

# SUPPLEMENTARY INFORMATION

for

## Genome-wide association analysis for chronic venous disease identifies *EFEMP1* and *KCNH8* as susceptibility loci

Eva Ellinghaus, PhD<sup>1,¶</sup>; David Ellinghaus, PhD<sup>1,¶,\*</sup>; Petra Krusche, MD<sup>2</sup>; Aljoscha Greiner, MD<sup>2</sup>; Claudia Schreiber, MD<sup>3</sup>; Susanna Nikolaus, MD<sup>3</sup>; Christian Gieger, PhD<sup>4</sup>; Konstantin Strauch, PhD<sup>4,5</sup>; Wolfgang Lieb, MD<sup>6</sup>; Philip Rosenstiel, MD<sup>1</sup>; Norbert Frings, MD<sup>2</sup>; Andreas Fiebig, PhD<sup>1</sup>; Stefan Schreiber, MD<sup>1,3</sup>; Andre Franke, PhD<sup>1</sup>

- 1 Institute of Clinical Molecular Biology, Christian-Albrechts-University of Kiel, 24105 Kiel, Germany
- 2 Capio Mosel-Eifel-Clinic, 56864 Bad Bertrich, Germany
- 3 Department of General Medicine, University Hospital Schleswig-Holstein, Campus Kiel, 24105 Kiel, Germany
- 4 Institute of Genetic Epidemiology, Helmholtz Zentrum München, German Research Center for Environmental Health, 85764 Neuherberg, Germany
- 5 Institute of Medical Informatics, Biometry and Epidemiology, Chair of Genetic Epidemiology, Ludwig-Maximilians-Universität, 80539 Munich, Germany
- 6 PopGen Biobank, Institute of Epidemiology, University Hospital Schleswig-Holstein, 24105 Kiel, Germany

¶These authors contributed equally to this work

\*To whom correspondence should be addressed:

Dr. David Ellinghaus

Institute of Clinical Molecular Biology

Christian-Albrechts-University of Kiel

University Hospital Schleswig Holstein · Campus Kiel

Rosalind-Franklin-Str. 12 · 24105 Kiel, Germany

Delivery address: Am Botanischen Garten 11, 24118 Kiel, Germany

Phone: +49 431 500 15131

Fax: +49 431 500 15178

eMail: [d.ellinghaus@ikmb.uni-kiel.de](mailto:d.ellinghaus@ikmb.uni-kiel.de)

## Supplementary Tables

### Supplementary Table 1. Case/control panels used in the analysis.

**Panel A:** German discovery panel (GWAS) after quality control. ‘Group’ refers to the scientific group that provided samples.

Country	Group	No. Cases (C2, C3, C4)	No. Controls	Platform
Germany	Capio Mosel-Eifel-Clinic	323	-	Affymetrix 5.0
	CAU Kiel & PopGen	-	1228	Affymetrix 6.0
	KORA S3 (Helmholtz Center Munich)	-	1773	Affymetrix 500k Set
	KORA S4 (Helmholtz Center Munich)	-	1618	Affymetrix 6.0
Total		323	4619	

**Panel B:** Stage one replication in German population after quality control.

Country	Group	No. Cases (C2, C3, C4)	No. Controls	Platform
Germany	Capio Mosel-Eifel-Clinic	1258	-	Sequenom & TaqMan
	CAU Kiel & PopGen	-	1925	

**Panel C:** Stage two replication in German population after quality control.

Country	Group	No. Cases (C2, C3, C4)	No. Controls	Platform
Germany	Capio Mosel-Eifel-Clinic	688	-	TaqMan
	CAU Kiel & PopGen	-	1221	

**Supplementary Table 2. Results of replication stage one genotyping and association testing of 70 SNPs in an independent German panel of 1,258 CVD cases and 1,925 controls (panel B, Supplementary Table 1).**

CHR	BP	SNP	P <sub>GWAS</sub>	A1	A2	AF <sub>cases</sub>	AF <sub>controls</sub>	P	OR	95% CI
1	50,336,554	rs4285748	1.82×10 <sup>-4</sup>	T	C	0.05	0.04	0.46	1.10	0.86-1.39
1	76,441,522	rs17097780	2.37×10 <sup>-7</sup>	T	A	0.09	0.08	0.27	1.11	0.92-1.33
2	53,885,058	rs11682721	1.08×10 <sup>-4</sup>	G	A	0.4	0.42	0.17	0.93	0.84-1.03
2	56,095,994	rs17278665	5.18×10 <sup>-5</sup>	G	C	0.18	0.14	8.47×10 <sup>-5</sup>	1.32	1.15-1.51
2	106,373,169	rs13028719	9.23×10 <sup>-4</sup>	C	T	0.28	0.28	0.48	0.96	0.86-1.08
2	128,309,469	rs12464981	3.05×10 <sup>-4</sup>	G	C	0.41	0.40	0.27	1.06	0.96-1.17
2	142,027,067	rs3856359	3.62×10 <sup>-4</sup>	T	A	0.08	0.07	0.09	1.18	0.98-1.43
2	154,163,944	rs1347927	7.65×10 <sup>-5</sup>	G	A	0.39	0.35	3.29×10 <sup>-3</sup>	1.17	1.05-1.30
2	218,588,976	rs6436007	1.33×10 <sup>-7</sup>	A	G	0.10	0.10	0.42	1.07	0.91-1.27
2	223,658,198	rs11689692	1.04×10 <sup>-4</sup>	A	T	0.13	0.13	0.81	1.02	0.88-1.18
3	19,334,695	rs727139	1.69×10 <sup>-4</sup>	G	A	0.18	0.22	4.54×10 <sup>-5</sup>	0.77	0.67-0.87
3	65,638,580	rs1499496	8.85×10 <sup>-5</sup>	C	T	0.07	0.07	0.72	1.04	0.85-1.27
3	73,379,546	rs12637430	1.13×10 <sup>-4</sup>	C	T	0.23	0.21	0.10	1.11	0.98-1.25
4	30,949,606	rs4692493	8.89×10 <sup>-4</sup>	A	T	0.47	0.46	0.76	1.02	0.92-1.12
4	107,359,768	rs6819528	7.29×10 <sup>-5</sup>	T	G	0.47	0.50	0.05	0.90	0.82-1.00
4	110,499,958	rs13118855	1.18×10 <sup>-4</sup>	G	A	0.10	0.11	0.10	0.87	0.74-1.03
4	110,571,210	rs1029453	2.42×10 <sup>-4</sup>	A	C	0.10	0.12	0.07	0.86	0.73-1.01
5	26,963,586	rs2127426	1.03×10 <sup>-7</sup>	C	G	0.17	0.17	0.45	0.95	0.83-1.09
5	64,495,058	rs1895431	4.90×10 <sup>-4</sup>	T	C	0.32	0.33	0.38	0.95	0.86-1.06
5	91,049,451	rs331385	6.09×10 <sup>-5</sup>	G	A	0.45	0.42	0.01	1.15	1.03-1.27
5	115,800,559	rs10077720	4.94×10 <sup>-5</sup>	A	G	0.10	0.08	0.01	1.26	1.06-1.51
5	127,684,411	rs11241956	1.97×10 <sup>-4</sup>	G	A	0.31	0.34	4.64×10 <sup>-3</sup>	0.86	0.77-0.95
5	171,128,464	rs6555969	3.88×10 <sup>-4</sup>	T	C	0.33	0.33	0.60	1.03	0.92-1.15
6	2,623,930	rs9503233	5.24×10 <sup>-4</sup>	A	G	0.21	0.21	0.45	0.95	0.84-1.08
6	33,020,842	rs3128953	1.20×10 <sup>-12</sup>	A	G	0.24	0.22	0.07	1.12	0.99-1.27
6	55,347,790	rs9396099	8.13×10 <sup>-4</sup>	T	C	0.26	0.23	1.93×10 <sup>-3</sup>	1.20	1.07-1.35
7	24,005,443	rs17148598	3.91×10 <sup>-7</sup>	G	A	0.08	0.08	0.57	0.95	0.78-1.14
7	26,891,016	rs2030136	3.93×10 <sup>-4</sup>	C	T	0.26	0.22	6.98×10 <sup>-5</sup>	1.27	1.13-1.43
7	31,445,598	rs1860793	5.99×10 <sup>-4</sup>	C	T	0.34	0.36	0.04	0.89	0.80-0.99
7	47,445,812	rs834601	3.95×10 <sup>-4</sup>	C	T	0.49	0.50	0.32	0.95	0.86-1.05
7	52,104,548	rs10278047	1.76×10 <sup>-4</sup>	T	C	0.22	0.25	3.03×10 <sup>-3</sup>	0.83	0.74-0.94
7	77,686,838	rs6958027	3.43×10 <sup>-4</sup>	T	G	0.44	0.42	0.30	1.06	0.95-1.17
7	77,868,102	rs10953499	6.86×10 <sup>-5</sup>	A	G	0.17	0.19	0.07	0.89	0.78-1.01
7	83,134,459	rs11982066	4.28×10 <sup>-4</sup>	C	T	0.41	0.43	0.12	0.92	0.83-1.02
7	115,708,814	rs12532028	8.00×10 <sup>-6</sup>	A	C	0.10	0.09	0.58	1.05	0.88-1.24
7	153,638,248	rs6973850	1.59×10 <sup>-7</sup>	T	C	0.12	0.11	0.36	1.08	0.92-1.26
8	59,857,097	rs16924316	1.58×10 <sup>-4</sup>	C	T	0.12	0.13	0.20	0.90	0.77-1.05

8	73,409,704	rs12547029	1.25×10 <sup>-4</sup>	T	C	0.42	0.42	0.91	0.99	0.90-1.10
8	73,419,786	rs1346958	5.39×10 <sup>-5</sup>	A	C	0.38	0.38	0.93	1.01	0.91-1.11
8	114,888,083	rs12334329	3.06×10 <sup>-7</sup>	A	G	0.13	0.13	0.95	1.01	0.87-1.17
8	133,247,788	rs4736558	7.37×10 <sup>-4</sup>	T	A	0.24	0.24	0.98	1.00	0.89-1.12
9	8,117,601	rs4742453	7.97×10 <sup>-4</sup>	A	G	0.12	0.12	0.44	1.06	0.91-1.24
9	22,740,232	rs7872081	5.30×10 <sup>-6</sup>	A	G	0.06	0.05	0.18	1.16	0.94-1.44
9	28,299,438	rs7855717	1.30×10 <sup>-4</sup>	C	T	0.29	0.30	0.55	0.97	0.87-1.08
9	71,198,014	rs17479566	3.11×10 <sup>-5</sup>	T	C	0.20	0.23	3.71×10 <sup>-3</sup>	0.83	0.74-0.94
10	6,677,767	rs10795331	7.79×10 <sup>-5</sup>	T	C	0.46	0.47	0.53	0.97	0.88-1.07
10	95,116,029	rs787629	1.20×10 <sup>-4</sup>	A	C	0.27	0.27	0.78	1.02	0.91-1.14
10	128,526,537	rs10794128	4.66×10 <sup>-5</sup>	C	T	0.35	0.33	0.03	1.13	1.01-1.25
11	63,948,354	rs4980525	5.14×10 <sup>-4</sup>	C	T	0.46	0.47	0.55	0.97	0.88-1.07
11	104,140,745	rs7950244	4.33×10 <sup>-5</sup>	C	A	0.07	0.07	0.24	0.89	0.73-1.08
12	27,031,461	rs7306179	5.20×10 <sup>-4</sup>	A	T	0.38	0.39	0.23	0.94	0.85-1.04
12	101,637,505	rs10778104	1.56×10 <sup>-4</sup>	A	C	0.40	0.37	0.06	1.10	1.00-1.22
12	106,299,118	rs7957583	8.97×10 <sup>-4</sup>	A	T	0.38	0.40	0.16	0.93	0.84-1.03
12	114,713,898	rs7962292	9.98×10 <sup>-4</sup>	T	C	0.35	0.35	0.82	1.01	0.91-1.13
13	19,875,086	rs547308	1.40×10 <sup>-4</sup>	C	T	0.41	0.41	0.94	1.00	0.90-1.10
13	24,208,829	rs7987909	6.30×10 <sup>-4</sup>	G	A	0.11	0.11	0.85	0.98	0.84-1.16
15	30,137,080	rs1403253	1.03×10 <sup>-4</sup>	G	A	0.15	0.13	0.13	1.12	0.97-1.29
15	70,817,223	rs8033107	3.11×10 <sup>-4</sup>	T	C	0.23	0.21	0.21	1.08	0.96-1.22
17	5,391,690	rs2309399	9.81×10 <sup>-4</sup>	A	G	0.43	0.44	0.45	0.96	0.87-1.07
17	68,256,295	rs236562	3.02×10 <sup>-5</sup>	G	A	0.41	0.42	0.39	0.96	0.86-1.06
18	3,656,488	rs1791398	8.30×10 <sup>-4</sup>	G	A	0.21	0.21	0.43	0.95	0.84-1.08
18	5,620,114	rs12455403	6.81×10 <sup>-6</sup>	C	T	0.18	0.18	0.55	0.96	0.84-1.10
18	28,619,690	rs276931	9.36×10 <sup>-4</sup>	G	A	0.31	0.33	0.15	0.92	0.83-1.03
19	3,709,018	rs570896	6.17×10 <sup>-4</sup>	G	C	0.31	0.31	0.92	1.01	0.90-1.12
19	16,337,588	rs10469462	1.04×10 <sup>-4</sup>	C	T	0.17	0.20	0.02	0.86	0.75-0.98
19	16,343,847	rs12984337	6.79×10 <sup>-5</sup>	T	A	0.28	0.28	0.92	0.99	0.89-1.11
19	16,368,528	rs4808478	2.98×10 <sup>-5</sup>	A	C	0.26	0.28	0.08	0.90	0.81-1.01
20	2,100,348	rs6112914	9.70×10 <sup>-4</sup>	A	G	0.20	0.20	0.48	0.96	0.84-1.08
20	61,018,265	rs6061531	4.34×10 <sup>-7</sup>	C	T	0.14	0.14	0.59	1.04	0.90-1.20
21	39,284,835	rs2836050	2.18×10 <sup>-5</sup>	T	C	0.20	0.20	0.73	1.02	0.90-1.16

**CHR:** chromosome; **BP:** position (NCBI build 37/hg19 coordinates) of SNP marker; **SNP:** SNP rs-id (if available from dbSNP130); **P<sub>GWAS</sub>:** *P*-value in the initial genome-wide phase; **A1:** minor allele; **A2:** major allele; **AF<sub>cases</sub>:** allele frequency of minor allele A1 in cases; **AF<sub>controls</sub>:** allele frequency of minor allele A1 in controls; **P:** *P*-value; **OR:** corresponding odds ratio and **95% CI:** 95% confidence interval with respect to minor allele.

**Supplementary Table 3. Results of replication stage two genotyping and association testing of 3 SNPs in an independent German panel of 688 CVD cases and 1,221 controls (panel C, Supplementary Table 1).**

CHR	BP	SNP	P <sub>GWAS</sub>	A1	A2	AF <sub>cases</sub>	AF <sub>controls</sub>	P	OR	95% CI
2	56,095,994	rs17278665	5.18×10 <sup>-5</sup>	G	C	0.17	0.15	0.07	1.18	0.99-1.41
3	19,334,695	rs727139	1.69×10 <sup>-4</sup>	G	A	0.16	0.21	1.53×10 <sup>-4</sup>	0.72	0.60-0.85
7	26,891,016	rs2030136	3.93×10 <sup>-4</sup>	C	T	0.26	0.24	0.17	1.11	0.95-1.30

**CHR:** chromosome; **BP:** position (NCBI build 37/hg19 coordinates) of SNP marker; **SNP:** SNP rs-id (if available from dbSNP130); **P<sub>GWAS</sub>:** *P*-value in the initial genome-wide phase; **A1:** minor allele; **A2:** major allele; **AF<sub>cases</sub>:** allele frequency of minor allele A1 in cases; **AF<sub>controls</sub>:** allele frequency of minor allele A1 in controls; **P:** *P*-value; **OR:** corresponding odds ratio and **95% CI:** 95% confidence interval with respect to minor allele.

**Supplementary Table 4. Results of association testing of 3 SNPs in panel B and C with adjustments made for sex, age at ascertainment and body mass index (BMI) if available.** No notably different results were obtained in the sex-, age- and BMI-adjusted analyses.

CHR	BP	SNP	A1	A2	P	panel B				panel B & C				
						no adjustment		BMI* adjusted	P	no adjustment		sex & age adjusted		
						OR	95% CI	P	OR	P	OR	95% CI	P	OR
2	56,095,994	rs17278665	G	C	8.47×10 <sup>-5</sup>	1.32	1.15-1.51	7.81×10 <sup>-5</sup>	1.32	2.59×10 <sup>-5</sup>	1.26	1.13-1.41	4.54×10 <sup>-6</sup>	1.32
3	19,334,695	rs727139	G	A	4.54×10 <sup>-5</sup>	0.77	0.67-0.87	6.33×10 <sup>-5</sup>	0.77	3.39×10 <sup>-8</sup>	0.75	0.68-0.83	4.99×10 <sup>-8</sup>	0.74
7	26,891,016	rs2030136	C	T	6.98×10 <sup>-5</sup>	1.27	1.13-1.43	2.18×10 <sup>-4</sup>	1.25	6.84×10 <sup>-5</sup>	1.21	1.10-1.33	2.53×10 <sup>-5</sup>	1.24

**CHR:** chromosome; **BP:** position (NCBI build 37/hg19 coordinates) of SNP marker; **SNP:** SNP rs-id (if available from dbSNP130); **A1:** minor allele; **A2:** major allele; **P/OR:** *P*-value, corresponding odds ratio and **95% CI:** 95% confidence interval with respect to minor allele.

\*BMI was available for the cases of panel B and C, but only for the controls of panel B.

**Supplementary Table 5. Functional annotation of three newly identified disease associations and all variants in high LD ( $r^2 > 0.8$ ) with these variants.** All variants were annotated using CADD<sup>1</sup> v1.3 (GRCh37/hg19;), fathmm<sup>2</sup> v2.3, MutationTaster<sup>3</sup> (NCBI build 37, Ensemble 69) and HaploReg<sup>4</sup> v4.1 to determine position annotations, including the closest gene, the effect on SNPs on splice sites and functional consequences (**Methods**).











**SNP:** SNP rs ID; **CHR:** Chromosome; **BP:** position (NCBI build 37/hg19 coordinates) of SNP marker; **Ref:** reference allele; **Anc:** ancestral allele; **Alt:** observed allele; **Type:** event type (“SNV”: single nucleotide variant, ‘DEL’: deletion, ‘INS’: insertion); **AnnoType:** ‘CodingTranscript’, ‘Intergenic’, ‘MotifFeature’, ‘NonCodingTranscript’, ‘RegulatoryFeature’, ‘Transcript’; **Consequence:** ‘3PRIME\_UTR’, ‘5PRIME\_UTR’, ‘DOWNSTREAM’, ‘INTERGENIC’, ‘INTRONIC’, ‘NON\_SYNONYMOUS’, ‘SYNONYMOUS’, ‘REGULATORY’, ‘STOP\_GAINED’, ‘STOP\_LOST’, ‘SPLICE\_SITE’, ‘CANONCAL\_SPLICE’, ‘UPSTREAM’, ‘NONCODING\_CHANGE’; **GC:** percent GC in a window of +/- 75 bp; **CpG:** percent CpG in a window of +/-75 bp; **prPhCons:** primate PhastCons conservation score (excl. human); **mamPhCons:** mammalian PhastCons conservation score (excl. human); **verPhCons:** vertebrate PhastCons conservation score (excl. human); **priPhyloP:** primate PhyloP score (excl. human); **mamPhyloP:** mammalian PhyloP score (excl. human); **verPhyloP:** vertebrate PhyloP score (excl. human); **GerpN:** neutral evolution score defined by GERP++ (GERP: Genomic Evolutionary Rate Profiling); **GerpS:** rejected substitution score defined by GERP++; **GerpRS:** Gerp element score; **GerpRSpval:** Gerp element *P*-value ; **GeneName:** name of gene provided in ENSEMBL annotation; **Intron:** intron number/total number of introns; **Grantham:** Grantham score; **PolyPhenVal:** PolyPhen score; **SIFTval:** SIFT score; **RawScore:** raw CADD score. Raw values have relative meaning, with higher values indicating that a variant is more likely not to be observed and therefore more likely to have deleterious effects; **PHRED:** PHRED-like ( $-10 \times \log_{10}(\text{rank}/\text{total})$ ) scaled C-score. A scaled C-score of  $\geq 10$  indicates a prediction of the 10% most deleterious substitutions that you can do to the human genome, a score of  $\geq 20$  indicates the 1% most deleterious; **Non-Coding score:** predictions are given as *P*-values [0-1], values  $> 0.5$  are predicted to be deleterious, values  $< 0.5$  are predicted to be neutral/benign; **Non-Coding Groups:** non-coding predictor (for positions in intergenic regions, introns or non-coding genes) using a subset of 4 of these feature groups: A (46-Way Sequence Conservation based on multiple sequence alignment scores, at the nucleotide level, of 46 vertebrate genomes compared to the human genome), B (Histone Modifications based on ChIP-Seq peak calls for histone modifications), C (Transcription Factor Binding Sites (TFBS PeakSeq) based on PeakSeq peak calls for various transcription factors), D (Open Chromatin based on DNase-Seq peak calls); **pred\_index:** ‘disease causing’: probably deleterious, ‘disease causing automatic’: known to be deleterious, ‘polymorphism’: probably harmless, ‘polymorphism automatic’: known to be harmless; **probability:** probability of the prediction, i.e. a value close to 1 indicates a high security of the prediction.; **features:** summary of

affected features; **splice site - effect**: donor/acceptor splice site increased/decreased/gained/lost; **splice site - gDNA position**: gDNA position of splice site; **splice site - score**: confidence score for wild-type (wt) and/or mutated (mu) splice site as generated by NNSplice<sup>5</sup>; **Sequence constraint by GERP**: ‘yes’ indicates that the region is called as constrained (vs. non-constrained) by the mammalian conservation algorithm GERP; **Sequence constraint by SiPhy**: ‘yes’ indicates that the region is called as constrained (vs. non-constrained) by the mammalian conservation algorithm SiPhy-omega; **NHGRI/EBI GWAS hits**: GWAS results (PMID, phenotype, *P*-value) obtained from the NHGRI catalog (accessed October 31, 2015); **RefSeq genes**: RefSeq genes from the UCSC Genome Browser; **dbSNP func annot**: annotations of functional consequences from dbSNP.

**Supplementary Table 6. List of known eQTLs for the three newly identified disease associations and all variants in high LD ( $r^2 > 0.8$ ) with these variants.** All variants were annotated using HaploReg<sup>4</sup> v4.1 to identify SNPs with an effect on gene expression (see **Methods**).

			HaploReg		
SNP	CHR	BP	eQTL resource id*	Selected eQTL hits	P-val
rs17278665	2	56,095,994	GTEEx2015_v6	Skin Sun Exposed Lower leg, EFEMP1	2.25×10 <sup>-6</sup>
			Hao2012	Lung, EFEMP1	3.45×10 <sup>-7</sup>
rs11899888	2	56,102,744	.	.	.
rs13064845	3	19,312,697	.	.	.
rs727139	3	19,334,695	.	.	.
rs212838	7	26,698,194	GTEEx2015_v6	Thyroid, HOXA5	2.26×10 <sup>-5</sup>
			GTEEx2015_v6	Whole Blood, SKAP2	4.26×10 <sup>-7</sup>
			Lappalainen2013	Lymphoblastoid EUR exonlevel, ENSG00000005020.8	8.82×10 <sup>-7</sup>
			Westra2013	Whole Blood, SKAP2	2.23×10 <sup>-122</sup>
rs3757654	7	26,711,104	GTEEx2015_v6	Thyroid, HOXA5	1.20×10 <sup>-5</sup>
			GTEEx2015_v6	Whole Blood, SKAP2	3.47×10 <sup>-7</sup>
			Lappalainen2013	Lymphoblastoid EUR exonlevel, ENSG00000005020.8	3.45×10 <sup>-6</sup>
			Westra2013	Whole Blood, SKAP2	4.57×10 <sup>-119</sup>
rs212848	7	26,722,155	GTEEx2015_v6	Thyroid, HOXA5	1.28×10 <sup>-5</sup>
			GTEEx2015_v6	Whole Blood, SKAP2	3.41×10 <sup>-7</sup>
			Lappalainen2013	Lymphoblastoid EUR exonlevel, ENSG00000005020.8	1.44×10 <sup>-6</sup>
rs212850	7	26,724,279	GTEEx2015_v6	Thyroid, HOXA5	1.38×10 <sup>-5</sup>
			GTEEx2015_v6	Whole Blood, SKAP2	3.80×10 <sup>-7</sup>
			Lappalainen2013	Lymphoblastoid EUR exonlevel, ENSG00000005020.8	1.37×10 <sup>-6</sup>
			Westra2013	Whole Blood, SKAP2	9.41×10 <sup>-123</sup>
rs212856	7	26,729,205	GTEEx2015_v6	Thyroid, HOXA5	1.41×10 <sup>-5</sup>
			GTEEx2015_v6	Whole Blood, SKAP2	3.89×10 <sup>-7</sup>
			Lappalainen2013	Lymphoblastoid EUR exonlevel, ENSG00000005020.8	1.34×10 <sup>-6</sup>
rs212857	7	26,730,200	GTEEx2015_v6	Thyroid, HOXA5	1.23×10 <sup>-5</sup>
			GTEEx2015_v6	Whole Blood, SKAP2	7.76×10 <sup>-7</sup>
			Lappalainen2013	Lymphoblastoid EUR exonlevel, ENSG00000005020.8	1.38×10 <sup>-6</sup>
			Westra2013	Whole Blood, SKAP2	2.28×10 <sup>-122</sup>
rs4719877	7	26,736,220	GTEEx2015_v6	Thyroid, HOXA5	1.31×10 <sup>-5</sup>
			GTEEx2015_v6	Whole Blood, SKAP2	3.44×10 <sup>-7</sup>
			Lappalainen2013	Lymphoblastoid EUR exonlevel, ENSG00000005020.8	1.16×10 <sup>-6</sup>
			Westra2013	Whole Blood, SKAP2	1.87×10 <sup>-122</sup>
rs6461962	7	26,746,304	GTEEx2015_v6	Thyroid, HOXA5	1.41×10 <sup>-5</sup>
			GTEEx2015_v6	Whole Blood, SKAP2	6.87×10 <sup>-7</sup>
			Lappalainen2013	Lymphoblastoid EUR exonlevel, ENSG00000005020.8	1.89×10 <sup>-6</sup>
			Westra2013	Whole Blood, SKAP2	6.70×10 <sup>-123</sup>
rs6972378	7	26,753,074	GTEEx2015_v6	Thyroid, HOXA5	1.38×10 <sup>-5</sup>
			GTEEx2015_v6	Whole Blood, SKAP2	5.77×10 <sup>-7</sup>
			Lappalainen2013	Lymphoblastoid EUR exonlevel, ENSG00000005020.8	1.75×10 <sup>-6</sup>
			Westra2013	Whole Blood, SKAP2	2.32×10 <sup>-123</sup>
rs6951065	7	26,756,162	GTEEx2015_v6	Thyroid, HOXA5	1.27×10 <sup>-5</sup>
			GTEEx2015_v6	Whole Blood, SKAP2	3.51×10 <sup>-7</sup>
			Lappalainen2013	Lymphoblastoid EUR exonlevel, ENSG00000005020.8	1.80×10 <sup>-6</sup>
			Westra2013	Whole Blood, SKAP2	6.91×10 <sup>-124</sup>
rs6964817	7	26,756,424	GTEEx2015_v6	Thyroid, HOXA5	1.27×10 <sup>-5</sup>
			GTEEx2015_v6	Whole Blood, SKAP2	3.49×10 <sup>-7</sup>
			Lappalainen2013	Lymphoblastoid EUR exonlevel, ENSG00000005020.8	1.67×10 <sup>-6</sup>
			Westra2013	Whole Blood, SKAP2	6.91×10 <sup>-124</sup>
rs2106905	7	26,766,851	GTEEx2015_v6	Thyroid, HOXA5	1.22×10 <sup>-5</sup>
			GTEEx2015_v6	Whole Blood, SKAP2	2.44×10 <sup>-7</sup>
			Lappalainen2013	Lymphoblastoid EUR exonlevel, ENSG00000005020.8	1.41×10 <sup>-6</sup>
			Westra2013	Whole Blood, SKAP2	1.82×10 <sup>-120</sup>
rs6461963	7	26,772,491	GTEEx2015_v6	Thyroid, HOXA5	1.22×10 <sup>-5</sup>
			GTEEx2015_v6	Whole Blood, SKAP2	2.43×10 <sup>-7</sup>
			Lappalainen2013	Lymphoblastoid EUR exonlevel, ENSG00000005020.8	1.77×10 <sup>-6</sup>
			Westra2013	Whole Blood, SKAP2	2.30×10 <sup>-126</sup>

rs4722630	7	26,773,364	GTEEx2015_v6	Thyroid, HOXA5	1.22×10 <sup>-5</sup>
			GTEEx2015_v6	Whole Blood, SKAP2	2.43×10 <sup>-7</sup>
			Lappalainen2013	Lymphoblastoid EUR exonlevel, ENSG00000005020.8	2.19×10 <sup>-6</sup>
			Westra2013	Whole Blood, SKAP2	3.60×10 <sup>-126</sup>
rs6971397	7	26,782,564	GTEEx2015_v6	Thyroid, HOXA5	1.18×10 <sup>-5</sup>
			GTEEx2015_v6	Whole Blood, SKAP2	1.41×10 <sup>-7</sup>
			Lappalainen2013	Lymphoblastoid EUR exonlevel, ENSG00000005020.8	1.01×10 <sup>-6</sup>
			Westra2013	Whole Blood, SKAP2	3.60×10 <sup>-126</sup>
rs727450	7	26,787,642	GTEEx2015_v6	Thyroid, HOXA5	9.08×10 <sup>-6</sup>
			GTEEx2015_v6	Whole Blood, SKAP2	3.25×10 <sup>-7</sup>
			Lappalainen2013	Lymphoblastoid EUR exonlevel, ENSG00000005020.8	1.50×10 <sup>-6</sup>
			Westra2013	Whole Blood, SKAP2	5.16×10 <sup>-126</sup>
rs10281261	7	26,789,472	GTEEx2015_v6	Thyroid, HOXA5	1.11×10 <sup>-5</sup>
			GTEEx2015_v6	Whole Blood, SKAP2	1.37×10 <sup>-7</sup>
			Lappalainen2013	Lymphoblastoid EUR exonlevel, ENSG00000005020.8	1.45×10 <sup>-6</sup>
			Westra2013	Whole Blood, SKAP2	5.16×10 <sup>-126</sup>
rs7457076	7	26,793,215	GTEEx2015_v6	Thyroid, HOXA5	9.39×10 <sup>-6</sup>
			GTEEx2015_v6	Whole Blood, SKAP2	1.27×10 <sup>-7</sup>
			Lappalainen2013	Lymphoblastoid EUR exonlevel, ENSG00000005020.8	1.32×10 <sup>-6</sup>
			Westra2013	Whole Blood, SKAP2	6.55×10 <sup>-126</sup>
rs2189017	7	26,795,073	GTEEx2015_v6	Thyroid, HOXA5	8.16×10 <sup>-6</sup>
			GTEEx2015_v6	Whole Blood, SKAP2	1.23×10 <sup>-7</sup>
			Lappalainen2013	Lymphoblastoid EUR exonlevel, ENSG00000005020.8	1.37×10 <sup>-6</sup>
			Westra2013	Whole Blood, SKAP2	6.07×10 <sup>-126</sup>
rs2189019	7	26,798,581	GTEEx2015_v6	Thyroid, HOXA5	7.90×10 <sup>-6</sup>
			GTEEx2015_v6	Whole Blood, SKAP2	1.23×10 <sup>-7</sup>
			Lappalainen2013	Lymphoblastoid EUR exonlevel, ENSG00000005020.8	1.60×10 <sup>-6</sup>
			Westra2013	Whole Blood, SKAP2	6.07×10 <sup>-126</sup>
rs73061894	7	26,802,085	GTEEx2015_v6	Thyroid, HOXA5	7.48×10 <sup>-6</sup>
			GTEEx2015_v6	Whole Blood, SKAP2	1.21×10 <sup>-7</sup>
			Lappalainen2013	Lymphoblastoid EUR exonlevel, ENSG00000005020.8	1.04×10 <sup>-6</sup>
rs117217399	7	26,805,212	Lappalainen2013	Lymphoblastoid EUR exonlevel, ENSG00000005020.8	1.34×10 <sup>-6</sup>
rs10486478	7	26,819,465	GTEEx2015_v6	Thyroid, HOXA5	1.62×10 <sup>-6</sup>
			GTEEx2015_v6	Whole Blood, SKAP2	3.69×10 <sup>-7</sup>
			Lappalainen2013	Lymphoblastoid EUR exonlevel, ENSG00000005020.8	1.61×10 <sup>-7</sup>
			Westra2013	Whole Blood, SKAP2	3.98×10 <sup>-149</sup>
rs3801850	7	26,820,577	GTEEx2015_v6	Thyroid, HOXA5	3.64×10 <sup>-7</sup>
			GTEEx2015_v6	Whole Blood, SKAP2	6.89×10 <sup>-6</sup>
			Lappalainen2013	Lymphoblastoid EUR exonlevel, ENSG00000005020.8	2.93×10 <sup>-8</sup>
			Westra2013	Whole Blood, SKAP2	2.36×10 <sup>-149</sup>
rs12536560	7	26,822,876	GTEEx2015_v6	Thyroid, HOXA5	3.64×10 <sup>-7</sup>
			GTEEx2015_v6	Whole Blood, SKAP2	7.14×10 <sup>-6</sup>
			Lappalainen2013	Lymphoblastoid EUR exonlevel, ENSG00000005020.8	3.78×10 <sup>-8</sup>
			Westra2013	Whole Blood, SKAP2	1.91×10 <sup>-149</sup>
rs73065504	7	26,825,984	GTEEx2015_v6	Thyroid, HOXA5	9.16×10 <sup>-7</sup>
			GTEEx2015_v6	Whole Blood, SKAP2	1.19×10 <sup>-7</sup>
			Lappalainen2013	Lymphoblastoid EUR exonlevel, ENSG00000005020.8	3.73×10 <sup>-8</sup>
rs10267950	7	26,826,757	GTEEx2015_v6	Thyroid, HOXA5	3.64×10 <sup>-7</sup>
			GTEEx2015_v6	Whole Blood, SKAP2	1.04×10 <sup>-5</sup>
			Lappalainen2013	Lymphoblastoid EUR exonlevel, ENSG00000005020.8	3.48×10 <sup>-8</sup>
			Westra2013	Whole Blood, SKAP2	1.51×10 <sup>-149</sup>
rs10272303	7	26,827,815	GTEEx2015_v6	Thyroid, HOXA5	3.64×10 <sup>-7</sup>
			GTEEx2015_v6	Whole Blood, SKAP2	1.04×10 <sup>-5</sup>
			Lappalainen2013	Lymphoblastoid EUR exonlevel, ENSG00000005020.8	3.10×10 <sup>-8</sup>
rs3801846	7	26,828,653	GTEEx2015_v6	Thyroid, HOXA5	3.92×10 <sup>-7</sup>
			Lappalainen2013	Lymphoblastoid EUR exonlevel, ENSG00000005020.8	3.44×10 <sup>-8</sup>
			Westra2013	Whole Blood, SKAP2	1.11×10 <sup>-149</sup>



rs1327988	7	26,828,965	GTEX2015_v6	Thyroid, HOXA5	3.64×10 <sup>-7</sup>
			Lappalainen2013	Lymphoblastoid EUR exonlevel, ENSG00000005020.8	3.18×10 <sup>-8</sup>
			Westra2013	Whole Blood, SKAP2	8.95×10 <sup>-150</sup>
rs3801844	7	26,829,759	GTEX2015_v6	Thyroid, HOXA5	3.64×10 <sup>-7</sup>
			Lappalainen2013	Lymphoblastoid EUR exonlevel, ENSG00000005020.8	3.16×10 <sup>-8</sup>
			Westra2013	Whole Blood, SKAP2	8.95×10 <sup>-150</sup>
rs3801843	7	26,830,201	GTEX2015_v6	Thyroid, HOXA5	3.64×10 <sup>-7</sup>
			Lappalainen2013	Lymphoblastoid EUR exonlevel, ENSG00000005020.8	3.34×10 <sup>-8</sup>
			Westra2013	Whole Blood, SKAP2	8.95×10 <sup>-150</sup>
rs3801842	7	26,830,263	GTEX2015_v6	Thyroid, HOXA5	3.64×10 <sup>-7</sup>
			Lappalainen2013	Lymphoblastoid EUR exonlevel, ENSG00000005020.8	3.07×10 <sup>-8</sup>
			Westra2013	Whole Blood, SKAP2	8.95×10 <sup>-150</sup>
rs73065517	7	26,830,555	GTEX2015_v6	Thyroid, HOXA5	3.64×10 <sup>-7</sup>
			Lappalainen2013	Lymphoblastoid EUR exonlevel, ENSG00000005020.8	4.16×10 <sup>-8</sup>
rs28733208	7	26,830,825	GTEX2015_v6	Thyroid, HOXA5	3.64×10 <sup>-7</sup>
			Lappalainen2013	Lymphoblastoid EUR exonlevel, ENSG00000005020.8	3.98×10 <sup>-8</sup>
rs73065521	7	26,831,409	GTEX2015_v6	Thyroid, HOXA5	3.64×10 <sup>-7</sup>
			Lappalainen2013	Lymphoblastoid EUR exonlevel, ENSG00000005020.8	3.14×10 <sup>-8</sup>
rs3801840	7	26,832,986	GTEX2015_v6	Thyroid, HOXA5	3.64×10 <sup>-7</sup>
			GTEX2015_v6	Whole Blood, SKAP2	1.04×10 <sup>-5</sup>
			Lappalainen2013	Lymphoblastoid EUR exonlevel, ENSG00000005020.8	3.76×10 <sup>-8</sup>
			Westra2013	Whole Blood, SKAP2	1.41×10 <sup>-150</sup>
rs1327989	7	26,833,563	GTEX2015_v6	Thyroid, HOXA5	3.52×10 <sup>-7</sup>
			Lappalainen2013	Lymphoblastoid EUR exonlevel, ENSG00000005020.8	3.33×10 <sup>-8</sup>
			Westra2013	Whole Blood, SKAP2	1.14×10 <sup>-150</sup>
rs6964094	7	26,834,380	GTEX2015_v6	Thyroid, HOXA5	4.10×10 <sup>-7</sup>
			GTEX2015_v6	Whole Blood, SKAP2	1.17×10 <sup>-6</sup>
			Lappalainen2013	Lymphoblastoid EUR exonlevel, ENSG00000005020.8	3.18×10 <sup>-8</sup>
			Westra2013	Whole Blood, SKAP2	5.02×10 <sup>-147</sup>
rs6956088	7	26,835,727	GTEX2015_v6	Thyroid, HOXA5	3.07×10 <sup>-7</sup>
			Lappalainen2013	Lymphoblastoid EUR exonlevel, ENSG00000005020.8	4.27×10 <sup>-8</sup>
rs10276471	7	26,837,940	GTEX2015_v6	Thyroid, HOXA5	2.56×10 <sup>-7</sup>
			Lappalainen2013	Lymphoblastoid EUR exonlevel, ENSG00000005020.8	1.58×10 <sup>-8</sup>
rs4722641	7	26,838,677	GTEX2015_v6	Thyroid, HOXA5	1.32×10 <sup>-6</sup>
			GTEX2015_v6	Whole Blood, SKAP2	7.60×10 <sup>-6</sup>
			Lappalainen2013	Lymphoblastoid EUR exonlevel, ENSG00000005020.8	1.35×10 <sup>-8</sup>
rs10225924	7	26,839,954	GTEX2015_v6	Thyroid, HOXA5	2.19×10 <sup>-7</sup>
			GTEX2015_v6	Whole Blood, SKAP2	1.03×10 <sup>-5</sup>
			Lappalainen2013	Lymphoblastoid EUR exonlevel, ENSG00000005020.8	1.50×10 <sup>-8</sup>
rs10229837	7	26,840,652	GTEX2015_v6	Thyroid, HOXA5	5.88×10 <sup>-7</sup>
			GTEX2015_v6	Whole Blood, SKAP2	9.85×10 <sup>-6</sup>
			Lappalainen2013	Lymphoblastoid EUR exonlevel, ENSG00000005020.8	1.39×10 <sup>-8</sup>
rs12532395	7	26,841,702	GTEX2015_v6	Thyroid, HOXA5	1.92×10 <sup>-7</sup>
			GTEX2015_v6	Whole Blood, SKAP2	1.03×10 <sup>-5</sup>
			Lappalainen2013	Lymphoblastoid EUR exonlevel, ENSG00000005020.8	1.32×10 <sup>-8</sup>
rs12539719	7	26,844,852	GTEX2015_v6	Thyroid, HOXA5	3.77×10 <sup>-7</sup>
			GTEX2015_v6	Whole Blood, SKAP2	9.70×10 <sup>-8</sup>
			Lappalainen2013	Lymphoblastoid EUR exonlevel, ENSG00000005020.8	4.02×10 <sup>-8</sup>
rs4722644	7	26,845,386	GTEX2015_v6	Thyroid, HOXA5, 4.66927614117146e-07	7.47×10 <sup>-6</sup>
			GTEX2015_v6	Whole Blood, SKAP2	7.47×10 <sup>-6</sup>
			Lappalainen2013	Lymphoblastoid EUR exonlevel, ENSG00000005020.8	1.56×10 <sup>-8</sup>
rs12333525	7	26,847,762	GTEX2015_v6	Thyroid, HOXA5	4.67×10 <sup>-7</sup>
			Lappalainen2013	Lymphoblastoid EUR exonlevel, ENSG00000005020.8	1.07×10 <sup>-8</sup>
rs10244986	7	26,849,558	GTEX2015_v6	Thyroid, HOXA5	4.67×10 <sup>-7</sup>
			GTEX2015_v6	Whole Blood, SKAP2	7.58×10 <sup>-6</sup>
			Lappalainen2013	Lymphoblastoid EUR exonlevel, ENSG00000005020.8	1.49×10 <sup>-8</sup>

rs73067435	7	26,850,195	GTEEx2015_v6	Thyroid, HOXA5	4.67×10 <sup>-7</sup>
			GTEEx2015_v6	Whole Blood, SKAP2	7.45×10 <sup>-6</sup>
			Lappalainen2013	Lymphoblastoid EUR exonlevel, ENSG00000005020.8	1.49×10 <sup>-8</sup>
rs73067437	7	26,850,313	GTEEx2015_v6	Thyroid, HOXA5	3.26×10 <sup>-7</sup>
			GTEEx2015_v6	Whole Blood, SKAP2	1.18×10 <sup>-7</sup>
			Lappalainen2013	Lymphoblastoid EUR exonlevel, ENSG00000005020.8	1.50×10 <sup>-8</sup>
rs57135974	7	26,850,699	GTEEx2015_v6	Thyroid, HOXA5	4.67×10 <sup>-7</sup>
			GTEEx2015_v6	Whole Blood, SKAP2	7.35×10 <sup>-6</sup>
			Lappalainen2013	Lymphoblastoid EUR exonlevel, ENSG00000005020.8	1.46×10 <sup>-8</sup>
rs58760954	7	26,850,776	GTEEx2015_v6	Thyroid, HOXA5	4.67×10 <sup>-7</sup>
			GTEEx2015_v6	Whole Blood, SKAP2	7.33×10 <sup>-6</sup>
			Lappalainen2013	Lymphoblastoid EUR exonlevel, ENSG00000005020.8	1.26×10 <sup>-8</sup>
rs73067443	7	26,851,074	GTEEx2015_v6	Thyroid, HOXA5	4.67×10 <sup>-7</sup>
			GTEEx2015_v6	Whole Blood, SKAP2	7.27×10 <sup>-6</sup>
			Lappalainen2013	Lymphoblastoid EUR exonlevel, ENSG00000005020.8	1.47×10 <sup>-8</sup>
rs3801837	7	26,851,361	GTEEx2015_v6	Thyroid, HOXA5	4.67×10 <sup>-7</sup>
			GTEEx2015_v6	Whole Blood, SKAP2	7.23×10 <sup>-6</sup>
			Lappalainen2013	Lymphoblastoid EUR exonlevel, ENSG00000005020.8	1.21×10 <sup>-8</sup>
rs3801836	7	26,851,400	GTEEx2015_v6	Thyroid, HOXA5	5.37×10 <sup>-7</sup>
			GTEEx2015_v6	Whole Blood, SKAP2	3.01×10 <sup>-6</sup>
			Lappalainen2013	Lymphoblastoid EUR exonlevel, ENSG00000005020.8	1.27×10 <sup>-8</sup>
rs3801834	7	26,852,062	GTEEx2015_v6	Thyroid, HOXA5	4.77×10 <sup>-7</sup>
			Lappalainen2013	Lymphoblastoid EUR exonlevel, ENSG00000005020.8	1.22×10 <sup>-8</sup>
rs3801833	7	26,852,212	GTEEx2015_v6	Thyroid, HOXA5	4.67×10 <sup>-7</sup>
			GTEEx2015_v6	Whole Blood, SKAP2	7.17×10 <sup>-6</sup>
			Lappalainen2013	Lymphoblastoid EUR exonlevel, ENSG00000005020.8	1.58×10 <sup>-8</sup>
rs3801832	7	26,852,280	GTEEx2015_v6	Thyroid, HOXA5	4.67×10 <sup>-7</sup>
			GTEEx2015_v6	Whole Blood, SKAP2	7.14×10 <sup>-6</sup>
			Lappalainen2013	Lymphoblastoid EUR exonlevel, ENSG00000005020.8	1.46×10 <sup>-8</sup>
rs7802715	7	26,852,720	GTEEx2015_v6	Thyroid, HOXA5	5.52×10 <sup>-6</sup>
			GTEEx2015_v6	Whole Blood, SKAP2	5.16×10 <sup>-6</sup>
			Lappalainen2013	Lymphoblastoid EUR exonlevel, ENSG00000005020.8	1.00×10 <sup>-8</sup>
rs7785815	7	26,852,728	GTEEx2015_v6	Thyroid, HOXA5	5.52×10 <sup>-6</sup>
			Lappalainen2013	Lymphoblastoid EUR exonlevel, ENSG00000005020.8	1.17×10 <sup>-8</sup>
rs114978183	7	26,852,740	.	.	.
rs115453073	7	26,852,741	GTEEx2015_v6	Thyroid, HOXA5	5.45×10 <sup>-6</sup>
			GTEEx2015_v6	Whole Blood, SKAP2	6.61×10 <sup>-6</sup>
			Lappalainen2013	Lymphoblastoid EUR exonlevel, ENSG00000005020.8	1.85×10 <sup>-8</sup>
rs7785832	7	26,852,760	GTEEx2015_v6	Thyroid, HOXA5	4.69×10 <sup>-7</sup>
			GTEEx2015_v6	Whole Blood, SKAP2	6.48×10 <sup>-6</sup>
			Lappalainen2013	Lymphoblastoid EUR exonlevel, ENSG00000005020.8	1.95×10 <sup>-8</sup>
			Westra2013	Whole Blood, SKAP2	1.08×10 <sup>-153</sup>
rs17315334	7	26,852,860	GTEEx2015_v6	Thyroid, HOXA5	4.69×10 <sup>-7</sup>
			GTEEx2015_v6	Whole Blood, SKAP2	6.47×10 <sup>-6</sup>
			Lappalainen2013	Lymphoblastoid EUR exonlevel, ENSG00000005020.8	2.02×10 <sup>-8</sup>
rs73067456	7	26,852,998	GTEEx2015_v6	Thyroid, HOXA5	4.67×10 <sup>-7</sup>
			GTEEx2015_v6	Whole Blood, SKAP2	7.00×10 <sup>-6</sup>
			Lappalainen2013	Lymphoblastoid EUR exonlevel, ENSG00000005020.8	1.48×10 <sup>-8</sup>
rs3801831	7	26,853,470	GTEEx2015_v6	Thyroid, HOXA5	4.67×10 <sup>-7</sup>
			GTEEx2015_v6	Whole Blood, SKAP2	6.98×10 <sup>-6</sup>
			Lappalainen2013	Lymphoblastoid EUR exonlevel, ENSG00000005020.8	4.27×10 <sup>-8</sup>
			Westra2013	Whole Blood, SKAP2	1.48×10 <sup>-153</sup>
rs3823938	7	26,853,741	GTEEx2015_v6	Thyroid, HOXA5	4.67×10 <sup>-7</sup>
			GTEEx2015_v6	Whole Blood, SKAP2	6.97×10 <sup>-6</sup>
			Lappalainen2013	Lymphoblastoid EUR exonlevel, ENSG00000005020.8	7.89×10 <sup>-8</sup>

rs3823936	7	26,853,830	GTEX2015_v6	Thyroid, HOXA5	4.67×10-7
			GTEX2015_v6	Whole Blood, SKAP2	6.97×10-6
			Lappalainen2013	Lymphoblastoid EUR exonlevel, ENSG00000005020.8	6.67×10-8
			Westra2013	Whole Blood, SKAP2	1.48×10-153
rs3823934	7	26,853,934	GTEX2015_v6	Thyroid, HOXA5	3.07×10-7
			GTEX2015_v6	Whole Blood, SKAP2	1.18×10-7
			Lappalainen2013	Lymphoblastoid EUR exonlevel, ENSG00000005020.8	7.81×10-8
			Westra2013	Whole Blood, SKAP2	1.19×10-153
rs3823933	7	26,853,966	GTEX2015_v6	Thyroid, HOXA5	4.67×10-7
			GTEX2015_v6	Whole Blood, SKAP2	6.97×10-6
			Lappalainen2013	Lymphoblastoid EUR exonlevel, ENSG00000005020.8	5.93×10-8
			Westra2013	Whole Blood, SKAP2	1.19×10-153
rs3823932	7	26,853,980	GTEX2015_v6	Thyroid, HOXA5	4.67×10-7
			GTEX2015_v6	Whole Blood, SKAP2	6.97×10-6
			Lappalainen2013	Lymphoblastoid EUR exonlevel, ENSG00000005020.8	7.33×10-8
			Westra2013	Whole Blood, SKAP2	1.19×10-153
rs3823931	7	26,854,026	GTEX2015_v6	Thyroid, HOXA5	4.67×10-7
			GTEX2015_v6	Whole Blood, SKAP2	6.97×10-6
			Lappalainen2013	Lymphoblastoid EUR exonlevel, ENSG00000005020.8	6.35×10-8
rs10262414	7	26,854,266	GTEX2015_v6	Thyroid, HOXA5	4.67×10-7
			GTEX2015_v6	Whole Blood, SKAP2	2.65×10-6
			Lappalainen2013	Lymphoblastoid EUR exonlevel, ENSG00000005020.8	6.76×10-8
			Westra2013	Whole Blood, SKAP2	8.07×10-154
rs12532199	7	26,854,375	GTEX2015_v6	Thyroid, HOXA5	4.67×10-7
			GTEX2015_v6	Whole Blood, SKAP2	6.97×10-6
			Lappalainen2013	Lymphoblastoid EUR exonlevel, ENSG00000005020.8	5.98×10-8
rs12537323	7	26,854,388	GTEX2015_v6	Thyroid, HOXA5	4.68×10-7
			GTEX2015_v6	Whole Blood, SKAP2	6.96×10-6
			Lappalainen2013	Lymphoblastoid EUR exonlevel, ENSG00000005020.8	5.99×10-8
			Westra2013	Whole Blood, SKAP2	1.08×10-153
rs28617235	7	26,854,671	GTEX2015_v6	Thyroid, HOXA5	4.67×10-7
			GTEX2015_v6	Whole Blood, SKAP2	6.97×10-6
			Lappalainen2013	Lymphoblastoid EUR exonlevel, ENSG00000005020.8	6.40×10-8
rs12532796	7	26,854,828	GTEX2015_v6	Thyroid, HOXA5	3.07×10-7
			GTEX2015_v6	Whole Blood, SKAP2	1.18×10-7
			Lappalainen2013	Lymphoblastoid EUR exonlevel, ENSG00000005020.8	6.76×10-8
			Westra2013	Whole Blood, SKAP2	1.08×10-153
rs3801830	7	26,854,870	GTEX2015_v6	Thyroid, HOXA5	4.74×10-7
			GTEX2015_v6	Whole Blood, SKAP2	6.98×10-6
			Lappalainen2013	Lymphoblastoid EUR exonlevel, ENSG00000005020.8	6.15×10-8
			Westra2013	Whole Blood, SKAP2	4.83×10-150
rs3801829	7	26,854,996	GTEX2015_v6	Thyroid, HOXA5	4.67×10-7
			GTEX2015_v6	Whole Blood, SKAP2	6.97×10-6
			Lappalainen2013	Lymphoblastoid EUR exonlevel, ENSG00000005020.8	6.01×10-8
rs10266759	7	26,855,282	GTEX2015_v6	Thyroid, HOXA5	4.67×10-7
			GTEX2015_v6	Whole Blood, SKAP2	6.97×10-6
			Lappalainen2013	Lymphoblastoid EUR exonlevel, ENSG00000005020.8	4.99×10-8
rs3801828	7	26,855,284	GTEX2015_v6	Thyroid, HOXA5	4.67×10-7
			GTEX2015_v6	Whole Blood, SKAP2	6.97×10-6
			Lappalainen2013	Lymphoblastoid EUR exonlevel, ENSG00000005020.8	6.41×10-8
			Westra2013	Whole Blood, SKAP2	1.08×10-153
rs6948584	7	26,855,732	GTEX2015_v6	Thyroid, HOXA5	4.36×10-7
			GTEX2015_v6	Whole Blood, SKAP2	7.38×10-6
			Lappalainen2013	Lymphoblastoid EUR exonlevel, ENSG00000005020.8	1.55×10-8

rs6948657	7	26,855,943	GTEEx2015_v6	Thyroid, HOXA5	4.43×10 <sup>-7</sup>
			GTEEx2015_v6	Whole Blood, SKAP2	6.72×10 <sup>-6</sup>
			Lappalainen2013	Lymphoblastoid EUR exonlevel, ENSG00000005020.8	1.52×10 <sup>-8</sup>
			Westra2013	Whole Blood, SKAP2	1.08×10 <sup>-153</sup>
rs73067472	7	26,855,959	GTEEx2015_v6	Thyroid, HOXA5	2.76×10 <sup>-7</sup>
			GTEEx2015_v6	Whole Blood, SKAP2	1.14×10 <sup>-7</sup>
			Lappalainen2013	Lymphoblastoid EUR exonlevel, ENSG00000005020.8	1.46×10 <sup>-8</sup>
rs3823930	7	26,856,525	GTEEx2015_v6	Thyroid, HOXA5	4.43×10 <sup>-7</sup>
			GTEEx2015_v6	Whole Blood, SKAP2	6.72×10 <sup>-6</sup>
			Lappalainen2013	Lymphoblastoid EUR exonlevel, ENSG00000005020.8	1.29×10 <sup>-8</sup>
rs3823929	7	26,856,600	GTEEx2015_v6	Thyroid, HOXA5	3.85×10 <sup>-7</sup>
			GTEEx2015_v6	Whole Blood, SKAP2	3.84×10 <sup>-6</sup>
			Lappalainen2013	Lymphoblastoid EUR exonlevel, ENSG00000005020.8	1.51×10 <sup>-8</sup>
rs3801827	7	26,856,626	GTEEx2015_v6	Thyroid, HOXA5	4.43×10 <sup>-7</sup>
			GTEEx2015_v6	Whole Blood, SKAP2	6.72×10 <sup>-6</sup>
			Lappalainen2013	Lymphoblastoid EUR exonlevel, ENSG00000005020.8	1.61×10 <sup>-8</sup>
rs3801826	7	26,856,720	GTEEx2015_v6	Thyroid, HOXA5	4.43×10 <sup>-7</sup>
			GTEEx2015_v6	Whole Blood, SKAP2	6.72×10 <sup>-6</sup>
			Lappalainen2013	Lymphoblastoid EUR exonlevel, ENSG00000005020.8	1.49×10 <sup>-8</sup>
rs3801825	7	26,856,959	GTEEx2015_v6	Thyroid, HOXA5	4.43×10 <sup>-7</sup>
			GTEEx2015_v6	Whole Blood, SKAP2	6.72×10 <sup>-6</sup>
			Lappalainen2013	Lymphoblastoid EUR exonlevel, ENSG00000005020.8	1.35×10 <sup>-8</sup>
			Westra2013	Whole Blood, SKAP2	8.56×10 <sup>-154</sup>
rs73067475	7	26,857,120	GTEEx2015_v6	Thyroid, HOXA5	4.43×10 <sup>-7</sup>
			GTEEx2015_v6	Whole Blood, SKAP2	6.72×10 <sup>-6</sup>
			Lappalainen2013	Lymphoblastoid EUR exonlevel, ENSG00000005020.8	1.41×10 <sup>-8</sup>
rs73067476	7	26,857,240	GTEEx2015_v6	Thyroid, HOXA5	4.43×10 <sup>-7</sup>
			GTEEx2015_v6	Whole Blood, SKAP2	6.72×10 <sup>-6</sup>
			Lappalainen2013	Lymphoblastoid EUR exonlevel, ENSG00000005020.8	1.43×10 <sup>-8</sup>
rs115447617	7	26,857,751	Lappalainen2013	Lymphoblastoid EUR exonlevel, ENSG00000005020.8	1.54×10 <sup>-8</sup>
rs111373218	7	26,859,152	GTEEx2015_v6	Thyroid, HOXA5	1.03×10 <sup>-5</sup>
			Lappalainen2013	Lymphoblastoid EUR exonlevel, ENSG00000005020.8	2.46×10 <sup>-9</sup>
rs17315775	7	26,860,134	GTEEx2015_v6	Thyroid, HOXA5	4.43×10 <sup>-7</sup>
			GTEEx2015_v6	Whole Blood, SKAP2	6.71×10 <sup>-6</sup>
			Lappalainen2013	Lymphoblastoid EUR exonlevel, ENSG00000005020.8	1.37×10 <sup>-8</sup>
			Westra2013	Whole Blood, SKAP2	8.56×10 <sup>-154</sup>
rs28404962	7	26,860,324	GTEEx2015_v6	Thyroid, HOXA5	4.43×10 <sup>-7</sup>
			GTEEx2015_v6	Whole Blood, SKAP2	6.71×10 <sup>-6</sup>
			Lappalainen2013	Lymphoblastoid EUR exonlevel, ENSG00000005020.8	1.51×10 <sup>-8</sup>
rs13438513	7	26,860,633	GTEEx2015_v6	Thyroid, HOXA5	4.43×10 <sup>-7</sup>
			GTEEx2015_v6	Whole Blood, SKAP2	6.71×10 <sup>-6</sup>
			Lappalainen2013	Lymphoblastoid EUR exonlevel, ENSG00000005020.8	1.84×10 <sup>-8</sup>
			Westra2013	Whole Blood, SKAP2	8.56×10 <sup>-154</sup>
rs13438514	7	26,860,688	GTEEx2015_v6	Thyroid, HOXA5	4.45×10 <sup>-7</sup>
			GTEEx2015_v6	Whole Blood, SKAP2	6.53×10 <sup>-6</sup>
			Lappalainen2013	Lymphoblastoid EUR exonlevel, ENSG00000005020.8	1.67×10 <sup>-8</sup>
			Westra2013	Whole Blood, SKAP2	8.56×10 <sup>-154</sup>
rs55739485	7	26,860,741	GTEEx2015_v6	Thyroid, HOXA5	4.42×10 <sup>-7</sup>
			GTEEx2015_v6	Whole Blood, SKAP2	6.72×10 <sup>-6</sup>
			Lappalainen2013	Lymphoblastoid EUR exonlevel, ENSG00000005020.8	1.53×10 <sup>-8</sup>
rs3801823	7	26,861,031	GTEEx2015_v6	Thyroid, HOXA5	4.32×10 <sup>-7</sup>
			GTEEx2015_v6	Whole Blood, SKAP2	8.96×10 <sup>-6</sup>
			Lappalainen2013	Lymphoblastoid EUR exonlevel, ENSG00000005020.8	1.66×10 <sup>-8</sup>
rs3801822	7	26,861,201	GTEEx2015_v6	Thyroid, HOXA5	4.42×10 <sup>-7</sup>
			GTEEx2015_v6	Whole Blood, SKAP2	6.72×10 <sup>-6</sup>
			Lappalainen2013	Lymphoblastoid EUR exonlevel, ENSG00000005020.8	2.09×10 <sup>-8</sup>

rs3801821	7	26,861,206	GTEEx2015_v6	Thyroid, HOXA5	4.42×10 <sup>-7</sup>
			GTEEx2015_v6	Whole Blood, SKAP2	6.72×10 <sup>-6</sup>
			Lappalainen2013	Lymphoblastoid EUR exonlevel, ENSG00000005020.8	1.98×10 <sup>-8</sup>
			Westra2013	Whole Blood, SKAP2	8.56×10 <sup>-154</sup>
rs3801820	7	26,861,353	GTEEx2015_v6	Thyroid, HOXA5	4.42×10 <sup>-7</sup>
			GTEEx2015_v6	Whole Blood, SKAP2	6.72×10 <sup>-6</sup>
			Lappalainen2013	Lymphoblastoid EUR exonlevel, ENSG00000005020.8	1.51×10 <sup>-8</sup>
			Westra2013	Whole Blood, SKAP2	8.56×10 <sup>-154</sup>
rs7785689	7	26,861,609	GTEEx2015_v6	Thyroid, HOXA5	4.42×10 <sup>-7</sup>
			GTEEx2015_v6	Whole Blood, SKAP2	6.72×10 <sup>-6</sup>
			Lappalainen2013	Lymphoblastoid EUR exonlevel, ENSG00000005020.8	1.41×10 <sup>-8</sup>
rs7785711	7	26,861,640	GTEEx2015_v6	Thyroid, HOXA5	4.29×10 <sup>-7</sup>
			GTEEx2015_v6	Whole Blood, SKAP2	7.32×10 <sup>-6</sup>
			Lappalainen2013	Lymphoblastoid EUR exonlevel, ENSG00000005020.8	1.33×10 <sup>-8</sup>
rs7785798	7	26,861,943	GTEEx2015_v6	Thyroid, HOXA5	4.42×10 <sup>-7</sup>
			GTEEx2015_v6	Whole Blood, SKAP2	6.72×10 <sup>-6</sup>
			Lappalainen2013	Lymphoblastoid EUR exonlevel, ENSG00000005020.8	1.71×10 <sup>-8</sup>
rs7785918	7	26,861,959	GTEEx2015_v6	Thyroid, HOXA5	4.35×10 <sup>-7</sup>
			GTEEx2015_v6	Whole Blood, SKAP2	6.93×10 <sup>-6</sup>
			Lappalainen2013	Lymphoblastoid EUR exonlevel, ENSG00000005020.8	1.59×10 <sup>-8</sup>
rs12540045	7	26,863,174	GTEEx2015_v6	Thyroid, HOXA5	4.43×10 <sup>-7</sup>
			GTEEx2015_v6	Whole Blood, SKAP2	6.71×10 <sup>-6</sup>
			Lappalainen2013	Lymphoblastoid EUR exonlevel, ENSG00000005020.8	1.43×10 <sup>-8</sup>
rs10248500	7	26,863,294	GTEEx2015_v6	Thyroid, HOXA5	4.43×10 <sup>-7</sup>
			GTEEx2015_v6	Whole Blood, SKAP2	6.71×10 <sup>-6</sup>
			Lappalainen2013	Lymphoblastoid EUR exonlevel, ENSG00000005020.8	1.53×10 <sup>-8</sup>
rs17315929	7	26,864,173	GTEEx2015_v6	Thyroid, HOXA5	4.43×10 <sup>-7</sup>
			GTEEx2015_v6	Whole Blood, SKAP2	6.65×10 <sup>-6</sup>
			Lappalainen2013	Lymphoblastoid EUR exonlevel, ENSG00000005020.8	5.25×10 <sup>-8</sup>
			Westra2013	Whole Blood, SKAP2	5.78×10 <sup>-154</sup>
rs10227673	7	26,865,299	GTEEx2015_v6	Thyroid, HOXA5	4.43×10 <sup>-7</sup>
			GTEEx2015_v6	Whole Blood, SKAP2	6.71×10 <sup>-6</sup>
			Lappalainen2013	Lymphoblastoid EUR exonlevel, ENSG00000005020.8	1.15×10 <sup>-8</sup>
rs7805766	7	26,865,610	GTEEx2015_v6	Thyroid, HOXA5	4.43×10 <sup>-7</sup>
			GTEEx2015_v6	Whole Blood, SKAP2	1.11×10 <sup>-5</sup>
			Lappalainen2013	Lymphoblastoid EUR exonlevel, ENSG00000005020.8	1.16×10 <sup>-8</sup>
			Westra2013	Whole Blood, SKAP2	5.37×10 <sup>-159</sup>
rs17375272	7	26,866,497	GTEEx2015_v6	Thyroid, HOXA5	2.97×10 <sup>-7</sup>
			Lappalainen2013	Lymphoblastoid EUR exonlevel, ENSG00000005020.8	1.30×10 <sup>-8</sup>
rs3801819	7	26,866,698	GTEEx2015_v6	Thyroid, HOXA5	2.76×10 <sup>-7</sup>
			GTEEx2015_v6	Whole Blood, SKAP2	1.14×10 <sup>-7</sup>
			Lappalainen2013	Lymphoblastoid EUR exonlevel, ENSG00000005020.8	1.39×10 <sup>-8</sup>
			Westra2013	Whole Blood, SKAP2	1.94×10 <sup>-154</sup>
rs61354389	7	26,868,343	GTEEx2015_v6	Thyroid, HOXA5	4.43×10 <sup>-7</sup>
			GTEEx2015_v6	Whole Blood, SKAP2	6.71×10 <sup>-6</sup>
			Lappalainen2013	Lymphoblastoid EUR exonlevel, ENSG00000005020.8	1.10×10 <sup>-8</sup>
rs10225700	7	26,869,880	GTEEx2015_v6	Thyroid, HOXA5	2.26×10 <sup>-6</sup>
			GTEEx2015_v6	Whole Blood, SKAP2	8.05×10 <sup>-6</sup>
			Lappalainen2013	Lymphoblastoid EUR exonlevel, ENSG00000005020.8	3.62×10 <sup>-8</sup>
rs10241234	7	26,869,887	GTEEx2015_v6	Thyroid, HOXA5	3.78×10 <sup>-6</sup>
			GTEEx2015_v6	Whole Blood, SKAP2	3.39×10 <sup>-6</sup>
			Lappalainen2013	Lymphoblastoid EUR exonlevel, ENSG00000005020.8	4.69×10 <sup>-8</sup>
rs10225904	7	26,869,888	GTEEx2015_v6	Thyroid, HOXA5	3.78×10 <sup>-6</sup>
			GTEEx2015_v6	Whole Blood, SKAP2	3.39×10 <sup>-6</sup>
			Lappalainen2013	Lymphoblastoid EUR exonlevel, ENSG00000005020.8	4.34×10 <sup>-8</sup>

rs28550029	7	26,872,581	GTEEx2015_v6	Thyroid, HOXA5	4.43×10 <sup>-7</sup>			
			GTEEx2015_v6	Whole Blood, SKAP2	6.71×10 <sup>-6</sup>			
			Lappalainen2013	Lymphoblastoid EUR exonlevel, ENSG00000005020.8	1.40×10 <sup>-8</sup>			
rs114236648	7	26,874,639	Lappalainen2013	Lymphoblastoid EUR exonlevel, ENSG00000005020.8	2.20×10 <sup>-8</sup>			
			rs12534899	7	26,877,189	GTEEx2015_v6	Thyroid, HOXA5	4.43×10 <sup>-7</sup>
						GTEEx2015_v6	Whole Blood, SKAP2	6.71×10 <sup>-6</sup>
Lappalainen2013	Lymphoblastoid EUR exonlevel, ENSG00000005020.8	1.46×10 <sup>-8</sup>						
rs12534921	7	26,877,210	Westra2013	Whole Blood, SKAP2	7.96×10 <sup>-155</sup>			
			GTEEx2015_v6	Thyroid, HOXA5	4.43×10 <sup>-7</sup>			
			GTEEx2015_v6	Whole Blood, SKAP2	6.71×10 <sup>-6</sup>			
rs73069507	7	26,877,536	Lappalainen2013	Lymphoblastoid EUR exonlevel, ENSG00000005020.8	2.10×10 <sup>-8</sup>			
			GTEEx2015_v6	Thyroid, HOXA5	4.43×10 <sup>-7</sup>			
			GTEEx2015_v6	Whole Blood, SKAP2	6.71×10 <sup>-6</sup>			
rs73069510	7	26,877,567	Lappalainen2013	Lymphoblastoid EUR exonlevel, ENSG00000005020.8	2.30×10 <sup>-8</sup>			
			GTEEx2015_v6	Thyroid, HOXA5	3.32×10 <sup>-7</sup>			
			GTEEx2015_v6	Whole Blood, SKAP2	6.71×10 <sup>-6</sup>			
rs3801816	7	26,881,177	Lappalainen2013	Lymphoblastoid EUR exonlevel, ENSG00000005020.8	1.86×10 <sup>-8</sup>			
			GTEEx2015_v6	Thyroid, HOXA5	4.57×10 <sup>-7</sup>			
			GTEEx2015_v6	Whole Blood, SKAP2	6.99×10 <sup>-6</sup>			
			Westra2013	Whole Blood, SKAP2	5.64×10 <sup>-155</sup>			
rs3801815	7	26,881,292	Lappalainen2013	Lymphoblastoid EUR exonlevel, ENSG00000005020.8	9.22×10 <sup>-9</sup>			
			GTEEx2015_v6	Thyroid, HOXA5	4.76×10 <sup>-7</sup>			
			GTEEx2015_v6	Whole Blood, SKAP2	2.22×10 <sup>-6</sup>			
			Westra2013	Whole Blood, SKAP2	5.64×10 <sup>-155</sup>			
rs3801814	7	26,881,302	Lappalainen2013	Lymphoblastoid EUR exonlevel, ENSG00000005020.8	1.90×10 <sup>-8</sup>			
			GTEEx2015_v6	Thyroid, HOXA5	4.67×10 <sup>-7</sup>			
			GTEEx2015_v6	Whole Blood, SKAP2	2.65×10 <sup>-6</sup>			
			Westra2013	Whole Blood, SKAP2	4.73×10 <sup>-155</sup>			
rs12535784	7	26,882,067	Lappalainen2013	Lymphoblastoid EUR exonlevel, ENSG00000005020.8	1.74×10 <sup>-8</sup>			
			GTEEx2015_v6	Thyroid, HOXA5	4.83×10 <sup>-7</sup>			
			GTEEx2015_v6	Whole Blood, SKAP2	6.05×10 <sup>-6</sup>			
			Westra2013	Whole Blood, SKAP2	7.01×10 <sup>-155</sup>			
rs4719882	7	26,882,252	Lappalainen2013	Lymphoblastoid EUR exonlevel, ENSG00000005020.8	1.60×10 <sup>-8</sup>			
			GTEEx2015_v6	Thyroid, HOXA5	3.04×10 <sup>-7</sup>			
			GTEEx2015_v6	Whole Blood, SKAP2	6.58×10 <sup>-6</sup>			
rs1982995	7	26,884,135	Lappalainen2013	Lymphoblastoid EUR exonlevel, ENSG00000005020.8	8.99×10 <sup>-7</sup>			
			GTEEx2015_v6	Thyroid, HOXA5	3.48×10 <sup>-7</sup>			
			GTEEx2015_v6	Whole Blood, SKAP2	5.50×10 <sup>-6</sup>			
rs10236221	7	26,884,671	Lappalainen2013	Lymphoblastoid EUR exonlevel, ENSG00000005020.8	1.51×10 <sup>-8</sup>			
			GTEEx2015_v6	Thyroid, HOXA5	4.87×10 <sup>-7</sup>			
			GTEEx2015_v6	Whole Blood, SKAP2	5.81×10 <sup>-6</sup>			
rs10270187	7	26,885,885	Lappalainen2013	Lymphoblastoid EUR exonlevel, ENSG00000005020.8	1.44×10 <sup>-8</sup>			
			GTEEx2015_v6	Thyroid, HOXA5	4.97×10 <sup>-7</sup>			
			GTEEx2015_v6	Whole Blood, SKAP2	7.15×10 <sup>-6</sup>			
			Westra2013	Whole Blood, SKAP2	7.01×10 <sup>-155</sup>			
rs10227529	7	26,886,309	Lappalainen2013	Lymphoblastoid EUR exonlevel, ENSG00000005020.8	1.79×10 <sup>-7</sup>			
			GTEEx2015_v6	Thyroid, HOXA5	5.53×10 <sup>-7</sup>			
			GTEEx2015_v6	Whole Blood, SKAP2	7.09×10 <sup>-6</sup>			
rs3801813	7	26,887,069	Lappalainen2013	Lymphoblastoid EUR exonlevel, ENSG00000005020.8	7.77×10 <sup>-9</sup>			
			GTEEx2015_v6	Thyroid, HOXA5	4.19×10 <sup>-7</sup>			
			GTEEx2015_v6	Whole Blood, SKAP2	6.20×10 <sup>-6</sup>			
			Westra2013	Whole Blood, SKAP2	8.72×10 <sup>-155</sup>			
rs3801812	7	26,887,615	Lappalainen2013	Lymphoblastoid EUR exonlevel, ENSG00000005020.8	1.62×10 <sup>-8</sup>			
			GTEEx2015_v6	Thyroid, HOXA5	4.97×10 <sup>-7</sup>			
			GTEEx2015_v6	Whole Blood, SKAP2	7.13×10 <sup>-6</sup>			
			Westra2013	Whole Blood, SKAP2	1.08×10 <sup>-154</sup>			

rs10272190	7	26,887,942	GTEEx2015_v6	Thyroid, HOXA5	4.97×10 <sup>-7</sup>
			GTEEx2015_v6	Whole Blood, SKAP2	7.12×10 <sup>-6</sup>
			Lappalainen2013	Lymphoblastoid EUR exonlevel, ENSG00000005020.8	1.58×10 <sup>-8</sup>
			Westra2013	Whole Blood, SKAP2	1.08×10 <sup>-154</sup>
rs10227231	7	26,888,074	GTEEx2015_v6	Thyroid, HOXA5	9.43×10 <sup>-7</sup>
			GTEEx2015_v6	Whole Blood, SKAP2	1.07×10 <sup>-6</sup>
			Lappalainen2013	Lymphoblastoid EUR exonlevel, ENSG00000005020.8	1.26×10 <sup>-8</sup>
			Westra2013	Whole Blood, SKAP2	1.08×10 <sup>-154</sup>
rs10272457	7	26,888,155	GTEEx2015_v6	Thyroid, HOXA5	4.97×10 <sup>-7</sup>
			GTEEx2015_v6	Whole Blood, SKAP2	7.11×10 <sup>-6</sup>
			Lappalainen2013	Lymphoblastoid EUR exonlevel, ENSG00000005020.8	1.51×10 <sup>-8</sup>
			Westra2013	Whole Blood, SKAP2	1.08×10 <sup>-154</sup>
rs10272603	7	26,888,277	GTEEx2015_v6	Thyroid, HOXA5	4.97×10 <sup>-7</sup>
			GTEEx2015_v6	Whole Blood, SKAP2	7.11×10 <sup>-6</sup>
			Lappalainen2013	Lymphoblastoid EUR exonlevel, ENSG00000005020.8	1.65×10 <sup>-8</sup>
			GTEEx2015_v6	Thyroid, HOXA5	2.92×10 <sup>-7</sup>
rs6461979	7	26,888,918	GTEEx2015_v6	Whole Blood, SKAP2	1.51×10 <sup>-7</sup>
			Lappalainen2013	Lymphoblastoid EUR exonlevel, ENSG00000005020.8	1.49×10 <sup>-8</sup>
			Westra2013	Whole Blood, SKAP2	1.35×10 <sup>-154</sup>
			GTEEx2015_v6	Thyroid, HOXA5	3.35×10 <sup>-7</sup>
rs17376018	7	26,889,862	GTEEx2015_v6	Whole Blood, SKAP2	1.03×10 <sup>-5</sup>
			Lappalainen2013	Lymphoblastoid EUR exonlevel, ENSG00000005020.8	1.41×10 <sup>-8</sup>
			GTEEx2015_v6	Thyroid, HOXA5	4.45×10 <sup>-7</sup>
			GTEEx2015_v6	Whole Blood, SKAP2	6.36×10 <sup>-6</sup>
rs4722647	7	26,890,446	Lappalainen2013	Lymphoblastoid EUR exonlevel, ENSG00000005020.8	1.10×10 <sup>-8</sup>
			GTEEx2015_v6	Thyroid, HOXA5	4.68×10 <sup>-7</sup>
			GTEEx2015_v6	Whole Blood, SKAP2	6.78×10 <sup>-6</sup>
			Lappalainen2013	Lymphoblastoid EUR exonlevel, ENSG00000005020.8	1.37×10 <sup>-8</sup>
rs3934268	7	26,890,572	GTEEx2015_v6	Thyroid, HOXA5	4.67×10 <sup>-7</sup>
			GTEEx2015_v6	Whole Blood, SKAP2	6.78×10 <sup>-6</sup>
			Lappalainen2013	Lymphoblastoid EUR exonlevel, ENSG00000005020.8	1.31×10 <sup>-8</sup>
			Westra2013	Whole Blood, SKAP2	1.35×10 <sup>-154</sup>
rs2031811	7	26,890,672	GTEEx2015_v6	Thyroid, HOXA5	4.69×10 <sup>-7</sup>
			Lappalainen2013	Lymphoblastoid EUR exonlevel, ENSG00000005020.8	8.30×10 <sup>-9</sup>
			Westra2013	Whole Blood, SKAP2	4.99×10 <sup>-141</sup>
			GTEEx2015_v6	Thyroid, HOXA5	4.67×10 <sup>-7</sup>
rs2030136	7	26,891,016	GTEEx2015_v6	Whole Blood, SKAP2	6.78×10 <sup>-6</sup>
			Lappalainen2013	Lymphoblastoid EUR exonlevel, ENSG00000005020.8	9.69×10 <sup>-9</sup>
			Westra2013	Whole Blood, SKAP2	6.51×10 <sup>-155</sup>
			GTEEx2015_v6	Thyroid, HOXA5	8.76×10 <sup>-7</sup>
rs7804356	7	26,891,665	Lappalainen2013	Lymphoblastoid EUR exonlevel, ENSG00000005020.8	1.18×10 <sup>-8</sup>
			GTEEx2015_v6	Thyroid, HOXA5	2.75×10 <sup>-7</sup>
			GTEEx2015_v6	Whole Blood, SKAP2	1.43×10 <sup>-7</sup>
			Lappalainen2013	Lymphoblastoid EUR exonlevel, ENSG00000005020.8	1.64×10 <sup>-8</sup>
rs7804620	7	26,891,831	GTEEx2015_v6	Thyroid, HOXA5	4.67×10 <sup>-7</sup>
			GTEEx2015_v6	Whole Blood, SKAP2	6.75×10 <sup>-6</sup>
			Lappalainen2013	Lymphoblastoid EUR exonlevel, ENSG00000005020.8	1.69×10 <sup>-8</sup>
			Westra2013	Whole Blood, SKAP2	1.35×10 <sup>-154</sup>
rs73069533	7	26,892,325	GTEEx2015_v6	Thyroid, HOXA5	4.59×10 <sup>-7</sup>
			GTEEx2015_v6	Whole Blood, SKAP2	8.97×10 <sup>-6</sup>
			Lappalainen2013	Lymphoblastoid EUR exonlevel, ENSG00000005020.8	1.69×10 <sup>-8</sup>
			Westra2013	Whole Blood, SKAP2	1.35×10 <sup>-154</sup>
rs10486483	7	26,892,440	GTEEx2015_v6	Thyroid, HOXA5	4.68×10 <sup>-7</sup>
			GTEEx2015_v6	Whole Blood, SKAP2	6.74×10 <sup>-6</sup>
			Lappalainen2013	Lymphoblastoid EUR exonlevel, ENSG00000005020.8	1.44×10 <sup>-8</sup>
			GTEEx2015_v6	Thyroid, HOXA5	4.59×10 <sup>-7</sup>
rs3801810	7	26,892,531	GTEEx2015_v6	Whole Blood, SKAP2	8.97×10 <sup>-6</sup>
			Lappalainen2013	Lymphoblastoid EUR exonlevel, ENSG00000005020.8	1.69×10 <sup>-8</sup>
			Westra2013	Whole Blood, SKAP2	1.35×10 <sup>-154</sup>
			GTEEx2015_v6	Thyroid, HOXA5	4.68×10 <sup>-7</sup>
rs3823928	7	26,893,317	GTEEx2015_v6	Whole Blood, SKAP2	6.74×10 <sup>-6</sup>
			Lappalainen2013	Lymphoblastoid EUR exonlevel, ENSG00000005020.8	1.44×10 <sup>-8</sup>
			GTEEx2015_v6	Thyroid, HOXA5	4.68×10 <sup>-7</sup>
			GTEEx2015_v6	Whole Blood, SKAP2	6.74×10 <sup>-6</sup>

rs7791513	7	26,893,357	GTEEx2015_v6	Thyroid, HOXA5	5.40×10-7
			GTEEx2015_v6	Whole Blood, SKAP2	6.40×10-6
			Lappalainen2013	Lymphoblastoid EUR exonlevel, ENSG00000005020.8	1.42×10-8
			Westra2013	Whole Blood, SKAP2	1.35×10-154
rs3213853	7	26,893,524	GTEEx2015_v6	Thyroid, HOXA5	4.68×10-7
			GTEEx2015_v6	Whole Blood, SKAP2	6.74×10-6
			Lappalainen2013	Lymphoblastoid EUR exonlevel, ENSG00000005020.8	1.32×10-8
rs6461980	7	26,894,123	GTEEx2015_v6	Thyroid, HOXA5	4.68×10-7
			GTEEx2015_v6	Whole Blood, SKAP2	6.70×10-6
			Lappalainen2013	Lymphoblastoid EUR exonlevel, ENSG00000005020.8	1.40×10-8
rs12536145	7	26,894,860	GTEEx2015_v6	Thyroid, HOXA5	4.69×10-7
			GTEEx2015_v6	Whole Blood, SKAP2	6.22×10-6
			Lappalainen2013	Lymphoblastoid EUR exonlevel, ENSG00000005020.8	1.31×10-8
rs10242572	7	26,895,065	GTEEx2015_v6	Thyroid, HOXA5	4.70×10-7
			GTEEx2015_v6	Whole Blood, SKAP2	6.83×10-6
			Lappalainen2013	Lymphoblastoid EUR exonlevel, ENSG00000005020.8	2.09×10-8
			Westra2013	Whole Blood, SKAP2	9.34×10-143
rs10256250	7	26,895,080	GTEEx2015_v6	Thyroid, HOXA5	4.70×10-7
			GTEEx2015_v6	Whole Blood, SKAP2	6.07×10-6
			Lappalainen2013	Lymphoblastoid EUR exonlevel, ENSG00000005020.8	1.53×10-8
			Westra2013	Whole Blood, SKAP2	1.35×10-154
rs3801809	7	26,895,456	GTEEx2015_v6	Thyroid, HOXA5	4.73×10-7
			GTEEx2015_v6	Whole Blood, SKAP2	5.56×10-6
			Lappalainen2013	Lymphoblastoid EUR exonlevel, ENSG00000005020.8	1.29×10-8
rs3801808	7	26,896,555	GTEEx2015_v6	Thyroid, HOXA5	4.92×10-7
			GTEEx2015_v6	Whole Blood, SKAP2	4.36×10-6
			Lappalainen2013	Lymphoblastoid EUR exonlevel, ENSG00000005020.8	3.77×10-8
			Westra2013	Whole Blood, SKAP2	2.11×10-150
rs3801807	7	26,897,439	GTEEx2015_v6	Thyroid, HOXA5	4.91×10-7
			GTEEx2015_v6	Whole Blood, SKAP2	4.34×10-6
			Lappalainen2013	Lymphoblastoid EUR exonlevel, ENSG00000005020.8	1.32×10-8
			Westra2013	Whole Blood, SKAP2	1.16×10-154
rs3801805	7	26,897,640	GTEEx2015_v6	Thyroid, HOXA5	1.63×10-6
			GTEEx2015_v6	Whole Blood, SKAP2	2.40×10-6
			Lappalainen2013	Lymphoblastoid EUR exonlevel, ENSG00000005020.8	1.81×10-8
			Westra2013	Whole Blood, SKAP2	1.35×10-154
rs10486485	7	26,900,356	GTEEx2015_v6	Thyroid, HOXA5	1.05×10-6
			GTEEx2015_v6	Whole Blood, SKAP2	5.74×10-7
			Lappalainen2013	Lymphoblastoid EUR exonlevel, ENSG00000005020.8	3.82×10-8
			Westra2013	Whole Blood, SKAP2	7.16×10-155
rs3801803	7	26,901,740	GTEEx2015_v6	Thyroid, HOXA5	8.85×10-7
			GTEEx2015_v6	Whole Blood, SKAP2	1.41×10-6
			Lappalainen2013	Lymphoblastoid EUR exonlevel, ENSG00000005020.8	2.58×10-8
			Westra2013	Whole Blood, SKAP2	7.16×10-155
rs73069539	7	26,903,661	GTEEx2015_v6	Thyroid, HOXA5	4.23×10-7
			GTEEx2015_v6	Whole Blood, SKAP2	6.23×10-9
			Lappalainen2013	Lymphoblastoid EUR exonlevel, ENSG00000005020.8	5.06×10-8
rs17323934	7	26,904,330	GTEEx2015_v6	Thyroid, HOXA5	7.33×10-7
			GTEEx2015_v6	Whole Blood, SKAP2	9.46×10-9
			Lappalainen2013	Lymphoblastoid EUR exonlevel, ENSG00000005020.8	3.67×10-9
			Westra2013	Whole Blood, SKAP2	1.58×10-161
rs73069540	7	26,904,770	GTEEx2015_v6	Thyroid, HOXA5	3.72×10-7
			GTEEx2015_v6	Whole Blood, SKAP2	5.40×10-9
			Lappalainen2013	Lymphoblastoid EUR exonlevel, ENSG00000005020.8	2.54×10-9
rs73069541	7	26,904,928	GTEEx2015_v6	Thyroid, HOXA5	3.71×10-7
			GTEEx2015_v6	Whole Blood, SKAP2	5.29×10-9
			Lappalainen2013	Lymphoblastoid EUR exonlevel, ENSG00000005020.8	2.24×10-9



rs12540388	7	26,905,731	GTEX2015_v6	Thyroid, HOXA5	3.60×10 <sup>-7</sup>
			GTEX2015_v6	Whole Blood, SKAP2	4.85×10 <sup>-9</sup>
			Lappalainen2013	Lymphoblastoid EUR exonlevel, ENSG00000005020.8	3.00×10 <sup>-9</sup>
			Westra2013	Whole Blood, SKAP2	3.49×10 <sup>-163</sup>
rs2391362	7	26,907,123	GTEX2015_v6	Thyroid, HOXA5	3.35×10 <sup>-7</sup>
			GTEX2015_v6	Whole Blood, SKAP2	5.01×10 <sup>-9</sup>
			Lappalainen2013	Lymphoblastoid EUR exonlevel, ENSG00000005020.8	4.56×10 <sup>-9</sup>
			Westra2013	Whole Blood, SKAP2	1.64×10 <sup>-162</sup>
rs76438067	7	26,911,904	GTEX2015_v6	Thyroid, HOXA5	4.30×10 <sup>-7</sup>
			GTEX2015_v6	Whole Blood, SKAP2	2.29×10 <sup>-9</sup>
			Lappalainen2013	Lymphoblastoid EUR exonlevel, ENSG00000005020.8	3.77×10 <sup>-9</sup>

## \*eQTL resources used by HaploReg

id	pmid	name	source
Montgomery2010	20220756	Transcriptome genetics using second generation sequencing in a Caucasian population	From NCBI eQTL Browser, <a href="http://www.ncbi.nlm.nih.gov/projects/gap/eqtl/index.cgi">http://www.ncbi.nlm.nih.gov/projects/gap/eqtl/index.cgi</a>
Schadt2008	18462017	Mapping the genetic architecture of gene expression in human liver	From NCBI eQTL Browser, <a href="http://www.ncbi.nlm.nih.gov/projects/gap/eqtl/index.cgi">http://www.ncbi.nlm.nih.gov/projects/gap/eqtl/index.cgi</a>
Gibbs2010	20485568	Abundant quantitative trait loci exist for DNA methylation and gene expression in human brain	From NCBI eQTL Browser, <a href="http://www.ncbi.nlm.nih.gov/projects/gap/eqtl/index.cgi">http://www.ncbi.nlm.nih.gov/projects/gap/eqtl/index.cgi</a>
Stranger2008	17873874	Population genomics of human gene expression	From NCBI eQTL Browser, <a href="http://www.ncbi.nlm.nih.gov/projects/gap/eqtl/index.cgi">http://www.ncbi.nlm.nih.gov/projects/gap/eqtl/index.cgi</a>
GTEX2015_v6	25954001	The Genotype-Tissue Expression (GTEx) pilot analysis: Multitissue gene regulation in humans	From GTEx Portal, Release v6, from <a href="http://www.gtexportal.org/static/datasets/gtex_analysis_v6/single_tissue_eqtl_data/GTex_Analysis_V6_eQTLs.tar.gz">http://www.gtexportal.org/static/datasets/gtex_analysis_v6/single_tissue_eqtl_data/GTex_Analysis_V6_eQTLs.tar.gz</a>
Li2014	24907074	Expression QTL-based analyses reveal candidate causal genes and loci across five tumor types	From Table S1. cis-eQTL for mRNA were exported from each tab of the Excel file.
Westra2013	24013639	Systematic identification of trans eQTLs as putative drivers of known disease associations	All associations from 2012-12-21-CisAssociationsProbeLevelFDR0.5.txt
Hao2012	23209423	Lung eQTLs to help reveal the molecular underpinnings of asthma	All cis and trans eQTLs from Tables S2A and S2B were combined in the provided Excel and the p value in the field "pvalue.fix" was used
Koopmann2014	24846176	Genome-wide identification of expression quantitative trait loci (eQTLs) in human heart	All associations from Table S1
Fairfax2014	24604202	Innate immune activity conditions the effect of regulatory variants upon monocyte gene expression	All associations from Tables S2B and S3B. Tables were flattened by the four treatments and filtered for "NA" reported as a p-value.
Grundberg2011	21283786	Global Analysis of the Impact of Environmental Perturbation on cis-Regulation of Gene Expression	All associations from Table S6
Lappalainen2013	24037378	Transcriptome and genome sequencing uncovers functional variation in humans	All gene- and exon-level eQTLs from EUR and YRI. Gene-level eQTL genes are translated from ENSG to HGNC names when possible; exons are named by probe name.
Ramasamy2014	25174007	Genetic variability in the regulation of gene expression in ten regions of the human brain	All associations reported in Table S2

**SNP:** SNP rs ID; **CHR:** Chromosome; **BP:** position (NCBI build 37/hg19 coordinates) of SNP marker; **eQTL resource id:** publication that reports SNP as eQTL; **Selected eQTL hits:** selection of eQTL hits, shown are cell types/tissues and the respective gene; **P-val:** *P*-value of eQTL for the respective gene in the respective cell type/tissue.

**Supplementary Table 7. Gene expression of genes *EFEMP1*, *SKAP2* and *KCNH8* across multiple tissues/cell lines from ENCODE data.**

Expression data was obtained from website [http://promoter.bx.psu.edu/ENCODE/search\\_human.php](http://promoter.bx.psu.edu/ENCODE/search_human.php).

tissues/ cell lines	cell	Description	RPKM <sub>EFEMP1</sub>	RPKM <sub>SKAP2</sub>	RPKM <sub>KCNH8</sub>
A549_cell_longNonPolyA	A549	epithelial cell line derived from a lung carcinoma tissue	17.08	2.83	0.00
A549_cell_longPolyA			42.21	17.01	0.00
A549_cytosol_longPolyA			55.50	5.02	0.00
A549_nucleus_longPolyA			53.59	1.38	0.00
AG04450_cell_longNonPolyA	AG04450	fetal lung fibroblast	93.71	4.50	0.00
AG04450_cell_longPolyA			352.58	13.02	0.00
BJ_cell_longNonPolyA	BJ	skin fibroblast	273.33	1.04	0.00
BJ_cell_longPolyA			1124.11	3.90	0.00
CD20_Plus_cell_longNonPolyA	CD20+	B cells	0.00	9.58	4.77
CD20_Plus_cell_longPolyA			0.00	11.93	13.12
GM12878_cell_longNonPolyA	GM12878	B-lymphocyte, lymphoblastoid	1.73	3.24	0.00
GM12878_cell_longPolyA			0.51	18.12	0.04
GM12878_cytosol_longNonPolyA			0.00	1.20	0.00
GM12878_cytosol_longPolyA			0.84	9.24	0.00
GM12878_nucleolus_total			0.13	2.83	0.00
GM12878_nucleus_longNonPolyA			0.70	4.59	0.05
GM12878_nucleus_longPolyA			0.58	2.74	0.05
H1_hESC_cell_longNonPolyA	H1-hESC	embryonic stem cells	20.87	0.32	6.60
H1_hESC_cell_longPolyA			19.29	0.60	3.09
HAoAF_cell_total	HAoAF	Aortic Adventitial Fibroblasts	216.59	7.72	0.00
HAoEC_cell_total	HAoEC	Aortic Endothelial Cells (thoracic)	565.17	11.85	0.00
HAoSMC_cell_total	HAoSMC	Human Aortic Smooth Muscle Cells	694.76	0.79	0.00
HBdMEC_cell_total	HBdMEC	Human Bladder Microvascular Endothelial Cells	4649.97	11.11	0.00
HBdSMC_cell_total	HBdSMC	Human Bladder Smooth Muscle Cells	269.47	3.17	0.00
HBepC_cell_total	HBepC	Human Bronchial Epithelial Cells	36.44	0.93	0.00
HBSMC_cell_total	HBSMC	Human Bronchial Smooth Muscle Cells	281.68	3.90	0.00
HCAEC_cell_total	HCAEC	Human Coronary Artery Endothelial Cells	1332.16	5.90	0.00
HCASMC_cell_total	HCASMC	Human Coronary Artery Smooth Muscle Cells	1929.37	0.43	0.00

HCH_cell_total	HCH	Undifferentiated Chondrocytes	1068.59	1.34	0.00
HCM_cell_total	HCM	Human Cardiac Myocytes	939.91	1.01	0.00
HDBEC_cell_total	HDBEC	Human Dermal Blood Endothelial Cells	5896.16	17.95	0.02
HDLEC_cell_total	HDLEC	Human Dermal Lymphatic Endothelial Cells	1031.65	9.53	0.00
HDMEC_cell_total	HDMEC	Human Dermal Microvascular Endothelial Cells	2030.59	7.00	0.00
HeLa_S3_cell_longNonPolyA	HeLa-S3	cervical carcinoma	0.00	1.24	0.00
HeLa_S3_cell_longPolyA			89.60	4.08	0.00
HeLa_S3_cytosol_longPolyA			95.21	2.90	0.00
HeLa_S3_nucleus_longNonPolyA			19.14	1.62	0.00
HeLa_S3_nucleus_longPolyA			60.50	0.51	0.00
HepG2_cell_longNonPolyA	HepG2	hepatocellular carcinoma	0.00	1.38	0.00
HepG2_cell_longPolyA			0.00	3.49	0.00
HepG2_cytosol_longNonPolyA			0.00	1.48	0.00
HepG2_cytosol_longPolyA			0.00	3.06	0.00
HepG2_nucleus_longNonPolyA			0.22	2.49	0.00
HepG2_nucleus_longPolyA			0.00	0.80	0.00
HFDPC_cell_total	HFDPC	Follicle Dermal Papilla Cells	30.68	3.01	0.00
hMSC_AT_cell_total	hMSC-AT	Undifferentiated Mesenchymal Stem Cells from subcutaneous abdomen adipose tissue	30.54	9.13	0.00
hMSC_BM_cell_total	hMSC-BM	Undifferentiated Mesenchymal Stem Cells from femoral head	7.10	7.27	0.00
hMSC_UC_cell_total	hMSC-UC	Undifferentiated Mesenchymal Stem Cells from matrix (Wharton's Jelly)	6.71	7.42	0.00
HNEpC_cell_total			93.39	4.02	0.00
HOB_cell_total	HOB	Undifferentiated Osteoblasts femoral head	422.49	6.06	0.00
HPAEC_cell_total	HPAEC	Human Pulmonary Artery Endothelial Cells	3247.85	9.84	0.00
HPASMC_cell_total	HPASMC	Human Pulmonary Artery Smooth Muscle Cells	1547.67	4.19	0.00
HPC_PL_cell_total	HPC-PL	Undifferentiated Pericytes	4.98	2.37	0.00
HPF_cell_total	HPF	Human Pulmonary Fibroblasts	258.70	8.53	0.00
HPIEpC_cell_total	HPIEpC	Placental Epithelial Cells amniotic membrane	104.99	4.18	0.00
HPMEC_cell_total	HPMEC	Human Pulmonary Microvascular Endothelial Cells	4270.35	7.02	0.00
HRCEpC_cell_total	HRCEpC	Human Renal Cortical Epithelial Cells	125.07	31.05	0.00
HSaVEC_cell_total	HSaVEC	Saphenous Vein Endothelial Cells from thigh	1336.69	11.39	0.00
HSMM_cell_longNonPolyA	HSMM	Human Skeletal Muscle Myoblast	281.80	0.38	0.00
HSMM_cell_longPolyA			1147.55	2.37	0.00

HTEpC_cell_total	HTEpC	Human Tracheal Epithelial Cells	88.08	4.24	0.00
HTSMC_cell_total	HTSMC	Human Tracheal Smooth Muscle Cells	143.50	7.33	0.00
HUAEC_cell_total	HUAEC	Human Aortic Endothelial Cells	651.03	3.35	0.00
HUASMC_cell_total	HUASMC	Human Umbilical Artery Smooth Muscle Cells	47.92	6.03	0.00
HUtSMC_cell_total	HUtSMC	Human Uterine Smooth Muscle Cells	4.04	3.22	0.00
HUVEC_cell_longNonPolyA	HUVEC	umbilical vein endothelial cells	3868.04	2.53	0.00
HUVEC_cell_longPolyA			7680.14	3.20	0.00
HUVEC_cytosol_longPolyA			7580.66	7.87	0.00
HUVEC_nucleus_longNonPolyA			970.35	2.82	0.00
HUVEC_nucleus_longPolyA			2714.06	2.84	0.00
HVMF_cell_total	HVMF	villous mesenchymal fibroblast cells	6.10	3.42	0.00
HWP_cell_total	HWP	Undifferentiated White Preadipocytes, subcutaneous adipose tissue from abdomen / upper arm	25.77	10.94	0.00
IMR90_cell_longPolyA	IMR90	fetal lung fibroblasts	192.50	6.78	0.00
IMR90_cell_total			123.90	3.79	0.00
IMR90_cytosol_longPolyA			186.03	2.40	0.00
IMR90_nucleus_longPolyA			49.86	0.36	0.00
K562_cell_longNonPolyA	K562	leukemia	0.81	1.53	0.00
K562_cell_longPolyA			4.74	4.43	0.00
K562_chromatin_total			0.37	1.23	0.00
K562_cytosol_longNonPolyA			0.00	0.46	0.00
K562_cytosol_longPolyA			1.40	1.50	0.00
K562_nucleolus_total			0.59	0.38	0.00
K562_nucleoplasm_total			0.41	0.85	0.00
K562_nucleus_longNonPolyA			0.46	0.89	0.00
K562_nucleus_longPolyA			0.28	0.36	0.00
Liver_STL004_Liver_STL011_cell_total		Liver	3.63	1.77	0.06
MCF_7_cell_longNonPolyA	MCF-7	mammary gland, adenocarcinoma	10.68	0.43	0.00
MCF_7_cell_longPolyA			14.85	14.35	0.00
MCF_7_cytosol_longPolyA			20.45	11.04	0.00
MCF_7_nucleus_longPolyA			6.19	3.88	0.03
Monocytes_CD14_Plus_cell_longNonPolyA	Monocytes-CD14+	Monocytes-CD14+ are CD14-positive cells from human leukapheresis production	0.00	6.62	0.00
Monocytes_CD14_Plus_cell_longPolyA			0.00	8.48	0.00
NHDF_cell_total	NHDF	Dermal Fibroblasts from temple / breast	8.06	9.32	0.00

NHEK_cell_longNonPolyA	NHEK	epidermal keratinocytes	95.88	4.13	0.00
NHEK_cell_longPolyA			195.39	12.39	0.00
NHEK_cytosol_longPolyA			96.90	10.77	0.00
NHEK_nucleus_longNonPolyA			26.50	4.34	0.00
NHEK_nucleus_longPolyA			132.83	7.81	0.00
NHEM_f_M2_cell_total	NHEM.f_M2	Epidermal Melanocytes from foreskin	3.77	2.27	0.00
NHEM_M2_cell_total	NHEM_M2	Epidermal Melanocytes (adult) from cheek / temple	2.84	2.66	0.28
NHLF_cell_longNonPolyA	NHLF	Normal Human Lung Fibroblasts	88.40	0.67	0.00
NHLF_cell_longPolyA			416.63	14.80	0.00
SK_N_SH_cell_longPolyA	SK-N-SH	neuroblastoma	1042.44	0.93	0.05
SK_N_SH_cytosol_longPolyA			1098.83	1.46	0.15
SK_N_SH_nucleus_longPolyA			467.35	0.26	0.16
SK_N_SH_RA_cell_longNonPolyA	SK-N-SH_RA	neuroblastoma cell line, treatment: differentiated with retinoic acid	26.23	0.29	1.70
SK_N_SH_RA_cell_longPolyA			91.36	1.96	1.89
SkMC_cell_total	SKMC	skeletal muscle cells	74.16	12.48	0.00

**RPKM**: reads per kilobase per million mapped reads

**Supplementary Table 8. Regulatory features of three newly identified disease associations and all variants in high LD ( $r^2 > 0.8$ ) with these variants.** All variants were annotated using CADD<sup>1</sup> v1.3 (GRCh37/hg19;), MutationTaster<sup>3</sup> (NCBI build 37, Ensemble 69) and HaploReg<sup>4</sup> v4.1 to determine regulatory features including chromatin state, protein binding annotation and the effect of SNPs on regulatory motifs (**Methods**).











Gene	Transcript	Region	Strand	Start	End	Score	Chrom	Strand	Start	End	Score	Chrom	Strand	Start	End	Score
rs3801805	26,897,640	Transcript	INTRONIC	68.28	24.80	36.92	1	1	44.0918	139	13991					
rs10486485	26,900,356	Transcript	INTRONIC	15.88	11.52	2.00				3849	16707					
rs3801803	26,901,740	Transcript	INTRONIC	7.24	10.28	7.12	1	1	37.5219	2465	18091					
rs73069539	26,903,661	RegulatoryFeature	REGULATORY	0.44	52.20	15.88	121.80	3	0.02	4	4	44.4919	544	2012		
rs17323934	26,904,330	RegulatoryFeature	REGULATORY	1.78	15.72	4.00	30.00	52	86	448.4156	33	20681				
H3K9Ac, Histone, Histone 3 Lysine 9 Acetylation H3K9me3, Histone, Histone 3 Lysine 9 Tri-Methylation H3K4me3, Histone, Histone 3 Lysine 4 Tri-Methylation H3K27me3, Histone, Histone 3 Lysine 27 Tri-Methylation H3K27ac, Histone, Histone 3 Lysine 27 Acetylation H2AZ, Histone, Histone 2A variant 2	H3K9me1, Histone, Histone 3 Lysine 4 Mono-Methylation H3K4me1, Histone, Histone 3 Lysine 4 Mono-Methylation H3K27me3, Histone, Histone 3 Lysine 27 Tri-Methylation	H3K9Ac, Histone, Histone 3 Lysine 9 Acetylation D2F6, Open Chromatin, DNaseI Hypersensitive Site E2F6, Transcription Factor, E2F6 Transcription Factor Binding H3K4me3, Histone, Histone 3 Lysine 4 Tri-Methylation H3K4me3, Histone, Histone 3 Lysine 4 Di-Methylation H3K27me3, Histone, Histone 3 Lysine 27 Tri-Methylation H2AZ, Histone, Histone 2A variant 2 H3K36ac, Histone, Histone 3 Lysine 27 Acetylation H3K18Ac, Histone, Histone 3 Lysine 9 Acetylation H3K27ac, Histone, Histone 3 Lysine 27 Acetylation Promoter Associated, Regulatory Feature, Promoter like regulatory feature PoliI, Polymerase, RNA Polymerase II	DNase1, Open Chromatin, DNaseI Hypersensitive Site HEY1, Transcription Factor, HEY1 Transcription Factor Binding H3K4me3, Histone, Histone 3 Lysine 4 Tri-Methylation ELF1, Transcription Factor, ELF1 Transcription Factor Binding H3K27me3, Histone, Histone 3 Lysine 27 Tri-Methylation H3K36ac, Histone, Histone 3 Lysine 36 Acetylation H3K18Ac, Histone, Histone 3 Lysine 9 Acetylation TFAP1, Transcription Factor, TFAP1 Transcription Factor Binding H2AF1, Transcription Factor, H2AF1 Transcription Factor Binding Max, Transcription Factor, Max TF binding PoliI, Polymerase, RNA Polymerase II Cos, Transcription Factor, Cos TF binding H2BK2ac, Histone, Histone 28 Lysine 5 Acetylation H2BK12ac, Histone, Histone 28 Lysine 12 Acetylation H3K9Ac, Histone, Histone 3 Lysine 9 Acetylation FAIRE, Open Chromatin, Formaldehyde-Assisted Isolation of Regulatory Elements ATF3, Transcription Factor, ATF3 Transcription Factor Binding E2F6, Transcription Factor, E2F6 Transcription Factor Binding Yy1, Transcription Factor, Yy1 Transcription Factor Binding Ap2gamma, Transcription Factor, Ap2gamma Transcription Factor Binding USF1, Transcription Factor, USF1 Transcription Factor Binding FOSL2, Transcription Factor, FOSL2 Transcription Factor Binding H3K4me3, Histone, Histone 3 Lysine 4 Tri-Methylation H3K27me3, Histone, Histone 3 Lysine 27 Tri-Methylation H2AZ, Histone, Histone 2A variant 2 H3K9Ac, Histone, Histone 3 Lysine 9 Acetylation Promoter Associated, Regulatory Feature, Promoter like regulatory feature													



#abbreviation description

## Epigenome IDs

Epigenome ID (EID)	Group	Mnemonic	Description
E017	IMR90	LNG.IMR90	IMR90 fetal lung fibroblasts Cell Line
E002	ESC	ESC.WA7	ES-WA7 Cells
E008	ESC	ESC.H9	H9 Cells
E001	ESC	ESC.I3	ES-I3 Cells
E015	ESC	ESC.HUES6	HUES6 Cells
E014	ESC	ESC.HUES48	HUES48 Cells
E016	ESC	ESC.HUES64	HUES64 Cells
E003	ESC	ESC.H1	H1 Cells
E024	ESC	ESC.4STAR	ES-UCSF4 Cells
E020	iPSC	IPSC.20B	iPS-20b Cells
E019	iPSC	IPSC.18	iPS-18 Cells
E018	iPSC	IPSC.15b	iPS-15b Cells
E021	iPSC	IPSC.DF.6.9	iPS DF 6.9 Cells
E022	iPSC	IPSC.DF.19.11	iPS DF 19.11 Cells
E007	ES-deriv	ESDR.H1.NEUR.PROG	H1 Derived Neuronal Progenitor Cultured Cells
E009	ES-deriv	ESDR.H9.NEUR.PROG	H9 Derived Neuronal Progenitor Cultured Cells
E010	ES-deriv	ESDR.H9.NEUR	H9 Derived Neuron Cultured Cells
E013	ES-deriv	ESDR.CD56.MESO	hESC Derived CD56+ Mesoderm Cultured Cells
E012	ES-deriv	ESDR.CD56.ECTO	hESC Derived CD56+ Ectoderm Cultured Cells
E011	ES-deriv	ESDR.CD184.ENDO	hESC Derived CD184+ Endoderm Cultured Cells
E004	ES-deriv	ESDR.H1.BMP4.MESO	H1 BMP4 Derived Mesendoderm Cultured Cells
E005	ES-deriv	ESDR.H1.BMP4.TROP	H1 BMP4 Derived Trophoblast Cultured Cells
E006	ES-deriv	ESDR.H1.MSC	H1 Derived Mesenchymal Stem Cells
E062	Blood & T-cell	BLD.PER.MONUC.PC	Primary mononuclear cells from peripheral blood
E034	Blood & T-cell	BLD.CD3.PPC	Primary T cells from peripheral blood
E045	Blood & T-cell	BLD.CD4.CD25I.CD127.TMEMPC	Primary T cells effector/memory enriched from peripheral blood
E033	Blood & T-cell	BLD.CD3.CPC	Primary T cells from cord blood
E044	Blood & T-cell	BLD.CD4.CD25.CD127M.TREGPC	Primary T regulatory cells from peripheral blood
E043	Blood & T-cell	BLD.CD4.CD25M.TPC	Primary T helper cells from peripheral blood
E039	Blood & T-cell	BLD.CD4.CD25M.CD45RA.NPC	Primary T helper naive cells from peripheral blood
E041	Blood & T-cell	BLD.CD4.CD25M.IL17M.PL.TPC	Primary T helper cells PMA-I stimulated
E042	Blood & T-cell	BLD.CD4.CD25M.IL17P.PL.TPC	Primary T helper 17 cells PMA-I stimulated
E040	Blood & T-cell	BLD.CD4.CD25M.CD45RO.MPC	Primary T helper memory cells from peripheral blood 1
E037	Blood & T-cell	BLD.CD4.MPC	Primary T helper memory cells from peripheral blood 2
E048	Blood & T-cell	BLD.CD8.MPC	Primary T CD8+ memory cells from peripheral blood
E038	Blood & T-cell	BLD.CD4.NPC	Primary T helper naive cells from peripheral blood
E047	Blood & T-cell	BLD.CD8.NPC	Primary T CD8+ naive cells from peripheral blood
E029	HSC & B-cell	BLD.CD14.PC	Primary monocytes from peripheral blood
E031	HSC & B-cell	BLD.CD19.CPC	Primary B cells from cord blood
E035	HSC & B-cell	BLD.CD34.PC	Primary hematopoietic stem cells
E051	HSC & B-cell	BLD.MOB.CD34.PC.M	Primary hematopoietic stem cells G-CSF-mobilized Male
E050	HSC & B-cell	BLD.MOB.CD34.PC.F	Primary hematopoietic stem cells G-CSF-mobilized Female
E036	HSC & B-cell	BLD.CD34.CC	Primary hematopoietic stem cells short term culture
E032	HSC & B-cell	BLD.CD19.PPC	Primary B cells from peripheral blood
E046	HSC & B-cell	BLD.CD56.PC	Primary Natural Killer cells from peripheral blood
E030	HSC & B-cell	BLD.CD15.PC	Primary neutrophils from peripheral blood
E026	Mesench	STRM.MRW.MSC	Bone Marrow Derived Cultured Mesenchymal Stem Cells
E049	Mesench	STRM.CHON.MRW.DR.MSC	Mesenchymal Stem Cell Derived Chondrocyte Cultured Cells
E025	Mesench	FAT.ADIP.DR.MSC	Adipose Derived Mesenchymal Stem Cell Cultured Cells
E023	Mesench	FAT.MSC.DR.ADIP	Mesenchymal Stem Cell Derived Adipocyte Cultured Cells
E052	Myosat	MUS.SAT	Muscle Satellite Cultured Cells
E055	Epithelial	SKIN.PEN.FRSK.FIB.01	Foreskin Fibroblast Primary Cells skin01
E056	Epithelial	SKIN.PEN.FRSK.FIB.02	Foreskin Fibroblast Primary Cells skin02
E059	Epithelial	SKIN.PEN.FRSK.MEL.01	Foreskin Melanocyte Primary Cells skin01
E061	Epithelial	SKIN.PEN.FRSK.MEL.03	Foreskin Melanocyte Primary Cells skin03
E057	Epithelial	SKIN.PEN.FRSK.KER.02	Foreskin Keratinocyte Primary Cells skin02
E058	Epithelial	SKIN.PEN.FRSK.KER.03	Foreskin Keratinocyte Primary Cells skin03
E028	Epithelial	BRST.HMEC.35	Breast variant Human Mammary Epithelial Cells (vHMEC)
E027	Epithelial	BRST.MYO	Breast Myoepithelial Primary Cells
E054	Neurosph	BRN.GANGEM.DR.NRSPHR	Ganglion Eminence derived primary cultured neurospheres
E053	Neurosph	BRN.CRTX.DR.NRSPHR	Cortex derived primary cultured neurospheres
E112	Thymus	THYM	Thymus
E093	Thymus	THYM.FET	Fetal Thymus



E071	Brain	BRN.HIPP.MID	Brain Hippocampus Middle
E074	Brain	BRN.SUB.NIG	Brain Substantia Nigra
E068	Brain	BRN.ANT.CAUD	Brain Anterior Caudate
E069	Brain	BRN.CING.GYR	Brain Cingulate Gyrus
E072	Brain	BRN.INF.TMP	Brain Inferior Temporal Lobe
E067	Brain	BRN.ANG.GYR	Brain Angular Gyrus
E073	Brain	BRN.DL.PRFRTL.CRTX	Brain_Dorsolateral_Prefrontal_Cortex
E070	Brain	BRN.GRM.MTRX	Brain Germinal Matrix
E082	Brain	BRN.FET.F	Fetal Brain Female
E081	Brain	BRN.FET.M	Fetal Brain Male
E063	Adipose	FAT.ADIP.NUC	Adipose Nuclei
E100	Muscle	MUS.PSOAS	Psoas Muscle
E108	Muscle	MUS.SKLT.F	Skeletal Muscle Female
E107	Muscle	MUS.SKLT.M	Skeletal Muscle Male
E089	Muscle	MUS.TRNK.FET	Fetal Muscle Trunk
E090	Muscle	MUS.LEG.FET	Fetal Muscle Leg
E083	Heart	HRT.FET	Fetal Heart
E104	Heart	HRT.ATR.R	Right Atrium
E095	Heart	HRT.VENT.L	Left Ventricle
E105	Heart	HRT.VNT.R	Right Ventricle
E065	Heart	VAS.AOR	Aorta
E078	Sm. Muscle	GI.DUO.SM.MUS	Duodenum Smooth Muscle
E076	Sm. Muscle	GI.CLN.SM.MUS	Colon Smooth Muscle
E103	Sm. Muscle	GI.RECT.SM.MUS	Rectal Smooth Muscle
E111	Sm. Muscle	GI.STMC.MUS	Stomach Smooth Muscle
E092	Digestive	GI.STMC.FET	Fetal Stomach
E085	Digestive	GI.S.INT.FET	Fetal Intestine Small
E084	Digestive	GI.L.INT.FET	Fetal Intestine Large
E109	Digestive	GI.S.INT	Small Intestine
E106	Digestive	GI.CLN.SIG	Sigmoid Colon
E075	Digestive	GI.CLN.MUC	Colonic Mucosa
E101	Digestive	GI.RECT.MUC.29	Rectal Mucosa Donor 29
E102	Digestive	GI.RECT.MUC.31	Rectal Mucosa Donor 31
E110	Digestive	GI.STMC.MUC	Stomach Mucosa
E077	Digestive	GI.DUO.MUC	Duodenum Mucosa
E079	Digestive	GI.ESO	Esophagus
E094	Digestive	GI.STMC.GAST	Gastric
E099	Other	PLCNT.AMN	Placenta Amnion
E086	Other	KID.FET	Fetal Kidney
E088	Other	LNG.FET	Fetal Lung
E097	Other	OVRV	Ovary
E087	Other	PANC.ISLT	Pancreatic Islets
E080	Other	ADRL.GLND.FET	Fetal Adrenal Gland
E091	Other	PLCNT.FET	Placenta
E066	Other	LIV.ADLT	Liver
E098	Other	PANC	Pancreas
E096	Other	LNG	Lung
E113	Other	SPLN	Spleen
E114	ENCODE2012	LNG.A549.ETOHO02.CNCR	A549 EtOH 0.02pct Lung Carcinoma Cell Line
E115	ENCODE2012	BLD.DND41.CNCR	Dnd41 TCell Leukemia Cell Line
E116	ENCODE2012	BLD.GM12878	GM12878 Lymphoblastoid Cells
E117	ENCODE2012	CRVX.HELAS3.CNCR	HeLa-S3 Cervical Carcinoma Cell Line
E118	ENCODE2012	LIV.HEPG2.CNCR	HepG2 Hepatocellular Carcinoma Cell Line
E119	ENCODE2012	BRST.HMEC	HMEC Mammary Epithelial Primary Cells
E120	ENCODE2012	MUS.HSMM	HSMM Skeletal Muscle Myoblasts Cells
E121	ENCODE2012	MUS.HSMMT	HSMM cell derived Skeletal Muscle Myotubes Cells
E122	ENCODE2012	VAS.HUVEC	HUVEC Umbilical Vein Endothelial Primary Cells
E123	ENCODE2012	BLD.K562.CNCR	K562 Leukemia Cells
E124	ENCODE2012	BLD.CD14.MONO	Monocytes-CD14+ RO01746 Primary Cells
E125	ENCODE2012	BRN.NHA	NH-A Astrocytes Primary Cells
E126	ENCODE2012	SKIN.NHDFAD	NHDF-Ad Adult Dermal Fibroblast Primary Cells
E127	ENCODE2012	SKIN.NHEK	NHEK-Epidermal Keratinocyte Primary Cells
E128	ENCODE2012	LNG.NHLF	NHLF Lung Fibroblast Primary Cells
E129	ENCODE2012	BONE.OSTEO	Osteoblast Primary Cells



**chromatin core models**

**Core 15-state model (5 marks, 127 epigenomes)**

State No	Mnemonic	Description
1	TssA	Active TSS
2	TssAFlnk	Flanking Active TSS
3	TxFlnk	Transcr. at gene 5' and 3'
4	Tx	Strong transcription
5	TxWk	Weak transcription
6	EnhG	Genic enhancers
7	Enh	Enhancers
8	ZNF/Rpts	ZNF genes & repeats
9	Het	Heterochromatin
10	TssBiv	Bivalent/Poised TSS
11	BivFlnk	Flanking Bivalent TSS/Enh
12	EnhBiv	Bivalent Enhancer
13	ReprPC	Repressed PolyComb
14	ReprPCWk	Weak Repressed PolyComb
15	Quies	Quiescent/Low

**Chromatin state model based on imputed data (25 state, 12 marks, 127 epigenomes)**

State No	Mnemonic	Description
1	TssA	Active TSS
2	PromU	Promoter Upstream TSS
3	PromD1	Promoter Downstream TSS 1
4	PromD2	Promoter Downstream TSS 2
5	Tx5'	Transcribed - 5' preferential
6	Tx	Strong transcription
7	Tx3'	Transcribed - 3' preferential
8	TxWk	Weak transcription
9	TxReg	Transcribed & regulatory (Prom/Enh)
10	TxEnh5'	Transcribed 5' preferential and Enh
11	TxEnh3'	Transcribed 3' preferential and Enh
12	TxEnhW	Transcribed and Weak Enhancer
13	EnhA1	Active Enhancer 1
14	EnhA2	Active Enhancer 2
15	EnhAF	Active Enhancer Flank
16	EnhW1	Weak Enhancer 1
17	EnhW2	Weak Enhancer 2
18	EnhAc	Primary H3K27ac possible Enhancer
19	DNase	Primary DNase
20	ZNF/Rpts	ZNF genes & repeats
21	Het	Heterochromatin
22	PromP	Poised Promoter
23	PromBiv	Bivalent Promoter
24	ReprPC	Repressed Polycomb
25	Quies	Quiescent/Low

**SNP:** SNP rs ID; **CHR:** Chromosome; **BP:** position (NCBI build 37/hg19 coordinates) of SNP marker; **AnnoType:** ‘CodingTranscript’, ‘Intergenic’, ‘MotifFeature’, ‘NonCodingTranscript’, ‘RegulatoryFeature’, ‘Transcript’; **Consequence:** ‘3PRIME\_UTR’, ‘5PRIME\_UTR’, ‘DOWNSTREAM’, ‘INTERGENIC’, ‘INTRONIC’, ‘NON\_SYNONYMOUS’, ‘SYNONYMOUS’, ‘REGULATORY’, ‘STOP\_GAINED’, ‘STOP\_LOST’, ‘SPLICE\_SITE’, ‘CANONCAL\_SPLICE’, ‘UPSTREAM’, ‘NONCODING\_CHANGE’; **EncExp:** maximum ENCODE expression value; **EncH3K27Ac:** maximum ENCODE H3K27 acetylation level; **EncH3K4Me1:** maximum ENCODE H3K4 methylation level; **EncH3K4Me3:** maximum ENCODE H3K4 trimethylation level; **tOverlapMotifs:** number of overlapping predicted transcription factor motifs; **motifDist:** Reference minus alternate allele difference in nucleotide frequency within an predicted overlapping motif; **TFBS:** number of different overlapping Chromatin immunoprecipitation (ChIP) transcription factor binding sites; **TFBSPeaks:** number of overlapping ChIP transcription factor binding site peaks summed over different cell types/tissue; **TFBSPeaksMax:** maximum value of overlapping ChIP transcription factor binding site peaks across cell types/tissue; **minDistTSS:** distance to closest transcribed sequence start (TSS); **minDistTSE:** distance to closest transcribed sequence end (TSE); **regulatory features:** regulatory features from the Ensembl Regulation database, e.g. histone modification sites, open chromatin or transcription factor binding sites); **Chromatin States:** chromatin 15-state core model learned on five core chromatin marks: shown are the ‘Epigenome ID’ and the predicted chromatin state (state number and mnemonic); **Chromatin States Imputed:** chromatin 25-state core model learned on 12 core chromatin marks: shown are the ‘Epigenome ID’ and the predicted chromatin state (state number and mnemonic); **Chromatin Marks:** shown are the ‘Epigenome ID’ and the chromatin marks H3K4me1 and H3K27ac as enhancers and H3K4me3 and H3K9ac as promoters; **Promoter histone marks:** cell type/tissue in which promoter chromatin mark was found; **Enhancer histone marks:** cell type/tissue in which enhancer chromatin mark was found; **DNase:** ‘Epigenome ID’ in which DNase hypersensitivity site was found; **Proteins bound by ChIP (ENCODE):** DNA-associated proteins identified through ChIP-sequencing by the ENCODE<sup>6</sup> project; **Regulatory motifs altered:** shown are regulatory motifs altered by the variant.

**Supplementary Table 9. Prevalence of different CVD grades in the study population (panel A-C, see Supplementary Table 1) according to the CEAP classification.**

<b>CVD grade</b>	<b>Total (n=2,269)</b>	<b>Males (n=641; 28.3%)</b>	<b>Females (n=1,628; 71.7%)</b>
C2	383 (16.9%)	85 (13.3%)	298 (18.3%)
C3	1,488 (65.6%)	378 (59.0%)	1110 (68.2%)
C4	398 (17.5%)	178 (27.8%)	220 (13.5%)

**Supplementary Table 10. Characteristics of medical history of CVD patients (panel A-C, Supplementary Table 1).**

	<b>All patients (n=2,269)</b>	<b>SD (range)</b>	<b>Males (n=641)</b>	<b>SD (range)</b>	<b>Females (n=1,628)</b>	<b>SD (range)</b>
Age at ascertainment	55.4 years	11.1 years (21-82)	55.3 years	11.6 years (22-80)	55.5 years	11.0 years (21-82)
Age at onset	32.3 years	12.0 years (5-86)	36.2 years	13.0 years (5-86)	30.8 years	11.2 years (6-72)
Duration of disease	23.1 years	12.9 years (0-75)	19.0 years	12.0 years (0-59)	24.7 years	12.9 years (0-75)
BMI	26.7	4.8 (14-66)	27.4	3.7 (20-53)	26.5	5.1 (14-66)

Given are overall and sex-specific means, standard deviations (SD) and ranges. Duration of disease was calculated as the difference between age at onset and age at ascertainment.

**Supplementary Table 11. Personal characteristics of healthy control individuals (panel A-C, Supplementary Table 1).**

	<b>Controls (n=4,374)</b>	<b>SD (range)</b>	<b>Males (n=2,167)</b>	<b>SD (range)</b>	<b>Females (n=2,207)</b>	<b>SD (range)</b>
Age at ascertainment	49.4 years	15.9 years (18-90)	50.2 years	15.6 years (18-81)	48.6 years	16.2 years (18-90)
BMI*	25.9	4.5 (17-61)	26.4	4.0 (17-61)	25.4	4.9 (17-58)

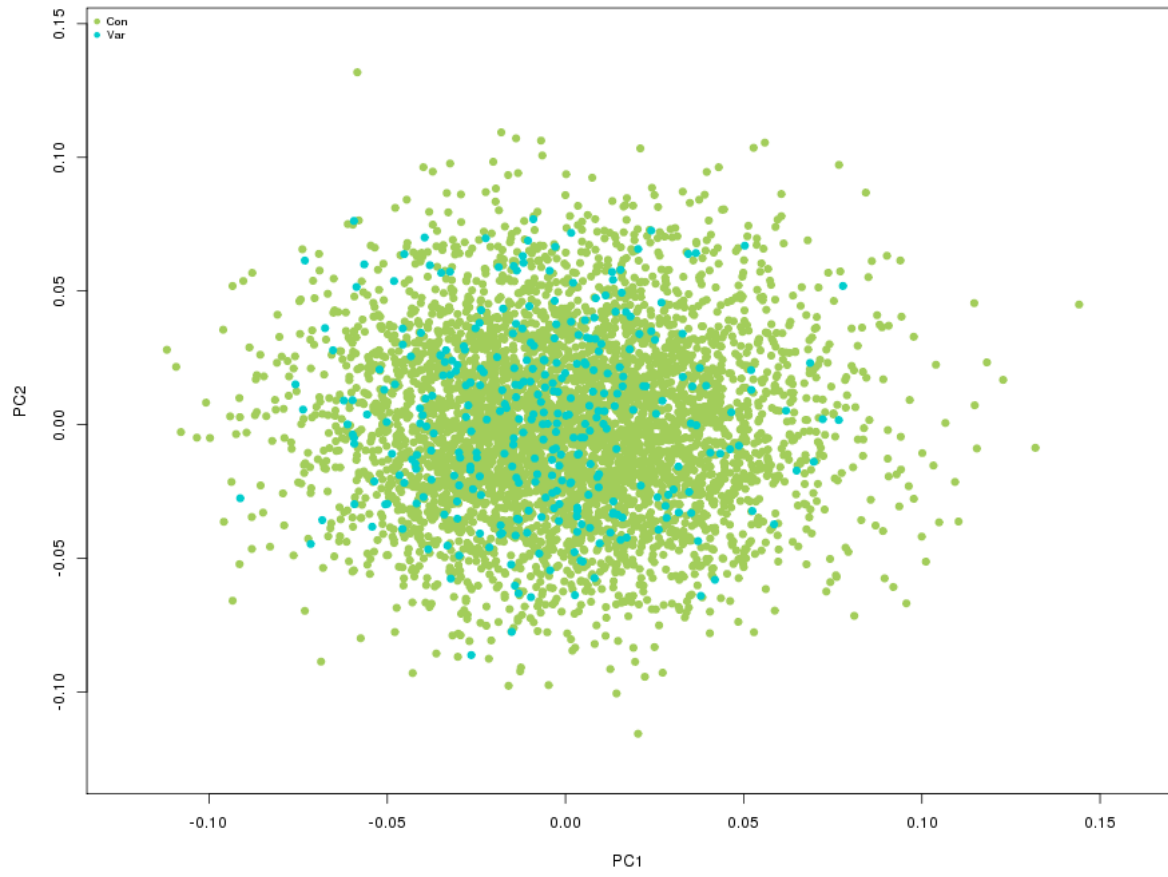
Given are the overall and sex-specific means, standard deviations (SD) and ranges. Age at ascertainment was available for all controls from the CAU Kiel and PopGen (4,374 controls; panels A-C in **Supplementary Table 1**).

\*BMI was available only for the controls of panel A and B from the CAU Kiel and PopGen (3,153 controls; 1,622 males and 1,531 females).

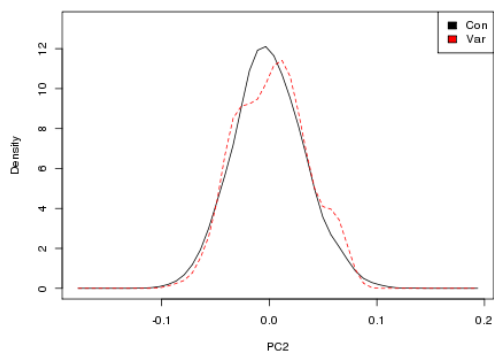
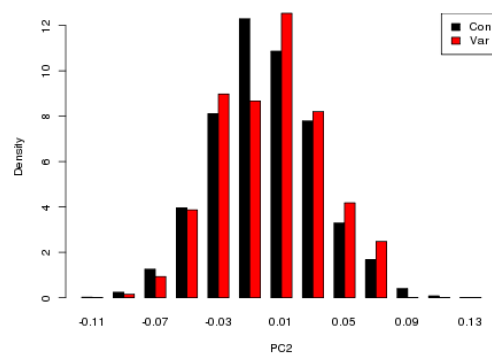
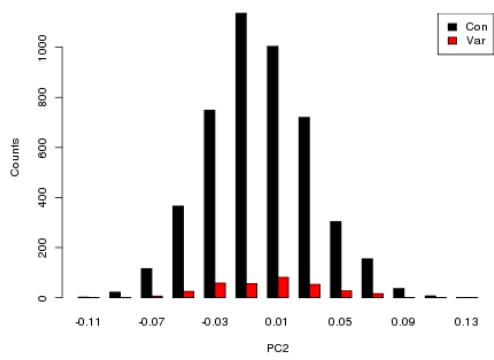
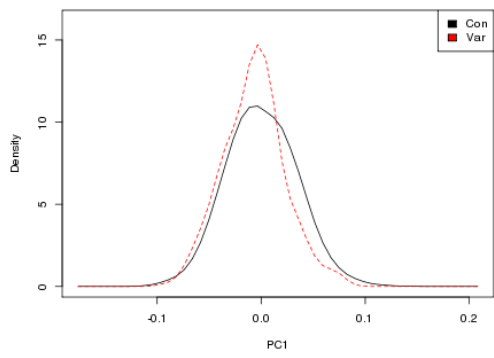
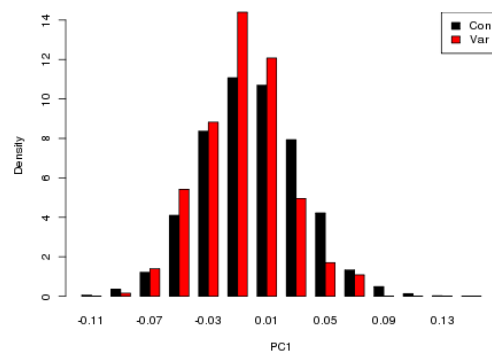
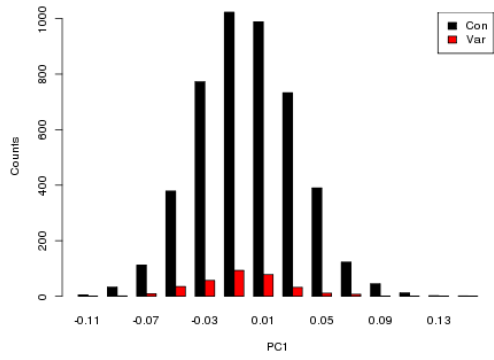
## Supplementary Figures

**Supplementary Figure 1. Principal component analysis of QCed CVD discovery panel (panel A, Supplementary Table 1).** (a) All GWAS samples are plotted on the first two principal components, colored by cases (Var: cyan) and controls (Con: green), respectively. The outlier detection procedure, as implemented in EIGENSTRAT<sup>7</sup> using default parameters ( $\sigma=6.0$ ,  $\text{maxiter}=5$ ,  $\text{topk}=10$ ), revealed no population outliers. (b) Distribution of cases and controls along the first ten principal components stratified by phenotype. (c) Principal component analysis of combined genetic data from GWAS samples and 26 reference populations from the 1000 Genomes Project (<http://www.internationalgenome.org/category/population/>). GWAS samples (grey crosses) are anchored near the CEU population, with values smaller than the median plus/minus five times the interquartile range ( $\text{median} \pm 5 \times \text{IQR}$ ; red square) of GWAS samples for the first two principal components. Super population code: AFR, African; AMR, Ad Mixed American, EAS, East Asian, EUR, European; SAS, South Asian; Population code: CHB, Han Chinese in Beijing, China; JPT, Japanese in Tokyo, Japan; CHS, Southern Han Chinese; CDX, Chinese Dai in Xishuangbanna, China; KHV, Kinh in Ho Chi Minh City, Vietnam; CEU, Utah Residents (CEPH) with Northern and Western Ancestry; TSI, Toscani in Italia; FIN, Finnish in Finland; GBR, British in England and Scotland; IBS, Iberian Population in Spain; YRI, Yoruba in Ibadan, Nigeria; LWK, Luhya in Webuye, Kenya; GWD, Gambia; MSL, Mende in Sierra Leone; ESN, Esan in Nigeria; ASW, Americans of African Ancestry in SW USA; ACB, African Caribbeans in Barbados; MXL, Mexican Ancestry from Los Angeles USA; PUR, Puerto Ricans from Puerto Rico; CLM, Colombians from Medellin, Colombia; PEL, Peruvians from Lima, Peru; GIH, Gujarati Indian from Houston, Texas; PJI, Punjabi from Lahore, Pakistan; BEB, Bengali from Bangladesh; STU, Sri Lankan Tamil from the UK; ITU, Indian Telugu from the UK.

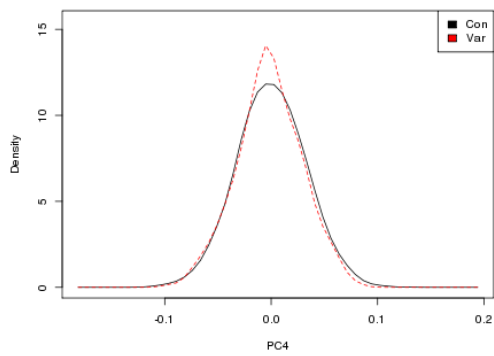
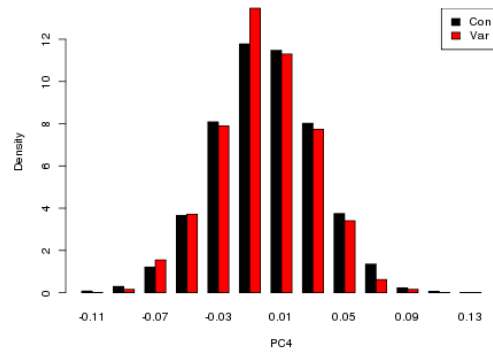
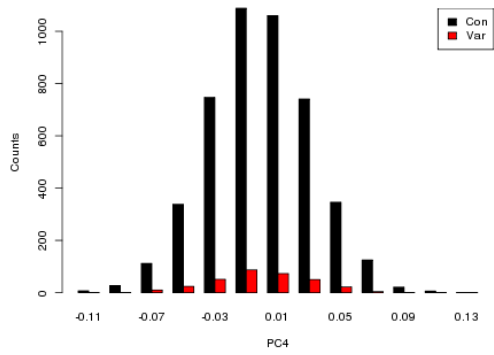
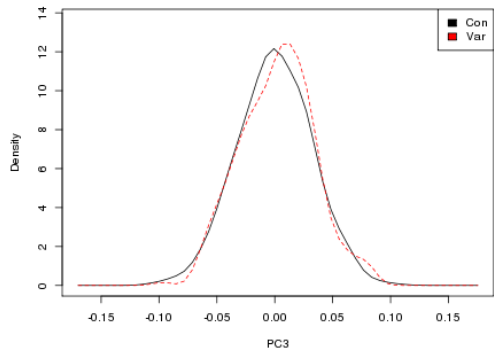
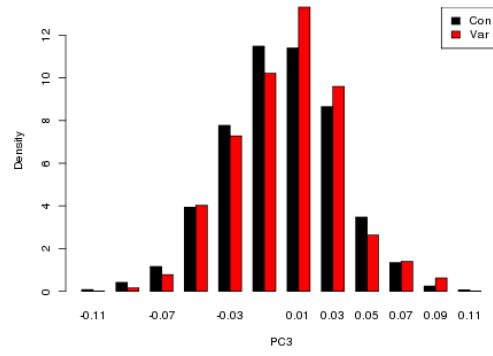
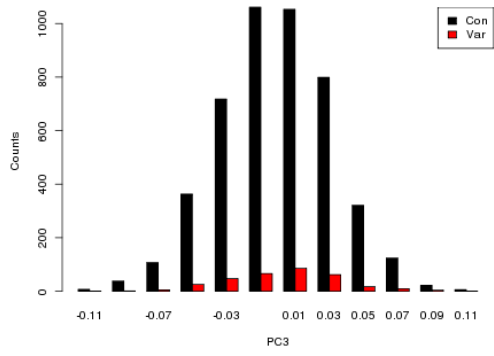
**(a)**

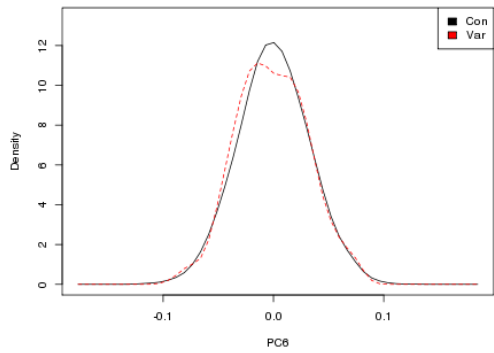
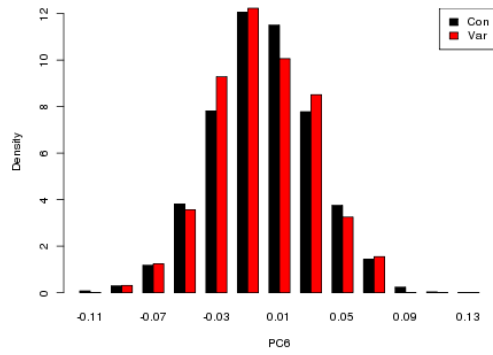
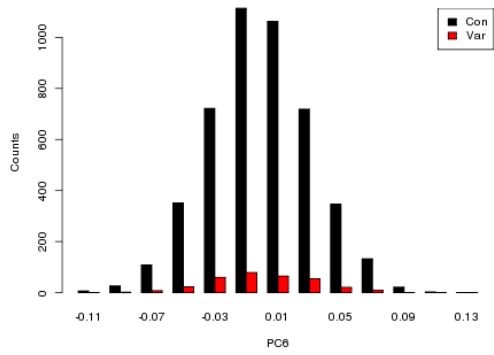
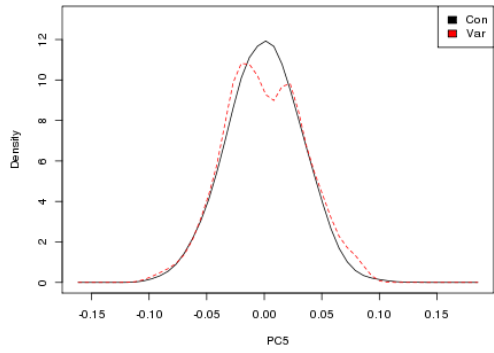
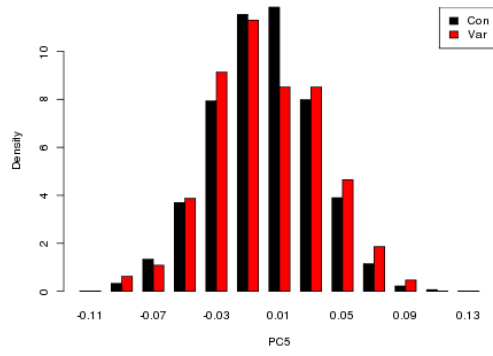
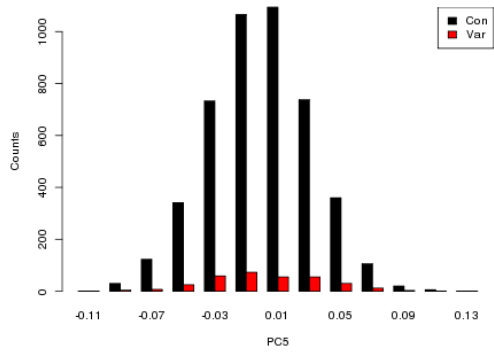


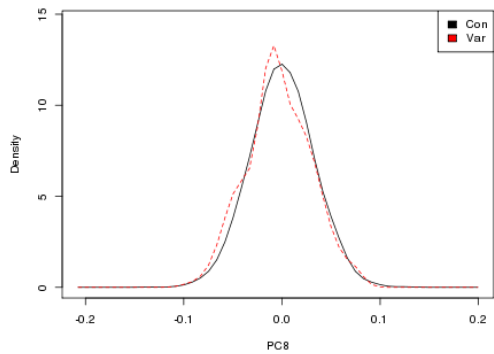
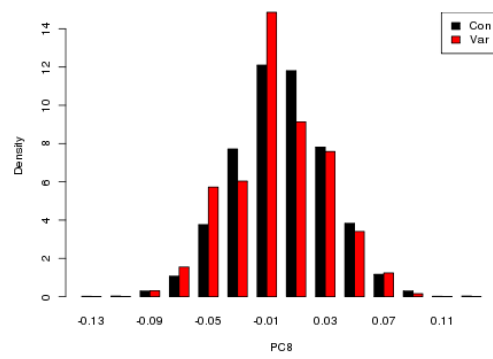
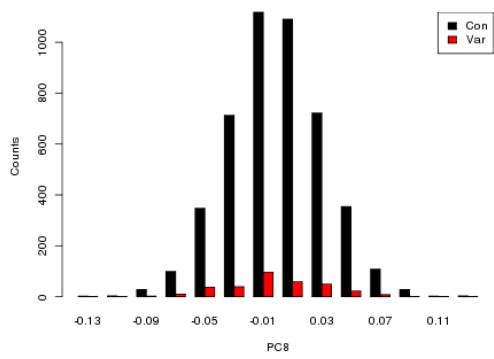
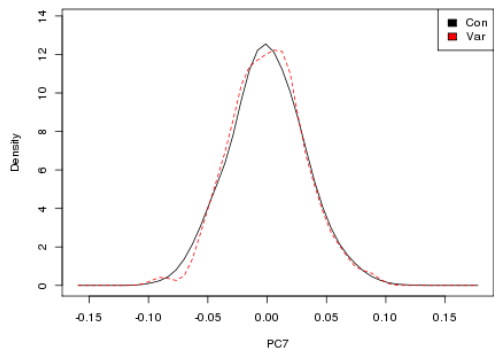
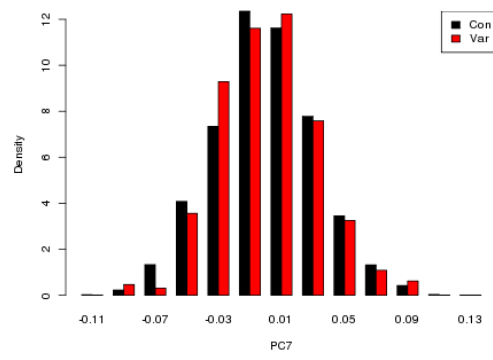
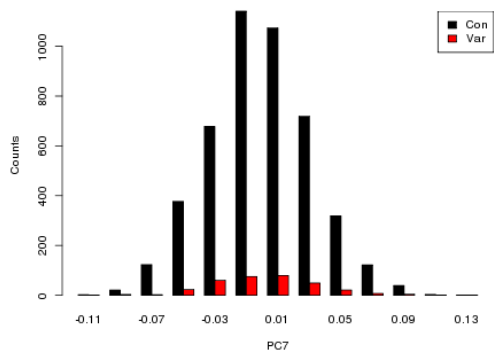
(b)

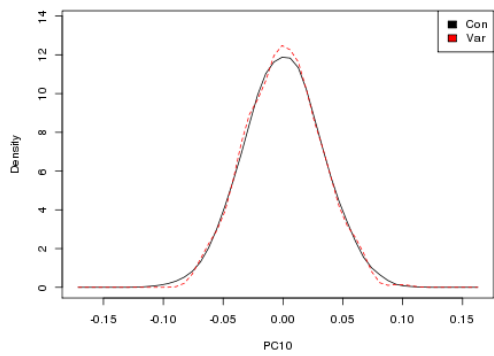
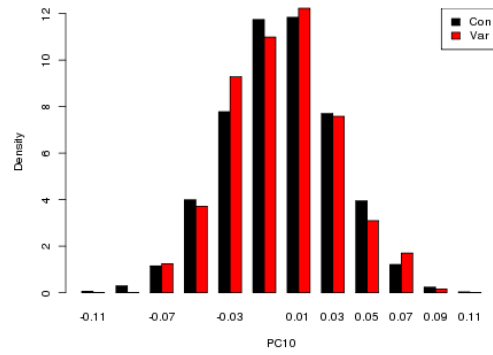
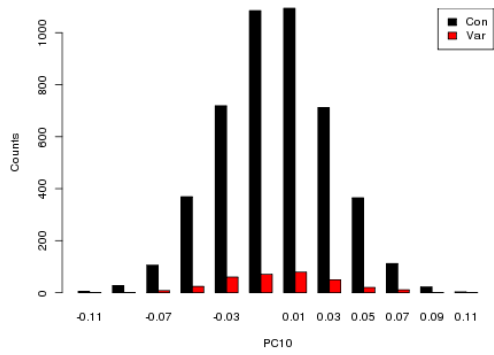
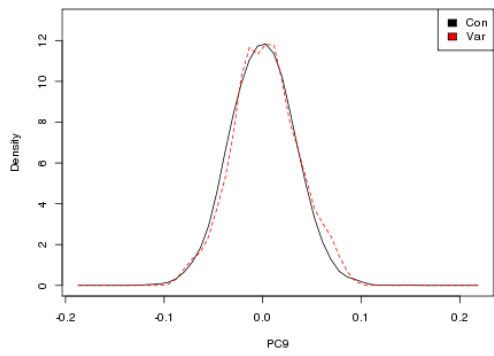
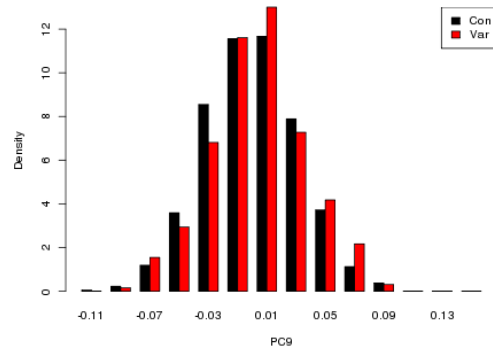
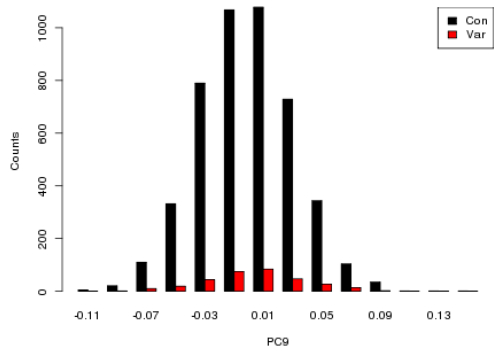




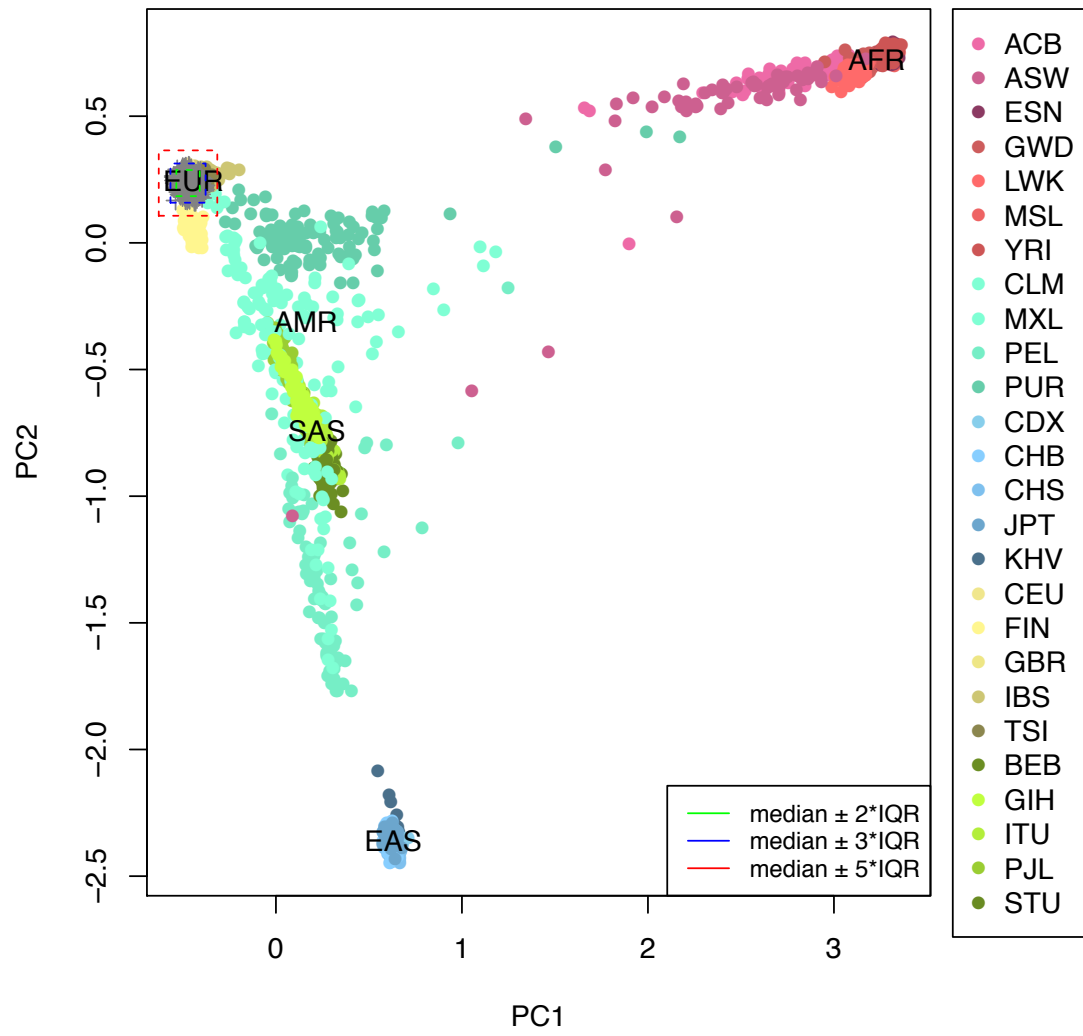




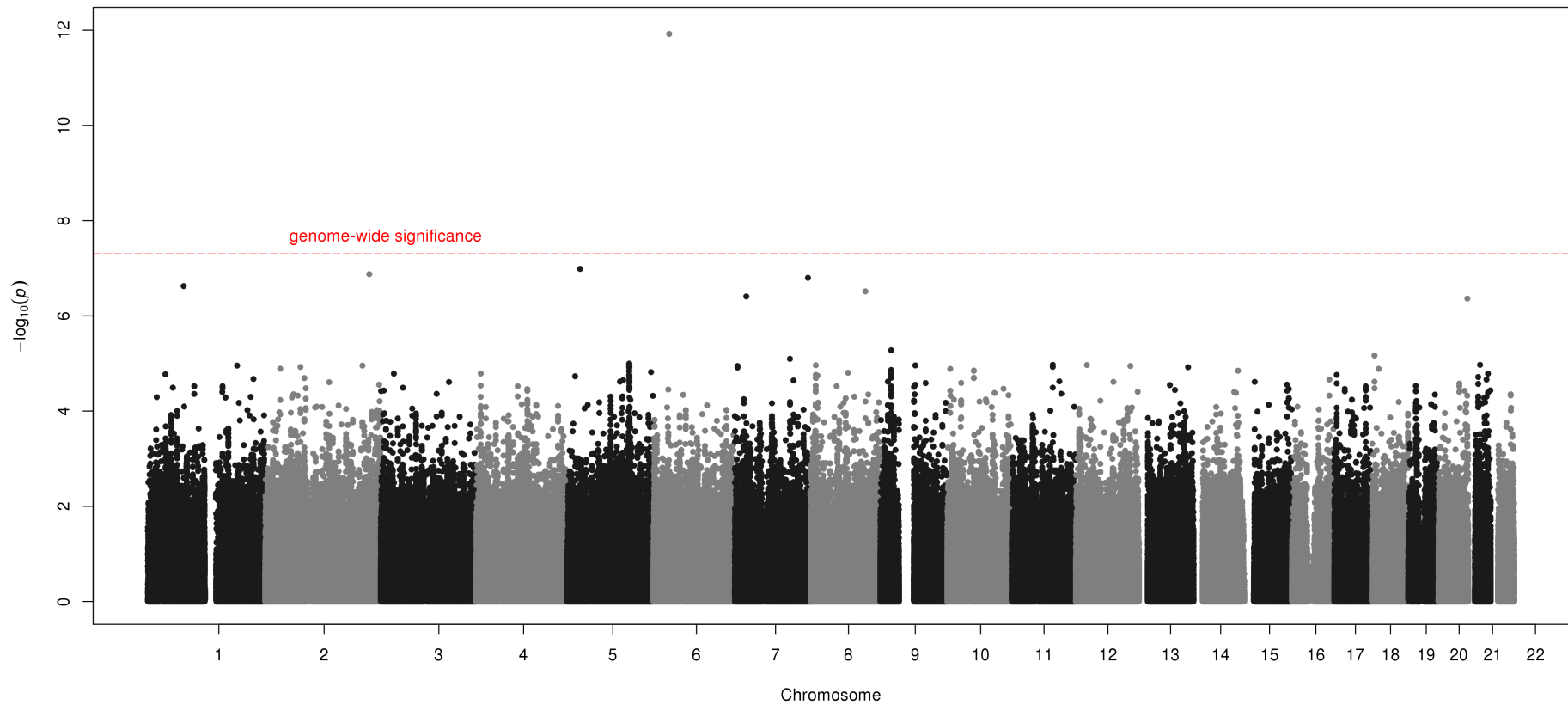




c)

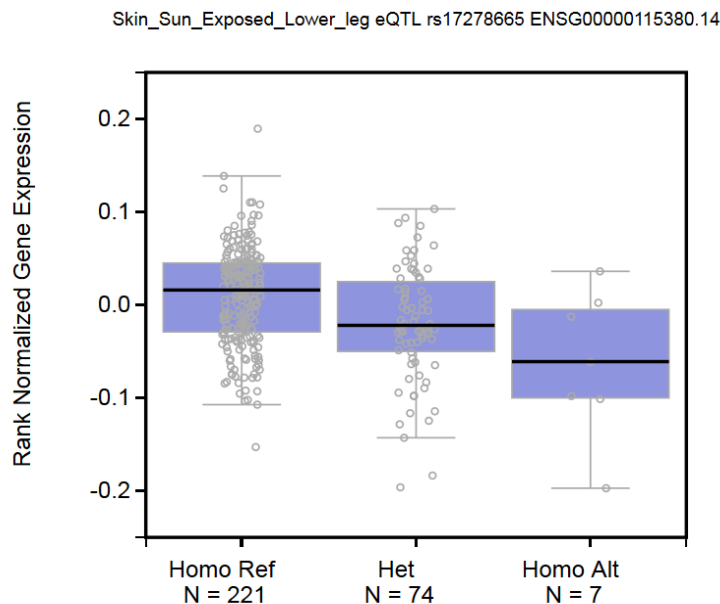


**Supplementary Figure 2. Manhattan plot of association statistics for CVD discovery panel (panel A in Supplementary Table 1).** Red horizontal line indicates the genome-wide significance threshold of  $5 \times 10^{-8}$ . The genome-wide significant locus did not replicate at  $P < 0.05$  in an independent German panel of 1,258 CVD cases and 1,925 control individuals (panel B, **Supplementary Table 1**).

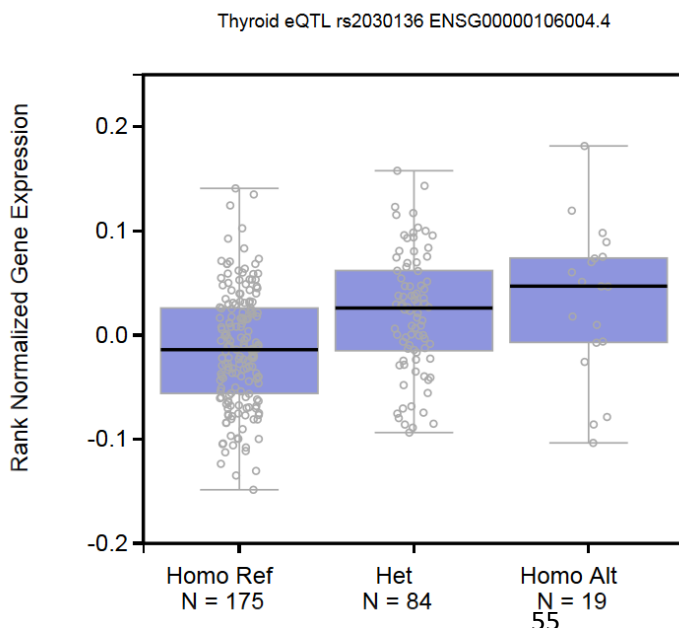


**Supplementary Figure 3. eQTL box plots generated with the GTEx<sup>8</sup> online tool and GTEx analysis release V6. (a)** SNP rs17278665 is a significant eQTL ( $FDR \leq 5\%$ ) for *EFEMP1* with decreased expression associated with the minor allele in sun exposed skin of the lower leg ( $P=2.25 \times 10^{-6}$ ).<sup>8</sup> SNP rs17278665 is also an eQTL for *EFEMP1* in lung tissue ( $P=3.45 \times 10^{-7}$ , no box plot data available from the respective study)<sup>9</sup> (**Supplementary Table 6**). **(b)** SNP rs2030136 is a significant eQTL ( $FDR \leq 5\%$ ) for *HOXA5* with increased expression associated with the minor allele in thyroid tissue ( $P=4.70 \times 10^{-7}$ ).<sup>8</sup> SNP rs2030136 is also an eQTL for *SKAP2* in lymphoblastoid cell lines ( $P=8.30 \times 10^{-9}$ , no box plot data available from the respective study)<sup>10</sup> and whole blood ( $P=4.99 \times 10^{-141}$ , no box plot data available from the respective study)<sup>11</sup> (**Supplementary Table 6**).

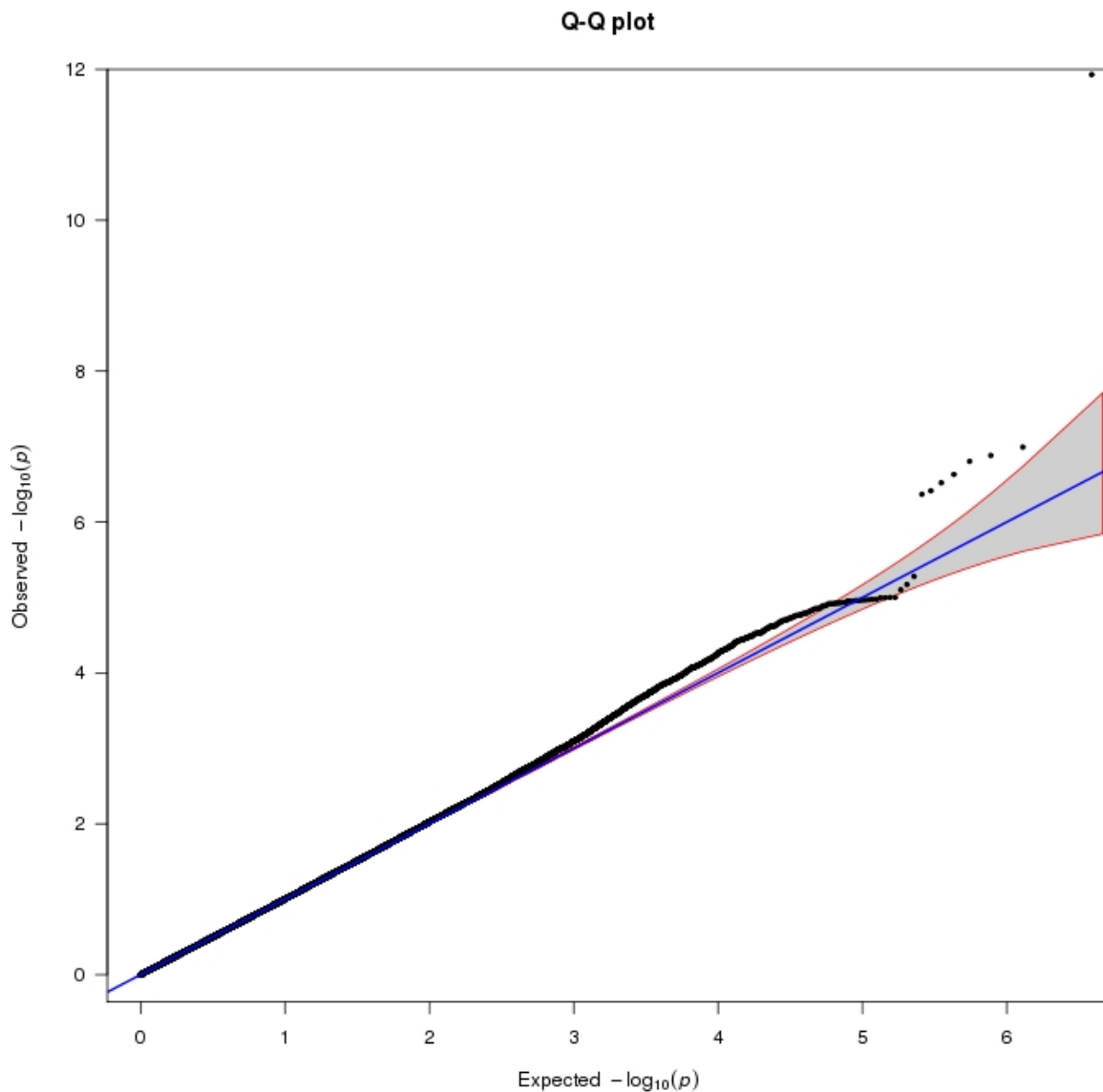
**(a)**



**(b)**



**Supplementary Figure 4. Quantile-quantile (Q-Q) plot for the CVD discovery panel (panel A, Supplementary Table 1).** Only markers that passed the quality criteria were used for plotting. The 2.5th and 97.5th centiles of the distribution under random sampling and the null hypothesis form the 95% concentration band. The genomic inflation factor  $\lambda$  is defined as the ratio of the medians of the sample  $\chi^2$  test statistics and the 1-d.f.  $\chi^2$  distribution  $(0.455)^{12}$ . Since  $\lambda$  inferred from 1,934,349 SNPs showed moderate inflation ( $\lambda=1.156$ ), genomic control correction was applied to results by correction of  $\chi^2$  test statistics using factor  $\lambda$  and multiplying SNP standard errors by the square root of the inflation factor, thus minimizing artificial differences in allele frequencies due to different genotyping platforms.





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