

Position (felCat9.0)	Ref	Alt	Gene Symbol	ENSEMBL Gene ID	ENSEMBL Transcript ID	Variant Type	Effect
A1:10292960	AC	A	n/a	ENSFAG00000031325	ENSFCAT00000027051	frameshift variant	HIGH
A1:83744719	C	A	n/a	ENSFAG00000044316	ENSFCAT00000050411	stop gained	HIGH
A1:86845101	CTTTG	C	n/a	ENSFAG00000035456	ENSFCAT00000049036	frameshift variant	HIGH
A2:73126183	C	T	n/a	ENSFAG00000045328	ENSFCAT00000050264	splice acceptor variant & non coding transcript variant	HIGH
A2:158967084	CTTCAGGT	C	CLCN1	ENSFAG00000007278	ENSFCAT00000007280	splice donor variant & coding sequence variant	HIGH
					ENSFCAT00000041228	splice donor variant & coding sequence variant	HIGH
A3:24651557	AGC	A	NCOA6	ENSFAG00000002306	ENSFCAT00000002306	frameshift variant	HIGH
					ENSFCAT00000062029	frameshift variant	HIGH
					ENSFCAT00000065640	frameshift variant	HIGH
A3:92580969	C	T	DUSP11 C2orf78	ENSFAG00000019224 ENSFAG000000026552	ENSFCAT00000019207	stop gained	HIGH
					ENSFCAT00000028153	upstream gene variant	MODIFIER
A3:93084265	CTG	C	n/a	ENSFAG00000008616	ENSFCAT00000008621	splice acceptor variant & intron variant	HIGH
					ENSFCAT00000046912	splice acceptor variant & intron variant	HIGH
					ENSFCAT00000047760	splice acceptor variant & intron variant	HIGH
					ENSFCAT00000052808	splice acceptor variant & intron variant	HIGH
B1:125723765	C	T	STPG2	ENSFAG00000040357	ENSFCAT00000050738	stop gained	HIGH
B1:187565548	GA	G	ZCHC4	ENSFAG00000023396	ENSFCAT00000031715	frameshift variant	HIGH
B4:96602070	T	TG	RAB3IP	ENSFAG00000015554	ENSFCAT00000015559	intron variant	MODIFIER
			n/a	ENSFAG00000046521	ENSFCAT00000058555	frameshift variant	HIGH
B4:121638249	AGG	A	RF00563	ENSFAG00000029370	ENSFCAT00000024452	upstream gene variant	MODIFIER
			SLC25A3	ENSFAG00000025087	ENSFCAT00000025087	frameshift variant	HIGH
				ENSFAG00000033040	ENSFCAT00000033040	frameshift variant	HIGH
				ENSFAG00000062282	ENSFCAT00000062282	frameshift variant	HIGH
B4:121638253	A	ATT	RF00563	ENSFAG00000029370	ENSFCAT00000024452	upstream gene variant	MODIFIER
			SLC25A3	ENSFAG00000025087	ENSFCAT00000025087	frameshift variant	HIGH
				ENSFAG00000033040	ENSFCAT00000033040	frameshift variant	HIGH
				ENSFAG00000062282	ENSFCAT00000062282	frameshift variant	HIGH
B4:127520915	T	C	n/a	ENSFAG00000036585	ENSFCAT00000043931	intron variant	MODIFIER
					ENSFCAT00000058001	start lost	HIGH
C1:12054155	C	T	n/a	ENSFAG00000044602	ENSFCAT00000042892	splice donor variant	HIGH
C1:106226700	GGA	G	SEMA6C	ENSFAG00000005557	ENSFCAT00000005559	frameshift variant	HIGH
					ENSFCAT00000043019	frameshift variant	HIGH
					ENSFCAT00000053872	3 prime UTR variant	MODIFIER
					ENSFCAT00000060036	downstream gene variant	MODIFIER
					ENSFCAT00000065994	3 prime UTR variant	MODIFIER
					ENSFCAT00000057479	start lost	HIGH
C1:107022276	C	T	n/a	ENSFAG00000041899	ENSFCAT00000008221	frameshift variant	HIGH
					ENSFCAT00000024736	intron variant	MODIFIER
					ENSFCAT00000038569	intron variant	MODIFIER
					ENSFCAT00000061772	frameshift variant	HIGH
C1:194039351	CTT	C	CREB1	ENSFAG00000008220	ENSFCAT000000031236	frameshift variant	HIGH
					ENSFCAT00000001167	stop gained	HIGH
					ENSFCAT00000001168	upstream gene variant	MODIFIER
D1:16618864	C	T	NLRX1	ENSFAG00000001167	ENSFCAT00000001167	stop gained	HIGH
			PDZD3	ENSFAG00000001168	ENSFCAT000000037361	upstream gene variant	MODIFIER
			NLRX1	ENSFAG00000001167	ENSFCAT00000043500	stop gained	HIGH
D2:33115866	A	AGCTG	TYSND1	ENSFAG00000044166	ENSFCAT00000060677	downstream gene variant	MODIFIER
					ENSFCAT00000062792	frameshift variant	HIGH
D2:59476548	G	GTGGGTGACAGGAGCTG GGATGGAGGCTCTGCCA CCAGGCCCTGTCTCAAT C	PYROXD2	ENSFAG00000009346	ENSFCAT00000009349	frameshift variant	HIGH
E1:9602491	T	TC	n/a	ENSFAG000000031027	ENSFCAT00000028891	intron variant	MODIFIER
			PIGL	ENSFAG00000010824	ENSFCAT00000065686	frameshift variant	HIGH
E1:38746457	CG	C	HOXB3	ENSFAG00000006408	ENSFCAT00000006410	upstream gene variant	MODIFIER
			MIR10A	ENSFAG00000016475	ENSFCAT00000016479	downstream gene variant	MODIFIER
			HOXB4	ENSFAG00000041872	ENSFCAT00000062069	frameshift variant	HIGH
			GPR179	ENSFAG00000005812	ENSFCAT00000005815	stop gained	HIGH
E1:51024511	AGGGGCGCTGGGTGGCGCAGTC GGTTAAGCGTCCGATTCAGCCAG GTCACGATCTCGCGTCCGTGAGT TTGAGCCCCGCTGGGCTCTGG GCTGATGGCTCAGAGCTGGAGC CTGTTTCTGATTCTGTCTCCCTC TCTCTGCCCCCTCCCCGTTCTATG CTCTGCTCTCTCTGCCAAAAAT AAATAAATGTTAAAAAAAATTT AAAAAAAAAAAAAAAAAAGAA ATTCTAAGC	A	n/a	ENSFAG00000042903	ENSFCAT00000053189	splice donor variant & non coding transcript exon variant & intron variant	HIGH
E2:13446733	T	C	LIPE	ENSFAG000000022678	ENSFCAT00000026802	stop lost	HIGH
			n/a	ENSFAG000000037892	ENSFCAT00000045735	intron variant	MODIFIER
E2:61620435	T	A	n/a	ENSFAG00000044866	ENSFCAT00000050297	upstream gene variant	MODIFIER

					ENSFECAT00000052013	splice acceptor variant & non coding transcript variant	HIGH
					ENSFECAT00000055872	splice acceptor variant & non coding transcript variant	HIGH
F1:70503299	C	G	DCST1	ENSFCAG00000018847	ENSFECAT00000018172	start lost	HIGH
					ENSFECAT00000058210	start lost	HIGH
			DCST2	ENSFCAG00000040157	ENSFECAT00000053006	intron variant	MODIFIER

Table 1. A list of variants unique to the proband that were called high impact by VEP and the effect in all associated transcripts.

Position (felCat9.0)	Ref	Alt	Gene Symbol	ENSEMBL Gene ID	ENSEMBL Transcript ID	Variant Type	Effect
A1:71234391	A	G	CLYBL	ENSCFCAG00000031043	ENSCFCAT00000034646	missense variant	MODERATE
A1:84177713	C	T	n/a	ENSCFCAG00000022087	ENSCFCAT00000050511	intron variant	MODIFIER
A1:91378473	T	A	BTNL9	ENSCFCAG00000000991	ENSCFCAT00000002092	missense variant	MODERATE
A1:111919100	G	A	SHROOM1	ENSCFCAG00000022651	ENSCFCAT00000000991	missense variant	MODERATE
A1:113672271	G	T	CAMLG	ENSCFCAG00000026813	ENSCFCAT00000034165	missense variant	MODERATE
A1:116898026	C	T	CDC25C	ENSCFCAG00000031992	ENSCFCAT00000054663	missense variant	MODERATE
A1:151761784	G	A	n/a	ENSCFCAG00000022528	ENSCFCAT00000022809	missense variant	MODERATE
A1:151762007	C	G	n/a	ENSCFCAG00000022528	ENSCFCAT00000034960	missense variant	MODERATE
A1:159686340	ATCT	A	TTC37	ENSCFCAG00000010817	ENSCFCAT00000022858	missense variant	MODERATE
A1:200642178	T	G	SPINK13	ENSCFCAG00000041823	ENSCFCAT00000028713	missense variant	MODERATE
A1:208845978	T	C	C7	ENSCFCAG00000031466	ENSCFCAT00000055412	missense variant	MODERATE
A1:215879699	C	T	ZFR	ENSCFCAG00000000354	ENSCFCAT00000029163	missense variant	MODERATE
A1:226396297	G	A	CDH18	ENSCFCAG00000013641	ENSCFCAT00000010822	inframe deletion	MODERATE
A1:227755754	C	T	n/a	ENSCFCAG00000027514	ENSCFCAT00000059783	missense variant	MODERATE
A1:228450994	G	A	MYO10	ENSCFCAG00000010325	ENSCFCAT00000031719	missense variant	MODERATE
A1:234525747	C	T	TAS2R1	ENSCFCAG00000012611	ENSCFCAT00000000354	missense variant	MODERATE
A2:1898631	G	A	THOP1	ENSCFCAG00000015785	ENSCFCAT00000013645	missense variant	MODERATE
A2:3447731	T	C	SEMA6B	ENSCFCAG00000019404	ENSCFCAT00000042027	missense variant	MODERATE
A2:6705658	G	C	n/a	ENSCFCAG00000018988	ENSCFCAT00000065136	missense variant	MODERATE
A2:8445900	G	A	LDLR	ENSCFCAG00000004895	ENSCFCAT00000031977	missense variant & splice region variant	MODERATE
A2:12858503	C	T	CCDC194	ENSCFCAG00000046586	ENSCFCAT00000010331	missense variant	MODERATE
A2:13640385	C	T	ELL	ENSCFCAG00000009739	ENSCFCAT00000047832	missense variant	MODERATE
A2:18819664	G	A	UBA7	ENSCFCAG00000010680	ENSCFCAT00000012613	missense variant	MODERATE
A2:20512376	C	T	n/a	ENSCFCAG00000019068	ENSCFCAT00000015789	missense variant	MODERATE
A2:20896501	G	C	GLYCTK	ENSCFCAG00000015709	ENSCFCAT00000019041	missense variant	MODIFIER
A2:20896867	C	T	MIR135A1	ENSCFCAG00000030718	ENSCFCAT00000063409	missense variant	MODERATE
A2:20947076	G	A	DNAH1	ENSCFCAG00000015710	ENSCFCAT00000018708	missense variant & splice region variant	MODERATE
A2:20954818	G	A	DNAH1	ENSCFCAG00000015710	ENSCFCAT00000048996	missense variant	MODERATE
A2:20969580	C	T	DNAH1	ENSCFCAG00000015710	ENSCFCAT00000066000	missense variant	MODERATE
A2:20975304	C	T	DNAH1	ENSCFCAG00000015710	ENSCFCAT00000066710	missense variant	MODERATE
A2:20986918	A	G	DNAH1	ENSCFCAG00000015710	ENSCFCAT00000056716	missense variant	MODERATE
A2:20995868	T	G	BAP1	ENSCFCAG00000015711	ENSCFCAT0000009741	missense variant	MODERATE
A2:21355829	C	T	ITIH3	ENSCFCAG00000015723	ENSCFCAT00000009741	missense variant	MODERATE
A2:23306612	A	G	CACNA2D3	ENSCFCAG00000010121	ENSCFCAT00000010683	missense variant	MODERATE
A2:66307090	A	G	SPATA48	ENSCFCAG00000038962	ENSCFCAT00000019101	missense variant	MODERATE
A2:77886608	T	C	SFRP4	ENSCFCAG00000025247	ENSCFCAT00000015713	missense variant	MODERATE
A2:88971202	G	T	PCLO	ENSCFCAG00000002901	ENSCFCAT00000025862	missense variant	MODIFIER
A2:100778052	C	T	DYNC111	ENSCFCAG00000011808	ENSCFCAT00000015713	missense variant	MODERATE
A2:107175563	T	G	VWDE	ENSCFCAG00000024923	ENSCFCAT00000015713	missense variant	MODERATE
A2:107381020	C	G	SCIN	ENSCFCAG00000005088	ENSCFCAT00000015713	missense variant	MODERATE
A2:124558814	C	T	ITPRID1	ENSCFCAG00000000212	ENSCFCAT00000015713	missense variant	MODERATE

A2:124620226	T	A	PPP1R17	ENSFCA00000033490	ENSFCA00000047229	missense variant	MODERATE
					ENSFCA00000052229	missense variant	MODERATE
					ENSFCA00000054005	missense variant	MODERATE
A2:166065585	C	T	KMT2C	ENSFCA000000008632	ENSFCA000000008634	missense variant	MODERATE
A2:166065695	C	T	KMT2C	ENSFCA000000008632	ENSFCA000000008634	missense variant	MODERATE
A3:28959300	A	G	n/a	ENSFCA000000040585	ENSFCA00000032363	missense variant	MODERATE
					ENSFCA00000038516	missense variant	MODERATE
A3:56844395	CGCGGTACATG	C	C2orf40	ENSFCA000000023685	ENSFCA000000030878	inframe deletion	MODERATE
A3:81535408	C	T	PUS10	ENSFCA000000008019	ENSFCA00000008021	missense variant	MODERATE
			n/a	ENSFCA000000028969	ENSFCA000000052531	missense variant	MODERATE
			n/a	ENSFCA000000028969	ENSFCA000000022328	upstream gene variant	MODIFIER
A3:84687830	G	C	AFTP	ENSFCA000000028719	ENSFCA000000023897	missense variant	MODERATE
					ENSFCA000000048766	missense variant	MODERATE
A3:89709015	G	A	PCYOX1	ENSFCA000000030129	ENSFCA000000031948	missense variant	MODERATE
			n/a	ENSFCA000000045921	ENSFCA000000058356	downstream gene variant	MODIFIER
			n/a	ENSFCA000000045921	ENSFCA000000062195	downstream gene variant	MODIFIER
A3:101970247	A	G	DNAH6	ENSFCA000000000768	ENSFCA000000000768	missense variant	MODERATE
A3:107592982	C	T	DUSP2	ENSFCA000000010694	ENSFCA000000010696	missense variant	MODERATE
B1:1888286	G	A	MYOM2	ENSFCA000000004727	ENSFCA00000004734	missense variant	MODERATE
					ENSFCA000000061368	missense variant	MODERATE
B1:7091214	C	G	n/a	ENSFCA000000046076	ENSFCA000000058055	missense variant	MODERATE
B1:17112211	A	G	CYP4V2	ENSFCA000000001547	ENSFCA000000001547	missense variant	MODERATE
B1:21265815	G	A	MTMR7	ENSFCA000000011961	ENSFCA000000011964	missense variant	MODERATE
B1:21634070	C	A	MICU3	ENSFCA000000018279	ENSFCA000000018303	missense variant	MODERATE
					ENSFCA000000049756	missense variant	MODERATE
					ENSFCA000000024267	missense variant	MODERATE
B1:42845162	T	C	AP3M2	ENSFCA000000031044	ENSFCA000000051861	missense variant	MODERATE
					ENSFCA000000065406	missense variant	MODERATE
					ENSFCA000000037425	missense variant	MODERATE
B1:65297575	A	G	TMEM192	ENSFCA000000038577	ENSFCA000000037425	missense variant	MODERATE
B1:67245701	G	T	NAF1	ENSFCA000000031652	ENSFCA000000061274	missense variant	MODERATE
B1:115410999	C	T	EGF	ENSFCA000000000506	ENSFCA000000000506	missense variant	MODERATE
					ENSFCA000000024046	missense variant	MODERATE
					ENSFCA000000034737	missense variant	MODERATE
					ENSFCA000000063173	missense variant	MODERATE
B1:115593791	A	G	n/a	ENSFCA000000000105	ENSFCA000000000105	intron variant	MODIFIER
					ENSFCA000000033262	intron variant	MODIFIER
					ENSFCA000000034598	upstream gene variant	MODIFIER
					ENSFCA000000035284	missense variant	MODERATE
					ENSFCA000000046626	upstream gene variant	MODIFIER
B1:126201532	A	G	n/a	ENSFCA000000035537	ENSFCA000000000105	intron variant	MODIFIER
					ENSFCA000000000105	intron variant	MODIFIER
B1:137463098	C	G	n/a	ENSFCA000000030152	ENSFCA000000041970	missense variant	MODERATE
B1:138153680	G	A	WDFY3	ENSFCA000000002946	ENSFCA000000049515	missense variant	MODERATE
B1:147580951	C	A	AREG	ENSFCA000000000370	ENSFCA000000002946	missense variant	MODERATE
					ENSFCA00000000370	5 prime UTR variant	MODIFIER
B1:203565473	G	A	n/a	ENSFCA000000046639	ENSFCA000000051865	missense variant	MODERATE
B1:203703029	G	T	KIAA0232	ENSFCA000000010024	ENSFCA000000066858	missense variant	MODERATE
B2:3482796	G	A	n/a	ENSFCA000000024356	ENSFCA000000010026	missense variant	MODERATE
B2:11920840	G	T	NUP153	ENSFCA000000011583	ENSFCA000000031800	missense variant	MODERATE
					ENSFCA000000011588	missense variant	MODERATE
					ENSFCA000000039376	missense variant	MODERATE
B2:18445365	G	A	n/a	ENSFCA000000038080	ENSFCA000000047179	downstream gene variant	MODIFIER
					ENSFCA000000040685	missense variant	MODERATE
B2:103086853	T	C	REV3L	ENSFCA000000028995	ENSFCA000000058597	missense variant	MODERATE
					ENSFCA000000025874	missense variant	MODERATE
B2:110042154	C	T	MCM9	ENSFCA000000028710	ENSFCA000000053725	missense variant	MODERATE
					ENSFCA000000026168	missense variant	MODERATE
B2:125733137	G	A	MTFR2	ENSFCA000000030038	ENSFCA000000036278	intron variant	MODIFIER
B2:127552387	C	T	ARFGEF3	ENSFCA000000013904	ENSFCA000000031085	missense variant	MODERATE
B2:152910321	C	T	UNC93A	ENSFCA000000006973	ENSFCA000000013910	missense variant	MODERATE
					ENSFCA00000006975	missense variant	MODERATE
B3:1157394	C	T	MINAR1	ENSFCA000000030462	ENSFCA000000045258	missense variant	MODERATE
B3:2077050	T	C	CEMIP	ENSFCA000000012937	ENSFCA000000022980	missense variant	MODERATE
B3:2113570	G	A	CEMIP	ENSFCA000000012937	ENSFCA000000012941	missense variant	MODERATE
B3:5551356	C	T	UNC45A	ENSFCA00000006097	ENSFCA000000006099	downstream gene variant	MODIFIER
					ENSFCA000000061787	downstream gene variant	MODIFIER
B3:48283707	C	T	RCCD1	ENSFCA00000006098	ENSFCA000000006100	missense variant	MODERATE
B3:60524114	T	G	LIPC	ENSFCA000000008009	ENSFCA000000008012	missense variant	MODERATE
B3:71343323	G	A	RVR3	ENSFCA000000000742	ENSFCA000000019156	missense variant	MODERATE
					ENSFCA000000000742	missense variant	MODERATE
B3:76288090	C	T	THTPA	ENSFCA000000012059	ENSFCA000000012062	downstream gene variant	MODIFIER
			JPH4	ENSFCA000000018964	ENSFCA000000018381	downstream gene variant	MODIFIER
			AP1G2	ENSFCA000000040037	ENSFCA000000042508	missense variant	MODERATE

B3:9971839	A	G	KLHDC1	ENSFCA00000036283	ENSFCAT00000038314	missense variant	MODERATE
B3:99820836	C	G	NEMF	ENSFCA00000018946	ENSFCAT00000019170	missense variant	MODERATE
			KLHDC2	ENSFCA00000031902	ENSFCA00000028217	downstream gene variant	MODIFIER
					ENSFCA00000037457	downstream gene variant	MODIFIER
B3:100769651	G	T	ABHD12B	ENSFCA00000002777	ENSFCAT00000002777	downstream gene variant	MODIFIER
			PYGL	ENSFCA00000002778	ENSFCA00000002778	missense variant	MODERATE
					ENSFCA00000040133	missense variant	MODERATE
B3:124433403	C	T	TMED8	ENSFCA00000011732	ENSFCAT00000011734	missense variant	MODERATE
B3:124439476	G	T	TMED8	ENSFCA00000011732	ENSFCAT00000011734	missense variant	MODERATE
B3:124468274	T	C	NOXRED1	ENSFCA00000039154	ENSFCAT00000046884	downstream gene variant	MODIFIER
			n/a	ENSFCA00000042664	ENSFCAT00000060305	missense variant	MODERATE
B3:124719696	A	T	ALKB1	ENSFCA00000037897	ENSFCAT00000037675	missense variant	MODERATE
			SLRP	ENSFCA00000036326	ENSFCAT00000039204	upstream gene variant	MODIFIER
			SNW1	ENSFCA00000001637	ENSFCAT00000001637	intron variant	MODIFIER
B3:124734389	C	T	SLRP	ENSFCA00000036326	ENSFCAT00000039204	missense variant & splice region variant	MODERATE
			TTC7B	ENSFCA00000001583	ENSFCA00000001583	missense variant	MODERATE
ENSFCA00000058981	missense variant	MODERATE					
ENSFCA00000063691	missense variant	MODERATE					
ENSFCA00000067224	missense variant	MODERATE					
B3:139320529	C	T	DDX24	ENSFCA00000010229	ENSFCA00000010232	missense variant	MODERATE
					ENSFCA00000065890	missense variant	MODERATE
B3:139329845	C	T	DDX24	ENSFCA00000010229	ENSFCA00000010232	missense variant	MODERATE
					ENSFCA00000065890	missense variant	MODERATE
B3:140225968	C	T	DICER1	ENSFCA00000003535	ENSFCA00000003537	missense variant	MODERATE
					ENSFCA00000037515	missense variant	MODERATE
B4:5397248	G	A	n/a	ENSFCA00000009007	ENSFCA00000009009	missense variant	MODERATE
B4:25101091	G	A	MASTL	ENSFCA00000014240	ENSFCA00000014244	missense variant	MODERATE
					ENSFCA00000034455	missense variant	MODERATE
B4:46708262	T	G	KLRK1	ENSFCA00000040686	ENSFCAT00000053631	missense variant	MODERATE
B4:46708276	T	C	KLRK1	ENSFCA00000040686	ENSFCAT00000053631	missense variant	MODERATE
B4:46993758	T	C	n/a	ENSFCA00000025057	ENSFCA00000022489	missense variant & splice region variant	MODERATE
					ENSFCA00000050581	missense variant & splice region variant	MODERATE
B4:81064790	A	G	KRT74	ENSFCA00000000290	ENSFCA00000000288	missense variant	MODERATE
					ENSFCA00000050616	missense variant	MODERATE
B4:81072162	A	C	KRT74	ENSFCA00000000290	ENSFCA00000000288	missense variant	MODERATE
					ENSFCA00000050616	missense variant	MODERATE
B4:81164526	A	G	KRT4	ENSFCA00000000295	ENSFCAT00000000295	intron variant	MODIFIER
			KRT1	ENSFCA00000042595	ENSFCAT00000055089	missense variant	MODERATE
B4:82671186	C	T	ZNF385A	ENSFCA00000010154	ENSFCA00000010157	missense variant	MODERATE
					ENSFCA00000053337	intron variant	MODIFIER
B4:115206387	G	A	LUM	ENSFCA00000003764	ENSFCA00000003763	missense variant	MODERATE
					ENSFCA00000033907	missense variant	MODERATE
B4:126019306	G	A	STAB2	ENSFCA00000007290	ENSFCAT00000007294	missense variant	MODERATE
B4:126124621	G	A	STAB2	ENSFCA00000007290	ENSFCAT00000007294	missense variant	MODERATE
B4:126578829	G	A	TXNRD1	ENSFCA00000003645	ENSFCA00000003645	intron variant	MODIFIER
					ENSFCA00000044250	missense variant	MODERATE
B4:127520710	T	G	n/a	ENSFCA00000036585	ENSFCA00000043931	missense variant	MODERATE
					ENSFCA00000058001	intron variant	MODIFIER
B4:127520870	T	C	n/a	ENSFCA00000036585	ENSFCA00000043931	missense variant & splice region variant	MODERATE
					ENSFCA00000036585	ENSFCA00000058001	missense variant & splice region variant
B4:127524132	C	G	n/a	ENSFCA00000036585	ENSFCA00000043931	missense variant	MODERATE
					ENSFCA00000058001	upstream gene variant	MODIFIER
C1:1984637	G	A	MEGF6	ENSFCA00000031840	ENSFCA00000032322	missense variant	MODERATE
					ENSFCA00000048790	missense variant	MODERATE
C1:7687386	C	T	CAS21	ENSFCA00000014593	ENSFCA00000014597	missense variant	MODERATE
					ENSFCA00000054128	missense variant	MODERATE
					ENSFCA00000058061	missense variant	MODERATE
C1:18305859	G	A	NIPAL3	ENSFCA00000002067	ENSFCA00000002067	downstream gene variant	MODIFIER
					ENSFCA00000036295	missense variant	MODERATE
C1:24065622	T	C	COL16A1	ENSFCA00000015669	ENSFCA00000015679	intron variant	MODIFIER
					ENSFCA00000044147	missense variant	MODERATE
C1:30550610	C	T	MACF1	ENSFCA00000045633	ENSFCA00000044017	missense variant	MODERATE
					ENSFCA00000055061	missense variant	MODERATE
					ENSFCA00000055111	missense variant	MODERATE
					ENSFCA00000064822	missense variant	MODERATE
C1:42573761	G	A	TUT4	ENSFCA00000003246	ENSFCA00000003246	missense variant	MODERATE
C1:51611279	T	C	DOCK7	ENSFCA00000022720	ENSFCA00000032284	missense variant	MODERATE
					ENSFCA00000048381	missense variant	MODERATE
C1:73288084	G	A	CLCA2	ENSFCA00000008210	ENSFCAT00000008212	missense variant	MODERATE

C1:104413373	C	T	PDZK1	ENSFCAG00000013368	ENSFCAT00000013371	missense variant	MODERATE
			n/a	ENSFCAG00000024477	ENSFCAT00000045829	missense variant	MODERATE
C1:107100857	T	G	TCHHL1	ENSFCAG00000046571	ENSFCAT00000052456	missense variant	MODERATE
C1:113656609	A	G	MYO7B	ENSFCAG00000013176	ENSFCAT00000013180	missense variant	MODERATE
					ENSFCAT00000036804	intron variant	MODIFIER
					ENSFCAT00000038750	intron variant	MODIFIER
C1:113705761	C	A	IWS1	ENSFCAG00000009026	ENSFCAT00000009028	missense variant	MODERATE
					ENSFCAT00000037913	intron variant	MODIFIER
C1:154085770	G	A	IFIH1	ENSFCAG00000023951	ENSFCAT00000007305	missense variant	MODERATE
					ENSFCAT00000026735	missense variant	MODERATE
					ENSFCAT00000061411	missense variant	MODERATE
C1:161995864	GGCCGCGGCC	G	SP5	ENSFCAG00000044347	ENSFCAT00000050366	inframe deletion	MODERATE
C1:164281618	G	A	n/a	ENSFCAG00000008059	ENSFCAT00000054583	missense variant	MODERATE
C1:164350082	T	C	CDCA7	ENSFCAG00000005476	ENSFCAT00000005479	missense variant	MODERATE
					ENSFCAT00000042357	missense variant	MODERATE
C1:165241721	G	GGAA	CIR1	ENSFCAG00000022845	ENSFCAT00000050183	missense variant	MODERATE
					ENSFCAT00000030886	inframe insertion	MODERATE
C1:165593144	A	T	CHRNA1	ENSFCAG00000014103	ENSFCAT00000014107	missense variant	MODERATE
					ENSFCAT00000054627	missense variant	MODERATE
					ENSFCAT00000065105	missense variant	MODERATE
C1:169226811	G	A	n/a	ENSFCAG00000046559	ENSFCAT00000054332	missense variant	MODERATE
					ENSFCAT00000057628	missense variant	MODERATE
C1:195274259	G	A	n/a	ENSFCAG00000041864	ENSFCAT00000031236	missense variant	MODERATE
C1:203392796	T	C	AAMP	ENSFCAG00000002372	ENSFCAT00000002372	upstream gene variant	MODIFIER
					ENSFCAT00000053285	upstream gene variant	MODIFIER
			TMBIM1	ENSFCAG00000002374	ENSFCAT00000002374	downstream gene variant	MODIFIER
					n/a	ENSFCAG00000040250	ENSFCAT00000053237
C1:219283421	C	T	COL6A3	ENSFCAG00000012720	ENSFCAT00000012724	missense variant	MODERATE
					ENSFCAT00000051285	missense variant	MODERATE
					ENSFCAT00000052409	missense variant	MODERATE
					ENSFCAT00000056790	missense variant	MODERATE
					ENSFCAT00000063857	missense variant	MODERATE
C1:220011772	A	G	n/a	ENSFCAG00000046551	ENSFCAT00000052753	missense variant	MODERATE
C1:221728541	G	C	ANKMY1	ENSFCAG00000012464	ENSFCAT00000012469	missense variant	MODERATE
					ENSFCAT00000036897	missense variant	MODERATE
					ENSFCAT00000037799	missense variant	MODERATE
					ENSFCAT00000039736	missense variant	MODERATE
					ENSFCAT00000040710	missense variant	MODERATE
C1:221768848	C	T	CAPN10	ENSFCAG00000012470	ENSFCAT00000012473	missense variant	MODERATE
					ENSFCAT00000036494	missense variant	MODERATE
					ENSFCAT00000063353	intron variant	MODIFIER
					ENSFCAT00000063602	intron variant	MODIFIER
C1:221791166	C	T	GPR35	ENSFCAG00000024715	ENSFCAT00000027678	missense variant	MODERATE
C2:650249	G	A	SPATC1L	ENSFCAG00000014816	ENSFCAT00000014820	missense variant	MODERATE
C2:1931874	G	GTGAGCACACGGGCT	TSPEAR	ENSFCAG00000001772	ENSFCAT00000001772	intron variant	MODIFIER
					ENSFCAT00000035693	inframe insertion	MODERATE
			n/a	ENSFCAG00000033709	ENSFCAT00000034604	downstream gene variant	MODIFIER
C2:2111583	C	T	TRPM2	ENSFCAG00000009173	ENSFCAT00000009175	missense variant	MODERATE
					ENSFCAT00000056225	missense variant	MODERATE
					ENSFCAT00000060156	missense variant	MODERATE
			RF00285	ENSFCAG00000035800	ENSFCAT00000033694	downstream gene variant	MODIFIER
ENSFCAG00000034468	ENSFCAT00000036764	downstream gene variant		MODIFIER			
C2:2117668	GTCC	G	TRPM2	ENSFCAG00000009173	ENSFCAT00000009175	inframe deletion	MODERATE
					ENSFCAT00000056225	inframe deletion	MODERATE
					ENSFCAT00000060156	inframe deletion	MODERATE
			RF00285	ENSFCAG00000035800	ENSFCAT00000033694	upstream gene variant	MODIFIER
ENSFCAG00000034468	ENSFCAT00000036764	upstream gene variant		MODIFIER			
C2:2170115	G	C	CFAP410	ENSFCAG00000040115	ENSFCAT00000040416	missense variant	MODERATE
					ENSFCAT00000059030	missense variant	MODERATE
C2:2359138	C	T	n/a	ENSFCAG00000004487	ENSFCAT00000004491	missense variant	MODERATE
					ENSFCAT00000037759	missense variant	MODERATE
					ENSFCAT00000008440	downstream gene variant	MODIFIER
C2:8372249	C	T	VPS26C	ENSFCAG00000012830	ENSFCAT00000012834	missense variant	MODERATE
					ENSFCAT00000047540	missense variant	MODERATE
					ENSFCAT00000050115	missense variant	MODERATE
					ENSFCAT00000060256	missense variant	MODERATE
C2:48628614	G	A	TFG	ENSFCAG00000025659	ENSFCAT00000027637	missense variant	MODERATE
					ENSFCAT00000050806	missense variant	MODERATE
					ENSFCAT00000059364	missense variant	MODERATE
C2:55837148	G	A	RETNLB	ENSFCAG00000036016	ENSFCAT00000049101	missense variant	MODERATE
C2:84089300	C	A	SEN2P	ENSFCAG00000042538	ENSFCAT00000055178	missense variant	MODERATE
C2:95840959	G	A	NCEH1	ENSFCAG00000028457	ENSFCAT00000024541	missense variant	MODERATE
C2:107223987	C	A	IL12A	ENSFCAG00000011130	ENSFCAT00000011133	missense variant	MODERATE
C2:116164833	C	T	TSC22D2	ENSFCAG00000031850	ENSFCAT00000023131	missense variant	MODERATE
					ENSFCAT00000047483	missense variant	MODERATE

C2:156182225	G	A	DCLK3	ENSFCAG00000022392	ENSFCAT00000032249	missense variant	MODERATE	
					ENSFCAT00000063849	missense variant	MODERATE	
D1:1515464	C	T	MMP13	ENSFCAG00000000118	ENSFCAT00000000118	missense variant	MODERATE	
D1:12166851	G	A	NNMT	ENSFCAG000000037839	ENSFCAT00000037849	missense variant	MODERATE	
					ENSFCAT00000001167	missense variant	MODERATE	
D1:16618344	G	A	NLRX1	ENSFCAG000000001167	ENSFCAT00000043500	missense variant	MODERATE	
					ENSFCAT00000001168	upstream gene variant	MODIFIER	
			PDZD3	ENSFCAG000000001168	ENSFCAT00000037361	upstream gene variant	MODIFIER	
D1:16627280	G	A	PDZD3	ENSFCAG000000001168	ENSFCAT00000001168	downstream gene variant	MODIFIER	
					ENSFCAT00000037361	downstream gene variant	MODIFIER	
			CCDC153	ENSFCAG000000001169	ENSFCAT00000001169	missense variant	MODERATE	
D1:16629401	C	T	PDZD3	ENSFCAG000000001168	ENSFCAT00000001168	downstream gene variant	MODIFIER	
					ENSFCAT00000037361	downstream gene variant	MODIFIER	
			CCDC153	ENSFCAG000000001169	ENSFCAT00000001169	missense variant	MODERATE	
D1:23231738	A	T	RPUSD4	ENSFCAG000000029789	ENSFCAT00000022983	missense variant	MODERATE	
D1:40592747	A	G	SESN3	ENSFCAG000000005081	ENSFCAT00000005082	missense variant	MODERATE	
					ENSFCAT00000056681	missense variant	MODERATE	
					ENSFCAT00000057835	missense variant	MODERATE	
D1:61137254	C	CCCCCGG	ARAP1	ENSFCAG000000013192	ENSFCAT000000013196	protein altering variant	MODERATE	
					ENSFCAT00000055940	protein altering variant	MODERATE	
D1:89038010	A	G	QSER1	ENSFCAG000000015204	ENSFCAT000000015210	missense variant	MODERATE	
					ENSFCAT00000040727	missense variant	MODERATE	
					ENSFCAT00000062010	missense variant	MODERATE	
D1:109146850	A	AGGC	RTN3	ENSFCAG000000023202	ENSFCAT000000024505	downstream gene variant	MODIFIER	
					ENSFCAT00000066093	downstream gene variant	MODIFIER	
			C11orf95	ENSFCAG000000032639	ENSFCAT00000033573	inframe insertion	MODERATE	
					ENSFCAT00000055356	downstream gene variant	MODIFIER	
D1:110362164	C	T	CAPN1	ENSFCAG000000001438	ENSFCAT000000001438	missense variant	MODERATE	
					ENSFCAT00000058310	missense variant	MODERATE	
D1:113819532	G	T	CCND1	ENSFCAG000000008358	ENSFCAT000000008360	missense variant	MODERATE	
					ENSFCAT00000058874	downstream gene variant	MODIFIER	
D2:6677819	C	A	A1CF	ENSFCAG000000011779	ENSFCAT000000011182	missense variant	MODERATE	
					ENSFCAT00000067045	missense variant	MODERATE	
D2:8660764	A	C	FAS	ENSFCAG000000033129	ENSFCAT000000036758	missense variant	MODERATE	
					ENSFCAT00000001248	missense variant	MODERATE	
D2:62911000	C	T	ACTR1A	ENSFCAG000000001248	ENSFCAT000000058888	downstream gene variant	MODIFIER	
			MFSD13A	ENSFCAG000000023881	ENSFCAT000000028865	downstream gene variant	MODIFIER	
D2:64308953	C	T	COL17A1	ENSFCAG000000004299	ENSFCAT000000004301	missense variant	MODERATE	
					ENSFCAT000000046187	missense variant	MODERATE	
D3:19138629	C	A	GSTT4	ENSFCAG000000036794	ENSFCAT000000045313	missense variant	MODERATE	
D3:19160731	C	A	n/a	ENSFCAG000000022623	ENSFCAT000000022132	downstream gene variant	MODIFIER	
					ENSFCAG000000035422	ENSFCAT000000040173	missense variant	MODERATE
D3:55423196	G	C	DSG2	ENSFCAG000000031128	ENSFCAT000000023465	missense variant	MODERATE	
D3:55423210	G	A	DSG2	ENSFCAG000000031128	ENSFCAT000000023465	missense variant	MODERATE	
					ENSFCAT00000008816	missense variant	MODERATE	
D4:11831268	C	T	n/a	ENSFCAG000000008814	ENSFCAT000000058936	missense variant	MODERATE	
					ENSFCAT00000060196	missense variant	MODERATE	
					ENSFCAT00000061958	missense variant	MODERATE	
					ENSFCAT00000064926	missense variant	MODERATE	
D4:17623266	G	A	PCSK5	ENSFCAG000000027896	ENSFCAT000000024077	missense variant	MODERATE	
					ENSFCAT00000050249	missense variant	MODERATE	
					ENSFCAT00000059461	5 prime UTR variant	MODIFIER	
D4:18650803	G	G	FOXB2	ENSFCAG000000046576	ENSFCAT000000056177	inframe deletion	MODERATE	
D4:29961035	T	C	n/a	ENSFCAG000000002729	ENSFCAT000000002729	missense variant	MODERATE	
D4:61261685	G	C	FRMPD1	ENSFCAG000000011920	ENSFCAT000000011923	missense variant	MODERATE	
			TRMT10B	ENSFCAG000000035239	ENSFCAT000000040640	upstream gene variant	MODIFIER	
D4:61380631	A	G	n/a	ENSFCAG000000036372	ENSFCAT000000036949	missense variant	MODERATE	
					ENSFCAT000000008629	missense variant	MODERATE	
D4:81633846	G	A	CDK5RAP2	ENSFCAG000000008625	ENSFCAT000000054488	missense variant	MODERATE	
					ENSFCAT000000057225	missense variant	MODERATE	
D4:81969996	G	A	PSMD5	ENSFCAG000000001313	ENSFCAT000000001312	downstream gene variant	MODIFIER	
					ENSFCAT000000057489	downstream gene variant	MODIFIER	
			B3GNT10	ENSFCAG000000022327	ENSFCAT000000029226	missense variant	MODERATE	

D4:91123222	G	C	LAMC3	ENSFACAG00000005118	ENSFACAT00000005119	missense variant	MODERATE
E1:14535925	A	T	MNT	ENSFACAG00000006315	ENSFACAT000000035165	missense variant	MODERATE
					ENSFACAT000000058918	downstream gene variant	MODIFIER
E1:15201590	G	A	SCARF1	ENSFACAG000000044300	ENSFACAT000000062099	missense variant	MODERATE
E1:16753714	C	T	NSRP1	ENSFACAG000000039697	ENSFACAT000000035282	missense variant	MODERATE
E1:16968924	C	T	EFCAB5	ENSFACAG00000004649	ENSFACAT00000004651	missense variant	MODERATE
			n/a	ENSFACAG000000044178	ENSFACAT000000057662	upstream gene variant	MODIFIER
E1:17328108	C	T	ABHD15	ENSFACAG00000002756	ENSFACAT00000002756	upstream gene variant	MODIFIER
			TP53I13	ENSFACAG00000002757	ENSFACAT00000002757	missense variant	MODERATE
			GIT1	ENSFACAG00000002758	ENSFACAT000000042777	downstream gene variant	MODIFIER
					ENSFACAT000000047071	downstream gene variant	MODIFIER
E1:30545053	C	T	TSPOAP1	ENSFACAG000000012264	ENSFACAT000000012268	missense variant	MODERATE
					ENSFACAT000000044954	missense variant	MODERATE
E1:30636189	G	A	MKS1	ENSFACAG000000012261	ENSFACAT000000012264	missense variant	MODERATE
			n/a	ENSFACAG000000034800	ENSFACAT000000052159	missense variant	MODERATE
					ENSFACAT000000033967	downstream gene variant	MODIFIER
E1:30682939	G	A	OR4D1	ENSFACAG000000044467	ENSFACAT000000030441	missense variant	MODERATE
E1:31056420	C	T	CCDC182	ENSFACAG000000001699	ENSFACAT000000001699	missense variant	MODERATE
E1:36539075	C	T	MBTD1	ENSFACAG000000006743	ENSFACAT000000006745	upstream gene variant	MODIFIER
					ENSFACAT000000041488	upstream gene variant	MODIFIER
			UTP18	ENSFACAG000000031396	ENSFACAT000000029778	missense variant	MODERATE
					ENSFACAT000000030760	missense variant	MODERATE
E1:37265200	G	A	EME1	ENSFACAG000000012332	ENSFACAT000000012335	downstream gene variant	MODIFIER
			LRRC59	ENSFACAG000000031770	ENSFACAT000000022309	missense variant	MODERATE
E1:41039872	G	A	MED24	ENSFACAG000000014993	ENSFACAT000000014997	intron variant	MODIFIER
					ENSFACAT000000041870	missense variant	MODERATE
					ENSFACAT000000043432	intron variant	MODIFIER
					ENSFACAT000000046079	intron variant	MODIFIER
					ENSFACAT000000051893	intron variant	MODIFIER
			RF01211	ENSFACAG000000033090	ENSFACAT000000034933	downstream gene variant	MODIFIER
			CSF3	ENSFACAG000000040124	ENSFACAT000000043767	downstream gene variant	MODIFIER
					ENSFACAT000000063974	downstream gene variant	MODIFIER
E1:41147641	A	G	CASC3	ENSFACAG000000014998	ENSFACAT000000015002	missense variant	MODERATE
E1:44190339	G	C	HDAC5	ENSFACAG000000002199	ENSFACAT000000002199	missense variant	MODERATE
					ENSFACAT000000036901	missense variant	MODERATE
E1:44239695	G	C	C17orf53	ENSFACAG000000008021	ENSFACAT000000008023	missense variant & splice region variant	MODERATE
					ENSFACAT000000065449	intron variant	MODIFIER
E1:44726300	G	A	DBF4B	ENSFACAG000000023490	ENSFACAT000000023498	missense variant	MODERATE
					ENSFACAT000000060046	missense variant	MODERATE
E1:45347104	C	A	n/a	ENSFACAG000000046546	ENSFACAT000000053446	missense variant	MODERATE
					ENSFACAT000000061422	intron variant	MODIFIER
E1:45347109	G	A	n/a	ENSFACAG000000046546	ENSFACAT000000053446	missense variant	MODERATE
					ENSFACAT000000061422	intron variant	MODIFIER
E1:45347329	C	T	n/a	ENSFACAG000000046546	ENSFACAT000000053446	missense variant	MODERATE
					ENSFACAT000000061422	intron variant	MODIFIER
E1:61100712	C	T	RNF213	ENSFACAG000000000945	ENSFACAT000000000947	missense variant	MODERATE
					ENSFACAT000000040505	missense variant	MODERATE
E1:61769120	C	T	TEPSIN	ENSFACAG000000007574	ENSFACAT000000007575	missense variant	MODERATE
E2:391338	G	A	ZNF132	ENSFACAG000000034735	ENSFACAT000000037402	missense variant	MODERATE
E2:3164628	C	G	SBK2	ENSFACAG000000018172	ENSFACAT000000019391	missense variant	MODERATE
			n/a	ENSFACAG000000005639	ENSFACAT000000005641	upstream gene variant	MODIFIER
E2:3275072	C	T	TMEM238	ENSFACAG000000042544	ENSFACAT000000051974	missense variant	MODERATE
			TMEM190	ENSFACAG000000044046	ENSFACAT000000058607	downstream gene variant	MODIFIER
			n/a	ENSFACAG000000005637	ENSFACAT000000027091	missense variant	MODERATE
E2:3296678	C	G	COX6B2	ENSFACAG000000045733	ENSFACAT000000054765	upstream gene variant	MODIFIER
					ENSFACAT000000062952	upstream gene variant	MODIFIER
E2:4510569	A	C	n/a	ENSFACAG000000000122	ENSFACAT000000000122	missense variant	MODERATE
E2:7229279	C	T	n/a	ENSFACAG00000002858	ENSFACAT000000000274	missense variant	MODERATE
					ENSFACAT000000002858	missense variant	MODERATE
					ENSFACAT000000013695	missense variant	MODERATE
E2:7279785	T	C	n/a	ENSFACAG0000000046174	ENSFACAT0000000063304	missense variant	MODERATE
E2:10578436	G	A	PTGIR	ENSFACAG000000027533	ENSFACAT000000031423	missense variant	MODERATE
E2:11383116	A	T	PPP1R13L	ENSFACAG000000033518	ENSFACAT000000033821	missense variant	MODERATE
E2:12882208	G	A	LYPD3	ENSFACAG000000025558	ENSFACAT000000022021	missense variant	MODERATE
E2:12980296	C	G	n/a	ENSFACAG000000042962	ENSFACAT000000050319	missense variant	MODERATE
E2:12980375	C	A	n/a	ENSFACAG000000042962	ENSFACAT000000050319	missense variant	MODERATE
E2:12980725	T	C	n/a	ENSFACAG000000042962	ENSFACAT000000050319	missense variant	MODERATE
E2:13013310	T	C	n/a	ENSFACAG000000044759	ENSFACAT000000053023	missense variant	MODERATE
E2:13163878	T	C	GRIK5	ENSFACAG000000012730	ENSFACAT000000012733	missense variant	MODERATE

					ENSF00000055225	missense variant	MODERATE
					ENSF00000057632	missense variant	MODERATE
					ENSF00000062684	missense variant	MODERATE
					ENSF00000063192	missense variant	MODERATE
					ENSF00000065353	missense variant	MODERATE
E2:13328101	C	T	ERF	ENSF00000030101	ENSF00000029651	missense variant	MODERATE
E2:32289680	C	T	n/a	ENSF00000030405	ENSF00000028957	missense variant	MODERATE
			KATNB1	ENSF00000002687	ENSF00000002687	missense variant	MODERATE
E2:36806817	A	G	KIFC3	ENSF00000002689	ENSF000000035824	downstream gene variant	MODIFIER
					ENSF000000037293	downstream gene variant	MODIFIER
					ENSF000000045877	downstream gene variant	MODIFIER
					ENSF000000051211	downstream gene variant	MODIFIER
E3:774328	T	C	DNAAF5	ENSF00000005248	ENSF000000038033	missense variant	MODERATE
E3:797033	C	T	DNAAF5	ENSF00000005248	ENSF000000038033	missense variant	MODERATE
E3:797036	G	A	DNAAF5	ENSF00000005248	ENSF000000038033	missense variant	MODERATE
E3:1683102	C	T	MAD1L1	ENSF00000005389	ENSF00000005390	missense variant	MODERATE
E3:2251003	G	A	IQCE	ENSF000000025676	ENSF000000027142	missense variant	MODERATE
					ENSF000000036337	missense variant	MODERATE
E3:2304718	C	T	BRAT1	ENSF000000031391	ENSF000000032068	missense variant	MODERATE
E3:2312083	G	A	BRAT1	ENSF000000031391	ENSF000000032068	missense variant	MODERATE
					ENSF00000008563	missense variant	MODERATE
E3:2503530	G	A	CARD11	ENSF00000008561	ENSF000000036684	missense variant	MODERATE
					ENSF000000061721	missense variant	MODERATE
E3:5354310	T	C	EIF2AK1	ENSF00000008465	ENSF000000024848	intron variant	MODIFIER
			ANKRD61	ENSF000000027842	ENSF000000033423	missense variant	MODERATE
E3:7130119	G	C	n/a	ENSF000000023135	ENSF000000046678	missense variant	MODERATE
E3:7878324	C	T	ZAN	ENSF00000006851	ENSF00000006853	missense variant	MODERATE
					ENSF000000054763	missense variant	MODERATE
E3:7878456	A	G	ZAN	ENSF00000006851	ENSF00000006853	missense variant	MODERATE
					ENSF000000054763	missense variant	MODERATE
					ENSF00000008710	intron variant	MODIFIER
E3:15165314	G	T	n/a	ENSF00000008708	ENSF000000034285	intron variant	MODIFIER
					ENSF000000053064	upstream gene variant	MODIFIER
					ENSF000000063874	missense variant	MODERATE
E3:20493402	G	A	PRSS53	ENSF00000002602	ENSF00000002602	missense variant	MODERATE
			ZNF668	ENSF00000008954	ENSF00000008956	intron variant	MODIFIER
			ZNF646	ENSF000000021918	ENSF000000027839	downstream gene variant	MODIFIER
F1:17988451	T	C	CENPL	ENSF000000013044	ENSF000000013048	missense variant	MODERATE
					ENSF000000006475	missense variant	MODERATE
F1:20764018	C	T	ASTN1	ENSF00000006473	ENSF000000039511	missense variant	MODERATE
					ENSF000000053463	missense variant	MODERATE
F1:23027052	A	C	SOAT1	ENSF00000007880	ENSF00000007882	intron variant	MODIFIER
			n/a	ENSF000000036745	ENSF000000060726	intron variant	MODIFIER
F1:23028528	G	C	SOAT1	ENSF00000007880	ENSF00000007882	intron variant	MODIFIER
			n/a	ENSF000000036745	ENSF000000060726	intron variant	MODIFIER
					ENSF000000047543	missense variant	MODERATE
F1:28499581	C	T	CACNA1E	ENSF000000015444	ENSF000000015448	missense variant	MODERATE
					ENSF000000032756	missense variant	MODERATE
					ENSF000000047607	missense variant	MODERATE
F1:30078275	C	T	CEP350	ENSF000000014835	ENSF000000014843	missense variant	MODERATE
			IARS2	ENSF000000002597	ENSF000000002597	missense variant	MODERATE
F1:59343562	C	G	BPNT1	ENSF000000026600	ENSF000000045775	upstream gene variant	MODIFIER
F1:60046003	C	G	HLX	ENSF000000034331	ENSF000000039455	missense variant	MODERATE
F1:61342644	C	T	HHIPL2	ENSF000000002257	ENSF000000002257	missense variant	MODERATE
F1:62240122	G	A	ALDH9A1	ENSF000000004312	ENSF000000004313	missense variant	MODERATE
F1:68196401	G	A	CD1B	ENSF000000008648	ENSF000000008650	missense variant	MODERATE
F1:69370149	G	A	BCAN	ENSF00000006557	ENSF000000006559	missense variant	MODERATE
					ENSF000000058402	missense variant	MODERATE
			TMEM79	ENSF000000011787	ENSF000000011789	missense variant	MODERATE
F1:69629361	A	G	GLMP	ENSF000000011788	ENSF000000011790	downstream gene variant	MODIFIER
					ENSF000000058515	downstream gene variant	MODIFIER
			SMG5	ENSF000000011783	ENSF000000050065	upstream gene variant	MODIFIER
F1:69787409	G	A	RAB25	ENSF000000011775	ENSF000000011777	missense variant	MODERATE
			LAMTOR2	ENSF000000028596	ENSF000000023659	downstream gene variant	MODIFIER
					ENSF000000043261	downstream gene variant	MODIFIER
F1:70279561	G	T	RUSC1	ENSF000000024900	ENSF000000026005	missense variant	MODERATE
			RUSC1	ENSF000000024900	ENSF000000055419	upstream gene variant	MODIFIER
			FDP5	ENSF000000039350	ENSF000000034725	downstream gene variant	MODIFIER
F1:70502868	G	A	DCST1	ENSF000000018847	ENSF000000018172	missense variant	MODERATE
					ENSF000000058210	missense variant	MODERATE
			DCST2	ENSF000000040157	ENSF000000053006	synonymous variant	LOW

F1:70503591	G	A	DCST1	ENSFCA00000018847	ENSFCAT00000018172	upstream gene variant	MODIFIER
					ENSFCAT00000058210	upstream gene variant	MODIFIER
F1:70932845	T	C	DCST2	ENSFCA00000040157	ENSFCAT00000053006	missense variant	MODERATE
					ENSFCAT00000028335	missense variant	MODERATE
			IL6R	ENSFCA00000026747	ENSFCAT00000057276	missense variant	MODERATE
F2:81054785	C	T	COL22A1	ENSFCA00000040466	ENSFCAT00000031166	upstream gene variant	MODIFIER
					ENSFCAT00000056098	missense variant & splice region variant	MODERATE
F2:81202027	C	T	COL22A1	ENSFCA00000040466	ENSFCAT00000056098	missense variant	MODERATE
F2:82973708	T	C	SLC45A4	ENSFCA00000010846	ENSFCAT00000010851	missense variant	MODERATE
F2:83794075	C	T	TSNARE1	ENSFCA00000010752	ENSFCAT00000010755	missense variant	MODERATE
					ENSFCAT00000043081	missense variant	MODERATE
X:10709501	C	T	OFD1	ENSFCA00000014870	ENSFCAT00000014879	missense variant	MODERATE
					ENSFCAT00000042140	missense variant	MODERATE
					ENSFCAT00000062574	missense variant	MODERATE
X:10709512	G	T	OFD1	ENSFCA00000014870	ENSFCAT00000014879	missense variant	MODERATE
					ENSFCAT00000042140	missense variant	MODERATE
					ENSFCAT00000062574	missense variant	MODERATE
X:26130933	A	G	NR0B1	ENSFCA00000026851	ENSFCAT00000023728	missense variant	MODERATE
X:39321509	T	C	EFHC2	ENSFCA00000007765	ENSFCAT00000007767	missense variant	MODERATE
X:104748999	G	C	TENM1	ENSFCA00000008355	ENSFCAT00000008357	missense variant	MODERATE
					ENSFCAT00000046755	missense variant	MODERATE
					ENSFCAT00000066641	missense variant	MODERATE

Table 2. A list of variants unique to the proband that were called moderate impact by VEP and the effect in all associated transcripts.