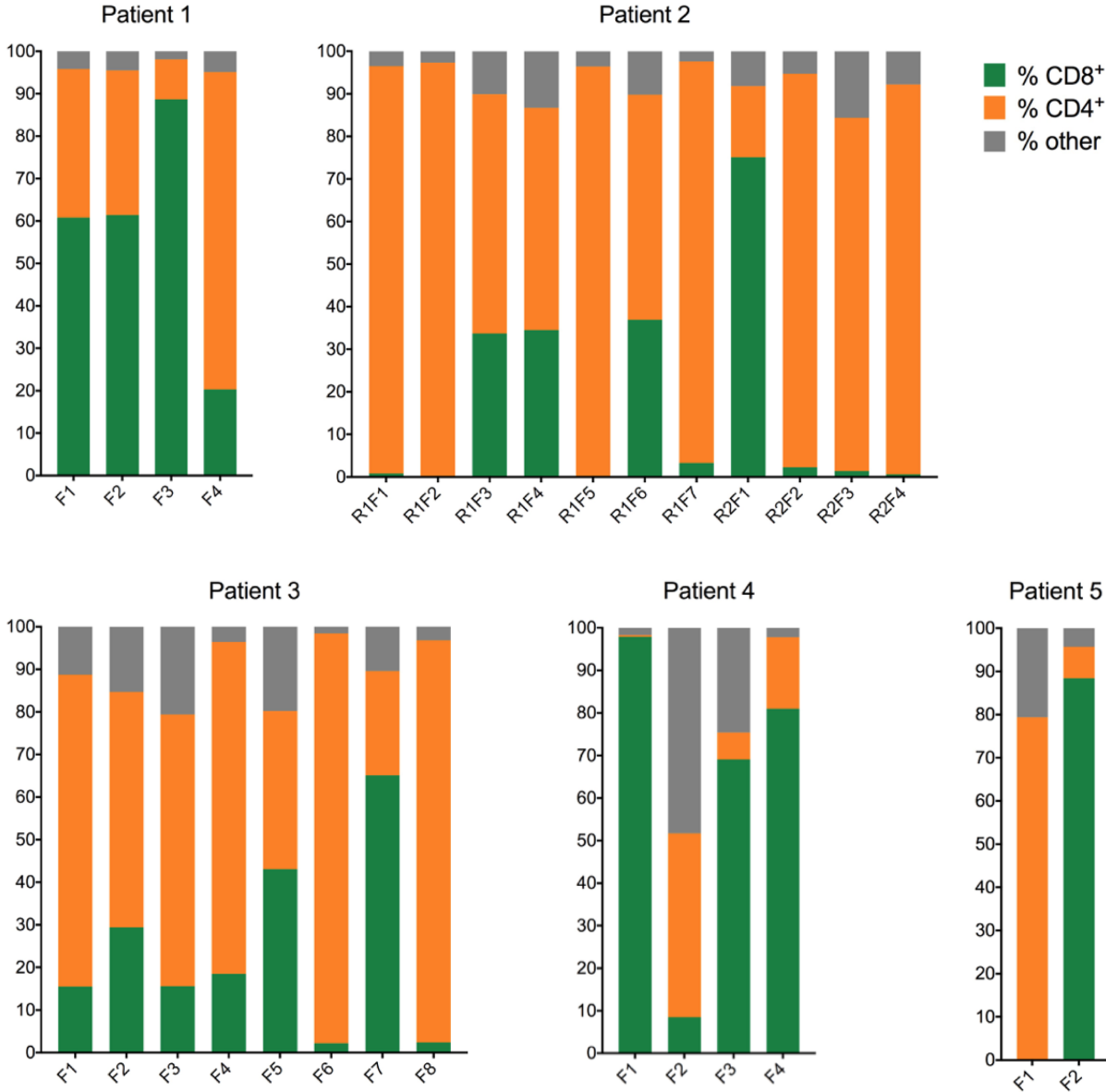
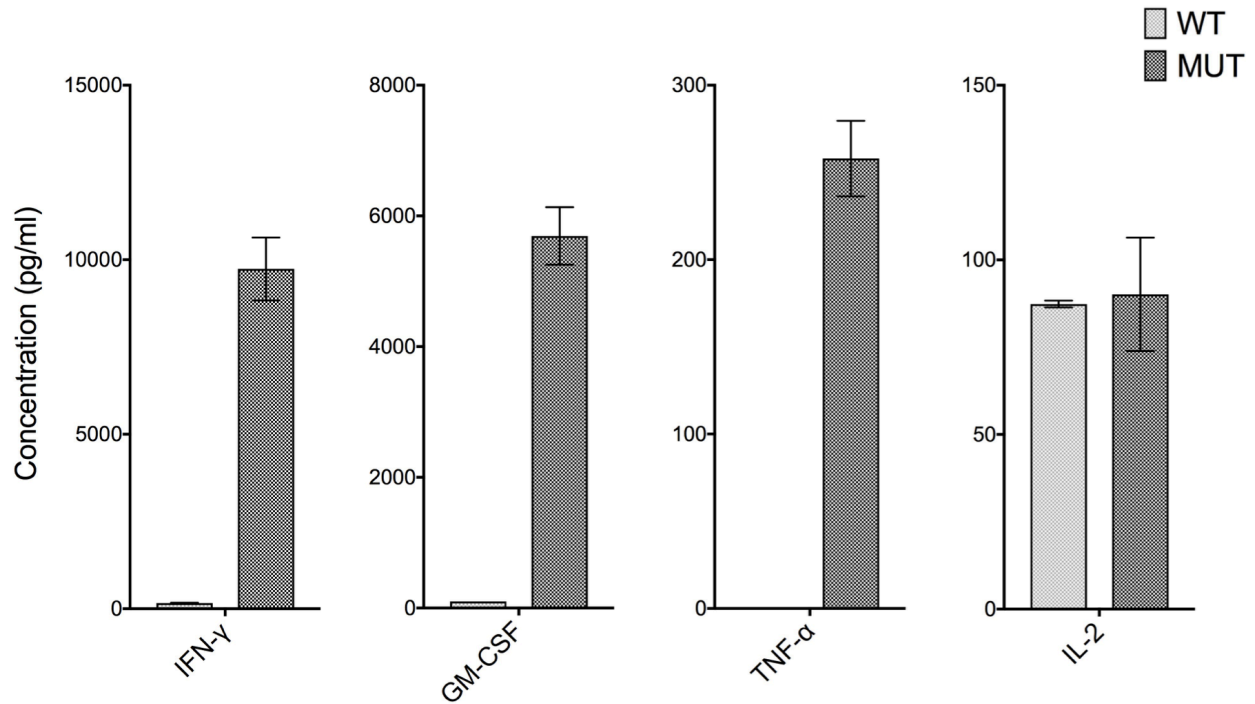


# Supplementary Figure 1



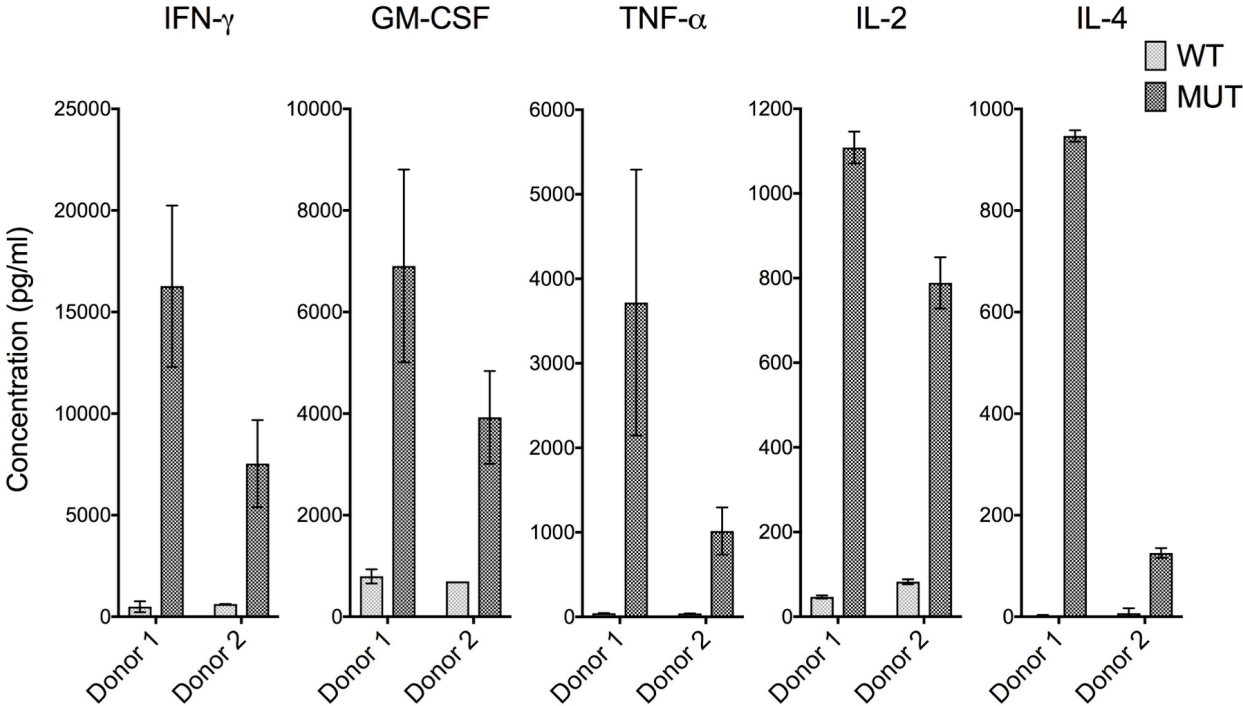
**Supplementary Figure 1.** The data was gated on live CD3<sup>+</sup> cells. Bar graphs represent the proportions of CD8<sup>+</sup> and CD4<sup>+</sup> cells (y axis) in each of the TIL cultures (x axis). The category “others” includes CD56<sup>+</sup> NKT cells and other uncharacterized cell types.

## Supplementary Figure 2



**Supplementary Figure 2.** F3 TILs were co-cultured with EBV-B cells pulsed with wild-type (WT) and mutant (MUT) CTBP1 25-mer peptide, followed by IFN- $\gamma$ , GM-CSF, TNF- $\alpha$ , IL-2 and IL-4 ELISA on the co-culture supernatants. IL-4 ELISA was negative (not depicted). Bars represent average reads from two duplicate wells; error bars represent SD.

### Supplementary Figure 3



**Supplementary Figure 3.** CTBP1<sup>Q277R</sup>-TCR transduced CD4<sup>+</sup> T cells from the two donors were co-cultured overnight with EBV-B cells stimulated with WT or MUT CTBP1 25-mer peptide, followed by IFN-γ, GM-CSF, TNF-α, IL-2 and IL-4 ELISA on the co-culture supernatants. Bars represent average reads from two duplicate wells; error bars represent SD.

**Supplementary Table 1.** Nonsynonymous mutations, including single-nucleotide variants (SNV) and frameshift insertions/deletions, were identified from the tumor specimens of five patients. Mutations are listed in the order in which they were incorporated into the tandem-minigenes (TMGs).

**Patient #1**

TMG	Gene name	Mutation type	cDNA change	Amino acid change	Wild type minigene sequence	Mutated minigene sequence
1	ACAN	nonsynonymous SNV	c.C4347A	p.S1449R	VLETTAPGVVEISGLPSGEVLETST	VLETTAPGVVEIRGLPSGEVLETST
1	ACSL3	nonsynonymous SNV	c.G866A	p.G289E	LPSDIAVIMYTSGSTGLPKGVMISH	LPSDIAVIMYTSESTGLPKGVMISH
1	ADAM33	nonsynonymous SNV	c.C2033T	p.P678L	NHNCHCAPGWAPPFCDKPGFGGSM	NHNCHCAPGWAPLFCDKPGFGGSM
1	ADAM33	nonsynonymous SNV	c.C304T	p.P102S	TITATVLQAGLHPSVTSQALVAAWT	TITATVLQAGLHSSVTSQALVAAWT
1	AK130486	nonsynonymous SNV	c.T116A	p.L39H	GRVARGRPGTPPLPHRPLPRGAGPG	GRVARGRPGTPPHRPLPRGAGPG
1	ANAPC5	nonsynonymous SNV	c.T161G	p.L54R	NEMSRTGEGAVSLMERRRLNQLLLP	NEMSRTGEGAVSRMERRRLNQLLLP
1	CCDC87	nonsynonymous SNV	c.G1546C	p.E516Q	SDHLHFDQGGLVPAADKDWSTFLS	SDHLHFDQGGLVQPAADKDWSTFLS
1	CDC42BPG	nonsynonymous SNV	c.T2779A	p.C927S	FCHTTCAPQAPPQVPPDLLRTALG	FCHTTCAPQAPPSPVPPDLLRTALG
1	EIF3B	nonsynonymous SNV	c.C350A	p.S117Y	GEAPGEQARDERSDSRAQAVSEADAG	GEAPGEQARDERYDSRAQAVSEADAG
1	ELF3	nonsynonymous SNV	c.G281A	p.R94Q	KNKYDASAI DFSRCMDGATLCNCA	KNKYDASAI DFSQCMDGATLCNCA
1	WIZ	nonsynonymous SNV	c.C496A	p.P166T	HLDLPAQPPPLAFRRVFPVEDTP	HLDLPAQPPPLATFRRVFPVEDTP
1	CDKN1A	nonframeshift deletion	c.107_109del	p.36_37del	CNHRDFFCSGAMSEPA GDV RQNPCG	CNHRDFFCSGAMPAGDVRQNPCGS
2	DNAH3	nonsynonymous SNV	c.C9780G	p.I3260M	MSKGNILEDETAIKVLSSSKVLSEE	MSKGNILEDETAMKVLSSSKVLSEE
2	CTBP1	nonsynonymous SNV	c.A830G	p.Q277R	ARGGLVDEKALAQALKEGRIRGAAL	ARGGLVDEKALARALKEGRIRGAAL
2	C1QTNF4	nonsynonymous SNV	c.A58G	p.T20A	LLGPAACWALGPTPGGSSELRSF	LLGPAACWALGPAPGGSSELRSF
2	FAM133A	nonsynonymous SNV	c.G199A	p.E67K	SKALAEFEEKMNENWKKELEKSREK	SKALAEFEEKMKNWKKELEKSREK
2	FAM134A	nonsynonymous SNV	c.G838A	p.E280K	APPGGDEPLAETESEAE LAGFSP	APPGGDEPLAETKSESEAE LAGFSP
2	FAM134A	nonsynonymous SNV	c.G850A	p.E284K	GDEPLAETESEAE LAGFSPVVDV	GDEPLAETESKAE LAGFSPVVDV
2	FAM194B	nonsynonymous SNV	c.T415C	p.Y139H	YLGKEEHLEEEYLGKEGYLEKEDY	YLGKEEHLEEEH LGKEGYLEKEDY
2	FAM47B	nonsynonymous SNV	c.A773T	p.H258L	HLRVDPPETGVSHLCPEPPKTLVSS	HLRVDPPETGVSLLCPEPPKTLVSS
2	FAM47C	nonsynonymous SNV	c.G2102T	p.R701L	HLRPEPPETGVSRLHPEPPKTRVSS	HLRPEPPETGVSLHPEPPKTRVSS
2	FAM65A	nonsynonymous SNV	c.G1439A	p.G480E	SVEAVGPESLAWGSPPTHPATHG	SVEAVGPESLAWEPSPTHPATHG

2	FAM83C	nonsynonymous SNV	c.C1369T	p.R457W	LLPRSRPLLQFHRGAPALSRFPENG	LLPRSRPLLQFHWGAPALSRFPENG
2	FGFR3	nonsynonymous SNV	c.A1118G	p.Y373C	EELVEADEAGSVYAGILSYGVGFLL	EELVEADEAGSVCAGILSYGVGFLL
3	FGL1	nonsynonymous SNV	c.C494A	p.T165N	EYWLGNKNLHFLTTQEDYTLKIDLA	EYWLGNKNLHFLNTQEDYTLKIDLA
3	FICD	nonsynonymous SNV	c.G1267A	p.E423K	VRPFIRFIAKCTETTLDTLLFATTE	VRPFIRFIAKCTKTTLDLTLFATTE
3	FOXD4	nonsynonymous SNV	c.C320A	p.S107Y	ASEDARQPAKPPSSYIALITMAILQ	ASEDARQPAKPPSYIALITMAILQ
3	GALR2	nonsynonymous SNV	c.C820T	p.R274C	GQFPLTRATYALRILSHLVSYANSC	GQFPLTRATYALCILSHLVSYANSC
3	GAPVD1	nonsynonymous SNV	c.G670A	p.E224K	EDHLETDPNKLIERFSPSQEKLFG	EDHLETDPNKLIKRFSPSQEKLFG
3	GLI1	nonsynonymous SNV	c.C3233G	p.S1078C	DQRGSSGHTPPSPGPPNMAVGNMSV	DQRGSSGHTPPPCGPPNMAVGNMSV
3	HIST1H1C	nonsynonymous SNV	c.G468C	p.K156N	GATPKKSAKKTPKKAKKPAATVTK	GATPKKSAKKTPNKAKKPAATVTK
3	HLA-DQB1	nonsynonymous SNV	c.G703A	p.V235I	QSESAQSKMLSGVGGFVLGLIFLGL	QSESAQSKMLSGIGGFVLGLIFLGL
3	HLA-DRB5	nonsynonymous SNV	c.G505A	p.A169T	EVRWFRNSQEEKAGVVSTGLIQNGD	EVRWFRNSQEEKGVVSTGLIQNGD
3	HRNR	nonsynonymous SNV	c.A3262T	p.S1088C	HGQHGSTSGQSSSCGQHGASSGQSS	HGQHGSTSGQSSCCGQHGASSGQSS
3	IGFN1	nonsynonymous SNV	c.A6127G	p.R2043G	EAGYRKDLGAPERIGSGSKAGFRDG	EAGYRKDLGAPEGIGSGSKAGFRDG
3	IGFN1	nonsynonymous SNV	c.A6132G	p.I2044M	AGYRKDLGAPERIGSGSKAGFRDGL	AGYRKDLGAPERMGSGSKAGFRDGL
4	IGSF9B	nonsynonymous SNV	c.C598T	p.R200W	VSDGSLTVTSVSREDRGAYTCRAYS	VSDGSLTVTSVSWEDRGAYTCRAYS
4	IVL	nonsynonymous SNV	c.A1428T	p.Q476H	EQEEKQLELPEQEQGVKHLEKQEA	EQEEKQLELPEQHEQGVKHLEKQEA
4	KANSL1	nonsynonymous SNV	c.G13T	p.D5Y	MFLADVPTSLHFQSMK	MFLAYVPTSLHFQSMK
4	KDM6A	nonsynonymous SNV	c.C3361G	p.H1121D	FVRVVSAGNLLSHVGHITLGMNTVQ	FVRVVSAGNLLSDVGHITLGMNTVQ
4	KDM6A	nonsynonymous SNV	c.G3535C	p.D1179H	FVVPEGYWGVLNDFCEKNNLNFLMG	FVVPEGYWGVLNHFCEKNNLNFLMG
4	KIAA1683	nonsynonymous SNV	c.C3199T	p.R1067C	PPSVVMLVGSSPRTCHTCGRTPTR	PPSVVMLVGSSPCTCHTCGRTPTR
4	KIF21B	nonsynonymous SNV	c.G1452C	p.E484D	EAIGALIQNYIREIEELRTKLESE	EAIGALIQNYIRDIEELRTKLESE
4	KMT2D	frameshift deletion	c.15430delC	p.Q5144fs	LCPMHKIKGPCEQELSSFAVFRVY	LCPMHKIKGPCEKS
4	KRTAP10-1	nonsynonymous SNV	c.G604T	p.V202F	KPVCKPICCVPCSGASTSCCQQS	KPVCKPICCVPCSGASTSCCQQS
4	LEMD2	nonsynonymous SNV	c.G601C	p.E201Q	RAGPAGAARARPEVGRRLERWLSRL	RAGPAGAARARPVGRRLERWLSRL
4	LHX9	nonsynonymous SNV	c.C1034T	p.P345L	GVDKADGTSLPAPPSADSGALTPPG	GVDKADGTSLPALPSADSGALTPPG
4	LOC100129195	nonsynonymous SNV	c.C534G	p.I178M	QPPLAAAAFIPELLLTINRNPEKS	QPPLAAAAFIPEMLLTINRNPEKS
5	MACF1	nonsynonymous SNV	c.G1240A	p.E414K	EEEWGKLIIEMLEREKSLRPAVERL	EEEWGKLIIEMLKREKSLRPAVERL
5	MELK	nonsynonymous SNV	c.G532C	p.E178Q	QTCCGSLAYAAPQLIQGKSYLGSEA	QTCCGSLAYAAPQLIQGKSYLGSEA

5	MGA	nonsynonymous SNV	c.A767G	p.K256R	ITQLKIDYNPFAKGRDDGLNNKPQ	ITQLKIDYNPFARGFRDDGLNNKPQ
5	MUC12	nonsynonymous SNV	c.T9325A	p.Y3109N	TTAGLSEKSTTFYSSPRSPDTTLSP	TTAGLSEKSTTFNSSPRSPDTTLSP
5	MUC21	nonsynonymous SNV	c.A289T	p.T97S	SGISTATNSEFSTVSSGISIATNSE	SGISTATNSEFSSVSSGISIATNSE
5	MUC22	nonsynonymous SNV	c.G3305T	p.G1102V	AGSETTTTTSTEGSETTTASTEGSE	AGSETTTTTSTEVSETTTASTEGSE
5	MYEF2	nonsynonymous SNV	c.G92A	p.R31Q	HLQPAEPPGEPREPHPAEAEKQQP	HLQPAEPPGEPREQEPHAEAEKQQP
5	MYOM2	nonsynonymous SNV	c.C2696T	p.S899L	VRAVNANGVGKPSDTSEPVLEARP	VRAVNANGVGKPLDTSEPVLEARP
5	NARR	nonsynonymous SNV	c.G172A	p.E58K	PPDGTTPRQLAAEESPRPRVIFGTP	PPDGTTPRQLAAKESPRPRVIFGTP
5	NAV2	nonsynonymous SNV	c.G5313T	p.K1771N	VGSNIESDSKKKKRKNWLRSSFQKA	VGSNIESDSKKKNRKNWLRSSFQKA
5	NAV2	nonsynonymous SNV	c.G5313T	p.K1771N	VGSNIESDSKKKKRKNWVNLRSSF	VGSNIESDSKKKNRKNWVNLRSSF
5	NBPF10	nonsynonymous SNV	c.A841G	p.S281G	NVSMVVSAGPLSSEKAEMNILEINE	NVSMVVSAGPLSGEKAEMNILEINE
6	NEK4	nonsynonymous SNV	c.G2110A	p.E704K	DGDYGEGKGQTNEINALVQLMTQTL	DGDYGEGKGQTNKINALVQLMTQTL
6	OGFRL1	nonsynonymous SNV	c.G578T	p.R193I	IEEFKKTKEAIRRFLLAYKMMLEFF	IEEFKKTKEAIRFLLAYKMMLEFF
6	OR10Z1	nonsynonymous SNV	c.C728T	p.S243L	AEGQKKAFASTCASHLTVVHHYGCA	AEGQKKAFASTCALHLTVVHHYGCA
6	PIK3CA	nonsynonymous SNV	c.G353A	p.G118D	GNREEKILNREIGFAIGMPVCEFDM	GNREEKILNREIDFAIGMPVCEFDM
6	PLCG2	nonsynonymous SNV	c.G2671T	p.D891Y	KQQGDPPEFATDRVEELFEWFQSI	KQQGDPPEFATYRVEELFEWFQSI
6	PRDM5	nonsynonymous SNV	c.C649T	p.H217Y	GKKFPVKQALQRHFEQHQETCRGDA	GKKFPVKQALQRYFEQHQETCRGDA
6	PRDM5	nonsynonymous SNV	c.C649T	p.H217Y	GKKFPVKQALQRHVLQCTAKSSLKE	GKKFPVKQALQRYVLQCTAKSSLKE
6	PRSS1	nonsynonymous SNV	c.G238A	p.E80K	LRLGKMDGRGGGEENLLAAADSPEQ	LRLGKMDGRGGKENLLAAADSPEQ
6	RAET1L	nonsynonymous SNV	c.A191G	p.Y64C	VQGQVDEKTFLLHYDCGNKTVTPVSP	VQGQVDEKTFLLHDCGNKTVTPVSP
6	RALB	nonsynonymous SNV	c.A128G	p.Y43C	TLQFMYDEFVEDYEPTKADSYRKKV	TLQFMYDEFVEDCEPTKADSYRKKV
6	RALB	nonsynonymous SNV	c.A191G	p.Y64C	KKVEGILEFVEDYEPTKADSYRKKV	KKVEGILEFVEDCEPTKADSYRKKV
6	RAPGEF5	nonsynonymous SNV	c.G1388A	p.R463Q	DLVSKSLEASGRIYVYRKDLADTL	DLVSKSLEASGQIYVYRKDLADTL
7	RBM6	nonsynonymous SNV	c.A224G	p.Y75C	PPFANVEEHSFSYGARDGPHGDYRG	PPFANVEEHSFSCGARDGPHGDYRG
7	RBMXL3	nonsynonymous SNV	c.C1864A	p.H622N	AYSGGHDSSSWSHRYGGGGRYEEYR	AYSGGHDSSSWSNRYGGGGRYEEYR
7	RBMXL3	nonsynonymous SNV	c.A2179T	p.T727S	SLDANSGGRSPDYSRGRDSSSQSD	SLDANSGGRSPDYSRGRDSSSQSD
7	RDH12	nonsynonymous SNV	c.C271G	p.Q91E	AASEIRVDTKNSQVLRKLDLSDTK	AASEIRVDTKNSEVLVRKLDLSDTK
7	RECQL4	nonsynonymous SNV	c.G1324A	p.E442K	PEPLVSPQPVPVPSLDPTVLPY	PEPLVSPQPVPVPSLDPTVLPY
7	RHOB	nonsynonymous SNV	c.T137C	p.I46T	YVPTVFENYVADIEVDGKQVELALW	YVPTVFENYVADTEVDGKQVELALW

7	RP2	nonsynonymous SNV	c.C12A	p.F4L	MGCFFSKRRKADKESR	MGCLFSKRRKADKESR
7	RRBP1	nonsynonymous SNV	c.T845C	p.V282A	KKVDTPNQGKKVEGAPTQGRKAEG	KKVDTPNQGKKAEGAPTQGRKAEG
7	SBK3	nonsynonymous SNV	c.C104G	p.P35R	QRLVELTTSRVTPVRSLRDQYHLIR	QRLVELTTSRVTRVRSRLRDQYHLIR
7	SCN1A	nonsynonymous SNV	c.C3636A	p.F1212L	RGKQWWNLRRCTCFRIVEHNWFETFI	RGKQWWNLRRTCLRIVEHNWFETFI
7	SEMA4G	nonsynonymous SNV	c.G212T	p.G71V	LLLEEASARLLVGARGALFSL SAND	LLLEEASARLLVVARGALFSL SAND
7	SH2B1	nonsynonymous SNV	c.G1921C	p.E641Q	QRQQGEQSR SAGEEVPVHPRSEAGS	QRQQGEQSR SAGQEVVHPRSEAGS
8	SLCO4A1	nonsynonymous SNV	c.C1380G	p.F460L	RLRGS AVIKFCLFCTVVSLLGILVF	RLRGS AVIKFCLLCTVVSLLGILVF
8	SPHK1	nonsynonymous SNV	c.A128C	p.E43A	GAPAA TAPGGEGEPHSR PCDARLGS	GAPAA TAPGGEGAPHSR PCDARLGS
8	SPTBN5	nonsynonymous SNV	c.G2515A	p.E839K	PGESLRNPGPWSEASCHPGPGDAWK	PGESLRNPGPWSKASCHPGPGDAWK
8	SYNE2	nonsynonymous SNV	c.T4163C	p.F1388S	HLDKCLKMLDMSFKDAERGD DTSCE	HLDKCLKMLDMSKDAERGD DTSCE
8	TAF4	nonsynonymous SNV	c.G2131T	p.A711S	SSSVQRTAGKTAATVTSALQPPVLS	SSSVQRTAGKTASTVTSALQPPVLS
8	TCF21	nonsynonymous SNV	c.G218T	p.S73I	KAPT KKSPLSGVSQEGKQVQRNAAN	KAPT KKSPLSGVIQEGKQVQRNAAN
8	TCRBV5S6 A3N2T	nonsynonymous SNV	c.G20T	p.C7F	MGPGLLCWALLC LLGAGSV	MGPGLLFWALLC LLGAGSV
8	TDRD9	nonsynonymous SNV	c.C388A	p.P130T	SVTCIPGTTYKY PDLPI SRYKEEVV	SVTCIPGTTYKY TDLPI SRYKEEVV
8	TENC1	nonsynonymous SNV	c.C4033T	p.R1345C	ITLTDNQRKLFRRHY PVNSITFSS	ITLTDNQRKLFRCRHY PVNSITFSS
8	TEX36	nonsynonymous SNV	c.C58G	p.H20D	NPPSDKDGRWFP HIGLTQKT PESIT	NPPSDKDGRWFP DIGLTQKT PESIT
8	THADA	nonsynonymous SNV	c.G5629A	p.E1877K	PEMLCHLQRMVSE QCHLLSQFFREL	PEMLCHLQRMVSKQCHLLSQFFREL
8	TMEM178B	nonsynonymous SNV	c.C703T	p.R235C	VAGINFELSRYPRYLYGLPDDISHG	VAGINFELSRYPCYLYGLPDDISHG
9	TRO	frameshift deletion	c.428_429del	p.K143fs	SVTAQPKKANKMKRV TAAAAQGSQS	SVTAQPKKANKMKS YCQGS PRLPIP NWP
9	TUBGCP3	nonframeshift deletion	c.1241_1255del	p.414_419del	TRDFPTRDFPTRDFP MHVFPTRVFP	TRDFPTRDFPTRVFPTRVFPTRVWH
9	UBE3A	nonsynonymous SNV	c.G977C	p.R326T	SKLPLAAQGKLIRLWSKY NADQIRR	SKLPLAAQGKLITLWSKY NADQIRR
9	UNK	nonsynonymous SNV	c.G1946T	p.R649L	AFLSGP GAAELARLRQELDEANSTI	AFLSGP GAAELALLRQELDEANSTI
9	USP47	nonsynonymous SNV	c.A1196G	p.Y399C	ISKSGLEKNSLIYELFSVMVHSGSA	ISKSGLEKNSLICELFSVMVHSGSA
9	VEGFB	nonsynonymous SNV	c.A425C	p.H142P	DSAVKPDRAATPHHRPQRSVPGWD	DSAVKPDRAATPPHRPQRSVPGWD
9	VWA5B2	nonsynonymous SNV	c.G268A	p.G90S	RRRSQAACCRALGPGLTPTPRRCA	RRRSQAACCRALSPGLTPTPRRCA
9	WNK3	nonsynonymous SNV	c.G985A	p.V329I	APEMYEEHYDESVDVYAFGMCMLEM	APEMYEEHYDESIDVYAFGMCMLEM
9	ZAN	nonsynonymous SNV	c.C2843A	p.P948H	ISTEKLTIPT EKPTISPEKLTIPTE	ISTEKLTIPT EKHTISPEKLTIPTE
9	ZBTB39	nonsynonymous SNV	c.G877A	p.E293K	EHSKDPGFGQMD ELQLEDLGDDDLQ	EHSKDPGFGQMD KLQLEDLGDDDLQ

9	ZFP36L2	nonsynonymous SNV	c.G745A	p.E249K	GTRDALHLGFPREPRPKLHHSLSFS	GTRDALHLGFPKPRPKLHHSLSFS
9	ZNF14	nonsynonymous SNV	c.C340T	p.H114Y	PHECSFCGRDFIHHSSLNRHMRSHT	PHECSFCGRDFIYHSSLNRHMRSHT
10	ZNF197	nonsynonymous SNV	c.C1301A	p.A434E	GEKPYKCNECGKAFSQAAYLLNHQR	GEKPYKCNECGKEFSQAAYLLNHQR
10	ZNF208	nonsynonymous SNV	c.G1474T	p.A492S	PYKCEECDKATHAGEKPYKCEECGK	PYKCEECDKATHSGEKPYKCEECGK
10	ZNF430	nonsynonymous SNV	c.T1233A	p.N411K	KFYKCEECGKGFNWSSTLTKHKRIH	KFYKCEECGKGFKWSSTLTKHKRIH
10	ZNF430	nonsynonymous SNV	c.T1234A	p.W412R	FYKCEECGKGFNWSSTLTKHKRIHT	FYKCEECGKGFNRSSTLTKHKRIHT
10	ZNF460	nonsynonymous SNV	c.G248T	p.G83V	PEEVSLQEQLAQVPRYSYLGQAMD	PEEVSLQEQLAQVPRYSYLGQAMD
10	ZNF548	nonsynonymous SNV	c.G1141T	p.V381F	VCGELFRYNSSLVKHWRNHTGERPY	VCGELFRYNSSLFKHWRNHTGERPY
10	ZNF606	nonsynonymous SNV	c.C310G	p.Q104E	LETYGHLLSVGNQIAKPEVISLLEQ	LETYGHLLSVGNEIAKPEVISLLEQ
10	ZNF626	nonsynonymous SNV	c.A764G	p.K255R	TLTTHKRNHTGEKPYKCDKCGKAFM	TLTTHKRNHTGERPYKCDKCGKAFM
10	ZNF732	nonsynonymous SNV	c.A1332T	p.K444N	DLNKHKIIHTGEKPYKCEECGKAFG	DLNKHKIIHTGENPYKCEECGKAFG
10	ZNF853	nonsynonymous SNV	c.G585T	p.Q195H	QEQQVLQQEQLQQVQEQQLLQQQ	QEQQVLQQEQLHQVQEQQLLQQQ
10	ZNF91	nonsynonymous SNV	c.T1321A	p.W441R	PFKCKEKGKAFIWSSTLTRHKRIHT	PFKCKEKGKAFIRSSTLTRHKRIHT
10	ZNFX1	nonsynonymous SNV	c.C5527G	p.R1843G	ERVQIVSAIGYPRGHWFKCRNGHIY	ERVQIVSAIGYPGGHWFKCRNGHIY
11	LNP1	frameshift insertion	c.207_208insT CTCATGAGG ACCAGGAAT TCCGATGCC G	p.H69fs	PRIPSSDCHPRRSHEDQEFRCRSH	PRIPSSDCHPRRSHEDQEFRCRLMRTKNFDVATYGITENT QRMGHSRSHWNQKEDPIPKLRNFQSPLNGNCALEPSVLPW DLKAERREMKENA
11	ZFP36L1	frameshift insertion	c.453dupC	p.K152fs	GIHELRSLTRHPKYKTELCRTFHTI	GIHELRSLTRHPQVQDGAVPHLPHHRLPLRAPLPLHPQR
11	ZNF41	frameshift insertion	c.1335dupG	p.K446fs	TGEKPYVCADCGKAFIQKSHFNTHQ	TGEKPYVCADCGELHPEITFQHTSENSYWRKAV



## Patient #2

TMG	Gene name	Mutation type	cDNA change	Amino acid change	Wild type minigene sequence	Mutated minigene sequence
1	AHNAK2	nonsynonymous SNV	c.A11876C	p.K3959T	GARLEGDLSLADKDMTAKDSKFKMP	GARLEGDLSLADTDMTAKDSKFKMP
1	AHNAK2	nonsynonymous SNV	c.G2557A	p.E853K	PSFGVSAPGKSMEDSVDSAPKVEA	PSFGVSAPGKSMKDSVDVSAPKVEA
1	AK293147	nonsynonymous SNV	c.T116C	p.L39P	SLAWLNRVPPTPLSPPPPTGPASS	SLAWLNRVPPTPLSPPPPTGPASS
1	AK293147	nonsynonymous SNV	c.T20C	p.L7P	MCPPQPLSPYQSADRPSVPLPPWS	MCPPQPPSPYQSADRPSV
1	AKNA	nonsynonymous SNV	c.T1666G	p.W556G	AGWGRKPWDRGWGGGEAWGGGWLQ	AGWGRKPWDRGWGGGEAWGGGWLQ
1	AQP10	nonsynonymous SNV	c.T107C	p.L36P	CLAEFLGVFVLMMLLTQGAVAQAVTS	CLAEFLGVFVLMPLTQGAVAQAVTS
1	ATP2A3	nonsynonymous SNV	c.A1166T	p.Y389F	CLLHEFTISGTTYTPEGEVRQGDQP	CLLHEFTISGTTFTPEGEVRQGDQP
1	BAGE	nonsynonymous SNV	c.C26A	p.A9E	MAARAVFLALSAQLLQARLMKEESP	MAARAVFLELSAQLLQARLMK
1	BAGE3	nonsynonymous SNV	c.C26A	p.A9E	MAAGAVFLALSAQLLQARLMKEESP	MAAGAVFLELSAQLLQARLMK
1	BAGE3	nonsynonymous SNV	c.C26A	p.A9E	MAAGVVFLALSAQLLQARLMKEESP	MAAGVVFLELSAQLLQARLMK
1	C11orf95	nonsynonymous SNV	c.C88A	p.L30M	PGPAVASARGRRLPPAGSSGSAEPE	PGPAVASARGRRMPPAGSSGSAEPE
1	CCNK	nonsynonymous SNV	c.A1217C	p.H406P	TDLPKVQIPPAHPAPVHQPPPLPH	TDLPKVQIPPPAPPAPVHQPPPLPH
2	CDC27	nonsynonymous SNV	c.A1276G	p.I426V	ALCSYNCKEAINILSHLPSHHYNTG	ALCSYNCKEAINVLSHLPSHHYNTG
2	CDH19	nonsynonymous SNV	c.C1189A	p.P397T	QGSFVGVVSATDPDNRKSPIRYSIT	QGSFVGVVSATDNDNRKSPIRYSIT
2	CELSR3	nonsynonymous SNV	c.G5486A	p.G1829D	LDRGLLSVTVTRSGRASHLLLDQV	LDRGLLSVTVTRDSGRASHLLLDQV
2	CFD	nonsynonymous SNV	c.G83T	p.G28V	AACAAPRGRILGGREAEAHARPYM	AACAAPRGRILVGREAEAHARPYM
2	CHORDC1	nonsynonymous SNV	c.G313T	p.A105S	DEPMTNLELKISASLKQALDKLKLS	DEPMTNLELKISSSLKQALDKLKLS
2	CNTN4	nonsynonymous SNV	c.T2813G	p.V938G	ALDNESEVKGYKVLRYRWNRSSTSV	ALDNESEVKGYKGLRYRWNRSSTSV
2	CNTNAP3B	nonsynonymous SNV	c.T3517C	p.F1173L	TSGFTGCLSAVRFGRAAPLKAALRP	TSGFTGCLSAVRLGRAAPLKAALRP
2	CNTNAP3B	nonsynonymous SNV	c.T3517C	p.F1173L	TSGFTGCLSAVRFCAAPLKAALRP	TSGFTGCLSAVRLGCAAPLKAALRP
2	CNTROB	nonsynonymous SNV	c.A289C	p.T97P	FQGLRDALDSEHTRRKHCERHIQSL	FQGLRDALDSEHPRRKHCERHIQSL
2	CNTROB	nonsynonymous SNV	c.C1424A	p.P475Q	WDEANQLLSTTLPPNPPAPPAGPS	WDEANQLLSTTLQPPNPPAPPAGPS
2	COL4A5	nonsynonymous SNV	c.G1228C	p.D410H	PPGFPPGERGQKGDGEPGGSIPGPP	PPGFPPGERGQKGHEGPPGISIPGPP
2	COL9A2	nonsynonymous SNV	c.G296T	p.G99V	GPRGPPGRAGEKGDGEGSPGIRGPQG	GPRGPPGRAGEKVDGEGSPGIRGPQG
3	CTBP1	nonsynonymous SNV	c.G640T	p.A214S	FYDPYLSDGVERALGLQRVSTLQDL	FYDPYLSDGVERSLGLQRVSTLQDL
3	CTNS	nonsynonymous SNV	c.T404A	p.L135H	QRVSWPAIGFLVLAWLFAFVTMIVA	QRVSWPAIGFLVHAWLFAFVTMIVA

3	CYTH2	nonsynonymous SNV	c.A394C	p.T132P	GHHSAPVGPLPPTPQTQLLPSHWQL	GHHSAPVGPLPPPQTQLLPSHWQL
3	DAZL	nonsynonymous SNV	c.G10A	p.A4T	MSTANPETPNSTISREASTQSSSAA	MSTTNPETPNSTISRE
3	DAZL	nonsynonymous SNV	c.G70A	p.A24T	ARLTPLPHESTANPETPNSTISRE	ARLTPLPHESTTNPETPNSTISRE
3	DHRS4L2	nonsynonymous SNV	c.A373T	p.S125C	APRPGASHPATPSIQTLRQPLHLSP	APRPGASHPATPCIQTLRQPLHLSP
3	DLEU7	nonsynonymous SNV	c.C169A	p.P57T	HVSTAPARRSGPPRARPGGREERG	HVSTAPARRSGPTRARPGGREERG
3	DNM1P41	nonsynonymous SNV	c.C484T	p.R162C	ICVVTLPTHVLQRAEHLHAPDPCGT	ICVVTLPTHVLQCAEHLHAPDPCGT
3	DTYMK	nonsynonymous SNV	c.G495T	p.E165D	MVDASKSIEAVHEDIRVLSAIRT	MVDASKSIEAVHDDIRVLSAIRT
3	DUX2	nonsynonymous SNV	c.G599C	p.G200A	TGLPAPHVPCAPGALPQGAFVSQAA	TGLPAPHVPCAPAALPQGAFVSQAA
3	DUX4L4	nonsynonymous SNV	c.C997A	p.P333T	AAWEPQAGEAPPPQPAPPDASARQG	AAWEPQAGEAPPTQPAPPDASARQG
3	DYRK1B	nonsynonymous SNV	c.A1703C	p.D568A	MTGGRPPLPPDDPATLGPLHLGRG	MTGGRPPLPPDAPATLGPLHLGRG
4	EML6	nonsynonymous SNV	c.T134G	p.V45G	GKEVVYFVAGVGVVYNTREHSQKFF	GKEVVYFVAGVGGVYNTREHSQKFF
4	SREBF1	nonsynonymous SNV	c.C2A	p.A1D	AVQLFLCDLLLVRVTRSLWRQQPPA	DVQLFLCDLLLVV
4	DAZL	nonsynonymous SNV	c.G124A	p.A42T	MSSSLLLLPKSTANPETPNSTISRE	MSSSLLLLPKSTTNPETPNSTISRE
4	FAM118A	nonsynonymous SNV	c.T593C	p.V198A	WGVLCPGSCSHFAVNY	WGVLCPGSCSHFAAVNY
4	OLFM2	nonsynonymous SNV	c.T50C	p.M17T	LLKIGAVLSTMAMVTNWMSQTLPSL	LLKIGAVLSTMATVTNWMSQTLPSL
4	SUB1	nonsynonymous SNV	c.G167A	p.S56N	ETSRALSSSKQSSSRDDNMFQELF	ETSRALSSSKQSNSSRDDNMFQELF
4	SUB1	nonsynonymous SNV	c.G167A	p.S56N	ETSRALSSSKQSSSRDDNMFQV	ETSRALSSSKQSNSSRDDNMFQV
4	TCEAL4	nonsynonymous SNV	c.T268G	p.W90G	KNGYGRGKGWGWGRVESREAGPSV	KNGYGRGKGWGGGRVESREAGPSV
4	CCNG2	nonsynonymous SNV	c.T955C	p.F319L	AQAGVQWPDLSFQPPTRFKRFSC	AQAGVQWPDLSLQPPTRFKRFSC
4	UQCRC2	nonsynonymous SNV	c.G23A	p.G8D	MKLLTRAGSFSFTKLPNGLVIASLE	MKLLTRADSFSFTKLPNGLV
4	ARHGDI	nonsynonymous SNV	c.T575C	p.L192P	RTTDRRMCPQPLPSPYQSADRPSV	RTTDRRMCPQPPSPYQSADRPSV
4	MGST3	nonsynonymous SNV	c.T371C	p.I124T	RLASLGHTGRRRIVLGH	RLASLGHTGRRRTVLGH
5	SAMD8	nonsynonymous SNV	c.C95A	p.A32E	PPPPLEALHSGEAGRAPDSDGGSDA	PPPPLEALHSGEAGRAPDSDGGSDA
5	ZNF6	nonsynonymous SNV	c.A902T	p.K301I	NLTTHKKIHTGEKPYKCEECGKAFK	NLTTHKKIHTGEIPYKCEECGKAFK
5	UBQLN4	nonsynonymous SNV	c.A508C	p.T170P	QRRVYARGPHFLTSHASHIFSNRGF	QRRVYARGPHFLPSHASHIFSNRGF
5	TMEM184A	nonsynonymous SNV	c.A383T	p.E128V	YYVYFDSVRDCYEAFVIYSFLSLC	YYVYFDSVRDCYVAFVIYSFLSLC
5	TMEM184A	nonsynonymous SNV	c.A383T	p.E128V	YYVYFDSVRDCYEAS	YYVYFDSVRDCYVAS
5	CNTROB	nonsynonymous SNV	c.A583C	p.T195P	FQGLRDALDSEHTRRK	FQGLRDALDSEHPRRK

5	GOLGA6L2	nonsynonymous SNV	c.G2605C	p.A869P	GEDVGAGGEDVGAGGDAREGGEDTR	GEDVGAGGEDVPGGDAREGGEDTR
5	GOLGA6L2	nonsynonymous SNV	c.G820C	p.A274P	RQEKMQREQEDVGAGGDAREGGEDTR	RQEKMQREQEDVPGGDAREGGEDTR
5	GOLGA6L2	nonsynonymous SNV	c.C2542G	p.P848A	GEDAGAGGEDAGPGGEDVPGGEDV	GEDAGAGGEDAGAGGEDVPGGEDV
5	DPP7	nonsynonymous SNV	c.C545T	p.P182L	AQLGCAATLAAFPSESTRTWPRVG	AQLGCAATLAAFLSESTRTWPRVG
5	ALDH1A3	nonsynonymous SNV	c.T116C	p.L39P	LEVKFTKVRRAPLPPDGRGPLRWAA	LEVKFTKVRRAPPPDGRGPLRWAA
5	CFD	nonsynonymous SNV	c.G104T	p.G35V	WAWAAPPRGRILGGREAEAHARPYM	WAWAAPPRGRILVGREAEAHARPYM
6	IGHV3-13	nonsynonymous SNV	c.G49A	p.V17I	LSWVFLVAILEGVQCEVQLVESGGG	LSWVFLVAILEGIQCEVQLVESGGG
6	IGHV3-38	nonsynonymous SNV	c.G70T	p.V24L	GILKGVQCEVQLVESGGGLVQPRGS	GILKGVQCEVQLLESGGGLVQPRGS
6	ZNF99	nonsynonymous SNV	c.C2862A	p.S954R	KPYKCEERGKAFSHFSRLTKHRIIH	KPYKCEERGKAFRHSRLTKHRIIH
6	NBPF1	nonsynonymous SNV	c.C3113T	p.T1038M	KGKGGKRRRGRSTKRRRRRGRKEGE	KGKGGKRRRGRSMKRRRRRGRKEGE
6	NBPF1	nonsynonymous SNV	c.G3030T	p.E1010D	SCLPYGSSFYALEEKHVGFSLDVGE	SCLPYGSSFYALDEKHVGFSLDVGE
6	NBPF1	nonsynonymous SNV	c.A2936T	p.Q979L	RELLEAVEPEVLQDSLDRCYSTPSS	RELLEAVEPEVLLDSLDRCYSTPSS
6	HLA-DQB2	nonsynonymous SNV	c.G748A	p.G250S	IFLGLGLIIRHRGQKGRGPPPPAGL	IFLGLGLIIRHRSQKGRGPPPPAGL
6	DUX4L4	nonsynonymous SNV	c.C814A	p.P272T	PPHPGKSREDRDPQRDGLPGCAVA	PPHPGKSREDRDTQRDGLPGCAVA
6	DUX4L4	nonsynonymous SNV	c.G1032T	p.Q344H	PPQPAPPDASARQGQMGPAPSQA	PPQPAPPDASARHGQMGPAPSQA
6	AC026202.3	nonsynonymous SNV	c.G541T	p.A181S	AAGIILKRGGLPADHRTPRSPRSPA	AAGIILKRGGLPDHRTPRSPRSPA
6	MUC-B1	nonsynonymous SNV	c.G206A	p.G69D	RKPQQLQRRPAGGTGQRRGSGCSPS	RKPQQLQRRPAGDTGQRRGSGCSPS
6	MUC-B1	nonsynonymous SNV	c.G404C	p.R135T	QRRGSAAPVPTSRGTGQRRGSAAPA	QRRGSAAPVPTSTGTGQRRGSAAPA
7	EVX2	nonsynonymous SNV	c.G1145T	p.G382V	ASSAAAAGAPPSGGSAPCSCLSCHS	ASSAAAAGAPPSVGSAPCSCLSCHS
7	FBXL19-AS1	nonsynonymous SNV	c.A88C	p.T30P	SLSRSPLSTTLGTSPACGLEIPPTS	SLSRSPLSTTLGPSPACGLEIPPTS
7	FNTA	nonsynonymous SNV	c.A63C	p.Q21H	EAAQGGEPGQPAQPPQPHPPPPQQ	EAAQGGEPGQPAHPPQPHPPPPQQ
7	FRG1B	nonsynonymous SNV	c.A163G	p.N55D	VFQNGKMALLASNSCFIRCNEAGDI	VFQNGKMALLASDSCFIRCNEAGDI
7	FRG1B	nonsynonymous SNV	c.A19G	p.N7D	MALLASNSCFIRCNEAGDIEAKSKT	MALLASDSCFIRCNEAGDI
7	FRG2B	nonsynonymous SNV	c.C463T	p.R155C	AHHRGRSRACTGRSKRHRSRALGVQ	AHHRGRSRACTGCSKRHRSRALGVQ
7	FRMPD4	nonsynonymous SNV	c.C2629A	p.Q877K	LGALEALSVSEEQQTSDNSGVAILR	LGALEALSVSEEKQTSNSGVAILR
7	FTH1	nonsynonymous SNV	c.G46T	p.D16Y	ASTSQVRQNYHQDSEAAINRQINLE	ASTSQVRQNYHQYSEAAINRQINLE
7	GAREML	nonsynonymous SNV	c.C1099T	p.P367S	LWAHQGPEGLVRPPGLDLISFGAA	LWAHQGPEGLVRSPGLDLISFGAA
7	GATA1	nonsynonymous SNV	c.C919A	p.R307S	LTMRKDGQTRNRKASGKGKKKRGGS	LTMRKDGQTRNSKASGKGKKKRGGS

7	GDPD2	nonsynonymous SNV	c.G160A	p.A54T	KVLLLLIMLLVAAGLVGLDIQWQQE	KVLLLLIMLLVATGLVGLDIQWQQE
7	GID4	nonsynonymous SNV	c.A137C	p.H46P	RRQRAGGRPSRPHPARARPGLSLPA	RRQRAGGRPSRPPPARARPGLSLPA
8	GOLGA6L2	nonsynonymous SNV	c.C401A	p.A134D	GTPSSFNLALSQAFRGSPLGCVSTS	GTPSSFNLALSQDFRGSPLGCVSTS
8	GOLGA8K	nonsynonymous SNV	c.G1702A	p.A568T	GPGAPAPQELGAADKHGDLREVSLT	GPGAPAPQELGATDKHGDLESLT
8	GOLGA8K	nonsynonymous SNV	c.A1319G	p.Q440R	GEHLDSEGEAAPQPMPSVPEDLESR	GEHLDSEGEAAPRPMPSVPEDLESR
8	HCFC1	nonsynonymous SNV	c.A1762C	p.T588P	IVKTMAVTPGTTTLPATVKVASSPV	IVKTMAVTPGTTPLPATVKVASSPV
8	HEBP2	nonsynonymous SNV	c.G31A	p.A11T	MAEPLQPDPGAEDAAAQAVETPGW	MAEPLQPDPGTAEDAAAQAVETP
8	HLA-DQB2	nonsynonymous SNV	c.G748A	p.G250S	IFLGLGLIIRHRGQKGRGPPPPAGN	IFLGLGLIIRHRSQKGRGPPPPAGN
8	HLA-DRB5	nonsynonymous SNV	c.C165A	p.F55L	ECHFFNGTERVRFLLHRDIYNQEDL	ECHFFNGTERVRLLRDIYNQEDL
8	HS6ST1	nonsynonymous SNV	c.C261A	p.D87E	LERSLRFDKMGDDVIVFLHIQKTGG	LERSLRFDKMGDEVIVFLHIQKTGG
8	HYDIN	nonsynonymous SNV	c.C14623A	p.L4875M	TFSTECRMPDIALPSQFVVPANSEG	TFSTECRMPDIAMPSQFVVPANSEG
8	INSR	nonsynonymous SNV	c.C37T	p.P13S	MATGGRRGAAAAPLLVAVAALLLGA	MATGGRRGAAAASLLVAVAALLLGA
8	KANK3	nonsynonymous SNV	c.T1811C	p.V604A	SQAEPVARMLEGVRRLGPELLAHVV	SQAEPVARMLEGARRLGPELLAHVV
8	KRT18	nonsynonymous SNV	c.G252A	p.M84I	AGMGGIQNEKETMQSLNDRLASLYD	AGMGGIQNEKETIQSLNDRLASLYD
9	KRTAP4-8	nonsynonymous SNV	c.T283A	p.C95S	VCCQPTCCHPSCCISCCRPSCCVS	VCCQPTCCHPSCSISCCRPSCCVS
9	KRTAP9-8	nonsynonymous SNV	c.T179C	p.I60T	RPTCCQNTCCQPICVTSCCQPSCCS	RPTCCQNTCCQPTCVTSCCQPSCCS
9	LILRB1	nonsynonymous SNV	c.G955A	p.A319T	WSAPSDPLDILIAGQFYDRVLSVQ	WSAPSDPLDILITGQFYDRVLSVQ
9	LINC00273	nonsynonymous SNV	c.G595C	p.G199R	LVSRSRGREDTRAGRGAQTGKARDRK	LVSRSRGREDTRARRGAQTGKARDRK
9	LINC00937	nonsynonymous SNV	c.T443C	p.V148A	APGLRGRAGRAAVGQEGDRHPDRWR	APGLRGRAGRAAAGQEGDRHPDRWR
9	MCCC2	nonsynonymous SNV	c.T115C	p.S39P	SVASLGTQPDLSALYQENYKQMKKA	SVASLGTQPDLPALYQENYKQMKKA
9	METTL9	nonsynonymous SNV	c.G17A	p.G6D	MRLLAGWLCLSLASVWLARRMWTLR	MRLRADWLCLSLASVWLA
9	MROH5	nonsynonymous SNV	c.T608G	p.V203G	NMLVAICRHSWRVVAQHLETLLTG	NMLVAICRHSWRGVAQHLETLLTG
9	MUC4 (1)	nonsynonymous SNV	c.A12607T	p.T4203S	TPLPVTDTSSASTGHATPLPVTDT	TPLPVTDTSSASSGHATPLPVTDT
9	MUC4 (2)	nonsynonymous SNV	c.T10634C	p.V3545A	HATPLPVTSLSSVSTGDTTPLPVT	HATPLPVTSLSSASTGDTTPLPVT
9	MUC4 (3)	nonsynonymous SNV	c.T8810C	p.V2937A	QATPLPVTSLSSVSTGDTTPLPVT	QATPLPVTSLSSASTGDTTPLPVT
9	MUC4 (4)	nonsynonymous SNV	c.T8572C	p.S2858P	TTPLPVTDASSVSTGHATSLPVTDA	TTPLPVTDASSVPTGHATSLPVTDA
10	MUC4	nonsynonymous SNV	c.C6554T	p.A2185V	HATSLPVTDTSSASTGHATPLPVT	HATSLPVTDTSSVSTGHATPLPVT
10	MUC4	nonsynonymous SNV	c.C6506T	p.A2169V	HATSLPVTDTSSASTGHATSLPVT	HATSLPVTDTSSVSTGHATSLPVT

10	MUC4	nonsynonymous SNV	c.C5056A	p.L1686I	STGHATPLPVTGLSSATDDTTRLP	STGHATPLPVTGISSATDDTTRLP
10	MYH1	nonsynonymous SNV	c.G3718C	p.E1240Q	MKMEIDDLASNMETVSKAKGNLEKM	MKMEIDDLASNMQTVSKAKGNLEKM
10	NBPF1	nonsynonymous SNV	c.G3030T	p.E1010D	LGQPYRSAYVYSLEEYQLGLALDVDR	LGQPYRSAYVYSLDEYQLGLALDVDR
10	NBPF1	nonsynonymous SNV	c.A2936T	p.Q979L	RELLDEKEPEVLQDSLDRCYSTPSG	RELLDEKEPEVLLDSLDRCYSTPSG
10	NBPF14	nonsynonymous SNV	c.A323G	p.Q108R	DEDVQVEEAQKVKSSAPREVQKTE	DEDVQVEEAQKVRKSSAPREVQKTE
10	NBPF8	nonsynonymous SNV	c.A548G	p.Q183R	DEDVQVEEAEKVKSSAPREVQKAE	DEDVQVEEAEKVRKSSAPREVQKAE
10	NCOA4	nonsynonymous SNV	c.G709A	p.V237M	VQNHQDPCKVEEVCANRANPCTSAE	VQNHQDPCKVEEMCRANRANPCTSAE
10	NEK2	nonsynonymous SNV	c.A37G	p.T13A	MPSRAEDYEVLYTIGTGSYGRQKI	MPSRAEDYEVLYAIGTGSYGRQKI
10	NHS	nonsynonymous SNV	c.C155T	p.P52L	PPGRRDLDEVEAPGPEEPARAVPAP	PPGRRDLDEVEALGPEEPARAVPAP
10	OR4F15	nonsynonymous SNV	c.C919T	p.H307Y	KVAMRRLCSRLAHFTKIL	KVAMRRLCSRLAYFTKIL
11	OR4K15	nonsynonymous SNV	c.G682T	p.G228C	TYVVSLIVADSGFLSLSSFLLVV	TYVVSLIVADSCFLSLSSFLLVV
11	PAN3-AS1	nonsynonymous SNV	c.A298G	p.T100A	SSSAAARGAAAATTIDGGGKEDAAG	SSSAAARGAAAAATTIDGGGKEDAAG
11	PCDHB7	nonsynonymous SNV	c.C1744T	p.P582S	APCTEPLPRAAEPGYLVTKVAVDG	APCTEPLPRAAESGYLVTKVAVDG
11	PCMTD1	nonsynonymous SNV	c.G440C	p.R147T	AMKPEEPPQNLREKIMKLPESL	AMKPEEPPQNLTEKIMKLPESL
11	PCNT	nonsynonymous SNV	c.A3758G	p.Q1253R	HRLVLELESRRRLQQAQEAALR	HRLVLELESRRRLQQAQEAALR
11	PGAM4	nonsynonymous SNV	c.G719T	p.C240F	KPMQFLGDEETVCKAIEAVAAQGA	KPMQFLGDEETVFKAIEAVAAQGA
11	PGC	nonsynonymous SNV	c.T2A	p.MIK	MKWMVVVLVCLQLLEAAVVKVPLKK	KKWMVVVLVCLQL
11	PLEKHA4	nonsynonymous SNV	c.G740C	p.R247P	PLSRPPSLSLPRPSAPARRPPAP	PLSRPPSLSLPPRSAPARRPPAP
11	POM121	nonsynonymous SNV	c.T2266C	p.Y756H	PAPPSMIKVVPAVPTPIHPIFGGA	PAPPSMIKVVPAHVPTPIHPIFGGA
11	POTEM	nonsynonymous SNV	c.C280A	p.L94I	TSGDHDDSAMKTLRSKMGKWCCHCF	TSGDHDDSAMKTIRSKMGKWCCHCF
11	pp8961	nonsynonymous SNV	c.G419T	p.G140V	SVSGGADAVPLEGAPAVRSKS	SVSGGADAVPLEVAPAVRSKS
11	PRB2	nonsynonymous SNV	c.G326A	p.R109Q	KPQGPPQGDKSRSPRPPGKQGP	KPQGPPQGDKSQSPRPPGKQGP
12	PRB3	nonsynonymous SNV	c.C562T	p.P188S	PPHPGKPEGPPQGGNQSQGP	PPHPGKPEGPPSQQGGNQSQGP
12	RAB3GAP1	nonsynonymous SNV	c.G2126A	p.G709D	SCLLEQPEVLVTGAGRGHAGRIHK	SCLLEQPEVLVTDAGRGHAGRIHK
12	RAB40A	nonsynonymous SNV	c.T458C	p.F153S	AQAYAERLGVTFEVSPLCNFNIE	AQAYAERLGVTFEVSPLCNFNIE
12	RAD21-AS1	nonsynonymous SNV	c.C115T	p.P39S	VPVRLRSQKPPPPRRSGAGAMTI	VPVRLRSQKPPSPPRRSGAGAMTI
12	RARG	nonsynonymous SNV	c.T25C	p.C9R	MLALPLPPCWSGAPPGEESAAAGP	MLALPLPPRWSGAPPGEES
12	RPL23AP32	nonsynonymous SNV	c.C139G	p.Q47E	HKKKKIRMSPTFQRPKTLRLWRPPR	HKKKKIRMSPTFERPKTLRLWRPPR

12	RPL30	nonsynonymous SNV	c.A268G	p.K90E	SQVQAILLPQPPK	SQVQAILLPQPE
12	RPS14P3	nonsynonymous SNV	c.T422G	p.L141R	IEDVTPIPSDSTLRKGVTVVAVCEQ	IEDVTPIPSDSTRRKGVTVVAVCEQ
12	RPS15	nonsynonymous SNV	c.G140C	p.R47P	QLYSARQRRRLNRGLRRKQHSLLKR	QLYSARQRRRLNPGLRRKQHSLLKR
12	RTL1	nonsynonymous SNV	c.G3836C	p.R1279P	APPSHTAATHPPPRHLMDPQVLEF	APPSHTAATHPPPRHLMDPQVLEF
12	SFPQ	nonsynonymous SNV	c.G400A	p.A134T	GSAPPASSSAPPATPPTSGAPPGSG	GSAPPASSAPPTTPPTSGAPPGSG
12	SLC34A3	nonsynonymous SNV	c.C1367A	p.A456D	LQVALIHFFFNLAGILLWYLVPALR	LQVALIHFFFNLDGILLWYLVPALR
13	SPDYE1	nonsynonymous SNV	c.G1001A	p.R334H	DPEHWVWARDRARLS	DPEHWVWARDRAHLS
13	SPDYE3	nonsynonymous SNV	c.C1018A	p.L340I	APGVDPSPPRRSLGCKRKRECLDES	APGVDPSPPRRSIGCKRKRECLDES
13	SPEN	nonsynonymous SNV	c.A9613C	p.T3205P	AKTPDAKAAPTTPAPVVPVPLPA	AKTPDAKAAPTPPAPVVPVPLPA
13	SREBF1	nonsynonymous SNV	c.C2903A	p.A968D	LATTPASSSIDKAVQLFLCDLLLVV	LATTPASSIDKDVQLFLCDLLLVV
13	SUB1	nonsynonymous SNV	c.G167A	p.S56N	ETSRALSSSKQSSSRDDNMFQIGK	ETSRALSSSKQSNSSRDDNMFQIGK
13	THAP7	nonsynonymous SNV	c.C61A	p.R21S	GCCTRDTRERNRGISFHRLPKKDN	GCCTRDTRERNSGISFHRLPKKDN
13	TLE2	nonsynonymous SNV	c.A664C	p.T222P	KSCDSSPPQDASTPGPSSASHLCQL	KSCDSSPPQDASPPGPSSASHLCQL
13	TMEM184A	nonsynonymous SNV	c.A383T	p.E128V	YYVYFDSVRDCYEAFFVIYSFLSLCF	YYVYFDSVRDCYVAFVIYSFLSLCF
13	TPSD1	nonsynonymous SNV	c.A65G	p.E22G	HPQWVLTAAHCEVPVQLREHLYYQ	HPQWVLTAAHCVGPVQLREHLYYQ
13	TPSD1	nonsynonymous SNV	c.A251G	p.E84G	HPQWVLTAAHCEVPDIKDLAALRVQ	HPQWVLTAAHCVGPDIKDLAALRVQ
13	TTBK1	nonsynonymous SNV	c.G1402A	p.A468T	VAAQQPASRSHGAAPALDTAITSSR	VAAQQPASRSHGTAPALDTAITSSR
13	TTBK1	nonsynonymous SNV	c.G3538A	p.A1180T	VAAQQPASRSHGAAPALDTAITSRL	VAAQQPASRSHGTAPALDTAITSRL
14	TUBB4Q	nonsynonymous SNV	c.G313C	p.G105R	CLQGFQLTHSLGGGTGSGMGTLIS	CLQGFQLTHSLGRGTGSGMGTLIS
14	UGT1A3	nonsynonymous SNV	c.G775C	p.G259R	DILSHASVWLFRRDFVMDYPRPIMP	DILSHASVWLFRRDFVMDYPRPIMP
14	UGT1A3	nonsynonymous SNV	c.A850G	p.R284G	NMVFINGINCANRKLPSQEFAYIN	NMVFINGINCANGKPLSQEFAYIN
14	UQCRC2	nonsynonymous SNV	c.G23A	p.G8D	MKLLTRAGSFSRFYSLKVAP	MKLLTRADSFYSLKVAP
14	USP32	nonsynonymous SNV	c.G2149T	p.A717S	KDMSWPEEMSFISNSSKIDRHKVPT	KDMSWPEEMSFISNSSKIDRHKVPT
14	USP35	nonsynonymous SNV	c.T515A	p.L172H	HRLLCQQLVRCGRFRCPAEGEEG	HRLLCQQLVRCGRFRCPAEGEEG
14	WASH3P	nonsynonymous SNV	c.C571A	p.Q191K	LRSMKEREKLEKKQKEQEVVRSQ	LRSMKEREKLEKKKQKEQEVVRSQ
14	X97876	nonsynonymous SNV	c.A146C	p.E49A	FINSESVGNNTRELFPDGDGSCQWS	FINSESVGNNTRELFPDGDGSCQWS
14	XRCC1	nonsynonymous SNV	c.C1292A	p.P431Q	KPPTPEETKAASQVLQEDIDIEGVQ	KPPTPEETKAASQVLQEDIDIEGVQ
14	ZC3H11A	nonsynonymous SNV	c.C1650A	p.D550E	EAKEASGETTGVDITKIQVKRCETM	EAKEASGETTGEITKIQVKRCETM

14	ZNF100	nonsynonymous SNV	c.C1439A	p.T480N	RSSQLTAHKMIHTGEKPYKCEECGK	RSSQLTAHKMIHNGEKPYKCEECGK
14	ZNF107	nonsynonymous SNV	c.A1135G	p.T379A	QSSNLTEHKKIHTAEKSYKCEECGK	QSSNLTEHKKIHAAEKSYKCEECGK
15	ZNF181	nonsynonymous SNV	c.T692A	p.I231N	LISHSGEKPYKCIIECGKAFSHVSSL	LISHSGEKPYKCEECGKAFSHVSSL
15	ZNF181	nonsynonymous SNV	c.G785T	p.G262V	HTGEKPYECMNCGKSFVSHLIEH	HTGEKPYECMNCVKSFSRVSHLIEH
15	ZNF34	nonsynonymous SNV	c.C325A	p.L109M	VLDVQGTSGKEHLRVNSPALGTRTE	VLDVQGTSGKEHMRVNSPALGTRTE
15	ZNF429	nonsynonymous SNV	c.T1876C	p.C626R	IHTREKPYKCEECAKAFTRSSRLTQ	IHTREKPYKCEERAKAFTRSSRLTQ
15	ZNF43	nonsynonymous SNV	c.G820A	p.E274K	STLTKHKRIHTGEKPYTCEECGKAF	STLTKHKRIHTGKKPYTCEECGKAF
15	ZNF430	nonsynonymous SNV	c.A1303G	p.K435E	TGEKPYKCEQCCKAFNESSNLTAHK	TGEKPYKCEQCCEAFNESSNLTAHK
15	ZNF676	nonsynonymous SNV	c.C1317G	p.H439Q	GKAFSWSSSLTEHKRIHAGEKPYKC	GKAFSWSSSLTEQKRIHAGEKPYKC
15	ZNF681	nonsynonymous SNV	c.A14T	p.K5I	MEPLKFRDVAIEFSLEEWQCLDTIQ	MEPLIFRDVAIEFSLEE
15	ZNF746	nonsynonymous SNV	c.G118A	p.G40R	QAARLLSLEGRGTMAEKKLADCEKT	QAARLLSLEGRTRMAEKKLADCEKT
15	ZP3	nonsynonymous SNV	c.G580A	p.V194I	SRNMIYITCHLKVTLAEQDPDELNK	SRNMIYITCHLKITLAEQDPDELNK
15	ARMCX4	nonsynonymous SNV	c.G5440C; c.A5465G	p.G1814R p.E1822G	YMGSCVGAEEAGAGAEAGAGAEAGAG	YMGSCVGAEEAGARAEAGAGAGAGAGAEAGAE
15	BC101079	nonsynonymous SNV	c.T476C c.T479G	p.L159P p.L160R	RHSGATRCCFSELLSNLHSLGNKKT	RHSGATRCCFSEPRSNLHSLGNKKT
16	DUX2	nonsynonymous SNV	c.C524T c.G550A c.G555T	p.P175L p.G184S p.E185D	LCSAAPGGGHPAPSWVAFHAHTGEWGTGL PAPHVPC	LCSAAPGGGHPALSWVAFHAHTSDWGTGLPAPHVPC
16	DUX4L4	nonsynonymous SNV	c.C1063A	p.Q355K	RQQMQGIPAPSQALQEPAPWSALP	RQQMQGIPAPSKALQEPAPWSALP
16	ZNF66	nonsynonymous SNV	c.G1110T c.C1127G	p.W370C p.T376R	PYKCEECGKAFNWSSHLTTHKRIHTGEK YK	PYKCEECGKAFNCSSHLTRHKRIHTGEKPYK
16	ZNF724	nonsynonymous SNV	c.A1043G c.C1057G c.A1075G c.G1105A	p.N348S p.L353V p.I359V p.E369K	KPYKCEECGKAFNVSSLTQHKRIHTGEK PYKCEECGKAFNVSSLT	KPYKCEECGKAFVSSSTVTQHKRVHTGEKPYKCKEKGKAF NVSSLT
16	FAM182B	nonsynonymous SNV	c.G85A c.A88G	p.G29R p.I30V	WMWNVEEEEHEVGICTWGGQHCPCPA	WMWNVEEEEHEVRVCTWGGQHCPCPA
16	FAM182B	nonsynonymous SNV	c.T394G c.G431C	p.W132G p.G144A	QAAVSDASLGPESWQEGCRPGLTSGQHG GRDGR	QAAVSDASLGPESWQEGCRPGLTSAQHGGRDGR
16	DUX4L4	nonsynonymous SNV	c.C880A c.T896A c.C923A c.T928G	p.Q294K p.L299H p.P308Q p.W310G	AVAQPGPAQAGPQGGVLPPTSQGSQW WGWGRGPQVAGAA	AVAQPGPAQAGPKQGGVHAPPTSQGSQWGWGRGPQVAG AA
16	HNRNPL	nonsynonymous SNV	c.A224C c.T230C	p.H75P p.L77P	AQVATAPSMGTPHPLPHPSMALTPTA	AQVATAPSMGTPPPPHPSMALTPTA
16	ICOSLG	nonsynonymous SNV	c.A1240G c.A1253G c.T1255C	p.M414V p.E418G p.W419R	VQGGSRHRRPAPMGCEVWQAPAPSPRG VS	VQGGSRHRRPAPVGCPRVQAPAPSPRGVS

17	LINC00273	nonsynonymous SNV	c.G1049T c.G1063A c.C1067T c.C1070T c.C1091A	p.G350V p.G355R p.T356M p.P357L p.A364E	RGAGGEPARSGAGHAPDGTPTGTATARAA SQPLGRRPGPA	RGAGGEPARSGAVHAPDRMLGTATAREASQPLGRRPGPA
17	LOC440563	nonsynonymous SNV	c.G624T c.A625T c.A635G	p.Q208H p.S209C p.E212G	SLENLEKIEKEQSKQEVEVKNAKSEEEQ	SLENLEKIEKEHCKQGVEVKNAKSEEEQ
17	LOC729732	nonsynonymous SNV	c.C146T c.C160A	p.A49V p.R54S	GPGHGQGTATSWARTGERGAGPGPACPR CA	GPGHGQGTATSWVRTGESGAGPGPACPRCA
17	LOC100132 831	nonsynonymous SNV	c.G85C c.A89C	p.A29P p.H30P	VLMRRSMAEGEPAHPASDVLCSLAN	VLMRRSMAEGEPPPPASDVLCSLAN
17	LOC100288 255	nonsynonymous SNV	c.G303C c.C329G	p.M101I p.T110R	SSSYRENCRKRKMSKDCQDTAGNCEPEK ECCLS	SSSYRENCRKRKISSKDCQDRAGNCEPEKCSLS
17	MUC4	nonsynonymous SNV	c.G10847C c.A10864G	p.R3616P p.T3622A	TDTSSASTGDTTRLPVTDTSASTGQATPL P	TDTSSASTGDTTLPVTDASSASTGQATPLP
17	USP17L11	nonsynonymous SNV	c.T85G c.G101A c.C104G	p.F29V p.R34Q p.T35S	FSKLTSSRPDAFAEIQRTSLPEKSPLSCET	FSKLTSSRPDAVAEIQSSLPEKSPLSCET
17	ZNF737	nonsynonymous SNV	c.A1153G c.A1156G c.A1192C c.G1210A c.T1217A c.A1223G c.T1231G	p.T385A p.T386A p.K398Q p.E404K p.F406Y p.Y408C p.S411A	ECGKAFNWSSHLTTHKRIHTGEKPYKCEE CGEAFKYSSSLTTHKIIHTGQQ	ECGKAFNWSSHLAAHKRIHTGEKPYQCEECGKAYKCSSALT THKIIHTGQQ
17	FANCD2	frameshift deletion	c.1278_1278del	p.426_426del	EQLLQSTFSVHYLVLKDMCSSILSL	EQLLQSTFSVHYFLRICVHPFCRWLRVCFTL
18	PLA2G2C	frameshift deletion	c.26delT	p.L9fs	MKVIAILTLFLCSPHSSFW	MKVIAILTPSSAPPTAVSGSFRGGSNTSRGEVPSHITDMA ATVGLGKIGSPWMTLTGTAPHLPLPTRSS
18	RUNX1-IT1	frameshift deletion	c.186_187del	p.62_63del	LSGEIKVSLSLSLYLTHHTHTHTH	LSGEIKVSLSLSLSHHTHTHTHTHREREREREREREASIQ NSHA
18	HGC6.3	frameshift deletion	c.404delT	p.F135fs	HTAGVMTPTASSFPTHCRGNPCHL	HTAGVMTPTASSPHTAGGMTPTAFSSPNTAGGMTPTASCS PHTHCRGSGA
18	RPS14P3	frameshift insertion	c.429dupG	p.K143fs	DVTPIPSDSTLRKGVTVVAVCEQDS	DVTPIPSDSTLRKGGHRGRRL
18	KRT18	frameshift insertion	c.174_175ins A	p.M58fs	ISVSRSTFRGGMGSGGLATGIAGG	ISVSRSTFRGGMRVRGPGHRDSRGSGRNHRPEREGDHAK PERPPGLPGQSEEPGDREPEAGEQNPALGEEGTPGQRLEP LLQDHRGPEGSDLRKYCGQCPHRSAD
18	FAM182B	frameshift insertion	c.446_447ins GCCG	p.R149fs	CRPGLTSGQHGGRRDR	CRPGLTSGQHGGRRPWKVA
19	ADRA2C	frameshift insertion	c.48dupG	p.A16fs	PALAAALAVAAAAGPNASGAGERGS	PALAAALAVAAAAGPQCERRREGQRRCQCLGGFLGAAA RPVLGGRGGRAGCRGGLPHRLHRGQQRAGGDRRADQPGA ARATEPLPGVAGLGRHPGGHAGHALLVQRAHGLLVLRAG VRRRVPGARCAVLHLVDRASVCHQGPLLVGDAGRVRQPE AHTTPRQGHHRGRVAHLGRHLLPAAGLALPPARRRRLPAV RPQRRDLVHPVLLHRLLLRALPHHGPLRAHLPSGQAAHAH AQREARPRGPRRCVP
19	LINC00273	nonframeshift deletion	c.1024_1050del	p.342_350del	CRHRLLLLRGAGGEPARSGAGHAPD	CRHRLLLLRGAGHAPDGTPTGTATAR



20	TMEM181	frameshift insertion	c.43dupG	p.G14fs	GAAPSPTQASSRGGGPGPPAPTRAV	GAAPSPTQASSRGGGAGSPSPHPRRVQLLACSRRRSPERTGSE SCAPPHLLPRTCSSSSVPPSAPAARSVVLGEERGRRRRHKGW SGGTGTREAAARHGRRVPCL
20	ZNF91	nonframeshift insertion	c.898_899ins AAT	p.S300delinsKC	KPYKCEECGKAFFRSSTLAKHKRIH	KPYKCEECGKAFFCRSSTLAKHKRIH

### Patient #3

TMG	Gene name	Mutation type	cDNA change	Amino acid change	Wild type minigene sequence	Mutated minigene sequence
1	AACS	nonsynonymous SNV	c.C1354A	p.L452I	DIISCFMGHNFSLPVYKGEIQARNL	DIISCFMGHNFSPVYKGEIQARNL
1	ADARB2	nonsynonymous SNV	c.G703A	p.G235R	LFQEFEPAPRPLAGGRPGDAALL	LFQEFEPAPRPLAGGRPGDAALL
1	AK302451	nonsynonymous SNV	c.A442T	p.T148S	QSSRTSSTCSCQTSRTSSTSCSYQS	QSSRTSSTCSCQSSRTSSTSCSYQS
1	AK302451	nonsynonymous SNV	c.A139T	p.T47S	QTSSTCSCQSSGTSSTSCNWQSSRI	QTSSTCSCQSSGSSSTSCNWQSSRI
1	ALPK1	nonsynonymous SNV	c.A332G	p.K111R	ATGTWLYRNE SDKVLVQSVCIQIRG	ATGTWLYRNE SDRVLVQSVCIQIRG
1	ANKRD30A	nonsynonymous SNV	c.C688A	p.P230T	EGTSAGTPDEAAPLAERTPDTAESL	EGTSAGTPDEAATLAERTPDTAESL
1	ANXA13	nonsynonymous SNV	c.A521G	p.D174G	EGDDVDKDLAQDAKDLYDAGEGRW	EGDDVDKDLAQGAKDLYDAGEGRW
1	ARHGAP5	frameshift deletion	c.1630_1633del	p.K544fs	QKLAPDRESLLK HIGFVYHPTKET	QKLAPDRESLLI
1	ARHGEF18	nonsynonymous SNV	c.C3047A	p.P1016H	AERPEVARRDSAPTENRLAKSDVPI	AERPEVARRDSAHTENRLAKSDVPI
1	ARL13A	frameshift deletion	c.84delC	p.G28fs	EETRRNVTIPII GLNNSGKTVLVEA	EETRRNVTIPIIG
1	ARMCX4	nonsynonymous SNV	c.G2435C	p.W812S	TLPGAKNKVRGNWNAVSKAGAGMDT	TLPGAKNKVRGNSNAVSKAGAGMDT
1	ARMCX4	nonsynonymous SNV	c.A2894T	p.K965I	VASFQGEVLP GAKNKVRGNSNAVPK	VASFQGEVLP GAINKVRGNSNAVPK
2	ARMCX4	nonsynonymous SNV	c.T3887G	p.V1296G	GNMSSVSYWAGVVDQAGGGSWAGTS	GNMSSVSYWAGVGDQAGGGSWAGTS
2	ARMCX4	nonsynonymous SNV	c.G2436C	p.W812C	TLPGAKNKVRGNWNAVSKAGAGMDT	TLPGAKNKVRGNCAVSKAGAGMDT
2	ASAH2	nonsynonymous SNV	c.T17A	p.F6Y	MAKRTFSNLETFLIFLLV	MAKRTYSNLETFLIFLLV
2	ASAP1	nonsynonymous SNV	c.C2964A	p.D988E	QLSDLPPKPQMKDLPPKPQLGDLLA	QLSDLPPKPQMKELPPKPQLGDLLA
2	ASF1B	nonsynonymous SNV	c.C367T	p.R123C	YYVNNEYLNPELRENPPMKPDFSQL	YYVNNEYLNPELCENPPMKPDFSQL
2	ASPM	nonsynonymous SNV	c.G2542A	p.A848T	ISLEDNSDVTGLAMFILNRLWNPD	ISLEDNSDVTGLTMFILNRLWNPD
2	ZNF98	nonsynonymous SNV	c.G724C	p.E242Q	RIHTGKKPYKCEECGKAFNRLSHLT	RIHTGKKPYKCEQCGKAFNRLSHLT
2	AX746755	nonsynonymous SNV	c.C546G	p.I182M	VLHTFFLSPAGIILACTILLTFFLS	VLHTFFLSPAGIMLACTILLTFFLS
2	AX747493	nonsynonymous SNV	c.T131A	p.F44Y	LAQQGPQHDPMAFPSTQSTCSSGGV	LAQQGPQHDPMAYPSTQSTCSSGGV
2	AX747493	nonsynonymous SNV	c.T167A	p.V56D	FPSTQSTCSSGGVPEMSLAQQAQAFQY	FPSTQSTCSSGGDPEMSLAQQAQAFQY
2	AX747493	nonsynonymous SNV	c.T202A	p.Y68N	VPMSLAQQAQAFQYQPVAFSTQSTS	VPMSLAQQAQAFQNPVAFSTQSTS
2	AX747985	nonsynonymous SNV	c.T170A	p.L57Q	PNPTSSTESLGYLSSLSQPEPL	PNPTSSTESLGYQSSLSQPEPL
3	AXL	nonsynonymous SNV	c.C162A	p.H54Q	ACTSSQGPSSWTHWLPVETPEGVPL	ACTSSQGPSSWTQWLPVETPEGVPL
3	C2CD2L	nonsynonymous SNV	c.A134G	p.D45G	YARGLWLARARGDRGPGPALAGEPA	YARGLWLARARGGRGPGPALAGEPA

3	C2orf16	nonsynonymous SNV	c.G4988T	p.R1663L	ERSHRSPERSHRSSSERRHRSPSQ	ERSHRSPERSHLSSSERRHRSPSQ
3	C2orf16	nonsynonymous SNV	c.G5012T	p.R1671L	ERSHRSSERRHRSPSQRSHRGPSE	ERSHRSSERRHLSPSQRSHRGPSE
3	CAD	nonsynonymous SNV	c.C4810G	p.Q1604E	EQQTVAAVLMVAQLTQRSVHICHVA	EQQTVAAVLMVAELTQRSVHICHVA
3	CAMSAP3	nonsynonymous SNV	c.C362G	p.P121R	ARRGTVPALPERPVREADLRHQPIIL	ARRGTVPALPERRVREADLRHQPIIL
3	CCDC106	nonsynonymous SNV	c.C374G	p.A125G	GPRRMEGDSRGGAGGEASDPESAAS	GPRRMEGDSRGGGGGEASDPESAAS
3	CCDC112	nonsynonymous SNV	c.A79C	p.T27P	AVAGAVAGAGAATGTGVGATPAPQQ	AVAGAVAGAGAAPGTGVGATPAPQQ
3	CCDC17	nonsynonymous SNV	c.C461T	p.A154V	FRTRARRVAEMEAQSRALQLRGEEL	FRTRARRVAEMEVQSRALQLRGEEL
3	CCNB3	nonsynonymous SNV	c.C2516G	p.T839S	FKEPLALQEEPSTEKEAVLKEPSVD	FKEPLALQEEPSSEKEAVLKEPSVD
3	CCND1	nonsynonymous SNV	c.G58A	p.A20T	CEVETIRRAYPDANLLNDRVLRAML	CEVETIRRAYPDTNLLNDRVLRAML
3	CDH22	nonsynonymous SNV	c.G1168A	p.E390K	QAIVRVAVTDVDEPPEFRPPSGLLE	QAIVRVAVTDVDKPEFRPPSGLLE
4	COL11A1	nonsynonymous SNV	c.C1648G	p.P550A	LGPRGTPGAPGQPGMAGVDGPPGPK	LGPRGTPGAPGQAGMAGVDGPPGPK
4	COL1A1	nonsynonymous SNV	c.C3023T	p.P1008L	PPGPMGPPGLAGPPGESGREGAPGA	PPGPMGPPGLAGLPGESGREGAPGA
4	COL6A5	nonsynonymous SNV	c.A671T	p.H224L	YKEGAVDADMVQVHFPISCQKDSLAD	YKEGAVDADMVQLFPISCQKDSLAD
4	CRLF1	nonsynonymous SNV	c.G214A	p.E72K	CSVHGDPPGATAEGLYWTLNGRRLP	CSVHGDPPGATAKGLYWTLNGRRLP
4	CYLC2	nonsynonymous SNV	c.C519A	p.D173E	SKKGKKDAEKGDATESEDEKGGGA	SKKGKKDAEKGKESATESEDEKGGGA
4	CYP2D6	nonsynonymous SNV	c.G955A	p.V319I	AVIHEVQRFGDIVPLGVTHMTRSRI	AVIHEVQRFGDIPLGVTHMTRSRI
4	CYP2D6	nonsynonymous SNV	c.A911G	p.Y304C	RRPEMGDQAHMPYTTAVIHEVQRFG	RRPEMGDQAHMPCTTAVIHEVQRFG
4	CYP3A5	nonsynonymous SNV	c.C914T	p.A305V	LELAAQSIIFIFAGYETTSSVLSFT	LELAAQSIIFVGYETTSSVLSFT
4	DCC	nonsynonymous SNV	c.C1382T	p.T461I	WRPPAEAKGNIQFTVFFSREGDNR	WRPPAEAKGNIQFTVFFSREGDNR
4	DHTKD1	nonsynonymous SNV	c.C1258T	p.R420C	ATRLAFEYQRQFRKDVIIIDLLCYRQ	ATRLAFEYQRQFCKDVIIIDLLCYRQ
4	DPCR1	nonsynonymous SNV	c.G1413C	p.K471N	GPTENRETTANEKTTLSPVEPTENR	GPTENRETTANENTLSPVEPTENR
4	DPCR1	nonsynonymous SNV	c.C2747T	p.P916L	GERTPFTNEKTPSSAEPTEHGERT	GERTPFTNEKTTLSSAEPTEHGERT
5	DPCR1	nonsynonymous SNV	c.C2687A	p.S896Y	EMTPLANEKTTLSPAEPTEGERTP	EMTPLANEKTTLYPAEPTEGERTP
5	DUSP2	nonsynonymous SNV	c.G653T	p.R218L	SASCPNHFEGLFRYKSIPIVEDNQMV	SASCPNHFEGLFLYKSIPIVEDNQMV
5	EGF	nonsynonymous SNV	c.G2945A	p.R982H	RCQYRDLKWWELRHAGHGQQQKVIV	RCQYRDLKWWELHHAGHGQQQKVIV
5	ELFN2	nonsynonymous SNV	c.C2045G	p.A682G	LNSPLDRLPLVPAGSGGGSGGGGGI	LNSPLDRLPLVPGSGGGSGGGGGI
5	GABARAPL2	nonsynonymous SNV	c.G6T	p.W2C	MWIIRKRIQLPSEK	MCIIRKRIQLPSEK
5	SLC25A3	nonsynonymous SNV	c.C293T	p.A98V	LDLVKCRMQVCFACWTRAY	LDLVKCRMQVCFVCWTRAY

5	AACS	nonsynonymous SNV	c.C19A	p.L7I	MGHNFSLPVYKGEIQARNL	MGHNFSIPVYKGEIQARNL
5	CCDC80	frameshift deletion	c.50delT	p.F17fs	SMGQFSVFFFFFFWAQKKIASLSHS	SMGQFSVFFFFFFSGHRKKLLA
5	ASF1B	nonsynonymous SNV	c.C367T	p.R123C	YYVNNEYLNPELRENPPMKPDFSQV	YYVNNEYLNPELCENPPMKPDFSQV
5	TIMM50	nonsynonymous SNV	c.A235C	p.T79P	ALWLAGLLGAGGTVSVVYIFDPILV	ALWLAGLLGAGGPVSVVYIFDPILV
5	ABCG2	nonsynonymous SNV	c.G10T	p.D4Y	SDGDQTLGQTRKIKT	SDGYQTLGQTRKIKT
5	ADAMTS12	nonsynonymous SNV	c.A2785G	p.K929E	VLKPNKGTISNGKNPPTLKPVPPPT	VLKPNKGTISNGENPPTLKPVPPPT
6	MAGEC1	nonsynonymous SNV	c.T331C	p.S111P	GFPQSPLQIPVSSSSSTLL	GFPQSPLQIPVSPSSSSTLL
6	WDR19	nonsynonymous SNV	c.G1087A	p.D363N	YRHPVSVKKIFPDPNGTRLVFIDEK	YRHPVSVKKIFPNPNGTRLVFIDEK
6	ZNF66	nonsynonymous SNV	c.G1332T	p.K444N	KAFSRSSILTTHKIIHTGEKPYECE	KAFSRSSILTTHNIIHTGEKPYECE
6	MSLNL	nonsynonymous SNV	c.C320A	p.P107Q	HLPMHTVYTPCTPSPTRAHSCHLPV	HLPMHTVYTPCTQSPTRAHSCHLPV
6	RNF181	nonsynonymous SNV	c.G252C	p.R84S	AVFQVRWGQVFSRSGSLGGRACSSS	AVFQVRWGQVFSSSGSLGGRACSSS
6	PHF8	nonsynonymous SNV	c.C261G	p.F87L	HKGKPVKGTGSPTFVRELRSRTFD	HKGKPVKGTGPTLVRELRSRTFD
6	GOLGA6L2	nonsynonymous SNV	c.C1709G	p.A570G	DVGAGGEDAGSGAEDVGGGEDVGA	DVGAGGEDAGSGGEDVGGGEDVGA
6	TOMM5	nonsynonymous SNV	c.G209A	p.R70Q	GHPLTLRGHRGLRTELNGSGEQGGS	GHPLTLRGHRGLQTELNGSGEQGGS
6	RABL6	nonsynonymous SNV	c.T1141G	p.W381G	RGALGRRGQWPPWGGGRACHCLGRH	RGALGRRGQWPPGGGGRACHCLGRH
6	ZNF841	nonsynonymous SNV	c.C1016A	p.T339K	QLVRHQIIHTGETPYKCNCEGKVFR	QLVRHQIIHTGEKPYKCNCEGKVFR
6	LST1	nonsynonymous SNV	c.T31G	p.W11G	MIYVSTGAWGWAGSCFWQWFCP	MIYVSTGAWGGAGSCFWQWFCP
6	ZNF99	nonsynonymous SNV	c.A2715C	p.K905N	CGKGFNNFSTLMKHKIIHTGEKPYK	CGKGFNNFSTLMNHKIIHTGEKPYK
7	FAM194B	nonframeshift deletion	c.404_421del	p.135_141del	EEEEYLGKEEHL EEEEEYLGKEGYLE	EEEEYLGKEEHLGKEGYLEKEDYIE
7	FAM47C	nonsynonymous SNV	c.T937A	p.S313T	TGVDPDLCLEPPKSRVSHLRPEPSET	TGVDPDLCLEPPKTRVSHLRPEPSET
7	FGA	nonsynonymous SNV	c.T937A	p.S313T	GPGSTGNRNPSSGTGGTATWKPGS	GPGSTGNRNPSTGTGGTATWKPGS
7	FOXC2	nonsynonymous SNV	c.A1153C	p.T385P	NLAAGQEGALAAATGHHHQHHGHHHP	NLAAGQEGALAAPGHHHQHHGHHHP
7	FZD1	nonframeshift insertion	c.1768_1769insCCC	p.A590delinsAP	AIPCPHLQAGGGAPPHPPMSPDFTV	AIPCPHLQAGGGAPPHPPMSPDFT
7	GABARAPL2	nonsynonymous SNV	c.G186T	p.W62C	LVPSDITVAQFMWIIRKRIQLPSEK	LVPSDITVAQFMCIIRKRIQLPSEK
7	H3F3A	nonsynonymous SNV	c.C344G	p.A115G	GLFEDTNLCAIHAKRVTIMPKDIQL	GLFEDTNLCAIHGKRVTIMPKDIQL
7	HMCN1	nonsynonymous SNV	c.A15862G	p.I5288V	CKDGTHQCRYNQICENTRGSYRCVC	CKDGTHQCRYNQVCENTRGSYRCVC
7	HOXA4	nonsynonymous SNV	c.A344C	p.Q115P	GYRGGASPRPPPEQPPAQAKGPA	GYRGGASPRPPPEQPPAQAKGPA
7	HOXA6	nonsynonymous SNV	c.G107C	p.R36T	QLPLYQAGYDALRPPASYGASSLP	QLPLYQAGYDALTPPASYGASSLP

7	IKBKE	nonsynonymous SNV	c.C1465T	p.H489Y	MRPGLGYNEEQIHKLDKVNFSHLAK	MRPGLGYNEEQIYKLDKVNFSHLAK
7	IMPA2	nonsynonymous SNV	c.G719T	p.R240I	HCWDLAAATVIIIEAGGIVIDTSGG	HCWDLAAATVIIIIEAGGIVIDTSGG
8	IQCA1	nonsynonymous SNV	c.G190A	p.V64I	IFRNLENYVDQFVHPQKRILIRKVL	IFRNLENYVDQFIHPQKRILIRKVL
8	IWS1	nonsynonymous SNV	c.T1238G	p.V413G	EDEEKASAKKSRVSDADSDSDAV	EDEEKASAKKSRGVSDADSDSDAV
8	KRTAP10-4	nonsynonymous SNV	c.A1026T	p.Q342H	PVCSGASSCCQSSCQPACCTTSC	PVCSGASSCCQHSSCQPACCTTSC
8	KRTAP10-4	nonsynonymous SNV	c.T787A	p.S263T	CQQSSCQPACCTSSPCQACCVPVC	CQQSSCQPACCTTSPCQACCVPVC
8	KRTAP4-9	nonsynonymous SNV	c.C211G	p.Q71E	CCQPTCSRPSCCQTTCRRTCYRPS	CCQPTCSRPSCECTTCRRTCYRPS
8	LINC00273	nonsynonymous SNV	c.C383T	p.A128V	HRGQPPEPSSLRALHADPIPSGLSP	HRGQPPEPSSLRVLHADPIPSGLSP
8	LINC00273	nonsynonymous SNV	c.T341A	p.L114H	PSMKAAPRLPHRLRHRGQPPEPSSL	PSMKAAPRLPHRHRHRGQPPEPSSL
8	LOC554223	nonframeshift deletion	c.438_458del	p.146_153del	RLSDATSPRMEPRAPWMEQEGPEY	RLSDATSPRMEPEGPEYWEETGT
8	LOC554223	nonframeshift deletion	c.331_351del	p.111_117del	ARQRRHESQDGAAGAVDGAGGAGIL	ARQRRHESQDGAGGAGILGRGDRDR
8	LONRF3	nonsynonymous SNV	c.C443G	p.A148G	VAAEETGAAAAAAATEVWDGFKCRK	VAAEETGAAAAAGATEVWDGFKCRK
8	LRP10	frameshift deletion	c.1250_1269del	p.V417fs	RCRDEKCVYETWVCDGQPCADGSD	RCRDEKCVYETWVCGRQ
8	LRRC37A6P	nonsynonymous SNV	c.T997A	p.L333M	SNLTQATVQPLDLGLTTTPESTTEI	SNLTQATVQPLDMGLTTTPESTTEI
9	LST1	nonsynonymous SNV	c.T47G	p.L16R	RNDDICIYGGLGLGLLLLAVVLLS	RNDDICIYGGLGRGLLLLAVVLLS
9	MAGEC1	nonsynonymous SNV	c.T925C	p.S309P	GFPQSPLQIPVSSSSSTLLSLFQS	GFPQSPLQIPVSPSSSSTLLSLFQS
9	MCF2L2	nonsynonymous SNV	c.T3314G	p.L1105R	PAGATAGFQARALRPRTSAQES	PAGATAGFQARARRPRTSAQES
9	MGAT4B	nonsynonymous SNV	c.C701A	p.P234H	IHSGLLEVISPSHFYPDFSRLRES	IHSGLLEVISPSHHFYPDFSRLRES
9	MLIP	nonsynonymous SNV	c.C794T	p.T265M	LDVGGAVVEESATYFQTTAHSTPFS	LDVGGAVVEESAMYFQTTAHSTPFS
9	MMP21	nonsynonymous SNV	c.A64C	p.T22P	TLLLCWLAAPWPTQPESLFHSRDRS	TLLLCWLAAPWPPQPESLFHSRDRS
9	MTMR11	nonsynonymous SNV	c.T7G	p.W3G	MWWGGRGQSFNIAPQ	MWGGGRGQSFNIAPQ
9	MUC12	nonsynonymous SNV	c.G2248C	p.A750P	EPTSYHSSPGSTATTHFPDSSTTSG	EPTSYHSSPGSTPTTHFPDSSTTSG
9	MUC17	nonsynonymous SNV	c.C1293A	p.S431R	DSKTFVTTASEASSPTTAEDTSIA	DSKTFVTTASEARSSPTTAEDTSIA
9	MUC21	nonsynonymous SNV	c.T243A	p.H81Q	SNGVSIVTNSEFHTTSSGISTATNS	SNGVSIVTNSEFQTTSSGISTATNS
9	MUC21	nonsynonymous SNV	c.C877A	p.P293T	GISTVTNSESSTPSSGANTATNSES	GISTVTNSESSTTSSGANTATNSES
9	MUC22	nonsynonymous SNV	c.G253T	p.A85S	SETTSASTMASTAAFTTGSETNTAS	SETTSASTMASTSAFTTGSETNTAS
10	MYC	nonsynonymous SNV	c.A217C	p.T73P	SEDIWKKFELLPTPLSPRRSGLC	SEDIWKKFELLPPPLSPRRSGLC
10	NR0B1	nonsynonymous SNV	c.T271A	p.Y91N	ILYSMLTSAKQTYAAPKAPEATLGP	ILYSMLTSAKQTNAAPKAPEATLGP

10	NRTN	nonsynonymous SNV	c.T490G	p.C164G	RRLRRERVRAQPCCRPTAYEDEVSF	RRLRRERVRAQPGRPTAYEDEVSF
10	PCDH12	nonsynonymous SNV	c.C2385G	p.H795Q	GQAGEPCEVGQSHKDVDKEAMMEAG	GQAGEPCEVGQSQKDVDKEAMMEAG
10	PCDHGA12	nonsynonymous SNV	c.G409A	p.E137K	DINDNAPYFRESELEIKISENAATE	DINDNAPYFRESKLEIKISENAATE
10	PDE1C	nonsynonymous SNV	c.G256C	p.D86H	VHDPRPPEEILADELPQLDSPEVLV	VHDPRPPEEILAHHELPQLDSPEVLV
10	PDE1C	nonsynonymous SNV	c.G256C	p.D86H	VHDPRPPEEILADELPQLDSSEVLV	VHDPRPPEEILAHHELPQLDSSEVLV
10	PHF8	nonsynonymous SNV	c.C369G	p.F123L	HKGKPVKTGSPTFVRELRSRTFDSS	HKGKPVKTGSPTLVRELRSRTFDSS
10	POLE	nonsynonymous SNV	c.G3796C	p.E1266Q	ILGQPPALGTSQEEWLWVLRFHKKK	ILGQPPALGTSQEEWLWVLRFHKKK
10	POLG2	nonsynonymous SNV	c.G199C	p.E67Q	EHPEAPGSGEGSEALLEICQRRHFL	EHPEAPGSGEGSQALLEICQRRHFL
10	POLR2A	nonsynonymous SNV	c.C5744A	p.T1915N	TPTSPKYSPTSPTYSPKYSPTS	TPTSPKYSPTSPNYSPTS
10	PRNP	nonsynonymous SNV	c.T124G	p.W42G	ASWWWLGAASWWLGAAPWWLWGT	ASWWWLGAASWWLGAAPWWLWGT
11	PRSS1	nonsynonymous SNV	c.C44G	p.A15G	PLLILTFVAAAALAPFDDDDKIVGG	PLLILTFVAAAALGAPFDDDDKIVGG
11	PRSS1	nonsynonymous SNV	c.C47T	p.A16V	LLILTFVAAAALAPFDDDDKIVGGY	LLILTFVAAAALAVPFDDDDKIVGGY
11	PRSS1	nonsynonymous SNV	c.C49T	p.P17S	LILTFVAAAALAPFDDDDKIVGGYN	LILTFVAAAALASFDDDDKIVGGYN
11	RBM11	nonsynonymous SNV	c.G603C	p.Q201H	WTHQQPSDSLDYQMTAPLNSASVS	WTHQQPSDSLDYHMTAPLNSASVS
11	RBMXL3	nonsynonymous SNV	c.C1654A	p.R552S	SYGQSHRYGGEGRYEYRGRSHDAHS	SYGQSHRYGGEGSYEYRGRSHDAHS
11	RBMXL3	nonsynonymous SNV	c.G1256T	p.G419V	SSNSYGQSHHYGGEGRYEYRGRSH	SSNSYGQSHHYGVEGRYEEYRGRSH
11	RERE	nonsynonymous SNV	c.A1415G	p.N472S	RRIKTRTASTPVNTSPRPPSSEFLD	RRIKTRTASTPVSTSPRPPSSEFLD
11	RING1	nonsynonymous SNV	c.A760C	p.S254R	GTLGPPSPGAPSPPEPGGEIELVF	GTLGPPSPGAPRPEPGGEIELVF
11	RUNX1	nonsynonymous SNV	c.A644C	p.H215P	QLRRTAMRVSPHHPAPTNPASLN	QLRRTAMRVSPHHPAPTNPASLN
11	RUNX1	nonsynonymous SNV	c.A20C	p.H7P	MRVSPHHPAPTNPASLN	MRVSPHHPAPTNPASLN
11	SF3B1	nonsynonymous SNV	c.G2704A	p.E902K	EQLIDGILYAFQEQTTEDSVMLNGF	EQLIDGILYAFQKQTTEDSVMLNGF
11	SHC3	nonsynonymous SNV	c.C271G	p.R91G	SGLRGLSSAARERAGARLSGSCSAP	SGLRGLSSAAREGAGARLSGSCSAP
12	SIKE1	nonsynonymous SNV	c.G275A	p.R92K	ENTQIRDLQENRELWISLEEHQDA	ENTQIRDLQENKELWISLEEHQDA
12	SLC22A24	nonsynonymous SNV	c.G10C	p.D4H	MGFDVLLDQVGGMGRF	MGFHVLLDQVGGMGRF
12	SPATA7	nonsynonymous SNV	c.G193A	p.E65K	RREKLKKELAQCEKEFKLTKTAMRA	RREKLKKELAQCKEKEFKLTKTAMRA
12	SPNS1	nonsynonymous SNV	c.A1235C	p.E412A	GGA AFLGTAIFIEADRRRAQLHVQG	GGA AFLGTAIFIAADRRRAQLHVQG
12	SRRM5	nonsynonymous SNV	c.T1326A	p.S442R	SPNKARDCSRSRSPYKARDRSRSRS	SPNKARDCSRSRSPYKARDRSRSRS
12	SSTR5	nonsynonymous SNV	c.C807G	p.I269M	FAGCWLPFFTVNIVNLAVALPQEPA	FAGCWLPFFTVNMVNLAVALPQEPA

12	ST7L	nonsynonymous SNV	c.C1405G	p.H469D	NLLQCTWEGTFHHVSVYPKKELPLF	NLLQCTWEGTFHDVSVYPKKELPLF
12	ST7L	nonsynonymous SNV	c.C1498G	p.H500D	ETADRELLPTFHHVSVYPKKELPLF	ETADRELLPTFHDVSVYPKKELPLF
12	STK10	nonsynonymous SNV	c.C714G	p.H238Q	TLIEMAQIEPPHHELNPMRVLLKIA	TLIEMAQIEPPHQELNPMRVLLKIA
12	SYNPO	nonsynonymous SNV	c.A2050C	p.T684P	GIEAQDRRESLTPSPWPWGASRPP	GIEAQDRRESLPPSPWPWGASRPP
12	TCHH	nonsynonymous SNV	c.A3417T	p.Q1139H	EREKRRRQELERQYREEEVQQEEE	EREKRRRQELERHYREEEVQQEEE
12	TEX15	nonsynonymous SNV	c.G5324C	p.R1775T	HDQLVELLEETKREKNSYYVFLKYK	HDQLVELLEETKTEKNSYYVFLKYK
13	TIMM50	nonsynonymous SNV	c.A544C	p.T182P	ALWLAGLLGAGGTVSVVYIFGNP	ALWLAGLLGAGGPVSVVYIFGNP
13	TLE2	nonsynonymous SNV	c.G439A	p.V147M	KVPICIPARRDLVDSASLASSLGS	KVPICIPARRDLMDSASLASSLGS
13	TOPAZ1	nonsynonymous SNV	c.A169C	p.T57P	ENKQKRRMVARATPGRGEVESDKSV	ENKQKRRMVARAPPGRGEVESDKSV
13	TPSAB1	nonsynonymous SNV	c.G68T	p.G23V	VLASRAYAAPAPGQALQRVGIVGGQ	VLASRAYAAPAPVQALQRVGIVGGQ
13	TPSAB1	nonsynonymous SNV	c.G83A	p.R28Q	AYAAPAPGQALQRVGIVGGQEAPRS	AYAAPAPGQALQVIVGGQEAPRS
13	TPSAB1	nonsynonymous SNV	c.T86C	p.V29A	YAAPAPGQALQRVGIVGGQEA PRSK	YAAPAPGQALQ RAGIVGGQEA PRSK
13	TRAPPC12	nonsynonymous SNV	c.G1912A	p.E638K	SAFLHLGQNNFAEHRFFTEILRMD	SAFLHLGQNNFAKAHRFFTEILRMD
13	TUBGCP6	nonsynonymous SNV	c.C3717G	p.C1239W	ENVSDVAPIRRCNTHGHVSDASIS	ENVSDVAPIRSRWNTHGHVSDASIS
13	TUBGCP6	nonsynonymous SNV	c.G3566T	p.G1189V	DMAPARPRWNTHGHVSDASISLGES	DMAPARPRWNTHVHVSDASISLGES
13	UBA1	nonsynonymous SNV	c.G1828T	p.V610L	SGTLGTKGNVQVVIPFLTESYSSSQ	SGTLGTKGNVQVLIPFLTESYSSSQ
14	UCN2	nonsynonymous SNV	c.G151A	p.A51T	TPRPAASESPSAAPTWPWAAQSHCS	TPRPAASESPSATPTWPWAAQSHCS
14	USP49	nonsynonymous SNV	c.G679C	p.A227P	TPRDAGPAASRPALPTSRRVPAAT	TPRDAGPAASRPPALPTSRRVPAAT
14	VEZF1	nonsynonymous SNV	c.A1402C	p.T468P	SPLTLTPVNLPTPV TAPVNIAHPV	SPLTLTPVNLPPPVTAPVNIAHPV
14	ZAN	nonsynonymous SNV	c.T2456A	p.M819K	ETTISTEKPSIPMEKPTLPTEETTT	ETTISTEKPSIPKEKPTLPTEETTT
14	ZNF117	nonsynonymous SNV	c.G899C	p.S300T	QSSTLTTHKRIHSGEKPYKCEECGK	QSSTLTTHKRIHTGEKPYKCEECGK
14	ZNF12	nonsynonymous SNV	c.C1754A	p.T585N	GEKPYECSECGKTFQNSALNRHQR	GEKPYECSECGKNFCQNSALNRHQR
14	ZNF135	nonsynonymous SNV	c.A1408G	p.T470A	GEKPYGCNECGKTFSSHSSLSQHER	GEKPYGCNECGKAFSSHSSLSQHER
14	ZNF135	nonsynonymous SNV	c.C1189G	p.Q397E	KAFSSHSSSLTKHQRIHTGEKPYECH	KAFSSHSSSLTKHERIHTGEKPYECH
14	ZNF14	nonsynonymous SNV	c.C956G	p.S319C	YECKEKGKAFYFASFAHVIIHTG	YECKEKGKAFYFASFAHVIIHTG
14	ZNF154	nonsynonymous SNV	c.C996A	p.S332R	HRVHTGERPYKCSECGKSFSQRSAL	HRVHTGERPYKCRECGKSFSQRSAL
14	ZNF180	nonsynonymous SNV	c.G1530T	p.Q510H	CNQCCKSFSWSSQLVAHQRTHTGEK	CNQCCKSFSWSSHLVAHQRTHTGEK
14	ZNF208	nonsynonymous SNV	c.A2157T	p.K719N	NLMEHKRIHTGEKPYKCEECGKSFS	NLMEHKRIHTGENPYKCEECGKSFS

15	ZNF208	nonsynonymous SNV	c.A1849G	p.T617A	GEKPYKCEECGKTFSKVSTLTTHKA	GEKPYKCEECGKAFSKVSTLTTHKA
15	ZNF211	nonsynonymous SNV	c.C933A	p.S311R	RRVHTGERPYECSECGKSFSQNFSL	RRVHTGERPYECRECGKSFSQNFSL
15	ZNF28	nonsynonymous SNV	c.A2049T	p.R683S	VFNQQAHLAQHQRVHTGEKPYKCNE	VFNQQAHLAQHQSVHTGEKPYKCNE
15	ZNF283	nonsynonymous SNV	c.T1251A	p.S417R	CKECGKAFNCGSSLIQHERIHTGEK	CKECGKAFNCGSRLIQHERIHTGEK
15	ZNF304	nonsynonymous SNV	c.C1071A	p.H357Q	SRSSHLVQHQRHTGERPYKCNKCG	SRSSHLVQHQRHTGERPYKCNKCG
15	ZNF345	nonsynonymous SNV	c.A1185T	p.E395D	SKLIQHQLIHTGERPYECKEKGKSF	SKLIQHQLIHTGDRPYECKEKGKSF
15	ZNF429	nonsynonymous SNV	c.T838A	p.S280T	RYSTLTTHKRIHSGEKPYKCDECGK	RYSTLTTHKRIHTGEKPYKCDECGK
15	ZNF430	nonsynonymous SNV	c.T1231A	p.W411R	FYKCEECGKGFNWSSTLTKHKRIHT	FYKCEECGKGFNRSSSTLTKHKRIHT
15	ZNF431	nonsynonymous SNV	c.C1008A	p.F336L	AFNQSSTLSTHKFIHAGEKPYKCEE	AFNQSSTLSTHKLIHAGEKPYKCEE
15	ZNF460	nonsynonymous SNV	c.T1318A	p.W440R	AFTRMSGLTRHQWIHTGEKPYVCIQ	AFTRMSGLTRHQRIHTGEKPYVCIQ
15	ZNF460	nonsynonymous SNV	c.T1326A	p.H442Q	TRMSGLTRHQWIHTGEKPYVCIQCG	TRMSGLTRHQWIHTGEKPYVCIQCG
15	ZNF501	nonsynonymous SNV	c.G123C	p.Q41H	KFFTQRSSLTQHQRHHRGEKPYVCS	KFFTQRSSLTQHHRHHRGEKPYVCS
16	ZNF530	nonsynonymous SNV	c.G1249A	p.G417S	RPYVCSECGKSFQKSVLIQHQRVH	RPYVCSECGKSFQKSVLIQHQRVH
16	ZNF616	nonsynonymous SNV	c.C1475A	p.P492H	LAAHQRIHTGEKPYKNECGKVFQ	LAAHQRIHTGEKHYKNECGKVFQ
16	ZNF675	nonsynonymous SNV	c.G1443T	p.K481N	EHKKIHSGEIPYKCEECGKAFKHSS	EHKKIHSGEIPYNCEECGKAFKHSS
16	ZNF679	nonsynonymous SNV	c.C942A	p.H314Q	GKAFSLSSSLTYHKRIHTGEKPYTC	GKAFSLSSSLTYQKRIHTGEKPYTC
16	ZNF679	nonsynonymous SNV	c.G1181C	p.S394T	CEECDKAFKWSSTLANHKSMTGEK	CEECDKAFKWSSTLANHKSMTGEK
16	ZNF700	nonsynonymous SNV	c.G1305C	p.Q435H	CKQCGKAFRSASQLRVHGGTHTGEK	CKQCGKAFRSASHLRVHGGTHTGEK
16	ZNF708	nonsynonymous SNV	c.G722T	p.R241I	CGKAFNQSSTLTRHKIHTGEKLYK	CGKAFNQSSTLTIHKIHTGEKLYK
16	ZNF709	nonsynonymous SNV	c.T1460G	p.F487C	PYECKQCGKAFSFSFRMHERTHT	PYECKQCGKAFSCSSFRMHERTHT
16	ZNF728	nonsynonymous SNV	c.G1145C	p.S382T	CEECGKAFSWPSSLTEHKRIHAGDK	CEECGKAFSWPSSLTEHKRIHAGDK
16	ZNF728	nonsynonymous SNV	c.C963A	p.F321L	EKPYKCEECGKAFNRSSNLMEHKRI	EKPYKCEECGKALNRSSNLMEHKRI
16	ZNF729	nonsynonymous SNV	c.G2471T	p.C824F	TVHKVIHTGEKPKCEECGKAFKHF	TVHKVIHTGEKPKCEECGKAFKHF
16	ZNF729	nonsynonymous SNV	c.G3082A	p.A1028T	REKLYKCEECVKAFNFSALMKHKI	REKLYKCEECVKTFNFSALMKHKI
17	ZNF787	nonsynonymous SNV	c.C926G	p.A309G	AHQRAQHGDGLGAAGGEEPAHICVE	AHQRAQHGDGLGGAGGEEPAHICVE
17	ZNF799	nonsynonymous SNV	c.C1845A	p.H615Q	LHRHKKTHWKKTHTGENPYGCKEKG	LHRHKKTHWKKTHTGENPYGCKEKG
17	ZNF813	nonsynonymous SNV	c.C1684A	p.Q562K	PYKNECGKVFNQKAHLARHRLHT	PYKNECGKVFNKAHLARHRLHT
17	ZNF841	nonsynonymous SNV	c.C1364A	p.T455K	QLVRHQIHTGETPYKNECGKVFF	QLVRHQIHTGETPYKNECGKVFF



17	ZNF92	nonsynonymous SNV	c.A635T	p.E212V	SSTLTKHKRIHTEEKPYKCEECGKA	SSTLTKHKRIHTVEKPYKCEECGKA
17	ZNF93	nonsynonymous SNV	c.C886A	p.Q296K	PYKCEECGKAFNQSSTLTKHKKIHT	PYKCEECGKAFNKSSTLTKHKKIHT
17	ZNF93	nonsynonymous SNV	c.A1502T	p.K501I	AFNQSSSLTKHKKIHTGEKPYKCEE	AFNQSSSLTKHKIIHTGEKPYKCEE
17	ATP5F1	frameshift insertion	c.56_57insGG	p.A19fs	SVPAGSGSELRNAGQGQWGQLGDATQ	SVPAGSGSELRNAGGRAGDSSVTRPRTQTGRRPETPLRQQTP RGAPLSLRPSPEGTATLLLARIASMAPSCFPAVALMR

## Patient #4

TMG	Gene name	Mutation type	cDNA change	Amino acid change	Wild type minigene sequence	Mutated minigene sequence
1	ABCA1	frameshift insertion	c.6596_6597insC	p.K2199fs	LARIFSILSQSKRHLHIEDYSVSQT	LARIFSILSQSKNATPHRRLLCFSDNT
1	ABTB2	nonsynonymous SNV	c.G1213C	p.D405H	CPQMESMENPNLDPPRMTLNNERPF	CPQMESMENPNLHPPRMTLNNERPF
1	AFF4	nonsynonymous SNV	c.G3322A	p.E1108K	YVQVTSNFLYATEIWDQAEQLSKEQ	YVQVTSNFLYATKIWDQAEQLSKEQ
1	AKT1S1	nonsynonymous SNV	c.A496C	p.T166P	GSLSEETPAGPPTCSVPPASALPTQ	GSLSEETPAGPPCSVPPASALPTQ
1	ANKRD36	nonsynonymous SNV	c.C817G	p.P273A	PINSNPVSSQKQPALKATSGKEDSI	PINSNPVSSQQAALKATSGKEDSI
1	ANKRD36	nonsynonymous SNV	c.C47G	p.T16S	ILKNAASVFSETTSLEVSSQKQPAL	ILKNAASVFSETSSLEVSSQKQPAL
1	ANKRD36	nonsynonymous SNV	c.T1320G	p.N440K	TNRRTISQQSAENLDAACGIDKTEN	TNRRTISQQSAEKLDAACGIDKTEN
1	ASIC4	nonsynonymous SNV	c.G556A	p.G186S	STSTLHGLGRACGPGPHGLRRTLWA	STSTLHGLGRACSPGPHGLRRTLWA
1	ATF4	nonsynonymous SNV	c.A450C	p.K150N	VNPIGHLPESLTKPDQVAPFTFLQP	VNPIGHLPESLTNPQVAPFTFLQP
1	BLOC1S3	nonsynonymous SNV	c.G577A	p.E193K	RLLPDIRGVPGTEPEKDPGPA	RLLPDIRGVPGTKPEKDPGPA
1	C17orf53	nonsynonymous SNV	c.A1159C	p.T387P	LVTAASRTPQQPTHSTRAKTRRFP	LVTAASRTPQQPPHSTRAKTRRFP
1	CACNA1A	frameshift deletion	c.3984delT	p.F1328fs	LDFIVVSGALVAFAGTGNKSGKDIN	LDFIVVSGALVALPSLAIKEKTSTRLNPSSESSGCYDLLK PSSGCQSSRLCLTVW
2	CCNB3	nonsynonymous SNV	c.T2622A	p.F874L	LQEKPSIEQEALFKRHSALWEKPST	LQEKPSIEQEALLKRHSALWEKPST
2	CDK5R1	nonsynonymous SNV	c.G3A	p.M1I	MGTVLSLSPSYRK	IGTVLSLSPSYRK
2	CEP131	nonsynonymous SNV	c.C1670G	p.P557R	DSQQEGWVPEAGPGPLELGSEVSTS	DSQQEGWVPEAGRGPLELGSEVSTS
2	CHD2	nonsynonymous SNV	c.G4164T	p.M1388I	GEVKDDGLEKSPMKKKQKKKENKEN	GEVKDDGLEKSPIKKKQKKKENKEN
2	CHKA	nonsynonymous SNV	c.G94C	p.G32R	LSCGSGSAAPAGVGGQRDAASDLE	LSCGSGSAAPAPRVGGQRDAASDLE
2	CNTNAP3B	nonsynonymous SNV	c.G841C	p.D281H	DQHWHSVLIELLDTQVNFVVDKHTH	DQHWHSVLIELLHTQVNFVVDKHTH
2	COL6A3	nonsynonymous SNV	c.C2588T	p.P863L	FVSRIVRRLNIGPSKVRVGVVQFSN	FVSRIVRRLNIGLSKVRVGVVQFSN
2	CORO6	nonsynonymous SNV	c.G883A	p.E295K	LCGKGDSSIRYFEITDEPPFVHYLN	LCGKGDSSIRYFKITDEPPFVHYLN
2	DAXX	nonsynonymous SNV	c.T73A	p.S25T	DDEDEAAAQPGPSHPLPNAASPGAE	DDEDEAAAQGPPTHPLPNAASPGAE
2	DDX11L1	nonsynonymous SNV	c.G181A	p.E61K	SVPSVAESLHHPEITFLTAFLCLPSF	SVPSVAESLHHPKITFLTAFLCLPSF
2	DHX33	nonsynonymous SNV	c.C176T	p.S59L	GGRRQQPPLAQPSASPYPEAVELQR	GGRRQQPPLAQPLASPYPEAVELQR
2	DNAJC6	nonsynonymous SNV	c.C140T	p.A47V	VGGKQRVNAGAAARSPARQPPDRAS	VGGKQRVNAGAAVRSPARQPPDRAS
3	DPCR1	nonsynonymous SNV	c.C1127A	p.S376Y	HGERTANENTTPSPAEPTEHGERTA	HGERTANENTTPYPAEPTEHGERTA
3	DVL2	nonsynonymous SNV	c.C275T	p.S92L	CFNGRVVSWLVSSDNPQPEMAPPVH	CFNGRVVSWLVSLDNPQPEMAPPVH

3	EFCAB4A	nonsynonymous SNV	c.G440C	p.R147P	PPSQVGLGPRPSRQAQSHLAGLPGS	PPSQVGLGPRPSPQAQSHLAGLPGS
3	ELF2	nonsynonymous SNV	c.G1315A	p.A439T	QASGQTPPRVISAVIKGPEVKSEAV	QASGQTPPRVISTVIKGPVKSEAV
3	SPSB2	nonsynonymous SNV	c.C409G	p.R137G	GSNSESWGWDIGRGLYHQSKGPGA	GSNSESWGWDIGGGKLYHQSKGPGA
3	ZNF614	nonsynonymous SNV	c.C1024T	p.P342S	LIVHQRTHTEKPYICSECGKGFTM	LIVHQRTHTEKSYICSECGKGFTM
3	TATDN1	frameshift deletion	c.168delA	p.K56fs	TNGILIFLLPKKK	TNGILIFLLPKKN
3	ATXN2L	nonsynonymous SNV	c.A37C	p.S13R	MAFAAALSPSPASGPLFALNRRLHQ	MAFAAALSPSPARGPLFALNRRLHQ
3	FAM9C	nonsynonymous SNV	c.G298A	p.D100N	YNPYSRLYELVDDKTVLNAFFSYIA	YNPYSRLYELVDNKTVLNAFFSYIA
3	ZNF724	nonsynonymous SNV	c.C766A	p.R256S	HKIIHTGEKSYKREECGKAFNISSH	HKIIHTGEKSYKSEECGKAFNISSH
3	ZNF676	nonsynonymous SNV	c.G1730C	p.S577T	ECGKAFKSSSTVSYHKKIHTGENP	ECGKAFKSSSTVTYHKKIHTGENP
3	RPS4X	nonsynonymous SNV	c.G349A	p.E117K	TKGRFAVHRITPEEAKPC	TKGRFAVHRITPKEAKPC
4	PRR31	nonsynonymous SNV	c.A500T	p.E167V	RGHAGPPEGWAGEGGVSPGLTPLLG	RGHAGPPEGWAGVGGVSPGLTPLLG
4	ZNF99	nonsynonymous SNV	c.G2537T	p.R846M	ECGKAFFNSALRKHKIIHTGEKPY	ECGKAFFNSALMKHKIIHTGEKPY
4	ERC2	nonsynonymous SNV	c.G295A	p.G99R	NRAVYGGRTAMGSSPNIASAGLSH	NRAVYGGRTAMRSSPNIASAGLSH
4	ERCC2	nonsynonymous SNV	c.G175C	p.V59L	IYCSRTVPEIEKVIEELRKLNFYE	IYCSRTVPEIEKLIEELRKLNFYE
4	ERICH1	nonsynonymous SNV	c.G851T	p.R284L	EDGVDTIEEDLTRAGEEDGKDTREE	EDGVDTIEEDLTRAGEEDGKDTREE
4	FAM120B	nonsynonymous SNV	c.C1405A	p.P469T	PRQEVPMYTGSEPRQEVPMYGPES	PRQEVPMYTGSETRQEVPMYGPES
4	FAM194B	nonframeshift deletion	c.404_421del	p.135_141del	EEEEYLGKEEHLKEEYLGKEGYLE	EEEEYLGKEEHLGKEGYLEKEDYIE
4	FAM194B	nonsynonymous SNV	c.G260T	p.G87V	GKEEYLKEEYLGKEEHLKEEYLE	GKEEYLKEEYLVKEEHLKEEYLE
4	FEV	nonsynonymous SNV	c.A607C	p.T203P	GFSYWPGPGAATAAAATAALYSPSP	GFSYWPGPGAAPAAAAATAALYSPSP
4	FKBP11	nonsynonymous SNV	c.T33A	p.H11Q	MTLRPSLLPLHLLLLLLLLSAAVC	MTLRPSLLPLQLLLLLLLLLSAAVC
4	GP6	nonsynonymous SNV	c.T1820C	p.V607A	HCTLARVTKPDFVSKNTGMNMSITL	HCTLARVTKPDFASKNTGMNMSITL
4	GSDMD	nonsynonymous SNV	c.A446C	p.H149P	RWARRGQGRAGPHLPQATLLFIYL	RWARRGQGRAGPPLPQATLLFIYL
5	HAND2	nonsynonymous SNV	c.G262C	p.G88R	DHSHYGGVPPGAGPPGLGPRPVKR	DHSHYGGVPPGARPPGLGPRPVKR
5	HSD17B6	nonsynonymous SNV	c.T317A	p.L106H	TQWVKEHVGDRLWGLVNNAGILTP	TQWVKEHVGDGRHWGLVNNAGILTP
5	HSF4	nonsynonymous SNV	c.G125C	p.S42T	DPGTDHLIRWSPSGTSFLVSDQSRF	DPGTDHLIRWSPGTGTSFLVSDQSRF
5	JAG1	nonsynonymous SNV	c.A556C	p.T186P	TGVAHFYQIRVTCDDYYYGFGCNK	TGVAHFYQIRVPCDDYYYGFGCNK
5	KANSL1	frameshift insertion	c.2109dupA	p.S704fs	KPFDKIKPPKLSLKHRAPMPGSLP	KPFDKIKPPKLLIA
5	KIAA1244	nonsynonymous SNV	c.C6525G	p.I2175M	REWLGRVGRVYDIIV	REWLGRVGRVYDMIV

5	KRT2	nonsynonymous SNV	c.A1789T	p.S597C	SGGGYSGGGKHS SGGGSRGGSSSG	SGGGYSGGGKHCSGGGSRGGSSSG
5	KRTAP1-3	nonsynonymous SNV	c.C121G	p.Q41E	CCETSCCQPSCCQTSFCGFPSFSTS	CCETSCCQPSCCETSFCGFPSFSTS
5	LOC407835	nonframeshift deletion	c.200_214del	p.67_72del	AFLTQKAKV GELKDDDFERTSELDA	AFLTQKAKV GELKRTSELDA GNGGV
5	LOC642846	nonsynonymous SNV	c.G344A	p.S115N	QLINDRCVDMQRSRHEK KGAEEK	QLINDRCVDMQRNRHEK KGAEEK
5	LOC642846	nonsynonymous SNV	c.G344A	p.S115N	QLINDRCVDMQRSRHGSHRDHGVA A	QLINDRCVDMQRNRHGSHRDHGVA A
5	LOC642846	nonsynonymous SNV	c.G794A	p.S265N	QLINDRCVDMQRSRHVA VFSVFLC	QLINDRCVDMQRNRHVA VFSVFLC
6	LPA	nonsynonymous SNV	c.G65A	p.S22N	LLLFLKSAAPEQSHVVQDCYHGDGQ	LLLFLKSAAPEQNHVVQDCYHGDGQ
6	MAGEC1	nonsynonymous SNV	c.T617C	p.F206S	PQSPLQIPVSRFSSTLLSIFQSSP	PQSPLQIPVSRSSSTLLSIFQSSP
6	MAGEC1	nonsynonymous SNV	c.A2376T	p.Q792H	SSFFSYTLASLLQSSHESQPSPPEG	SSFFSYTLASLLHSSHESQPSPPEG
6	MAML2	nonsynonymous SNV	c.G199A	p.D67N	EGRYERGRAESSDRERESTLQLLSL	EGRYERGRAESSNRERESTLQLLSL
6	MAP6	nonsynonymous SNV	c.C394T	p.R132W	VMRQDYRAWK VQRPEPSCRPRSEYQ	VMRQDYRAWK VQWPEPSCRPRSEYQ
6	MEMO1	nonsynonymous SNV	c.T446C	p.L149P	ALSESKEQEFGLFSKY LADPSNLF	ALSESKEQEFGKPF SKYLADPSNLF
6	MUC12	nonsynonymous SNV	c.C10193A	p.S3398Y	PGSTATTHFPDSSTTSGRSEESTAS	PGSTATTHFPDSYTTSGRSEESTAS
6	MUC12	nonsynonymous SNV	c.T10404A	p.S3468R	TPGLEKSTTFHSSPRSPATLSPA	TPGLEKSTTFHRSRSPATLSPA
6	MUC16	nonsynonymous SNV	c.A33440T	p.E11147V	VEASSAVLTVSPEVPGMVTSLVTSS	VEASSAVLTVSPVPGMVTSLVTSS
6	MUC22	nonsynonymous SNV	c.G4273A	p.A1425T	SEATTSSAAGSEATTTSTEGSETTT	SEATTSSAAGSETTTTSTEGSETTT
6	MYO3B	nonsynonymous SNV	c.G283A	p.D95N	NVVKFYGMFYKADHC VGGQLWLVL E	NVVKFYGMFYKANHC VGGQLWLVL E
6	NFATC2IP	nonsynonymous SNV	c.T773A	p.L258H	QEDEVVLVEGPTLPETPRLFPLKIR	QEDEVVLVEGPTHETPRLFPLKIR
7	NOS3	nonsynonymous SNV	c.G346C	p.G116R	VFPRKLQGRPSGPPAPEQLLSQAR	VFPRKLQGRPSRPPAPEQLLSQAR
7	NSUN5P2	nonsynonymous SNV	c.G140C	p.W47S	AMLLDPRQAPMSWMPVPPQAIKTSH	AMLLDPRQAPMSSMPVPPQAIKTSH
7	NUP93	nonsynonymous SNV	c.G40A	p.E14K	DTEGFGELLQAEQLAAETEGISEL	DTEGFGELLQAKQLAAETEGISEL
7	OSBPL1A	nonsynonymous SNV	c.A1142C	p.K381T	ENGEIDQASEEKKRLEEKQRAARKN	ENGEIDQASEEKTRLEEKQRAARKN
7	PAX7	nonsynonymous SNV	c.A883C	p.T295P	AFNHLLPGGFPPGMPPLPPYQLPD	AFNHLLPGGFPPGMPPLPPYQLPD
7	PITPNM1	nonsynonymous SNV	c.A2441C	p.Q814P	WKGSELATDPPAQAAPSTTSEVVK	WKGSELATDPPAPPAAPSTTSEVVK
7	PPP1R10	nonsynonymous SNV	c.A2524T	p.S842C	RPHEGPGGSMGGSGGHRPHEGPGHG	RPHEGPGGSMGGCGGHRPHEGPGHG
7	PRDM9	nonsynonymous SNV	c.T2379A	p.S793R	CGRGFRDKSNLLSHQRTHTEKPYV	CGRGFRDKSNLLRHQRTHTEKPYV
7	PRRC2A	nonsynonymous SNV	c.C3833T	p.T1278I	NAARGSEGKPSLTPASAPGPEEAL	NAARGSEGKPSLILPASAPGPEEAL
7	PRSS3	nonsynonymous SNV	c.G125T	p.G42V	YQVSLNSGSHFCGGLISEQWVVSA	YQVSLNSGSHFCVGLISEQWVVSA

7	PRSS3	nonsynonymous SNV	c.T137A	p.I46N	LNSGSHFCGGSLISEQWVVSAAHCY	LNSGSHFCGGSLNSEQWVVSAAHCY
7	PRSS3	nonsynonymous SNV	c.C141G	p.S47R	NSGSHFCGGSLISEQWVVSAAHCYK	NSGSHFCGGSLIREQWVVSAAHCYK
8	PRSS37	nonsynonymous SNV	c.G449A	p.R150Q	WSQENSGRHPDLRQNLEAPVMSDRE	WSQENSGRHPDLQQNLEAPVMSDRE
8	PRSS37	nonsynonymous SNV	c.G446A	p.R149Q	DWSQENSRHPDLRQNLEAPVMSDRE	DWSQENSRHPDLQQNLEAPVMSDRE
8	PRSS37	nonsynonymous SNV	c.G410A	p.R137Q	HNSHEQGRHPDLRQNLEAPVMSDRE	HNSHEQGRHPDLQQNLEAPVMSDRE
8	RBMXL3	nonsynonymous SNV	c.G2102A	p.R701H	YSGGHDSSSQSNRYGGGGRYEYERG	YSGGHDSSSQSNHYGGGGRYEYERG
8	RILPL1	nonsynonymous SNV	c.A88T	p.I30F	NVAELTVMDVYDIASLVGHEFERVI	NVAELTVMDVYDFASLVGHEFERVI
8	RP1L1	nonsynonymous SNV	c.C5726A	p.P1909Q	WEVQPESEGAEAPEAEKEAQPETES	WEVQPESEGAEAEAEKEAQPETES
8	SAMD1	nonsynonymous SNV	c.G61C	p.A21P	PPETAAAATTAASSSAASPHYQE	PPETAAAATTAAPASSAASPHYQE
8	SH3GL1P1	nonsynonymous SNV	c.G527C	p.R176P	MPLPSDSLVPPrPSIHTGWHPRQV	MPLPSDSLVPPrPSIHTGWHPRQV
8	SLC25A5	nonsynonymous SNV	c.T811C	p.F271L	RKIARDEGGKAFKGAWSNVLRGMG	RKIARDEGGKAFKGAWSNVLRGMG
8	SLC9A8	nonsynonymous SNV	c.G1783A	p.E595K	RQGPGSEDDEQELL	RQGPGSEDDEQKLL
9	SOGA1	nonsynonymous SNV	c.G4418T	p.W1473L	WNLHQGKQNGSAWARSTTRDSPVL	WNLHQGKQNGSALARSTTRDSPVL
9	SON	nonsynonymous SNV	c.G2232T	p.Q744H	DSQMLASNTMDSQMLASNTMDSQML	DSQMLASNTMDSHMLASNTMDSQML
9	SOX5	nonsynonymous SNV	c.C703T	p.P235S	QARLSKQHLEKYPDYKYKPRPKRTC	QARLSKQHLEKYSYDYKYKPRPKRTC
9	SPEN	nonsynonymous SNV	c.T10105A	p.C3369S	VQSTQPAQPAPPCPPSQLGQPGQPP	VQSTQPAQPAPPPPSQLGQPGQPP
9	SPEN	nonsynonymous SNV	c.T10114A	p.S3372T	TQPAQPAPPCPPSQLGQPGQPPSSK	TQPAQPAPPCPPTQLGQPGQPPSSK
9	SRSF9	nonsynonymous SNV	c.T286G	p.W96G	VEFPRTYGGRGGWPRGGRNGPPTRR	VEFPRTYGGGGPRGGRNGPPTRR
9	TCEB3CL	nonsynonymous SNV	c.G1495A	p.G499S	AGSSQAPSLGDGDGGSVSGGGSSN	AGSSQAPSLGDSGDGGSVSGGGSSN
9	TCHH	nonsynonymous SNV	c.G3193A	p.G1065R	EEELQQEEEQLLGEERTRRRQELE	EEELQQEEEQLLREERTRRRQELE
9	TCHH	nonsynonymous SNV	c.C2933G	p.P978R	LQQKEEQLLGEPEKRRRQEREKKY	LQQKEEQLLGEEREKRRRQEREKKY
9	THSD7B	nonsynonymous SNV	c.G770T	p.C257F	EACIVEGELLQQCPRYSWRTSEWKE	EACIVEGELLQQFPRYSWRTSEWKE
9	TMEM131	nonsynonymous SNV	c.T919A	p.F307I	IRIKTNASDSTEIILPVEVEVTTA	IRIKTNASDSTEIILPVEVEVTTA
9	TMEM251	nonsynonymous SNV	c.G34A	p.D12N	MLAFSEMPKPPDYSELSDSLTLAV	MLAFSEMPKPPNYSELSDSLTLAV
10	TNS3	nonsynonymous SNV	c.G1570A	p.G524S	CHKSSQNSLLSDGFGSNVGEDPQGT	CHKSSQNSLLSDSFGSNVGEDPQGT
10	TREX2	nonsynonymous SNV	c.T452A	p.L151Q	PRDTVCLDTLPALRGLDRAHSHGTR	PRDTVCLDTLPAQRGLDRAHSHGTR
10	TTN	nonsynonymous SNV	c.G38836T	p.V12946L	APKEVVPEKKVPVTPPKKPEVPPVK	APKEVVPEKKVPLTPPKKPEVPPVK
10	TUBBP5	nonsynonymous SNV	c.A421T	p.T141S	PDNFIFGELRARTGVRLLSQGSSKS	PDNFIFGELRARGVRLLSQGSSKS

10	TUBGCP3	nonframeshift deletion	c.1241_1255del	p.414_419del	TRDFPTRDFPTRDFPMPHVFPTRVFP	TRDFPTRDFPTRVFPTRVFPTRVWH
10	UBXN4	nonsynonymous SNV	c.G1504A	p.G502R	QDDGEDENNTWNGNSTQQM	QDDGEDENNTWNRNSTQQM
10	USP48	frameshift deletion	c.1975delT	p.S659fs	EDILCPHAVFFFSKYIFLNSGELCI	EDILCPHAVFFFPNIFF
10	WEE1	nonsynonymous SNV	c.A253C	p.S85R	PERRRSPGPAPGSPGELEEDLLPLG	PERRRSPGPAPGRPGELEEDLLPLG
10	ZCCHC6	nonsynonymous SNV	c.A421G	p.R141G	RKDSFQENEDGYRWQDTRGCRTVRR	RKDSFQENEDGYGWQDTRGCRTVRR
10	ZFP62	nonsynonymous SNV	c.A2328T	p.K776N	KAFRNSSGLTVHKRIHTGEKPYECD	KAFRNSSGLTVHNRIHTGEKPYECD
10	ZMYND15	nonsynonymous SNV	c.A1883C	p.Q628P	MAVATGGGTSPQPNNPFRSPFRLRA	MAVATGGGTSPPPNPFRSPFRLRA
10	ZNF100	nonsynonymous SNV	c.G1262T	p.R421I	GFNWSSALTKHKRIHTGEKPYKCEE	GFNWSSALTKHKIHTGEKPYKCEE
11	ZNF112	nonsynonymous SNV	c.C1849T	p.H617Y	CEECGKGFSSSHLQGHQRVHTGEK	CEECGKGFSSSYLQGHQRVHTGEK
11	ZNF112	nonsynonymous SNV	c.G1859C	p.G620A	CGKGFSSSHLQGHQRVHTGEKPFK	CGKGFSSSHLQAHQRVHTGEKPFK
11	ZNF208	nonsynonymous SNV	c.G1323T	p.M441I	ECGKAFNWSSNLMEHKKIHTGETPY	ECGKAFNWSSNLIEHKKIHTGETPY
11	ZNF25	nonsynonymous SNV	c.C983A	p.A328D	CKECRKCFYQKSALTVHQRTHHTGEK	CKECRKCFYQKSDLTVHQRTHHTGEK
11	ZNF257	nonsynonymous SNV	c.C1417A	p.H473N	CEECGKAFNQSSHLTQHKKIHTGEK	CEECGKAFNQSSNLTQHKKIHTGEK
11	ZNF257	nonsynonymous SNV	c.C1081A	p.Q361K	CGKAFNRSSHLTQHKKIHTKEKPYK	CGKAFNRSSHLTKHKIHTKEKPYK
11	ZNF257	nonsynonymous SNV	c.A821T	p.Q274L	CGKAFNRSSHITLHKRIHNREKPFK	CGKAFNRSSHITLHKRIHNREKPFK
11	ZNF30	nonsynonymous SNV	c.G1296C	p.K432N	QRIHTGEKPYECKEKGKAFISRHQL	QRIHTGEKPYECNECGKAFISRHQL
11	ZNF30	nonsynonymous SNV	c.G1297A	p.E433K	RIHTGEKPYECKEKGKAFISRHQLT	RIHTGEKPYECKKCGKAFISRHQLT
11	ZNF37A	nonsynonymous SNV	c.T1373A	p.L458Q	KFFCYYSGFTEHLRRHTGEKPFGCN	KFFCYYSGFTEHQRRHTGEKPFGCN
11	ZNF431	nonsynonymous SNV	c.G810C	p.K270N	KPFKCEECGKAFKQSSLTTHKIIH	KPFKCEECGKAFNQSSLTTHKIIH
11	ZNF469	nonsynonymous SNV	c.A604C	p.T202P	SFTSTNYTSPSATPRPPAPGPPQSR	SFTSTNYTSPSAPRPPAPGPPQSR
12	ZNF519	nonsynonymous SNV	c.A1353T	p.Q451H	KAFNRGSHLTRHQRIHTGEKSFKCE	KAFNRGSHLTRHHRIHTGEKSFKCE
12	ZNF595	nonsynonymous SNV	c.C101G	p.T34R	KPYKCEECGKAFRRSTTLNEHKKIH	KPYKCEECGKAFRRSTTLNEHKKIH
12	ZNF626	nonsynonymous SNV	c.T914A	p.I305K	AFNRSSTLTTHKKIHTGEKPYKCEE	AFNRSSTLTTHKKIHTGEKPYKCEE
12	ZNF678	nonsynonymous SNV	c.C1129A	p.Q377K	RHKRIHTGEKPYQCEECGKTFNRCS	RHKRIHTGEKPYKCEECGKTFNRCS
12	ZNF678	nonsynonymous SNV	c.G1352T	p.R451M	KAFNKFSSLTQHRRHTGVKPYKCE	KAFNKFSSLTQHMRHTGVKPYKCE
13	ZNF695	nonsynonymous SNV	c.G1112T	p.R371M	KAFNQSSHLTEHRRHTGEKPYKCE	KAFNQSSHLTEHMRIHTGEKPYKCE
13	ZNF721	nonsynonymous SNV	c.G1849A	p.E617K	KIHTGEKLYKCEECGKDFVWYTDLN	KIHTGEKLYKCEKCGKDFVWYTDLN
13	ZNF735	nonsynonymous SNV	c.A893T	p.K298I	NHKRIHTGERPYKCEECGKAHSVSS	NHKRIHTGERPYICEECGKAHSVSS

13	ZNF790	nonsynonymous SNV	c.C985A	p.Q329K	KAFSQRSHLIKHQRIHTGEKPYECK	KAFSQRSHLIHKRIHTGEKPYECK
13	ZNF813	nonsynonymous SNV	c.A1048T	p.T350S	QTSSLTCHRRRLHTGEKPFKCNECGK	QTSSLTCHRRRLHSGEKPFKCNECGK
13	ZNF83	nonsynonymous SNV	c.G1210T	p.D404Y	HRIHTGEKPYKCDECGKVFSQNSYL	HRIHTGEKPYKCYECGKVFSQNSYL
13	ZNF85	nonsynonymous SNV	c.T1313A	p.I438K	GFKWPSTLTIHKIIHTGEKPYKCEE	GFKWPSTLTIHKKIHTGEKPYKCEE
13	ZNF850	nonsynonymous SNV	c.C841G	p.Q281E	QIHTGEKPYHCKQCGKSFTVGSTLI	QIHTGEKPYHCKEKGKSFTVGSTLI
13	ZNF93	nonsynonymous SNV	c.G1560T	p.Q520H	PYKCEECGKAFNQSSSLIKHKKIHT	PYKCEECGKAFNHSSTLIKHKKIHT
13	ZNF99	nonsynonymous SNV	c.G2248T	p.V750L	CGKAFKWSSKLTVHKVIHTAEKPCK	CGKAFKWSSKLTLLHKVIHTAEKPCK
13	ZNF99	nonsynonymous SNV	c.G1526T	p.C509F	TVHKVIHMEEKPCKCEECGKAFKHF	TVHKVIHMEEKPFKCEECGKAFKHF

## Patient #5

TMG	Gene name	Mutation type	cDNA change	Amino acid change	Wild type minigene sequence	Mutated minigene sequence
1	ABHD17A	nonsynonymous SNV	c.T196A	p.W66R	GTLRASSGAPGRWKLHLTERADFQY	GTLRASSGAPGRRKLHLTERADFQY
1	ABHD17A	nonsynonymous SNV	c.T113G	p.V38G	LAFLPPEATYSLVPEPEPGGGAGA	LAFLPPEATYSLGPEPEPGGGAGA
1	ACOT4	nonsynonymous SNV	c.G22A	p.E8K	MSATLILEPPGRCCWNEPVRIAVRG	MSATLILKPPGRCCWNEPVRIAVRG
1	AHNAK2	nonsynonymous SNV	c.C12629G	p.A4210G	DVKLPEGPLPKGAGLKGHLPKVQMP	DVKLPEGPLPKGGGLKGHLPKVQMP
1	AHNAK2	nonsynonymous SNV	c.C12022G	p.Q4008E	PKVEADVSLPSMQGDLKATDLSVQP	PKVEADVSLPSMEGDLKATDLSVQP
1	AHNAK2	nonsynonymous SNV	c.A9817G	p.S3273G	DVTAPDVEVSQPSMEVDVEAPGAKL	DVTAPDVEVSQPGMEVDVEAPGAKL
1	APOE	nonsynonymous SNV	c.C359A	p.A120D	RARLSKELQAAQARLGADMEDVCGR	RARLSKELQAAQDRLGADMEDVCGR
1	AQP1	nonsynonymous SNV	c.A161T	p.N54I	RLFDLYYYDGLANQQEQIRLTIDTT	RLFDLYYYDGLAIQEQEIRLTIDTT
1	AQP7	nonsynonymous SNV	c.G167A	p.R56H	SVADRDAVAVSLRHHGPGEQPSTAR	SVADRDAVAVSLHHHGPGEQPSTAR
1	AQP7	nonsynonymous SNV	c.G286A	p.A96T	AWLTGMLQLCLFAITDQENNPALPG	AWLTGMLQLCLFTITDQENNPALPG
1	ARHGAP2 3	nonsynonymous SNV	c.T4453C	p.S1485P	SLQSQPPIRRAASRLHQCL	SLQSQPPIRRAAPRLHQCL
1	ATF5	nonsynonymous SNV	c.T263C	p.L88P	LLPLEPPLPPGTLQPSPTPPDLEA	LLPLEPPLPPGTPPQPSPTPPDLEA
2	BAGE3	nonsynonymous SNV	c.G305A	p.C102Y	LLKDRRKMQTVQCATARETS	LLKDRRKMQTVQYATARETS
2	BANP	nonsynonymous SNV	c.G688A	p.E230K	EEDYPNGTWLGDENNPENRVRCAII	EEDYPNGTWLGDKNNPENRVRCAII
2	BC068290	nonsynonymous SNV	c.T611C	p.L204P	THVCLSRVPRRSLDPKKGFPDTRT	THVCLSRVPRRSPDPKKGFPDTRT
2	BRD8	nonsynonymous SNV	c.C458T	p.S153F	PRLVNESEMAMAVAFGHLNSTGVLLEV	PRLVNESEMAMAVAFGHLNSTGVLLEV
2	BRD8	nonsynonymous SNV	c.C548T	p.S183F	TSGVNESEMAMAVAFGHLNSTGVLLEV	TSGVNESEMAMAVAFGHLNSTGVLLEV
2	C17orf64	nonsynonymous SNV	c.C36A	p.D12E	MEASDGQGGEGDKPLEQVTNVSCLE	MEASDGQGGEGEKPLEQVTNVSCL
2	C2orf72	nonsynonymous SNV	c.C173T	p.P58L	RALLRDFARAVFPPEPGAAPGGAA	RALLRDFARAVFLPEPGAAPGGAA
2	CABP2	nonsynonymous SNV	c.T73C	p.S25P	PGAGALRTPCSGAPHQGAPAPAPA	PGAGALRTPCSGAPHQGAPAPAPA
2	CABP2	nonsynonymous SNV	c.T59C	p.L20P	PWRRGPKDPLQWLGSPPRGSCSPS	PWRRGPKDPLQWPGSPPRGSCSPS
2	CD209	nonsynonymous SNV	c.A731G	p.Q244R	KSKQQEIQELTQLKAAVERLCHPC	KSKQQEIQELTRLKAAVERLCHPC
2	CDRT1	nonsynonymous SNV	c.G334A	p.V112M	QGGNASLTECAHVRLHIAGHLPASR	QGGNASLTECAHMLRHIAGHLPASR
3	CHCHD10	nonsynonymous SNV	c.C89T	p.S30L	APSAHPPAHPPPSAAAPAPAPSGQP	APSAHPPAHPPPLAAAPAPAPSGQP
3	CLCF1	nonsynonymous SNV	c.C230A	p.A77D	NPPRLGAETLPRATVDLEVWRSND	NPPRLGAETLPRDTVDLEVWRSND
3	CLCN5	nonsynonymous SNV	c.T187A	p.L63I	REVGHHIIGDDNLS	REVGHHIIGDDNIS



3	CNKSR2	nonsynonymous SNV	c.T197A	p.I66N	DLGVSRIHQELILEAVDLLCALNY	DLGVSRIHQELNLEAVDLLCALNY
3	CR1	nonsynonymous SNV	c.C3275A	p.T1092N	TNRENFHYGSSVVTYRCNLGSRGRKV	TNRENFHYGSSVNYRCNLGSRGRKV
3	DCAF15	nonsynonymous SNV	c.C5A	p.A2E	MAPSSKSERNMSGAGSGGGGPGGAGG	MEPSSKSERNMSGAGSGGGGPGGAGG
3	EAF1	nonsynonymous SNV	c.C83A	p.A28D	LRLGESFEKRPRASFHTIRYDFKPA	LRLGESFEKRPRDSFHTIRYDFKPA
3	EFCAB6	nonsynonymous SNV	c.G680A	p.R227K	GLQVSSKGPLTKRNSINSRNESHKE	GLQVSSKGPLTKKNSINSRNESHKE
3	EIF2B5	nonsynonymous SNV	c.C782A	p.T261K	ISICSPQVAQLFTDNFDYQTRDDFV	ISICSPQVAQLFKDNFDYQTRDDFV
3	ENSA	nonsynonymous SNV	c.A335G	p.K112R	MKNKQLPSAGPDKNLVTGDHIPTPQ	MKNKQLPSAGPDRNLVTGDHIPTPQ
3	FAM136A	nonsynonymous SNV	c.A470G	p.N157S	FQDRLARCTMHCNDKAKDS	FQDRLARCTMHCSDKAKDS
3	ADAM7	nonsynonymous SNV	c.T2228C	p.L743S	EIHFLNQRTPESLESPTSFSSPHY	EIHFLNQRTPESSESPTSFSSPHY
4	ASCC2	nonsynonymous SNV	c.A606T	p.Q202H	KLQCLRWSKTPSQKKRKKTKPCSVT	KLQCLRWSKTPSHKKRKKTKPCSVT
4	NDRG4	nonsynonymous SNV	c.A59G	p.D20G	RFPEEKPLLRGQDATELEHDIETPY	RFPEEKPLLRGQGATELEHDIETPY
4	UNC93B1	nonsynonymous SNV	c.G1492A	p.V498M	LGSSLHMKAKLAVLLVTLVAAAVSY	LGSSLHMKAKLAMLLVTLVAAAVSY
4	RAB5B	nonsynonymous SNV	c.T50C	p.L17P	GVLKPGNPLPPLPPLQYPPPSTLS	GVLKPGNPLPFPPLPPLQYPPPSTLS
4	FBXO2	nonsynonymous SNV	c.G77A	p.S26N	GAWDTSGLTIKESVQPEEASPEEQ	GAWDTSGLTIKENVQPEEASPEEQ
4	DCAF15	nonsynonymous SNV	c.C2A	p.A1E	APSSKSERNMSGAGSGGGGPGGAGGK	EPSSKSERNMSGAG
4	ATP5A1	nonsynonymous SNV	c.A163G	p.N55D	RALSAGGRGAVANAAILHPWLLRLD	RALSAGGRGAVADAAILHPWLLRLD
4	FAM100A	nonsynonymous SNV	c.T197G	p.V66G	TRAAAGPAGAWRVSRGLSSCRQPSA	TRAAAGPAGAWRGRGLSSCRQPSA
4	SAMD8	nonsynonymous SNV	c.G53T	p.R18L	RHRPRLHSGSPPRAPPPLEALHSG	RHRPRLHSGSPPLAPPPLEALHSG
4	ZNF66	nonsynonymous SNV	c.G1261A	p.G421R	SSPLSKHKRIHTGEKPYKCEECGKA	SSPLSKHKRIHTREKPYKCEECGKA
4	SELENON	nonsynonymous SNV	c.G1148A	p.R383H	WQQELSWEEAARRLEVAMYPFKKVS	WQQELSWEEAARRHLEVAMYPFKKVS
4	C15orf40	nonsynonymous SNV	c.C219A	p.F73L	ELQVHATHTRLIFVFEETGSRHVA	ELQVHATHTRLILVFEETGSRHVA
5	MARCH10	nonsynonymous SNV	c.G391A	p.A131T	ASTSPDPVPESPAVLCYFPISCPTP	ASTSPDPVPESPTVLCYFPISCPTP
5	LRRC75B	nonsynonymous SNV	c.C169A	p.L57M	RRPERARQLLRLLRQDPGDSGPWRE	RRPERARQLLRMRQDPGDSGPWRE
5	CYP4F31P	nonsynonymous SNV	c.A200G	p.D67G	GPLHILGTDGNVDTTGKLGVLVPTPP	GPLHILGTDGNVGTGKLGVLVPTPP
5	PRR33	nonsynonymous SNV	c.A1276C	p.T426P	QGRLAGPSGGEHTPASLTRLPLFLYR	QGRLAGPSGGEHPPASLTRLPLFLYR
5	AC099489	nonsynonymous SNV	c.G1043A	p.R348Q	VTLRPLEEVWRERAEAMRRLQAWV	VTLRPLEEVWREAEAMRRLQAWV
5	ZNF724	nonsynonymous SNV	c.G1105A	p.E369K	KRIHTGEKPYKCEECGKAFNVSSTL	KRIHTGEKPYKCEECGKAFNVSSTL
5	LILRB3	nonsynonymous SNV	c.C560A	p.T187N	RGFQALFPVGPVTPSHS	RGFQALFPVGPVNPVSHS

5	AC092384.1	nonsynonymous SNV	c.C641G	p.A214G	FRICSPVPWTGTATEGQSVGTCLPD	FRICSPVPWTGTGTEGQSVGTCLPD
5	IGHV2-26	nonsynonymous SNV	c.G208T	p.A70S	IRQPPGKALEWLAHIFSNDEKSYST	IRQPPGKALEWLSHIFSNDEKSYST
5	ENSG00000240834	nonsynonymous SNV	c.T328A	p.C110S	SSLQPEDFATYYCQQANSFP	SSLQPEDFATYYSQQANSFP
5	DUX4L4	nonsynonymous SNV	c.C767T	p.A256V	PAPPDGALSHPQAPRWPPHPGKSRE	PAPPDGALSHPQVPRWPPHPGKSRE
5	DUX4L4	nonsynonymous SNV	c.G865C	p.A289P	LPGCAVAQPGPAQAGPQQGVLP	LPGCAVAQPGPPQAGPQQGVLP
6	DUX4L4	nonsynonymous SNV	c.G988A	p.A330T	VAGAAWEPQAGEAPPQPAPPDASA	VAGAAWEPQAGETPPPQPAPPDASA
6	ENSG00000267976	nonsynonymous SNV	c.G113T	p.C38F	FPSSLTSLPPSPSCHPWWPGRPG	FPSSLTSLPPSPSCHPWWPGRPG
6	LRRC2-AS1	nonsynonymous SNV	c.A282T	p.K94N	RKV TARPRMQSRKGTVLSPLCGQIP	RKV TARPRMQSRNGTVLSPLCGQIP
6	EPHB3	nonsynonymous SNV	c.G101A	p.G34D	PLLLLPLLLLPA GCRALETLM DTK	PLLLLPLLLLPA DCRALLETLM DTK
6	ESRRB	nonsynonymous SNV	c.A923T	p.D308V	DDKL VYAEDYIMDEEHSRLAGLLEL	DDKL VYAEDYIMVEEHSRLAGLLEL
6	ETV2	nonsynonymous SNV	c.C272A	p.A91D	SREFQLCDPKEVARLWGERKRKPGM	SREFQLCDPKEVDRLWGERKRKPGM
6	FAM115C	nonsynonymous SNV	c.A214G	p.T72A	LVVVSHEGYLSHTGLAPFLNAVSW	LVVVSHEGYLSHAGLAPFLNAVSW
6	FAM136A	nonsynonymous SNV	c.A260G	p.N87S	FQDRLARCTMHCNDKAKDSIDAGSK	FQDRLARCTMHCSDKAKDSIDAGSK
6	FAM211B	nonsynonymous SNV	c.C169A	p.L57M	RRPERARQLLRLLRQDLGLERTLLP	RRPERARQLLRMLRQDLGLERTLLP
6	FAM47A	nonsynonymous SNV	c.G262A	p.A88T	FLLPKISLRGPQADPKSGQKLLKK	FLLPKISLRGPQTDPKSGQKLLKK
6	FAM9C	nonsynonymous SNV	c.A461G	p.K154R	KEEQIKIFQEQQKRWQQDGKGTED	KEEQIKIFQEQQRRWQQDGKGTED
6	FARP1	nonsynonymous SNV	c.C2275A	p.P759T	LIGIDNLVVPGRPGSFLIRTPHLG	LIGIDNLVVPGRPGSFLIRTPHLG
7	FBXO2	nonsynonymous SNV	c.G26A	p.S9N	MDGDGDPESVGPPEEASPEEQ	MDGDGDPENVGPPEEASPEEQ
7	FRG1B	nonsynonymous SNV	c.G148C c.C149A	p.A50H	EQWEPVFQNGKMALLASNSCFIRC	EQWEPVFQNGKMHLLASNSCFIRC
7	FRG1B	nonsynonymous SNV	c.G4C c.C5A	p.A2H	MALLASNSCFIRCNEAGDIEAKSKT	MHLLASNSCFIRCNEAGDIEAKSKT
7	FUT3	nonsynonymous SNV	c.C974T	p.T325M	HARYLSYFRWRETLRPRFSWALDF	HARYLSYFRWREMLRPRFSWALDF
7	GABRD	nonsynonymous SNV	c.G1063C	p.D355H	AKVKVSRPRAEMDVRNAIVLFLSA	AKVKVSRPRAEMHVRNAIVLFLSA
7	GLCCI1	nonsynonymous SNV	c.A415G	p.S139G	RPRRSPESHRRSSPERRSPGPVC	RPRRSPESHRRSGPERRSPGPVC
7	GNA12	nonsynonymous SNV	c.C67T	p.R23C	CLLPAEAGGARERRAGSGARDAERE	CLLPAEAGGARECRAGSGARDAERE
7	GOLGA6L1	nonsynonymous SNV	c.A1874G	p.Q625R	EKMWEQEEKMCEQEEKMQEQUEKMR	EKMWEQEEKMCEREKMQEQUEKMR
7	GOLGA8DP	nonsynonymous SNV	c.A615C	p.K205N	LGKERLEAASQQKQLTAQLSLMAL	LGKERLEAASQQNQLTAQLSLMAL
7	GPR143	nonsynonymous SNV	c.A844G	p.K282E	LEMQTDINGGSLKPVRTAAKTTWFI	LEMQTDINGGSLKPVRTAAKTTWFI

7	GUSB	nonsynonymous SNV	c.T410G	p.V137G	SAHSYAIVWVNGVDLTLEHEGGYLPF	SAHSYAIVWVNGDLEHEGGYLPF
7	GXYLT1	nonsynonymous SNV	c.T351G	p.H117Q	SAIIFSIKPLQFHIFAEDQLHHSFK	SAIIFSIKPLQFQIFAEDQLHHSFK
8	GXYLT1	nonsynonymous SNV	c.T351G	p.H117Q	RYSLKIQPVKEKMLAVVACGERLEE	RYSLKIQPVKEKMLAVVACGERLEE
8	HGC6.3	nonsynonymous SNV	c.A365C	p.Q122P	TAGGMTPTASSSQHTAGVMTPTASS	TAGGMTPTASSSPHTAGVMTPTASS
8	IQSEC3	nonsynonymous SNV	c.A41G	p.Y14C	ESLLENPVRAVLYLKELTAIVQNQQ	ESLLENPVRAVLCKELTAIVQNQQ
8	IQSEC3	nonsynonymous SNV	c.C92T	p.T31I	TAIVQNQQSLIHTQRERIDELERRL	TAIVQNQQSLIHIQRERIDELERRL
8	IRS2	nonsynonymous SNV	c.A3244C	p.T1082P	GDYTEMAFGVAATPPQPIAAPPKPE	GDYTEMAFGVAAPPQPIAAPPKPE
8	KIAA1109	nonsynonymous SNV	c.A4823T	p.K1608M	NIQDTIAEIRRKISFKVVF	NIQDTIAEIRRMISFKVVF
8	KIAA1210	nonsynonymous SNV	c.A5119T	p.I1707F	RKKAKAWSHMAEITQ	RKKAKAWSHMAEFTQ
8	KRT18	nonsynonymous SNV	c.A201G	p.I67M	RGGMGSGGLATGIAGGLAGMGGIQN	RGGMGSGGLATGMAGGLAGMGGIQN
8	KRT6B	nonsynonymous SNV	c.G262A	p.G88R	SCAISGGYGSRRAGSYFGGAGSGF	SCAISGGYGSRRAGSYFGGAGSGF
8	KRTAP1-3	nonsynonymous SNV	c.T148C	p.F50L	SCCQTSFCGFPFSTSGTCSSSCCQ	SCCQTSFCGFPPLSTSGTCSSSCCQ
8	KRTAP4-6	nonsynonymous SNV	c.T580C	p.C194R	TTCYRPTCVISTCPRPLCCASSCC	TTCYRPTCVISTRPRPLCCASSCC
8	LEPREL2	nonsynonymous SNV	c.C167T	p.A56V	GLRAYAAGAWAPAVALLREALRSQA	GLRAYAAGAWAPVVALREALRSQA
9	LILRB3	nonsynonymous SNV	c.C560A	p.T187N	GGFQALFPVGPVTPSHRWRFTCYYY	GGFQALFPVGPVNPSHRWRFTCYYY
9	LOC10028 8778	nonsynonymous SNV	c.G379A	p.A127T	VLASAPPLPPSTAAPVGQGARQDDS	VLASAPPLPPSTTAPVGQGARQDDS
9	LOC10028 8778	nonsynonymous SNV	c.C475T	p.R159C	GGSSSLRPPDFWRAQCTRVDFLWVC	GGSSSLRPPDFWCAQCTRVDFLWVC
9	LOC39975 3	nonsynonymous SNV	c.G1725T	p.E575D	TAILLLAHGSREEVNETCGEGDGCT	TAILLLAHGSREDVNETCGEGDGCT
9	LOC40101 0	nonsynonymous SNV	c.C1603A	p.P535T	DTEGFLERGLGPLSTRRGVEEDKE	DTEGFLERGLGTLSTRRGVEEDKE
9	LOC64575 2	nonsynonymous SNV	c.G270C	p.K90N	NNQALSVLSKEQKQRLQEERLQE	NNQALSVLSKEQNQRLQEERLQE
9	MAFA	nonsynonymous SNV	c.G293A	p.S98N	GSSQAGGAPPPSGGPGAVGGTSGK	GSSQAGGAPPPNGGPGAVGGTSGK
9	MUC5B	nonsynonymous SNV	c.A5977C	p.T1993P	TPIPSSSLGTTWTRLSQTTTPTATM	TPIPSSSLGTTWPRLSQTTTPTATM
9	NACAD	nonsynonymous SNV	c.T1999C	p.S667P	SAPKPVAAATLVSQAAEGLTLPQD	SAPKPVAAATLVPPQAAEGLTLPQD
9	NBPF10	nonsynonymous SNV	c.G580A	p.A194T	VLESSAPREVQKAEESKVPEDSLEE	VLESSAPREVQKTEESKVPEDSLEE
9	NBPF15	nonsynonymous SNV	c.A715G	p.T239A	TKITFEEDKV DSTLIGSSSHVEWED	TKITFEEDKVDSALIGSSSHVEWED
9	NBPF8	nonsynonymous SNV	c.G342T	p.L114F	EGYSTLSIPPEMLASYKSYSSTFHS	EGYSTLSIPPEMFASYKSYSSTFHS
10	NDRG4	nonsynonymous SNV	c.A59G	p.D20G	RFPEEKPLLRGQDATELESSDAFLL	RFPEEKPLLRGQGATELESSDAFLL

10	NLGN4X	nonsynonymous SNV	c.C1434A	p.S478R	PTYFYAFYHHCQSEMKPSWADSAHG	PTYFYAFYHHCQREMKPSWADSAHG
10	NLGN4X	nonsynonymous SNV	c.C572T	p.A191V	EGTGNMIDGSILASYGNVIVITINY	EGTGNMIDGSILVSYGNVIVITINY
10	NOTCH2NL	nonsynonymous SNV	c.C563A	p.P188H	GQYCDSLYVPCAPSPCVNGGTCRQT	GQYCDSLYVPCAHSPCVNGGTCRQT
10	NPIPL1	nonsynonymous SNV	c.C26G	p.T9S	MVKLSIVLTPQFLSHDQSQLTKELQ	MVKLSIVLSPQFLSHDQSQLT
10	NSUN5P2	nonsynonymous SNV	c.A176C	p.Q59P	CSLCQEENEDMVQDALQQNPGAFRL	CSLCQEENEDMVPDALQQNPGAFRL
10	NSUN5P2	nonsynonymous SNV	c.A568C	p.K190Q	APSARRRMKTWYKMRCRSTRAPSG	APSARRRMKTWYQMRCRSTRAPSG
10	OR10G9	nonsynonymous SNV	c.C560A	p.A187D	HLYCDAPPILKLACADTSANEMVIF	HLYCDAPPILKLDACADTSANEMVIF
10	OR1D5	nonsynonymous SNV	c.T308C	p.F103S	AISYAGCLTQLYFLVSLVTLDNLIL	AISYAGCLTQLYSLVSLVTLDNLIL
10	OR2L8	nonsynonymous SNV	c.A206T	p.D69V	PMYFLLSQLSLIDLNYISTIVPKMA	PMYFLLSQLSLIVLNYISTIVPKMA
10	OR2M1P	nonsynonymous SNV	c.T175C	p.C59R	LAVMSYDCYIAICHPLRYTNLMRPK	LAVMSYDCYIAIRHPLRYTNLMRPK
10	OR2T3	nonsynonymous SNV	c.C344G	p.A115G	GIQMFFYLTLAGAEVFLAAMAYDR	GIQMFFYLTLAGGEVFLAAMAYDR
11	OR5H14	nonsynonymous SNV	c.C427T	p.R143W	LYPAIMTNGLCIRLLLSYVGGLLH	LYPAIMTNGLCIWLILLSYVGGLLH
11	PBX3	nonsynonymous SNV	c.G20A	p.R7K	MRGLSIRGAQEEDPPDPQLMRDNM	MRGLSIKGAQEEDPPDPQL
11	PBX3	nonsynonymous SNV	c.G62A	p.R21K	LCEIKEKTGLSIRGAQEEDPPDPQL	LCEIKEKTGLSIKGAQEEDPPDPQL
11	PCDHA10	nonsynonymous SNV	c.T67C	p.W23R	QCLLLSLLLLAAWEVGSQGLHYSVY	QCLLLSLLLLAAREVGSQGLHYSVY
11	PCMTD1	nonsynonymous SNV	c.G418A	p.E140K	EEDKHNEAMKEPPQNLLREKIMK	EEDKHNEAMKEKPPQNLLREKIMK
11	PGM2	nonsynonymous SNV	c.T542G	p.V181G	AVAEKQDSGEWRVFSGNELGALLGW	AVAEKQDSGEWRGFSGNELGALLGW
11	PKD1	nonsynonymous SNV	c.C841G	p.H281D	FPASPGATLVGPHGPLASGQLAAFH	FPASPGATLVGPDGPLASGQLAAFH
11	PLXND1	nonsynonymous SNV	c.A958G	p.M320V	AHYKIPEGASLAMLIDKKDNTLGR	AHYKIPEGASLAVSLIDKKDNTLGR
11	POTED	nonsynonymous SNV	c.T278C	p.M93T	GTSGDHENSFMKMLRSKMGKWCCHC	GTSGDHENSFMKTLRSKMGKWCCHC
11	PPP2CB	nonsynonymous SNV	c.G95A	p.C32Y	CKQLNENQVRTLCEKAKEILTKESN	CKQLNENQVRTLYEKAKEILTKESN
11	PPP2CB	nonsynonymous SNV	c.G43C	p.E15Q	DKAFTKELDQWVEQLNECKQLNENQ	DKAFTKELDQWVQQLNECKQLNENQ
11	PRAMEF20	nonsynonymous SNV	c.T206G	p.L69R	QAWPFLRPLGSLMKRPCPETFQAV	QAWPFLRPLGSRMKRPCPETFQAV
12	PRB1	nonsynonymous SNV	c.G446A	p.R149Q	KPQGPPPPQGDKSRSPRPPGKPPQP	KPQGPPPPQGDKSQSPRPPGKPPQP
12	PROS1	nonsynonymous SNV	c.T848C	p.F283S	EAVMDINKPGPLFKPENGLLETKVY	EAVMDINKPGPLSKPENGLLETKVY
12	PTDSS2	nonsynonymous SNV	c.G28A	p.G10R	MRRGERRDAGGPRPESVPAGRASL	MRRGERRDARGPRPESVPAGR
12	PWWP2B	nonsynonymous SNV	c.G298C	p.E100Q	ARGVQPPETTRPEPPPLVPLPAG	ARGVQPPETTRQPPPLVPLPAG
12	RAB4B	nonsynonymous SNV	c.G295T	p.V99L	IWDTAGQERFRSVTRSYYGAAGAL	IWDTAGQERFRLTRSYYGAAGAL

12	RBMS1	nonsynonymous SNV	c.T563A	p.M188K	SSGTSRGGVGFARMESTEKCEAVIGH	SSGTSRGGVGFARKESTEKCEAVIGH
12	RBMXL1	nonsynonymous SNV	c.T596C	p.L199P	RDSYGGPPRREPLSRRDVYLSPRD	RDSYGGPPRREPPSRRDVYLSPRD
12	RFPL4A	nonsynonymous SNV	c.G409A	p.A137T	QNRKEQAERFDTALCVLGTFRFTSG	QNRKEQAERFDTTLCVLGTFRFTSG
12	RNF187	nonsynonymous SNV	c.G73T	p.A25S	ALCQRAPREPVRADCGHRFCRACVV	ALCQRAPREPVRSDCGHRFCRACVV
12	RP9P	nonsynonymous SNV	c.G64T	p.G22W	RGSRWIRNCSGPGSRSGGGTTPSSC	RGSRWIRNCSGPWSRSGGGTTPSSC
12	SH2D3C	nonsynonymous SNV	c.G1270A	p.A424T	WGSTEHGVEVVLHLEAARTVAHHG	WGSTEHGVEVVLTHLEAARTVAHHG
12	SH3BP1	nonsynonymous SNV	c.A1869C	p.L623F	GSSLRAPTVPPLPPTPPQARRQS	GSSLRAPTVPFPPTPPQARRQS
13	SHISA7	nonsynonymous SNV	c.T14A	p.L5Q	MPALLLVLLASSAGQARARPSNAT	MPALQLLVLLASSAGQA
13	SLC25A5	nonsynonymous SNV	c.G132T	p.Q44H	VKLLLVQVHASKQITADKQYKGIID	VKLLLVQVHASKHITADKQYKGIID
13	SLC35G5	nonsynonymous SNV	c.G667T	p.A223S	RSLHFPSCLPTVAFLSGLVGLLGCV	RSLHFPSCLPTVFLSGLVGLLGCV
13	SLC9A6	nonsynonymous SNV	c.C1309A	p.L437I	GSNFQHMMMFAGLRGAMAFALAIRD	GSNFQHMMMFAGIRGAMAFALAIRD
13	SNRNP70	nonsynonymous SNV	c.G545C	p.R182P	DGRRVLVDVERGRTVKGWRPRRLGG	DGRRVLVDVERGPTVKGWRPRRLGG
13	SPATC1	nonsynonymous SNV	c.A979C	p.T327P	EQVVPASVPTSPTTPTVTVLASAP	EQVVPASVPTSPTTPTVTVLASAP
13	SPPL2A	nonsynonymous SNV	c.C58T	p.L20F	SPAGAALLWGFLQLTAAQEAILHA	SPAGAALLWGFLFQLTAAQEAILHA
13	STAT5A	nonsynonymous SNV	c.T8A	p.F3Y	MKFKAEVQSNRGLTKENLVFLAQKL	MKYKAEVQSNRGLTK
13	STAT5A	nonsynonymous SNV	c.T1454A	p.F485Y	LWPQLCEALNMKFKAEVQSNRGLTK	LWPQLCEALNMKYKAEVQSNRGLTK
13	STMN1	nonsynonymous SNV	c.A122T	p.K41M	KESVPEFPLSPPKKKDLSEEIQKK	KESVPEFPLSPPMKKDLSEEIQKK
13	TINCR	nonsynonymous SNV	c.T313C	p.F105L	TLRGTSALAQDIFPATPLPKPCPLP	TLRGTSALAQDILPATPLPKPCPLP
13	TMEM184A	nonsynonymous SNV	c.C250T	p.P84S	HQIYLHLRSYTVPEQRYIIRLLLI	HQIYLHLRSYTVSQEQRYIIRLLLI
14	TMEM201	nonsynonymous SNV	c.C71A	p.A24E	PTAGLAGGLGVTACAAAGVLLYRIA	PTAGLAGGLGVTECAAAGVLLYRIA
14	TPTE	nonsynonymous SNV	c.G1237A	p.D413N	EKMTSSDVVAGSD	EKMTSSDVVAGSN
14	TRIM28	nonsynonymous SNV	c.C59G	p.P20R	ASAAAASAASGSPGPEGSAGGEKR	ASAAAASAASGRGPEGSAGGEKR
14	UGT2B4	nonsynonymous SNV	c.G1075A	p.G359R	LTWFQYHSLDVTGFLACVATVIFI	LTWFQYHSLDVTRFLLACVATVIFI
14	UGT2B4	nonsynonymous SNV	c.C622T	p.P208S	PQKVLWRFDGNKPDTLGLNTRLYKW	PQKVLWRFDGNKSDTLGLNTRLYKW
14	UPF3A	nonsynonymous SNV	c.C282A	p.H94Q	EQLEEQLRPLPAHDYFEFFAADLSL	EQLEEQLRPLPAQDYFEFFAADLSL
14	USP1	nonsynonymous SNV	c.C599T	p.A200V	LNPMYEGYLQHDVQEVLCILGNIQ	LNPMYEGYLQHDVQEVLCILGNIQ
14	VPS13C	nonsynonymous SNV	c.G10237T	p.A3413S	PFQGA VQGPEEF AEGLVIGVRSFLG	PFQGA VQGPEEF SEGLVIGVRSFLG
14	WDR13	nonsynonymous SNV	c.T13C	p.W5R	MAAVWQVLAVDARYNAYRTPTFPQ	MAAVRQVLAVDARYNA

14	WDR24	nonsynonymous SNV	c.G2207A	p.C736Y	RPMSRGGWVCDRCHRCASMCVCHH	RPMSRGGWVCDRYHRCASMCVCHH
14	X97876	nonsynonymous SNV	c.A632G	p.Y211C	CKSRKEQKETTFTVMRLAATDLLF	CKSRKEQKETTFTVMRLAATDLLF
14	XYLT1	nonsynonymous SNV	c.C190T	p.R64C	GGGEQPPAPAPRRERRDLPAEPAA	GGGEQPPAPAPCRERRDLPAEPAA
15	ZBTB47	nonsynonymous SNV	c.A1076G	p.Q359R	EEEEEGEAGGKQGPRGSRSSRADP	EEEEEGEAGGKRGPGRSSRADP
15	ZCCHC6	nonsynonymous SNV	c.C304T	p.P102S	KVFTKIYKGEKKPEIFVDGWNIYFF	KVFTKIYKGEKKSEIFVDGWNIYFF
15	ZCCHC6	nonsynonymous SNV	c.C3604T	p.P1202S	PVLQEIYKGEKKPEIFVDGWNIYFF	PVLQEIYKGEKKSEIFVDGWNIYFF
15	ZMIZ1	nonsynonymous SNV	c.T251C	p.M84T	PQLSHPPDMPNNMAALEKPLSHPMQ	PQLSHPPDMPNNTAALEKPLSHPMQ
15	ZNF273	nonsynonymous SNV	c.A1383C	p.E461D	KIIHTGAKPYKCEECGSAFRAFSTL	KIIHTGAKPYKCEECGSAFRAFSTL
15	ZNF429	nonsynonymous SNV	c.C1634A	p.A545D	GEKPYKCEECGKAFNRSSRLTQHKK	GEKPYKCEECGKDFNRSSRLTQHKK
15	ZNF717	nonsynonymous SNV	c.G1588A	p.A530T	CKSFLTVHQRTHAGEKPYACNECGK	CKSFLTVHQRTHTGEKPYACNECGK
15	ZNF721	nonsynonymous SNV	c.T503A	p.L168H	EECGKAFNRSTNLTAHKRIHNREKA	EECGKAFNRSTNHTAHKRIHNREKA
15	ZNF737	nonsynonymous SNV	c.C1157G	p.T386R	CGKAFNWSSHLTTHKRIHTGEKPYK	CGKAFNWSSHLTRHKRIHTGEKPYK
15	ZNF93	nonsynonymous SNV	c.A995G	p.K332R	KAFKYSRILTTHKRIHTGEKPYKCN	KAFKYSRILTTHRIHTGEKPYKCN
15	ZNF99	nonsynonymous SNV	c.A2348G	p.H783R	KHFSALRKHKIIHTGKKPYKCEECG	KHFSALRKHKIIRTGKKPYKCEECG
15	MUC2	nonsynonymous SNV	c.T4373C	p.I1458T	TTPPPTTTPSPPISTTTTTPPTTTP	TTPPPTTTPSPPTSTTTTTPPTTTP
16	MUC2	nonsynonymous SNV	c.C4463T c.C4472T c.C4483T	p.T1488I p.T1491I p.P1495S	TPSPPTTTPSPPTTTTTTTPPTTTPSP	TPSPPTTTPSPPTITTTTPSPTTTPSP
16	MUC4	nonsynonymous SNV	c.A8317G c.T8330C	p.N2773D p.V2777A	ASTGHATPLPVTNTSSVSTGHATPLHVTS	ASTGHATPLPVDTSASTGHATPLHVTS
16	PLEC	nonsynonymous SNV	c.A13646C	p.Y4549S	GSGFSMTFSSSSSSSGYGRRYASG	GSGFSMTFSSSSSSSGYGRRYASG
16	ADAMTS7	nonsynonymous SNV	c.T3875C c.T3878C	p.L1292P p.L1293P	DSELWPTVGVASLLPPIAPLPEMKV	DSELWPTVGVASPPPIAPLPEMKV
16	CLIC6	nonsynonymous SNV	c.G775A c.T791A c.A805G	p.G259S p.V264D p.S269G	GDSVEAGDPAGDGVEAGVPAGDSVEAEG PAGDSMD	GDSVEAGDPAGDSVEAGDPAGDGVEAEGPAGDSMD
16	DSCAML1	nonsynonymous SNV	c.G6145C c.A6151C	p.G2049R p.T2051P	PAPPAGTAPPAPGPTPAEPTAPSAAP	PAPPAGTAPPAPRPPPAEPTAPSAAP
16	ENSG0000 0163046	nonsynonymous SNV	c.C533T c.G553C	p.A178V p.E185Q	VKNKAGHTPLLLAIRKRSEIIVEFLTKNA NA	VKNKAGHTPLLLVIRKRSEIIVEFLTKNANA
16	KRT18	nonsynonymous SNV	c.G76A c.C79T c.C83A c.G94T c.C102A	p.A26T p.R27W p.P28Q p.A32S p.S34R	RSLGVSQAPSYGARPVSSAASVYAGAGGS GS	RSLGVSQAPSYGTWQVSSARVYAGAGGSGS
16	LINC00273	nonsynonymous SNV	c.G257A	p.R86Q	ARDRRPLGRGRRGAAQTFGQTRGGPRCP GSHR	ARDRRPLGRGRRRAAFTFGQTRGGPRCPGSHR
16	RBMX	nonsynonymous SNV	c.G356T c.C382G	p.G119V p.P128A	PLPVKRGPPRSGGPPPKRSAPSGPVRSSSG MGG	PLPVKRGPPRSGVPPPKRSAASGPVRSSSGMGG

17	X97876	nonsynonymous SNV	c.C433G c.T434C c.C490A c.G464A c.A406G	p.L145A p.P164T p.R155H p.M136V	RGSNALQVVISLMVALSLFCHLQTLCTP GLRTTSHCHVNPFPQCVLGLLEPN	RGSNALQVVISLVVALSLFCHAQTQLCTPGLHTTSHCHVNTF PGQCVLGLLEPN
17	OVGP1	nonframeshift insertion	c.543_544insACT GGACAGAAGA CCCTGACC	p.P182delinsTGQK TLTP	KTLTPVGHQSVTPVSHQSVSPGGTT	KTLTPVGHQSVTTGQKTLTPVSHQSVSPGGTT
17	MAFA	nonframeshift insertion	c.603_604insGCC GCCAC	p.H202delinsAAH H	HAAHHHHAHHHHHHHHHHGGAGHG	HAAHHHHAHHHHAHHHHHHHHGGAGHG
17	ZC3H3	nonframeshift insertion	c.2639_2640insC AA	p.S880delinsSK	SSSKASSSSSSSSPPASLDHEAPS	SSSKASSSSSSSSKPPASLDHEAPS
17	GOLGA6B	nonframeshift deletion	c.1528_1530del	p.510_510del	PGEGDGGQHLDSSEEEAPRPTNIP	PGEGDGGQHLDSSEEEAPRPTNIPE
17	AGAP6	frameshift insertion	c.54dupC	p.D18fs	TCRVHPSVSLFEDQQQGSVCPSESE	TCRVHPSVSLFEDPAAGVGVSL
17	PTPN4	frameshift insertion	c.549dupT	p.N183fs	SRVAPGTPVSYLNVSNKSCFQEYVL	SRVAPGTPVSYLNCFK
17	CYP4F3IP	frameshift insertion	c.345dupT	p.P115fs	LIVWWLPFVAVSPCLF	LIVWWLPFVAVSPLFVL
17	ZNF717	frameshift insertion	c.397_398insAA	p.T133fs	NTSTQERVELGKTFNLNSNHVLNLI	NTSTQERVELGKKHLI
18	DQ580909	frameshift insertion	c.744dupC	p.H248fs	QALHLLPAPARLHPMVSLPKEERRE	QALHLLPAPARLHPDGLPAQGGEKRKDAPRWWTSAATTLTSL LLLPCVFHDLEVVGNPRLKTRGEFRLQDDM
18	X97876	frameshift insertion	c.633_634insTC	p.Y211fs	CKSRKEQKETTFFYTMRLAATDLLF	CKSRKEQKETTFFYSQLCGWLPPTCCSLCW
18	HDAC1	frameshift deletion	c.36delT	p.C12fs	MAQTQGTRRKVCYYYDGDVGNYYY	MAQTQGTRRKVCTTTTGMLEITIMDKATQS
18	LOC39975 3	frameshift deletion	c.1731delC	p.N577fs	ILLLAHGSREEVNETCGEGDGCTAL	ILLLAHGSREEVKRPVGRETAARHSIWAARGMWSWSSS
18	SMAD6	frameshift deletion	c.336delC	p.G112fs	AGSSLLDVAEPGGPGWLPESDCETV	AGSSLLDVAEPGRAGCPRTARR
18	BC033456	frameshift deletion	c.156delT	p.F52fs	PEYSRFSLLFFFFPQHLNSQLPRLS	PEYSRFSLLFFFFPSTLTLSFPDFLQ
18	ABHD17A	frameshift deletion	c.635delC	p.S212fs	ASRYECAAVVLHSPPLTSGMRVAFPD	ASRYECAAVVLHCRSPRACASPSPTPRRPTASTPSLTSRRCP SRLPCSSSTARTR
19	ABHD17A	frameshift deletion	c.635delC	p.S212fs	ASRYECAAVVLHSPPLTSGMRVAFPD	ASRYECAAVVLHCRSPRACASPSPTPRRPTASTPSLSS
19	ABHD17A	frameshift deletion	c.541delC	p.R181fs	PRATSVPRWCCTRRSPRACASPSPT	PRATSVPRWCCTAAHLGHARRLPRHQEDLLRRLP
19	RUNX1- IT1	frameshift deletion	c.186_187del	p.62_63del	LSGEIKVLSLSLYLTHTHTHTH	LSGEIKVLSLSLSHTHTHTHTHTHREEREREREREREASIQ NSHA
19	HGC6.3	frameshift deletion	c.499_502del	p.167_168del	GNDPHCLLLPHTLQGE	GNDPHCLLLPHTCRGSS
19	ADAMTSL 1	frameshift deletion	c.30delC	p.F10fs	MSGVFCFFFFLGVPEAEVTWF	MSGVFCFFFFSS
19	ARID1A	nonsynonymous SNV	c.G107T	p.G36V	LKKAQQQREEAGGEAAAAAAAAAERG	LKKAQQQREEAVGEAAAAAAAAAERG
19	CAPNS1	nonsynonymous SNV	c.C15A	p.N5K	MFLVNSFLKGGGGGGGGGLGGGL	MFLVKSFLKGGGGGGGG
19	EPHA8	nonsynonymous SNV	c.G2756A	p.S919N	TVSRCPPPAFVRSCFDLRGGSGGGG	TVSRCPPPAFVRNCFDLRGGSGGGG
19	GRIK5	nonsynonymous SNV	c.C4T	p.P2S	MPAELLLLLIVAFASPCQVLSLR	MSAELLLLLIVAF