Supplementary Table 3. Continued

Gene	Reference position	Chromosome	Coding sequence	Coverage	Allele change	Amino acid change	Functional predictions by SIFT	Patient no.
NBPF15	126071852	1	4	159	A < G	S	N	4
HRNR	129676605	1	2	214	G < A	S	N	4
HRNR	129676984	1	2	52	G < C	NS	NO	3
HRNR	129677003	1	2	88	C < T	NS	N	4
FLG[a]	129766583	1	2	89	C < G	NS	N	2
FLG	129767962	1	2	102	C < T	NS	N	1
FLG	129768306	1	2	39	T < C	NS	N	4
FLG	129768312	1	2	59	C < G	NS	N	4
FLG	129771039	1	2	449	G < A	NS	N	2
FLG	129771228	1	2	298	C < G	NS	N	4
FLG	129773236	1	2	135	T < C	NS	N	2
FLG	129773862	1	2	222	G < C	NS	N	4
FLG	129774814	1	2	232	T < C	NS	N	4
PGLYRP3	130769598	1	2	118	C < T	S	N	4
CLK2	132724561	1	8	17	C < T	NS	D	3
CLK2	132724562	1	8	17	G < T	S	N	3
MSTO1	133072971	1	11	33	T < G	S	N	3
GON4L	133214185	1	27	50	C < A	NS	D	2
IQGAP3	134016387	1	12	66	C < G	NS	N	4
PEA15	137673244	1	3	25	A < T	NS	N	3
HSPA6	138985040	1	1	34	C < T	NS	D	4
NUF2	140800188	1	8	38	C < A	NS	NO	3
FAM78B	143529898	1	2	204	C < G	S	N	4
F5	147009112	1	10	162	C < T	NS	N	4
FAM5C[a]	167558142	1	7	33	G < A	NS	N	1
ZBTB41	174618823	1	10	13	A < C	NS	NO	2
KIF21B[a]	178450152	1	18	56	T < C	S	N	1
TMEM9	178602981	1	4	127	A < G	S	N	4
ELF3	179471218	1	2	54	C < G	S	N	4
PPP1R12B	180023641	1	21	22	C < A	NS	N	3
KDM5B	180267325	1	1	13	G < C	NS	D	1
CHI3L1	180642801	1	5	141	T < C	NS	D	4
FAM71A	189989294	1	1	133	T < C	NS	N	3
MIA3	200015587	1	13	40	T < C	S	N	4
JMJD4	205110357	1	6	53	C < T	S	N	4
OBSCN[a]	205602418	1	8	22	T < C	NS	D	1
RHOU	206063445	1	2	52	C < G	S	N	4
GNPAT	208576822	1	2	47	C < T	NS	D	3
LYST	213162183	1	3	14	G < T	NS	N	2
ADSS	221776216	1	7	22	A < C	NS	D	3
ADSS	221776218	1	7	22	C < T	S	N	3
KIF26B	223037622	1	11	110	C < T	S	N	4
LOC391343	227830117	2	1	41	T < G	NS	NR	4
LOC391343	227830313	2	1	16	G < C	S	NR	3
PXDN	228577682	2	17	144	G < C	NS	N	4
ODC1	237355517	2	10	27	G < A	S	N	4
APOB	247947498	2	16	24	A < C	NS	N	3
APOB	247947499	2	16	24	A < T	NS	N	3
ALK	256154817	2	15	59	A < G	S	N	4
FSHR	275890251	2	10	21	G < T	NS	NO	3
C2orf63	282104344	2	10	39	G < A	NS	N	4
CYP26B1	299058969	2	6	23	G < A	S	N	1
CCDC142	301407728	2	4	37	G < A	NS	NO	2
ST3GAL5	312787861	2	3	17	T < C	NS	N	4
KIAA1310	319723935	2	13	14	G < T	NS	N	3
ACTR1B	320724551	2	6	276	C < T	S	N	2
CHST10	323459632	2	5	57	G < C	S	N	4
MAP4K4	324943015	2	26	37	G < C	NS	D	3
SLC9A4	325569668	2	3	69	A < T	NS	D	3
TGFBRAP1	328335511	2	10	51	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.
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
CTNNB1	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

Supplementary Table 3. Continued

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
TNIK[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

Supplementary Table 3. Continued

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
LRRC16A	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

Supplementary Table 3. Continued

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

Supplementary Table 3. Continued

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
ZFHX4	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	1818755429	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

Supplementary Table 3. Continued

Gene	Reference position	Chromosome	Coding sequence	Coverage	Allele change	Amino acid change	Functional predictions by SIFT	Patient no.
TBC1D3B	2384202011	17	5	27	C < T	NS	D	4
TBC1D3E	2384401830	17	9	36	A < T	NS	D	4
TBC1D3D	2385938140	17	2	61	A < G	NS	N	4
TBC1D3D	2385940014	17	4	85	C < A	NS	N	4
TBC1D3D	2385940944	17	6	147	G < A	NS	N	4
ERBB2	2387531879	17	17	51	A < G	NS	N	4
TOP2A	2388219990	17	9	19	T < C	S	N	3
KRT25	2388559828	17	4	159	G < A	S	N	2
KRT26	2388580603	17	1	18	A < T	S	N	3
KRT40	2388787496	17	6	48	A < G	S	N	4
KRTAP3-2	2388808375	17	1	95	T < C	NS	N	4
KRTAP1-1	2388849768	17	1	37	G < C	NS	D	4
KRTAP4-1	2388993082	17	2	33	G < C	NS	N	4
KRTAP4-1	2388993086	17	2	30	A < G	S	N	4
KRTAP9-4	2389058283	17	1	217	C < T	NS	D	4
KRTAP9-4	2389058336	17	1	39	A < C	NS	N	3
KRTAP9-9	2389063986	17	1	44	A < C	NS	N	4
TUBG1	2390418855	17	10	14	C < A	S	N	1
BRCA1	2390878791	17	13	31	C < T	NS	NO	3
NAGS	2391737375	17	6	45	G < A	NS	N	3
CDK5RAP3	2395703185	17	3	61	T < G	NS	D	2
FAM117A	2397445898	17	7	17	G < A	S	N	3
ITGA3	2397801147	17	6	33	T < C	S	N	4
NOG	2404324293	17	1	67	A < C	NS	N	3
MTMR4	2406238200	17	6	13	A < T	NS	NO	3
CSH2	2411602334	17	4	55	C < T	NS	N	4
GH2	2411610386	17	4	18	G < C	NS	N	1
TEX2	2411943294	17	1	21	A < T	NS	D	3
COG1	2420849730	17	7	78	C < T	S	N	4
GPR142[a]	2422019077	17	3	57	A < G	NS	D	1
UNK	2423468317	17	14	19	A < G	S	N	4
QRICH2	2423941144	17	4	44	T < G	NS	N	4
HRNBP3	2426764023	17	1	22	G < A	S	N	4
CBX4	2427461157	17	5	66	C < T	NS	N	2
RNF213	2427979649	17	9	73	G < A	S	N	4
MYL12A	2434375274	18	1	40	G < A	NS	N	3
MYL12A	2434375275	18	1	41	A < T	NS	D	3
AMAC1L1	2442732047	18	1	22	G < A	S	N	2
C18orf1	2444767403	18	5	65	A < G	S	D	3
LOC729774	2445483471	18	2	38	G < T	NS	NR	3
POTEC	2445664872	18	1	68	T < C	NS	N	2
CTAGE1	2448017831	18	1	35	C < T	NS	N	1
KCTD1	2452149054	18	1	18	T < G	NS	N	3
DSG4	2457014977	18	15	41	G < T	NS	NO	3
FAM59A	2457889818	18	4	30	G < T	S	N	3
FAM59A	2457889821	18	4	29	A < C	NS	D	3
MOCOS	2461870479	18	15	143	T < C	NS	N	4
SLC14A2	2471234230	18	4	39	G < A	NS	N	3
KIAA1632	2471505962	18	25	17	A < T	NS	N	3
KIAA1632	2471505963	18	25	17	A < T	NS	NO	3
FUSSEL18	2472796815	18	1	35	A < T	NS	D	1
ZBTB7C	2473578004	18	2	78	T < G	NS	D	4
ZBTB7C	2473588900	18	1	208	T < C	S	N	4
KIAA0427	2474259908	18	6	22	T < A	NS	N	3
CXXC1	2475832249	18	10	67	A < G	S	N	4
TCF4	2480799098	18	12	33	C < A	NS	D	3
TCF4	2481003232	18	3	19	C < A	S	N	3
CCBE1	2485008619	18	4	99	C < T	S	N	2
NETO1[a]	2498404004	18	3	115	A < T	NS	D	1
C19orf6	2506845912	19	4	13	T < G	NS	D	1

Supplementary Table 3. Continued

Gene	Reference position	Chromosome	Coding sequence	Coverage	Allele change	Amino acid change	Functional predictions by SIFT	Patient no.
ABCA7	2506887468	19	25	15	A < G	S	N	4
REXO1	2507659336	19	3	14	C < T	S	N	4
FAM108A1	2507714358	19	2	13	T < C	NS	N	4
PIP5K1C	2509476658	19	13	27	G < A	NS	N	3
MAP2K2	2509934448	19	6	47	C < T	NS	D	4
ACER1	2512145698	19	3	26	T < C	NS	N	4
LASS4	2514105354	19	7	55	T < C	S	N	4
MUC16	2514735984	19	51	113	T < C	NS	N	4
MUC16	2514735995	19	51	104	C < T	NS	N	4
MUC16	2514783318	19	5	66	C < T	NS	N	4
MUC16	2514793493	19	3	59	C < T	NS	N	4
MUC16	2514797580	19	3	46	T < C	S	N	3
ICAM3	2516182922	19	2	22	T < C	NS	N	4
MAST1	2518717997	19	26	18	G < A	S	N	3
CYP4F12	2521526643	19	5	51	T < C	NS	N	4
OR10H2	2521572582	19	1	89	C < T	S	N	4
AP1M1	2522077812	19	11	81	C < G	NS	D	3
CPAMD8	2522819358	19	17	55	G < A	NS	N	3
KIAA1683	2524110904	19	2	62	C < G	NS	N	4
ISYNA1	2524280086	19	7	57	T < C	S	N	4
KIAA0892	2525185534	19	7	61	G < A	S	N	3
ZNF536	2533672403	19	3	28	T < C	S	N	4
GPI	2537503303	19	7	71	C < G	S	N	4
CD22	2538462717	19	5	17	C < G	S	N	3
C19orf15	2541461368	19	1	88	T < C	NS	N	4
MAP4K1	2541732174	19	14	25	G < A	NS	N	4
CAPN12	2541857821	19	18	71	A < G	S	N	4
LGALS4[a]	2541932950	19	3	30	C < A	NS	D	1
ECH1	2541939937	19	9	29	C < T	NS	D	4
PLEKHG2	2542544839	19	12	22	C < T	S	N	3
FCGBP	2543017507	19	21	76	G < A	S	N	4
FCGBP	2543053201	19	6	13	G < T	S	N	1
SNRPA	2543896811	19	2	60	A < G	S	N	4
CYP2F1	2544255597	19	1	31	G < A	S	N	4
ERF	2545386691	19	4	32	G < A	S	N	4
PSG3	2545867483	19	4	156	C < A	S	N	4
PSG8	2545901763	19	2	60	C < A	NS	D	3
CEACAM20	2547650657	19	7	58	T < C	NS	NR	4
ERCC2	2548501717	19	6	71	T < G	S	N	4
EMP3	2551464282	19	2	40	G < T	NS	NO	1
TMEM143	2551479358	19	6	19	A < C	NS	D	3
PTH2	2552559200	19	2	22	G < C	NS	N	3
SHANK1	2553853011	19	2	18	T < C	NS	N	1
ZNF808[a]	2555691982	19	3	39	G < A	NS	N	1
ZNF765	2556544681	19	3	19	C < G	NS	N	1
ZNF765	2556544684	19	3	19	T < C	S	N	1
ZNF761	2556586232	19	2	16	G < A	S	NR	4
LILRB3[a]	2557359732	19	3	208	G < C	NS	N	2
LILRA1	2557740721	19	5	61	T < C	NS	N	2
KIR2DL4	2557949666	19	3	17	C < G	NS	N	3
KIR3DL1	2557963191	19	3	109	A < G	S	N	4
KIR2DS4	2557982650	19	3	37	T < G	S	NR	2
KIR2DS4	2557982701	19	3	36	G < T	NS	NR	2
KIR2DS4	2557982728	19	3	21	G < C	NS	NR	2
RDH13	2558201492	19	1	28	C < T	NS	N	4
RDH13	2558201493	19	1	29	G < T	NS	N	4
ZFP28	2559692350	19	3	33	C < A	S	N	3
ZNF550[a]	2560701027	19	1	67	C < T	NS	D	1
ZSCAN22	2561483213	19	2	17	T < G	NS	N	3
KIR2DS1	2561871082	19	3	153	A < G	S	NR	2

Supplementary Table 3. Continued

Gene	Reference position	Chromosome	Coding sequence	Coverage	Allele change	Amino acid change	Functional predictions by SIFT	Patient no.
SIGLEC1	2565630685	20	3	19	C < T	NS	N	4
PAK7	2571487871	20	4	131	C < T	NS	N	4
FLRT3	2576252200	20	1	17	T < G	NS	D	3
CST9L	2585490888	20	2	216	T < G	NS	N	4
BPI	2595650275	20	11	148	A < G	NS	N	4
LBP	2595672219	20	2	23	G < A	S	N	4
LBP[a]	2595691968	20	10	25	G < T	S	N	1
KIAA1219	2595847726	20	10	14	T < G	NS	D	3
PTPRT	2599404902	20	31	37	C < A	NS	D	3
SEMG2	2602544774	20	2	42	A < G	S	N	4
ZNF335	2603273210	20	21	32	A < G	S	N	4
PCK1	2614832083	20	3	52	A < G	S	N	4
CTSZ	2616266012	20	5	51	A < G	S	N	4
OGFR	2620085220	20	4	20	G < A	S	N	4
KCNQ2	2620689776	20	14	14	C < G	NS	N	4
LOC100132288	2622007697	21	2	24	C < T	NS	NR	4
LOC100288017	2623267208	21	1	18	G < A	NS	NR	2
POTED	2623681322	21	1	60	G < A	NS	N	2
KRTAP13-2	2640442822	21	1	81	A < T	NS	D	3
C21orf66	2642816546	21	12	14	A < C	NS	N	1
C21orf66	2642816547	21	12	14	C < T	NS	N	1
WRB	2649461227	21	2	73	G < T	NS	NO	4
WRB	2649461228	21	2	73	A < T	NS	D	4
DSCAM	2650145634	21	27	116	G < C	S	N	4
PRDM15	2651870131	21	31	65	G < A	S	N	4
PFKL	2654380692	21	4	19	C < T	S	N	4
KRTAP10-6	2654660390	21	1	137	G < A	S	N	4
KRTAP12-2	2654734983	21	1	59	C < T	NS	N	4
KRTAP12-2	2654735333	21	1	71	G < A	S	N	4
KRTAP12-2	2654735334	21	1	69	C < T	NS	N	4
COL6A2	2656200935	21	27	63	C < G	S	N	3
FTCD	2656222648	21	2	55	T < A	NS	D	2
CECR5	2658218162	22	6	14	T < G	NS	D	3
CECR2	2658624712	22	16	25	C < T	S	N	4
LOC100288065	2658662354	22	4	63	A < G	NS	N	4
TBX1	2660347982	22	4	51	C < T	S	N	4
ZNF280B[a]	2663338671	22	1	78	G < A	S	N	2
C22orf30	2672604693	22	3	17	C < T	NS	NO	3
ISX	2675974809	22	2	60	G < A	S	N	1
HMGXB4	2676157665	22	4	42	A < T	S	N	3
APOL1	2677147183	22	2	27	C < T	NS	N	1
TMPRSS6	2677959079	22	17	144	G < A	S	N	4
TMPRSS6	2677959089	22	17	133	A < G	NS	N	4
SSTR3	2678099174	22	1	18	G < A	S	N	4
APOBEC3A	2679853734	22	3	53	C < T	S	N	4
L3MBTL2	2682109341	22	5	24	C < T	S	N	4
NAGA	2682959967	22	3	46	C < T	S	N	4
TTLL12	2684071796	22	5	53	C < T	S	N	3
SCUBE1	2684110469	22	15	48	C < G	S	N	4
LOC100289317	2686220100	22	1	25	A < G	NS	NR	4
CELSR1	2687425845	22	1	58	A < G	NS	N	4
MAPK8IP2	2691494874	22	11	52	G < A	NS	N	4
CD99	2693971425	X	6	16	G < A	NS	D	4
PRKX	2694923444	X	2	88	G < A	S	N	4
ARHGAP6[a]	2702537703	X	4	81	G < A	NS	D	2
DMD	2723711682	X	37	92	T < C	NS	N	4
WAS	2739828502	X	11	13	G < T	NS	N	3
GATA1	2739931290	X	2	14	A < C	NS	N	3
GAGE12E	2740547782	X	1	62	G < T	NS	D	4
PAGE1[a]	2740686656	X	3	37	T < C	S	N	1

Supplementary Table 3. Continued

Gene	Reference position	Chromosome	Coding sequence	Coverage	Allele change	Amino acid change	Functional predictions by SIFT	Patient no.
USP27X	2740875896	X	1	39	G < C	NS	NR	3
TSPYL2	2744246047	X	6	76	A < G	NS	N	3
FAM120C	2745238526	X	14	66	C < A	NS	D	2
ITIH5L	2745914425	X	8	22	G < A	NS	N	3
MSN	2752987418	X	9	47	G < A	S	N	2
OPHN1	2755314567	X	20	47	T < C	NS	N	3
DGAT2L6	2757452592	X	5	20	C < T	S	N	3
LPAR4	2765991350	X	1	46	G < T	NS	D	3
LPAR4	2765991352	X	1	43	C < T	S	N	3
PCDH11X	2779854213	X	7	171	C < A	NS	N	3
SYTL4	2787924130	X	9	24	G < A	S	N	3
SYTL4	2787924131	X	9	24	T < C	NS	D	3
NXF5	2789077402	X	3	81	T < C	NS	D	1
NXF2	2789554971	X	10	19	C < T	S	N	3
CLDN2	2794152808	X	1	26	C < A	NS	N	3
CLDN2	2794152809	X	1	26	C < T	NS	N	3
TRPC5	2799176319	X	1	24	G < A	NS	D	3
TRPC5	2799176320	X	1	24	G < T	NS	N	3
RHOXF2B	2807087244	X	4	29	A < G	NS	N	3
PLAC1	2821580905	X	1	20	A < G	NS	N	3
RBMX	2823837181	X	8	40	G < C	NS	D	4
SLITRK4	2830598721	X	1	56	T < C	NS	N	3
NSDHL	2839816887	X	6	109	A < G	NS	D	3
MPP1	2841799005	X	5	36	T < C	S	N	3
MPP1	2841799006	X	5	36	T < C	NS	N	3
RBMY1D	2863471281	Y	11	23	T < C	S	N	1

SIFT, Sorting Intolerant From Tolerant; N, nonsynonymous mutation; D, deleterious; N, neutral; S, synonymous mutation; NO, nonsense mutation; NR, no record found.
[a]These genes were commonly mutated in the synchronously developed HCCs from patients 1 and 2.

Supplementary Table 4. Functional Relevance of Mutations Detected in HCC Tumors

Patient 1					
HCC 1	No. of mutated genes	51			
	Amino acid change (+)	38 (74.5%)			
	Functional loss[a] (+)	20 (39.2%)			
	KEGG pathway				
	Pathways in cancer	CTNNB1	PDGFRB	TP53	
	Wnt signaling pathway	FBXW11			
	PI3K-Akt signaling pathway	ITGB8			
	Others	ALDH1A1	HDAC9	SORBS2	RYR3
HCC 2	No. of mutated genes	79			
	Amino acid change (+)	58 (73.4%)			
	Functional loss[a] (+)	23 (29.1%)			
	KEGG pathway				
	Viral carcinogenesis	HDAC9	RB1	TP53	
	Wnt signaling pathway	FBXW11			
	PI3K-Akt signaling pathway	ITGB8			
	Others	ALDH1A1	NXF5		
Patient 2					
HCC 1	No. of mutated genes	39			
	Amino acid change (+)	20 (51.3%)			
	Functional loss[a] (+)	10 (25.6%)			
	KEGG pathway				
	Metabolic pathways	DBH			
	Others	AGRN			
HCC 2	No. of mutated genes	70			
	Amino acid change (+)	40 (57.1%)			
	Functional loss[a] (+)	20 (28.6%)			
	KEGG pathway				
	Metabolic pathways	ADSSL1	FTCD	RDH16	
	Others	ABCA13	BTRC	VWF	C4A
		GRM4			
Patient 3					
HCC 1	No. of mutated genes	30			
	Amino acid change (+)	20 (66.7%)			
	Functional loss[a] (+)	6 (20.0%)			
	KEGG pathway				
	Metabolic pathways	CYP1A2			
HCC 2	No. of mutated genes	276			
	Amino acid change (+)	208 (75.3%)			
	Functional loss[a] (+)	90 (32.6%)			
	KEGG pathway				
	Metabolic pathways	ACSM4	ADSS	UGT2B28	DHRS4L2
		GALNT5	ME1	POLE	NSDHL
		PIK3C2G			
	PI3K-Akt signaling pathway	COL6A6	HGF	ANGPT1	LPAR4
	Neuroactive ligand receptor	GLRA2			
	Others	CDK9	CA2	ABCC12	AP1M1
		GNPAT	GLYAT	RUVBL1	GDF9
		MYL12A	MLL3	SLC18A2	MAP4K4
		PRPF8	PIP4K2A	SLC9A4	NUP37
		VCP	TTN		

Supplementary Table 4. Continued

Patient 4					
HCC	No. of mutated genes	364			
	Amino acid change (+)	177 (48.6%)			
	Functional loss[a] (+)	46 (12.6%)			
	KEGG pathway				
	Metabolic pathways	ACSM5	ALPP	PNLIPRP1	
	MAPK signaling pathway	HSPA6	MAP2K2		
	PI3K-Akt signaling pathway	FLT4			
	Others	ECH1	CHI3L1	FURIN	CD99
		KCNJ12	ITGAE	TMPRSS4	REXO1L1
		RBMX	PLOD3	TUBA4A	PGA3

[a]The number of mutated genes predicted to be "damaging (deleterious)" by Sorting Intolerant From Tolerant (SIFT) functional impact predictions (http://provean.jcvi.org/index.php). The genes categorized in multiple pathways are shown in only one representative pathway.

Supplementary Table 5. List of 448 Indels in 409 Genes at a Frequency of >20% of Reads in 7 HCC Tumors From 4 Patients

Reference position	Gene	Chromosome	Coding sequence	Coverage	Allele change	Patient no.
7813482	ERRFI1	1	3	17	insA	3
12628605[a]	PRAMEF11	1	3	27	insC	2
12718307	PRAMEF7	1	2	47	insT	3
17358674[a]	PADI6	1	9	41	delG	4
17358674[a]	PADI6	1	9	62	delGT	2
26696284	ARID1A	1	2	19	delC	4
31395890[a]	SERINC2	1	9	17	insG	3
46770747	CYP4B1	1	8	42	delAT	4
46770748	CYP4B1	1	8	42	delT	4
52949277[a]	LOC100133211	1	1	42	delG	4
53189215	MAGOH	1	3	25	insA	3
54095320[a]	CDCP2	1	4	21	insC	4
62557547	ANGPTL3	1	4	17	insT	3
78876012	ELTD1	1	10	13	insA	3
89014597	GBP1	1	4	17	insA	2
90670304	BARHL2	1	2	20	insC	3
108974106	CLCC1	1	6	84	insG	4
122705624[a]	PDE4DIP	1	14	80	delG	4
122713730[a]	PDE4DIP	1	6	465	delT	1
131401399	DENND4B	1	12	43	insG	3
131474740	NUP210L	1	34	13	insG	3
131951749	SHE	1	3	112	insA	2
133072970	MSTO1	1	11	33	insA	3
133795656	CCT3	1	2	34	insT	3
133844355[a]	RHBG	1	9	40	delC	4
134043234[a]	TTC24	1	3	12	delC	3
136505109	IFI16	1	7	31	insT	3
146999551	F5	1	13	23	insT	3
151907797	GPR52	1	1	18	insA	2
162596811	C1orf25	1	10	26	insT	3
163815430	TPR	1	15	43	delT	3
174374155	CFHR4	1	5	18	insC	3
177507300	NR5A2	1	5	20	insT	3
201685738	NVL	1	6	32	insA	3
203543660	ACBD3	1	2	18	insG	3
208315870[a]	ARV1	1	3	146	delCT	1
208315871[a]	ARV1	1	3	144	delT	1
213162772	LYST	1	3	19	insG	3
214770350	RYR2	1	11	22	insG	3
219641707	PLD5	1	2	102	insA	1
224245114	AHCTF1	1	14	19	insG	3
230467928	RNASEH1	2	8	13	insT	3
236404886	ADAM17	2	19	29	insG	3
251086730[a]	LOC375190	2	8	40	insC	4
254371933	IFT172	2	38	18	insT	3
258505433[a]	SRD5A2	2	1	28	insG	2
267355164	SLC8A1	2	1	15	insG	3
282849120	EFEMP1	2	1	14	insA	3
287714203	PAPOLG	2	15	20	delT	3
288274645	USP34	2	15	14	insC	3
301408194	CCDC142	2	2	27	insC	3
302628924	C2orf3	2	3	22	insA	3
312276713	RETSAT	2	4	55	delC	2
314781292	RGPD2	2	5	27	insT	3
317672121[a]	LOC391405	2	4	43	delA	4
318709414	TRIM43	2	1	22	insA	3
325574912	SLC9A4	2	6	27	insT	3
329490991	RGPD3	2	20	29	insT	3
331565693	GCC2	2	22	18	insC	2
332910299	RGPD5	2	21	25	insA	3
333616906	RGPD7	2	8	21	delA	3

Supplementary Table 5. Continued

Reference position	Gene	Chromosome	Coding sequence	Coverage	Allele change	Patient no.
335725611	SLC20A1	2	8	14	insC	3
336700192	RABL2A	2	4	76	delG	1
340887331	DDX18	2	7	18	insT	3
350555029	IWS1	2	11	62	delG	3
355383031[a]	ZNF806	2	3	50	delC	4
355383457[a]	ZNF806	2	3	56	insA	4
355383669[a]	ZNF806	2	3	52	delA	4
361736116	NXPH2	2	2	48	insT	3
385464478	KCNH7	2	10	41	insT	3
387807820	COBLL1	2	2	23	insA	3
388452944	SCN2A	2	26	57	insT	3
400689016	TTC30A	2	1	32	insA	3
401650351	TTN	2	270	19	insG	3
401670059	TTN	2	242	25	insT	3
401800064	TTN	2	64	26	insA	3
402189048	SESTD1	2	14	31	insG	3
403038177	CWC22	2	11	16	insA	3
418958541	DNAH7	2	34	27	insC	3
439213578	XRCC5	2	13	22	delC	3
446013793	ACSL3	2	14	26	insT	3
446670417	SCG2	2	1	17	insA	3
446671148	SCG2	2	1	30	insC	3
456401277[a]	SAG	2	10	76	delA	2
463724350	AQP12B	2	1	26	delC	2
479793553[a]	GRIP2	3	10	55	insG	4
504388108	TTC21A	3	6	13	insA	3
509772719	ZNF852	3	3	20	delTC	4
509772720	ZNF852	3	3	19	delC	4
511646368	CCR5	3	1	24	insT	3
515483759[a]	SLC38A3	3	2	18	insG	4
538148646	GLT8D4	3	6	14	insC	3
538243406[a]	FLJ10213	3	1	12	insA	2
538564462	PDZRN3	3	10	24	insG	3
540846731[a]	LOC100288801	3	2	39	delG	2
540918687	ZNF717	3	4	18	delC	1
570208682	HHLA2	3	4	18	insT	3
570487430	DZIP3	3	10	45	insT	3
574780156	CD200R1	3	4	24	insA	3
587083495	ZNF148	3	6	14	insG	3
591946859[a]	LOC644974	3	6	36	delC	3
595459401	TOPBP1	3	26	16	insT	3
608303870	PLSCR2	3	4	59	insG	3
611616867	C3orf16	3	5	22	delCT	3
611616868	C3orf16	3	5	21	delT	3
612474548	SELT	3	4	38	insT	4
631967149	PHC3	3	10	13	insT	3
648156647	DGKG	3	2	44	insC	3
652237997[a]	CLDN16	3	1	271	delG	2
658673909	PAK2	3	12	16	insT	3
660166378	ZNF595	4	4	17	insA	1
662098266[a]	POLN	4	23	65	delG	3
696277331	FLJ16686	4	3	53	delC	1
725715810	TMPRSS11F	4	7	30	insA	3
728128121	LOC100129410	4	3	13	insC	2
752860649	UNC5C	4	14	17	insT	3
767688857[a]	EGF	4	24	16	insC	1
779934903	KIAA1109	4	39	17	insT	3
782346647	ANKRD50	4	3	37	insT	3
827268418	NEK1	4	4	17	insT	3
841124368	CDKN2AIP	4	3	16	insA	3
853638502	KIAA0947	5	14	24	insT	3

Supplementary Table 5. Continued

Reference position	Gene	Chromosome	Coding sequence	Coverage	Allele change	Patient no.
889026155[a]	CARD6	5	3	32	insT	2
891727995	PAIP1	5	2	16	insG	2
901348948	MAP3K1	5	13	24	insC	3
909928153	ADAMTS6	5	3	23	insG	3
914509435	SERF1B	5	3	15	insG	3
914509482	SERF1B	5	3	49	insA	3
915509411[a]	GTF2H2	5	13	31	insT	2
919186632	HEXB	5	11	43	insA	3
922917852	SCAMP1	5	7	37	insA	1
928534269	EDIL3	5	7	25	insA	3
931867232	CCNH	5	7	26	insT	3
956697390	EPB41L4A	5	11	13	insT	1
966610635	ZNF474	5	1	39	delT	1
972596345	SLC12A2	5	8	35	insT	3
980635083[a]	SMAD5	5	6	105	insC	1
985314450[a]	LOC100288105	5	1	14	delC	4
985640033[a]	PCDHB9	5	1	32	insA	1
985844899[a]	PCDHGA8	5	1	27	delC	3
992330379	SCGB3A2	5	1	14	delA	3
994446878[a]	TIGD6	5	1	136	delT	1
994476149	HMGXB3	5	6	14	delA	3
998157358	GRIA1	5	11	19	insC	3
1020539380	FAM153B	5	4	23	insC	3
1039337531	C6orf114	6	1	30	insA	3
1052252527	BTN2A2	6	2	44	insG	3
1054107191[a]	ZNF187	6	1	33	insG	4
1056096293[a]	FLJ45422	6	2	18	insT	2
1057247419[a]	MICA	6	5	27	delG	3
1082305754	DST	6	45	18	insT	3
1088830738	EYS	6	6	19	insT	3
1093406718	COL19A1	6	5	16	insA	3
1113248546	MDN1	6	15	14	insC	3
1113280524	MDN1	6	2	40	insA	3
1131602437[a]	FOXO3	6	2	64	insG	3
1133380782	SLC22A16	6	4	20	insA	3
1135037748	C6orf225	6	1	17	delC	3
1153093327	SAMD3	6	7	14	delC	3
1154647636	LOC643854	6	1	26	insT	3
1154648098	LOC643854	6	1	20	insC	3
1159216432	BCLAF1	6	2	13	delT	2
1161156444	PBOV1	6	1	36	insG	3
1182019086	RSPH3	6	6	43	insA	3
1200500350	RSPH10B2	7	19	23	insG	3
1206053594	VWDE	7	19	26	insA	4
1221518316	TAX1BP1	7	13	14	insA	3
1222659823[a]	KIAA0644	7	1	90	delC	4
1222659922[a]	KIAA0644	7	1	26	insC	3
1226974977	BBS9	7	7	19	insT	1
1228643735[a]	DPY19L1	7	18	22	delAT	4
1228643736[a]	DPY19L1	7	18	50	delT	1
1262731853	TYW1B	7	8	142	delA	4
1262954278	TRIM74	7	2	24	insA	2
1265555276	TRIM73	7	2	84	insT	2
1266437261[a]	FLJ37078	7	14	43	insC	2
1266593512[a]	ZP3	7	8	51	insG	1
1266763110[a]	POMZP3	7	5	83	delA	4
1278946055	C7orf62	7	1	20	insC	3
1283360469	HEPACAM2	7	4	29	insT	3
1283589759	CALCR	7	9	26	insT	3
1290893801[a]	ZAN	7	30	28	insG	3
1291366094	MOGAT3	7	2	24	insA	3