

Table S1. Genomic position/reference for all genotyped single-nucleotide variants (SNVs)

SNV name	Gene name	Chr	Genomic Position	Reference	Type of mutation	Alleles	Genotyping result	MAF case	MAF cont	Exac All MAF	Exac African	GnomAD All	GnomAD African
SPINK5_5	SPINK5	5	14744998	SPINK5:NM_001127698:exon3:c.C194T:p.T6 5M,SPINK5:NM_001127699:exon3:c.C194T:p.T65M,SPINK5:NM_006846:exon3:c.C194T:p.T65M	Missense	C/T	Non-polymorphic	0.062	0.07	0.0002	0		
SPINK5_6	SPINK5	5	147469114	SPINK5:NM_001127698:exon7:c.G532A:p.E178K,SPINK5:NM_006846:exon7:c.G532A:p.E178K	Missense	G/A	OK						
rs2303063	SPINK5	5	147480027	SPINK5:NM_001127698:exon13:c.G1103A:p.S368N	Missense	G/A	OK	0.34	0.4	0.5	0.24	0.5	0.24
rs2303067	SPINK5	5	147480955	SPINK5:NM_001127698:exon14:c.A1258C:p.K420E	Missense	G/A	OK	0.34	0.34	0.5	0.24	0.5	0.24
rs34393923	SPINK5	5	147481363	SPINK5:NM_001127699:exon15:c.G1322A:p.R441H,SPINK5:NM_006846:exon15:c.G1322A:p.R441H	Missense	G/A	Non-polymorphic		0.0022		0.024	0.0023	0.024
rs111662216	SPINK5	5	147496004-06	SPINK5:NM_001127698:exon22:c.2087_2089del:p.696_697del,SPINK5:NM_001127699:exon22:c.2087_2089del:p.696_697del,SPINK5:NM_006846:exon22:c.2087_2089del:p.696_697del	Non-frameshift /GTG deletion	/GTG	Two-cluster	0.003	0.004	0.0021	0.024		
rs3188691	SPINK5	5	147506583	SPINK5:NM_006846:exon30:c.A2905G:p.K969E,SPINK5:NM_01127698:exon31:c.A2995G:p.K99E	Missense	A/G	OK	0.063	0.068	0.003	0.025	0.0032	0.023
rs145120458	CRNM	1	152383382	CRNM:NM_016190:exon3:c.G176A:p.R59H	Missense	G/A	Non-polymorphic		0.002		0.0057	0.002	0.0058
rs35492900	CRNM	1	152382885	CRNM:NM_016190:exon3:c.G673C:p.E225Q	Missense	G/C	Non-polymorphic		0.00079		0.008	0.0008	0.0078
EMSY	c30orf11	11	76256857	C11orf30:NM_020193:exon20:c.T3290C:p.I1097T	Missense	T/C	Two-cluster	0.071	0.054				
rs2229462	TGM1	14	24711015	TGM1:NM_000359:exon3:c.G394A:p.D132N	Missense	G/A	Two-cluster	0.065	0.056	0.00081	0.0089	0.0007	0.0083
rs114998364	TGM3	20	2321094	TGM3:NM_003245:exon13:c.G1949A:p.G650E	Missense	G/A	OK	0.063	0.051	0.0027	0.031	0.0026	0.028
rs80058195	TGM5	15	43525791	TGM5:NM_004245:exon11:c.T1724C:p.V575A,TGM5:NM_201631:exon12:c.T1970C:p.V657A	Missense	T/C	Two-cluster	0.044	0.044	0.0028	0.032	0.0029	0.032
rs140779290	TGM5	15	43545103	TGM5:NM_004245:exon5:c.A470G:p.N157S,TGM5:NM_201631:exon6:c.A716G:p.N239S	Missense	A/G	Two-cluster	0.057	0.061	8.246e-06	0	0.000018	0.00004
rs138507857	TGM5	15	43552428	TGM5:NM_201631:exon3:c.C258G:p.S86R	Missense	C/G	Two-cluster	0.05	0.06	0.00024	0	0.00022	0.00004
rs16961655	DSG1	18	28916495	DSG1:NM_001942:exon9:c.C1184G:p.T395S	Missense	C/G	Non-polymorphic		0.0025		0.027	0.0026	0.027
rs112401974	DSG1	18	28934485	DSG1:NM_001942:exon15:c.C2326T:p.P776S	Missense	C/T	Non-polymorphic		3.295e-05		0.00038	0.000039	0.00045
DSG1_1	DG51	18	28935224	DSG1:NM_001942:exon15:c.A3065T:p.H1022L	Missense	A/G	Non-polymorphic						
DSG1_3	DSG1	18	28935262	DSG1:NM_001942:exon15:c.G3103A:p.A1035T	Missense	G/A	Non-polymorphic						
rs200219673	DSG4	18	28966732	DSG4:NM_001134453:exon3:c.G166A:p.A56T,DSG4:NM_177986:exon3:c.G166A:p.A56T	Missense	G/A	Two-cluster	0.006	0.0025	9.076e-05	0.00028	0.000052	0.00026
rs36040686	DSG4	18	28968349	S79L,DSG4:NM_177986:exon4:c.C236T:p.S79L	Missense	C/T	Two-cluster	0.077	0.065	0.042	0.046	0.0041	0.0033
rs143110911	DSG4	18	28968862	DSG4:NM_001134453:exon5:c.G398A:p.R133Q,DSG4:NM_177986:exon5:c.G398A:p.R133Q	Missense	G/A	Non-polymorphic			9.082e-05	9.718e-05	0.000079	0.00024
rs140750904	DSG4	18	28972253	DSG4:NM_001134453:exon8:c.G955A:p.D319N,DSG4:NM_177986:exon8:c.G955A:p.D319N	Missense	G/A	OK	0.039	0.023	0.0006	0	0.00051	0.00012
rs35378785	DSG4	18	28979427	DSG4:NM_001134453:exon9:c.G1198A:p.G400R,DSG4:NM_177986:exon9:c.G1198A:p.G400R	Missense	G/A	Two-cluster	0.024	0.013	0.0043	0.0046	0.043	0.0033
rs16961975	DSG3	18	29046606	DSG3:NM_001944:exon11:c.G1525A:p.V509M	Missense	G/A	Two-cluster	0.012	0.018	0.027	0.034	0.024	0.032
DSG3_2	DSG3	18	29055892	DSG3:NM_001944:exon16:c.T2669A:p.V890D	Missense	T/A	Two-cluster	0.01	0.018				
HRNR3	HRNR	1	152190027	HRNR:NM_001009931:exon3:c.C4078T:p.P1360S	Missense	G/A	Fail-manufacture						
HRNR2	HRNR	1	152191419	HRNR:NM_001009931:exon3:c.C2686T:p.R896C	Missense	G/A	OK	0.048	0.07				
rs111789738	HRNR	1	152191698	HRNR:NM_001009931:exon3:c.A2407G:p.S803G	Missense	T/C	OK	0.096	0.086	0.0021	0.024	0.0021	0.022
rs77898999	HRNR	1	152191712	HRNR:NM_001009931:exon3:c.C2393A:p.T798K	Missense	G/T	OK	0.023	0.019	0.0014	0.016	0.0014	0.015
HRNR1	HRNR	1	152191822	HRNR:NM_001009931:exon3:c.A2283C:p.Q761H	Missense	T/G	Fail-manufacture						
HRNR4	HRNR	1	152192541	HRNR:NM_001009931:exon3:c.T1564A:p.S522T	Missense	A/T	Non-polymorphic						
rs142030922	HRNR	1	152192605	HRNR:NM_001009931:exon3:c.G1500C:p.Q500H	Missense	C/G	Two-cluster	0.0057	0.007		0	0.00045	0.000041
rs148892565	HRNR	1	152192882	HRNR:NM_001009931:exon3:c.G1223A:p.R408H	Missense	C/T	Fail-manufacture						
rs187389918	HRNR	1	152193177	HRNR:NM_001009931:exon3:c.G928A:p.V310I	Missense	C/T	Non-polymorphic						
rs148822876	HRNR	1	152193390	HRNR:NM_001009931:exon3:c.G715C:p.G239R	Missense	C/G	Two-cluster	0.022	0.034	0.000199681	0.0008	0.00019	0.00024
rs139222671	HRNR	1	152193720	HRNR:NM_001009931:exon3:c.C385T:p.H129Y,HRNR:NM_001009931:exon3:c.C325A:p.Q109K	Missense	G/A	Non-polymorphic			3.296e-05	0.00019	0.00005	0.000041
		1	152193780		Missense	G/T	Two-cluster	0.036	0.023	6.59E-05	0	0.00072	0

Fail-manufacture: single-nucleotide polymorphisms (snps) for which primer/probes failed to be manufactured by the vendor. Snps marked non-polymorphic, indicate snps which were seen to be non-polymorphic in our Ethiopian case-control material. Two-cluster: snps for which no homozygotes were detected for the minor allele. OK: snps for which all 3 genotypes were observed. All snps marked two-cluster and OK were seen to follow H-W.