

Table S1 The list of nine highly variable genes in patient revealed by whole-exome analysis

Gene name	Chromosome	Position	dbSNP ID	REF	ALT	ExonicFunc	Gene	AChange	cytoBand	cosmic82	SIFT
AHNAK2	14	105411781	rs10438247	G	A	Missense SNV	NM_138420	AHNAK2:NM_138420:exon7:c.C10007T;p.P3336L	14q32.33	ID=COSM432734;OCCURENCE=1(thyroid)	0.014,D
AHNAK2	14	105413223	rs55797226	A	G	Synonymous SNV	NM_138420	AHNAK2:NM_138420:exon7:c.T8565C;p.D2855D	14q32.33	NA	NA
AHNAK2	14	105414810	rs10145032	C	G	Synonymous SNV	NM_138420	AHNAK2:NM_138420:exon7:c.G6978C;p.L2326L	14q32.33	NA	NA
AHNAK2	14	105414923	rs200761412	T	C	Missense SNV	NM_138420	AHNAK2:NM_138420:exon7:c.A6865G;p.K2289E	14q32.33	ID=COSM5438893;OCCURENCE=1(oesophagus)	0.148,T
AHNAK2	14	105417588	rs557610829	G	A	Synonymous SNV	NM_138420	AHNAK2:NM_138420:exon7:c.C4200T;p.P1400P	14q32.33	ID=COSM4147906;OCCURENCE=1(thyroid)	NA
AHNAK2	14	105417597	rs572861020	T	C	Synonymous SNV	NM_138420	AHNAK2:NM_138420:exon7:c.A4191G;p.P1397P	14q32.33	ID=COSM4147907;OCCURENCE=1(thyroid)	NA
AHNAK2	14	105417598	rs201134968	G	A	Missense SNV	NM_138420	AHNAK2:NM_138420:exon7:c.C4190T;p.P1397L	14q32.33	ID=COSM4147908;OCCURENCE=1(thyroid)	0.486,T
AHNAK2	14	105417837	rs76231332	A	G	Synonymous SNV	NM_138420	AHNAK2:NM_138420:exon7:c.T3951C;p.A1317A	14q32.33	NA	NA
AHNAK2	14	105418264	rs55650155	G	A	Missense SNV	NM_138420	AHNAK2:NM_138420:exon7:c.C3524T;p.A1175V	14q32.33	ID=COSM4147917;OCCURENCE=1(thyroid),1(large_intestine),1(liver)	1.0,T
AHNAK2	14	105418344	rs55791176	T	G	Missense SNV	NM_138420	AHNAK2:NM_138420:exon7:c.A3444C;p.E1148D	14q32.33	ID=COSM1368638;OCCURENCE=2(thyroid)	0.765,T
AHNAK2	14	105418391	rs11625007	C	T	Missense SNV	NM_138420	AHNAK2:NM_138420:exon7:c.G3397A;p.V1133I	14q32.33	ID=COSM1368639;OCCURENCE=2(central_nervous_system),1(lung),1(thyroid)	1.0,T
AHNAK2	14	105419243	NA	C	T	Missense SNV	NM_138420	AHNAK2:NM_138420:exon7:c.G2545A;p.G849S	14q32.33	NA	0.446,T
MUC4	3	195489009	rs2246901	C	A	Missense SNV	NM_004532,NM_018406,NM_138297	MUC4:NM_138297:exon12:c.G1600T;p.A534S,MUC4:NM_004532:exon13:c.G1753T;p.A585S,MUC4:NM_018406:exon14:c.G14461T;p.A4821S	3q29	ID=COSM4157536,COSM4157534,COSM4157535;OCCURENCE=2(thyroid)	0.241,T
MUC4	3	195489067	rs2246980	C	G	Synonymous SNV	NM_004532,NM_018406,NM_138297	MUC4:NM_138297:exon12:c.G1542C;p.S514S,MUC4:NM_004532:exon13:c.G1695C;p.S565S,MUC4:NM_018406:exon14:c.G14403C;p.S4801S	3q29	ID=COSM1421868,COSM1421867,COSM1421869;OCCURENCE=1(large_intestine)	NA
MUC4	3	195510762	rs2911273	G	A	Synonymous SNV	NM_018406	MUC4:NM_018406:exon2:c.C7689T;p.V2563V	3q29	ID=COSM149590,COSM149589;OCCURENCE=3(large_intestine),1(thyroid),1(upper_aerodigestive_tract),3(central_nervous_system),1(stomach),1(kidney)	NA
MUC4	3	195512446	rs550380149	G	A	Missense SNV	NM_018406	MUC4:NM_018406:exon2:c.C6005T;p.P2002L	3q29	NA	0.032,D
MUC4	3	195512597	rs6799339	G	A	Missense SNV	NM_018406	MUC4:NM_018406:exon2:c.C5854T;p.P1952S	3q29	ID=COSM4157817,COSM1042915;OCCURENCE=1(endometrium),2(central_nervous_system),15(upper_aerodigestive_tract),4(thyroid),2(large_intestine),2(haematopoietic_and_lymphoid_tissue),2(prostate)	0.14,T
MUC4	3	195513063	NA	G	A	Synonymous SNV	NM_018406	MUC4:NM_018406:exon2:c.C5388T;p.T1796T	3q29	NA	NA
MUC4	3	195513345	rs71321841	A	C	Synonymous SNV	NM_018406	MUC4:NM_018406:exon2:c.T5106G;p.V1702V	3q29	ID=COSM4002588,COSM4002587;OCCURENCE=2(haematopoietic_and_lymphoid_tissue),2(large_intestine),2(thyroid),1(upper_aerodigestive_tract)	NA
MUC4	3	195515008	rs201451131	C	G	Missense SNV	NM_018406	MUC4:NM_018406:exon2:c.G3443C;p.G1148A	3q29	ID=COSM1485032,COSM4591321;OCCURENCE=3(central_nervous_system),17(upper_aerodigestive_tract),1(prostate)	1.0,T
MUC4	3	195517321	rs1104760	G	A	Missense SNV	NM_018406	MUC4:NM_018406:exon2:c.C1130T;p.T377I	3q29	ID=COSM4005514,COSM4005515;OCCURENCE=1(urinary_tract)	1.0,T
SULT1A1	16	28617507	rs41278160	C	T	Synonymous SNV	NM_001055,NM_177529,NM_177530,NM_177534,NM_177536	SULT1A1:NM_177536:exon5:c.G411A;p.L137L,SULT1A1:NM_177534:exon6:c.G645A;p.L215L,SULT1A1:NM_001055:exon7:c.G645A;p.L215L,SULT1A1:NM_177529:exon7:c.G645A;p.L215L,SULT1A1:NM_177530:exon7:c.G645A;p.L215L	16p11.2	ID=COSM4415626,COSM4415625;OCCURENCE=2(soft_tissue),4(liver),1(large_intestine),1(thyroid),1(breast),2(haematopoietic_and_lymphoid_tissue)	NA
SULT1A1	16	28617572	rs117124912	G	A	Unknown	NM_001055,NM_177529,NM_177530,NM_177534,NM_177536	unknown	16p11.2	ID=COSN26728343;OCCURENCE=1(breast)	NA
SULT1A1	16	28618237	rs9282862	T	C	Unknown	NM_001055,NM_177529,NM_177530,NM_177534,NM_177536	unknown	16p11.2	ID=COSN17140510,COSN17144487,COSN17146316,COSN17133335,COSN17139650,COSN26727881,COSN17148089,COSN17141357,COSN17147203,COSN17135963,COSN17136811,COSN17135064,COSN17145383,COSN17152807,COSN17149916;OCCURENCE=14(soft_tissue),1(breast)	NA
SULT1A1	16	28618446	rs2925623	T	C	Unknown	NM_001055,NM_177529,NM_177530,NM_177534,NM_177536	unknown	16p11.2	ID=COSN17149917,COSN17136812,COSN17135964,COSN17145384,COSN17135065,COSN17133336,COSN17152808,COSN17147204,COSN17140511,COSN17139651,COSN17148090,COSN26728421,COSN17146317,COSN165161,COSN17144488,COSN17141358;OCCURENCE=14(soft_tissue),1(breast),1(stomach)	NA
SULT1A1	16	28618469	rs376487988	A	T	Unknown	NM_001055,NM_177529,NM_177530,NM_177534,NM_177536	unknown	16p11.2	ID=COSN15330618,COSN26728685,COSN19667205,COSN20345033;OCCURENCE=1(haematopoietic_and_lymphoid_tissue),1(breast),2(large_intestine)	NA
SULT1A1	16	28618636	rs3020804	A	G	Unknown	NM_001055,NM_177529,NM_177530,NM_177534,NM_177536	unknown	16p11.2	ID=COSN26732151,COSN19708983;OCCURENCE=1(breast),1(large_intestine)	NA
SULT1A1	16	28619911	rs1126447	T	C	Synonymous SNV	NM_001055,NM_177529,NM_177530,NM_177534	SULT1A1:NM_177534:exon2:c.A162G;p.V54V,SULT1A1:NM_001055:exon3:c.A162G;p.V54V,SULT1A1:NM_177529:exon3:c.A162G;p.V54V,SULT1A1:NM_177530:exon3:c.A162G;p.V54V	16p11.2	ID=COSM148063;OCCURENCE=1(haematopoietic_and_lymphoid_tissue),1(breast),13(soft_tissue),1(stomach)	NA
SULT1A1	16	28619920	rs1126446	A	G	Synonymous SNV	NM_001055,NM_177529,NM_177530,NM_177534	SULT1A1:NM_177534:exon2:c.T153C;p.T51T,SULT1A1:NM_001055:exon3:c.T153C;p.T51T,SULT1A1:NM_177529:exon3:c.T153C;p.T51T,SULT1A1:NM_177530:exon3:c.T153C;p.T51T	16p11.2	ID=COSM148064;OCCURENCE=1(stomach),13(soft_tissue),1(haematopoietic_and_lymphoid_tissue),1(breast),1(large_intestine)	NA
SULT1A1	16	28620120	rs34513973	C	T	Synonymous SNV	NM_001055,NM_177529,NM_177530,NM_177534	SULT1A1:NM_177534:exon1:c.G57A;p.P19PSULT1A1:NM_001055:exon2:c.G57A;p.P19PSULT1A1:NM_177529:exon2:c.G57A;p.P19PSULT1A1:NM_177530:exon2:c.G57A;p.P19P	16p11.2	ID=COSM3754830;OCCURENCE=1(haematopoietic_and_lymphoid_tissue),1(liver),1(large_intestine),4(soft_tissue)	NA
NBPF1	1	16890417	NA	T	G	Unknown	NM_017940	unknown	1p36.13	NA	NA
NBPF1	1	16890441	rs3863779	C	T	Unknown	NM_017940	UNKNOWN	1p36.13	ID=COSN17908214,COSN19726322,COSN20442428,COSN9365785;OCCURENCE=1(skin),2(large_intestine),1(kidney)	NA
NBPF1	1	16890484	rs12117084	G	C	Unknown	NM_017940	UNKNOWN	1p36.13	ID=COSN26856318,COSN17907833,COSN26856984,COSN20442427,COSN19670624,COSN26860000;OCCURENCE=1(skin),2(large_intestine),3(thyroid)	NA
NBPF1	1	16892282	rs202078823	C	A	Unknown	NM_017940	UNKNOWN	1p36.13	ID=COSN19624539,COSN20280143,COSN20401972,COSN23914604;OCCURENCE=1(large_intestine),3(haematopoietic_and_lymphoid_tissue)	NA
NBPF1	1	16893598	rs927016856	G	A	Unknown	NM_017940	unknown	1p36.13	ID=COSN1091962;OCCURENCE=1(liver)	NA
NBPF1	1	16903026	rs3897291	C	A	Unknown	NM_017940	unknown	1p36.13	ID=COSN8861695,COSN15653441;OCCURENCE=1(haematopoietic_and_lymphoid_tissue),1(upper_aerodigestive_tract)	NA
NBPF1	1	16907164	rs201839566	C	T	Unknown	NM_017940	unknown	1p36.13	ID=COSN19749507,COSN15655724,COSN19700541,COSN19629792;OCCURENCE=1(haematopoietic_and_lymphoid_tissue),1(upper_aerodigestive_tract),2(large_intestine)	NA
NBPF1	1	16918411	rs199725761	T	C	Unknown	NM_017940	UNKNOWN	1p36.13	ID=COSN19740211;OCCURENCE=1(large_intestine)	NA
ABO	9	136131188	rs8176749	C	T	Unknown	NM_020469	UNKNOWN	9q34.2	NA	NA
ABO	9	136131315	rs8176747	C	G	Unknown	NM_020469	UNKNOWN	9q34.2	ID=COSM3952439;OCCURENCE=1(lung)	NA
ABO	9	136131322	rs8176746	G	T	Unknown	NM_020469	UNKNOWN	9q34.2	NA	NA
ABO	9	136131415	rs8176743	C	T	Unknown	NM_020469	UNKNOWN	9q34.2	NA	NA
ABO	9	136131461	rs8176741	G	A	Unknown	NM_020469	UNKNOWN	9q34.2	NA	NA
ABO	9	136131592	rs7853989	G	C	Unknown	NM_020469	UNKNOWN	9q34.2	NA	NA
ABO	9	136132754	rs8176722	C	A	Unknown	NM_020469	unknown	9q34.2	NA	NA
MST1L	1	17084086	rs749078168	A	G	Missense SNV	NM_001271733	MST1L:NM_001271733:exon14:c.T1835C;p.I612T	1p36.13	NA	NA
MST1L	1	17084152	rs202123117	C	T	Missense SNV	NM_001271733	MST1L:NM_001271733:exon14:c.G1769A;p.G590D	1p36.13	ID=COSM1317624;OCCURENCE=1(large_intestine),2(upper_aerodigestive_tract),1(haematopoietic_and_lymphoid_tissue),1(breast),3(central_nervous_system)	NA
MST1L	1	17085046	rs200838083	C	G	Missense SNV	NM_001271733	MST1L:NM_001271733:exon11:c.G1429C;p.D477H	1p36.13	ID=COSM1491885,COSM1491886;OCCURENCE=1(thyroid),1(large_intestine),2(pancreas),1(kidney)	NA
MST1L	1	17086236	rs61769737	G	C	Unknown	NM_001271733	unknown	1p36.13	ID=COSN15107201,COSN19643620,COSN19618339;OCCURENCE=2(haematopoietic_and_lymphoid_tissue),1(oesophagus)	NA
MST1L	1	17086273	NA	T	A	Unknown	NM_001271733	unknown	1p36.13	ID=COSN20838787,COSN21670862;OCCURENCE=2(breast)	NA
MST1L	1	17087432	rs2446554	G	T	Unknown	NM_001271733	unknown	1p36.13	ID=COSN19672857;OCCURENCE=1(large_intestine)	NA
ZNF718	4	53310	rs761021532	C	T	Unknown	NM_001039127,NM_001286052,NM_001286053,NM_001286054,NM_182524	unknown	4p16.3	NA	NA
ZNF718	4	53360	rs75858144	C	G	Unknown	NM_001039127,NM_001286052,NM_001286053,NM_001286054,NM_182524	unknown	4p16.3	ID=COSN26958394;OCCURENCE=1(lung)	NA
ZNF718	4	59350	rs1045387254	A	G	Missense SNV	NM_001039127,NM_001286052,NM_182524	ZNF718:NM_001039127:exon2:c.A31G;p.I11V	4p16.3	ID=COSM3760687;OCCURENCE=2(large_intestine),1(thyroid),1(liver),1(adrenal_gland),1(prostate)	0.15,T
ZNF718	4	59469	rs6834940	A	G	Unknown	NM_001039127,NM_001286052,NM_001286053,NM_001286054,NM_001289931,NM_182524	unknown	4p16.3	ID=COSN19749630,COSN26670632,COSN19766576,COSN26679945;OCCURENCE=2(liver),2(large_intestine)	NA
ZNF718	4	60210	rs3908749	G	A	Unknown	NM_001039127,NM_001286052,NM_001286053,NM_001286054,NM_001289931,NM_182524	unknown	4p16.3	ID=COSN19615617,COSN20466351,COSN19695458,COSN17001606,COSN20335714,COSN19746410,COSN19618066,COSN20279079;OCCURENCE=4(large_intestine),1(prostate),3(haematopoietic_and_lymphoid_tissue)	NA
ZNF718	4	67801	rs77175674	C	T	Unknown	NM_001039127,NM_001286052,NM_001286053,NM_001286054,NM_001289931,NM_182524	unknown	4p16.3	ID=COSN19699753,COSN19751413;OCCURENCE=2(large_intestine)	NA
MUC12	7	100635014	rs542320663	C	T	Synonymous SNV	NM_001164462	MUC12:NM_001164462:exon2:c.C1170T;p.H390H	7q22.1	NA	NA
MUC12	7	100635205	rs4729631	C	T	Missense SNV	NM_001164462	MUC12:NM_001164462:exon2:c.C1361T;p.A454V	7q22.1	ID=COSM4004183,COSM4004182;OCCURENCE=2(haematopoietic_and_lymphoid_tissue)	0.044,D
MUC12	7	100637193	rs10247974	C	A	Missense SNV	NM_001164462	MUC12:NM_001164462:exon2:c.C3349A;p.P1117T	7q22.1	ID=COSM4004188,COSM4004189;OCCURENCE=1(haematopoietic_and_lymphoid_tissue),1(thyroid),6(upper_aerodigestive_tract)	0.0,D
MUC12	7	100638828	rs56914801	C	T	Missense SNV	NM_001164462	MUC12:NM_001164462:exon2:c.C4984T;p.P1662S	7q22.1	ID=COSM4004198,COSM4004197;OCCURENCE=2(haematopoietic_and_lymphoid_tissue),2(thyroid)	0.237,T
MUC12	7	100646938	rs199993063	C	G	Missense SNV	NM_001164462	MUC12:NM_001164462:exon2:c.C13094G;p.T4365R	7q22.1	NA	0.0,D
MUC12	7	100647073	rs140284525	C	T	Missense SNV	NM_001164462	MUC12:NM_001164462:exon2:c.C13229T;p.T4410M	7q22.1	ID	