79	T < C	NS	NR	4
EYS	1087198904	6	40	25	G < T	S	N	3
EYS	1087198906	6	40	19	G < C	NS	N	3
IMPG1	1099519071	6	2	37	T < C	NS	N	3

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Gene	Reference position	Chromosome	Coding sequence	Coverage	Allele change	Amino acid change	Functional predictions by SIFT	Patient no.
ME1	1106705882	6	10	19	T < A	NS	D	3
ME1	1106705883	6	10	17	T < A	NS	NO	3
GABRR1	1112675095	6	5	172	A < G	NS	N	2
MDN1	1113138459	6	88	90	G < A	S	N	4
WISP3	1134999625	6	2	157	T < G	NS	D	4
HS3ST5	1136996498	6	2	16	C < T	NS	N	3
COL10A1	1139059549	6	2	97	A < G	S	N	2
NKAIN2 ^a	1147293746	6	3	247	C < G	NS	NO	1
PERP ^a	1161034772	6	2	61	T < C	NS	N	1
HEBP2	1161351286	6	4	56	A < C	S	N	4
NHSL1	1161385538	6	4	32	G < T	NS	D	3
HIVEP2	1165711211	6	1	34	C < T	NS	D	3
SAMD5	1170447330	6	1	31	C < T	S	N	4
PCMT1	1172688307	6	1	41	T < C	NS	D	3
ZBTB2	1174303968	6	2	18	T < A	NS	N	3
SYNE1	1175082156	6	136	30	C < T	S	N	3
SYNJ2	1181103116	6	11	37	T < G	NS	N	3
TULP4	1181540258	6	13	24	A < C	NS	N	3
RSPH3	1182019083	6	6	43	A < T	NS	D	3
IGF2R	1183085535	6	16	66	A < G	S	N	4
AGPAT4	1184192475	6	3	85	C < T	NS	N	2
MLLT4	1190935073	6	19	75	A < C	S	N	4
FAM120B	1193244908	6	1	53	G < A	S	N	4
ADAP1	1194606192	7	6	69	G < A	S	N	4
MICALL2	1195144333	7	7	58	G < C	S	N	4
SDK1	1197713015	7	15	37	G < T	NS	D	3
RSPH10B	1199630320	7	18	54	C < G	S	N	3
VWDE	1206072045	7	12	19	G < T	NS	N	3
HDAC9 ^a	1212495378	7	16	176	A < T	NS	D	1
TMEM196	1213427541	7	3	117	G < T	S	N	2
ITGB8 ^a	1214065640	7	2	100	G < C	NS	D	1
C7orf10	1234451360	7	14	36	T < A	NS	N	3
C7orf10	1234451361	7	14	38	T < G	NS	NO	3
AEBP1	1237814514	7	18	22	A < T	NS	N	1
MYO1G	1238671649	7	11	87	G < A	S	N	4
C7orf65	1241361009	7	3	60	A < G	S	N	4
ABCA13	1242074221	7	33	18	A < C	NS	D	2
ABCA13	1242105751	7	39	33	C < T	S	N	3
LOC100289307	1263515207	7	2	24	G < T	NS	NR	2
MLXIPL	1263542408	7	7	13	C < T	NS	D	1
SPDYE5	1265649637	7	3	130	G < A	S	NR	4
POR	1266124069	7	2	11	G < T	S	N	3
HGF	1271854360	7	18	27	T < A	NS	D	3
SEMA3E	1273551762	7	11	83	C < T	S	N	4
SEMA3A	1274113176	7	17	291	T < C	S	N	4
FZD1	1281417809	7	1	72	G < T	S	N	3
SAMD9	1283257403	7	1	18	T < A	NS	N	1
PVRIG	1290339880	7	1	17	C < T	NS	N	1
MUC12	1291135269	7	1	14	C < T	S	N	1
MUC12	1291159789	7	5	42	C < T	S	N	4
MUC12	1291160135	7	5	46	G < A	NS	N	4
MUC12	1291160603	7	5	15	T < C	NS	N	4
MUC12	1291161436	7	5	14	G < C	S	N	4
MUC12	1291161644	7	5	52	C < T	NS	N	4
MUC12	1291165668	7	5	25	G < T	NS	D	4
MUC12	1291165672	7	5	29	A < C	S	N	4
MUC12	1291166156	7	5	79	C < G	NS	N	4
MUC12	1291166192	7	5	26	C < A	NS	N	4
MUC17	1291200140	7	3	165	G < A	NS	N	4
MUC17	1291201640	7	3	120	T < A	NS	N	4

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Gene	Reference position	Chromosome	Coding sequence	Coverage	Allele change	Amino acid change	Functional predictions by SIFT	Patient no.
MUC17	1291203834	7	3	37	T < C	S	N	2
PLOD3	1291376235	7	13	27	G < C	NS	D	4
LOC100132214	1292658130	7	12	31	A < G	NS	D	4
IFRD1	1302624679	7	8	44	T < G	S	N	4
POT1	1314997694	7	11	13	C < T	NS	N	2
POT1	1314997698	7	11	12	A < G	S	N	2
CALD1	1325140404	7	3	19	A < T	NS	N	3
TMEM140	1325371901	7	1	64	A < C	S	N	4
JHDM1D	1330288269	7	20	22	A < T	S	N	3
JHDM1D	1330288270	7	20	24	A < T	NS	D	3
TRBV7-7	1332617173	7	2	155	T < C	S	NR	4
TRBV20-1	1332996157	7	5	110	A < T	NS	NR	4
TRBV20-1	1332996161	7	5	114	C < T	NS	NR	4
LOC441294	1333766303	7	1	41	G < A	S	NR	3
LOC441294	1333766306	7	1	41	A < T	S	NR	3
CTAGE4	1334380174	7	1	29	A < T	NS	N	3
ARHGEF5L	1334381935	7	1	16	T < G	NS	D	2
EZH2	1339023228	7	5	106	C < G	NS	N	4
AGAP3	1341312932	7	7	15	G < T	S	N	1
MLL3	1342375605	7	36	28	G < A	NS	D	3
DLGAP2	1351315601	8	5	24	A < G	S	N	4
MCPH1	1356177925	8	13	40	C < T	S	N	4
FAM90A15	1356815370	8	4	30	C < G	NS	NR	2
TNKS	1359086641	8	2	19	A < T	NS	D	2
TNKS	1359086644	8	2	17	G < C	NS	D	2
RP1L1	1360114631	8	3	25	T < C	NS	N	4
RP1L1	1360116520	8	3	23	T < C	NS	N	2
C8orf74	1360204184	8	3	75	G < T	NS	D	4
MTUS1	1367102384	8	14	33	G < A	S	N	4
DOCK5	1374758768	8	10	113	G < A	S	N	4
C8orf41	1382968877	8	1	13	A < G	S	N	4
CHRNA6	1392210295	8	5	36	G < T	S	N	1
KCNB2	1420442160	8	2	17	A < C	NS	D	3
JPH1	1421750684	8	4	41	T < C	NS	N	3
ZFX4	1424211672	8	1	17	C < T	S	N	3
CA2	1432983132	8	6	24	G < A	NS	D	3
REXO1L1	1433167938	8	1	21	G < C	NS	D	4
LOC100289448	1433170685	8	1	17	G < C	NS	NR	4
RNF19A	1447714815	8	9	14	C < A	NS	N	2
ANGPT1	1454778129	8	4	54	G < T	NS	N	3
ANGPT1	1454778131	8	4	45	T < C	NS	D	3
COL14A1	1467659774	8	8	87	T < C	S	N	4
ZHX1	1470709988	8	1	42	G < T	NS	N	3
TG	1480325843	8	3	113	G < A	NS	N	2
COL22A1	1486144992	8	36	63	G < T	NS	N	4
FLJ43860	1488920394	8	19	104	G < A	S	NR	4
CYP11B2	1490439544	8	5	202	C < T	S	N	4
LY6H	1490684040	8	3	122	C < G	S	N	4
KIAA0020	1495552364	9	1	35	C < T	NS	N	4
CNTLN	1510054815	9	11	13	A < T	NS	NO	3
LINGO2	1520664425	9	1	19	G < T	S	N	3
PRSS3	1526511755	9	3	163	A < G	NS	N	4
PRSS3	1526512490	9	4	108	T < C	S	N	4
VCP	1527776116	9	8	30	T < C	NS	D	3
FAM75A1	1532072293	9	4	27	C < T	NS	N	2
ALDH1A1 ^a	1548388439	9	11	85	G < A	NS	D	1
TLE1	1557099343	9	9	23	G < T	NS	N	3
TLE1	1557099344	9	9	21	T < C	NS	N	3
WNK2	1568934596	9	27	77	A < G	NS	D	3
C9orf129	1568961550	9	2	47	T < C	NS	N	3

Supplementary Table 3. Continued

Gene	Reference position	Chromosome	Coding sequence	Coverage	Allele change	Amino acid change	Functional predictions by SIFT	Patient no.
PTCH1 ^a	1571085902	9	17	111	T < C	NS	N	1
GRIN3A	1577199512	9	9	20	A < T	NS	N	3
MUSK	1586295091	9	1	57	G < A	NS	N	3
FKBP15	1588814600	9	13	33	A < C	NS	N	4
COL27A1	1589933932	9	59	118	A < G	NS	N	4
ORM2	1589957833	9	4	37	G < C	NS	D	4
GSN	1596958694	9	17	159	T < C	S	N	4
MAPKAP1	1601110716	9	8	14	A < T	S	N	3
CDK9	1603414267	9	4	23	A < G	NS	D	3
C9orf78	1605455403	9	8	32	A < G	S	N	4
C9orf98	1608412608	9	9	23	T < A	NS	N	1
C9orf98	1608412610	9	9	24	G < A	S	N	1
GFI1B	1608580179	9	6	19	T < A	NS	D	1
GFI1B	1608580182	9	6	19	G < T	NS	NO	1
ABO	1608845331	9	7	39	C < T	NS	NR	4
ABO	1608845366	9	7	43	A < T	NS	NR	4
SARDH	1609287306	9	10	36	G < A	S	N	4
OLFM1	1610646021	9	2	93	T < C	S	N	4
PAEP	1611120049	9	4	108	C < A	NS	N	4
CACNA1B	1613566398	9	28	22	G < A	S	N	3
CACNA1B	1613566402	9	28	24	A < C	NS	N	3
PFKP	1617341440	10	9	103	C < T	S	N	4
AKR1CL2	1619063490	10	2	31	A < T	NS	D	3
ITIH2	1621971113	10	16	62	C < G	NS	N	4
BEND7	1627671894	10	7	32	T < A	S	N	4
ARMETL1	1629060660	10	2	54	T < A	NS	D	3
CUBN	1631072959	10	62	38	T < A	S	N	3
CUBN	1631276445	10	26	49	T < A	NS	N	3
MRC1L1	1632130724	10	24	11	G < T	NS	D	4
PIP4K2A	1637047251	10	6	18	T < A	NS	D	3
ARHGAP21	1639064800	10	25	109	C < T	NS	N	1
TMEM72	1656420929	10	5	46	C < T	S	N	4
ANUBL1	1657125773	10	5	19	C < T	S	N	4
ANXA8L2	1658602609	10	12	48	T < C	S	N	4
AGAP9	1658906406	10	1	38	G < T	NS	D	4
AGAP9	1658906463	10	1	30	T < G	NS	N	4
MSMB ^a	1662146277	10	2	105	A < G	NS	D	1
PCDH15	1666172531	10	34	14	G < T	NS	N	2
PCDH15 ^a	1666177751	10	32	70	A < G	S	N	1
TMEM26	1673760923	10	6	22	C < T	NS	N	3
HKDC1	1681600842	10	12	23	C < T	S	N	4
ADAMTS14	1683108476	10	21	79	A < G	NS	N	4
USP54	1685873867	10	15	93	G < A	NS	N	1
DLG5	1690161340	10	23	67	G < T	NS	N	4
FAM22B	1692061925	10	7	25	A < C	S	N	4
BMPR1A	1699271849	10	9	83	C < T	S	N	4
FAM25A	1699372551	10	2	93	A < G	S	N	4
MYOF	1705731605	10	20	24	G < C	NS	D	3
MYOF	1705731606	10	20	24	C < T	S	N	3
TLL2	1708736294	10	15	21	T < G	NS	N	3
TLL2	1708736297	10	15	22	A < T	NS	D	3
MMS19 ^a	1709816131	10	18	118	G < A	S	N	1
MMS19 ^a	1709816132	10	18	116	C < A	S	N	1
BTRC	1713888546	10	13	19	C < G	NS	D	2
POLL	1713935693	10	3	174	G < T	NS	N	4
PNLIPRP1	1728959073	10	12	57	T < C	NS	D	4
DMBT1	1734932783	10	13	13	G < T	NS	NO	1
DMBT1	1734934333	10	14	38	A < T	S	N	3
DMBT1	1734942466	10	20	106	T < C	S	N	4
CTBP2	1737305458	10	1	13	G < A	S	N	3

Supplementary Table 3. Continued

Gene	Reference position	Chromosome	Coding sequence	Coverage	Allele change	Amino acid change	Functional predictions by SIFT	Patient no.
MMP21	1738045784	10	7	33	G < A	NS	D	3
JAKMIP3	1744395894	10	10	74	A < G	NS	N	4
C10orf93	1745195014	10	4	39	G < A	S	N	2
KNDC1	1745437947	10	5	104	C < T	S	N	4
SYCE1	1745809057	10	13	80	T < C	NS	N	4
FRG2B	1745879355	10	4	105	C < T	S	N	4
SCGB1C1	1746098936	11	2	28	A < G	S	N	4
B4GALNT4	1746279024	11	8	44	G < A	S	N	4
CHID1	1746775660	11	11	37	G < A	NS	N	4
MUC2	1746998500	11	30	33	C < G	S	N	4
MUC2	1746998519	11	30	39	C < A	S	N	4
MUC5AC	1747163509	11	40	33	A < G	S	N	4
MUC5AC	1747176291	11	50	197	G < T	NS	N	4
MUC5AC	1747183167	11	59	41	G < A	NS	N	4
KRTAP5-3	1747534374	11	1	73	C < T	NS	N	3
TNNT3	1747860451	11	10	88	C < A	NS	D	2
ART1	1749586329	11	2	22	A < G	S	N	4
DCHS1	1752558224	11	6	111	C < T	NS	D	2
SOX6	1761982617	11	9	24	T < C	S	N	3
SAAL1	1764013726	11	9	29	T < C	NS	N	4
SAAL1	1764016154	11	7	38	T < A	NS	N	4
MRGPRX3	1764064511	11	1	277	G < A	S	NO	1
MRGPRX3	1764064669	11	1	70	T < C	NS	D	4
NAV2	1765806846	11	5	40	G < A	S	N	4
NAV2	1765972037	11	14	74	A < G	NS	D	3
FANCF	1768551685	11	1	24	C < T	NS	N	3
SLC5A12	1772648382	11	1	29	C < A	NS	NO	3
SLC5A12	1772648383	11	1	29	C < A	NS	N	3
MPPED2	1776462933	11	2	21	G < T	S	N	3
MPPED2	1776462935	11	2	20	C < G	NS	N	3
ZNF408	1792629936	11	4	54	T < A	NS	N	2
GLYAT	1800978539	11	3	36	G < C	NS	D	3
PGA3	1803475551	11	6	166	T < G	NS	D	4
AHNAK	1804789193	11	3	55	T < C	NS	N	4
SIPA1	1807916289	11	15	12	G < T	S	N	1
CATSPER1	1808286286	11	7	52	C < T	NS	N	4
RBM4B	1808942484	11	1	34	A < G	NS	D	3
TPCN2	1811349628	11	19	316	T < C	NS	N	4
FADD	1812550653	11	2	45	G < T	NS	N	2
C11orf30	181875429	11	19	54	T < C	S	N	4
GDPD4	1819477756	11	8	38	C < T	NS	D	3
ALG8	1820310415	11	13	26	G < A	S	N	3
GAB2	1820434380	11	5	79	A < G	S	N	4
FAT3	1835072125	11	17	53	G < T	NS	N	4
PANX1	1836411162	11	4	21	C < A	NS	N	3
PIWIL4	1836824979	11	9	76	G < C	NS	N	4
CWC15	1837197727	11	5	30	A < G	NS	NR	2
TMEM133	1843211533	11	1	84	A < C	NS	N	4
TRPC6 ^a	1843723722	11	2	65	C < T	S	N	1
TMEM123	1844621025	11	3	11	G < A	NS	D	2
ZC3H12C	1852355676	11	2	23	G < T	NS	N	3
LAYN	1853779209	11	7	40	G < A	NS	N	2
ZW10	1855955650	11	15	49	A < C	NS	N	3
CEP164	1859631014	11	31	58	G < T	S	N	4
DSCAML1	1859657009	11	25	62	G < A	S	N	4
DSCAML1	1859751449	11	4	51	G < T	NS	N	4
IL10RA	1860212261	11	4	49	A < G	S	N	4
TMPRSS4	1860336319	11	12	79	C < T	NS	D	4
BCL9L	1861117811	11	8	14	G < T	NS	D	1
CCDC84	1861234319	11	10	45	C < G	S	N	2

Supplementary Table 3. Continued

Gene	Reference position	Chromosome	Coding sequence	Coverage	Allele change	Amino acid change	Functional predictions by SIFT	Patient no.
ZNF202	1865948687	11	2	14	C < A	NS	NO	3
HSN2	1878252896	12	1	49	T < C	S	N	1
VWF	1883406765	12	25	19	T < C	NS	D	2
ACSM4	1884701983	12	11	32	G < T	NS	D	3
PRB2	1888770834	12	3	58	T < C	S	N	4
PRB2	1888770905	12	3	100	G < T	NS	N	2
PRB2	1888771519	12	3	100	C < T	NS	N	4
PIK3C2G	1895940574	12	25	19	C < T	NS	D	3
SLCO1C1	1898110764	12	9	41	A < G	S	N	4
ARID2	1920469949	12	15	35	T < C	S	N	3
AMIGO2	1921696779	12	1	29	C < A	NS	N	3
KRT86	1926923866	12	5	158	G < A	NS	N	4
KRT86	1926923877	12	5	167	C < G	S	N	4
KRT2	1927263989	12	9	24	C < A	NS	D	3
KRT2	1927263991	12	9	24	C < A	NS	D	3
NCKAP1L	1929139765	12	18	111	C < T	S	N	4
RDH16	1931575957	12	1	144	G < T	NS	D	2
LRP1	1931814492	12	54	29	C < T	S	N	4
LRIG3	1933499258	12	13	28	T < A	NS	D	3
TMEM5	1938398588	12	1	72	G < A	S	N	4
RASSF9	1960454767	12	1	16	T < A	NS	NO	3
C12orf12	1965572650	12	1	74	C < A	NS	N	3
NUP37	1976695411	12	7	27	A < C	NS	D	3
USP30	1983694022	12	8	37	C < T	S	N	4
C12orf51	1986840857	12	36	47	T < C	S	N	2
DDX54	1987789501	12	7	85	G < A	S	N	4
PLBD2	1987987438	12	5	27	C < T	NS	NO	3
SDSL	1988046794	12	4	40	G < T	NS	N	3
MED13L	1990588227	12	24	18	C < A	NS	D	3
CIT	1994346828	12	24	71	T < C	S	N	4
ORAI1	1996254022	12	2	53	C < T	S	N	4
B3GNT4	1996814014	12	1	20	C < G	NS	N	4
CLIP1	1996970528	12	4	25	G < A	S	N	2
SBNO1	1997954707	12	3	23	T < C	NS	N	3
SETD8	1998000144	12	3	24	C < T	S	N	2
GPR133	2005609817	12	9	29	A < G	S	N	3
POLE	2007276853	12	12	66	T < C	NS	D	3
PGAM5	2007320186	12	6	59	C < T	NS	N	4
TPTE2	2008847341	13	18	64	A < G	S	N	4
TPTE2	2008847342	13	18	61	T < A	NS	N	4
PARP4	2013890787	13	15	79	G < A	S	N	1
SLC7A1	2018953795	13	2	58	G < C	S	N	4
NBEA	2024476994	13	7	190	T < C	S	N	4
DCLK1	2025231759	13	11	53	G < T	S	N	4
KBTBD6	2030552300	13	1	29	C < T	NS	N	4
MED4	2037507221	13	3	16	T < A	NS	D	3
RB1	2037880563	13	20	49	G < A	S	D	1
RCBTB1	2038988090	13	1	88	G < A	S	N	4
PCDH9	2056646696	13	1	23	T < A	NS	NO	3
PCDH9	2056646999	13	1	39	C < A	NS	N	3
KLF12	2063234134	13	4	43	C < G	NS	N	3
COL4A1	2099524302	13	37	135	T < A	S	N	4
C13orf16	2100677265	13	2	112	C < T	S	N	4
ATP11A	2102082860	13	29	44	T < C	S	N	4
RASA3	2103108926	13	21	105	C < T	S	N	4
POTEG	2104010042	14	1	119	C < T	NS	N	4
P704P	2104476800	14	1	60	C < T	S	N	4
NDRG2	2105942492	14	15	51	G < C	NS	D	4
HAUS4	2107873469	14	7	11	G < T	S	N	2
HOMEZ	2108202700	14	2	14	A < T	NS	N	3

Supplementary Table 3. Continued

Gene	Reference position	Chromosome	Coding sequence	Coverage	Allele change	Amino acid change	Functional predictions by SIFT	Patient no.
DHRS4	2108891643	14	4	116	C < T	NS	NO	4
DHRS4L2	2108914889	14	1	132	G < T	NS	D	3
DHRS4L2	2108916099	14	2	35	G < A	S	N	4
GZMH	2109533483	14	3	196	G < C	NS	N	4
GZMH	2109533541	14	3	38	C < A	NS	D	3
C14orf182	2134929075	14	1	27	A < T	NS	D	3
MAP4K5	2135379887	14	13	28	G < A	NS	D	3
PPM1A	2145206418	14	2	80	G < T	NS	N	3
SYNE2	2148878083	14	7	85	A < G	NS	D	1
PLEKHG3	2149666422	14	14	36	C < A	NS	D	2
PLEKHG3	2149666423	14	14	38	C < T	NS	D	2
GPHN	2151839368	14	6	21	G < A	S	N	3
SIPA1L1	2156512152	14	1	22	A < T	S	N	3
DIO2	2165125971	14	3	63	C < T	S	N	3
FLRT2	2170544935	14	1	20	G < T	NS	N	3
DDX24	2178978135	14	6	101	A < C	NS	D	2
BEGAIN	2185466810	14	4	97	T < C	S	N	4
C14orf73	2188025488	14	2	16	T < G	S	N	2
TMEM179	2189527477	14	1	28	C < G	NS	N	2
ADSSL1	2189666079	14	10	107	G < A	NS	D	2
AHNAK2	2189862548	14	7	57	G < A	S	N	4
AHNAK2	2189862844	14	7	26	A < C	NS	N	4
LOC727832	2192485885	15	8	14	A < G	NS	N	1
C15orf2	2196518354	15	1	17	C < G	S	N	3
GOLGA8G	2200364560	15	8	34	T < A	NS	D	3
GOLGA8G	2200368232	15	3	23	T < G	NS	D	4
CHRNA7	2203996850	15	7	134	G < A	S	N	4
RYR3	2205498123	15	33	72	T < G	S	N	4
RYR3	2205507714	15	37	44	T < G	NS	D	1
SRP14	2211874811	15	5	170	G < A	S	N	4
STARD9	2214531200	15	23	19	A < T	NS	N	3
DMXL2	2223337705	15	18	32	A < G	NS	N	4
RNF111	2230919345	15	7	17	C < T	S	N	3
ANXA2	2232187466	15	12	29	G < A	NS	D	2
ITGA11	2240170436	15	14	59	G < A	S	N	4
GOLGA6B	2244497986	15	4	49	G < A	S	N	4
GOLGA6	2245910065	15	15	18	G < T	NS	N	1
CYP1A2	2246588818	15	1	41	A < G	NS	D	3
GOLGA6C	2247104814	15	11	23	A < T	NS	N	4
GOLGA6C	2247104889	15	11	35	G < A	NS	N	4
GOLGA6C	2247106801	15	13	22	G < A	NS	N	2
CSPG4	2247528218	15	3	39	G < A	NS	D	2
SGK269	2248972147	15	3	11	G < C	NS	N	3
KIAA1024	2251295855	15	1	14	C < A	NS	N	3
AP3B2	2254828471	15	20	55	A < G	S	N	1
LOC100288732	2260124462	15	5	24	G < C	NS	NR	3
LOC100288732	2260124464	15	5	27	T < A	NS	NR	3
KIF7	2261638402	15	3	16	T < G	NS	D	2
SEMA4B	2262210365	15	5	49	G < A	NS	N	4
FURIN	2262865860	15	3	115	T < G	NS	D	4
MEF2A	2271698884	15	9	28	A < C	NS	N	3
ADAMTS17	2271960760	15	22	65	T < C	NS	N	4
HBA2	2274131104	16	3	26	T < G	S	N	3
PDIA2	2274242118	16	2	51	G < C	S	N	4
JMJD8	2274641266	16	4	23	T < A	NS	N	3
PRSS22	2276813235	16	4	38	C < T	NS	N	4
CLDN9	2276971003	16	1	43	C < T	S	N	1
ALG1	2279037309	16	9	58	T < C	S	N	4
TMEM114	2282529635	16	1	33	A < T	NS	D	4
TEKT5	2284677496	16	5	142	T < C	NS	N	4

Supplementary Table 3. Continued

Gene	Reference position	Chromosome	Coding sequence	Coverage	Allele change	Amino acid change	Functional predictions by SIFT	Patient no.
NOMO2	2292457809	16	10	19	T < A	NS	N	4
TMC5	2293409286	16	16	94	C < T	NS	NO	1
ACSM5 ^a	2294342868	16	5	162	C < G	NS	N	1
ACSM5	2294348591	16	7	60	C < G	NS	D	4
NPIPL3	2295321650	16	8	32	A < G	S	N	4
OTOA	2295663836	16	23	35	C < T	NS	D	4
VWA3A	2296051856	16	20	40	C < T	NS	N	4
PRKCB	2298073558	16	10	19	C < T	NS	NO	3
KIAA0556	2301696816	16	27	72	G < A	NS	N	4
TUFM ^a	2302763330	16	6	66	C < A	S	N	2
INO80E	2303915676	16	3	14	A < G	S	N	3
POL3S	2305006549	16	3	23	T < C	NS	N	4
ERAF	2305447472	16	2	18	G < T	S	N	4
LOC100287647	2307849444	16	2	151	G < A	S	NR	1
ABCC12	2310775449	16	28	22	T < G	NS	D	3
ABCC12	2310775450	16	28	22	C < A	NS	D	3
ABCC12	2310796723	16	19	112	G < A	S	N	1
BRD7	2313015114	16	12	42	A < C	NS	NO	4
SALL1	2313830540	16	2	19	A < C	NS	D	3
CETP	2319673630	16	14	59	G < A	NS	N	4
SETD6	2321208022	16	5	48	G < A	S	N	4
CDH5	2329078423	16	2	105	C < T	S	N	4
PDPR	2332847939	16	17	49	C < T	S	N	4
PKD1L2	2343889813	16	7	21	G < A	NS	NR	4
MPHOSPH6	2344839892	16	5	61	G < A	S	N	4
CRISPLD2	2347537002	16	2	20	A < G	NS	N	4
FAM38A	2351390771	16	35	14	C < T	NS	N	4
FAM38A	2351393656	16	33	16	A < C	NS	D	1
WDR81	2354433283	17	1	18	G < A	S	N	4
WDR81	2354443084	17	10	67	C < T	S	N	4
TSR1	2355042000	17	1	34	G < A	NS	D	3
TRPV3	2356238371	17	7	155	C < T	S	N	4
TRPV3	2356249176	17	4	23	T < C	NS	N	4
ITGAE	2356435127	17	24	49	G < A	NS	D	4
ITGAE	2356463343	17	9	31	G < A	S	D	4
ZZEF1	2356772099	17	28	13	C < A	NS	N	3
GGT6	2357265990	17	1	13	G < A	NS	N	4
CXCL16	2357444046	17	3	22	C < T	S	N	4
TEKT1	2359518533	17	5	87	G < A	NS	D	4
AMAC1L3	2360187655	17	2	35	G < A	NS	N	4
AMAC1L3	2360187863	17	2	20	G < A	NS	N	4
AMAC1L3	2360188508	17	2	179	T < C	NS	N	4
TP53 ^a	2360379830	17	6	38	G < A	NS	D	1
MYH13	2363018801	17	28	98	G < A	S	N	4
COX10	2366807730	17	4	62	G < A	S	N	4
COX10	2366897810	17	6	105	C < T	S	N	4
FAM18B2	2368251449	17	5	57	A < G	NS	N	4
FAM18B2	2368259332	17	3	141	A < C	S	N	4
TBC1D26	2368443097	17	3	80	A < C	NS	D	4
TBC1D26	2368443106	17	3	90	A < G	NS	N	4
SHMT1	2371041192	17	7	30	C < T	S	N	3
LGALS9C	2371193284	17	4	95	C < T	S	N	4
LGALS9C	2371193294	17	4	83	G < A	NS	N	4
ULK2	2372501833	17	19	21	G < T	S	N	3
KCNJ12	2374121073	17	1	157	G < A	NS	D	4
KCNJ12	2374121499	17	1	55	C < T	NS	D	4
KCNJ12	2374121521	17	1	67	G < C	NS	N	4
KIAA0100	2376657621	17	24	46	A < C	NS	N	4
SUPT6H	2376730797	17	36	14	C < T	S	N	1
CCL8 ^a	2382349668	17	2	51	A < G	NS	N	1