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13 Table S5. Summary of possible driver mutations identified in this study

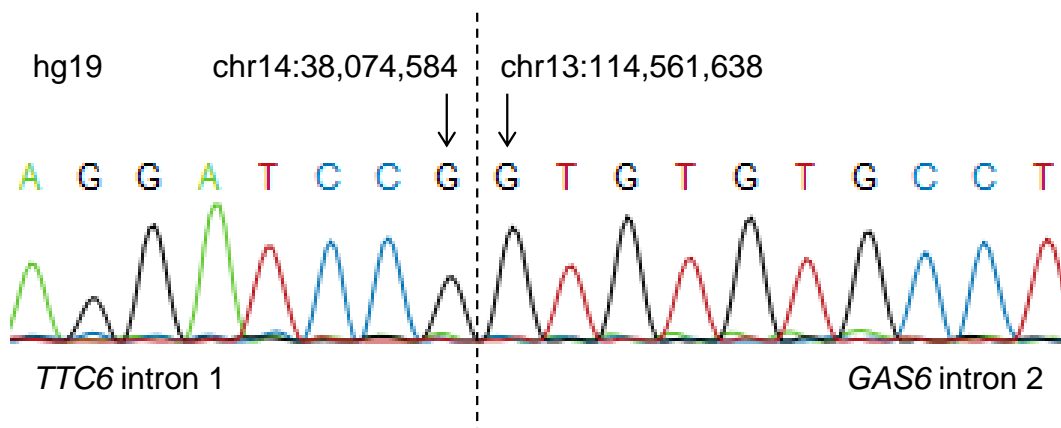
14 Table S6. Top 200 differentially expressed genes

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16

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19 **Figure S1. Validation of the genomic breakpoints involving *GAS6* and *FOXA1* in UPN1**



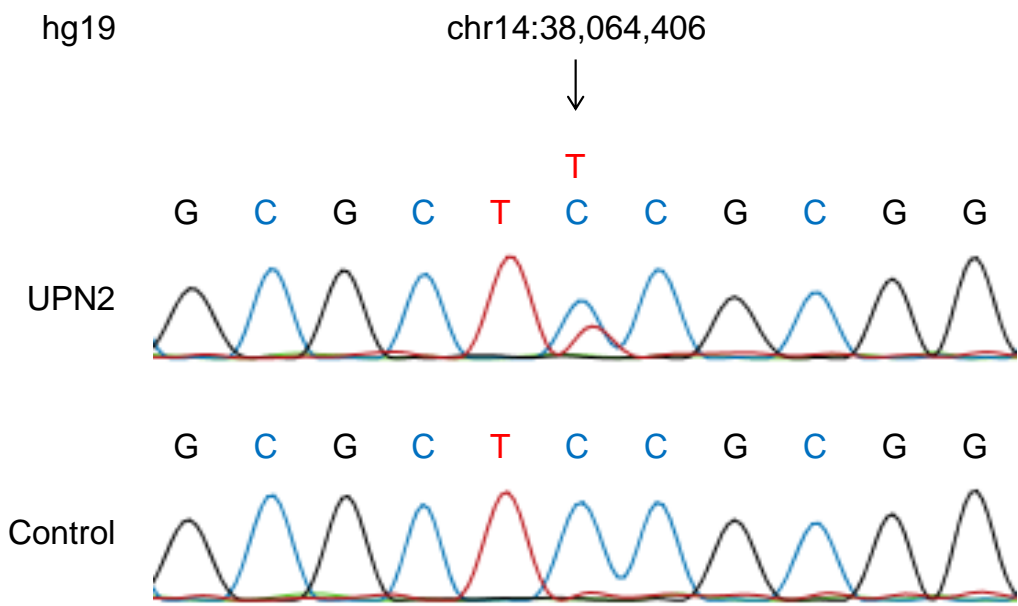
23 Sanger sequencing validating the structural variation identified in UPN1. The hg19 genome
24 coordinate is used to represent the positions of the breakpoint.

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26

27 **Figure S2. Validation of the *FOXA1* g.38064406G>A mutation identified in UPN2**

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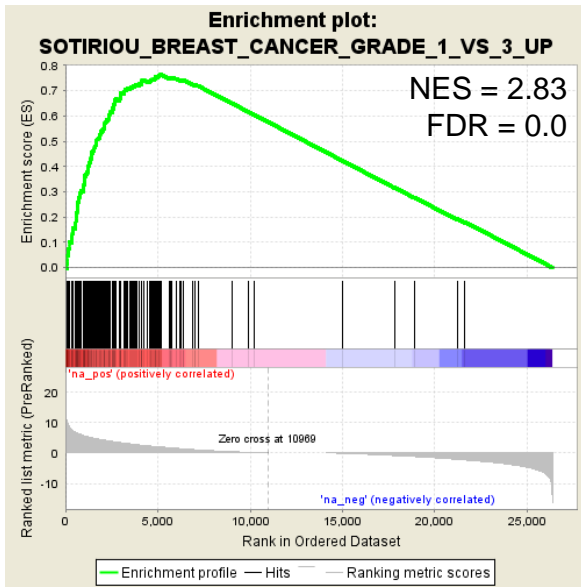
31 Sanger sequencing validating the presence of C>T (G>A) nucleotide alteration at 38,064,406 on
32 chromosome 14.

33

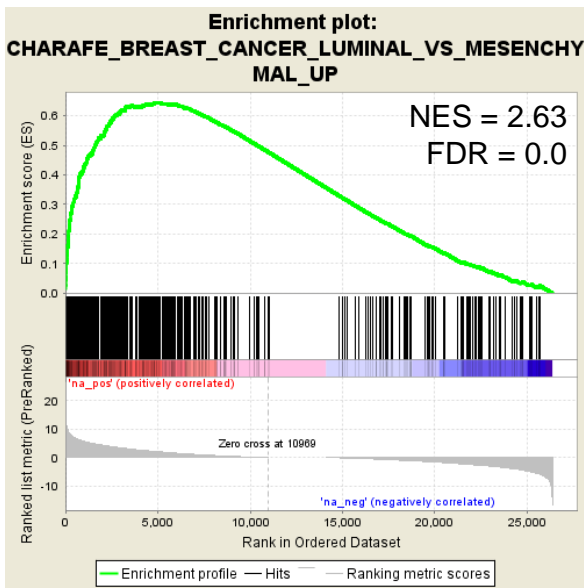
34

Figure S3. Additional gene set enrichment analyses

A



B



37 Enrichment plots showing the upregulation of genes associated with (A) high-grade breast cancer

38 (SOTIRIOU_BREAST_CANCER_GRADE_1_VS_3_UP, the most significantly enriched in the C2

39 gene set database) and (B) luminal breast cancer

40 (CHARAFE_BREAST_CANCER_LUMINAL_VS_MESENCHYMAL_UP, the fourth most

41 significantly enriched). NES, normalized enrichment score; FDR, false discovery rate.

Table S1. Detailed clinical characteristics of each patient

UPN	Age / gender	Age at onset	Primary location	Dermal invasion	Metastasis of SLN	Frozen / FFPE	Analysis	FOXA1 expression	ER expression
1	55 / F	53	Perineal	<i>In situ</i>	-	Frozen	WGS/RNA/TGS	+	+
2	70 / M	68	Penoscrotal	Invasive	-	Frozen	WGS/RNA/TGS	+	+
3	73 / F	72	Perineal	<i>In situ</i>	-	Frozen	RNA/TGS	+	+
4	65 / F	64	Perineal	Invasive	-	Frozen	TGS	+	+
5	89 / M	88	Penoscrotal	Invasive	+	Frozen	RNA/TGS	+	+
6	76 / F	75	Perineal	Invasive	-	Frozen	RNA/TGS	+	+
7	71 / F	68	Perineal	<i>In situ</i>	-	Frozen	RNA/TGS	+	+
8	79 / M	78	Penoscrotal	Invasive	-	Frozen	RNA/TGS	+	+
9	58 / M	53	Penoscrotal	Invasive	-	Frozen	RNA/TGS	+	NA
10	66 / M	65	Penoscrotal	<i>In situ</i>	-	Frozen	TGS	+	+
11	72 / M	67	Penoscrotal	Invasive	+	FFPE	WES/TGS	+	+
12	80 / M	79	Penoscrotal	<i>In situ</i>	-	FFPE	WES/TGS	+	+
13	60 / M	59	Penoscrotal	Invasive	+	FFPE	WES/TGS	+	+
14	67 / M	64	Inguinal	Invasive	+	FFPE	WES/TGS	+	-
15	70 / M	70	Perineal	Invasive	-	FFPE	WES/TGS	+	-
16	73 / F	70	Perineal	<i>In situ</i>	-	FFPE	WES/TGS	+	-
17	88 / F	84	Perineal	<i>In situ</i>	-	FFPE	WES/TGS	+	+
18	80 / F	79	Perineal	<i>In situ</i>	-	FFPE	WES/TGS	+	-
19	82 / F	79	Perineal	Invasive	-	FFPE	WES/TGS	+	-
20	72 / M	69	Perineal	Invasive	-	FFPE	WES/TGS	+	-
21	69 / F	65	Perineal	<i>In situ</i>	-	FFPE	WES/TGS	+	+
22	78 / M	75	Penoscrotal	Invasive	-	FFPE	TGS	+	+
23	82 / M	81	Penoscrotal	Invasive	-	FFPE	TGS	+	++
24	90 / F	83	Perineal	<i>In situ</i>	NA	FFPE	TGS	+	+
25	65 / M	57	Penoscrotal	<i>In situ</i>	NA	FFPE	TGS	+	+
26	76 / M	76	Penoscrotal	<i>In situ</i>	NA	FFPE	TGS	+	-
27	64 / F	58	Perineal	<i>In situ</i>	-	FFPE	TGS	+	++
28	62 / M	59	Penoscrotal	Invasive	-	FFPE	TGS	+	+
29	85 / M	84	Penoscrotal	Invasive	NA	FFPE	TGS	+	-
30	74 / F	66	Axillary	<i>In situ</i>	NA	FFPE	TGS	+	+
31	73 / M	68	Penoscrotal	<i>In situ</i>	NA	FFPE	TGS	+	-
32	81 / M	80	Penoscrotal	Invasive	-	FFPE	TGS	+	-
33	71 / M	61	Penoscrotal	Invasive	NA	FFPE	TGS	+	+
34	78 / M	77	Penoscrotal	Invasive	NA	FFPE	TGS	+	++
35	92 / F	NA	Perineal	Invasive	NA	FFPE	TGS	+	+
36	87 / F	NA	Axillary	<i>In situ</i>	NA	FFPE	TGS	+	++
37	64 / M	61	Penoscrotal	<i>In situ</i>	NA	FFPE	TGS	+	++
38	83 / M	NA	Penoscrotal	<i>In situ</i>	NA	FFPE	TGS	+	+

39	77/M	77	Penoscrotal	Invasive	-	Frozen	WES/TGS	NA	NA
40	57/F	54	Vulval	<i>In situ</i>	-	Frozen	WES/TGS	+	-
41	78/F	78	Perianal	<i>In situ</i>	-	Frozen	WES/TGS	+	-
42	77/M	76	Perianal	<i>In situ</i>	-	Frozen	WES/TGS	+	+
43	59/M	53	Penoscrotal	Invasive	-	Frozen	WES/TGS	+	-
44	68/F	67	Perianal	<i>In situ</i>	-	Frozen	WES/TGS	+	-
45	69/M	64	Mons pubis	Invasive	-	Frozen	WES/TGS	+	-
46	75/M	74	Inguinal	Invasive	+	Frozen	WES/TGS	+	+
47	74/M	74	Penoscrotal	<i>In situ</i>	-	Frozen	WES/TGS	+	+
48	71/M	71	Inguinal	<i>In situ</i>	-	Frozen	WES/TGS	+	-

44 SLN, sentinel lymph node: FFPE, formalin-fixed paraffin-embedded; ER, estrogen receptor; WGS,
45 whole-genome sequencing; RNA, RNA sequencing; TGS, targeted gene sequencing; WES,
46 whole-exome sequencing; NA, not available

Table S2. Genes and regions analyzed by targeted sequencing

Genes (coding regions + 25 bp targeted)							
<i>ABCB1</i>	<i>ABCC2</i>	<i>ABL1</i>	<i>ABL2</i>	<i>ACO1</i>	<i>ACOX1</i>	<i>ACRC</i>	<i>ACTG1</i>
<i>ACVR1B</i>	<i>ADAM28</i>	<i>ADGRB3</i>	<i>ADIPOR1</i>	<i>AGAP2</i>	<i>AHNAK</i>	<i>AIM1</i>	<i>AK2</i>
<i>AKT1</i>	<i>AKT2</i>	<i>AKT3</i>	<i>ALDH1B1</i>	<i>ALK</i>	<i>ALPK2</i>	<i>ANK3</i>	<i>ANTXR1</i>
<i>APC</i>	<i>AQP7</i>	<i>ARHGEF16</i>	<i>ARID1A</i>	<i>ARID2</i>	<i>ARMCX4</i>	<i>ASXL1</i>	<i>ATM</i>
<i>ATN1</i>	<i>ATP1A1</i>	<i>ATP6V0C</i>	<i>ATP8A1</i>	<i>ATP8B2</i>	<i>ATRX</i>	<i>BAHD1</i>	<i>BCL11A</i>
<i>BCLAF1</i>	<i>BRAF</i>	<i>BRCA1</i>	<i>BRCA2</i>	<i>BRD4</i>	<i>C11orf30</i>	<i>C11orf80</i>	<i>C14orf159</i>
<i>C16orf82</i>	<i>C16orf89</i>	<i>C1orf127</i>	<i>C1QL1</i>	<i>C8orf44</i>	<i>CACNA1C</i>	<i>CAMTA1</i>	<i>CASC5</i>
<i>CASP8</i>	<i>CAST</i>	<i>CBFB</i>	<i>CBL</i>	<i>CCDC116</i>	<i>CCDC148</i>	<i>CCDC40</i>	<i>CDA</i>
<i>CDC42BPA</i>	<i>CDH1</i>	<i>CDK20</i>	<i>CDK4</i>	<i>CDKAL1</i>	<i>CDKN1B</i>	<i>CDKN2A</i>	<i>CDKN2B</i>
<i>CEBPA</i>	<i>CFAP221</i>	<i>CHD2</i>	<i>CHD7</i>	<i>CHGB</i>	<i>CHIC2</i>	<i>CHRM5</i>	<i>CLCN3</i>
<i>CLPS</i>	<i>CNNM4</i>	<i>COBLL1</i>	<i>CREBBP</i>	<i>CRLF2</i>	<i>CROT</i>	<i>CRY1</i>	<i>CRY2</i>
<i>CRYBA4</i>	<i>CSF1R</i>	<i>CTCF</i>	<i>CTNNB1</i>	<i>CTNND2</i>	<i>CUL4B</i>	<i>CYB561D1</i>	<i>CYGB</i>
<i>CYP19A1</i>	<i>CYP26B1</i>	<i>CYP27B1</i>	<i>CYP2A6</i>	<i>CYP2B6</i>	<i>CYP2C19</i>	<i>CYP2C9</i>	<i>CYP2D6</i>
<i>CYP2E1</i>	<i>DCAF10</i>	<i>DDR1</i>	<i>DDR2</i>	<i>DDX3X</i>	<i>DDX53</i>	<i>DGKD</i>	<i>DHPS</i>
<i>DHX32</i>	<i>DHX38</i>	<i>DLC1</i>	<i>DNMT1</i>	<i>DNMT3A</i>	<i>DPCR1</i>	<i>DPYD</i>	<i>DST</i>
<i>DUSP14</i>	<i>DUSP4</i>	<i>DYNAP</i>	<i>EGFL6</i>	<i>EGFR</i>	<i>ELOVL2</i>	<i>EP300</i>	<i>EPHA7</i>
<i>EPPK1</i>	<i>ERBB2</i>	<i>ERBB3</i>	<i>ERBB4</i>	<i>ERG</i>	<i>ERP29</i>	<i>ERRFI1</i>	<i>ESR1</i>
<i>ESR2</i>	<i>EYA1</i>	<i>EZH2</i>	<i>FAM208A</i>	<i>FANCD2</i>	<i>FAT1</i>	<i>FBXW7</i>	<i>FDXACB1</i>
<i>FGFR1</i>	<i>FGFR2</i>	<i>FGFR3</i>	<i>FGFR4</i>	<i>FITM1</i>	<i>FLG</i>	<i>FLNB</i>	<i>FLT1</i>
<i>FLT3</i>	<i>FLT4</i>	<i>FRMD7</i>	<i>FRYL</i>	<i>FSTL5</i>	<i>FUT7</i>	<i>GATA3</i>	<i>GLIS3</i>
<i>GLUD1</i>	<i>GNA11</i>	<i>GNAQ</i>	<i>GNAS</i>	<i>GNE</i>	<i>GNPTAB</i>	<i>GPC5</i>	<i>GPR116</i>
<i>GPR179</i>	<i>GPRC6A</i>	<i>GRHL2</i>	<i>GSTP1</i>	<i>GTPBP4</i>	<i>H3F3A</i>	<i>H3F3B</i>	<i>HDAC9</i>
<i>HECTD4</i>	<i>HIST1H1A</i>	<i>HIST1H1B</i>	<i>HIST1H1C</i>	<i>HIST1H1D</i>	<i>HIST1H1E</i>	<i>HIST1H1T</i>	<i>HIST1H2AA</i>
<i>HIST1H2AB</i>	<i>HIST1H2AC</i>	<i>HIST1H2AD</i>	<i>HIST1H2AE</i>	<i>HIST1H2AG</i>	<i>HIST1H2AH</i>	<i>HIST1H2AI</i>	<i>HIST1H2AJ</i>
<i>HIST1H2AK</i>	<i>HIST1H2AL</i>	<i>HIST1H2AM</i>	<i>HIST1H2BA</i>	<i>HIST1H2BB</i>	<i>HIST1H2BC</i>	<i>HIST1H2BD</i>	<i>HIST1H2BE</i>
<i>HIST1H2BF</i>	<i>HIST1H2BG</i>	<i>HIST1H2BH</i>	<i>HIST1H2BI</i>	<i>HIST1H2BJ</i>	<i>HIST1H2BK</i>	<i>HIST1H2BL</i>	<i>HIST1H2BM</i>
<i>HIST1H2BN</i>	<i>HIST1H2BO</i>	<i>HIST1H3A</i>	<i>HIST1H3B</i>	<i>HIST1H3C</i>	<i>HIST1H3D</i>	<i>HIST1H3E</i>	<i>HIST1H3F</i>
<i>HIST1H3G</i>	<i>HIST1H3H</i>	<i>HIST1H3I</i>	<i>HIST1H3J</i>	<i>HIST1H4A</i>	<i>HIST1H4B</i>	<i>HIST1H4C</i>	<i>HIST1H4D</i>
<i>HIST1H4E</i>	<i>HIST1H4F</i>	<i>HIST1H4G</i>	<i>HIST1H4H</i>	<i>HIST1H4I</i>	<i>HIST1H4J</i>	<i>HIST1H4K</i>	<i>HIST1H4L</i>
<i>HIST2H2AA3</i>	<i>HIST2H2AA4</i>	<i>HIST2H2AB</i>	<i>HIST2H2AC</i>	<i>HIST2H2BE</i>	<i>HIST2H2BF</i>	<i>HIST2H3A</i>	<i>HIST2H3C</i>
<i>HIST2H3D</i>	<i>HIST2H4A</i>	<i>HIST2H4B</i>	<i>HIST3H2A</i>	<i>HIST3H2BB</i>	<i>HIST3H3</i>	<i>HIST4H4</i>	<i>HLA-B</i>
<i>HMCN1</i>	<i>HNF1A</i>	<i>HOXA3</i>	<i>HPS5</i>	<i>HRAS</i>	<i>HRNR</i>	<i>HSPG2</i>	<i>IDH1</i>
<i>IDH2</i>	<i>IDI1</i>	<i>IGSF10</i>	<i>IGSF22</i>	<i>IKZF1</i>	<i>IL1RN</i>	<i>IL2RA</i>	<i>IL2RB</i>
<i>IL2RG</i>	<i>IL6ST</i>	<i>ILKAP</i>	<i>INO80D</i>	<i>INPP4B</i>	<i>IQSEC1</i>	<i>ITGAV</i>	<i>JAG2</i>
<i>JAK1</i>	<i>JAK2</i>	<i>JAK3</i>	<i>JARID2</i>	<i>JMJD6</i>	<i>KALRN</i>	<i>KANK2</i>	<i>KDM1A</i>
<i>KDM6A</i>	<i>KDM7A</i>	<i>KDR</i>	<i>KIAA0391</i>	<i>KIT</i>	<i>KMT2A</i>	<i>KMT2C</i>	<i>KNG1</i>
<i>KRAS</i>	<i>KRTAP10-2</i>	<i>KRTAP4-11</i>	<i>KSR2</i>	<i>LAMA2</i>	<i>LAMC1</i>	<i>LCK</i>	<i>LCLAT1</i>
<i>LCTL</i>	<i>LILRB5</i>	<i>LMTK3</i>	<i>LOXL4</i>	<i>LRCH2</i>	<i>LRP6</i>	<i>LRR1</i>	<i>LTK</i>
<i>MACF1</i>	<i>MAP2K1</i>	<i>MAP2K2</i>	<i>MAP2K4</i>	<i>MAP3K1</i>	<i>MAPK1</i>	<i>MED13</i>	<i>MED23</i>

<i>MED24</i>	<i>MET</i>	<i>MGA</i>	<i>MGAM2</i>	<i>MLH1</i>	<i>MOV10L1</i>	<i>MPL</i>	<i>MRT04</i>
<i>MST1R</i>	<i>MTOR</i>	<i>MTR</i>	<i>MUC4</i>	<i>MXRA5</i>	<i>MYB</i>	<i>MYC</i>	<i>MYD88</i>
<i>MYOCD</i>	<i>MYRF</i>	<i>NACA</i>	<i>NACC2</i>	<i>NADK</i>	<i>NBPF10</i>	<i>NCOR1</i>	<i>NEFH</i>
<i>NELL2</i>	<i>NF1</i>	<i>NIM1K</i>	<i>NLRP8</i>	<i>NME2</i>	<i>NOTCH1</i>	<i>NOTCH2</i>	<i>NPIPA5</i>
<i>NPIPB15</i>	<i>NPIPB5</i>	<i>NPM1</i>	<i>NRAS</i>	<i>OGDH</i>	<i>OPCML</i>	<i>OR4A16</i>	<i>OR52N1</i>
<i>PABPC1</i>	<i>PAGE1</i>	<i>PANX2</i>	<i>PCDHA9</i>	<i>PCDHB4</i>	<i>PCNX</i>	<i>PCSK1</i>	<i>PCSK6</i>
<i>PDGFRA</i>	<i>PDGFRB</i>	<i>PDK1</i>	<i>PDZD8</i>	<i>PHF6</i>	<i>PIGN</i>	<i>PIK3C2A</i>	<i>PIK3CA</i>
<i>PIK3R1</i>	<i>PITPNC1</i>	<i>PLA2G6</i>	<i>PLCL1</i>	<i>PLIN4</i>	<i>PLXDC2</i>	<i>PLXNA4</i>	<i>POTEG</i>
<i>PPFIBP1</i>	<i>PPP6C</i>	<i>PRUNE2</i>	<i>PSMB1</i>	<i>PSMB2</i>	<i>PSMB5</i>	<i>PSMD1</i>	<i>PSMD2</i>
<i>PTCH1</i>	<i>PTEN</i>	<i>PTGFRN</i>	<i>PTPN11</i>	<i>PTPRF</i>	<i>RAB40A</i>	<i>RAC1</i>	<i>RAF1</i>
<i>RARA</i>	<i>RARB</i>	<i>RARG</i>	<i>RB1</i>	<i>RET</i>	<i>RNASE1</i>	<i>RNF43</i>	<i>ROCK2</i>
<i>ROR1</i>	<i>ROS1</i>	<i>RPS6KB1</i>	<i>RRP9</i>	<i>RUNX1</i>	<i>RXRA</i>	<i>RXRB</i>	<i>RXRG</i>
<i>SACS</i>	<i>SCN2A</i>	<i>SCP2</i>	<i>SCRT1</i>	<i>SDHB</i>	<i>SEC31A</i>	<i>SERPINB3</i>	<i>SETDB1</i>
<i>SF3B1</i>	<i>SHH</i>	<i>SHOC2</i>	<i>SLC12A8</i>	<i>SLC22A1</i>	<i>SLC22A2</i>	<i>SLC2A1</i>	<i>SLC31A1</i>
<i>SLC34A2</i>	<i>SLC44A4</i>	<i>SLC45A3</i>	<i>SLC4A1AP</i>	<i>SLC4A7</i>	<i>SLC6A1</i>	<i>SLCO1B1</i>	<i>SLCO1B3</i>
<i>SMAD4</i>	<i>SMARCA4</i>	<i>SMARCB1</i>	<i>SMC5</i>	<i>SMG1</i>	<i>SMO</i>	<i>SMPD4</i>	<i>SNCAIP</i>
<i>SNX31</i>	<i>SOD2</i>	<i>SOGA1</i>	<i>SOS1</i>	<i>SPDYE1</i>	<i>SPEN</i>	<i>SPG11</i>	<i>SPRED1</i>
<i>SPTAN1</i>	<i>SRC</i>	<i>STAG2</i>	<i>STARD13</i>	<i>STK11</i>	<i>STK19</i>	<i>SUFU</i>	<i>SYNE1</i>
<i>TANC1</i>	<i>TAS2R38</i>	<i>TBL1XR1</i>	<i>TBX3</i>	<i>TBXA2R</i>	<i>TCEB3C</i>	<i>TCEB3CL</i>	<i>TEKT4</i>
<i>TET2</i>	<i>TIAL1</i>	<i>TM9SF2</i>	<i>TMEM51</i>	<i>TNFRSF25</i>	<i>TP53</i>	<i>TRIO</i>	<i>TRPV4</i>
<i>TRRAP</i>	<i>TSHZ3</i>	<i>TTC22</i>	<i>TUBA3E</i>	<i>TXNRD1</i>	<i>TYK2</i>	<i>UBE3B</i>	<i>UBE4A</i>
<i>UBTD2</i>	<i>UGT1A1</i>	<i>UGT2B11</i>	<i>UNC13A</i>	<i>UQCRFS1</i>	<i>USP19</i>	<i>USP4</i>	<i>USP6NL</i>
<i>VHL</i>	<i>VPS41</i>	<i>WASF3</i>	<i>WHSC1</i>	<i>WT1</i>	<i>WWP1</i>	<i>XIRP2</i>	<i>YES1</i>
<i>YIPF1</i>	<i>ZC3H7A</i>	<i>ZKSCAN2</i>	<i>ZMYM3</i>	<i>ZMYM4</i>	<i>ZNF141</i>	<i>ZNF195</i>	<i>ZNF208</i>
<i>ZNF217</i>	<i>ZNF236</i>	<i>ZNF285</i>	<i>ZNF429</i>	<i>ZNF479</i>	<i>ZNF528</i>	<i>ZNF534</i>	<i>ZNF587B</i>
<i>ZNF611</i>	<i>ZNF676</i>	<i>ZNF727</i>	<i>ZNF728</i>	<i>ZNF729</i>	<i>ZNF735</i>	<i>ZNF750</i>	<i>ZNF765</i>
<i>ZNF816</i>	<i>ZNF85</i>	<i>ZSWIM8</i>					

Regions

Whole UTR/exon/intron of *GAS6* (chr13:113819854-113864930 @ hg38, 44 kb)

Whole UTR/exon/intron of *FOXA1* and 200-kb upstream (chr14:37585355-37843301 @ hg38, 256 kb)

51 UTR, untranslated region

52

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Table S3. Nonsynonymous mutations identified by whole-genome sequencing

UPN	Gene	Reference	Nucleic acid change	Amino acid change	VAF	ClinVar	COSMIC
1	<i>ADAM11</i>	NM_002390	c.826-2A>C	(exon 11)	0.12	-	-
1	<i>AHNAK2</i>	NM_138420	c.6181G>C	p.V2061L	0.16	-	-
1	<i>BEAN1</i>	NM_001178020	c.244_245insTTTTTTAATG	p.H82Lfs*3	0.10	-	-
1	<i>CALML6</i>	NM_138705	c.433G>A	p.E145K	0.10	-	-
1	<i>CCDC141</i>	NM_173648	c.4018G>T	p.G1340C	0.10	-	-
1	<i>CNOT1</i>	NM_206999	c.4635A>T	p.L1545F	0.10	-	-
1	<i>CORIN</i>	NM_006587	c.618-3T>-	(exon 5)	0.11	-	-
1	<i>CORO2B</i>	NM_006091	c.881A>G	p.N294S	0.11	-	-
1	<i>EMX1</i>	NM_004097	c.241A>C	p.T81P	0.11	-	-
1	<i>F10</i>	NM_000504	c.626A>C	p.D209A	0.15	-	-
1	<i>FAM47C</i>	NM_001013736	c.1373G>A	p.R458Q	0.10	-	-
1	<i>G2E3</i>	NM_017769	c.1393G>T	p.G465C	0.11	-	-
1	<i>IQSEC2</i>	NM_001111125	c.3574T>G	p.Y1192D	0.18	-	-
1	<i>LLGL2</i>	NM_004524	c.1732C>T	p.Q578*	0.12	-	-
1	<i>LOC100129520</i>	NM_001195272	c.1148C>T	p.P383L	0.12	-	-
1	<i>MDK</i>	NM_002391	c.406+2T>G	(exon 4)	0.16	-	-
1	<i>MUC2</i>	NM_002457	c.4370C>T	p.P1457L	0.08	-	-
1	<i>MYBPC1</i>	NM_206820	c.2290G>T	p.A764S	0.07	-	-
1	<i>MYCBP2</i>	NM_015057	c.2434A>C	p.K812Q	0.16	-	-
1	<i>NFATC3</i>	NM_004555	c.91T>G	p.S31A	0.14	-	-
1	<i>NOS2</i>	NM_000625	c.2081T>C	p.I694T	0.10	-	-
1	<i>NRAS</i>	NM_002524	c.430A>C	p.T144P	0.10	-	-
1	<i>NUMB</i>	NM_003744	c.625G>A	p.E209K	0.12	-	-
1	<i>OBSCN</i>	NM_052843	c.11536T>C	p.Y3846H	0.09	-	-
1	<i>OR1L4</i>	NM_001005235	c.143T>C	p.L48P	0.11	-	-
1	<i>PAMR1</i>	NM_015430	c.509G>C	p.S170T	0.07	-	-
1	<i>PCM1</i>	NM_006197	c.1471C>A	p.Q491K	0.08	-	-
1	<i>PDSS2</i>	NM_020381	c.94A>C	p.I32L	0.13	-	-
1	<i>PLCL1</i>	NM_006226	c.2161C>G	p.L721V	0.11	-	-
1	<i>PRAMEF1</i>	NM_023013	c.41G>T	p.G14V	0.11	-	-
1	<i>PRAMEF6</i>	NM_001010889	c.1211T>G	p.I404R	0.15	-	-
1	<i>PRAMEF6</i>	NM_001010889	c.1222T>A	p.L408I	0.17	-	-
1	<i>PTPRH</i>	NM_002842	c.2227C>T	p.H743Y	0.14	-	-
1	<i>PTPRQ</i>	NM_001145026	c.3368G>A	p.G1123E	0.09	-	-
1	<i>RIMS1</i>	NM_014989	c.2054T>C	p.V685A	0.13	-	-
1	<i>SEPT8</i>	NM_015146	c.528C>G	p.D176E	0.10	-	-
1	<i>SOX8</i>	NM_014587	c.982C>A	p.P328T	0.15	-	-
1	<i>STXBP5</i>	NM_001127715	c.2205G>T	p.K735N	0.10	-	-
1	<i>TMCO4</i>	NM_181719	c.856T>G	p.W286G	0.12	-	-

1	<i>TRIM43</i>	NM_138800	c.1188_1189delAG	p.K398Tfs*10	0.14	-	-
1	<i>ZNF517</i>	NM_213605	c.1094A>C	p.D365A	0.11	-	-
2	<i>ABCA10</i>	NM_080282	c.2456C>T	p.P819L	0.18	-	-
2	<i>ADAMTS10</i>	NM_030957	c.457G>A	p.E153K	0.19	-	-
2	<i>APOE</i>	NM_000041	c.371C>T	p.A124V	0.29	-	-
2	<i>APOL5</i>	NM_030642	c.1141C>T	p.L381F	0.23	-	-
2	<i>C10orf90</i>	NM_001004298	c.193G>C	p.E65Q	0.20	-	-
2	<i>C14orf105</i>	NM_001283056	c.653-4T>-	(exon 6)	0.19	-	-
2	<i>CCDC168</i>	NM_001146197	c.8479G>C	p.E2827Q	0.20	-	-
2	<i>CCT6A</i>	NM_001762	c.1065+1G>C	(exon 9)	0.22	-	-
2	<i>CD300C</i>	NM_006678	c.494G>C	p.R165T	0.22	-	-
2	<i>CFAP54</i>	NM_001306084	c.387G>C	p.K129N	0.36	-	-
2	<i>CRYBG3</i>	NM_153605	c.5167G>A	p.E1723K	0.36	-	-
2	<i>DST</i>	NM_001144769	c.170C>T	p.S57L	0.15	-	-
2	<i>EQTN</i>	NM_020641	c.280C>G	p.L94V	0.41	-	-
2	<i>FAM129B</i>	NM_022833	c.587A>G	p.N196S	0.16	-	-
2	<i>FASN</i>	NM_004104	c.1771G>A	p.D591N	0.26	-	-
2	<i>FASTK</i>	NM_006712	c.1039+4C>G	(exon 5)	0.23	-	-
2	<i>FCAR</i>	NM_002000	c.460G>C	p.D154H	0.22	-	-
2	<i>FOXA1</i>	NM_001310135	g.38064406G>A	-	0.20	-	-
2	<i>FREM3</i>	NM_001168235	c.4063C>A	p.L1355I	0.25	-	-
2	<i>FUT9</i>	NM_006581	c.68T>C	p.M23T	0.15	-	-
2	<i>GGT1</i>	NM_013421	c.1673C>T	p.S558L	0.13	-	-
2	<i>GLDN</i>	NM_181789	c.295G>A	p.E99K	0.28	-	-
2	<i>GUCY1B3</i>	NM_000857	c.565G>C	p.E189Q	0.25	-	-
2	<i>HDGFRP3</i>	NM_016073	c.607-5T>-	(exon 6)	0.23	-	-
2	<i>HIST1H2BC</i>	NM_003526	c.72G>C	p.K24N	0.25	-	-
2	<i>HORMAD1</i>	NM_032132	c.872-5T>-	(exon 12)	0.34	-	-
2	<i>INTS7</i>	NM_015434	c.1816-5->T	(exon 14)	0.21	-	-
2	<i>LAMA2</i>	NM_000426	c.6307G>C	p.E2103Q	0.17	-	-
2	<i>MOCS1</i>	NM_005943	c.250+3G>A	(exon 1)	0.24	-	-
2	<i>MUC12</i>	NM_001164462	c.994C>A	p.P332T	0.20	-	-
2	<i>MUM1</i>	NM_032853	c.2020G>A	p.E674K	0.20	-	-
2	<i>MYPOP</i>	NM_001012643	c.337G>C	p.E113Q	0.22	-	-
2	<i>NDUFS5</i>	NM_004552	c.70G>A	p.E24K	0.25	-	-
2	<i>NFATC4</i>	NM_001288802	c.61G>C	p.E21Q	0.18	-	-
2	<i>NMRK2</i>	NM_170678	c.436G>A	p.D146N	0.24	-	-
2	<i>NOTCH1</i>	NM_017617	c.1904-3C>A	(exon 12)	0.18	-	-
2	<i>NR4A3</i>	NM_006981	c.1299T>G	p.H433Q	0.22	-	-
2	<i>NRIP1</i>	NM_003489	c.2650C>A	p.H884N	0.33	-	-
2	<i>PEAR1</i>	NM_001080471	c.133C>T	p.R45C	0.16	-	-
2	<i>PIK3CA</i>	NM_006218	c.241G>A	p.E81K	0.21	-	COSM271871

2	<i>POM121</i>	NM_172020	c.822delG	p.K275Rfs*59	0.23	-	-
2	<i>PRUNE2</i>	NM_015225	c.9051-3T>-	(exon 16)	0.16	-	-
2	<i>RGPD3</i>	NM_001144013	c.2107G>A	p.E703K	0.30	-	-
2	<i>RPTOR</i>	NM_020761	c.3670G>A	p.D1224N	0.16	-	-
2	<i>SALL1</i>	NM_002968	c.1763dupC	p.G589Rfs*6	0.26	-	-
2	<i>SLC18A3</i>	NM_003055	c.770_772delTGT	p.L257del	0.20	-	-
2	<i>SPATA13</i>	NM_001166271	c.755G>A	p.S252N	0.25	-	-
2	<i>SRL</i>	NM_001098814	c.530A>C	p.E177A	0.36	-	-
2	<i>SRSF10</i>	NM_054016	c.749G>A	p.R250K	0.17	-	-
2	<i>TAP1</i>	NM_000593	c.1039C>T	p.R347W	0.31	-	-
2	<i>THSD7A</i>	NM_015204	c.1630C>T	p.R544C	0.27	-	-
2	<i>TMEM182</i>	NM_144632	c.94C>G	p.L32V	0.23	-	-
2	<i>TMPRSS13</i>	NM_001077263	c.1136G>A	p.W379*	0.18	-	-
2	<i>TNK2</i>	NM_005781	c.2121C>G	p.F707L	0.22	-	-
2	<i>TP53BP2</i>	NM_005426	c.1973C>G	p.S658*	0.24	-	-
2	<i>TRIOBP</i>	NM_001039141	c.31G>A	p.E11K	0.21	-	-
2	<i>UBR1</i>	NM_174916	c.463G>C	p.E155Q	0.25	-	-
2	<i>USE1</i>	NM_018467	c.232-1G>C	(exon 4)	0.17	-	-
2	<i>ZBED5</i>	NM_021211	c.116T>C	p.L39S	0.17	-	-
2	<i>ZNF286B</i>	NM_001145045	c.1540C>G	p.Q514E	0.15	-	-
2	<i>ZNF292</i>	NM_015021	c.266G>A	p.R89Q	0.24	-	-

55 VAF, variant allele frequency; COSMIC, Catalogue Of Somatic Mutations In Cancer

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Table S4. Nonsynonymous mutations identified by whole-exome sequencing

UP N	Gene	Reference	Nucleic acid change	Amino acid change	VAF	ClinVar	COSMIC
12	<i>CNNM4</i>	NM_020184	c.727C>T	p.L243F	0.14	-	-
12	<i>EPPK1</i>	NM_031308	c.2521T>C	p.Y841H	0.07	-	-
12	<i>ILKAP</i>	NM_030768	c.714+1G>A	(exon 8)	0.21	-	-
12	<i>INO80D</i>	NM_017759	c.1638delA	p.K546Nfs*49	0.14	-	-
12	<i>PANX2</i>	NM_052839	c.508G>A	p.E170K	0.06	-	-
12	<i>STARD13</i>	NM_052851	c.2032G>A	p.V678I	0.07	-	-
12	<i>TNFRSF25</i>	NM_003790	c.998C>T	p.P333L	0.2	-	-
12	<i>TUBA3E</i>	NM_207312	c.322T>C	p.Y108H	0.05	-	-
13	<i>KRTAP10-2</i>	NM_198693	c.233C>T	p.S78L	0.18	-	-
13	<i>POTEG</i>	NM_00100535 6	c.35C>T	p.S12F	0.14	-	-
14	<i>ARHGEF16</i>	NM_014448	c.544G>A	p.E182K	0.14	-	-
14	<i>ARMCX4</i>	NM_00125615 5	c.3155C>A	p.S1052Y	0.37	-	-
14	<i>ATN1</i>	NM_001940	c.496C>A	p.P166T	0.13	-	-
14	<i>ATN1</i>	NM_001940	c.2558C>G	p.S853C	0.24	-	-
14	<i>BCL11A</i>	NM_018014	c.914C>T	p.P305L	0.18	-	-
14	<i>C16orf82</i>	NM_00114554 5	c.43G>C	p.E15Q	0.24	-	-
14	<i>C1orf127</i>	NM_00117075 4	c.1924G>A	p.D642N	0.13	-	-
14	<i>C1QL1</i>	NM_006688	c.478G>A	p.D160N	0.26	-	-
14	<i>CAMTA1</i>	NM_015215	c.1636G>C	p.E546Q	0.26	-	-
14	<i>CHD2</i>	NM_001271	c.3004G>T	p.A1002S	0.17	-	COSM370682 7
14	<i>CHRM5</i>	NM_012125	c.527G>A	p.R176Q	0.17	-	-
14	<i>COBLL1</i>	NM_014900	c.2462C>G	p.S821C	0.13	-	-
14	<i>CRYBA4</i>	NM_001886	c.74G>A	p.R25Q	0.17	-	-
14	<i>CYP27B1</i>	NM_000785	c.40C>T	p.R14C	0.25	-	-
14	<i>CYP2E1</i>	NM_000773	c.1478C>T	p.S493L	0.2	-	-
14	<i>DCAF10</i>	NM_024345	c.1172C>T	p.S391L	0.25	-	-
14	<i>DLC1</i>	NM_006094	c.2028C>A	p.F676L	0.44	-	-
14	<i>ELOVL2</i>	NM_017770	c.445C>T	p.H149Y	0.2	-	-
14	<i>EPHA7</i>	NM_004440	c.962C>A	p.S321Y	0.15	-	-
14	<i>EPPK1</i>	NM_031308	c.5620C>T	p.R1874C	0.34	-	-
14	<i>ERP29</i>	NM_006817	c.579G>C	p.E193D	0.17	-	-
14	<i>EYA1</i>	NM_000503	c.1318C>T	p.R440W	0.16	-	-
14	<i>FITM1</i>	NM_203402	c.196delT	p.F66Lfs*24	0.2	-	-

14	<i>FLG</i>	NM_002016	c.9896G>C	p.G3299A	0.18	-	-
14	<i>GLUD1</i>	NM_005271	c.283G>A	p.E95K	0.28	-	-
14	<i>GPC5</i>	NM_004466	c.232C>A	p.Q78K	0.23	-	-
14	<i>GPRC6A</i>	NM_148963	c.2393T>G	p.F798C	0.13	-	-
14	<i>HIST1H2BB</i>	NM_021062	c.72G>C	p.K24N	0.18	-	COSM116907 1
14	<i>HPS5</i>	NM_007216	c.244C>G	p.R82G	0.19	-	-
14	<i>HSPG2</i>	NM_005529	c.9352C>T	p.P3118S	0.18	-	-
14	<i>IGSF10</i>	NM_178822	c.4869G>C	p.K1623N	0.2	-	-
14	<i>IL1RN</i>	NM_000577	c.403G>A	p.D135N	0.19	-	-
14	<i>KDM7A</i>	NM_030647	c.379C>T	p.R127C	0.19	-	COSM286115 3
14	<i>KIAA0391</i>	NM_014672	c.1072A>G	p.I358V	0.25	-	-
14	<i>KNG1</i>	NM_000893	c.181G>A	p.E61K	0.24	-	-
14	<i>KSR2</i>	NM_173598	c.1402A>G	p.T468A	0.23	-	-
14	<i>LRCH2</i>	NM_020871	c.1522G>C	p.E508Q	0.34	-	-
14	<i>MACF1</i>	NM_012090	c.12435G>C	p.E4145D	0.22	-	-
14	<i>MED24</i>	NM_014815	c.700G>C	p.E234Q	0.17	-	COSM147952 2
14	<i>MGA</i>	NM_00108054 1	c.2137G>C	p.D713H	0.33	-	-
14	<i>MRTO4</i>	NM_016183	c.131C>G	p.S44C	0.14	-	-
14	<i>MTR</i>	NM_000254	c.3632C>T	p.P1211L	0.14	-	-
14	<i>MUC12</i>	NM_00116446 2	c.5866C>A	p.P1956T	0.16	-	-
14	<i>NACA</i>	NM_00111320 2	c.10G>A	p.E4K	0.21	-	-
14	<i>NACC2</i>	NM_144653	c.1362G>T	p.K454N	0.15	-	-
14	<i>NADK</i>	NM_023018	c.1315G>A	p.E439K	0.22	-	-
14	<i>NEFH</i>	NM_021076	c.2836G>C	p.E946Q	0.31	-	-
14	<i>NLRP8</i>	NM_176811	c.676G>C	p.E226Q	0.18	-	-
14	<i>OPCML</i>	NM_002545	c.665-5C>T	(exon 5)	0.22	-	-
14	<i>PAGE1</i>	NM_003785	c.55T>A	p.S19T	0.4	-	-
14	<i>PCSK1</i>	NM_000439	c.1813C>T	p.R605C	0.25	-	-
14	<i>PIK3CA</i>	NM_006218	c.1633G>A	p.E545K	0.21	RCV00001463 1	COSM763
14	<i>PIK3CA</i>	NM_006218	c.3172A>C	p.I1058L	0.14	-	COSM479744
14	<i>PITPNC1</i>	NM_012417	c.856G>A	p.E286K	0.24	-	-
14	<i>PLXNA4</i>	NM_020911	c.5513G>A	p.R1838Q	0.12	-	-
14	<i>PTGFRN</i>	NM_020440	c.359C>T	p.S120L	0.19	-	-
14	<i>PTPRF</i>	NM_002840	c.2356G>A	p.G786S	0.32	-	-
14	<i>RNASE1</i>	NM_002933	c.112C>T	p.R38W	0.21	-	-

14	<i>RNF43</i>	NM_017763	c.476G>A	p.W159*	0.17	-	-
14	<i>SACS</i>	NM_014363	c.9002T>C	p.I3001T	0.33	-	-
14	<i>SCN2A</i>	NM_021007	c.3906C>G	p.I1302M	0.15	-	-
14	<i>SCRT1</i>	NM_031309	c.844G>A	p.G282S	0.25	-	-
14	<i>SERPINB3</i>	NM_006919	c.1084G>C	p.E362Q	0.22	-	-
14	<i>SLC4A7</i>	NM_003615	c.1010C>T	p.S337L	0.17	-	-
14	<i>SMARCB1</i>	NM_003073	c.744_745insCCCA	p.D251Hfs*31	0.37	-	-
14	<i>SMPD4</i>	NM_017751	c.803A>G	p.Y268C	0.17	-	-
14	<i>TEKT4</i>	NM_144705	c.325A>G	p.M109V	0.23	-	-
14	<i>UBE4A</i>	NM_004788	c.562-1G>C	(exon 6)	0.17	-	-
14	<i>UGT2B11</i>	NM_001073	c.1201G>C	p.D401H	0.13	-	-
14	<i>USP6NL</i>	NM_014688	c.1882C>T	p.H628Y	0.26	-	-
14	<i>WWP1</i>	NM_007013	c.1807G>C	p.E603Q	0.15	-	-
14	<i>ZC3H7A</i>	NM_014153	c.1144C>T	p.L382F	0.25	-	-
14	<i>ZNF236</i>	NM_007345	c.821G>A	p.R274Q	0.19	-	-
14	<i>ZNF429</i>	NM_00100141 5	c.1649C>G	p.S550*	0.17	-	-
14	<i>ZNF534</i>	NM_00114393 8	c.1135G>C	p.E379Q	0.21	-	-
14	<i>ZNF727</i>	NM_00115952 2	c.317G>C	p.C106S	0.15	-	-
15	<i>C8orf44</i>	NM_019607	c.188A>G	p.K63R	0.4	-	-
15	<i>DYNAP</i>	NM_173629	c.374T>C	p.M125T	0.31	-	-
16	<i>AHNAK</i>	NM_001620	c.5341G>A	p.V1781I	0.14	-	-
16	<i>C14orf159</i>	NM_024952	c.497T>C	p.M166T	0.14	-	-
16	<i>CCDC116</i>	NM_152612	c.409C>T	p.R137C	0.13	-	-
16	<i>CCDC40</i>	NM_00124334 2	c.2884G>A	p.G962R	0.08	-	-
16	<i>CDK20</i>	NM_00117064 0	c.581C>T	p.T194M	0.06	-	-
16	<i>CLPS</i>	NM_00125259 7	c.1A>G	p.M1V	0.36	-	-
16	<i>DPCR1</i>	NM_080870	c.2878A>G	p.K960E	0.06	-	-
16	<i>DUSP14</i>	NM_007026	c.487C>T	p.R163C	0.11	-	-
16	<i>GPR179</i>	NM_00100433 4	c.235C>T	p.R79C	0.17	-	-
16	<i>JARID2</i>	NM_004973	c.2416G>A	p.V806I	0.21	-	-
16	<i>LMTK3</i>	NM_00108043 4	c.2951_2952insG	p.E985Rfs*506	0.1	-	-
16	<i>LRR1</i>	NM_152329	c.986G>A	p.R329Q	0.26	-	-
16	<i>PCDHB4</i>	NM_018938	c.1417A>C	p.S473R	0.41	-	-
16	<i>PCNX</i>	NM_014982	c.5567G>A	p.R1856H	0.08	-	-

16	<i>PLXDC2</i>	NM_032812	c.1268C>T	p.T423I	0.31	-	-
16	<i>SLC6A1</i>	NM_003042	c.650C>T	p.T217M	0.17	-	-
16	<i>TBXA2R</i>	NM_201636	c.1091C>T	p.A364V	0.28	-	-
16	<i>TCEB3C</i>	NM_145653	c.1150G>A	p.A384T	0.04	-	-
16	<i>UNC13A</i>	NM_00108042 1	c.1483C>T	p.R495C	0.13	-	-
16	<i>ZNF285</i>	NM_152354	c.254C>G	p.T85S	0.2	-	-
17	<i>ADGRB3</i>	NM_001704	c.3713C>T	p.S1238L	0.12	-	-
17	<i>AGAP2</i>	NM_00112277 2	c.788G>A	p.R263Q	0.1	-	-
17	<i>ATP8B2</i>	NM_020452	c.2188G>C	p.E730Q	0.09	-	-
17	<i>BAHD1</i>	NM_014952	c.1021G>C	p.E341Q	0.09	-	-
17	<i>CAST</i>	NM_173060	c.1618-3C>T	(exon 23)	0.1	-	-
17	<i>CHGB</i>	NM_001819	c.745C>T	p.H249Y	0.15	-	-
17	<i>CTNND2</i>	NM_001332	c.2654G>A	p.R885Q	0.12	-	-
17	<i>CYGB</i>	NM_134268	c.164C>T	p.S55L	0.08	-	-
17	<i>DDX53</i>	NM_182699	c.1189G>A	p.V397M	0.08	-	-
17	<i>HOXA3</i>	NM_030661	c.732G>A	p.M244I	0.09	-	-
17	<i>IDI1</i>	NM_004508	c.670G>A	p.E224K	0.27	-	-
17	<i>IL6ST</i>	NM_002184	c.2245T>A	p.S749T	0.06	-	-
17	<i>IQSEC1</i>	NM_014869	c.796G>A	p.E266K	0.13	-	-
17	<i>MOV10L1</i>	NM_018995	c.3352G>A	p.D1118N	0.12	-	-
17	<i>MUC16</i>	NM_024690	c.33380C>G	p.P11127R	0.1	-	-
17	<i>OR4N5</i>	NM_00100472 4	c.329G>C	p.G110A	0.06	-	-
17	<i>OR5M10</i>	NM_00100474 1	c.512C>G	p.S171C	0.08	-	-
17	<i>PIK3CA</i>	NM_006218	c.1624G>C	p.E542Q	0.08	-	COSM17442
17	<i>PLCL1</i>	NM_006226	c.482C>T	p.S161F	0.07	-	-
17	<i>UBTD2</i>	NM_152277	c.374C>T	p.P125L	0.23	-	-
17	<i>ZNF611</i>	NM_030972	c.949T>A	p.C317S	0.13	-	-
17	<i>ZNF728</i>	NM_00126771 6	c.505G>A	p.G169R	0.11	-	-
18	<i>AQP7</i>	NM_001170	c.101T>A	p.V34E	0.09	-	-
18	<i>HLA-B</i>	NM_005514	c.121C>A	p.R41S	0.31	-	-
18	<i>KANK2</i>	NM_015493	c.530C>T	p.P177L	0.33	-	-
18	<i>MUC2</i>	NM_002457	c.2770A>C	p.M924L	0.22	-	-
18	<i>ROR1</i>	NM_005012	c.2788G>A	p.E930K	0.06	-	-
18	<i>ZNF195</i>	NM_00124284 1	c.338A>G	p.N113S	0.26	-	-
19	<i>CYP26B1</i>	NM_019885	c.1190G>A	p.R397Q	0.14	-	-
19	<i>FUT7</i>	NM_004479	c.335G>A	p.R112Q	0.29	-	-

19	<i>IGSF22</i>	NM_173588	c.3247G>A	p.E1083K	0.21	-	-
19	<i>JAG2</i>	NM_002226	c.3702C>A	p.Y1234*	0.16	-	-
19	<i>MYRF</i>	NM_013279	c.1645G>C	p.G549R	0.12	-	-
19	<i>PCDHA9</i>	NM_014005	c.1061C>T	p.T354M	0.12	-	-
19	<i>PIK3CA</i>	NM_006218	c.1637A>G	p.Q546R	0.12	RCV00003867 2	COSM12459
19	<i>SLCO1B3</i>	NM_019844	c.1510T>A	p.C504S	0.15	-	-
19	<i>SOGA1</i>	NM_080627	c.2237T>C	p.F746S	0.13	-	-
19	<i>TTN</i>	NM_003319	c.31156C>T	p.R10386C	0.16	-	-
19	<i>UQCRFS1</i>	NM_006003	c.397G>A	p.V133I	0.13	-	-
21	<i>KRTAP4-11</i>	NM_033059	c.249C>G	p.S83R	0.26	-	-
21	<i>NPIP5</i>	NM_00113586 5	c.1549C>T	p.R517C	0.05	-	-
21	<i>SPDYE1</i>	NM_175064	c.791A>G	p.H264R	0.1	-	-
39	<i>ACOT2</i>	NM_006821	c.620G>C	p.R207P	0.23	-	-
39	<i>ADRB1</i>	NM_000684	c.790C>T	p.R264C	0.21	-	-
39	<i>ALDH3B2</i>	NM_000695	c.10G>A	p.E4K	0.2	-	-
39	<i>BCL11B</i>	NM_00128223 8	c.2078C>T	p.S693L	0.17	-	-
39	<i>BCL9</i>	NM_004326	c.2518dupG	p.L841Pfs*21	0.08	-	-
39	<i>C10orf71</i>	NM_00113519 6	c.832G>C	p.E278Q	0.09	-	-
39	<i>CACNA1E</i>	NM_00120529 4	c.5082+3G>A	(exon 36)	0.13	-	-
39	<i>CASP8AP2</i>	NM_00113766 7	c.1600G>C	p.D534H	0.46	-	-
39	<i>CCDC121</i>	NM_00114268 3	c.848T>C	p.I283T	0.14	-	-
39	<i>CHRNA10</i>	NM_00130303 4	c.445G>C	p.E149Q	0.12	-	-
39	<i>CHRNA7</i>	NM_000746	c.1066G>A	p.A356T	0.05	-	-
39	<i>CLDN18</i>	NM_016369	c.139G>A	p.E47K	0.19	-	-
39	<i>COQ4</i>	NM_016035	c.567G>C	p.Q189H	0.25	-	-
39	<i>DHX35</i>	NM_00119080 9	c.1963C>T	p.Q655X	0.28	-	-
39	<i>DLGAP1</i>	NM_00100380 9	c.811G>A	p.G271R	0.24	-	-
39	<i>EPB41L3</i>	NM_00128153 3	c.475G>A	p.E159K	0.48	-	-
39	<i>FAM160A1</i>	NM_00110997 7	c.1621G>A	p.E541K	0.17	-	-
39	<i>FASN</i>	NM_004104	c.6060_6062delCTC	p.2020delS	0.12	-	-

39	<i>FCRL1</i>	NM_00115939 7	c.733G>T	p.E245X	0.17	-	-
39	<i>GAREM1</i>	NM_00124240 9	c.1402C>T	p.L468F	0.23	-	-
39	<i>GON4L</i>	NM_00128285 6	c.4960C>T	p.Q1654X	0.18	-	-
39	<i>HAUS4</i>	NM_00116626 9	c.196C>T	p.Q66X	0.16	-	-
39	<i>HPCAL4</i>	NM_00128239 6	c.163-5C>-	(exon 4)	0.32	-	-
39	<i>HTRA1</i>	NM_002775	c.1297G>A	p.V433I	0.21	-	-
39	<i>KIAA0754</i>	NM_015038	c.4118dupT	p.M1374Hfs*35	0.16	-	-
39	<i>KIAA1549</i>	NM_00116466 5	c.2179C>T	p.L727F	0.27	-	-
39	<i>KIAA1958</i>	NM_133465	c.1513T>G	p.S505A	0.1	-	-
39	<i>LILRA2</i>	NM_00129027 0	c.1166C>G	p.S389C	0.07	-	-
39	<i>LONP1</i>	NM_004793	c.1686-1G>A	(exon 11)	0.24	-	-
39	<i>METTL10</i>	NM_212554	c.8C>G	p.S3W	0.15	-	-
39	<i>MRM1</i>	NM_024864	c.80G>A	p.G27E	0.27	-	-
39	<i>MTCL1</i>	NM_015210	c.3328G>A	p.E1110K	0.09	-	-
39	<i>MYO10</i>	NM_012334	c.3079G>A	p.D1027N	0.08	-	-
39	<i>MYO7B</i>	NM_00108052 7	c.6037G>A	p.E2013K	0.18	-	-
39	<i>NDUFS8</i>	NM_002496	c.412C>T	p.R138C	0.07	-	-
39	<i>NEUROD1</i>	NM_002500	c.223G>A	p.E75K	0.2	-	-
39	<i>NOC2L</i>	NM_015658	c.2221G>A	p.E741K	0.42	-	-
39	<i>OR2T8</i>	NM_00100552 2	c.81G>T	p.M27I	0.26	-	-
39	<i>OVGP1</i>	NM_002557	c.1555C>T	p.P519S	0.18	-	-
39	<i>PCDHA9</i>	NM_014005	c.544G>A	p.V182M	0.2	-	-
39	<i>PEG3</i>	NM_00114618 6	c.4486C>G	p.Q1496E	0.17	-	-
39	<i>PLPP6</i>	NM_203453	c.611C>T	p.S204L	0.14	-	-
39	<i>PMFBP1</i>	NM_031293	c.1213G>A	p.E405K	0.29	-	-
39	<i>POM121L12</i>	NM_182595	c.308C>T	p.P103L	0.11	-	-
39	<i>PRDM13</i>	NM_021620	c.707C>T	p.S236L	0.26	-	-
39	<i>PTPRT</i>	NM_007050	c.886G>A	p.E296K	0.18	-	-
39	<i>RNF213</i>	NM_00125607 1	c.13115T>G	p.L4372R	0.23	-	-
39	<i>RPS6KA2</i>	NM_021135	c.1901C>T	p.S634F	0.26	-	-
39	<i>RRBP1</i>	NM_004587	c.710T>G	p.L237R	0.13	-	-

39	<i>RRP7A</i>	NM_015703	c.712C>A	p.L238M	0.12	-	-
39	<i>RTL1</i>	NM_00113488 8	c.2972G>A	p.R991Q	0.08	-	-
39	<i>SALL3</i>	NM_171999	c.3718C>T	p.Q1240X	0.12	-	-
39	<i>SETD7</i>	NM_00130619 9	c.264C>G	p.D88E	0.12	-	-
39	<i>SHISA7</i>	NM_00114517 6	c.976+3G>A	(exon 3)	0.22	-	-
39	<i>SOX12</i>	NM_006943	c.856G>A	p.E286K	0.16	-	-
39	<i>SPATA31D1</i>	NM_00100167 0	c.1890G>T	p.Q630H	0.21	-	-
39	<i>TBX4</i>	NM_018488	c.1055G>A	p.R352Q	0.22	-	-
39	<i>THEMIS2</i>	NM_00103947 7	c.160C>T	p.Q54X	0.18	-	-
39	<i>TNKS1BP1</i>	NM_033396	c.3718G>A	p.E1240K	0.09	-	-
39	<i>TNR</i>	NM_003285	c.3219G>C	p.L1073F	0.3	-	-
39	<i>TP53BP1</i>	NM_00114197 9	c.5764G>A	p.V1922M	0.21	-	-
39	<i>TUBB4B</i>	NM_006088	c.1234G>A	p.E412K	0.19	-	-
39	<i>TULP4</i>	NM_00100746 6	c.1972T>A	p.F658I	0.24	-	-
39	<i>USP40</i>	NM_018218	c.488G>A	p.G163E	0.23	-	-
39	<i>ZNF711</i>	NM_021998	c.1442G>C	p.C481S	0.57	-	-
39	<i>ZNRF3</i>	NM_00120699 8	c.2221G>A	p.E741K	0.13	-	-
39	<i>ZSCAN10</i>	NM_00128241 5	c.925G>A	p.A309T	0.22	-	-
40	<i>ABCA1</i>	NM_005502	c.2140G>A	p.D714N	0.15	-	-
40	<i>ADH1A</i>	NM_000667	c.944G>A	p.W315X	0.08	-	-
40	<i>AMIGO2</i>	NM_181847	c.174C>A	p.N58K	0.16	-	-
40	<i>BTD</i>	NM_000060	c.809C>T	p.T270I	0.12	-	-
40	<i>C19orf24</i>	NM_017914	c.80A>G	p.E27G	0.17	-	-
40	<i>C2orf69</i>	NM_153689	c.1012G>A	p.D338N	0.13	-	-
40	<i>CAPN7</i>	NM_014296	c.1810G>A	p.E604K	0.34	-	-
40	<i>CARNS1</i>	NM_020811	c.214G>A	p.D72N	0.16	-	-
40	<i>CDCA5</i>	NM_080668	c.404A>G	p.D135G	0.18	-	-
40	<i>CENPF</i>	NM_016343	c.3115C>G	p.L1039V	0.3	-	-
40	<i>CTTNBP2</i>	NM_033427	c.3201C>G	p.S1067R	0.23	-	-
40	<i>CUX1</i>	NM_00120254 3	c.2893G>T	p.E965X	0.21	-	-
40	<i>CYFIP1</i>	NM_014608	c.44A>G	p.D15G	0.15	-	-

40	<i>DDX54</i>	NM_00111132 2	c.1505C>T	p.S502L	0.17	-	-
40	<i>DFNB31</i>	NM_00117342 5	c.334G>A	p.E112K	0.17	-	-
40	<i>DPEP3</i>	NM_00112975 8	c.1414C>T	p.H472Y	0.14	-	-
40	<i>DPEP3</i>	NM_00112975 8	c.590C>G	p.S197C	0.17	-	-
40	<i>EP300</i>	NM_001429	c.3798G>A	p.W1266X	0.37	-	-
40	<i>EPHB2</i>	NM_00130919 2	c.1237G>A	p.G413S	0.16	-	-
40	<i>EPHB3</i>	NM_004443	c.514_515insGCCGT	p.V174Afs*175	0.09	-	-
40	<i>ERBB3</i>	NM_001982	c.2773G>A	p.E925K	0.12	-	-
40	<i>FAM179B</i>	NM_00130812 0	c.280G>A	p.E94K	0.16	-	-
40	<i>FCHO1</i>	NM_00116135 9	c.1189G>A	p.E397K	0.35	-	-
40	<i>FSIP2</i>	NM_173651	c.16366G>A	p.D5456N	0.15	-	-
40	<i>GNPAT</i>	NM_00131635 0	c.35C>T	p.S12F	0.1	-	-
40	<i>GPC2</i>	NM_152742	c.1487-1G>A	(exon 10)	0.19	-	-
40	<i>GPRASP2</i>	NM_00118487 5	c.1795G>A	p.E599K	0.21	-	-
40	<i>GSX2</i>	NM_133267	c.791C>T	p.T264M	0.15	-	-
40	<i>HELZ2</i>	NM_033405	c.3260A>T	p.Q1087L	0.07	-	-
40	<i>HIST1H1T</i>	NM_005323	c.148G>A	p.E50K	0.12	-	-
40	<i>HMX2</i>	NM_005519	c.707C>T	p.S236L	0.06	-	-
40	<i>HTR6</i>	NM_000871	c.1207G>A	p.E403K	0.2	-	-
40	<i>KDM3B</i>	NM_016604	c.4789G>A	p.E1597K	0.18	-	-
40	<i>KIR2DL1</i>	NM_014218	c.854G>A	p.R285K	0.11	-	-
40	<i>LENG1</i>	NM_024316	c.130G>A	p.E44K	0.24	-	-
40	<i>LMAN1</i>	NM_005570	c.304G>A	p.E102K	0.18	-	-
40	<i>LTBR</i>	NM_00127098 7	c.1111G>A	p.E371K	0.13	-	-
40	<i>LTN1</i>	NM_015565	c.1417G>A	p.E473K	0.23	-	-
40	<i>LTN1</i>	NM_015565	c.1426G>T	p.E476X	0.22	-	-
40	<i>MAGEB17</i>	NM_00127730 7	c.730G>A	p.E244K	0.14	-	-
40	<i>MC5R</i>	NM_005913	c.788C>G	p.S263C	0.18	-	-
40	<i>MLLT4</i>	NM_00129196 4	c.4996C>T	p.Q1666X	0.16	-	-

40	<i>MTMR1</i>	NM_00130614 5	c.961G>A	p.E321K	0.22	-	-
40	<i>MUC4</i>	NM_018406	c.4775C>T	p.S1592L	0.05	-	-
40	<i>NEURL1</i>	NM_004210	c.790G>A	p.E264K	0.13	-	-
40	<i>NOP56</i>	NM_006392	c.1256A>T	p.D419V	0.06	-	-
40	<i>NPIP5</i>	NM_00113586 5	c.1861C>T	p.Q621X	0.08	-	-
40	<i>NXF5</i>	NM_032946	c.42C>G	p.F14L	0.2	-	-
40	<i>OVOL1</i>	NM_004561	c.151G>A	p.V51M	0.12	-	-
40	<i>PARVG</i>	NM_00113760 5	c.616C>G	p.P206A	0.15	-	-
40	<i>PCLO</i>	NM_014510	c.6511G>A	p.E2171K	0.21	-	-
40	<i>PDE12</i>	NM_177966	c.998A>G	p.N333S	0.24	-	-
40	<i>PDE4DIP</i>	NM_00100281 1	c.101C>T	p.S34L	0.07	-	-
40	<i>PELI3</i>	NM_00124313 6	c.163G>A	p.D55N	0.14	-	-
40	<i>PMFBP1</i>	NM_031293	c.1720C>G	p.Q574E	0.17	-	-
40	<i>POLD1</i>	NM_00130863 2	c.1682G>A	p.R561Q	0.16	-	-
40	<i>RDH11</i>	NM_00125265 0	c.407C>G	p.S136W	0.16	-	-
40	<i>RNF123</i>	NM_022064	c.922C>T	p.R308W	0.21	-	-
40	<i>RREB1</i>	NM_00100369 8	c.364G>A	p.E122K	0.2	-	-
40	<i>SIK2</i>	NM_015191	c.2125G>C	p.E709Q	0.15	-	-
40	<i>SKIDA1</i>	NM_207371	c.1520A>C	p.D507A	0.22	-	-
40	<i>SLC9A7</i>	NM_00125729 1	c.714G>C	p.K238N	0.34	-	-
40	<i>SRCAP</i>	NM_006662	c.2518C>G	p.R840G	0.16	-	-
40	<i>SUFU</i>	NM_00117813 3	c.1099G>C	p.E367Q	0.18	-	-
40	<i>SUMF2</i>	NM_00104246 9	c.167G>A	p.R56K	0.18	-	-
40	<i>TINAGL1</i>	NM_00120441 4	c.112C>T	p.R38W	0.16	-	-
40	<i>TMEM63A</i>	NM_014698	c.127C>T	p.Q43X	0.31	-	-
40	<i>TRAPPC13</i>	NM_00109375 6	c.617C>T	p.S206L	0.36	-	-
40	<i>TTN</i>	NM_003319	c.75965C>G	p.S25322C	0.15	-	-
40	<i>TTN</i>	NM_003319	c.76951C>T	p.H25651Y	0.13	-	-
40	<i>TTN</i>	NM_003319	c.77291C>T	p.S25764L	0.23	-	-

40	<i>TTN</i>	NM_003319	c.78890C>G	p.S26297C	0.19	-	-
40	<i>UCP2</i>	NM_003355	c.334G>C	p.E112Q	0.17	-	-
40	<i>USP29</i>	NM_020903	c.1907G>C	p.R636T	0.22	-	-
40	<i>WDR24</i>	NM_032259	c.1362C>G	p.I454M	0.23	-	-
40	<i>WDR38</i>	NM_00127637 6	c.614C>T	p.S205F	0.16	-	-
40	<i>ZSWIM5</i>	NM_020883	c.3397C>T	p.R1133C	0.17	-	-
41	<i>ARPP21</i>	NM_00126761 7	c.2216C>T	p.P739L	0.1	-	-
41	<i>ERBB2</i>	NM_00128993 7	c.929C>T	p.S310F	0.11	-	COSM48358
42	<i>ADAMTS16</i>	NM_139056	c.2738C>T	p.T913I	0.22	-	-
42	<i>ADRA2C</i>	NM_000683	c.607G>A	p.G203S	0.07	-	-
42	<i>AGRP</i>	NM_001138	c.161C>T	p.T54I	0.23	-	-
42	<i>APOH</i>	NM_000042	c.460C>T	p.R154C	0.18	-	-
42	<i>ATXN2</i>	NM_00131012 3	c.2290C>T	p.Q764X	0.23	-	-
42	<i>CAD</i>	NM_00130607 9	c.6229G>A	p.D2077N	0.17	-	-
42	<i>CAD</i>	NM_00130607 9	c.6327G>A	p.M2109I	0.21	-	-
42	<i>CDKN2A</i>	NM_000077	c.245T>G	p.V82G	0.21	-	-
42	<i>CEACAM5</i>	NM_00129148 4	c.546T>A	p.N182K	0.2	-	-
42	<i>CEP295</i>	NM_033395	c.5978A>G	p.K1993R	0.27	-	-
42	<i>CHRNA4</i>	NM_000744	c.1034G>A	p.R345H	0.12	-	-
42	<i>COL22A1</i>	NM_152888	c.359C>T	p.A120V	0.11	-	-
42	<i>DDX53</i>	NM_182699	c.784C>G	p.L262V	0.43	-	-
42	<i>DOT1L</i>	NM_032482	c.448G>C	p.E150Q	0.15	-	-
42	<i>FARP1</i>	NM_005766	c.2527G>A	p.E843K	0.08	-	-
42	<i>GTF3C4</i>	NM_012204	c.523G>A	p.D175N	0.29	-	-
42	<i>GYPC</i>	NM_016815	c.86G>C	p.R29T	0.16	-	-
42	<i>HHIPL2</i>	NM_024746	c.562G>A	p.D188N	0.12	-	-
42	<i>HSF1</i>	NM_005526	c.718T>G	p.Y240D	0.13	-	-
42	<i>KIR3DL2</i>	NM_00124286 7	c.486G>T	p.E162D	0.05	-	-
42	<i>KLK4</i>	NM_00130296 1	c.223G>A	p.V75M	0.06	-	-
42	<i>LIPE</i>	NM_005357	c.3049C>A	p.R1017S	0.25	-	-
42	<i>LRRN2</i>	NM_201630	c.1721G>A	p.R574H	0.13	-	-
42	<i>MAD1L1</i>	NM_00130452 5	c.349G>A	p.D117N	0.12	-	-

42	<i>OR51S1</i>	NM_00100475 8	c.137C>G	p.S46C	0.21	-	-
42	<i>OR6F1</i>	NM_00100528 6	c.422C>T	p.S141L	0.25	-	-
42	<i>PLK1</i>	NM_005030	c.1711G>A	p.G571S	0.06	-	-
42	<i>PPP1R26</i>	NM_014811	c.1834G>A	p.E612K	0.27	-	-
42	<i>RAB40C</i>	NM_021168	c.552G>A	p.W184X	0.09	-	-
42	<i>SH2D6</i>	NM_198482	c.490C>G	p.R164G	0.1	-	-
42	<i>SLC18A3</i>	NM_003055	c.358G>A	p.E120K	0.17	-	-
42	<i>SMAD3</i>	NM_00114510 4	c.654G>T	p.Q218H	0.14	-	-
42	<i>SMYD4</i>	NM_052928	c.892C>T	p.R298X	0.14	-	-
42	<i>SNAP25</i>	NM_003081	c.531G>C	p.Q177H	0.37	-	-
42	<i>SORCS3</i>	NM_014978	c.3202G>A	p.E1068K	0.21	-	-
42	<i>SPEF1</i>	NM_015417	c.553C>A	p.Q185K	0.1	-	-
42	<i>SRC</i>	NM_005417	c.142C>T	p.R48C	0.2	-	-
42	<i>TP63</i>	NM_00111498 0	c.1543G>A	p.E515K	0.18	-	-
42	<i>WNT7A</i>	NM_004625	c.1032G>C	p.E344D	0.12	-	-
43	<i>IRS2</i>	NM_003749	c.3751C>G	p.L1251V	0.07	-	-
43	<i>KRTAP3-3</i>	NM_033185	c.20G>A	p.R7Q	0.22	-	-
43	<i>MUC5B</i>	NM_002458	c.16800+2T>A	(exon 45)	0.06	-	-
43	<i>PCDHA2</i>	NM_018905	c.2252C>T	p.S751L	0.07	-	-
43	<i>TCOF1</i>	NM_000356	c.959C>T	p.A320V	0.14	-	-
44	<i>C19orf66</i>	NM_00130827 7	c.301G>A	p.D101N	0.06	-	-
44	<i>CYP2A6</i>	NM_000762	c.932G>A	p.R311H	0.06	-	-
44	<i>ENGASE</i>	NM_00104257 3	c.1185C>G	p.I395M	0.08	-	-
44	<i>FAM184B</i>	NM_015688	c.579G>A	p.M193I	0.11	-	-
44	<i>FARS2</i>	NM_006567	c.90G>T	p.Q30H	0.06	-	-
44	<i>FBN3</i>	NM_032447	c.8228G>A	p.R2743H	0.13	-	-
44	<i>FUT3</i>	NM_00109764 1	c.179_187delGCCCCACCC	p.58_60delPTR	0.21	-	-
44	<i>LOC389895</i>	NM_00127156 0	c.451G>T	p.A151S	0.11	-	-
44	<i>LRRC40</i>	NM_017768	c.131C>A	p.S44X	0.12	-	-
44	<i>MAP3K10</i>	NM_002446	c.1403G>A	p.R468Q	0.08	-	-
44	<i>MYO15A</i>	NM_016239	c.1375G>A	p.E459K	0.07	-	-
44	<i>PCDH8</i>	NM_002590	c.3110G>A	p.R1037H	0.12	-	-
44	<i>PLEC</i>	NM_201378	c.8846T>G	p.V2949G	0.05	-	-

44	<i>RHOBTB2</i>	NM_00116003 7	c.230G>A	p.R77Q	0.1	-	-
44	<i>RLIM</i>	NM_016120	c.176G>A	p.S59N	0.14	-	-
44	<i>SIGLEC12</i>	NM_033329	c.553A>G	p.T185A	0.13	-	-
44	<i>TP53</i>	NM_000546	c.827_830delCCTG	p.A276Vfs*68	0.11	-	-
44	<i>UBQLN3</i>	NM_017481	c.1400C>T	p.A467V	0.13	-	-
44	<i>ZNF423</i>	NM_00127162 0	c.1805A>G	p.Q602R	0.06	-	-
45	<i>DACT1</i>	NM_00107952 0	c.2009C>T	p.A670V	0.1	-	-
45	<i>DUSP9</i>	NM_001395	c.527C>A	p.S176Y	0.32	-	-
45	<i>ICT1</i>	NM_00130326 5	c.158A>C	p.Q53P	0.07	-	-
45	<i>ITPR3</i>	NM_002224	c.1954A>C	p.I652L	0.05	-	-
45	<i>KCNJ4</i>	NM_004981	c.100A>C	p.N34H	0.05	-	-
45	<i>KCNK12</i>	NM_022055	c.862G>T	p.V288L	0.06	-	-
45	<i>KLHL26</i>	NM_018316	c.280G>A	p.G94S	0.09	-	-
45	<i>LILRA5</i>	NM_181985	c.461T>G	p.L154R	0.12	-	-
45	<i>MFSD12</i>	NM_00128752 9	c.1196T>G	p.M399R	0.07	-	-
45	<i>MYO9B</i>	NM_00113006 5	c.499A>C	p.K167Q	0.05	-	-
45	<i>PARP9</i>	NM_00114610 2	c.440G>A	p.G147E	0.14	-	-
45	<i>PCDHB3</i>	NM_018937	c.1708G>A	p.A570T	0.08	-	-
45	<i>PLXNB2</i>	NM_012401	c.4127_4135delTCCTGGAGC	p.1374_1376delLE L	0.11	-	-
45	<i>PRDM16</i>	NM_022114	c.2555C>T	p.P852L	0.14	-	-
45	<i>SCN5A</i>	NM_000335	c.2182G>A	p.V728I	0.09	-	-
45	<i>SNX32</i>	NM_152760	c.1057G>T	p.D353Y	0.09	-	-
45	<i>ZNF646</i>	NM_014699	c.1513A>C	p.M505L	0.07	-	-
46	<i>ADCK2</i>	NM_052853	c.175G>A	p.E59K	0.08	-	-
47	<i>ALPK2</i>	NM_052947	c.3524C>T	p.S1175L	0.18	-	-
47	<i>AP3B2</i>	NM_004644	c.3099-3C>T	(exon 26)	0.29	-	-
47	<i>APBA2</i>	NM_00113041 4	c.1646C>T	p.S549L	0.23	-	-
47	<i>BEST4</i>	NM_153274	c.130G>T	p.A44S	0.29	-	-
47	<i>C9orf64</i>	NM_032307	c.157C>T	p.H53Y	0.25	-	-
47	<i>CNOT1</i>	NM_00126561 2	c.4291C>T	p.R1431X	0.27	-	-
47	<i>COQ4</i>	NM_00130594 2	c.31C>G	p.R11G	0.24	-	-

47	<i>CTNND2</i>	NM_001332	c.65C>T	p.S22L	0.42	-	-
47	<i>DAPK1</i>	NM_00128872 9	c.2641C>T	p.Q881X	0.32	-	-
47	<i>DCAF8</i>	NM_015726	c.1153G>A	p.E385K	0.23	-	-
47	<i>DDOST</i>	NM_005216	c.870A>C	p.E290D	0.13	-	-
47	<i>DDX49</i>	NM_019070	c.1024G>A	p.A342T	0.31	-	-
47	<i>EHMT2</i>	NM_00128941 3	c.1820G>A	p.R607Q	0.44	-	-
47	<i>FCRL1</i>	NM_00115939 7	c.666G>C	p.E222D	0.32	-	-
47	<i>GSG1</i>	NM_00108055 4	c.241G>A	p.D81N	0.12	-	-
47	<i>HELZ</i>	NM_014877	c.4696G>A	p.E1566K	0.16	-	-
47	<i>IFT140</i>	NM_014714	c.2856G>A	p.M952I	0.25	-	-
47	<i>IFT140</i>	NM_014714	c.3430G>A	p.E1144K	0.27	-	-
47	<i>INSRR</i>	NM_014215	c.1504C>T	p.R502C	0.08	-	-
47	<i>ITGAL</i>	NM_00111438 0	c.2434G>A	p.A812T	0.29	-	-
47	<i>ITPKA</i>	NM_002220	c.1273G>A	p.D425N	0.18	-	-
47	<i>KIF25</i>	NM_030615	c.898G>A	p.A300T	0.46	-	-
47	<i>KRT38</i>	NM_006771	c.1121C>T	p.S374F	0.21	-	-
47	<i>LOC441155</i>	NM_00127167 5	c.433G>A	p.E145K	0.29	-	-
47	<i>LRP1</i>	NM_002332	c.2608C>G	p.R870G	0.1	-	-
47	<i>MAGI1</i>	NM_00103305 7	c.2087C>T	p.A696V	0.17	-	-
47	<i>METTL14</i>	NM_020961	c.1343_1344delCA	p.H449Rfs*71	0.25	-	-
47	<i>MICB</i>	NM_00128916 0	c.426G>A	p.M142I	0.36	-	-
47	<i>MYH9</i>	NM_002473	c.3488C>T	p.S1163L	0.21	-	-
47	<i>NEU2</i>	NM_005383	c.277G>A	p.A93T	0.19	-	-
47	<i>NLRP7</i>	NM_00112725 5	c.331G>T	p.D111Y	0.22	-	-
47	<i>ODF1</i>	NM_024410	c.520G>A	p.E174K	0.07	-	-
47	<i>PDE4A</i>	NM_00124312 1	c.46G>T	p.E16X	0.27	-	-
47	<i>PTH</i>	NM_000315	c.236C>T	p.S79F	0.25	-	-
47	<i>RBM8A</i>	NM_005105	c.40G>A	p.D14N	0.16	-	-
47	<i>RPS12</i>	NM_001016	c.7G>A	p.E3K	0.33	-	-
47	<i>SEC62</i>	NM_003262	c.1156G>A	p.E386K	0.22	-	-
47	<i>SELE</i>	NM_000450	c.820G>A	p.E274K	0.2	-	-

47	<i>SEMA4A</i>	NM_00119330 2	c.1027G>A	p.A343T	0.19	-	-
47	<i>SFI1</i>	NM_00125832 6	c.982C>T	p.H328Y	0.31	-	-
47	<i>SLC26A9</i>	NM_052934	c.1634T>A	p.I545N	0.23	-	-
47	<i>SORCS1</i>	NM_052918	c.2058+1G>A	(exon 15)	0.25	-	-
47	<i>TMEM132E</i>	NM_00130443 8	c.50C>T	p.S17L	0.29	-	-
47	<i>TP53</i>	NM_00112611 5	c.128G>A	p.R43H	0.4	RCV00016106 5	COSM335599 4
47	<i>TPM2</i>	NM_003289	c.773-1->C	(exon 9)	0.18	-	-
47	<i>TUBGCP6</i>	NM_020461	c.3463C>T	p.R1155W	0.09	-	-
47	<i>ZIC3</i>	NM_003413	c.68G>A	p.R23H	0.08	-	-
47	<i>ZNF208</i>	NM_007153	c.205G>A	p.E69K	0.22	-	-
47	<i>ZNF787</i>	NM_00100283 6	c.508C>T	p.R170C	0.4	-	-
47	<i>ZNF831</i>	NM_178457	c.1100C>T	p.S367L	0.13	-	-
48	<i>ICT1</i>	NM_00130326 5	c.158A>C	p.Q53P	0.07	-	-

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Table S5. Summary of possible driver mutations identified in this study

UPN	Gene	Reference	Nucleic acid change	Amino acid change	VAF	ClinVar	COSMIC
1	<i>GAS6-FOXA1</i>	-	-	(fusion gene)	0.10	-	-
2	<i>FOXA1</i>	NM_001310135	g.38064406G>A	(promoter)	0.21	-	-
2	<i>HIST1H2BC</i>	NM_003526	c.72G>C	p.K24N	0.20	-	-
2	<i>PIK3CA</i>	NM_006218	c.241G>A	p.E81K	0.28	-	COSM271871
3	<i>FOXA1</i>	NM_001310135	g.38064406G>A	(promoter)	0.08	-	-
3	<i>PIK3CA</i>	NM_006218	c.2176G>A	p.E726K	0.20	VCV000376476	COSM446019
5	<i>PIK3CA</i>	NM_006218	c.3019G>C	p.G1007R	0.09	-	COSM17443
6	<i>FOXA1</i>	NM_001310135	g.38064406G>T	(promoter)	0.01	-	-
8	<i>FOXA1</i>	NM_001310135	g.38064406G>A	(promoter)	0.01	-	-
14	<i>FOXA1</i>	NM_001310135	g.38064406G>A	(promoter)	0.01	-	-
14	<i>HIST1H2BB</i>	NM_021062	c.72G>C	p.K24N	0.19	-	COSM1169071
14	<i>PIK3CA</i>	NM_006218	c.1633G>A	p.E545K	0.21	RCV000014631	COSM763
14	<i>PIK3CA</i>	NM_006218	c.3172A>C	p.I1058L	0.15	-	COSM479744
14	<i>SMARCB1</i>	NM_003073	c.744_745insCCCA	p.D251Hfs*31	0.37	-	-
17	<i>PIK3CA</i>	NM_006218	c.1624G>C	p.E542Q	0.08	-	COSM17442
18	<i>FOXA1</i>	NM_001310135	g.38064406G>A	(promoter)	0.29	-	-
19	<i>PIK3CA</i>	NM_006218	c.1637A>G	p.Q546R	0.13	RCV000038672	COSM12459
25	<i>FOXA1</i>	NM_001310135	g.38064406G>A	(promoter)	0.03	-	-
35	<i>FOXA1</i>	NM_001310135	g.38064406G>A	(promoter)	0.05	-	-
40	<i>CUX1</i>	NM_001202543	c.2893G>T	p.E965X	0.21	-	-
40	<i>EP300</i>	NM_001429	c.3798G>A	p.W1266X	0.37	-	-
41	<i>ERBB2</i>	NM_001289937	c.929C>T	p.S310F	0.11	-	COSM48358
44	<i>TP53</i>	NM_000546	c.827_830delCCTG	p.A276Vfs*68	0.11	-	-
46	<i>FOXA1</i>	NM_001310135	g.38064406G>A	(promoter)	0.04	-	-
47	<i>TP53</i>	NM_001126115	c.128G>A	p.R43H	0.4	RCV000161065	COSM3355994
48	<i>FOXA1</i>	NM_001310135	g.38064406G>A	(promoter)	0.01	-	-

Table S6. Top 200 differentially expressed genes

Gene	baseMean	Log2 fold change	adjusted p-value	FOXA1 target	FOXA1 pathway	Keratin family
<i>MLPH</i>	6291.8	3.59	3.2E-147			
<i>TSPAN1</i>	8694.4	6.38	1.2E-75			
<i>TMPRSS11D</i>	2394.2	7.92	5.6E-68			
<i>ANO7</i>	2972.0	7.61	5.2E-65			
<i>KRT2</i>	152699.4	-12.53	1.0E-56			Y
<i>GPX2</i>	2051.1	3.42	5.3E-55			
<i>MEF2A</i>	3745.1	-1.98	4.4E-53			
<i>PRR15</i>	1815.5	6.53	1.0E-52			
<i>ABCC11</i>	2896.6	8.88	1.3E-51			
<i>SOS2</i>	2029.9	-1.34	4.3E-51			
<i>SEC14L2</i>	3122.7	3.46	4.5E-51			
<i>HHIPL2</i>	647.6	7.81	9.3E-49			
<i>FGFR4</i>	4917.3	6.33	8.7E-47			
<i>RHCG</i>	7665.3	5.16	1.8E-46			
<i>KMO</i>	288.8	5.44	9.5E-46			
<i>MUC5AC</i>	9131.8	14.97	2.2E-44			
<i>STXBP5</i>	2264.0	-1.57	1.0E-43			
<i>GAL3ST2</i>	349.4	7.11	5.3E-42			
<i>TMEM132A</i>	8050.6	4.67	7.6E-42			
<i>SPDEF</i>	6669.8	4.75	1.1E-40	Y		
<i>JAK2</i>	1150.8	-1.87	3.0E-40			
<i>PGD</i>	4916.6	1.91	6.0E-40			
<i>NETO2</i>	717.7	4.19	1.8E-39			
<i>VSIG2</i>	1890.2	5.64	3.9E-39	Y		
<i>CAPN9</i>	1374.6	8.93	5.1E-39			
<i>FUT3</i>	1316.4	4.62	9.8E-39			
<i>NRK</i>	504.4	4.96	1.0E-37			
<i>PTHLH</i>	3723.3	7.46	2.7E-37			
<i>ANKRD12</i>	4283.4	-1.77	1.5E-36			
<i>SYT8</i>	2227.0	-5.27	1.6E-35			
<i>SYNE4</i>	291.5	3.09	1.9E-35			
<i>FOXA3</i>	1377.8	10.44	2.0E-35		Y	
<i>LRP4</i>	3358.7	-3.41	3.8E-35			
<i>BAALC</i>	1241.3	4.15	6.7E-35			
<i>PGAP1</i>	1509.0	-2.22	1.2E-34			
<i>SEPT10</i>	2239.2	-1.28	4.8E-34			
<i>YIF1B</i>	1907.5	1.33	1.1E-33			
<i>TFF1</i>	3927.8	15.44	2.1E-33		Y	

CAPN8	961.7	5.15	6.1E-33	
SLC44A4	3451.5	5.08	1.2E-32	
SPRR1A	26589.6	5.28	5.1E-32	
ZNF593	1131.7	1.82	7.6E-32	
PDK3	2532.0	-2.49	8.1E-32	
KRT6A	88088.3	5.13	1.0E-31	Y
GJB2	45826.9	3.76	3.1E-31	
KRT8	14812.9	4.20	7.1E-31	Y
KIAA1328	228.7	-1.58	1.1E-30	
CNDP2	7070.2	1.47	1.3E-30	
ERP27	489.3	3.04	3.0E-30	
LOC101929613	465.0	8.20	3.3E-30	
HBA1	175.5	-5.15	4.2E-30	
ANTXR1	4727.9	-2.74	7.7E-30	
ARRDC1	4439.7	2.09	4.2E-29	
GSTP1	37241.3	2.13	6.9E-29	
DST	24044.7	-1.69	7.0E-29	
TBC1D30	376.9	3.14	8.6E-29	
STRA6	1211.6	7.93	8.8E-29	
KRBA1	1154.2	1.52	1.0E-28	
LRRC31	305.9	8.21	1.0E-28	
ABCG4	654.2	5.68	7.5E-28	
PNMT	926.4	6.58	1.4E-27	
ASL	1303.0	1.52	1.8E-27	
CDKL1	202.4	-1.60	3.7E-27	
KIAA2026	1909.1	-1.15	4.0E-27	
AGR2	7079.3	6.04	4.3E-27	
RGMB	1959.2	-2.10	4.3E-27	
HDAC1	4293.1	1.10	5.0E-27	
CDON	2126.9	-3.22	5.2E-27	
CCDC103	97.3	2.54	5.2E-27	
EPHB1	241.5	-4.64	5.2E-27	
SPRY1	1088.7	-2.39	5.2E-27	Y
QKI	3722.9	-1.59	6.3E-27	
FOXA1	3816.5	4.22	9.7E-27	Y
COA3	2114.4	1.26	1.0E-26	
PCGF5	1751.2	-2.34	1.0E-26	
AKR7A3	397.6	4.39	2.2E-26	
CXCL17	334.3	6.60	2.3E-26	
POLD4	3714.0	1.83	2.9E-26	Y
MBNL1	5910.3	-1.26	3.0E-26	
VSIG1	959.5	5.51	3.5E-26	

<i>GNB4</i>	4613.4	-2.42	3.7E-26	
<i>B4GALNT2</i>	2949.3	6.99	4.3E-26	
<i>CEACAM6</i>	20151.5	4.52	7.0E-26	
<i>AEN</i>	1637.2	1.72	2.1E-25	
<i>ISG20</i>	1457.6	3.34	2.5E-25	
<i>FOXP2</i>	344.8	-3.21	2.7E-25	Y
<i>EFHD1</i>	3316.0	3.84	2.9E-25	
<i>MAT1A</i>	115.4	-5.73	3.4E-25	
<i>GBP6</i>	2157.5	4.46	4.0E-25	
<i>ST6GALNAC1</i>	355.0	3.40	5.4E-25	
<i>MBOAT7</i>	3696.0	1.23	5.5E-25	
<i>AP1M2</i>	3390.2	1.42	5.7E-25	
<i>CYP2S1</i>	929.3	2.00	5.9E-25	
<i>TNNI2</i>	431.1	-7.06	9.2E-25	
<i>PJA2</i>	4666.1	-1.12	1.2E-24	
<i>SMIM22</i>	857.8	4.15	1.7E-24	
<i>MUC7</i>	243.3	-11.56	1.7E-24	
<i>SOCS5</i>	1092.9	-1.04	2.0E-24	
<i>KRT19</i>	47366.4	4.40	2.2E-24	Y
<i>FAM122A</i>	973.4	-0.81	2.3E-24	Y
<i>CLN3</i>	2730.3	1.30	2.3E-24	
<i>TIMM8B</i>	1504.4	1.66	4.7E-24	
<i>SWI5</i>	525.3	1.12	5.0E-24	
<i>KYNU</i>	1437.2	4.38	5.4E-24	
<i>LDLRAD1</i>	166.0	6.12	6.3E-24	
<i>CYB561D2</i>	756.9	1.58	9.6E-24	
<i>MIR31HG</i>	105.8	6.81	1.1E-23	
<i>CAV2</i>	2831.8	-1.84	1.2E-23	
<i>CTDSPL</i>	3717.1	-1.65	1.9E-23	
<i>JADE1</i>	993.4	-2.11	2.4E-23	Y
<i>TUFM</i>	8269.6	1.17	2.4E-23	
<i>KLK13</i>	6683.4	4.52	3.2E-23	
<i>NR3C1</i>	3242.5	-2.03	3.2E-23	
<i>WDR47</i>	2324.4	-1.20	3.5E-23	
<i>FER</i>	1301.5	-1.08	3.7E-23	
<i>TRIM16</i>	2332.3	1.80	5.0E-23	
<i>BIK</i>	452.2	2.80	5.2E-23	
<i>LSM11</i>	307.4	-1.39	5.4E-23	
<i>ZNF654</i>	816.8	-1.10	6.5E-23	
<i>SNRNP35</i>	1345.2	1.00	8.8E-23	
<i>CSTA</i>	44252.8	2.89	9.2E-23	
<i>DDX39A</i>	2878.3	1.36	9.3E-23	

TOX3	730.9	4.39	1.1E-22		
AFP	625.6	11.63	1.1E-22		
USP12	1629.5	-1.25	1.3E-22		
SNTB2	1707.3	-1.34	1.5E-22		
HSP90AB1	20619.7	0.79	2.4E-22		
NFIX	11628.3	-2.05	2.7E-22		
ABCC1	5427.2	1.50	4.3E-22		
FARSA	3609.3	1.07	4.3E-22		
LINC00514	200.0	7.46	4.6E-22		
SOX11	651.4	7.26	4.7E-22		
MKLN1	2712.7	-1.00	4.7E-22		
TFAP2B	6897.3	3.36	6.1E-22		
DENND4C	3647.5	-1.59	6.6E-22		
CXCL14	28129.5	-2.26	7.8E-22		
ETV6	1958.7	-1.34	7.8E-22		
HM13	4588.3	1.34	8.1E-22		
LPIN1	775.7	-1.91	8.1E-22		
HID1	3461.8	2.96	9.5E-22	Y	
B9D1	421.0	1.82	9.7E-22		
KRT18	3956.5	3.56	1.0E-21		Y
OGFRL1	931.5	-1.80	1.1E-21		
SLC9A3R1	11639.6	2.70	1.4E-21	Y	
NPW	3371.2	7.23	1.6E-21		
BCAS1	1630.9	3.72	1.8E-21	Y	
ALDOA	36527.7	1.26	1.9E-21		
HMGB3	1136.6	1.91	1.9E-21		
SMPDL3B	468.2	3.75	1.9E-21		
CDHR1	12616.3	-3.78	1.9E-21		
SCAND1	2996.2	1.40	1.9E-21		
DYNC111	73.4	-4.68	2.1E-21		
SERPINB3	30391.2	5.83	2.6E-21		
CNGB1	116.0	6.74	2.6E-21		
TNFRSF21	2297.2	1.73	2.8E-21		
TMEM208	1784.5	1.35	3.0E-21	Y	
RPL7	457.1	-1.87	3.1E-21		
MRPL12	3375.2	1.65	3.1E-21		
KRT17	123592.3	5.21	4.1E-21	Y	Y
CXCL6	953.1	11.33	4.5E-21		
FAM175A	274.0	-1.84	5.3E-21		
MANF	3643.8	1.67	5.7E-21		
JOSD2	1264.5	1.10	7.0E-21		
PRSS27	2325.6	4.17	7.0E-21		

<i>FAM177B</i>	428.7	8.54	7.7E-21	
<i>SPRR1B</i>	56743.8	4.15	8.4E-21	
<i>ELK4</i>	2680.5	-1.25	8.4E-21	
<i>ZBTB33</i>	2119.2	-1.48	9.5E-21	
<i>SMAD5</i>	3665.5	-1.20	9.7E-21	Y
<i>HMGCS2</i>	8114.4	4.49	1.0E-20	
<i>CCT3</i>	9891.2	0.92	1.1E-20	
<i>ARHGAP31</i>	1436.3	-2.25	1.1E-20	
<i>MAML2</i>	906.0	-2.69	1.2E-20	
<i>MRPS33</i>	1246.5	1.16	1.2E-20	
<i>RAB30</i>	707.9	-2.06	1.3E-20	
<i>SLC25A39</i>	6605.8	1.28	1.3E-20	
<i>GNG12</i>	9709.2	-1.17	1.4E-20	
<i>KRT16</i>	63038.5	5.27	1.5E-20	Y
<i>PYCR1</i>	2667.7	3.32	1.6E-20	
<i>SNRNP25</i>	903.0	1.38	1.6E-20	
<i>ATP13A1</i>	4315.1	1.01	1.9E-20	
<i>PXK</i>	615.3	-1.68	3.0E-20	
<i>HIST1H2BH</i>	38.9	2.64	3.2E-20	
<i>RSBN1</i>	914.0	-1.36	3.4E-20	
<i>C9orf116</i>	117.3	2.93	3.7E-20	
<i>H2AFX</i>	2146.6	1.65	4.3E-20	
<i>PRKAB2</i>	1397.2	-1.54	4.7E-20	
<i>ATP7B</i>	1375.0	3.32	9.2E-20	
<i>PAX9</i>	355.6	4.57	9.8E-20	
<i>CCDC167</i>	373.0	1.75	1.2E-19	
<i>LBX2</i>	55.7	4.31	1.2E-19	
<i>CAPN13</i>	515.0	5.41	1.4E-19	
<i>AKAP11</i>	2649.5	-1.12	1.6E-19	
<i>ERVMER34-1</i>	156.1	3.04	1.7E-19	
<i>SLC26A2</i>	1874.4	-3.36	2.0E-19	
<i>KPNA2</i>	2185.7	2.25	2.0E-19	
<i>DSTYK</i>	1977.5	-1.30	2.4E-19	
<i>ALAS1</i>	3561.0	1.61	2.5E-19	
<i>GABRP</i>	708.4	-5.31	2.6E-19	
<i>SMAGP</i>	1552.4	1.48	2.8E-19	

66 Positive Log₂ fold change values indicate upregulation in patient samples. Y, yes

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