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Genome-Wide Association Study to Find Modifiers for Tetralogy of Fallot in the 22q11.2 Deletion Syndrome Identifies Variants in the *GPR98* Locus on 5q14.3

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Background—The 22q11.2 deletion syndrome (22q11.2DS; DiGeorge syndrome/velocardiofacial syndrome) occurs in 1 of 4000 live births, and 60% to 70% of affected individuals have congenital heart disease, ranging from mild to severe. In our cohort of 1472 subjects with 22q11.2DS, a total of 62% (n=906) have congenital heart disease and 36% (n=326) of these have tetralogy of Fallot (TOF), comprising the largest subset of severe congenital heart disease in the cohort.

Methods and Results—To identify common genetic variants associated with TOF in individuals with 22q11.2DS, we performed a genome-wide association study using Affymetrix 6.0 array and imputed genotype data. In our cohort, TOF was significantly associated with a genotyped single-nucleotide polymorphism (rs12519770, P=2.98×10^{-8}) in an intron of the adhesion *GPR98* (G-protein–coupled receptor V1) gene on chromosome 5q14.3. There was also suggestive evidence of association between TOF and several additional single-nucleotide polymorphisms in this region. Some genome-wide significant loci in introns or noncoding regions could affect regulation of genes nearby or at a distance. On the basis of this possibility, we examined existing Hi-C chromatin conformation data to identify genes that might be under shared transcriptional regulation within the region on 5q14.3. There are 6 genes in a topologically associated domain of chromatin with *GPR98*, including *MEF2C* (Myocyte-specific enhancer factor 2C). *MEF2C* is the only gene that is known to affect heart development in mammals and might be of interest with respect to 22q11.2DS.

Conclusions—In conclusion, common variants may contribute to TOF in 22q11.2DS and may function in cardiac outflow tract development. (***Circ Cardiovasc Genet*. 2017;10:e. DOI: 10.1161/CIRCGENETICS.116.001690.)**

Key Words: chromosomes ■ DiGeorge syndrome ■ genotype ■ ivelo-cardio-facial syndrome ■ tetralogy of Fallot

One of the greatest challenges in the area of human genetics is to understand the basis of phenotypic heterogeneity in known diseases. The 22q11.2 deletion syndrome (22q11.2DS; velocardiofacial syndrome/DiGeorge syndrome; Mendelian Inheritance in Man No. 192430, 188400) is one of the most common genomic disorders, occurring in 1 of 4000 live births.[1] Over 90% of affected individuals have a de novo, hemizygous 3 million base pair (Mb) deletion on chromosome 22q11.2.[2–4] All subjects with the deletion have features of the syndrome,

See Editorial by McBride and Ware
See Clinical Perspective

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*A list of the International 22q11.2 Consortium and IBBC is listed in Table 1 in the Data Supplement.

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but the clinical presentation is quite variable. For example, 60% to 70% of patients have congenital heart disease (CHD) involving the cardiac outflow tract (OFT) and aortic arch, whereas the rest have apparently normal cardiac structures.[5] Among the most serious defect observed in individuals with the 22q11.2DS is tetralogy of Fallot (TOF), which is defined by the presence of a ventricular septal defect, pulmonary stenosis, overriding aorta, and right ventricular hypertrophy. TOF is caused in part by failed migration or differentiation of second heart field mesodermal cells from the pharyngeal apparatus in embryos, needed to form or remodel the cardiac OFT.[6] TOF occurs in 1 of 2500 live births in the general population (Center for Disease Control and Prevention). Among individuals with the 22q11.2DS, 36% in our cohort has TOF, and among individuals with TOF, ≈15% have a 22q11.2 deletion.[7,8]

Among the genes in the deleted region on 22q11.2, *TBX1*, which encodes a T-box transcription factor, is the major candidate for CHD.[9–11] *Tbx1* is expressed in the second heart field mesoderm, which is disrupted in 22q11.2DS.[9–11] Global inactivation[9–11] or second heart field–specific inactivation[12] of *Tbx1* results in neonatal lethality with severe cardiac OFT defects. One hypothesis to explain variable phenotypic expression in the 22q11.2DS is the presence of pathogenic variants in *TBX1* on the haploid allele of 22q11.2. Previously, we tested whether common or rare single-nucleotide variants (SNVs) in the coding region of *TBX1* on the remaining allele of 22q11.2 were associated with CHD in 22q11.2DS subjects, but we did not find an association.[13] Another hypothesis is that there are copy number variations elsewhere in the genome that could explain differences in phenotypes. We previously found that a commonly occurring genomic duplication encompassing the glucose transporter gene, *SLC2A3*, was associated with CHD (P=2.68×10^{-4}).[14] This copy number variation occurred in 5.8% of individuals with 22q11.2DS and CHD and 1.1% of those with 22q11.2DS and no CHD. Recently, a partial duplication of a chromatin modifier, *KANSL1*, was associated with CHD in a Chilean 22q11.2DS cohort.[15] However, these copy number variations occurred in only some deleted subjects with CHD and thus do not explain the basis of phenotypic variability in the majority of patients.

Our goal was to identify common single-nucleotide polymorphisms (SNPs) that are associated with TOF in individuals with 22q11.2DS. We restricted our analyses to TOF because it is the largest single phenotypic category of severe CHD in our cohort. Restricting our analyses in this way may reduce heterogeneity in the genes that contribute to CHD in individuals with 22q11.2DS and thus may increase the power of a GWAS.

Methods

Human Subjects and Phenotype Data

We assembled a cohort of subjects with 22q11.2DS (Tables I and II in the Data Supplement). Subjects were previously recruited by the International Chromosome 22q11.2DS Consortium, the International 22q11.2 Brain Behavior Consortium (http://22q11-ibbc.org), and clinical groups that specialize in the treatment of individuals with 22q11.2DS. All subjects within the cohort had a clinical diagnosis of 22q11.2DS that was confirmed by the presence of a 22q11.2 deletion using fluorescence in situ hybridization or multiplex ligation-dependent probe amplification (SALSA MLPA kit P250 DiGeorge; MRC Holland, The Netherlands). Informed consent was obtained for all participants, and this study was conducted under an Internal Review Board-approved protocol at the Albert Einstein College of Medicine (CCI 1999-201).

For this study, we used previously collected genomic DNA and phenotypic and demographic information. We obtained echocardiogram and cardiology reports to confirm the specific CHD diagnosis (eg, TOF).

SNP Array Genotype and Data Quality Control

Genomic DNA from 1244 study subjects was array genotyped using Affymetrix GeneChip Genome-Wide SNP 6.0 array. The majority of samples were genotyped was at the Genomics Facility core laboratory of Albert Einstein College of Medicine. However, 37 samples were genotyped in the Advanced Genomics laboratory core at the Children's Research Institute (Milwaukee, WI) for clinical purposes,[16] and 191 Chilean samples were genotyped in the Center for Human Genetics, Clínica Alemana Universidad del Desarrollo, Santiago, Chile.[15]

The raw data from all arrays were processed through the same pipeline using the same criteria. Genotype data from arrays with contrast quality control scores ≤0.4 per sample, contrast quality control <1.7 per batch, and Median Absolute Pairwise Difference metric >0.35, were excluded. Genotypes were called using the Birdseed V2 Genotyping Algorithm (call rate: 99.02±0.02%). To account for batch effects, BEAGLECALL Version 1.0.1 software was used to rescore genotypes.[17] SNPs with call rates <95%, minor allele frequency <1%, or Hardy–Weinberg equilibrium P value <10^{-5} were excluded. In addition, samples that showed second-degree relatedness or closer, based on identity by state, were removed. For each subject, deletion size was determined using the $\log_2$ intensity ratio as estimated by the Copy Number Analysis Module of Golden Helix Powerseat Package. The CEL files and genotype data are being deposited to National Center for Biotechnology Information database of Genotypes and Phenotypes phs001339.v1.p1.

We performed imputation to increase the number of SNPs available for analysis. Only genotyped SNPs with minor allele frequency >1% were used for imputation. Haplotypes were prephased using SHAPEIT software,[18,19] and imputation was performed using IMPUTE2 with the 1000 Genomes Phase I data set as the reference panel.[20] Imputed SNPs with minor allele frequency ≤1% or imputation quality (INFO) scores ≤0.8 were excluded from the GWAS.

Statistical Methods

We used a case–control approach, in which individuals with 22q11.2DS and TOF were considered cases and individuals with 22q11.2DS without CHD were considered controls. We conducted principal component analyses to identify the PCs of race/ethnicity. Potential associations between TOF, sex, and deletion size were assessed using logistic regression adjusted for the first 4 PCs. A P value <0.05 was considered significant.

The association between TOF and each SNP was assessed by logistic regression analysis under an additive genetic model using data from all study subjects. These analyses were performed using SNPTEST v2.5.2 https://mathgen.stats.ox.ac.uk/genetics_software/snptest/snptest.html) and accounted for the genotyping accuracy and first 4 PCs of race/ethnicity.[21,22] A P value of 5×10^{-8} was used as the genome-wide significance cutoff for single association tests. For a meta-analysis, the cohort was split into groups determined by principal component analyses. Each group was analyzed separately using logistic regression, and the results were meta-analyzed using the inverse-variance method. Power for these analyses was assessed using QUANTO (http://biostats.usc.edu/Quanto.html). Manhattan plots and quantile–quantile (Q–Q) plots were generated using Golden Helix Powerseat. For regions of interest identified in the GWAS, regional association plots were generated using LocusZoom software (http://locuszoom.sph.umich.edu/locuszoom/).[23]

Conditional logistic regression analyses were performed to determine whether multiple variants within a region are independently associated with TOF in individuals with 22q11.2DS. Specifically, within a region, we conditioned on the genotyped SNP with the

lowest *P* value and the first 4 principal components of race/ethnicity and individually evaluated the association of TOF with each additional SNP in the region. Conditional analyses were conducted using SNPTEST v2.5.2.

Linkage Disequilibrium Analysis of Whole-Genome Sequence to Identify Variants in Linkage Disequilibrium With GWAS Findings

Whole-genome sequencing of a subset (n=397) of our 22q11.2DS samples was performed using the Illumina HiSeq2000 and HiSeq X Ten platform at Hudson Alpha Institute for Biotechnology (Huntsville, Alabama) as part of the International 22q11.2 Brain and Behavior Consortium to find genes for schizophrenia. Variant calling was performed using PEMapper software for read mapping to the hg38 (GRCh38) reference genome and PECaller software for variant calling.[24] CrossMap (http://crossmap.sourceforge.net/) was used to convert genome coordinates between hg38(GRCh38) and hg19 (GRCh37).[25] To follow-up on the top result from the GWAS (Results), the genomic region (chromosome 5 [chr5]:88 703 723–91 409 593) from *MEF2C* through *ARRDC3* was extracted from the whole-genome sequencing data. Functional annotation of SNVs was performed using the Variant Classification and the Annotate and Filter tools in the Golden Helix software. Nonexonic SNVs (intronic, intergenic) were removed, and predicted functional SNVs were used to generate an linkage disequilibrium (LD) matrix using Haploview. LD measurements of r^2 >0.8 were used to define LD haplotype blocks.

Mouse Embryo Analysis and Whole-Mount RNA In Situ Hybridization

Gene expression profiling was previously performed to identify differentially expressed genes in the second heart field mesoderm of wild-type mouse embryos.[12] Data from wild-type embryos were extracted for evaluation of specific expression levels in this tissue. For in situ hybridization, RNA probes were generated from mouse embryo cDNA using digoxigenin-uridine triphosphate (Roche Diagnostic Corp, Indianapolis, IN; Table III in the Data Supplement). Swiss Webster strain wild-type mouse embryos were isolated at day (E)9.5 or E10.5 and used for experiments.

Results

Description of the 22q11.2DS Population

A total of 1472 unrelated subjects with 22q11.2DS were ascertained through multiple sources (Tables I and II in the Data Supplement) and were genotyped on Affymetrix 6.0 arrays. Genotypes (n≈6.6 million SNPs with minor allele frequency >0.01 and INFO>0.8) were also imputed.

The characteristics of the study subjects are shown in Table 1. These subjects predominantly self-reported as white and non-Hispanic (Table 1). This was confirmed by principal component analyses using 4 PCs (we did not observe a difference between usage of 4 or 10 PCs) to estimate genetic ancestry (Figure I in the Data Supplement). The majority (191/217) of Hispanic subjects were recruited at the collection site in Santiago, Chile. Approximately 62% of the subjects (n=906) had CHD at birth. There are 4 sets of low copy repeats, termed LCR22A, B, C, and D, that span the 22q11.2 region. All of the subjects had a deletion of 1 allele of *TBX1*, which is located in the LCR22A-B interval, and the majority of subjects had the typical 3 Mb deletion flanked by LCR22A-D (Table 1; Table IV in the Data Supplement). In this cohort, neither sex nor deletion size were significantly associated with CHD in general or with TOF specifically (*P*>0.05).

GWAS to Identify Genetic Loci for TOF

The TOF phenotype comprised the largest individual group of subjects with severe intracardiac anomalies in our 22q11.2DS population (36%; Table 1; Figure 1). We conducted a GWAS to identify genetic variants associated with TOF. This analysis was based on data from 326 subjects with 22q11.2DS and TOF and 566 subjects with 22q11.2DS and normal cardiac anatomy. This study had a power of 80% to detect an odds ratio of >1.9 for a common SNP with an allele frequency >0.3 under a log-additive model at $P < 5\times10^{-8}$.

Subjects of all races and ethnicities were included, and associations were assessed using logistic regression adjusted with the first 4 PCs of race/ethnicity. As neither deletion size nor sex was significantly associated with TOF in this cohort, these variables were not included in the logistic models. The genomic inflation factor (λ=1.02) and the Q–Q plot (Figure II in the Data Supplement) provided little evidence of a systematic deviation from the expected distribution of the test statistic.

Three SNPs mapping to intron 61 of *GPR98* (G-protein–coupled receptor 98) were significantly associated with TOF. The genotyped SNP, rs12519770 ($P=2.98\times10^{-8}$),

Table 1. Characteristics of Study Subjects

Subject Characteristics	n (%)
Self-reported race	
White	1229 (83)
Black	34 (2)
Asian	11 (0.7)
Native American	1 (<0.1)
Mixed Ancestry	20 (1)
Unknown	177 (12)
Self-reported ethnicity	
Non-Hispanic	1255 (85)
Hispanic	217 (15)
Sex	
Male	717 (49)
Female	754 (51)
Unknown	1 (<0.1)
Congenital heart defect	
TOF	326 (22)
Other CHD	580 (39)
Normal (controls)	566 (38)
Deletion size	
Typical 3 Mb (LCR22A-D)	1366 (93)
Nested LCR22A-B	73 (5)
Nested LRC22A-C	26 (2)
Other	12 (1)

The cohort of 1472 samples is shown categorized based on self-reported race, ethnicity, and sex. The numbers and percentages in each category are indicated. A total of 906 subjects have congenital heart disease (CHD), whereas the rest have normal structures. The deletion sizes are indicated. LCR indicates low copy repeats; and TOF, tetralogy of Fallot.

and imputed SNPs, rs7720206 (P=2.22×10^{-8}) and chr5: 90 067 043:D (P=2.10×10^{-8}), showed the strongest association (Figure 2A, Table 2). These three SNPs seem to be in complete LD (Figure 2B and 2C). For rs12519770, the A allele was the risk allele with a frequency of 0.58 in TOF cases and 0.45 in controls, conferring an odds ratio of 1.69 (P=3.2×10^{-8}) per copy of the A allele in the 22q11.2DS cohort (Table 2; Figure 2). There was also suggestive evidence of association between TOF and 2 additional groups of SNPs in *GPR98*. The top genotyped SNP in each cluster (rs6889138, rs6893710) is listed in Table 2 and illustrated in Figure 2B and 2C.

Although our initial GWAS adjusted for the first 4 PCs of race/ethnicity, the observed associations may still reflect bias because of uncontrolled confounding resulting from population stratification. Consequently, we repeated our analyses for the top SNP, rs12519770, after separating the cohort into 3 groups: white, Admixed, and African, as determined by principal component analyses (Figure I in the Data Supplement). The P value for this SNP was significant in the meta-analysis (P=4.43×10^{-8}; Table V in the Data Supplement), suggesting that the observed association is unlikely to be the result of population stratification.

Within the 5q14.3 region, there seemed to be 3 clusters of SNPs that were associated with TOF. We refer to these as clusters 1, 2, and 3, and the clusters are ranked in ascending order based on the P value for the top SNP within the cluster. To determine whether >1 variant was independently associated with TOF, we performed conditional analyses in which we conditioned on the genotyped SNP in *GPR98* with the smallest P value in cluster 1 (rs12519770; P=2.98×10^{-8}) and individually evaluated the association of TOF with each of the additional SNPs in the 5q14.3 region (n=1344 SNPs, Table VI in the Data Supplement). In these conditional analyses, there was suggestive evidence for association with 1 SNP (rs6893710, P=3.92x10^{-5}). This variant was the top SNP in the third cluster of associated genes (Table 2). The association of the top SNP in the second cluster (rs6889138) was attenuated in the conditional analysis (P=0.002).

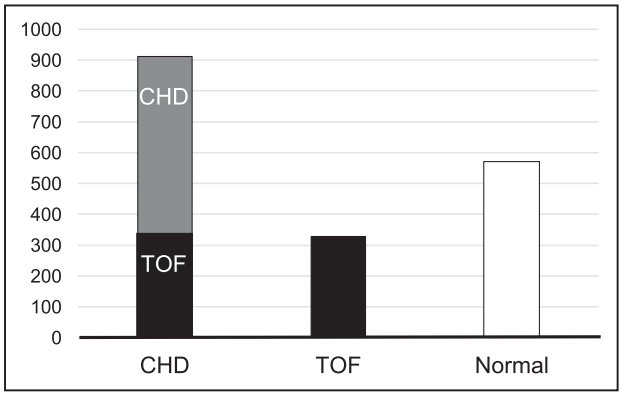

Figure 1. Distribution of cardiovascular phenotypes in 1472, 22q11.2 deletion syndrome (22q11.2DS) subjects. The number of subjects (*y* axis) sorted into phenotypes (*x* axis) is shown in the bar graph. All individuals have a hemizygous 22q11.2 deletion. The most serious cardiovascular diagnoses with the largest number of subjects is tetralogy of Fallot (TOF; n=326; black bar) among a total with congenital heart disease (CHD; n=906; gray bar) when compared with those with no intracardiac or aortic arch anomalies as detected by echocardiogram summary and cardiology report (white bar).

Definition of the 5q14.3 Locus

To identify nonsynonymous variants that may be in LD with the rs12519770 and to narrow the region containing the association signal on 5q14.3 based on LD, we performed an LD analysis using existing whole-genome sequence data from 397 individuals with 22q11.2DS (http://22q11-ibbc.org; unpublished data, International 22q11.2 Brain and Behavior Consortium authors in Supplementary Table 1, 2017). These individuals comprise a subset of the samples genotyped on Affymetrix 6.0 arrays and were selected based on psychiatric but not cardiovascular phenotype. There were 9680 SNVs identified in the 2.7 Mb region around *GPR98* (chr5: 8 799 640–90 704 983). There were 161 coding SNVs (102 nonsynonymous, 58 synonymous, and 1 splicing) and 115 SNVs in the 3′ or 5′-untranslated regions for a total of 276 SNVs (Table VII in the Data Supplement). None of these SNVs were in LD with rs12519770. The SNP, rs6893710, was in weak LD with the synonymous variant, rs41304884 (*GPR98*, NM_032119.3, c.16164G>A;D′=0.859, r^2=0.389; Figure 2B). There were no nonsynonymous variants related to our association signal.

Most GWAS signals that have been previously discovered are in intergenic regions and may mark transcriptional regulatory regions in the genome rather than genes themselves. To test this for TOF in 22q11.2DS, we examined the LD pattern from available whole-genome sequencing data on the same 397 22q11.2DS subjects to narrow the interval with SNPs showing the strongest association. We narrowed down the association signal to a 104.7 kb region on chromosome 5 (chr5: 90 057 563–90 162 285; Figure 2C and 2D, red block). Most of the common SNPs with P values <10^{-5} were located in this region (Figure 2C). A similar LD pattern has been observed in the white subset from the 1000 Genomes Project (Figure III in the Data Supplement). Thus, we were able to narrow the region of the association signal.

Genes Mapping to 5q14.3

Because the SNPs found within the intron of *GPR98* might affect its regulation or, instead, the regulation of other genes in the region, we examined local chromosome conformation forming topologically associated domains (TADs).[26–28] To investigate higher-order chromatin-mediated looping, we extracted data for the 5q14.3 interval from the Hi-C browser.[29] There was chromatin interaction data for 28 different cell lines ranging from H1 embryonic stem cells to cancer cell lines.[29–31] We focused on TAD contact domains in a 3.3 Mb region including *GPR98* (Figure 3; Figure IV in the Data Supplement). Because the 104.7 kb region with genetic association is within the *GPR98* locus, it is possible that variants might affect its expression or that of nearby genes. In addition to *GPR98*, this region includes 6 additional genes (Figure 3; Figure IV in the Data Supplement), including 5 protein coding genes within the 2.3 Mb TAD: *MEF2C* (Myocyte enhancer factor 2C), *CETN3* (Centrin 3), *MBLAC2* (Metallo-β-lactamase domain containing 2), *POLR3G* (RNA polymerase III subunit G), and *LYSMD3* (LysM, putative peptidoglycan-binding, domain containing 3). One gene, *ARRDC3* (Arrestin domain

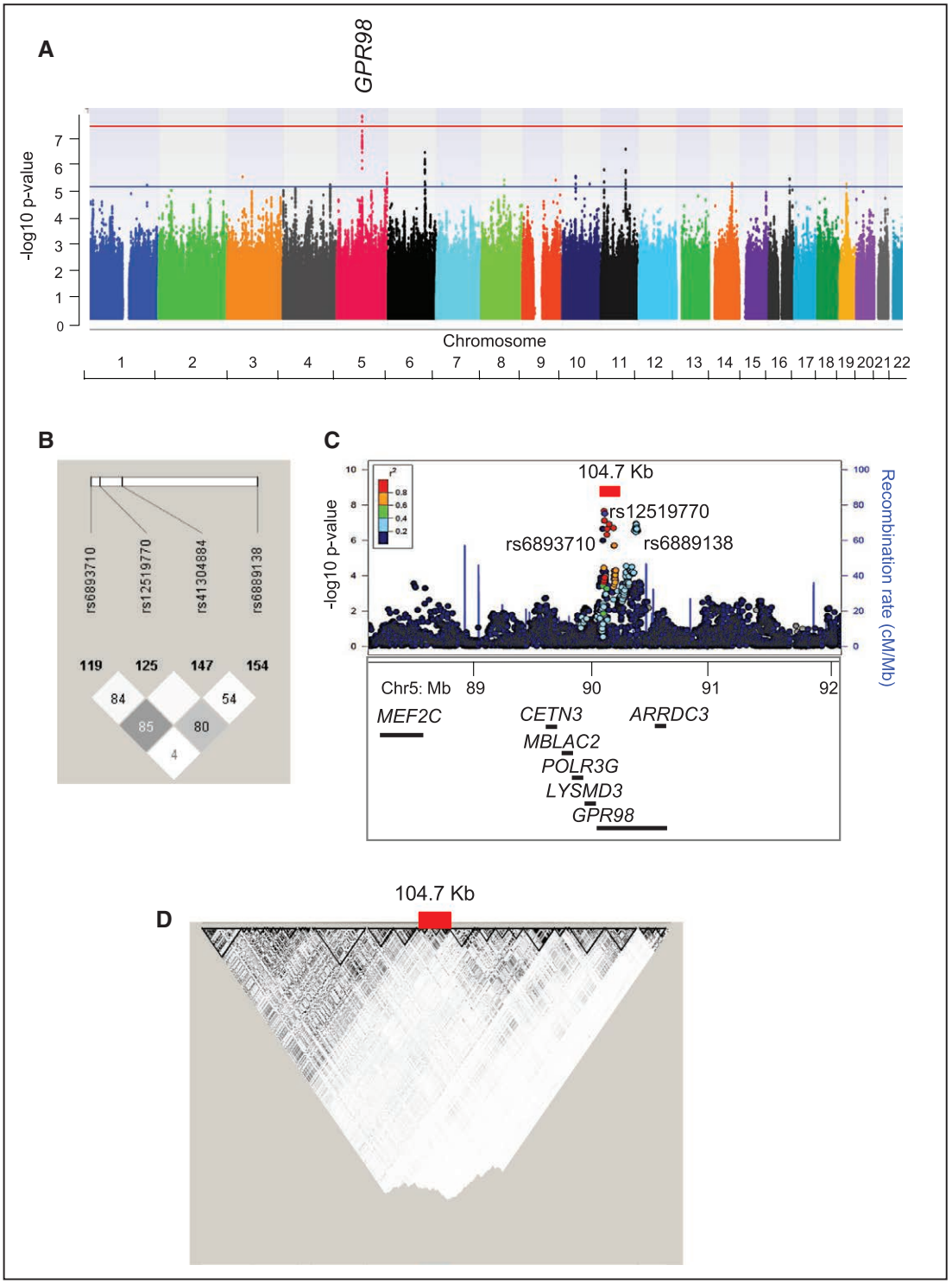

Figure 2. Genome-wide association results for tetralogy of Fallot (TOF) in 22q11.2 deletion syndrome (22q11.2DS). **A**, Values in the Manhattan plot for TOF vs controls were plotted against their respective positions on the autosomal chromosomes. The red line represents the genome-wide significance threshold (P=5×10^{-8}). The blue line represents the threshold for suggestive association (P=1×10^{-5}). A single locus marked by the *GPR98* (G-protein–coupled receptor V1) gene reached genome-wide significance. **B**, LD matrix of selected, predicted functional SNPs with top P values on the 5q14.3 region from WGS (Methods). The LD with respect to associated single-nucleotide polymorphisms (SNPs) with highest P values in the region is shown. The LD plot is based on r^2 values. Key: r^2=0 is given in white, 0<r^2<1 is given in shades of grey and r^2=1 is given in black. The pairwise D′ values are provided in the boxes. Nine SNPs are in modest LD (D′=0.80, r^2=0.23) with rs12519770. The G allele of the top genotyped SNP, rs6889138, located in intron 74 of *GPR98* and had a minor allele frequency (MAF) of 0.30 in TOF cases, and 0.21 in controls giving an odds ratio (OR) of 1.68 (P=1.72×10^{-7}) per copy in the 22q11.2DS cohort. There were 14 SNPs in the second group that had suggestive association with TOF and were in modest LD with SNP, rs12519770 (D′=0.84, r^2=0.02). The top SNP, rs6893710, is located in intron 47 of *GPR98* and had a MAP of 0.058 in TOF cases, but 0.015 in controls, giving an OR of 4.05 (P=1.04×10^{-6}) per copy of the C allele in the 22q11.2DS cohort (Table 2). **C**, LocusZoom plot of region of association at rs12519770 on 5q14.3 indicating $-\log_{10} P$ values (y axis) against the chromosomal positions of SNPs (x axis). The genotyped (*Continued*)

Figure 2 Continued. SNP with the strongest association signal in each locus is represented as a purple diamond; the other SNPs are colored according to the extent of LD (correlation r^2 is based on CEU HapMap haplotypes) with this SNP. Estimated recombination rates (GRCh37/hg19, CEU; 1000 Genomes Project 2012) are shown as light blue lines. Genes are indicated below the LocusZoom plot. **D**, Fine mapping of whole-genome sequencing (WGS; MAF>0.05) based on LD and imputed SNPs from the arrays was used to narrow the TOF signal to a 104.7 Kb region (chromosome 5 [chr5]: 90 057 563–90 162 285) as shown. Most of the common SNPs from this region have *P* values $<10^{-5}$. ARRDC3 indicates Arrestin domain containing 3; CETN3, Centrin 3; CEU, Northern Europeans who are Utah residents part of the CEPH collection; GPR98, G-protein–coupled receptor V1; LYSMD3, LysM, putative peptidoglycan-binding, domain containing 3; MBLAC2, Metallo-β-lactamase domain containing 2; MEF2C, Myocyte-specific enhancer factor 2C; and POLR3G, RNA polymerase III subunit G.

containing 3), maps downstream of *GPR98*, but it is in a different TAD (Figure 3). The same domain structure marked by TAD triangles occurred in most of the cell lines that were examined (Figure IV in the Data Supplement).

We next determined whether any of the genes are expressed in the pharyngeal apparatus or heart in embryos. Probes were generated, and in situ hybridization was successfully performed for *Gpr98, Cetn3*, *Lysmd3*, and *Mef2c* in mouse embryos at E9.5 and E10.5, when the cardiac OFT is expanding (Figure 3). Although *Gpr98* is weakly expressed at E9.5, it is strongly expressed in the neural tube region, particularly the hindbrain[32] (Figure 3). *Cetn3*, important in the cilia[33] for centrosome reproduction,[34] and *Lysmd3*, of unknown function, are ubiquitously expressed, although *Cetn3* has lower expression levels in the heart itself (Figure 3). *Mef2c* encodes a MADS box transcription factor, and it is expressed in the pharyngeal apparatus including the second heart field mesoderm,[35] as indicated in Figure 3. Among the genes in the TAD, *MEF2C* is the only one specifically expressed in cardiac progenitor cells known to be required for heart development.[36]

The second heart field mesodermal progenitor cell populations forming the cardiac OFT lie within the distal pharyngeal apparatus. We then examined expression levels of genes in the 5q14.3 region in existing Affymetrix microarray data from the microdissected distal pharyngeal apparatus.[12] The purpose was to determine whether any of the genes are expressed in this critical tissue. We compared expression levels of genes on 5q14.3 to the highest expressed genes (*Actb, Gapdh*), *Tbx1* expression, and the lowest expressed genes (*Il6, Olfr299*) in this tissue as shown in Figure 3C. All of the genes are expressed in the pharyngeal apparatus, albeit *Gpr98* is expressed at the lowest level, as can be also seen in Figure 3B.

The Encyclopedia of DNA Elements functional genomics data were examined in the 104.7 kb region of LD with SNP, rs12519770, to identify possible regulatory regions. We found 3 possible regulatory regions defined by binding of multiple transcription factors, with one close to rs12519770 (Figure V in the Data Supplement). None of the SNPs that were genotyped in our study lie within these putative regulatory regions; however, critical embryonic regulatory regions could be different, and they have not yet been defined.

Discussion

We identified genome-wide significant associations between TOF and several SNPs in an intron of *GPR98* in our 22q11.2DS cohort. We narrowed the associated region to a 104.7 kb interval. This interval may harbor functional variants that are in LD with the associated SNPs or noncoding variants that regulate the expression of genes within a broad TAD on 5q14.3.

GPR98 contains 90 exons, spans over 610 kb, and encodes a member of the adhesion-G protein-coupled receptor family of receptors. The GPR98 protein binds calcium and is weakly expressed early in mouse embryonic development at E9.5, but it becomes more strongly expressed in the future brain and neural tube by E10.5. It is the largest of the 7-transmembrane receptors and has important functions in hearing and vision.[37,38] Recessive mutations cause Usher syndrome type 2C (Mendelian Inheritance in Man No. 605472), which is characterized by congenital hearing loss and progressive retinitis pigmentosa.[39] There are multiple splice variants present in *GPR98*, and it is not known if all isoforms have similar functions. Thus, it is possible that ≥1 splice variants could have a function in neural crest cells deriving from the neural tube. Neural crest cells are a migratory population of progenitor cells in the pharyngeal apparatus, which contribute to cardiac OFT septation. There are no reports of a possible function of *GPR98* in the cardiovascular system and no known connections to human TOF.

CETN3 is another gene of note in the 5q14.3 region because it encodes a centrin protein that functions in the cytoskeleton of centrosomes and cilia.[40] Cilia are critically important for conferring left right assymetry during embryonic development and when disrupted is associated with human cardiac anomalies[22] Because laterality defects are not commonly found in association with 22q11.2DS, more work would need

Table 2. Top SNPs in *GPR98* Associated With TOF

SNP	Chr:Position (bp)	Cluster	Risk Allele	Other Allele	RAF	OR (95% CI)	*P* Value	Conditional *P* Value*	Conditional *P* Value†
rs12519770	5:90073277	1	A	G	0.5	1.69 (1.39–2.06)	2.98E-08	NA	NA
rs6889138	5:90335635	2	G	A	0.24	1.68 (1.35–2.10)	1.72E-07	0.00199523	NA
rs6893710	5:90058041	3	C	T	0.03	4.05 (2.27–7.24)	1.04E-06	3.92E-05	6.11E-05

The top genotyped single-nucleotide polymorphisms (SNPs) in each cluster of SNPs identified for tetralogy of Fallot (TOF) are shown. Chr:Position (bp), chromosome and positions are indicated according to NCBI36/hg18 (March 2006), and allele coding was based on the positive strand. The cluster numbers that the SNPs belong to are indicated. *GPR98* indicates G-protein–coupled receptor V1; NA, not associated; OR, odds ration; and RAF, risk allele frequency in the whole cohort.

*Conditional *P* value: analysis was conditioned on SNP rs12519770.

†Conditional *P* value: individually evaluated the association of TOF with 1344 SNPs in the 5q14.3 region (Table VI in the Data Supplement).

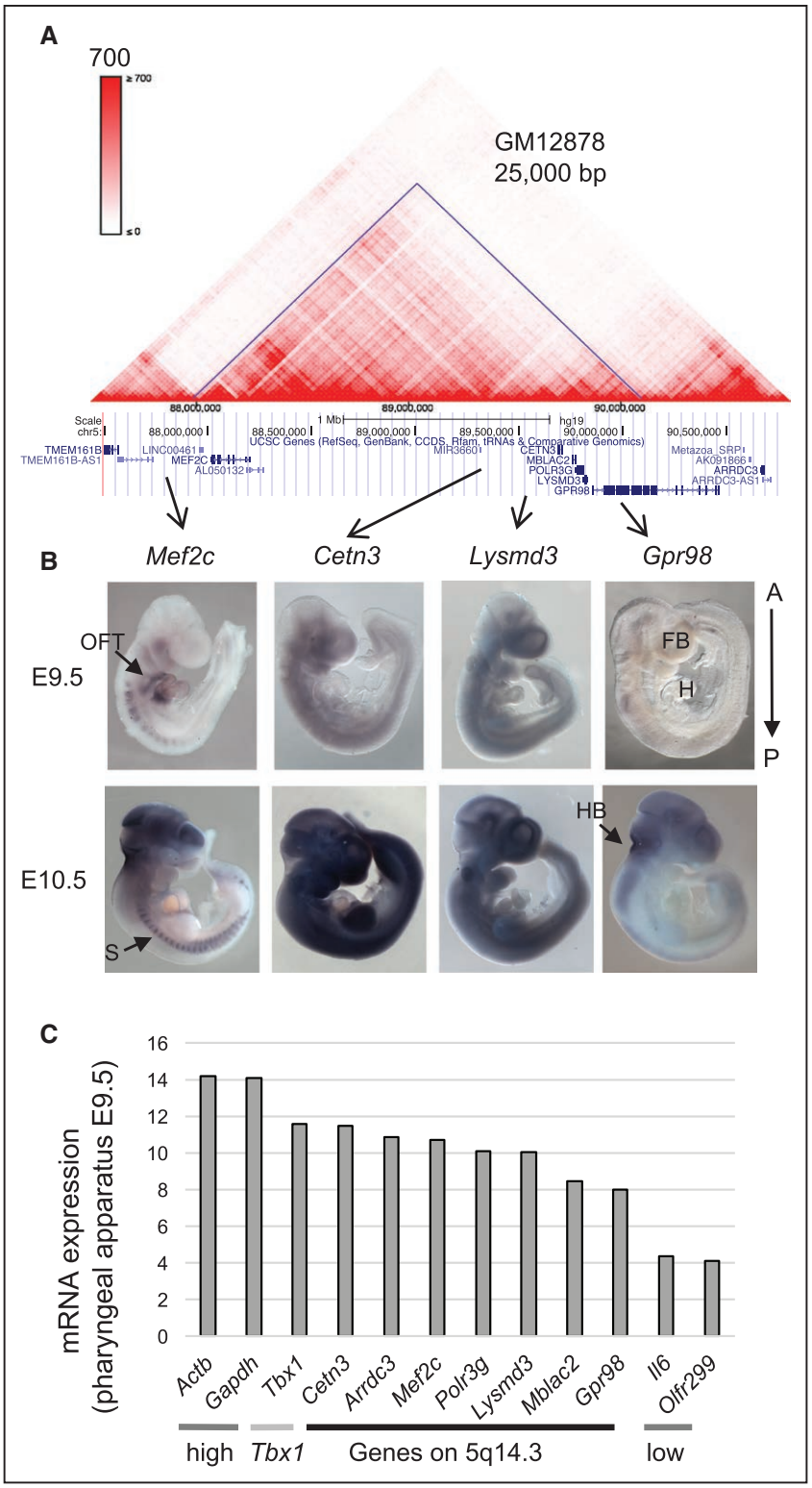

Figure 3. Representative heatmap of chromatin conformation for the *MEF2C* (Myocyte-specific enhancer factor 2C)-*GPR98* (G-protein–coupled receptor V1) interval and in situ hybridization of selected genes in mouse embryos. **A**, Chromatin conformation map extracted from Hi-C data for the human lymphoblastoid cell line, GM12878, from the HapMap sample set.[29] Similar results were found using other cell types (Figure III in the Data Supplement). The intensity of each pixel represents the normalized number of contacts between a pair of loci. The purple line marking the triangle depicts the contact domain[31] between the *GPR98* and the *MEF2C* locus. **B**, Lateral views of E9.5 and E10.5 wild-type embryos following in situ hybridization of antisense probes for *Gpr98*, *Cetn3* (Centrin 3), *Mef2c,* and *Lysmd3* (LysM, putative peptidoglycan-binding, domain containing 3). Purple color indicates mRNA expression. **C**, Bar graph of relative gene expression levels from Affymetrix microarrays for the microdissected distal pharyngeal apparatus from E9.5 stage mouse embryos (*y* axis). Individual genes are shown for comparison, including representative housekeeping genes with high expression (*Actb*, *Gapdh*), genes on chromosome 5q14.3 (*Cetn3, Arrdc3, Mef2c, Polr3g, Lysmd3, Mblac2,* and *Gpr98*) and representative low expressing genes (*Il6, Olfr299*), in descending order according to expression. A indicates anterior; Arrdc3 indicates Arrestin domain containing 3; Cetn3, Centrin 3; FB, forebrain; H, heart; HB, hindbrain; Lysmd3, LysM, putative peptidoglycan-binding, domain containing 3; Mblac2, Metallo-β-lactamase domain containing 2; OFT, cardiac outflow tract; P, posterior; Polr3g, RNA polymerase III subunit G; and S, somites.

to be done to provide additional support of the role of *CETN3* as a modifier of TOF in these individuals.

Among the remaining 5 genes (*MEF2C, POLR3G, MBLAC2, LYSMD3,* and *ARRDC3*), *MEF2C* is of particular interest because it encodes a transcription factor required in the second heart field mesoderm of the pharyngeal apparatus during embryogenesis for cardiac OFT development.[41,42] Because haploinsufficiency of *TBX1* is important for 22q11.2DS, and both *MEF2C* and *TBX1* are expressed in the second heart field progenitor cells, it is possible that *TBX1* might act in the same genetic pathway as *MEF2C*. Further, studies in mouse models indicate that *Tbx1* may be a negative regulator of *Mef2c*.[43] This suggests that genetic variants in the 5q14.3 locus that may be associated with *MEF2C* expression levels could act as genetic modifiers of 22q11.2DS, with the

caveat that causation will require direct experimental support. Further, ISL1 transcription factor (ISL LIM homeobox 1 transcription factor) and GATA (transcription factor) proteins bind and regulate both *Mef2c* and *Nkx2-5* (NK2 homeobox 5 transcription factor) cardiac development genes.[44] One hypothesis to test in the future by direct experimentation would be that rare DNA variants in *MEF2C*, *ISL1,* or *NKX2-5* affect cardiac OFT formation in individuals with 22q11.2DS.

In recent years, higher-order chromatin structure technologies have demonstrated that chromatin interactions occur in a nonrandom manner along the chromosome arms, which are separated into regions of highly interacting chromatin.[30,45–47] Relevant to this, the 104.7 kb region found with top associated SNPs to TOF shows a possible regulatory connection with *MEF2C* located 2 Mb upstream of *GPR98* using available Hi-C, chromatin conformation data.[29] One limitation of using Hi-C data to draw conclusions about gene regulation is that these data indicate that the 2 genes may reside within the same topological region but do not prove that there is a definitive regulatory connection. Further chromatin conformation data in cell progenitors relevant to cardiac OFT development, followed by direct experimental approaches, will be required to define this interaction further. In addition, a better understanding of the regulatory landscape in this region will be needed to identify the mechanism(s) responsible for the association to TOF we have observed in the 5q14.3 region.

Further support for *MEF2C,* as a possible modifier gene, comes from a recent GWAS of circulating VEGF (vascular endothelial growth factor) levels in blood in adults.[48] Although this is a study of adults, factors that regulate VEGF levels may be similar throughout life and could affect fetal development. Previously, it was found that absence of one of the VEGF isoforms causes a phenocopy of 22q11.2DS in mouse models.[49] In this recent GWAS of VEGF levels in adults, *MEF2C* and *JMJD1C* (Jumonji domain containing 1C) were found among the 6 loci with significant association to VEGF levels. *JMJD1C* is relevant because we previously found significant enrichment of rare predicted exonic variants in *JMJD1C* in whole-exome sequence from 184 22q11.2DS subjects in which the cases were enriched for TOF.[50] This provides a potential biological connection between common and previous rare variant analyses for 22q11.2DS. However, in regards to the general population, neither haploinsufficiency nor mutation of *MEF2C* in humans has been associated with any type of CHD thus far.[51–53] Further proof that *MEF2C* is a cardiac disease gene in the general population will require future genetic studies.

The number of samples we obtained, even after 25 years of collection, is quite small for a GWAS of a complex trait. Thus, one of the limitations of the study was the lack of a true replication cohort. Another limitation of our study is possible population stratification. The majority of our cohort was self-reported as white. Nevertheless, a subset had different ethnicities requiring statistical correction in the analysis. Further, we examined each ethnicity separately and combined each group by performing a meta-analysis. We found that the top SNP in *GPR98* was still statistically significant. In addition, the odds ratio was in the same direction in all the subcohorts, supporting our findings despite the limitations.

In this report, we used available bioinformatic data to interpret our data. However, this study did not provide proof of causation. This will need to be done by performing functional studies including development of animal models. Despite the limitations of the study, this is the first GWAS to identify common variants that may modify the cardiac phenotype in a large cohort of individuals with 22q11.2DS.

Conclusions

A GWAS of TOF in 22q11.2DS has identified a significant locus on 5q14.3 harboring potential genetic risk factors. Several genes reside in this locus including *MEF2C* that is a known gene for cardiac OFT development in animal models. Further work needs to be done to ascertain whether *MEF2C* or other genes in this region act as modifiers of TOF in 22q11.2DS.

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Appendix

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Disclosures

None.

References

1. McDonald-McGinn DM, Sullivan KE, Marino B, Philip N, Swillen A, Vorstman JA, et al. 22q11.2 deletion syndrome. *Nat Rev Dis Primers*. 2015;1:15071. doi: 10.1038/nrdp.2015.71.
2. Lindsay EA, Greenberg F, Shaffer LG, Shapira SK, Scambler PJ, Baldini A. Submicroscopic deletions at 22q11.2: variability of the clinical picture and delineation of a commonly deleted region. *Am J Med Genet*. 1995;56:191–197. doi: 10.1002/ajmg.1320560216.
3. Lindsay EA, Goldberg R, Jurecic V, Morrow B, Carlson C, Kucherlapati RS, et al. Velo-cardio-facial syndrome: frequency and extent of 22q11 deletions. *Am J Med Genet*. 1995;57:514–522. doi: 10.1002/ajmg.1320570339.
4. Morrow B, Goldberg R, Carlson C, Das Gupta R, Sirotkin H, Collins J, et al. Molecular definition of the 22q11 deletions in velo-cardio-facial syndrome. *Am J Hum Genet*. 1995;56:1391–1403.
5. Burn J Goodship J. Developmental genetics of the heart. *Curr Opin Genet Dev*. 1996;6:322–325.
6. Kelly RG. The second heart field. *Curr Top Dev Biol*. 2012;100:33–65. doi: 10.1016/B978-0-12-387786-4.00002-6.
7. Goldmuntz E, Clark BJ, Mitchell LE, Jawad AF, Cuneo BF, Reed L, et al. Frequency of 22q11 deletions in patients with conotruncal defects. *J Am Coll Cardiol*. 1998;32:492–498.
8. Chessa M, Butera G, Bonhoeffer P, Iserin L, Kachaner J, Lyonnet S, et al. Relation of genotype 22q11 deletion to phenotype of pulmonary vessels in tetralogy of Fallot and pulmonary atresia-ventricular septal defect. *Heart*. 1998;79:186–190.
9. Lindsay EA, Vitelli F, Su H, Morishima M, Huynh T, Pramparo T, et al. Tbx1 haploinsufficieny in the DiGeorge syndrome region causes aortic arch defects in mice. *Nature*. 2001;410:97–101. doi: 10.1038/35065105.
10. Merscher S, Funke B, Epstein JA, Heyer J, Puech A, Lu MM, et al. TBX1 is responsible for cardiovascular defects in velo-cardio-facial/DiGeorge syndrome. *Cell*. 2001;104:619–629.
11. Jerome LA Papaioannou VE. DiGeorge syndrome phenotype in mice mutant for the T-box gene, Tbx1. *Nature Genet*. 2001;27:286–291.
12. Racedo SE, Hasten E, Lin M, Devakanmalai GS, Guo T, Ozbudak EM, et al. Reduced dosage of β-catenin provides significant rescue of cardiac outflow tract anomalies in a Tbx1 conditional null mouse model of 22q11.2 deletion syndrome. *PLoS Genet*. 2017;13:e1006687. doi: 10.1371/journal.pgen.1006687.
13. Guo T, McDonald-McGinn D, Blonska A, Shanske A, Bassett AS, Chow E, et al; International Chromosome 22q11.2 Consortium. Genotype and cardiovascular phenotype correlations with TBX1 in 1,022 velo-cardio-facial/DiGeorge/22q11.2 deletion syndrome patients. *Hum Mutat*. 2011;32:1278–1289. doi: 10.1002/humu.21568.
14. Mlynarski EE, Sheridan MB, Xie M, Guo T, Racedo SE, McDonald-McGinn DM, et al; International Chromosome 22q11.2 Consortium. Copy-Number Variation of the glucose transporter gene SLC2A3 and congenital heart defects in the 22q11.2 deletion syndrome. *Am J Hum Genet*. 2015;96:753–764. doi: 10.1016/j.ajhg.2015.03.007.
15. León LE, Benavides F, Espinoza K, Vial C, Alvarez P, Palomares M, et al. Partial microduplication in the histone acetyltransferase complex member KANSL1 is associated with congenital heart defects in 22q11.2 microdeletion syndrome patients. *Sci Rep*. 2017;7:1795. doi: 10.1038/s41598-017-01896-w.
16. Tomita-Mitchell A, Mahnke DK, Struble CA, Tuffnell ME, Stamm KD, Hidestrand M, et al. Human gene copy number spectra analysis in congenital heart malformations. *Physiol Genomics*. 2012;44:518–541. doi: 10.1152/physiolgenomics.00013.2012.
17. Browning BL Yu Z. Simultaneous genotype calling and haplotype phasing improves genotype accuracy and reduces false-positive associations for genome-wide association studies. *Am J Hum Genet*. 2009;85:847–861.
18. Delaneau O, Marchini J, Zagury JF. A linear complexity phasing method for thousands of genomes. *Nat Methods*. 2011;9:179–181. doi: 10.1038/nmeth.1785.
19. Delaneau O, Zagury JF, Marchini J. Improved whole-chromosome phasing for disease and population genetic studies. *Nat Methods*. 2013;10:5–6. doi: 10.1038/nmeth.2307.
20. Howie B, Fuchsberger C, Stephens M, Marchini J, Abecasis GR. Fast and accurate genotype imputation in genome-wide association studies through pre-phasing. *Nat Genet*. 2012;44:955–959. doi: 10.1038/ng.2354.
21. Liu JZ, Tozzi F, Waterworth DM, Pillai SG, Muglia P, Middleton L, et al; Wellcome Trust Case Control Consortium. Meta-analysis and imputation refines the association of 15q25 with smoking quantity. *Nat Genet*. 2010;42:436–440. doi: 10.1038/ng.572.
22. Willaredt MA, Gorgas K, Gardner HA, Tucker KL. Multiple essential roles for primary cilia in heart development. *Cilia*. 2012;1:23. doi: 10.1186/2046-2530-1-23.
23. Pruim RJ, Welch RP, Sanna S, Teslovich TM, Chines PS, Gliedt TP, et al. LocusZoom: regional visualization of genome-wide association scan results. *Bioinformatics*. 2010;26:2336–2337. doi: 10.1093/bioinformatics/btq419.
24. Johnston HR, Chopra P, Wingo TS, Patel V, Epstein MP, Mulle JG, et al; International Consortium on Brain and Behavior in 22q11.2 Deletion Syndrome. PEMapper and PECaller provide a simplified approach to whole-genome sequencing. *Proc Natl Acad Sci USA*. 2017;114:E1923–E1932. doi: 10.1073/pnas.1618065114.
25. Zhao H, Sun Z, Wang J, Huang H, Kocher JP, Wang L. CrossMap: a versatile tool for coordinate conversion between genome assemblies. *Bioinformatics*. 2014;30:1006–1007. doi: 10.1093/bioinformatics/btt730.

26. Bulger M, Groudine M. Looping versus linking: toward a model for long-distance gene activation. *Genes Dev*. 1999;13:2465–2477.
27. Schoenfelder S, Clay I, Fraser P. The transcriptional interactome: gene expression in 3D. *Curr Opin Genet Dev*. 2010;20:127–133. doi: 10.1016/j.gde.2010.02.002.
28. Kagey MH, Newman JJ, Bilodeau S, Zhan Y, Orlando DA, van Berkum NL, et al. Mediator and cohesin connect gene expression and chromatin architecture. *Nature*. 2010;467:430–435. doi: 10.1038/nature09380.
29. Dixon JR, Jung I, Selvaraj S, Shen Y, Antosiewicz-Bourget JE, Lee AY, et al. Chromatin architecture reorganization during stem cell differentiation. *Nature*. 2015;518:331–336. doi: 10.1038/nature14222.
30. Dixon JR, Selvaraj S, Yue F, Kim A, Li Y, Shen Y, et al. Topological domains in mammalian genomes identified by analysis of chromatin interactions. *Nature*. 2012;485:376–380. doi: 10.1038/nature11082.
31. Rao SS, Huntley MH, Durand NC, Stamenova EK, Bochkov ID, Robinson JT, et al. A 3D map of the human genome at kilobase resolution reveals principles of chromatin looping. *Cell*. 2014;159:1665–1680. doi: 10.1016/j.cell.2014.11.021.
32. McMillan DR, Kayes-Wandover KM, Richardson JA, White PC. Very large G protein-coupled receptor-1, the largest known cell surface protein, is highly expressed in the developing central nervous system. *J Biol Chem*. 2002;277:785–792. doi: 10.1074/jbc.M108929200.
33. Kumamoto N, Gu Y, Wang J, Janoschka S, Takemaru K, Levine J, et al. A role for primary cilia in glutamatergic synaptic integration of adult-born neurons. *Nat Neurosci*. 2012;15:399–405, S1. doi: 10.1038/nn.3042.
34. Middendorp S, Küntziger T, Abraham Y, Holmes S, Bordes N, Paintrand M, et al. A role for centrin 3 in centrosome reproduction. *J Cell Biol*. 2000;148:405–416.
35. Verzi MP, McCulley DJ, De Val S, Dodou E, Black BL. The right ventricle, outflow tract, and ventricular septum comprise a restricted expression domain within the secondary/anterior heart field. *Dev Biol*. 2005;287:134–145. doi: 10.1016/j.ydbio.2005.08.041.
36. Lin Q, Schwarz J, Bucana C, Olson EN. Control of mouse cardiac morphogenesis and myogenesis by transcription factor MEF2C. *Science*. 1997;276:1404–1407.
37. Sun JP, Li R, Ren HZ, Xu AT, Yu X, Xu ZG. The very large G protein coupled receptor (Vlgr1) in hair cells. *J Mol Neurosci*. 2013;50:204–214. doi: 10.1007/s12031-012-9911-5.
38. McMillan DR White PC. Studies on the very large G protein-coupled receptor: from initial discovery to determining its role in sensorineural deafness in higher animals. *Adv Exp Med Biol*. 2010;706:76–86.
39. Weston MD, Luijendijk MW, Humphrey KD, Möller C, Kimberling WJ. Mutations in the VLGR1 gene implicate G-protein signaling in the pathogenesis of Usher syndrome type II. *Am J Hum Genet*. 2004;74:357–366. doi: 10.1086/381685.
40. Laoukili J, Perret E, Middendorp S, Houcine O, Guennou C, Marano F, et al. Differential expression and cellular distribution of centrin isoforms during human ciliated cell differentiation in vitro. *J Cell Sci*. 2000;113 (pt 8):1355–1364.
41. Potthoff MJ, Olson EN. MEF2: a central regulator of diverse developmental programs. *Development*. 2007;134:4131–4140.
42. Barnes RM, Harris IS, Jaehnig EJ, Sauls K, Sinha T, Rojas A, et al. MEF2C regulates outflow tract alignment and transcriptional control of Tdgf1. *Development*. 2016;143:774–779. doi: 10.1242/dev.126383.
43. Pane LS, Zhang Z, Ferrentino R, Huynh T, Cutillo L, Baldini A. Tbx1 is a negative modulator of Mef2c. *Hum Mol Genet*. 2012;21:2485–2496. doi: 10.1093/hmg/dds063.
44. Takeuchi JK, Mileikovskaia M, Koshiba-Takeuchi K, Heidt AB, Mori AD, Arruda EP, et al. Tbx20 dose-dependently regulates transcription factor networks required for mouse heart and motoneuron development. *Development*. 2005;132:2463–2474. doi: 10.1242/dev.01827.
45. Nora EP, Lajoie BR, Schulz EG, Giorgetti L, Okamoto I, Servant N, et al. Spatial partitioning of the regulatory landscape of the X-inactivation centre. *Nature*. 2012;485:381–385. doi: 10.1038/nature11049.
46. Hou C, Li L, Qin ZS, Corces VG. Gene density, transcription, and insulators contribute to the partition of the Drosophila genome into physical domains. *Mol Cell*. 2012;48:471–484. doi: 10.1016/j.molcel.2012.08.031.
47. Sexton T, Yaffe E, Kenigsberg E, Bantignies F, Leblanc B, Hoichman M, et al. Three-dimensional folding and functional organization principles of the Drosophila genome. *Cell*. 2012;148:458–472. doi: 10.1016/j.cell.2012.01.010.
48. Choi SH, Ruggiero D, Sorice R, Song C, Nutile T, Vernon Smith A, et al. Six novel loci associated with circulating VEGF levels identified by a meta-analysis of genome-wide association studies. *PLoS Genet*. 2016;12:e1005874. doi: 10.1371/journal.pgen.1005874.
49. Stalmans I, Lambrechts D, De Smet F, Jansen S, Wang J, Maity S, et al. VEGF: a modifier of the del22q11 (DiGeorge) syndrome? *Nat Med*. 2003;9:173–182. doi: 10.1038/nm819.
50. Guo T, Chung JH, Wang T, McDonald-McGinn DM, Kates WR, Hawuła W, et al. Histone modifier genes alter conotruncal heart phenotypes in 22q11.2 deletion syndrome. *Am J Hum Genet*. 2015;97:869–877. doi: 10.1016/j.ajhg.2015.10.013.
51. Zweier M, Gregor A, Zweier C, Engels H, Sticht H, Wohlleber E, et al; Cornelia Kraus. Mutations in MEF2C from the 5q14.3q15 microdeletion syndrome region are a frequent cause of severe mental retardation and diminish MECP2 and CDKL5 expression. *Hum Mutat*. 2010;31:722–733. doi: 10.1002/humu.21253.
52. Hotz A, Hellenbroich Y, Sperner J, Linder-Lucht M, Tacke U, Walter C, et al. Microdeletion 5q14.3 and anomalies of brain development. *Am J Med Genet A*. 2013;161A:2124–2133. doi: 10.1002/ajmg.a.36020.
53. Bienvenu T, Diebold B, Chelly J, Isidor B. Refining the phenotype associated with MEF2C point mutations. *Neurogenetics*. 2013;14:71–75. doi: 10.1007/s10048-012-0344-7.

CLINICAL PERSPECTIVE

The 22q11.2 deletion syndrome (DiGeorge syndrome/velocardiofacial syndrome) occurs in 1 of 4000 live births, and 60% to 70% of affected individuals have congenital heart disease, ranging from mild to severe. In our cohort of 1472 subjects with 22q11.2 deletion syndrome, a total of 62% (n=906) have congenital heart disease and 36% (n=326) of these have tetralogy of Fallot, comprising the largest subset of severe congenital heart disease in the cohort. One of the main questions clinicians are interested in understanding is the basis for variable phenotypic expression. To address this question, using 22q11.2 deletion syndrome as a model, we performed a genome-wide association study and found tetralogy of Fallot was significantly associated with a genetic interval on chromosome 5q14.3. The associated region is within an intron of GPR98. This intron may contain regulatory sequences that could effect expression of *GPR98* or other genes in the region. Among them, *MEF2C* (Myocyte-specific enhancer factor 2C) is the only gene that is known to affect heart development. On the basis of this work, common DNA variants on 5q14.3 may explain, in part, why phenotypic variation occurs despite the fact that they have the same deletion. Further work will need to be done to determine whether the same genetic variants can alter *MEF2C* expression and risk for congenital heart disease in the general population or alter clinical outcomes. In the future, understanding how genetic variations influence phenotype in both genetic syndromes and in more common disease will make it possible to improve genetic counseling, rehabilitation, clinical outcomes, and in the creation of novel therapeutics.

Circulation
Cardiovascular Genetics

Genome-Wide Association Study to Find Modifiers for Tetralogy of Fallot in the 22q11.2 Deletion Syndrome Identifies Variants in the *GPR98* Locus on 5q14.3

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on behalf of the International 22q11.2 Consortium/Brain and Behavior Consortium*

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Supplementary Table 1. Complete list of investigators that are part of the International 22q11.2 Brain and Behavior Consortium. The list is organized alphabetically. Membership indicated at the bottom of the table.

Collection Site	Number
Children Hospital of Philadelphia, PA, USA	396
Upstate Medical University, NY, USA	255
University of Toronto, Canada	132
KU Leuven, Belgium	103
Maastricht University, Netherlands	63
Tel Aviv University, Israel	74
University of Geneva, Geneva, Switzerland	65
Children's Healthcare of Atlanta, GA, USA	41
Bambino Gesu Hospital , Rome, Italy	34
University Hospital Timone, Marseilles, France	27
UCLA, CA, USA	22

Medical College of Wisconsin, WI, USA	36
Polish Mother's Memorial Hospital, Lodz, Poland	9
Boston Children's Hospital, MA, USA	6
UC Davis, USA	4
Others	14
Universidad del Desarrollo, Santiago, Chile	191
Total	1472

Supplementary Table 2. Number of the 22q11DS collected from each site.

Gene	Forward primer	Reverse primer
Gpr98	5'-GGGGAATTAACCCTCACTAAAGGGGTGTATGCTGTCTACGCCCA-3'	5'-GGGGTAATACGACTCACTATAGGGAAAACATAAAGTCCCTGCAGACTA-3'
Cetn3	5'-GGGGAATTAACCCTCACTAAAGGGGCTCTGAGAGGTGAGCTTGT-3'	5'-GGGGTAATACGACTCACTATAGGGAGGATGCTTGGTTTTGTTCCT-3'
Mef2c	5'-GGGGAATTAACCCTCACTAAAGGGCACCGGAACGAATTCCACTCC-3'	5'-GGGGTAATACGACTCACTATAGGGCCGGGTCTGTCCAAACCTCTA-3'
Lysmd3	5'-GGGGAATTAACCCTCACTAAAGGGAGGACAGCTAGAACGTCATGG-3'	5'-GGGGTAATACGACTCACTATAGGGAGAGAACCATCAGCTGGCAC-3'

Supplementary Table 3. Primers used for generating antisense RNA probes for *in situ* hybridization of mouse embryos of selected genes in the 5q14.3 locus. Primers shown in a 5'

Size of Deletion	Total	Control	CHD	TOF	CHD%	TOF%
LCR22A-D	1366	541	825	302	0.603953148	0.358244365
LCR22A-B	73	28	45	12	0.616438356	0.3
LCR22A-C	26	7	19	4	0.730769231	0.363636364
Others	7	5	2	2	0.285714286	0.285714286

Supplementary Table 4. Number of individuals with different sized deletion in the 22q11.2DS cohort. Control: individuals with 22q11.2DS but no detectable heart or aortic arch anomaly. Case: individuals with 22q11.2DS with a heart or aortic arch anomaly. CHD%: or TOF% is the percent of CHD or TOF, respectively among those with particular deletion sizes as indicated.

			CEU		AMR		AFR		Meta-analysis	
	CHD	Risk Alelele	OR(95% CI)	P	OR(95% CI)	P	OR(95% CI)	P	OR(95% CI)	P
rs12519770	Heart	T	1.83(1.37,2.46)	0.000622	1.35(0.79,2.33)	0.003	2.92(0.82,10.33)	0.0003	1.54(1.28,1.86)	7.045E-07
rs12519770	TOF	T	2.09(1.46,3.00)	0.00000874	1.54(0.77,3.12)	0.0464	2.20(0.52,9.30)	0.003	1.69(1.39,2.06)	4.43E-08

Supplementary Table 5. Comparison of races and ethnicities in the cohort. Meta-analysis of top SNP in TOF compared with combined CHD (heart) in different ethnicities of the cohort (CEU = Caucasian; AMR = Admixed mixed American; AFR = African).

SNP	Chr:Position(bp)	info	Risk Allele	Other allele	RAF	OR (95% CI)	P
rs154570	chr5:89854312	0.99713	G	C	0.54	1.16(0.96,1.41)	0.108433
rs154569	chr5:89854325	0.999644	G	A	0.52	1.07(0.88,1.3)	0.289311
rs154568	chr5:89854448	0.987062	C	G	0.53	1.13(0.93,1.37)	0.125697
rs192362302	chr5:89854669	0.917565	C	T	0.02	2.02(4.24,0.96)	0.0495458
rs75224084	chr5:89855959	0.91092	A	G	0.04	1.09(1.84,0.64)	0.804165
rs1700510	chr5:89857264	0.994967	C	A	0.63	1.08(0.89,1.32)	0.303013
rs40205	chr5:89857574	1	G	A	0.85	1(0.76,1.32)	0.854277
rs35980777	chr5:89857625	0.978298	A	G	0.92	1.12(0.78,1.6)	0.534306
rs12188935	chr5:89859089	0.990055	T	C	0.06	1.33(2.01,0.88)	0.203236
rs79256827	chr5:89860983	0.877508	G	A	0.03	1.06(1.93,0.58)	0.884283
rs474800	chr5:89861312	0.999752	C	T	0.63	1.07(0.88,1.31)	0.329808
rs623761	chr5:89861410	0.999768	T	G	0.63	1.07(0.88,1.3)	0.354982
rs552314	chr5:89862615	0.99975	T	C	0.63	1.07(0.88,1.31)	0.328167
rs688956	chr5:89863279	1	C	T	0.63	1.07(0.88,1.3)	0.354068
rs150388444	chr5:89864977	0.918665	C	T	0.01	1.88(5.32,0.67)	0.211819
rs154573	chr5:89865123	0.996894	C	T	0.52	1.08(0.89,1.31)	0.274391
rs257855	chr5:89865807	0.991784	A	C	0.85	1(0.76,1.32)	0.892088
rs625135	chr5:89865854	1	G	A	0.52	1.13(0.93,1.37)	0.240403
rs154572	chr5:89866872	0.994842	G	C	0.53	1.13(0.93,1.37)	0.18206
rs154571	chr5:89867164	0.99498	C	T	0.14	1(1.31,0.76)	0.906761
rs62375070	chr5:89867373	0.850328	A	G	0.15	1.04(1.36,0.8)	0.831416
rs13162241	chr5:89867828	0.979273	G	A	0.98	1.45(0.69,3.06)	0.322378
rs7724946	chr5:89868779	0.892817	A	G	0.99	2.13(0.9,5.04)	0.175071
chr5:89868937:I	chr5:89868937	0.963025	TCA	T	0.53	1.19(0.98,1.44)	0.056195
rs75647854	chr5:89869465	0.985056	C	T	0.08	1.07(1.53,0.75)	0.634576
rs34007784	chr5:89869506	0.896321	A	G	0.95	1.07(0.7,1.64)	0.726098
chr5:89869718:D	chr5:89869718	0.858468	TAG	T	0.04	1.18(1.89,0.73)	0.515122
rs250366	chr5:89870897	0.993498	C	T	0.53	1.19(0.99,1.45)	0.0530192
rs189691	chr5:89871375	0.993906	T	C	0.83	1.02(0.79,1.32)	0.772653
rs11741740	chr5:89871514	0.99602	C	T	0.71	1.18(0.96,1.45)	0.0923793
rs154566	chr5:89872082	0.976117	G	C	0.53	1.15(0.95,1.4)	0.0870448
rs6888355	chr5:89872545	0.993963	C	T	0.69	1.26(1.02,1.54)	0.0424831
rs6452896	chr5:89874126	0.845975	C	T	0.99	2.21(0.92,5.31)	0.187102
rs150754269	chr5:89875326	0.850432	G	A	0.03	1.35(2.52,0.72)	0.392257
rs567441	chr5:89877032	0.986108	A	G	0.53	1.25(1.03,1.52)	0.0228293
rs144389486	chr5:89877567	0.902359	G	A	0.02	2(4.16,0.96)	0.0483368
rs656679	chr5:89879259	0.995185	C	T	0.52	1.26(1.04,1.52)	0.0218583
chr5:89879662:D	chr5:89879662	0.993902	A	AAT	0.54	1.17(0.96,1.42)	0.067613
rs171629	chr5:89880752	0.998191	A	G	0.83	1.04(0.8,1.34)	0.702052
rs250367	chr5:89881344	0.998283	C	T	0.83	1.05(0.81,1.35)	0.64203
rs498491	chr5:89881445	0.996651	T	G	0.71	1.17(0.95,1.44)	0.101247
rs688960	chr5:89881853	0.997855	G	A	0.71	1.18(0.96,1.45)	0.086341
rs173660	chr5:89882068	1	G	A	0.83	1.03(0.8,1.34)	0.718805
rs645652	chr5:89882327	1	G	A	0.71	1.18(0.96,1.46)	0.0811599
rs154559	chr5:89882618	0.996902	T	G	0.83	1.05(0.81,1.35)	0.709961
rs13190272	chr5:89883114	0.987407	T	C	0.92	1.09(0.77,1.56)	0.579082
rs13153138	chr5:89883370	0.9401	A	G	0.92	1.26(0.9,1.77)	0.299195
rs154561	chr5:89883395	0.950072	A	T	0.18	1.03(1.33,0.8)	0.948803
rs154562	chr5:89883521	0.997629	T	C	0.83	1.03(0.8,1.33)	0.731268
rs154563	chr5:89883692	0.992783	G	A	0.82	1.11(0.86,1.42)	0.517709
rs154564	chr5:89883820	0.997407	T	C	0.82	1.1(0.86,1.42)	0.524645
rs152714	chr5:89884527	0.99761	G	C	0.82	1.1(0.86,1.41)	0.567024
rs34719862	chr5:89884878	0.994653	G	A	0.92	1.07(0.75,1.53)	0.664218
rs11738532	chr5:89887223	0.995214	C	T	0.92	1.07(0.75,1.53)	0.664962
rs168743	chr5:89887683	0.997954	C	T	0.74	1.12(0.9,1.39)	0.397036
rs154565	chr5:89887799	1	A	T	0.82	1.1(0.86,1.41)	0.536896
rs11954191	chr5:89888435	0.996077	A	G	0.99	2.77(1.07,7.17)	0.244059
rs57536334	chr5:89888562	0.956266	A	T	0.84	1.03(0.79,1.33)	0.782496
rs58598764	chr5:89888563	0.95814	A	C	0.84	1.01(0.78,1.31)	0.8244
rs491241	chr5:89888600	0.99433	T	C	0.71	1.19(0.97,1.47)	0.0707188
rs11957754	chr5:89888798	0.994452	C	T	0.99	2.78(1.07,7.19)	0.239315
rs171630	chr5:89889392	0.969301	C	T	0.61	1.27(1.04,1.55)	0.0149883
rs145850407	chr5:89889393	0.970957	G	A	0.08	1.36(1.96,0.94)	0.100974
rs62375073	chr5:89889650	0.845059	T	C	0.15	1.06(1.38,0.81)	0.753574

SNP	Chr:Position(bp)	info	Risk Allele	Other allele	RAF	OR (95% CI)	P
rs250369	chr5:89890131	0.97973	C	G	0.63	1.2(0.98,1.47)	0.0605701
rs11955985	chr5:89890197	0.989456	T	G	0.99	2.81(1.09,7.27)	0.22199
rs28399862	chr5:89890234	0.99593	T	C	0.92	1.07(0.75,1.53)	0.666172
rs250371	chr5:89890485	0.975279	T	C	0.17	1.02(1.32,0.79)	0.974216
rs28429067	chr5:89890555	0.84387	C	T	0.95	1.1(0.71,1.71)	0.600632
rs191486470	chr5:89891652	0.952384	G	C	0.9	1.19(0.86,1.63)	0.469146
rs173661	chr5:89891927	0.8003	C	T	0.61	1.26(1.04,1.53)	0.00981713
rs34260190	chr5:89892370	0.834095	C	A	0.94	1.29(0.86,1.93)	0.278488
rs138105777	chr5:89892467	0.91434	C	T	0.03	1.42(2.57,0.79)	0.155278
rs138811677	chr5:89892774	0.907119	A	G	0.02	1.86(3.75,0.92)	0.060632
rs17613704	chr5:89893336	0.898312	C	T	0.03	1.44(2.64,0.79)	0.253535
rs17541517	chr5:89894201	1	T	C	0.92	1.08(0.75,1.54)	0.652455
rs73179739	chr5:89894313	0.985031	C	T	0.99	2.93(1.14,7.51)	0.177748
rs27197	chr5:89894564	0.99964	C	T	0.56	1.28(1.06,1.56)	0.0102029
rs27196	chr5:89895019	0.998427	A	T	0.54	1.19(0.98,1.44)	0.0444804
rs149321	chr5:89895303	1	T	C	0.55	1.24(1.02,1.51)	0.0134574
rs152715	chr5:89895786	0.994636	T	C	0.16	1.01(1.31,0.78)	0.91549
rs11738461	chr5:89896125	1	T	C	0.92	1.08(0.75,1.54)	0.652455
rs617379	chr5:89896143	0.999884	G	C	0.71	1.18(0.96,1.46)	0.0833057
rs34301936	chr5:89896758	1	A	G	0.92	1.08(0.75,1.54)	0.652455
rs154558	chr5:89896791	0.993952	T	C	0.83	1.07(0.83,1.37)	0.565971
rs154557	chr5:89897229	1	G	A	0.83	1.06(0.82,1.36)	0.599426
rs11745908	chr5:89897340	1	T	A	0.92	1.08(0.75,1.54)	0.652455
rs154556	chr5:89898169	0.998769	G	A	0.83	1.03(0.79,1.33)	0.753087
rs17541704	chr5:89899019	1	A	G	0.92	1.08(0.75,1.54)	0.652455
rs73179758	chr5:89899638	0.932724	A	G	0.99	2.12(0.88,5.1)	0.289025
rs28480058	chr5:89899704	0.992325	T	C	0.52	1.23(1.02,1.49)	0.0327574
chr5:89899711:D	chr5:89899711	0.978165	G	GCA	0.7	1.19(0.97,1.47)	0.060734
rs187278	chr5:89899982	0.995192	T	A	0.83	1.05(0.82,1.35)	0.644161
rs60195623	chr5:89901283	0.864027	A	G	0.99	2.36(0.93,6.01)	0.209057
rs502596	chr5:89901315	1	G	A	0.71	1.18(0.96,1.46)	0.0838663
chr5:89901748:I	chr5:89901748	0.863987	GA	G	0.91	1.11(0.79,1.55)	0.46787
rs10514330	chr5:89901848	1	T	C	0.92	1.09(0.76,1.56)	0.602232
rs257848	chr5:89901944	0.996703	G	A	0.83	1.03(0.79,1.32)	0.763016
rs637442	chr5:89902846	0.999286	G	T	0.71	1.18(0.96,1.46)	0.080146
rs257847	chr5:89902952	1	A	G	0.56	1.3(1.07,1.57)	0.0075006
rs68096329	chr5:89905197	0.994345	C	T	0.17	1.02(1.32,0.78)	0.951799
rs10044044	chr5:89905562	0.999294	G	A	0.83	1.05(0.82,1.36)	0.620837
rs9293546	chr5:89906540	1	A	G	0.83	1.03(0.8,1.33)	0.724621
rs17541912	chr5:89906863	0.999489	C	A	0.92	1.09(0.76,1.56)	0.605329
rs490812	chr5:89907016	0.999296	C	A	0.62	1.24(1.02,1.51)	0.0285838
rs185504733	chr5:89907957	0.908212	G	T	0.02	1.95(3.95,0.96)	0.0485682
rs1700511	chr5:89908392	0.999769	A	G	0.71	1.19(0.96,1.46)	0.0760732
rs1673381	chr5:89908521	0.970958	A	G	0.54	1.3(1.07,1.58)	0.00605457
rs16868839	chr5:89908590	1	T	C	0.83	1.02(0.79,1.32)	0.78084
rs190085228	chr5:89908765	0.908212	T	C	0.02	1.95(3.95,0.96)	0.0485682
rs640042	chr5:89908905	1	T	C	0.94	1.06(0.7,1.6)	0.707528
rs60651883	chr5:89909272	1	G	A	0.83	1.02(0.79,1.32)	0.78084
rs17542086	chr5:89909493	0.999482	A	C	0.92	1.09(0.76,1.56)	0.605374
rs649449	chr5:89909779	0.999552	C	A	0.56	1.29(1.07,1.57)	0.00765838
rs116207662	chr5:89910249	0.878462	A	T	0.94	1.18(0.79,1.76)	0.374521
rs13167847	chr5:89910250	0.876045	A	T	0.94	1.18(0.79,1.76)	0.369904
rs116714351	chr5:89910251	0.878462	A	T	0.94	1.18(0.79,1.76)	0.374521
rs3112773	chr5:89911824	0.999552	T	C	0.56	1.29(1.07,1.57)	0.00765838
rs13158577	chr5:89912407	0.97768	G	A	0.98	1.45(0.69,3.05)	0.325845
rs13159048	chr5:89912649	0.999482	T	C	0.92	1.09(0.76,1.56)	0.605374
rs10462487	chr5:89914614	0.987536	C	T	0.17	1.04(1.35,0.8)	0.897346
rs1673379	chr5:89914687	0.995689	G	A	0.7	1.25(1.01,1.53)	0.045168
rs41311333	chr5:89914925	0.864655	T	G	0.05	1.15(1.84,0.72)	0.580211
rs146198134	chr5:89915573	0.976004	C	T	0.08	1.31(1.88,0.91)	0.172115
rs150744447	chr5:89915939	0.958773	A	G	0.84	1.02(0.78,1.32)	0.820026
rs36164590	chr5:89916129	0.848626	A	G	0.88	1.03(0.77,1.39)	0.751955
rs9283788	chr5:89916261	0.986519	A	G	0.3	1.23(1.51,1)	0.0431282
rs9283789	chr5:89916269	0.980496	T	C	0.35	1.23(1.51,1.01)	0.0305985

SNP	Chr:Position(bp)	info	Risk Allele	Other allele	RAF	OR (95% CI)	P
rs71637308	chr5:89916574	0.999162	T	C	0.92	1.09(0.76,1.55)	0.606542
rs145678506	chr5:89917287	0.916823	G	A	0.01	1.88(5.29,0.67)	0.212286
rs7709691	chr5:89918232	0.999976	A	C	0.83	1.02(0.79,1.32)	0.780478
rs16868864	chr5:89918334	0.976345	G	T	0.08	1.34(1.92,0.93)	0.124362
rs6891275	chr5:89918704	0.939705	T	C	0.34	1.11(1.36,0.91)	0.235709
rs6891287	chr5:89918725	0.990043	T	G	0.48	1.24(1.5,1.02)	0.0284906
rs6870337	chr5:89918813	0.931099	G	C	0.31	1.17(1.44,0.95)	0.0922348
rs4916803	chr5:89919005	0.999976	G	A	0.83	1.02(0.79,1.32)	0.780478
rs75900541	chr5:89919187	0.886843	A	G	0.98	1.4(0.72,2.75)	0.262901
rs4916804	chr5:89919243	0.999734	A	T	0.83	1.02(0.79,1.32)	0.777916
rs1316970	chr5:89919363	0.99084	G	C	0.48	1.24(1.51,1.03)	0.0254172
rs4916805	chr5:89919404	0.999795	A	G	0.83	1.02(0.79,1.32)	0.781203
rs66977476	chr5:89919820	0.99905	G	A	0.83	1.02(0.79,1.32)	0.785366
rs72782722	chr5:89919826	0.995481	G	A	0.83	1.02(0.79,1.31)	0.805063
rs11954050	chr5:89919850	0.982035	A	T	0.99	2.43(0.98,6.07)	0.30822
rs41311331	chr5:89921116	0.982058	C	T	0.99	2.43(0.98,6.07)	0.307989
rs113976155	chr5:89921510	0.907469	A	C	0.16	1.02(1.33,0.78)	0.949363
rs111421188	chr5:89921519	0.92977	A	G	0.85	1.01(0.77,1.33)	0.854843
rs111653093	chr5:89921660	0.999299	G	A	0.83	1(0.78,1.29)	0.923129
rs7726624	chr5:89921811	0.992337	C	G	0.29	1.19(1.47,0.97)	0.0644546
rs6891840	chr5:89922060	0.998017	G	T	0.44	1.3(1.57,1.07)	0.00732946
rs11749693	chr5:89922077	0.992112	G	A	0.92	1.09(0.76,1.55)	0.59949
rs6892132	chr5:89922445	1	A	G	0.44	1.3(1.57,1.07)	0.0075006
rs60359999	chr5:89922763	0.999972	A	G	0.83	1.02(0.79,1.32)	0.780441
rs45478296	chr5:89923667	0.999044	T	A	0.83	1.02(0.79,1.32)	0.780131
rs66683260	chr5:89923923	0.997414	T	C	0.92	1.09(0.76,1.55)	0.611726
rs2366771	chr5:89924976	0.998755	C	A	0.29	1.18(1.46,0.96)	0.086072
rs56360530	chr5:89925410	0.999941	T	C	0.83	1.02(0.79,1.32)	0.780139
rs55918113	chr5:89925578	0.995199	G	A	0.83	1.02(0.79,1.32)	0.774622
rs4574581	chr5:89925895	0.968825	G	A	0.3	1.22(1.5,0.99)	0.0353823
rs56002033	chr5:89926009	0.976954	C	T	0.84	1.04(0.8,1.34)	0.699594
rs12653924	chr5:89927354	0.999964	A	G	0.44	1.3(1.57,1.07)	0.00748818
chr5:89927880:D	chr5:89927880	0.999757	G	GT	0.83	1.02(0.79,1.32)	0.781628
rs4916807	chr5:89928238	0.999898	A	G	0.83	1.02(0.79,1.32)	0.779724
rs10072105	chr5:89928375	0.998863	T	C	0.29	1.19(1.46,0.96)	0.0746282
rs17614588	chr5:89928991	0.997408	G	A	0.92	1.09(0.76,1.55)	0.61177
rs10462488	chr5:89929066	0.999891	G	C	0.83	1.02(0.79,1.32)	0.779648
chr5:89929175:D	chr5:89929175	0.991664	CTT	C	0.17	1(1.3,0.77)	0.910476
chr5:89929215:I	chr5:89929215	0.989588	T	TA	0.3	1.16(1.43,0.94)	0.114773
rs10462489	chr5:89929352	0.970699	G	A	0.84	1.03(0.8,1.34)	0.706462
rs2366772	chr5:89929903	0.999873	A	G	0.45	1.29(1.57,1.06)	0.00855722
chr5:89930623:D	chr5:89930623	0.998956	C	CAGTT	0.83	1.02(0.79,1.32)	0.779263
chr5:89930628:D	chr5:89930628	0.857627	A	AG	0.87	1.01(0.76,1.35)	0.864749
chr5:89930940	chr5:89930940	0.907577	G	A	0.02	1.95(3.96,0.96)	0.0480107
chr5:89931159:D	chr5:89931159	0.994042	C	CA	0.83	1.03(0.8,1.33)	0.719267
rs57437217	chr5:89931662	0.99986	G	T	0.83	1.02(0.79,1.32)	0.779347
rs16868892	chr5:89932319	0.999853	G	T	0.83	1.02(0.79,1.32)	0.779271
rs6878917	chr5:89932354	0.998877	G	A	0.29	1.18(1.46,0.96)	0.0916042
rs16868894	chr5:89932411	0.999853	T	C	0.83	1.02(0.79,1.32)	0.779271
rs6883872	chr5:89933134	0.999026	C	T	0.29	1.19(1.46,0.96)	0.0750702
rs17542921	chr5:89933196	0.997074	G	A	0.93	1.07(0.74,1.54)	0.674609
chr5:89933287:D	chr5:89933287	0.989913	TAG	T	0.36	1.22(1.5,1)	0.048883
rs182990046	chr5:89933637	0.907565	G	A	0.02	1.95(3.96,0.96)	0.0479993
rs6889828	chr5:89934074	0.999359	A	G	0.83	1.02(0.79,1.32)	0.784778
rs4916680	chr5:89934428	0.99669	C	T	0.83	1.03(0.8,1.33)	0.723206
rs13182944	chr5:89934796	0.996208	A	G	0.93	1.05(0.73,1.51)	0.747243
rs68045375	chr5:89934843	0.999805	C	A	0.83	1.02(0.79,1.32)	0.778781
rs4588621	chr5:89934964	0.999813	A	G	0.45	1.29(1.57,1.06)	0.00854413
rs9790907	chr5:89935158	0.998976	T	C	0.29	1.19(1.46,0.96)	0.0750505
rs59496152	chr5:89935586	0.999231	C	T	0.83	1.02(0.79,1.32)	0.771114
rs58132310	chr5:89936075	0.998987	G	A	0.83	1.02(0.79,1.32)	0.763003
rs7701844	chr5:89936235	0.998988	G	A	0.83	1.02(0.79,1.32)	0.760359
rs7707357	chr5:89937312	0.999948	G	A	0.83	1.03(0.8,1.33)	0.743538
rs5028525	chr5:89937327	1	T	G	0.45	1.27(1.55,1.05)	0.00992478

SNP	Chr:Position(bp)	info	Risk Allele	Other allele	RAF	OR (95% CI)	P
rs67556741	chr5:89937430	0.999972	G	A	0.83	1.03(0.8,1.33)	0.743318
rs11952742	chr5:89937694	1	C	T	0.83	1.02(0.79,1.31)	0.808977
rs11955749	chr5:89937719	0.999972	C	A	0.83	1.03(0.8,1.33)	0.743318
rs1344030	chr5:89938434	0.99458	T	G	0.83	1.03(0.8,1.33)	0.742607
rs2366773	chr5:89938587	0.994315	C	T	0.29	1.19(1.46,0.96)	0.0742765
rs2366774	chr5:89939067	0.946421	G	A	0.29	1.19(1.47,0.97)	0.0636535
rs2366775	chr5:89940036	0.950251	G	C	0.44	1.27(1.54,1.04)	0.0101612
rs16868901	chr5:89940489	1	C	G	0.17	1.02(1.31,0.79)	0.808977
chr5:89940739:D	chr5:89940739	0.973119	A	AT	0.92	1.08(0.75,1.54)	0.625266
rs148779191	chr5:89940905	0.913277	G	C	0.01	1.63(4.35,0.61)	0.312148
rs34774423	chr5:89941240	0.991523	T	A	0.93	1.1(0.77,1.58)	0.556234
rs72782749	chr5:89941244	0.830677	C	T	0.04	1.33(2.16,0.82)	0.21239
rs17543150	chr5:89942313	0.993527	A	C	0.93	1.08(0.75,1.56)	0.617116
rs16868906	chr5:89942405	0.964259	C	T	0.15	1.04(1.37,0.79)	0.814636
chr5:89942586:I	chr5:89942586	0.969383	C	CT	0.14	1.03(1.35,0.78)	0.888551
chr5:89942587:D	chr5:89942587	0.96861	TTC	T	0.14	1.04(1.36,0.79)	0.854403
rs4916808	chr5:89942791	0.976118	A	G	0.35	1.2(1.47,0.98)	0.0766895
rs950692	chr5:89943433	0.978944	A	G	0.15	1.04(1.37,0.79)	0.830154
rs2366777	chr5:89943571	1	G	T	0.31	1.16(1.43,0.95)	0.0993161
rs16868917	chr5:89943655	0.921542	A	G	0.08	1.25(1.79,0.87)	0.193604
rs13164404	chr5:89943691	0.995052	T	C	0.93	1.08(0.75,1.56)	0.617676
rs12186867	chr5:89943699	0.991894	G	A	0.45	1.22(1.49,1.01)	0.0281312
rs16868924	chr5:89943938	0.981941	T	C	0.15	1.03(1.36,0.79)	0.876752
rs16868927	chr5:89944053	0.981148	T	C	0.15	1.03(1.35,0.78)	0.897495
rs10056775	chr5:89944311	0.988918	T	C	0.92	1.24(0.88,1.75)	0.251161
rs6898209	chr5:89944514	0.998829	T	C	0.32	1.15(1.42,0.94)	0.122853
rs191710706	chr5:89944946	0.847867	G	A	0.03	1.42(2.61,0.77)	0.268904
rs9293549	chr5:89945088	0.973656	G	T	0.36	1.21(1.48,0.99)	0.0620636
rs11951632	chr5:89945222	0.983473	G	A	0.15	1.03(1.36,0.79)	0.870603
chr5:89945880:D	chr5:89945880	0.984228	CCTTT	C	0.15	1.03(1.36,0.79)	0.871079
rs115002957	chr5:89946841	0.959782	A	G	0.98	1.3(0.69,2.45)	0.364726
rs190062495	chr5:89946934	0.898746	A	G	0.03	1.93(3.86,0.96)	0.0462919
rs72656677	chr5:89947020	0.985269	G	T	0.15	1.03(1.36,0.79)	0.869839
chr5:89947211:D	chr5:89947211	0.928824	TCTC	T	0.42	1.2(1.46,0.99)	0.041736
rs908818	chr5:89947247	0.991376	C	T	0.45	1.23(1.49,1.01)	0.0257669
rs13186124	chr5:89947695	0.995811	A	G	0.93	1.08(0.75,1.56)	0.61557
rs16868932	chr5:89947999	0.986214	T	C	0.15	1.03(1.36,0.79)	0.868371
rs10043564	chr5:89950272	1	A	G	0.46	1.19(1.44,0.98)	0.050234
rs41302840	chr5:89950570	0.998821	G	A	0.93	1.08(0.75,1.56)	0.614551
rs41308295	chr5:89950752	0.992712	G	A	0.14	1.04(1.37,0.79)	0.916084
rs10058765	chr5:89950787	0.998082	G	A	0.49	1.15(1.39,0.95)	0.131201
rs10060427	chr5:89951112	0.998353	G	A	0.49	1.15(1.39,0.95)	0.129454
rs61525440	chr5:89951441	0.994408	C	T	0.85	1.04(0.8,1.36)	0.658891
rs116551415	chr5:89951538	0.995747	G	T	0.09	1.29(1.84,0.9)	0.170796
rs11748911	chr5:89951585	1	C	A	0.93	1.06(0.74,1.53)	0.681859
rs12516985	chr5:89951749	0.998685	C	T	0.49	1.16(1.41,0.96)	0.111137
rs35667071	chr5:89951753	1	T	C	0.93	1.09(0.75,1.56)	0.611989
rs17621038	chr5:89952002	1	A	G	0.93	1.09(0.75,1.56)	0.611989
rs17621093	chr5:89952505	0.994326	G	A	0.09	1.24(1.76,0.87)	0.22177
rs10062714	chr5:89952531	1	C	T	0.49	1.17(1.41,0.96)	0.107298
rs10062924	chr5:89952801	1	C	A	0.49	1.17(1.41,0.96)	0.107298
rs10064606	chr5:89952979	0.997596	G	A	0.37	1.16(1.42,0.95)	0.0812227
rs28550273	chr5:89952997	0.994713	A	C	0.49	1.16(1.41,0.96)	0.105077
rs963866	chr5:89953175	0.998394	C	T	0.32	1.16(1.42,0.94)	0.118165
rs4379236	chr5:89953281	1	A	G	0.49	1.17(1.41,0.96)	0.107298
rs17621160	chr5:89953550	0.997695	A	T	0.93	1.09(0.76,1.57)	0.603563
rs17543819	chr5:89953849	0.996176	C	T	0.08	1.28(1.83,0.9)	0.190818
rs11958484	chr5:89954184	0.997843	T	C	0.37	1.17(1.44,0.96)	0.065726
rs16868957	chr5:89955125	1	T	G	0.49	1.17(1.41,0.96)	0.107298
rs62375083	chr5:89955691	0.947466	T	C	0.36	1.23(1.51,1.01)	0.0253523
rs4916809	chr5:89956765	1	A	G	0.86	1.01(0.76,1.33)	0.838113
rs4916810	chr5:89956783	0.998689	T	A	0.86	1.01(0.76,1.33)	0.834131
rs4916811	chr5:89956850	0.944242	A	G	0.49	1.14(1.39,0.94)	0.129933
rs63733862	chr5:89956851	0.935391	A	G	0.34	1.16(1.43,0.95)	0.0947723

SNP	Chr:Position(bp)	info	Risk Allele	Other allele	RAF	OR (95% CI)	P
rs4916813	chr5:89956966	1	T	C	0.32	1.14(1.4,0.93)	0.144515
rs4916681	chr5:89957017	1	C	T	0.49	1.13(1.37,0.93)	0.188508
rs4916814	chr5:89957090	0.99867	G	T	0.31	1.14(1.4,0.93)	0.144235
rs4916682	chr5:89957193	0.998521	C	T	0.31	1.14(1.4,0.93)	0.144924
rs62375094	chr5:89957362	0.996098	A	G	0.37	1.15(1.4,0.94)	0.127138
rs34490215	chr5:89957437	0.996711	A	G	0.93	1.09(0.76,1.58)	0.586371
rs115104361	chr5:89957759	0.875357	T	C	0.03	1.34(2.39,0.75)	0.357569
rs1988788	chr5:89958001	0.988853	C	G	0.49	1.11(1.34,0.91)	0.209542
rs145335577	chr5:89958156	0.979324	G	A	0.09	1.22(1.72,0.86)	0.243523
rs60562826	chr5:89958285	0.997837	T	C	0.86	1.01(0.77,1.33)	0.844444
rs12187830	chr5:89958570	0.989093	C	T	0.49	1.12(1.35,0.92)	0.220146
rs189686513	chr5:89960737	0.942855	T	G	0.47	1.05(1.27,0.87)	0.492476
rs180879610	chr5:89960738	0.901579	G	A	0.89	1.19(0.87,1.61)	0.173398
rs138904946	chr5:89960774	0.994646	T	A	0.09	1.31(1.86,0.91)	0.152254
rs189164060	chr5:89961325	0.94377	T	G	0.35	1.12(1.37,0.91)	0.209593
rs6878447	chr5:89962332	0.950963	C	T	0.5	1.09(1.32,0.9)	0.31739
rs6878918	chr5:89962438	0.838872	G	A	0.41	1.02(1.24,0.84)	0.770011
rs139872473	chr5:89962610	0.992002	T	C	0.09	1.25(1.78,0.88)	0.198483
rs190930957	chr5:89963215	0.97623	T	C	0.09	1.15(1.61,0.82)	0.361536
rs10942603	chr5:89963686	0.893997	G	A	0.33	1.14(1.4,0.93)	0.159221
rs11739825	chr5:89964089	0.872663	C	T	0.91	1.1(0.79,1.53)	0.501279
rs147437156	chr5:89964222	0.969152	A	C	0.14	1(1.32,0.76)	0.874236
rs11959408	chr5:89964298	0.911217	C	T	0.35	1.13(1.38,0.92)	0.17169
chr5:89964381:I	chr5:89964381	0.801954	C	CCGTCTTCTG	0.04	1.52(2.55,0.91)	0.0684044
rs7729205	chr5:89964417	0.882751	G	C	0.9	1.19(0.87,1.62)	0.188757
rs7729644	chr5:89964553	0.997314	T	G	0.86	1.01(0.77,1.33)	0.822485
rs11739958	chr5:89964571	0.886313	C	T	0.91	1.12(0.81,1.56)	0.396048
rs7729703	chr5:89964738	0.936714	C	G	0.3	1.09(1.34,0.88)	0.331195
rs6861067	chr5:89966285	0.985868	G	A	0.38	1.09(1.33,0.89)	0.262016
rs6861525	chr5:89966759	0.980453	C	T	0.48	1.09(1.32,0.9)	0.255667
rs74938671	chr5:89966896	0.990665	A	C	0.09	1.21(1.71,0.86)	0.256688
rs6866075	chr5:89967029	0.887866	A	C	0.9	1.18(0.86,1.63)	0.255337
rs11955565	chr5:89967158	0.993479	A	G	0.23	1.09(1.37,0.87)	0.500503
chr5:89967949:I	chr5:89967949	0.858202	CT	C	0.91	1.14(0.82,1.59)	0.356175
rs35680172	chr5:89968089	0.886772	C	A	0.91	1.13(0.81,1.56)	0.39365
rs1028191	chr5:89968626	1	T	A	0.68	1.14(0.93,1.4)	0.144515
rs1028192	chr5:89968773	0.927485	G	A	0.29	1.11(1.37,0.9)	0.237356
rs6875517	chr5:89970677	0.991433	C	T	0.85	1(0.76,1.32)	0.878516
rs75935830	chr5:89972731	0.994464	T	G	0.08	1.28(1.83,0.9)	0.200923
rs58582125	chr5:89972806	0.994176	T	C	0.84	1.17(0.9,1.51)	0.274852
rs16868964	chr5:89973018	0.995501	A	G	0.85	1.07(0.82,1.4)	0.628747
rs1033290	chr5:89973621	0.945764	C	A	0.3	1.14(1.4,0.93)	0.155079
rs10514331	chr5:89974082	0.995861	A	G	0.85	1.07(0.82,1.41)	0.62741
rs7716829	chr5:89974638	1	C	A	0.36	1.22(1.49,0.99)	0.0414412
rs7703530	chr5:89975009	0.968481	T	C	0.31	1.15(1.41,0.93)	0.138971
rs7721027	chr5:89975012	0.996092	G	A	0.84	1.17(0.9,1.51)	0.27784
rs7721198	chr5:89975085	0.970307	A	G	0.31	1.15(1.41,0.93)	0.136583
rs6863710	chr5:89975629	1	T	A	0.32	1.16(1.42,0.94)	0.120651
rs17544471	chr5:89975780	0.995085	T	C	0.08	1.29(1.84,0.9)	0.189599
rs55835641	chr5:89975818	0.994652	G	A	0.36	1.22(1.5,1)	0.0346033
rs17621772	chr5:89975894	0.994691	C	A	0.08	1.28(1.83,0.89)	0.201137
rs56255871	chr5:89975996	0.999321	G	T	0.36	1.21(1.48,0.99)	0.0454299
rs62375106	chr5:89976169	0.997649	A	C	0.36	1.21(1.48,0.99)	0.0442682
rs16868970	chr5:89976575	0.991634	T	C	0.14	1(1.32,0.76)	0.887177
rs62375107	chr5:89977598	0.800547	G	A	0.13	1.15(1.53,0.86)	0.350147
rs10045394	chr5:89977642	0.949648	A	C	0.35	1.19(1.45,0.97)	0.063379
chr5:89979027:D	chr5:89979027	0.981025	T	TTTTA	0.85	1.08(0.83,1.41)	0.591025
rs75880085	chr5:89979134	0.849064	C	G	0.01	3.02(10.41,0.88)	0.0403448
rs4916683	chr5:89979380	0.99979	T	C	0.36	1.21(1.48,0.99)	0.0461467
rs17544552	chr5:89979518	0.994873	C	T	0.08	1.29(1.84,0.9)	0.188611
rs4916684	chr5:89979589	0.999303	G	A	0.32	1.15(1.42,0.94)	0.12982
rs41303352	chr5:89979691	0.997745	G	A	0.85	1.09(0.83,1.43)	0.549047
rs4916685	chr5:89979698	0.999981	C	T	0.36	1.21(1.48,0.99)	0.0458658
rs16868972	chr5:89979750	0.997906	T	G	0.84	1.18(0.91,1.53)	0.232523

SNP	Chr:Position(bp)	info	Risk Allele	Other allele	RAF	OR (95% CI)	P
rs6880239	chr5:89980365	0.99979	A	G	0.36	1.21(1.48,0.99)	0.0461467
rs36045161	chr5:89980787	0.998638	T	A	0.93	1.08(0.75,1.56)	0.636309
chr5:89981024:D	chr5:89981024	0.997046	T	TGTTTA	0.85	1.09(0.83,1.42)	0.553058
rs28517617	chr5:89981135	0.999046	T	C	0.32	1.16(1.42,0.95)	0.125184
chr5:89981457:D	chr5:89981457	0.998653	A	AAAAT	0.93	1.08(0.75,1.56)	0.636454
rs10062423	chr5:89981980	0.838744	T	G	0.89	1.18(0.87,1.61)	0.409194
rs57255621	chr5:89982179	0.857462	T	C	0.98	1.76(0.86,3.61)	0.117935
rs147329258	chr5:89982515	0.875972	A	T	0.95	1.22(0.81,1.85)	0.191575
rs56816222	chr5:89983056	0.973083	T	G	0.92	1.16(0.82,1.65)	0.469724
rs13171056	chr5:89983349	0.998675	G	T	0.93	1.06(0.73,1.53)	0.708253
rs145501488	chr5:89983479	0.917789	C	A	0.08	1.22(1.77,0.85)	0.266638
rs192589194	chr5:89983780	0.999343	G	A	0.36	1.22(1.49,1)	0.0391976
rs146520661	chr5:89983903	0.873391	G	A	0.35	1.21(1.48,0.98)	0.0372547
rs16868980	chr5:89984881	1	T	C	0.36	1.21(1.48,0.99)	0.0457872
rs10037067	chr5:89985882	0.999878	A	G	0.36	1.22(1.5,1)	0.0352242
rs114172556	chr5:89986285	0.989113	T	G	0.98	1.18(0.64,2.19)	0.536498
rs73193232	chr5:89986519	0.998506	C	G	0.99	2.77(1.07,7.18)	0.045499
rs17621985	chr5:89986963	0.998599	G	T	0.93	1.08(0.75,1.56)	0.638242
rs16868985	chr5:89987053	0.998322	G	A	0.86	1.02(0.78,1.34)	0.785793
chr5:89987131:D	chr5:89987131	0.989118	CAT	C	0.36	1.24(1.51,1.01)	0.0223551
rs77332009	chr5:89988086	0.988474	A	T	0.41	1.09(1.32,0.89)	0.294961
rs10040165	chr5:89988412	0.997382	G	A	0.36	1.22(1.49,0.99)	0.0412419
rs193030567	chr5:89988464	0.901307	A	T	0.03	1.76(3.42,0.91)	0.0723276
rs2366926	chr5:89988504	0.999588	A	G	0.36	1.22(1.49,1)	0.0389777
rs10043836	chr5:89989158	0.9991	T	C	0.32	1.15(1.42,0.94)	0.129534
rs10035335	chr5:89989208	0.987347	A	G	0.36	1.22(1.5,1)	0.0347769
rs16876822	chr5:89989779	0.999371	G	A	0.36	1.22(1.49,1)	0.0387026
rs1878878	chr5:89990324	0.998019	A	G	0.31	1.14(1.4,0.93)	0.148087
rs10068473	chr5:89990545	0.998634	C	A	0.48	1.09(1.32,0.9)	0.268988
rs977906	chr5:89991389	0.995312	C	A	0.31	1.14(1.4,0.93)	0.156041
chr5:89991502:D	chr5:89991502	0.997179	A	AAC	0.85	1.1(0.84,1.44)	0.512512
rs55720814	chr5:89992204	0.997026	T	C	0.85	1.1(0.84,1.44)	0.513625
rs970319	chr5:89992339	0.997199	T	C	0.36	1.22(1.5,1)	0.0378604
chr5:89992596:I	chr5:89992596	0.977577	T	TAC	0.09	1.27(1.81,0.89)	0.207909
rs10942604	chr5:89993045	0.99847	T	C	0.36	1.21(1.48,0.99)	0.0429017
rs12188928	chr5:89993332	0.9952	C	T	0.48	1.09(1.32,0.9)	0.249398
rs73772469	chr5:89993742	0.99729	C	T	0.86	1.03(0.78,1.35)	0.747226
rs16868999	chr5:89994527	0.9983	G	A	0.36	1.21(1.48,0.99)	0.0428243
chr5:89994596:D	chr5:89994596	0.995154	GA	G	0.41	1.06(1.29,0.87)	0.457586
rs189135502	chr5:89994764	0.901871	G	C	0.03	1.75(3.4,0.9)	0.0750903
rs116533328	chr5:89994810	0.987055	C	T	0.98	1.14(0.62,2.1)	0.634204
rs12187771	chr5:89994983	0.894288	A	G	0.21	1.28(1.63,1.01)	0.0199666
rs142034716	chr5:89995607	0.911846	C	T	0.01	1.76(4.65,0.67)	0.232762
rs16869010	chr5:89995656	0.996531	A	T	0.36	1.21(1.48,0.99)	0.0433269
rs17622455	chr5:89995803	0.994392	A	G	0.08	1.26(1.8,0.88)	0.231304
rs16869013	chr5:89996329	0.996261	A	G	0.85	1.1(0.84,1.44)	0.513144
rs1878881	chr5:89996625	0.996141	A	C	0.85	1.1(0.84,1.44)	0.513221
rs67956713	chr5:89996684	0.997695	A	G	0.93	1.08(0.75,1.55)	0.648538
rs17622555	chr5:89996966	0.997735	A	T	0.93	1.08(0.75,1.55)	0.649243
rs10514332	chr5:89997048	0.994495	A	T	0.08	1.26(1.8,0.88)	0.231641
rs6452904	chr5:89997358	0.998877	T	C	0.32	1.15(1.41,0.93)	0.136219
rs6867004	chr5:89997506	0.988877	T	G	0.84	1.21(0.94,1.56)	0.165925
rs6452905	chr5:89997538	0.99891	T	G	0.32	1.15(1.41,0.93)	0.135914
rs77198275	chr5:89997634	0.936238	C	T	0.08	1.19(1.71,0.83)	0.322842
rs115766415	chr5:89997807	0.951872	C	T	0.97	1.29(0.72,2.33)	0.334858
rs192706921	chr5:89998242	0.903572	A	G	0.03	1.73(3.35,0.89)	0.080262
rs13174505	chr5:89998386	0.973098	T	A	0.92	1.16(0.82,1.64)	0.503726
rs10514333	chr5:89998454	0.997881	G	T	0.4	1.07(1.31,0.88)	0.353851
chr5:89998634:D	chr5:89998634	0.86642	CT	C	0.03	1.09(1.98,0.6)	0.810383
rs150429028	chr5:89998778	0.911399	T	A	0.01	1.76(4.65,0.67)	0.230756
rs76119383	chr5:89998957	0.869794	A	C	0.96	1.3(0.8,2.11)	0.222382
rs11959270	chr5:89999735	0.9948	A	G	0.86	1.03(0.78,1.35)	0.744766
rs10942605	chr5:89999954	1	T	C	0.4	1.08(1.32,0.89)	0.30935
rs10942606	chr5:89999968	1	A	G	0.32	1.16(1.42,0.94)	0.121757

SNP	Chr:Position(bp)	info	Risk Allele	Other allele	RAF	OR (95% CI)	P
rs16869016	chr5:90000210	0.868056	T	C	0.96	1.3(0.8,2.1)	0.228191
rs56369423	chr5:90000549	0.992487	C	A	0.36	1.22(1.5,1)	0.0339294
rs6894453	chr5:90000720	0.971817	C	A	0.36	1.23(1.5,1)	0.0294978
rs6880570	chr5:90001368	0.994683	G	T	0.85	1.1(0.84,1.43)	0.523501
rs764212	chr5:90001759	0.993949	G	C	0.08	1.26(1.81,0.88)	0.227612
rs1160121	chr5:90001988	0.998417	G	T	0.41	1.08(1.31,0.89)	0.364893
rs77614337	chr5:90002293	0.992488	A	G	0.08	1.26(1.8,0.88)	0.243913
rs1996551	chr5:90002596	1	C	T	0.86	1.02(0.78,1.34)	0.806054
rs1474119	chr5:90002885	0.993271	G	A	0.46	1.2(1.45,0.99)	0.0407203
rs1474120	chr5:90003053	0.994107	T	A	0.37	1.14(1.4,0.94)	0.139629
rs6869935	chr5:90003131	0.989911	A	G	0.16	1.13(1.48,0.87)	0.397655
rs2176624	chr5:90003274	0.992869	G	A	0.33	1.23(1.51,1.01)	0.0301043
rs4916686	chr5:90003594	0.997424	T	G	0.31	1.16(1.42,0.94)	0.113351
rs193121546	chr5:90003633	0.901186	C	T	0.03	1.72(3.3,0.89)	0.0821708
chr5:90003967:I	chr5:90003967	0.983705	C	CT	0.08	1.28(1.83,0.9)	0.195301
rs73193258	chr5:90004180	0.986056	T	C	0.09	1.07(1.49,0.77)	0.739168
rs73193259	chr5:90004474	0.978849	T	C	0.99	3(1.24,7.29)	0.0263454
rs62375135	chr5:90004977	0.824183	C	T	0.08	1.44(2.09,1)	0.0407014
rs2222242	chr5:90005214	0.997346	G	A	0.33	1.27(1.55,1.04)	0.0236634
rs28587199	chr5:90005534	0.995547	A	T	0.32	1.16(1.42,0.94)	0.115456
rs9293550	chr5:90005825	0.998689	G	A	0.32	1.16(1.43,0.95)	0.109949
rs1993474	chr5:90006552	0.992605	T	G	0.41	1.28(1.56,1.06)	0.0117986
rs7732169	chr5:90007196	0.995542	C	T	0.34	1.28(1.57,1.05)	0.0200104
rs7714506	chr5:90007288	0.990456	T	A	0.32	1.17(1.43,0.95)	0.100875
rs74836712	chr5:90008925	0.983003	G	A	0.08	1.28(1.83,0.9)	0.193266
chr5:90008979:I	chr5:90008979	0.858646	G	GT	0.45	1.01(1.22,0.83)	0.701579
rs10060365	chr5:90008988	0.971761	A	T	0.48	1.07(1.29,0.88)	0.334802
rs35312877	chr5:90009121	0.986868	A	G	0.93	1.04(0.72,1.5)	0.770899
rs6886668	chr5:90009248	0.968289	T	G	0.98	2.93(1.51,5.67)	0.00816381
rs13183331	chr5:90009927	0.986776	C	A	0.93	1.04(0.72,1.5)	0.774439
rs6891645	chr5:90009941	0.968293	T	C	0.98	2.93(1.51,5.67)	0.00647417
rs13165949	chr5:90010141	0.993928	C	T	0.99	1.48(0.68,3.22)	0.319391
rs10045236	chr5:90010486	0.999908	C	T	0.32	1.16(1.42,0.94)	0.12075
rs73175215	chr5:90011175	0.977777	A	T	0.99	3.01(1.24,7.31)	0.0251673
rs11960887	chr5:90011671	0.993462	A	G	0.85	1.01(0.77,1.33)	0.844454
rs10080113	chr5:90011696	0.968273	T	G	0.98	2.93(1.51,5.67)	0.00645967
rs7703588	chr5:90011949	0.999956	A	G	0.32	1.16(1.42,0.94)	0.120718
rs13157270	chr5:90012379	0.986776	A	G	0.93	1.04(0.72,1.5)	0.775575
rs10067181	chr5:90012614	0.996421	A	T	0.34	1.28(1.57,1.05)	0.0210468
rs3927265	chr5:90012703	0.999972	G	A	0.32	1.16(1.42,0.94)	0.120693
rs73175221	chr5:90012778	0.974095	G	A	0.99	3.02(1.25,7.32)	0.0241662
rs150073216	chr5:90013506	0.982318	T	C	0.08	1.28(1.83,0.9)	0.190746
rs13355001	chr5:90013833	0.968273	T	C	0.98	2.93(1.51,5.67)	0.00645934
rs76580665	chr5:90013856	0.98231	G	A	0.08	1.28(1.83,0.9)	0.190687
rs111456280	chr5:90013949	0.977558	A	G	0.99	3.01(1.24,7.31)	0.0250669
rs13167491	chr5:90014237	0.998244	C	A	0.32	1.16(1.42,0.94)	0.119
rs73175282	chr5:90014299	0.977695	G	A	0.99	3.01(1.24,7.31)	0.0250895
rs115956065	chr5:90014526	0.984092	C	A	0.98	3.46(1.6,7.48)	0.00819089
rs1014750	chr5:90014827	1	T	C	0.32	1.16(1.42,0.94)	0.120634
rs1568308	chr5:90014906	1	C	T	0.36	1.2(1.47,0.98)	0.0587103
rs3942393	chr5:90014936	0.999957	C	T	0.36	1.2(1.47,0.98)	0.0586172
chr5:90015128:D	chr5:90015128	0.981509	ACT	A	0.08	1.29(1.84,0.9)	0.185342
rs4364416	chr5:90015175	0.956878	C	T	0.98	2.92(1.51,5.62)	0.00570358
rs6881412	chr5:90015792	1	C	T	0.31	1.14(1.41,0.93)	0.135715
rs6452906	chr5:90016129	1	A	G	0.32	1.16(1.42,0.94)	0.120634
chr5:90016442:D	chr5:90016442	0.985721	A	AT	0.93	1.04(0.72,1.5)	0.786621
rs16869032	chr5:90016871	0.993376	A	G	0.86	1.01(0.76,1.32)	0.86018
rs139716327	chr5:90017060	0.97482	A	G	0.99	2.89(1.25,6.69)	0.0248698
rs146638672	chr5:90017092	0.956571	C	T	0.06	1.08(1.62,0.72)	0.763524
rs12657264	chr5:90017137	0.995057	T	C	0.32	1.19(1.46,0.97)	0.072406
rs35851488	chr5:90017178	0.953582	G	A	0.36	1.15(1.41,0.94)	0.132441
rs1533603	chr5:90017416	0.844706	T	G	0.38	1.1(1.34,0.9)	0.241051
chr5:90017469:I	chr5:90017469	0.994052	T	TG	0.42	1.19(1.44,0.98)	0.0881297
rs17552645	chr5:90017593	0.98603	C	T	0.93	1.04(0.72,1.5)	0.776632

SNP	Chr:Position(bp)	info	Risk Allele	Other allele	RAF	OR (95% CI)	P
rs17552645	chr5:90017593	0.98603	C	T	0.93	1.04(0.72,1.5)	0.776632
chr5:90017696:D	chr5:90017696	0.965895	CTG	C	0.49	1.21(1.47,1)	0.0397983
rs73175286	chr5:90018113	0.951487	T	C	0.98	2.81(1.49,5.3)	0.00518323
rs12652606	chr5:90018385	0.968493	G	A	0.33	1.16(1.42,0.95)	0.110057
rs10036258	chr5:90018414	0.958948	C	A	0.33	1.19(1.46,0.97)	0.063281
rs10036980	chr5:90019080	0.999874	C	A	0.32	1.16(1.42,0.94)	0.120757
rs76742855	chr5:90019854	0.981967	A	G	0.08	1.29(1.85,0.9)	0.178492
rs78971704	chr5:90020082	0.976529	T	C	0.05	1.53(2.49,0.94)	0.0950882
rs13171303	chr5:90020100	0.985979	T	G	0.93	1.04(0.72,1.5)	0.777026
rs62375138	chr5:90020192	0.825597	G	A	0.08	1.4(2.03,0.97)	0.0578176
rs13153584	chr5:90020283	0.972406	T	A	0.43	1.17(1.42,0.97)	0.107534
rs147798076	chr5:90020459	0.982943	G	A	0.99	1.54(0.68,3.47)	0.300483
rs7723259	chr5:90020868	0.999621	A	C	0.32	1.16(1.42,0.94)	0.121091
rs16869042	chr5:90020923	0.998977	T	G	0.36	1.2(1.47,0.98)	0.0589273
rs16869043	chr5:90021292	0.984671	A	G	0.14	1(1.32,0.76)	0.884513
rs4916815	chr5:90021499	0.810501	G	T	0.5	1.14(1.38,0.94)	0.146876
rs7724806	chr5:90021691	0.973172	C	T	0.32	1.17(1.43,0.95)	0.120045
rs28407508	chr5:90021905	0.975329	A	G	0.36	1.17(1.44,0.96)	0.0957766
rs17623602	chr5:90022341	0.962516	T	C	0.05	1.61(2.65,0.97)	0.0685637
rs10070074	chr5:90022407	1	T	C	0.38	1.03(1.25,0.84)	0.601863
rs7730204	chr5:90022584	1	T	G	0.79	1.04(0.82,1.31)	0.622318
rs4916687	chr5:90022712	1	A	G	0.2	1.02(1.3,0.8)	0.999231
rs4916688	chr5:90022780	0.98971	G	A	0.79	1.09(0.86,1.38)	0.401382
rs4916689	chr5:90022918	0.956491	T	C	0.22	1.03(1.31,0.82)	0.922876
rs4392662	chr5:90023335	0.864328	T	A	0.42	1.23(1.5,1.02)	0.0345084
rs988470	chr5:90023336	0.847056	T	A	0.35	1.29(1.57,1.06)	0.0117437
rs73772476	chr5:90024214	0.801409	G	A	0.06	1.11(1.69,0.72)	0.641714
rs10074104	chr5:90024277	0.943877	T	C	0.35	1.3(1.59,1.06)	0.0138739
rs2366928	chr5:90024735	0.884559	G	A	0.27	1.17(1.45,0.94)	0.0989217
rs73772478	chr5:90025171	0.802743	T	C	0.06	1.11(1.69,0.72)	0.641596
rs141197244	chr5:90025784	0.918712	T	G	0.99	5.31(1.84,15.38)	0.000312919
rs2030272	chr5:90025842	0.946549	C	G	0.44	1.26(1.53,1.04)	0.0143463
rs139635848	chr5:90025887	0.803888	G	A	0.06	1.11(1.69,0.72)	0.641047
rs4916816	chr5:90026142	0.859762	A	C	0.19	1.27(1.64,0.99)	0.032242
rs951116	chr5:90026261	0.894014	A	G	0.3	1.36(1.68,1.11)	0.00394824
rs2366929	chr5:90026634	0.909816	T	C	0.5	1.27(1.54,1.05)	0.00889703
rs4916690	chr5:90026688	0.999184	G	T	0.2	1.02(1.3,0.8)	0.989727
rs2443071	chr5:90026987	0.941692	T	C	0.36	1.29(1.58,1.06)	0.0160213
rs2203824	chr5:90027231	0.939894	C	A	0.98	3.46(1.81,6.62)	0.000504157
rs2203825	chr5:90027280	0.939886	A	T	0.98	3.46(1.81,6.62)	0.000504768
rs2030273	chr5:90027635	0.885679	G	T	0.28	1.19(1.47,0.96)	0.0833656
rs2460176	chr5:90027676	0.891457	A	T	0.3	1.35(1.66,1.1)	0.00540748
rs6452908	chr5:90028159	1	T	C	0.2	1.02(1.3,0.8)	0.979945
rs2460177	chr5:90028405	1	C	G	0.73	1.11(0.89,1.37)	0.270384
rs62373962	chr5:90029246	0.867847	G	A	0.05	1.06(1.66,0.68)	0.779292
rs2460178	chr5:90030185	0.880195	G	A	0.27	1.17(1.45,0.94)	0.0971332
rs1988093	chr5:90030442	0.930581	G	A	0.44	1.21(1.47,1)	0.0302062
rs72782766	chr5:90032844	0.986508	T	C	0.95	1.01(0.66,1.56)	0.886208
rs189367032	chr5:90032981	0.9167	T	A	0.03	1.51(2.77,0.83)	0.172783
rs2460181	chr5:90033213	0.943577	G	A	0.46	1.2(1.45,0.99)	0.0318534
chr5:90033232:D	chr5:90033232	0.919333	T	TAGA	0.99	5.28(1.84,15.12)	0.000296366
rs2460182	chr5:90033441	0.887435	G	A	0.31	1.14(1.4,0.92)	0.184867
rs2222243	chr5:90033697	0.946423	A	C	0.45	1.26(1.53,1.04)	0.0157191
chr5:90033762:D	chr5:90033762	0.936638	A	ACT	0.99	5.34(1.9,15.02)	0.000242106
rs990124	chr5:90033830	0.965102	G	C	0.47	1.21(1.46,0.99)	0.0270347
rs1988195	chr5:90033879	0.909254	T	G	0.27	1.21(1.5,0.97)	0.0604615
rs2443067	chr5:90034302	0.994878	A	G	0.46	1.29(1.56,1.06)	0.0098014
rs2460185	chr5:90034401	0.994024	C	T	0.46	1.28(1.56,1.06)	0.010739
rs2443066	chr5:90034784	0.995307	A	G	0.46	1.29(1.56,1.06)	0.0103708
rs62373963	chr5:90035887	0.929045	C	G	0.08	1.13(1.63,0.78)	0.5151
rs2222244	chr5:90036089	1	A	G	0.32	1.15(1.41,0.94)	0.161119
rs73177403	chr5:90036895	0.959527	T	C	0.99	5.38(1.94,14.9)	0.000228216
rs2460186	chr5:90036980	0.998649	A	G	0.46	1.29(1.56,1.06)	0.00992444
rs2460187	chr5:90038450	0.999858	G	T	0.46	1.29(1.57,1.06)	0.00949228

SNP	Chr:Position(bp)	info	Risk Allele	Other allele	RAF	OR (95% CI)	P
rs4916818	chr5:90038529	0.998478	C	T	0.2	1.02(1.3,0.8)	0.976534
rs59411130	chr5:90038549	0.960941	C	A	0.99	5.74(2.09,15.76)	0.000119007
rs10514334	chr5:90039078	1	T	C	0.2	1(1.27,0.79)	0.864355
rs57252870	chr5:90041153	0.960996	A	C	0.99	5.09(2,12.98)	0.000157206
rs2438356	chr5:90041161	0.998014	G	A	0.48	1.13(1.36,0.93)	0.145955
rs2438357	chr5:90041225	0.999955	G	A	0.46	1.29(1.57,1.06)	0.00946719
rs13171868	chr5:90041574	0.972169	C	T	0.99	1.29(0.54,3.1)	0.570568
rs2247747	chr5:90041898	0.999982	G	A	0.46	1.29(1.57,1.06)	0.00946327
rs7704041	chr5:90042321	0.999684	A	T	0.2	1.02(1.3,0.8)	0.979694
rs2438359	chr5:90042502	1	C	T	0.46	1.29(1.57,1.06)	0.00945779
rs73177407	chr5:90043047	0.960695	T	C	0.99	5.09(2,12.96)	0.000157801
rs2460188	chr5:90043198	0.997982	A	G	0.48	1.13(1.36,0.93)	0.145738
rs2438367	chr5:90043247	1	C	T	0.46	1.29(1.57,1.06)	0.00945779
rs181401225	chr5:90043603	0.91764	T	G	0.03	1.52(2.78,0.83)	0.169635
rs2438371	chr5:90043616	0.994018	C	A	0.46	1.29(1.56,1.06)	0.00994534
rs2203822	chr5:90043898	0.80875	A	G	0.06	1.1(1.68,0.72)	0.666815
rs2438372	chr5:90044215	0.998719	G	A	0.46	1.29(1.56,1.06)	0.0100782
chr5:90044283:D	chr5:90044283	0.992862	ATCTTG	A	0.46	1.28(1.55,1.06)	0.0115503
rs150025752	chr5:90044322	0.800891	G	A	0.02	1.14(2.54,0.51)	0.762362
rs2222241	chr5:90044373	0.99907	T	G	0.46	1.3(1.58,1.07)	0.00800052
chr5:90046189:D	chr5:90046189	0.938798	C	CAT	0.99	5.53(2.03,15.07)	9.66315E-05
rs41311341	chr5:90046296	0.872381	T	C	0.97	1.03(0.59,1.81)	0.86912
rs1996549	chr5:90046893	0.988088	C	T	0.45	1.3(1.57,1.07)	0.00784829
rs73772485	chr5:90047008	0.80368	G	A	0.05	1.08(1.66,0.71)	0.725699
rs2203823	chr5:90047161	0.984603	G	A	0.32	1.17(1.43,0.95)	0.121367
chr5:90047395:D	chr5:90047395	0.960085	TAAC	T	0.08	1.28(1.84,0.9)	0.203171
rs2460162	chr5:90047491	0.985641	G	T	0.48	1.15(1.4,0.95)	0.0842741
rs73177409	chr5:90047950	0.94665	G	A	0.99	5.25(1.91,14.43)	0.000233926
chr5:90048833:D	chr5:90048833	0.986912	TC	T	0.45	1.3(1.58,1.07)	0.00718869
chr5:90048834:I	chr5:90048834	0.986912	C	CA	0.45	1.3(1.58,1.07)	0.00718869
rs2438337	chr5:90049057	0.984405	T	C	0.48	1.14(1.38,0.94)	0.105764
rs73772486	chr5:90049157	0.802395	G	A	0.05	1.08(1.66,0.71)	0.729006
chr5:90049162:I	chr5:90049162	0.949593	A	AT	0.37	1.26(1.54,1.03)	0.0220131
rs2438339	chr5:90049238	0.931435	A	G	0.26	1.16(1.44,0.93)	0.118936
rs71637311	chr5:90050357	0.981953	T	C	0.98	1.14(0.56,2.31)	0.720446
chr5:90051086:I	chr5:90051086	0.874523	C	CT	0.4	1.26(1.54,1.04)	0.0117374
chr5:90051087:I	chr5:90051087	0.880952	T	TTC	0.4	1.27(1.55,1.04)	0.0104619
chr5:90051088:I	chr5:90051088	0.806704	T	TC	0.37	1.23(1.51,1.01)	0.0203067
rs1158530	chr5:90051332	0.975481	G	A	0.48	1.12(1.36,0.92)	0.153126
rs2438347	chr5:90051591	0.933935	C	T	0.47	1.29(1.57,1.07)	0.00741412
rs60470187	chr5:90051963	0.918807	G	A	0.99	4.83(1.82,12.86)	0.000287085
chr5:90052263:I	chr5:90052263	0.979019	T	TC	0.45	1.31(1.6,1.08)	0.00490118
rs10062026	chr5:90052289	0.972114	G	A	0.36	1.25(1.53,1.02)	0.0270923
rs2438349	chr5:90052372	0.981233	C	T	0.48	1.14(1.38,0.94)	0.106473
rs2443065	chr5:90053056	0.982273	A	G	0.45	1.3(1.57,1.07)	0.00783553
chr5:90053195:I	chr5:90053195	0.971755	C	CT	0.47	1.16(1.4,0.95)	0.0898817
rs2443064	chr5:90053280	0.980712	T	C	0.45	1.29(1.57,1.07)	0.00790744
rs2438350	chr5:90054109	0.97739	C	T	0.45	1.29(1.57,1.06)	0.00855369
rs62373965	chr5:90054878	0.943789	A	T	0.8	1(0.79,1.27)	0.854718
rs189981450	chr5:90055106	0.91248	C	T	0.03	1.52(2.75,0.84)	0.160791
rs76883417	chr5:90055130	0.92855	A	C	0.99	2.66(1.19,5.98)	0.0851516
rs77855755	chr5:90055866	0.976503	C	A	0.05	1.54(2.46,0.97)	0.0684422
rs113072800	chr5:90055963	0.973259	A	C	0.97	4.86(2.56,9.22)	**3.64621E-07**
rs188585053	chr5:90056058	0.844787	A	G	0.99	2.06(0.9,4.72)	0.0733563
rs16869094	chr5:90056174	0.972004	A	G	0.06	1.52(2.35,0.98)	0.0500481
rs750275	chr5:90056400	0.874735	G	A	0.04	1.04(1.66,0.65)	0.915332
rs870054	chr5:90056838	0.875673	A	G	0.04	1.04(1.66,0.65)	0.918572
rs2460163	chr5:90057563	0.99838	A	G	0.45	1.32(1.6,1.09)	0.00433388
rs73772488	chr5:90057686	0.877614	T	A	0.04	1.04(1.66,0.65)	0.925213
rs57547536	chr5:90057869	0.858782	G	A	0.91	1.53(1.12,2.11)	0.00887968
rs6893549	chr5:90057977	0.92005	G	A	0.92	1.89(1.34,2.68)	**0.00037666**
rs6893710	chr5:90058041	1	T	C	0.97	4.05(2.27,7.24)	**1.61084E-06**
rs2438351	chr5:90058109	0.976154	G	A	0.4	1.17(1.42,0.96)	0.0448044
rs2460164	chr5:90058190	1	A	T	0.45	1.32(1.61,1.09)	0.00420891

SNP	Chr:Position(bp)	info	Risk Allele	Other allele	RAF	OR (95% CI)	P
rs2460164	chr5:90058190	1	A	T	0.45	1.32(1.61,1.09)	0.00420891
rs73177420	chr5:90058451	0.97769	C	T	0.98	3.72(1.9,7.28)	**5.48738E-05**
rs62373966	chr5:90058982	0.975459	T	G	0.06	1.34(2.02,0.9)	0.0669446
rs881289	chr5:90059001	0.97643	T	A	0.4	1.16(1.41,0.96)	0.0514993
rs2049009	chr5:90059406	0.972	A	G	0.49	1.36(1.65,1.12)	**0.000410594**
rs2443061	chr5:90059436	0.997737	T	A	0.45	1.32(1.61,1.09)	**0.00411455**
rs2443060	chr5:90059708	0.994842	T	A	0.46	1.3(1.58,1.07)	**0.00501887**
rs73772489	chr5:90059968	0.879363	G	A	0.04	1.02(1.63,0.64)	0.997658
rs2460165	chr5:90060082	0.975165	A	G	0.47	1.09(1.32,0.9)	0.276009
rs62373981	chr5:90060597	0.979155	A	T	0.06	1.57(2.44,1.01)	0.0366638
rs10514335	chr5:90060782	1	A	G	0.06	1.53(2.37,0.98)	0.0531603
rs73177426	chr5:90061032	0.960768	G	C	0.98	3.64(1.87,7.09)	**5.75124E-05**
rs80332298	chr5:90061287	0.911597	G	C	0.91	1.26(0.9,1.76)	0.32744
rs77236023	chr5:90061812	0.923431	T	C	0.68	1.25(1.02,1.53)	0.0127648
rs73772490	chr5:90062601	0.941974	G	T	0.97	1.27(0.72,2.25)	0.364701
rs2460166	chr5:90063183	0.968488	G	A	0.93	2.22(1.52,3.24)	**6.97113E-05**
rs73177428	chr5:90063862	0.955835	G	T	0.98	3.42(1.76,6.65)	**0.000120101**
rs10514336	chr5:90064171	0.896828	C	T	0.02	1.08(2.19,0.53)	0.849493
rs115261920	chr5:90064561	0.958083	A	G	0.98	3.32(1.69,6.54)	**0.000208033**
rs12658994	chr5:90064784	0.951664	G	A	0.28	1.35(1.68,1.08)	**0.0061426**
rs2438352	chr5:90065570	0.993882	A	C	0.43	1.38(1.68,1.14)	**0.000226985**
rs2438353	chr5:90066246	1	T	C	0.43	1.43(1.73,1.18)	**2.74136E-05**
chr5:90067043:D	chr5:90067043	0.996653	TA	T	0.5	1.7(2.07,1.4)	**1.96988E-08**
rs12658034	chr5:90067887	0.984514	C	A	0.25	1.31(1.65,1.05)	0.0154166
rs7720206	chr5:90068209	0.996848	C	T	0.5	1.7(2.06,1.4)	**2.07962E-08**
rs73772491	chr5:90068343	0.982304	A	G	0.97	1.36(0.78,2.35)	0.282475
rs16869112	chr5:90069307	0.999265	A	C	0.02	1.08(2.03,0.57)	0.81825
rs2460167	chr5:90069563	1	A	G	0.43	1.39(1.68,1.14)	**0.000181959**
rs17624740	chr5:90069678	0.904292	T	C	0.08	1.38(1.99,0.95)	0.0838452
rs16869117	chr5:90069753	1	G	A	0.02	1.08(2.03,0.57)	0.818995
rs73772492	chr5:90070676	0.97794	G	A	0.97	1.33(0.75,2.35)	0.296331
rs13188479	chr5:90070979	0.991815	A	G	0.5	1.66(2.02,1.37)	**7.2975E-08**
rs12519770	chr5:90073277	1	A	G	0.5	1.69(2.06,1.39)	**2.9811E-08**
rs10514337	chr5:90073320	1	A	T	0.98	1.08(0.57,2.03)	0.818995
rs2438355	chr5:90073390	1	A	G	0.44	1.4(1.7,1.15)	**0.00012371**
rs3107223	chr5:90074097	0.996063	T	G	0.91	1.71(1.24,2.36)	**0.00638679**
rs41308293	chr5:90074646	0.908568	A	T	0.08	1.38(1.98,0.95)	0.0843279
rs35092519	chr5:90074759	0.976986	A	G	0.97	1.34(0.76,2.35)	0.283045
rs2443076	chr5:90075060	1	G	C	0.91	1.77(1.28,2.44)	0.0015852
rs2049010	chr5:90075770	0.959928	A	G	0.05	1.34(2.09,0.86)	0.162197
rs16869133	chr5:90075924	0.981223	A	G	0.02	1.08(2.02,0.58)	0.814673
chr5:90078672:I	chr5:90078672	0.994119	CAG	C	0.93	2.19(1.51,3.19)	**9.47194E-05**
chr5:90078674:I	chr5:90078674	0.992707	GA	G	0.93	2.2(1.52,3.19)	**8.98387E-05**
rs113674072	chr5:90078821	0.962578	G	A	0.98	3.28(1.68,6.41)	**0.000226989**
rs17554631	chr5:90079820	0.998726	A	G	0.2	1.53(1.97,1.19)	**0.00113715**
rs879571	chr5:90079972	0.998226	T	C	0.91	1.79(1.3,2.45)	**0.00183405**
rs2460171	chr5:90080575	0.993764	C	T	0.93	2.19(1.51,3.19)	**9.51967E-05**
rs2139191	chr5:90080797	0.991554	T	G	0.41	1.39(1.69,1.14)	**8.74469E-05**
rs147728824	chr5:90080947	0.980935	A	G	0.02	1.07(2,0.57)	0.83784
rs2945806	chr5:90081869	0.9827	T	C	0.93	2.24(1.54,3.25)	**6.71678E-05**
rs2460174	chr5:90082063	0.992094	G	A	0.93	2.2(1.51,3.19)	**9.06379E-05**
rs115997530	chr5:90082068	0.906079	G	C	0.01	1.59(4.17,0.61)	0.32271
rs2460175	chr5:90082396	0.991151	C	T	0.94	2.17(1.48,3.17)	0.000187452
rs10942608	chr5:90082594	1	C	T	0.2	1.53(1.98,1.19)	0.00111405
rs2443062	chr5:90083373	1	G	A	0.99	2.86(1.18,6.93)	0.0841518
rs11956622	chr5:90086216	0.971113	A	G	0.97	1.02(0.56,1.87)	0.847858
rs76310066	chr5:90087438	0.979869	A	G	0.03	1.04(1.87,0.57)	0.648595
rs60617564	chr5:90088460	0.977148	T	G	0.97	1.39(0.8,2.42)	0.229886
rs17554723	chr5:90089314	1	G	A	0.71	1.39(1.13,1.71)	0.000501136
chr5:90089525:D	chr5:90089525	0.999343	G	GAATT	0.71	1.4(1.13,1.72)	0.000396649
rs16869139	chr5:90089566	0.979662	A	C	0.03	1.03(1.87,0.57)	0.65343
rs16869140	chr5:90089651	0.979646	G	C	0.03	1.03(1.87,0.57)	0.653686
rs77395387	chr5:90089837	0.979652	T	C	0.03	1.03(1.87,0.57)	0.653886
rs79316782	chr5:90089981	0.97962	G	C	0.03	1.03(1.87,0.57)	0.654251

SNP	Chr:Position(bp)	info	Risk Allele	Other allele	RAF	OR (95% CI)	P
rs4916821	chr5:90090399	1	A	G	0.16	1.21(1.58,0.92)	0.136993
rs2438361	chr5:90090437	1	A	G	0.91	1.79(1.3,2.47)	0.00220614
rs114651485	chr5:90090671	0.97947	C	T	0.03	1.03(1.87,0.57)	0.654949
rs11745546	chr5:90090712	0.999572	G	C	0.71	1.39(1.13,1.71)	0.000495298
chr5:90090732:D	chr5:90090732	0.98131	AG	A	0.02	1.06(1.98,0.56)	0.865082
rs2438362	chr5:90090791	1	G	A	0.79	1.46(1.16,1.85)	0.00213394
rs148754405	chr5:90091430	0.928594	G	A	0.03	1.51(2.66,0.86)	0.148656
rs142342530	chr5:90091507	0.971477	A	G	0.97	1.4(0.8,2.44)	0.218183
rs76251327	chr5:90091585	0.976954	A	G	0.97	1.39(0.8,2.42)	0.229461
rs113802395	chr5:90091600	0.936089	A	G	0.94	1.07(0.7,1.62)	0.685504
rs2438364	chr5:90091816	0.989099	A	G	0.52	1.61(1.33,1.96)	**4.5485E-07**
rs145197019	chr5:90092525	0.976355	T	A	0.97	1.35(0.77,2.37)	0.267969
rs149067605	chr5:90093406	0.982163	A	G	0.91	1.73(1.25,2.41)	0.00610618
rs13180421	chr5:90094789	0.972757	G	A	0.99	1.17(0.48,2.88)	0.747109
rs2438368	chr5:90094855	0.996478	A	G	0.51	1.65(1.36,2)	**2.03502E-07**
rs140671922	chr5:90095574	0.85086	C	G	0.8	1.22(0.96,1.54)	0.0315881
rs2366932	chr5:90096696	0.925018	A	T	0.93	1.66(1.16,2.37)	0.0274733
rs2443073	chr5:90096792	0.826929	T	C	0.98	1.22(0.67,2.23)	0.642708
rs146994514	chr5:90096814	0.979719	C	T	0.02	1.08(2.02,0.57)	0.814204
rs2438370	chr5:90096921	0.826944	C	T	0.98	1.22(0.67,2.23)	0.642612
rs1983366	chr5:90097313	0.878906	G	C	0.91	1.62(1.18,2.24)	0.0103243
rs62373985	chr5:90099111	0.959484	C	T	0.05	1.44(2.25,0.92)	0.0931639
rs184909647	chr5:90099823	0.924708	G	A	0.03	1.52(2.67,0.87)	0.141914
rs73179304	chr5:90100408	0.946719	A	G	0.99	3.97(1.67,9.44)	**0.00058763**
rs112842366	chr5:90102825	0.946483	T	A	0.99	3.97(1.67,9.44)	**0.000592458**
rs16869151	chr5:90103678	0.981792	A	G	0.02	1.08(2.02,0.57)	0.820578
rs2438373	chr5:90103785	0.862569	T	G	0.04	1.73(2.86,1.05)	0.0922207
rs73179310	chr5:90104252	0.946242	A	G	0.99	3.97(1.67,9.43)	**0.00059412**
rs6860302	chr5:90104953	0.915397	T	C	0.04	1.33(2.24,0.79)	0.154814
rs75945334	chr5:90105280	0.981584	T	A	0.02	1.08(2.02,0.57)	0.817923
rs2438374	chr5:90107108	0.82592	A	G	0.02	1.22(2.23,0.67)	0.635094
rs60526684	chr5:90107838	0.934842	T	C	0.98	3.59(1.64,7.86)	**0.000375268**
rs56886929	chr5:90108039	0.945819	A	G	0.99	3.96(1.67,9.42)	**0.000601422**
rs114170072	chr5:90108056	0.904465	T	A	0.04	1.54(2.67,0.88)	0.122935
rs113336323	chr5:90108702	0.945792	G	A	0.99	3.96(1.67,9.42)	0.000601794
rs144457455	chr5:90108777	0.809904	C	T	0.98	1.38(0.73,2.61)	0.268462
rs2438375	chr5:90108906	0.806087	C	T	0.13	1.49(1.97,1.12)	0.011236
chr5:90109015:D	chr5:90109015	0.827355	TA	T	0.03	1.3(2.35,0.72)	0.497553
chr5:90109018:D	chr5:90109018	0.820242	TAGA	T	0.03	1.14(2.01,0.65)	0.801811
rs2443078	chr5:90109230	0.825215	T	C	0.02	1.23(2.24,0.67)	0.625043
rs2443079	chr5:90110224	1	A	G	0.49	1.67(2.02,1.37)	**1.10685E-07**
rs41304884	chr5:90111521	0.95551	G	A	0.99	3.89(1.63,9.31)	**0.000767278**
rs2443080	chr5:90111700	0.901463	C	T	0.96	1.5(0.88,2.54)	0.0824129
rs73179316	chr5:90111869	0.944538	C	T	0.99	3.95(1.66,9.41)	0.000611661
chr5:90112185:D	chr5:90112185	0.815012	GT	G	0.05	1.33(2.11,0.84)	0.151582
rs185666755	chr5:90112385	0.903638	C	A	0.03	1.03(1.9,0.56)	0.917116
rs188935231	chr5:90115911	0.825708	T	G	0.89	1.02(0.75,1.38)	0.727568
rs2438377	chr5:90118205	0.891708	C	T	0.12	1.32(1.76,0.98)	0.185865
rs145578574	chr5:90118239	0.940168	A	G	0.99	3.93(1.65,9.36)	0.000640334
rs12655163	chr5:90118880	0.986368	T	C	0.02	1.07(2.02,0.57)	0.832014
rs7711338	chr5:90119094	0.915026	A	T	0.04	1.53(2.68,0.88)	0.130723
chr5:90119123:I	chr5:90119123	0.934744	GA	G	0.99	3.87(1.63,9.18)	0.000696246
rs3763073	chr5:90119293	0.986413	C	T	0.02	1.07(2.02,0.57)	0.831667
rs2438378	chr5:90119324	0.896794	A	G	0.96	1.49(0.88,2.53)	0.0833998
rs2443081	chr5:90119922	0.896398	A	T	0.96	1.49(0.88,2.53)	0.0833939
rs73179326	chr5:90120710	0.938141	T	C	0.99	3.92(1.64,9.34)	0.000655175
rs2244265	chr5:90120963	0.869146	T	C	0.09	1.41(1.96,1.02)	0.12822
rs2438379	chr5:90121507	0.897344	G	T	0.11	1.28(1.73,0.95)	0.268437
rs6869135	chr5:90122164	0.910981	T	C	0.04	1.54(2.68,0.88)	0.128182
rs12655900	chr5:90124185	0.988317	C	G	0.02	1.08(2.03,0.57)	0.825252
rs2443083	chr5:90129284	0.900268	T	C	0.96	1.53(0.9,2.61)	0.0724244
rs2443084	chr5:90130560	0.867109	C	G	0.12	1.26(1.68,0.94)	0.327208
rs73179333	chr5:90131371	0.927571	G	A	0.99	3.85(1.61,9.22)	0.000747147
rs2438380	chr5:90131698	0.860991	T	C	0.11	1.21(1.63,0.89)	0.384297

SNP	Chr:Position(bp)	info	Risk Allele	Other allele	RAF	OR (95% CI)	P
rs74394442	chr5:90133379	0.989642	A	G	0.02	1.11(2.09,0.59)	0.749965
rs2438382	chr5:90133981	0.876977	G	A	0.96	1.45(0.87,2.42)	0.0822539
rs2438383	chr5:90134906	0.876576	T	A	0.96	1.45(0.88,2.42)	0.0818828
rs41304894	chr5:90136330	0.804139	C	G	0.99	1.37(0.57,3.29)	0.413927
rs16869163	chr5:90137108	1	A	G	0.02	1.12(2.1,0.59)	0.736567
rs112800433	chr5:90137252	0.933248	T	C	0.99	3.77(1.57,9.1)	0.000955322
chr5:90138361:D	chr5:90138361	0.932397	C	CCT	0.99	3.77(1.56,9.09)	0.000964565
rs78925640	chr5:90143846	0.868672	C	T	0.03	1.06(1.96,0.57)	0.860268
rs12657829	chr5:90145036	0.846313	A	G	0.03	1.06(1.97,0.58)	0.839744
rs140056399	chr5:90145236	0.80471	C	T	0.03	1.32(2.43,0.72)	0.406244
rs111543261	chr5:90145414	0.840314	T	C	0.03	1.07(1.97,0.58)	0.837303
rs2443086	chr5:90146380	0.86245	C	T	0.19	1.46(1.89,1.13)	0.00230333
rs2443091	chr5:90147447	1	C	G	0.51	1.65(1.36,2)	**1.88418E-07**
rs13159353	chr5:90148516	0.848666	C	G	0.6	1.34(1.1,1.63)	0.000366717
rs11744325	chr5:90150497	0.864395	A	T	0.72	1.42(1.15,1.75)	0.000149519
rs73179346	chr5:90151174	0.866648	C	A	0.97	1.58(0.94,2.65)	0.0717972
rs2247870	chr5:90151589	1	G	A	0.45	1.6(1.94,1.31)	**1.87066E-06**
rs2438338	chr5:90152302	0.984799	A	T	0.4	1.45(1.77,1.19)	**8.57653E-05**
rs2443089	chr5:90153758	0.964311	A	T	0.41	1.47(1.79,1.21)	**4.75995E-05**
rs2247546	chr5:90153915	0.993927	G	A	0.45	1.59(1.93,1.31)	**2.02541E-06**
rs2247419	chr5:90155414	0.922436	A	G	0.43	1.4(1.7,1.15)	0.000294821
rs2247415	chr5:90155553	0.981758	A	C	0.39	1.44(1.75,1.18)	0.000121964
rs6865577	chr5:90155713	0.822364	T	A	0.89	1.08(0.8,1.48)	0.419715
rs189592415	chr5:90155754	0.836664	G	A	0.99	2.66(1.2,5.93)	0.00864174
rs34368137	chr5:90156180	0.862471	C	T	0.24	1.16(1.46,0.93)	0.154308
rs117816198	chr5:90156688	0.84598	A	G	0.98	1.01(0.5,2.06)	0.94674
chr5:90158645:I	chr5:90158645	0.95842	C	CAT	0.37	1.2(1.47,0.99)	0.0589389
rs2438342	chr5:90160802	0.98494	G	A	0.39	1.43(1.74,1.17)	**0.000106096**
rs2438343	chr5:90161605	0.971301	T	C	0.39	1.4(1.71,1.15)	**0.000176938**
rs2438344	chr5:90161956	0.982834	C	T	0.39	1.46(1.78,1.2)	**5.52606E-05**
rs2443087	chr5:90162124	0.862825	A	G	0.3	1.46(1.79,1.18)	**4.98174E-05**
rs72782791	chr5:90162162	0.881415	G	A	0.73	1.43(1.16,1.77)	**0.000125139**
rs2438345	chr5:90162285	0.987712	T	C	0.39	1.45(1.77,1.19)	**7.77337E-05**
rs77174790	chr5:90164588	0.874463	T	G	0.97	1.52(0.91,2.56)	0.0942427
rs79636565	chr5:90166991	0.942205	G	T	0.96	1.44(0.92,2.27)	0.115489
rs11738573	chr5:90167106	0.901149	T	G	0.73	1.43(1.16,1.77)	**0.000158503**
rs3098356	chr5:90168011	1	C	A	0.39	1.48(1.8,1.22)	**4.09745E-05**
rs58729412	chr5:90169070	0.943499	T	C	0.31	1.14(1.4,0.92)	0.215072
rs10059629	chr5:90169163	0.819033	C	A	0.89	1.12(0.82,1.53)	0.295145
rs2366935	chr5:90169937	0.821477	C	A	0.89	1.11(0.82,1.52)	0.319266
rs62374006	chr5:90171615	0.898688	C	T	0.06	1.27(1.95,0.83)	0.118207
rs58884460	chr5:90171765	0.941985	C	T	0.9	1.05(0.76,1.44)	0.943875
rs6861214	chr5:90172331	0.932875	T	C	0.9	1.09(0.8,1.49)	0.880024
chr5:90173096:D	chr5:90173096	0.874896	T	TG	0.97	1.53(0.91,2.57)	0.0919519
rs10942614	chr5:90173527	0.932592	A	G	0.9	1.09(0.8,1.49)	0.887638
rs78750530	chr5:90174955	0.864994	G	C	0.07	1.18(1.75,0.79)	0.199229
rs7718692	chr5:90175446	0.866115	G	A	0.07	1.17(1.75,0.79)	0.204181
rs77146933	chr5:90176413	0.868152	A	G	0.97	1.51(0.9,2.53)	0.100018
rs78280156	chr5:90176646	0.868097	G	A	0.97	1.51(0.9,2.53)	0.100018
rs62374009	chr5:90177583	0.914898	T	G	0.06	1.21(1.85,0.79)	0.161095
rs6893164	chr5:90179724	1	C	T	0.88	1.1(0.82,1.48)	0.961489
rs111644494	chr5:90179984	0.88381	T	A	0.01	1.14(2.78,0.47)	0.794704
rs4916692	chr5:90180858	1	A	T	0.11	1.08(1.46,0.8)	0.920928
rs2438348	chr5:90181320	1	T	A	0.72	1.41(1.14,1.74)	0.000616302
rs77572045	chr5:90181647	0.868387	C	T	0.09	1.7(2.44,1.19)	0.00284234
rs2443068	chr5:90182824	1	A	G	0.3	1.38(1.71,1.11)	0.00657015
rs4637585	chr5:90185517	0.850763	C	T	0.41	1.29(1.58,1.06)	0.00418896
rs57107124	chr5:90185769	0.979095	C	T	0.31	1.37(1.7,1.11)	0.00539689
rs10045202	chr5:90186647	0.88152	T	C	0.41	1.3(1.58,1.06)	0.00423769
rs4279369	chr5:90188746	0.815462	A	G	0.05	1.15(1.76,0.75)	0.274145
rs4916826	chr5:90188763	0.996255	G	T	0.31	1.38(1.7,1.11)	0.00534315
rs4244205	chr5:90188886	0.945965	A	G	0.41	1.31(1.59,1.07)	0.00449923
rs4916827	chr5:90189111	1	C	G	0.31	1.36(1.68,1.1)	0.00654844
rs4916828	chr5:90189199	0.993395	A	G	0.32	1.36(1.68,1.1)	0.00686936

SNP	Chr:Position(bp)	info	Risk Allele	Other allele	RAF	OR (95% CI)	P
rs28437109	chr5:90189392	0.996001	T	C	0.32	1.37(1.69,1.11)	0.00569715
rs4580808	chr5:90190300	0.995093	G	T	0.41	1.32(1.61,1.08)	0.00376929
rs4496735	chr5:90190417	1	C	G	0.41	1.32(1.61,1.09)	0.00379627
chr5:90190912:I	chr5:90190912	0.98633	AT	A	0.9	1.01(0.73,1.39)	0.71435
rs6868618	chr5:90191256	0.997714	A	G	0.41	1.33(1.62,1.09)	0.00298973
rs76004104	chr5:90192012	0.987416	A	G	0.9	1.06(0.77,1.46)	0.912766
rs73771110	chr5:90192654	0.992173	T	A	0.9	1.02(0.74,1.41)	0.738669
chr5:90193344:I	chr5:90193344	0.948099	TAGCAAAACATC	T	0.91	1.04(0.75,1.44)	0.764704
rs10942617	chr5:90194112	1	C	G	0.9	1.05(0.76,1.44)	0.838641
rs6886778	chr5:90194841	0.991691	G	C	0.9	1.02(0.74,1.41)	0.727966
rs4916829	chr5:90195447	0.9904	T	G	0.31	1.42(1.75,1.15)	0.00209828
rs59343071	chr5:90196101	0.915344	A	G	0.95	1.16(0.75,1.78)	0.623452
rs58258039	chr5:90196383	0.98322	G	A	0.9	1.02(0.74,1.41)	0.744197
rs72784606	chr5:90196526	0.863292	C	T	0.1	1.56(2.2,1.11)	0.00915735
rs79115766	chr5:90197752	0.905591	T	G	0.96	1.11(0.69,1.8)	0.698412
rs4382198	chr5:90198472	0.977797	G	C	0.9	1.02(0.74,1.41)	0.734209
rs4464729	chr5:90198701	0.905715	G	A	0.96	1.14(0.71,1.83)	0.655299
rs10044202	chr5:90199564	0.982102	G	A	0.31	1.42(1.75,1.15)	0.00184334
rs4362971	chr5:90200022	0.976865	C	T	0.9	1.05(0.77,1.44)	0.853273
rs4916830	chr5:90201522	0.971992	A	G	0.9	1.05(0.77,1.44)	0.85735
rs10035307	chr5:90201999	0.973928	C	T	0.42	1.32(1.6,1.08)	0.00325001
rs78331558	chr5:90202490	0.967477	A	G	0.9	1.05(0.77,1.44)	0.852905
rs73771113	chr5:90202963	0.966088	C	T	0.9	1.05(0.77,1.44)	0.854525
rs73771115	chr5:90203249	0.965643	G	A	0.9	1.05(0.77,1.44)	0.857372
rs10061193	chr5:90203272	0.956467	T	C	0.12	1.01(1.36,0.75)	0.876172
rs73771117	chr5:90203488	0.959955	G	T	0.1	1.05(1.46,0.76)	0.492302
rs114113647	chr5:90203736	0.957363	C	T	0.1	1.05(1.46,0.76)	0.488936
rs73771118	chr5:90204509	0.94711	A	G	0.9	1.06(0.77,1.45)	0.873729
rs6452914	chr5:90204765	0.805846	G	T	0.44	1.04(1.26,0.86)	0.83443
rs6858917	chr5:90205193	0.942508	T	G	0.46	1.29(1.56,1.06)	0.00614939
rs6878896	chr5:90205537	0.964045	G	C	0.35	1.35(1.66,1.1)	0.0047644
rs73771119	chr5:90205700	0.930343	A	C	0.9	1.01(0.73,1.39)	0.798858
rs10474336	chr5:90206114	0.964797	T	A	0.35	1.35(1.66,1.1)	0.00405528
rs57190680	chr5:90206185	0.843523	C	A	0.32	1.01(1.24,0.82)	0.993173
rs36088173	chr5:90206282	0.930475	A	G	0.9	1.01(0.73,1.39)	0.798836
rs12658466	chr5:90206466	0.931943	G	A	0.9	1.01(0.73,1.39)	0.78055
rs6889986	chr5:90207399	0.963069	G	A	0.44	1.3(1.58,1.07)	0.00532609
rs6889598	chr5:90207409	0.930767	G	A	0.9	1.01(0.73,1.39)	0.797878
rs12520956	chr5:90208481	0.980084	G	A	0.34	1.34(1.65,1.09)	0.00634179
rs56851353	chr5:90208972	0.94269	A	G	0.1	1.01(1.39,0.73)	0.707978
rs61006221	chr5:90208978	0.942692	T	C	0.1	1.01(1.39,0.73)	0.70803
rs10052015	chr5:90209063	1	T	C	0.34	1.34(1.65,1.09)	0.00674727
rs10074525	chr5:90209824	0.824081	C	T	0.23	1.37(1.72,1.1)	0.00136682
rs78751985	chr5:90209848	0.912519	C	T	0.95	1.08(0.69,1.68)	0.802148
rs34088804	chr5:90210432	0.914584	A	G	0.29	1.01(1.24,0.82)	0.937086
rs12522395	chr5:90210518	1	C	G	0.66	1.34(1.09,1.65)	0.00674727
rs62374016	chr5:90211273	0.9253	T	C	0.29	1(1.24,0.81)	0.993603
rs10078496	chr5:90211505	0.871582	C	T	0.38	1.26(1.54,1.03)	0.022041
rs10078568	chr5:90211648	0.873735	C	G	0.43	1.23(1.5,1.01)	0.0337356
rs4916831	chr5:90212276	0.884922	A	G	0.48	1.25(1.52,1.03)	0.0116653
rs61424527	chr5:90212692	0.919963	G	A	0.28	1.03(1.28,0.84)	0.7463
rs12513628	chr5:90212696	0.935827	G	A	0.34	1.32(1.62,1.08)	0.00754036
rs138621664	chr5:90212700	0.864179	T	C	0.95	1.09(0.71,1.67)	0.73564
rs28368305	chr5:90212892	0.888124	G	A	0.48	1.25(1.52,1.03)	0.0120075
rs10060641	chr5:90213250	0.877513	T	C	0.38	1.26(1.53,1.03)	0.0234045
rs2367181	chr5:90215587	0.960945	G	A	0.29	1.04(1.29,0.84)	0.7239
rs7723305	chr5:90216806	0.97966	A	G	0.75	1.4(1.13,1.74)	0.000685582
chr5:90217622:I	chr5:90217622	0.82562	T	TG	0.06	1.37(2.07,0.91)	0.0548273
rs111930434	chr5:90217623	0.830549	T	G	0.06	1.35(2.04,0.9)	0.0611684
rs1976566	chr5:90217847	0.992266	A	C	0.26	1.41(1.75,1.14)	0.000734766
rs7724016	chr5:90217889	1	T	C	0.34	1.34(1.65,1.09)	0.00781648
rs78048019	chr5:90217918	0.929063	A	G	0.95	1.04(0.67,1.63)	0.924715
rs12054681	chr5:90217927	0.996643	C	A	0.34	1.35(1.66,1.09)	0.00701045
rs3114654	chr5:90218506	0.989245	T	G	0.35	1.06(1.3,0.87)	0.487298

SNP	Chr:Position(bp)	info	Risk Allele	Other allele	RAF	OR (95% CI)	P
rs75472091	chr5:90218544	0.923701	G	A	0.95	1.04(0.67,1.62)	0.935611
rs3114653	chr5:90218612	1	A	G	0.27	1.44(1.78,1.16)	0.000458123
rs3114652	chr5:90218696	1	T	C	0.35	1.05(1.29,0.86)	0.537042
chr5:90218846:I	chr5:90218846	0.982657	A	ACT	0.29	1.04(1.29,0.84)	0.731416
rs140285685	chr5:90220180	0.94551	G	C	0.33	1.36(1.68,1.11)	0.00277043
rs149048589	chr5:90220256	0.930859	G	A	0.32	1.43(1.76,1.15)	0.000594653
rs140697358	chr5:90220766	0.88447	T	C	0.32	1.34(1.64,1.09)	0.00160877
rs150974895	chr5:90220770	0.876861	G	A	0.32	1.32(1.62,1.07)	0.00285066
rs149487779	chr5:90220858	0.876175	G	A	0.3	1.45(1.8,1.17)	0.000303294
rs138673763	chr5:90220893	0.88768	C	G	0.95	1.19(0.78,1.8)	0.402766
rs36131676	chr5:90221351	0.97944	G	T	0.35	1.06(1.3,0.87)	0.481932
rs140865268	chr5:90223236	0.934011	G	C	0.32	1.39(1.72,1.12)	0.00212124
rs139912427	chr5:90223241	0.940339	A	G	0.32	1.37(1.69,1.11)	0.00284333
rs3114658	chr5:90223544	0.896452	T	C	0.3	1.34(1.65,1.09)	0.00209742
rs138587521	chr5:90223610	0.875457	C	T	0.29	1.33(1.65,1.07)	0.00677048
rs145601853	chr5:90223854	0.945903	C	T	0.33	1.31(1.62,1.07)	0.0100366
rs191321264	chr5:90224237	0.828444	G	A	0.06	1.06(1.62,0.7)	0.672229
rs59430498	chr5:90225291	0.86521	C	G	0.05	1.15(1.77,0.75)	0.293771
rs10035662	chr5:90225416	0.956055	A	C	0.33	1.32(1.62,1.07)	0.00936297
rs60681604	chr5:90225453	0.875084	G	T	0.62	1(0.82,1.22)	0.841694
rs57258230	chr5:90225461	0.907455	A	C	0.38	1.02(1.24,0.84)	0.657671
rs17556421	chr5:90225486	0.954445	G	A	0.33	1.31(1.61,1.06)	0.0117401
rs16869227	chr5:90225796	0.859417	A	G	0.05	1.02(1.56,0.67)	0.867171
rs3105793	chr5:90226061	1	A	G	0.71	1(0.81,1.24)	0.970953
rs61646974	chr5:90226122	0.858467	A	C	0.06	1.02(1.56,0.67)	0.873129
rs3105792	chr5:90226126	0.954156	G	T	0.35	1.04(1.27,0.85)	0.57961
rs16869229	chr5:90226282	0.863844	C	T	0.06	1.15(1.75,0.75)	0.282282
rs3105791	chr5:90226542	1	C	G	0.64	1.02(0.84,1.25)	0.746599
rs74501188	chr5:90226979	0.960358	G	A	0.21	1.4(1.79,1.1)	0.00640087
rs3105790	chr5:90227536	0.961838	G	T	0.48	1.05(1.27,0.86)	0.538759
rs3114655	chr5:90228059	0.969212	C	T	0.48	1.05(1.27,0.87)	0.519272
rs16876837	chr5:90228349	0.844727	A	G	0.05	1.02(1.56,0.67)	0.871679
rs7712313	chr5:90228398	0.978652	C	T	0.75	1.39(1.12,1.73)	0.000878105
rs7729840	chr5:90228400	0.976877	G	A	0.75	1.39(1.12,1.73)	0.000847935
rs3105789	chr5:90228431	0.963279	A	T	0.48	1.05(1.28,0.87)	0.502512
rs55941525	chr5:90228793	0.980788	T	C	0.75	1.4(1.13,1.74)	0.000859408
rs10942618	chr5:90229426	0.957903	T	G	0.47	1.04(1.26,0.86)	0.587685
rs11744005	chr5:90229530	0.977459	A	C	0.52	1.06(0.87,1.28)	0.470895
rs11750515	chr5:90229542	1	G	A	0.53	1.05(0.87,1.28)	0.478483
rs11744148	chr5:90229807	0.965248	G	A	0.21	1.4(1.79,1.1)	0.00599023
rs4415111	chr5:90230772	0.958723	G	A	0.53	1.06(0.87,1.28)	0.474728
rs10053303	chr5:90231189	0.915851	A	G	0.11	1.03(1.4,0.76)	0.960939
rs2367182	chr5:90231238	0.947493	C	T	0.53	1.06(0.88,1.29)	0.450921
rs6867530	chr5:90231248	0.958694	T	G	0.53	1.05(0.87,1.28)	0.484106
rs4916696	chr5:90233420	0.826199	G	A	0.8	1.03(0.81,1.31)	0.774671
rs10514340	chr5:90233667	0.805836	C	G	0.87	1.01(0.77,1.34)	0.926959
rs1818350	chr5:90233933	0.905978	T	C	0.11	1.12(1.54,0.82)	0.305839
rs58201771	chr5:90234007	0.893296	A	G	0.11	1.02(1.38,0.75)	0.953714
rs11749470	chr5:90235297	0.906388	T	C	0.56	1.06(0.87,1.28)	0.454745
rs1876633	chr5:90235367	0.959287	G	A	0.63	1.06(0.87,1.29)	0.499517
rs12187790	chr5:90236049	0.807103	G	A	0.83	1.38(1.07,1.77)	0.00150103
rs4916697	chr5:90236252	0.961428	A	T	0.63	1.06(0.87,1.29)	0.508316
rs57954532	chr5:90236366	0.880721	G	C	0.12	1.02(1.38,0.75)	0.959183
rs75836649	chr5:90236478	0.920183	T	G	0.05	1.24(1.92,0.8)	0.184968
chr5:90236647:D	chr5:90236647	0.898914	A	AC	0.56	1.06(0.87,1.28)	0.462155
rs66509877	chr5:90236895	0.838996	G	A	0.82	1.38(1.08,1.77)	0.00122741
rs73185149	chr5:90238058	0.863444	T	C	0.12	1(1.35,0.74)	0.941154
rs73185150	chr5:90238083	0.873789	C	A	0.12	1.02(1.38,0.76)	0.946523
rs12188655	chr5:90238295	0.977125	C	G	0.75	1.38(1.11,1.72)	0.000844257
rs1967413	chr5:90238905	0.891285	A	G	0.56	1.07(0.89,1.3)	0.36582
rs12055217	chr5:90239015	0.846901	C	A	0.12	1.02(1.36,0.76)	0.965843
rs4916698	chr5:90239135	0.875989	A	G	0.11	1.04(1.41,0.77)	0.899301
rs2063245	chr5:90239911	1	G	A	0.63	1.1(0.9,1.34)	0.335322
rs2063244	chr5:90240028	1	T	A	0.45	1.09(1.32,0.9)	0.304491

SNP	Chr:Position(bp)	info	Risk Allele	Other allele	RAF	OR (95% CI)	P
chr5:90241803:D	chr5:90241803	0.875709	A	AAAAG	0.59	1.02(0.84,1.23)	0.736433
chr5:90241804:D	chr5:90241804	0.891712	A	AAAG	0.58	1.05(0.86,1.27)	0.498631
rs76645775	chr5:90242010	0.921172	T	C	0.1	1.15(1.58,0.83)	0.252425
rs12651764	chr5:90242390	0.9727	C	G	0.05	1.3(2.03,0.83)	0.135731
rs6860216	chr5:90242703	0.96475	G	C	0.75	1.4(1.13,1.74)	0.000584692
rs6891672	chr5:90243899	1	C	T	0.69	1.02(0.83,1.26)	0.890183
rs13163934	chr5:90244104	0.964062	A	G	0.54	1.06(0.88,1.29)	0.45475
rs10942619	chr5:90244528	0.964892	T	A	0.54	1.06(0.88,1.29)	0.4482
rs4587106	chr5:90244654	0.898456	C	G	0.03	1.6(2.85,0.9)	0.0887495
rs7716083	chr5:90244912	0.964884	A	C	0.54	1.06(0.88,1.29)	0.441574
chr5:90245680:D	chr5:90245680	0.90692	CTT	C	0.12	1.03(1.39,0.76)	0.877859
rs10057134	chr5:90245713	0.964095	T	C	0.54	1.06(0.87,1.28)	0.472995
rs11741599	chr5:90245929	0.998708	T	C	0.69	1.02(0.83,1.26)	0.880482
rs60553010	chr5:90246067	0.880072	G	A	0.95	1(0.65,1.54)	0.979242
chr5:90246738:D	chr5:90246738	0.969588	A	AAC	0.75	1.41(1.13,1.75)	0.000409995
chr5:90246739:D	chr5:90246739	0.969093	A	AC	0.75	1.41(1.13,1.75)	0.000415756
rs7705201	chr5:90246842	1	G	T	0.54	1.08(0.89,1.3)	0.376904
chr5:90247304:D	chr5:90247304	0.908448	TTTTA	T	0.12	1.03(1.4,0.77)	0.852759
rs1828175	chr5:90248001	0.929957	T	A	0.1	1.13(1.56,0.82)	0.287093
rs16869247	chr5:90248442	0.909461	A	G	0.11	1.05(1.43,0.78)	0.823217
rs10052314	chr5:90248747	0.901643	C	T	0.12	1.03(1.4,0.77)	0.852477
rs4916832	chr5:90249035	0.912297	T	A	0.17	1.03(1.33,0.8)	0.806982
rs4916833	chr5:90249105	0.902901	G	A	0.7	1.03(0.83,1.27)	0.825781
rs7709513	chr5:90249154	0.899216	A	G	0.06	1.35(2.09,0.87)	0.073313
chr5:90249471:D	chr5:90249471	0.862347	C	CT	0.94	1.04(0.7,1.57)	0.92222
rs10213801	chr5:90249900	0.862054	A	G	0.54	1.02(0.84,1.24)	0.878666
rs6875416	chr5:90250631	1	T	A	0.8	1.49(1.16,1.91)	0.00131558
rs949789	chr5:90251093	1	C	T	0.74	1.46(1.18,1.82)	9.36047E-05
rs949787	chr5:90251205	0.811047	G	T	0.33	1.35(1.66,1.1)	0.000271906
rs1818351	chr5:90252077	0.872787	C	T	0.72	1.02(0.82,1.26)	0.897755
rs6452916	chr5:90252232	1	C	A	0.06	1.24(1.89,0.81)	0.138208
rs4916834	chr5:90252291	0.896544	C	T	0.95	1.07(0.7,1.64)	0.753718
rs6452917	chr5:90252429	0.989668	A	G	0.06	1.19(1.82,0.78)	0.181697
rs12522091	chr5:90252688	0.88487	T	A	0.94	1.1(0.73,1.67)	0.720186
rs2367183	chr5:90252893	0.987834	G	A	0.06	1.19(1.81,0.78)	0.184132
rs56219582	chr5:90253892	0.805762	A	G	0.28	1.43(1.79,1.15)	4.62927E-05
rs60875962	chr5:90254162	0.809143	G	T	0.27	1.47(1.84,1.18)	1.87871E-05
rs11744253	chr5:90254329	0.808905	G	A	0.28	1.43(1.79,1.15)	4.44198E-05
chr5:90254355:D	chr5:90254355	0.808075	TAAC	T	0.28	1.44(1.79,1.15)	3.97752E-05
rs10074585	chr5:90254998	0.856983	T	G	0.34	1.34(1.65,1.09)	0.0002704
rs6863794	chr5:90255381	0.946747	G	A	0.11	1.06(1.44,0.78)	0.432426
rs10942620	chr5:90255682	0.912195	C	T	0.11	1.09(1.49,0.8)	0.65496
rs7704653	chr5:90255685	0.839474	A	G	0.29	1.01(1.25,0.82)	0.998801
rs7724872	chr5:90255735	0.897728	C	T	0.94	1.21(0.81,1.82)	0.446639
rs35060258	chr5:90255753	0.924754	G	A	0.75	1.4(1.13,1.74)	0.000347823
rs7708774	chr5:90255965	0.93268	A	C	0.12	1.12(1.51,0.83)	0.496513
chr5:90255972:I	chr5:90255972	0.890653	AC	A	0.95	1.29(0.85,1.95)	0.290269
rs7709355	chr5:90256000	0.942964	T	G	0.88	1.02(0.76,1.37)	0.684129
rs7709365	chr5:90256026	0.913109	A	G	0.94	1.25(0.83,1.88)	0.356409
rs59665236	chr5:90256096	0.952849	T	C	0.11	1.05(1.43,0.77)	0.452477
rs16869285	chr5:90256819	0.918536	A	G	0.94	1.28(0.85,1.92)	0.301502
rs72656679	chr5:90256889	0.944223	T	G	0.88	1.01(0.75,1.37)	0.637584
rs16869289	chr5:90256972	0.918364	T	A	0.07	1.1(1.63,0.75)	0.27763
rs6874280	chr5:90257244	0.852088	A	G	0.28	1.02(1.26,0.82)	0.815485
chr5:90257366:D	chr5:90257366	0.907919	GGAGT	G	0.06	1.15(1.75,0.75)	0.195569
rs34381536	chr5:90257392	0.9905	G	A	0.73	1.42(1.15,1.76)	0.000222741
rs61130831	chr5:90258396	0.953655	A	G	0.88	1.09(0.81,1.46)	0.989237
rs12522571	chr5:90258584	1	A	G	0.19	1.56(2.01,1.21)	0.000476807
rs10079044	chr5:90259332	0.935033	A	C	0.33	1.48(1.82,1.2)	0.000114577
rs72656680	chr5:90260413	0.967971	C	G	0.06	1.11(1.67,0.73)	0.302683
rs12173258	chr5:90262194	0.936985	T	A	0.39	1.43(1.74,1.17)	0.00004591
rs12523056	chr5:90262475	0.949042	T	C	0.44	1.34(1.63,1.1)	0.00037713
rs16869291	chr5:90262492	0.971222	G	C	0.89	1.01(0.74,1.37)	0.654231
rs12523094	chr5:90262612	0.955804	T	C	0.45	1.36(1.66,1.12)	0.000207827

SNP	Chr:Position(bp)	info	Risk Allele	Other allele	RAF	OR (95% CI)	P
chr5:90263058:D	chr5:90263058	0.970409	A	ACT	0.95	1.11(0.72,1.71)	0.645907
rs10942621	chr5:90263169	0.990715	A	T	0.42	1.39(1.7,1.14)	0.000133007
rs11957689	chr5:90263347	0.996863	G	T	0.42	1.39(1.69,1.14)	0.000201171
rs6860111	chr5:90263581	0.999984	T	G	0.42	1.38(1.69,1.14)	0.000228808
rs6879563	chr5:90263863	0.999984	C	T	0.42	1.38(1.69,1.14)	0.000228808
rs6883495	chr5:90263916	0.998462	A	G	0.32	1.04(1.28,0.85)	0.604114
rs17570760	chr5:90264117	1	A	G	0.15	1.6(2.13,1.2)	0.000793432
rs6865267	chr5:90264248	1	T	C	0.42	1.38(1.69,1.14)	0.000228566
rs6884254	chr5:90264394	0.999984	A	G	0.42	1.38(1.69,1.14)	0.000228808
chr5:90264895:I	chr5:90264895	0.994573	G	GA	0.41	1.37(1.67,1.13)	0.000298751
rs6871542	chr5:90265658	0.999398	T	C	0.42	1.38(1.68,1.13)	0.00028335
rs12659201	chr5:90265809	0.991702	G	A	0.41	1.38(1.68,1.13)	0.000245646
rs12653061	chr5:90265815	0.990985	T	C	0.41	1.36(1.66,1.12)	0.000382439
rs10056755	chr5:90266068	0.988089	T	C	0.41	1.41(1.72,1.16)	0.000123028
rs140288466	chr5:90266322	0.996178	G	A	0.42	1.39(1.69,1.14)	0.000205498
rs143802918	chr5:90266357	0.977481	G	C	0.41	1.34(1.63,1.1)	0.000823104
rs141749194	chr5:90266399	0.940401	C	A	0.39	1.4(1.71,1.15)	0.000111557
rs143096144	chr5:90266422	0.999957	G	A	0.15	1.6(2.13,1.2)	0.000794674
rs142191948	chr5:90266469	0.985084	T	A	0.27	1.46(1.83,1.17)	0.00077172
rs6896506	chr5:90266668	0.996841	C	T	0.42	1.4(1.71,1.15)	0.000155534
rs145529324	chr5:90267092	0.988527	C	T	0.74	1.42(1.14,1.75)	0.000348315
rs13353995	chr5:90267114	0.984863	C	G	0.27	1.46(1.83,1.17)	0.000774474
chr5:90267241:I	chr5:90267241	0.978984	G	GA	0.27	1.45(1.82,1.16)	0.000832845
rs10051346	chr5:90267487	0.998317	G	C	0.42	1.4(1.71,1.15)	0.000144343
rs140921684	chr5:90267592	0.972473	G	A	0.95	1.12(0.73,1.73)	0.614534
rs138101960	chr5:90267990	0.964261	C	T	0.12	1.18(1.59,0.87)	0.338928
rs149531961	chr5:90268006	0.993861	A	C	0.73	1.43(1.16,1.78)	0.0002389
rs7721862	chr5:90269163	0.990196	G	A	0.73	1.44(1.16,1.78)	0.00021793
rs7704311	chr5:90269166	0.991749	T	A	0.41	1.42(1.73,1.17)	8.44693E-05
rs6452918	chr5:90269287	0.991461	C	T	0.41	1.42(1.73,1.17)	8.65767E-05
rs6452919	chr5:90269349	0.977891	C	A	0.41	1.39(1.7,1.14)	0.000157366
rs6452920	chr5:90269376	0.960439	G	A	0.41	1.38(1.68,1.13)	0.000239256
rs6452921	chr5:90269512	0.915844	T	C	0.42	1.41(1.71,1.15)	7.19024E-05
rs183425621	chr5:90269836	0.989151	G	A	0.41	1.41(1.72,1.16)	0.000110633
rs192718892	chr5:90270313	0.939266	G	A	0.95	1.21(0.79,1.84)	0.418037
rs6888672	chr5:90270600	0.97874	G	A	0.74	1.42(1.14,1.76)	0.000343462
rs6889021	chr5:90270760	0.968218	C	T	0.41	1.38(1.68,1.13)	0.000220394
rs7733911	chr5:90271619	0.992501	G	T	0.41	1.39(1.7,1.14)	0.000164928
chr5:90272028:D	chr5:90272028	0.905187	AGT	A	0.13	1.12(1.5,0.84)	0.178034
rs58853660	chr5:90272033	0.970704	G	A	0.14	1.04(1.38,0.79)	0.389076
chr5:90272103:I	chr5:90272103	0.883601	ATG	A	0.72	1.37(1.11,1.7)	0.000526129
chr5:90272111:I	chr5:90272111	0.888977	G	GTA	0.35	1.07(1.31,0.88)	0.378133
rs34879738	chr5:90272197	0.990987	G	A	0.74	1.42(1.15,1.76)	0.000323138
rs67257239	chr5:90272210	0.999984	A	G	0.42	1.38(1.69,1.14)	0.000228812
rs6859871	chr5:90272623	1	C	A	0.42	1.38(1.69,1.14)	0.000228566
rs6881586	chr5:90272756	1	T	C	0.42	1.38(1.69,1.14)	0.000228566
rs6860068	chr5:90272770	1	A	G	0.42	1.38(1.69,1.14)	0.000228566
rs6881722	chr5:90272798	1	T	G	0.42	1.38(1.69,1.14)	0.000228566
rs6860535	chr5:90272836	1	G	A	0.09	1.24(1.77,0.88)	0.068818
rs6865241	chr5:90273463	0.999095	A	G	0.32	1.04(1.28,0.85)	0.615363
rs73771151	chr5:90273801	0.954799	G	A	0.12	1.2(1.63,0.89)	0.263899
rs6865883	chr5:90273851	0.990173	A	C	0.41	1.38(1.68,1.13)	0.000226271
rs7711168	chr5:90274044	0.999466	A	G	0.73	1.42(1.14,1.76)	0.000359113
rs6870988	chr5:90274531	0.999436	A	G	0.42	1.38(1.68,1.13)	0.000244458
rs6871445	chr5:90274773	0.999464	A	G	0.42	1.38(1.68,1.13)	0.000246689
rs11747245	chr5:90274888	0.999504	A	G	0.42	1.38(1.68,1.13)	0.000248142
rs11744110	chr5:90274906	0.999509	T	A	0.42	1.38(1.68,1.13)	0.000248331
rs10056721	chr5:90275034	0.995457	G	A	0.09	1.23(1.74,0.86)	0.0870121
rs11747280	chr5:90275066	0.999563	A	T	0.42	1.38(1.68,1.13)	0.000250047
rs11740777	chr5:90275276	0.99099	C	T	0.27	1.48(1.85,1.19)	0.000461205
rs11744249	chr5:90275426	0.95119	T	A	0.16	1.61(2.14,1.22)	0.000828677
rs1995776	chr5:90275825	1	T	G	0.13	1.08(1.43,0.81)	0.321189
rs1995775	chr5:90275901	1	G	A	0.32	1.04(1.28,0.85)	0.61591
rs1995774	chr5:90276193	1	G	A	0.42	1.38(1.68,1.13)	0.000258541

SNP	Chr:Position(bp)	info	Risk Allele	Other allele	RAF	OR (95% CI)	P
rs10073898	chr5:90276298	1	A	G	0.42	1.38(1.68,1.13)	0.000258541
rs17633498	chr5:90276385	0.999219	G	A	0.73	1.42(1.14,1.76)	0.000366717
rs75538711	chr5:90276625	0.96734	C	T	0.15	1.61(2.14,1.21)	0.00100809
rs6882955	chr5:90276733	0.99374	C	A	0.28	1.44(1.8,1.16)	0.000868423
rs35299092	chr5:90276745	0.957189	G	A	0.72	1.41(1.14,1.74)	0.000324175
rs6887203	chr5:90277037	0.999629	A	G	0.42	1.38(1.68,1.13)	0.000243566
rs6868512	chr5:90277048	1	T	G	0.42	1.38(1.68,1.13)	0.000258541
rs115083075	chr5:90277124	0.811441	A	G	0.98	1.99(1.08,3.67)	0.0120391
rs11742679	chr5:90277675	0.999998	C	A	0.42	1.38(1.68,1.13)	0.000258499
chr5:90277785:I	chr5:90277785	0.962612	A	AAAT	0.4	1.36(1.66,1.12)	0.000407436
chr5:90277788:I	chr5:90277788	0.94499	T	TAA	0.4	1.35(1.64,1.1)	0.000566651
rs60325490	chr5:90277797	0.995206	T	C	0.42	1.37(1.67,1.13)	0.000289496
rs11742722	chr5:90277863	0.995525	C	T	0.42	1.37(1.67,1.13)	0.000279998
rs12188094	chr5:90278217	0.993094	T	A	0.73	1.44(1.16,1.78)	0.000252096
chr5:90278563:I	chr5:90278563	0.995831	C	CT	0.32	1.03(1.27,0.84)	0.676203
rs10054039	chr5:90279146	1	G	A	0.42	1.38(1.68,1.13)	0.000258541
rs12514684	chr5:90279293	1	C	T	0.42	1.36(1.66,1.12)	0.000371647
rs17633569	chr5:90279414	1	C	T	0.42	1.37(1.67,1.13)	0.000291393
rs16869305	chr5:90279442	1	G	A	0.42	1.37(1.67,1.13)	0.000291393
rs12187656	chr5:90279828	0.95035	C	T	0.76	1.47(1.18,1.84)	0.000117209
rs12186968	chr5:90279903	0.935622	T	C	0.75	1.39(1.12,1.73)	0.000678898
rs41305906	chr5:90281045	0.876082	A	G	0.03	1.36(2.38,0.78)	0.27636
rs62374052	chr5:90285075	0.89943	T	C	0.77	1.49(1.19,1.86)	4.52204E-05
chr5:90285392:D	chr5:90285392	0.911228	C	CT	0.66	1.15(0.94,1.41)	0.136156
rs10474338	chr5:90285899	0.838835	C	T	0.18	1.31(1.7,1.01)	0.019255
rs76312753	chr5:90286608	0.827788	T	C	0.01	1.54(3.71,0.64)	0.237106
rs17571029	chr5:90286831	0.909508	T	C	0.76	1.49(1.19,1.86)	4.99318E-05
rs78353640	chr5:90289347	0.877785	G	C	0.09	1.65(2.38,1.14)	0.00445603
rs11744033	chr5:90289617	0.899659	T	G	0.76	1.49(1.19,1.85)	5.14394E-05
rs4916836	chr5:90289914	0.875559	G	A	0.56	1.02(0.84,1.24)	0.998373
rs59805249	chr5:90289997	0.970415	C	T	0.1	1.55(2.17,1.11)	0.00273869
rs56099704	chr5:90290005	0.899664	C	A	0.76	1.49(1.19,1.85)	5.14209E-05
rs7721879	chr5:90290033	0.867933	T	C	0.46	1.01(1.23,0.84)	0.852432
chr5:90291981:I	chr5:90291981	0.848183	T	TG	0.2	1.24(1.58,0.97)	0.0447591
chr5:90293189:D	chr5:90293189	0.849015	GCTCT	G	0.2	1.26(1.61,0.98)	0.0338724
rs76392284	chr5:90294545	0.85437	C	T	0.2	1.29(1.65,1.01)	0.0220044
rs60553973	chr5:90294707	0.986181	G	A	0.1	1.58(2.23,1.12)	0.00295149
rs6452927	chr5:90296182	0.900088	A	G	0.76	1.48(1.19,1.85)	5.85561E-05
rs1967253	chr5:90297516	0.99664	A	G	0.1	1.62(2.28,1.15)	0.00188093
rs1967254	chr5:90297574	0.999885	A	G	0.1	1.67(2.35,1.18)	0.000856722
rs10942623	chr5:90297868	0.873613	T	G	0.45	1.02(1.24,0.84)	0.772341
rs13355688	chr5:90297911	0.861653	A	G	0.2	1.26(1.61,0.98)	0.0355885
rs10514341	chr5:90298632	0.848405	A	C	0.2	1.28(1.64,1)	0.0235821
rs10514342	chr5:90298762	0.900108	T	A	0.76	1.48(1.19,1.85)	0.00005849
rs1967255	chr5:90299069	0.999885	T	A	0.1	1.67(2.35,1.18)	0.000856722
rs1967256	chr5:90299223	1	G	C	0.1	1.67(2.35,1.18)	0.000858673
rs2367184	chr5:90299453	0.999338	A	G	0.1	1.67(2.35,1.18)	0.000848359
rs12521660	chr5:90299817	0.999344	G	C	0.1	1.67(2.35,1.18)	0.000848117
rs10053919	chr5:90300049	0.842676	T	G	0.2	1.26(1.6,0.98)	0.0329117
chr5:90300419:I	chr5:90300419	0.816164	ACT	A	0.59	1(0.82,1.22)	0.854449
rs16869328	chr5:90300468	0.999885	T	C	0.1	1.67(2.35,1.18)	0.000856722
chr5:90301196:I	chr5:90301196	0.986922	T	TA	0.1	1.59(2.25,1.12)	0.00257789
rs16869330	chr5:90301599	0.999885	A	G	0.1	1.67(2.35,1.18)	0.000856722
rs16869331	chr5:90301692	0.999861	T	A	0.1	1.67(2.35,1.18)	0.000856938
rs16869333	chr5:90301764	0.999885	G	A	0.1	1.67(2.35,1.18)	0.000856722
rs16869335	chr5:90301862	0.999263	G	A	0.1	1.67(2.35,1.18)	0.000846425
rs16869336	chr5:90301924	0.999885	G	T	0.1	1.67(2.35,1.18)	0.000856722
rs73187122	chr5:90302004	0.998756	C	T	0.1	1.67(2.35,1.18)	0.000848409
rs73187123	chr5:90302037	0.999885	C	T	0.1	1.67(2.35,1.18)	0.000856722
rs73187124	chr5:90302041	0.999885	T	C	0.1	1.67(2.35,1.18)	0.000856722
rs11954387	chr5:90302117	1	A	G	0.1	1.67(2.35,1.18)	0.000858673
chr5:90302590:I	chr5:90302590	0.999209	T	TA	0.1	1.67(2.35,1.18)	0.000856547
chr5:90303225:D	chr5:90303225	0.997799	CT	C	0.1	1.67(2.35,1.18)	0.000841552
rs58523233	chr5:90303339	0.863639	C	T	0.2	1.26(1.61,0.99)	0.0357005

SNP	Chr:Position(bp)	info	Risk Allele	Other allele	RAF	OR (95% CI)	P
rs6886773	chr5:90303618	0.887029	A	T	0.76	1.49(1.2,1.86)	3.08627E-05
rs11952452	chr5:90303764	0.994571	T	C	0.1	1.67(2.35,1.18)	0.000852448
rs11949884	chr5:90304531	0.892912	A	G	0.76	1.47(1.18,1.83)	0.000072659
rs7711918	chr5:90304577	0.87865	T	G	0.56	1.17(0.96,1.42)	0.0661353
chr5:90305474:I	chr5:90305474	0.86438	A	AT	0.19	1.31(1.68,1.02)	0.018986
chr5:90305479:I	chr5:90305479	0.863161	A	AT	0.19	1.31(1.68,1.02)	0.0185227
rs78720904	chr5:90305479	0.864374	A	T	0.19	1.31(1.68,1.02)	0.0189886
rs9293556	chr5:90305586	0.880042	T	C	0.46	1.02(1.24,0.84)	0.790708
rs10058791	chr5:90305962	0.879763	G	A	0.46	1.02(1.24,0.84)	0.793585
rs73187127	chr5:90305979	0.962341	A	G	0.1	1.68(2.36,1.2)	0.000549317
rs17571867	chr5:90306430	0.908483	C	T	0.03	2.56(5.2,1.26)	0.00515638
rs11951675	chr5:90306443	0.895349	T	G	0.76	1.48(1.18,1.84)	6.01272E-05
chr5:90306606:D	chr5:90306606	0.811584	AATTT	A	0.13	1.37(1.85,1.02)	0.0169884
rs16869341	chr5:90306939	0.88177	A	G	0.16	1.43(1.88,1.09)	0.00313156
rs7726848	chr5:90307152	0.871839	C	T	0.46	1.02(1.23,0.84)	0.819045
rs73771163	chr5:90307161	0.867353	A	G	0.16	1.42(1.86,1.08)	0.00303351
rs10058319	chr5:90309026	0.964846	G	A	0.68	1.06(0.86,1.3)	0.715327
rs4916838	chr5:90309735	0.967975	A	G	0.68	1.06(0.86,1.3)	0.706819
rs28875340	chr5:90309787	0.946578	C	T	0.37	1.38(1.68,1.12)	0.00104487
chr5:90310443:D	chr5:90310443	0.964057	G	GTA	0.68	1.06(0.86,1.3)	0.709737
rs1848481	chr5:90310720	0.97304	A	G	0.68	1.06(0.86,1.3)	0.694552
rs10055587	chr5:90311099	0.97514	G	T	0.68	1.06(0.86,1.3)	0.69084
rs4391205	chr5:90311482	0.977305	G	T	0.68	1.06(0.86,1.31)	0.686622
rs4916839	chr5:90311758	0.978981	T	C	0.68	1.06(0.86,1.31)	0.683915
rs4916840	chr5:90313045	0.98748	T	C	0.68	1.06(0.86,1.31)	0.671611
rs62376462	chr5:90313671	0.91906	C	T	0.76	1.49(1.19,1.85)	7.11478E-05
rs62376463	chr5:90314407	0.930896	G	C	0.75	1.49(1.2,1.86)	6.80287E-05
rs2367185	chr5:90314653	1	A	G	0.68	1.07(0.87,1.32)	0.566273
rs6452928	chr5:90314780	0.999095	A	C	0.69	1.07(0.87,1.32)	0.630632
rs1848487	chr5:90314865	0.99856	C	T	0.38	1.38(1.68,1.12)	0.00125134
rs1848486	chr5:90314905	0.998735	C	T	0.38	1.38(1.68,1.13)	0.00124844
rs1848485	chr5:90314923	1	C	G	0.69	1.07(0.87,1.32)	0.617895
rs1848484	chr5:90315036	0.999623	G	A	0.69	1.07(0.87,1.32)	0.611646
chr5:90315408:I	chr5:90315408	0.993509	GA	G	0.68	1.08(0.88,1.33)	0.547043
rs937920	chr5:90315818	1	C	G	0.38	1.42(1.73,1.16)	0.000512383
chr5:90315906:D	chr5:90315906	0.983275	CAT	C	0.38	1.39(1.7,1.13)	0.000834354
rs10038122	chr5:90316643	1	T	C	0.69	1.07(0.87,1.32)	0.617895
rs1848483	chr5:90316782	0.994086	A	G	0.68	1.07(0.87,1.31)	0.594912
rs1976565	chr5:90317558	0.948135	C	A	0.68	1.06(0.86,1.3)	0.713437
rs998788	chr5:90318391	0.892514	A	C	0.64	1.02(0.84,1.25)	0.842046
rs56258383	chr5:90319894	0.871536	A	G	0.76	1.49(1.2,1.86)	5.72504E-05
rs2697546	chr5:90321288	1	G	A	0.62	1.36(1.11,1.66)	0.0018724
rs2662283	chr5:90322028	1	T	G	0.62	1.36(1.11,1.67)	0.00166511
rs10514343	chr5:90322671	1	C	G	0.25	1.68(2.09,1.35)	3.62507E-07
chr5:90323846:D	chr5:90323846	0.852307	T	TTTTC	0.76	1.51(1.21,1.88)	2.27935E-05
rs2662282	chr5:90326582	0.992952	C	T	0.63	1.41(1.15,1.73)	0.000524829
rs2697547	chr5:90327430	0.933322	G	C	0.32	1.06(1.31,0.87)	0.648625
rs689544	chr5:90328537	0.892576	A	G	0.36	1.05(1.28,0.86)	0.691902
rs72784636	chr5:90328806	0.983041	A	G	0.94	1.06(0.71,1.59)	0.660138
rs2662281	chr5:90328927	0.892754	G	A	0.36	1.05(1.28,0.86)	0.676581
rs2662280	chr5:90329458	1	C	T	0.66	1.46(1.18,1.79)	0.000253177
rs2697548	chr5:90330959	0.963641	C	A	0.79	1.39(1.09,1.77)	0.00671718
rs188859916	chr5:90331504	0.849299	A	G	0.02	1.35(3.08,0.59)	0.488844
rs13179479	chr5:90331627	0.966851	C	T	0.62	1.39(1.13,1.7)	0.000539383
rs73771166	chr5:90331709	0.921666	G	A	0.76	1.67(1.34,2.08)	2.01616E-07
chr5:90331921:D	chr5:90331921	0.954003	AT	A	0.38	1.1(1.34,0.9)	0.356954
rs2662279	chr5:90333141	0.981497	A	G	0.61	1.34(1.1,1.64)	0.0013093
chr5:90333284:D	chr5:90333284	0.942578	T	TATC	0.76	1.65(1.33,2.06)	3.82314E-07
rs137855265	chr5:90333404	0.847808	C	T	0.02	2.15(4.88,0.95)	0.0433342
rs1818349	chr5:90333636	0.96653	C	T	0.78	1.34(1.05,1.7)	0.0135676
rs79665583	chr5:90333956	0.886271	G	A	0.11	1.29(1.77,0.94)	0.0670796
rs6888979	chr5:90335536	0.96465	A	G	0.75	1.67(1.34,2.08)	3.62026E-07
rs6889138	chr5:90335635	0.953164	A	G	0.76	1.68(1.35,2.1)	1.93061E-07
rs2048217	chr5:90336531	0.990056	T	C	0.36	1.13(1.38,0.93)	0.237251

SNP	Chr:Position(bp)	info	Risk Allele	Other allele	RAF	OR (95% CI)	P
chr5:90337352:I	chr5:90337352	0.88206	A	AT	0.11	1.29(1.77,0.94)	0.066466
rs2367186	chr5:90337413	0.98721	C	T	0.36	1.13(1.38,0.92)	0.265005
rs3850657	chr5:90337465	0.958369	G	A	0.75	1.71(1.38,2.13)	1.20248E-07
rs1858070	chr5:90337957	1	C	G	0.36	1.14(1.39,0.93)	0.221252
rs1828178	chr5:90339314	0.99873	C	T	0.36	1.14(1.4,0.93)	0.209176
rs55792004	chr5:90339780	0.975574	A	G	0.75	1.7(1.37,2.11)	1.54952E-07
rs1828177	chr5:90340068	0.998404	G	A	0.36	1.14(1.4,0.93)	0.209244
rs2463775	chr5:90340631	0.987568	T	C	0.61	1.36(1.11,1.66)	0.00109626
rs7449334	chr5:90342412	0.963032	A	G	0.78	1.29(1.02,1.63)	0.0233187
rs56106025	chr5:90342845	0.991562	A	G	0.75	1.69(1.36,2.1)	2.95882E-07
rs1830364	chr5:90342978	0.963498	A	C	0.78	1.29(1.02,1.63)	0.0235279
rs1876635	chr5:90343578	0.821758	G	A	0.88	1.02(0.76,1.38)	0.921679
rs188342788	chr5:90343840	0.806083	A	C	0.01	1.3(3.35,0.51)	0.576799
rs11955946	chr5:90344018	1	G	A	0.75	1.7(1.37,2.11)	2.47172E-07
rs11952993	chr5:90344149	1	C	T	0.75	1.7(1.37,2.11)	2.77345E-07
rs1818353	chr5:90344890	0.954528	A	T	0.16	1.24(1.62,0.95)	0.0888237
rs17572752	chr5:90344923	1	G	A	0.06	1.01(1.51,0.68)	0.908168
rs6861614	chr5:90346339	0.978874	G	T	0.76	1.67(1.34,2.08)	3.45947E-07
rs6880626	chr5:90346380	0.969626	T	A	0.76	1.67(1.34,2.08)	3.52442E-07
rs12189005	chr5:90347289	0.978572	C	A	0.76	1.67(1.34,2.08)	3.42522E-07
rs12188068	chr5:90348718	0.978104	T	A	0.76	1.67(1.34,2.08)	3.39224E-07
rs2697542	chr5:90348787	0.963646	A	G	0.78	1.35(1.06,1.72)	0.0100611
rs12188249	chr5:90349263	0.960578	G	A	0.76	1.67(1.34,2.08)	3.48802E-07
rs2010355	chr5:90349562	0.911569	C	A	0.53	1.37(1.13,1.66)	0.00066203
rs17572843	chr5:90350227	0.975872	A	C	0.76	1.67(1.35,2.09)	3.2591E-07
rs62376490	chr5:90350502	0.97588	G	T	0.76	1.67(1.35,2.09)	3.26144E-07
rs62376491	chr5:90350577	0.974197	A	G	0.76	1.68(1.35,2.09)	3.11983E-07
rs2697543	chr5:90350959	1	G	A	0.62	1.36(1.11,1.67)	0.00115456
rs16869392	chr5:90350998	0.957012	T	C	0.17	1.2(1.56,0.92)	0.116681
rs2697544	chr5:90351150	0.999046	A	T	0.61	1.35(1.11,1.65)	0.0011762
chr5:90351213:D	chr5:90351213	0.998328	T	TTAATGA	0.62	1.35(1.11,1.65)	0.00106519
rs2662284	chr5:90351463	0.999703	A	T	0.61	1.36(1.11,1.66)	0.000995876
rs7719311	chr5:90351699	0.999679	G	A	0.62	1.36(1.11,1.66)	0.00101912
rs79956589	chr5:90351791	0.957036	T	G	0.17	1.19(1.56,0.92)	0.116843
rs7723511	chr5:90351976	0.995567	G	A	0.62	1.36(1.11,1.66)	0.00121825
chr5:90352092:D	chr5:90352092	0.898365	A	AG	0.52	1.28(1.05,1.55)	0.00242233
rs6452930	chr5:90352220	0.999702	T	C	0.61	1.36(1.11,1.66)	0.000995714
rs2367274	chr5:90352302	0.999702	A	G	0.61	1.36(1.11,1.66)	0.000995714
rs4454077	chr5:90352309	0.999702	C	T	0.61	1.36(1.11,1.66)	0.000995714
rs4254936	chr5:90352452	0.995331	T	G	0.61	1.34(1.09,1.63)	0.00214973
rs6452934	chr5:90352694	0.999701	G	A	0.61	1.36(1.11,1.66)	0.000995432
rs77058932	chr5:90354099	0.956789	T	C	0.16	1.25(1.64,0.96)	0.0631486
rs2662278	chr5:90354265	0.999059	A	G	0.62	1.36(1.11,1.66)	0.00129986
rs12186662	chr5:90356197	0.920021	G	A	0.61	1.28(1.05,1.56)	0.00587332
chr5:90356207:D	chr5:90356207	0.936908	A	ATATATATATT	0.64	1.32(1.08,1.61)	0.00250293
rs66608099	chr5:90356564	0.812414	A	G	0.88	1.08(0.8,1.45)	0.632367
rs1852734	chr5:90356690	0.999011	G	A	0.62	1.36(1.11,1.66)	0.00130135
rs16869403	chr5:90356952	0.812396	G	C	0.88	1.08(0.8,1.45)	0.632517
rs10942624	chr5:90357301	0.998932	G	A	0.62	1.36(1.11,1.66)	0.000900059
rs13180409	chr5:90358300	1	G	A	0.62	1.36(1.11,1.66)	0.00103376
rs7706034	chr5:90361061	0.992223	A	G	0.62	1.36(1.11,1.66)	0.000949847
rs6865615	chr5:90361488	0.991993	G	A	0.62	1.37(1.12,1.67)	0.000772715
rs1819074	chr5:90362010	0.992494	A	G	0.62	1.36(1.12,1.67)	0.000983193
rs1819073	chr5:90362374	0.991897	A	G	0.62	1.36(1.11,1.67)	0.0010117
chr5:90363051:I	chr5:90363051	0.955514	TC	T	0.61	1.37(1.12,1.67)	0.000656725
rs6882081	chr5:90364101	0.954749	G	A	0.61	1.4(1.15,1.71)	0.000259981
rs6452935	chr5:90364625	0.885349	C	A	0.59	1.37(1.12,1.67)	0.000362108
rs78873707	chr5:90365337	0.934505	A	G	0.16	1.28(1.68,0.98)	0.0413362
rs187270236	chr5:90365703	0.892012	A	G	0.02	1.14(2.51,0.52)	0.802463
rs7733230	chr5:90366272	0.971407	A	G	0.63	1.38(1.13,1.69)	0.000639925
rs13152882	chr5:90366991	0.863776	A	G	0.52	1.33(1.1,1.62)	0.000293756
rs9293558	chr5:90368596	0.968513	T	C	0.79	1.34(1.05,1.7)	0.0181667
rs10058592	chr5:90369605	0.933244	A	G	0.16	1.27(1.66,0.97)	0.0488013
rs951948	chr5:90369887	0.862202	A	C	0.52	1.33(1.1,1.62)	0.000279779

SNP	Chr:Position(bp)	info	Risk Allele	Other allele	RAF	OR (95% CI)	P
rs12658697	chr5:90370142	0.861102	G	C	0.52	1.34(1.1,1.62)	0.000268543
rs76879861	chr5:90370635	0.846308	C	T	0.04	1.21(1.98,0.74)	0.467249
rs10037172	chr5:90371668	0.967472	C	T	0.79	1.34(1.05,1.71)	0.0170004
rs2007538	chr5:90374713	0.932564	T	C	0.16	1.28(1.68,0.98)	0.0435372
rs1819072	chr5:90375076	0.929601	G	A	0.16	1.26(1.65,0.96)	0.0544378
rs12652491	chr5:90378971	0.844359	T	C	0.52	1.32(1.09,1.6)	0.000440025
rs12519581	chr5:90379153	0.92567	A	G	0.16	1.27(1.66,0.97)	0.0470391
rs4386756	chr5:90379878	0.845824	G	A	0.52	1.32(1.09,1.6)	0.000381604
rs12657388	chr5:90380845	0.844066	G	A	0.52	1.32(1.09,1.6)	0.00035435
rs10454910	chr5:90382130	0.836218	A	C	0.54	1.29(1.06,1.56)	0.000870546
rs12153127	chr5:90383216	0.824366	T	C	0.55	1.29(1.06,1.56)	0.000663166
rs16869425	chr5:90385098	1	C	G	0.9	1.2(0.87,1.66)	0.216066
chr5:90385242:I	chr5:90385242	0.872276	T	TA	0.15	1.3(1.72,0.98)	0.0438435
rs10068613	chr5:90386514	0.903842	G	T	0.06	1.05(1.56,0.71)	0.685557
rs4244206	chr5:90389450	1	C	A	0.79	1.36(1.06,1.73)	0.015725
rs12518689	chr5:90390580	0.825609	A	G	0.57	1.22(1,1.48)	0.011742
rs4608951	chr5:90391034	0.81462	G	T	0.56	1.23(1.01,1.5)	0.00766685
rs76173152	chr5:90391920	0.822059	T	C	0.05	1.01(1.57,0.65)	0.929654
rs75117248	chr5:90391921	0.822067	A	C	0.05	1.01(1.57,0.65)	0.929727
rs1852731	chr5:90392621	1	A	G	0.11	1.12(1.54,0.82)	0.368053
rs2950849	chr5:90394625	0.962571	A	G	0.11	1.06(1.44,0.78)	0.591739
rs2367279	chr5:90395206	0.833335	T	G	0.86	1.16(0.88,1.53)	0.199048
rs59918121	chr5:90395861	0.893173	G	A	0.06	1.1(1.64,0.74)	0.593009
rs2973456	chr5:90395882	0.934476	T	A	0.1	1.11(1.54,0.8)	0.4417
rs59654682	chr5:90395918	0.934476	T	C	0.1	1.11(1.54,0.8)	0.4417
rs2973455	chr5:90396152	0.934485	G	A	0.1	1.11(1.54,0.8)	0.441593
rs2973454	chr5:90396790	0.934517	G	A	0.1	1.11(1.54,0.8)	0.441454
chr5:90396841:D	chr5:90396841	0.934517	ATC	A	0.1	1.11(1.54,0.8)	0.441454
rs2950850	chr5:90397333	0.934528	G	A	0.1	1.11(1.54,0.8)	0.441406
chr5:90397585:D	chr5:90397585	0.934298	AACAC	A	0.1	1.11(1.55,0.8)	0.440468
chr5:90397594:D	chr5:90397594	0.931322	ACACG	A	0.1	1.11(1.55,0.8)	0.445001
rs3749606	chr5:90397917	0.898484	C	T	0.07	1.07(1.59,0.73)	0.677225
rs3749605	chr5:90398014	0.898484	A	G	0.07	1.07(1.59,0.73)	0.677225
rs1986718	chr5:90398269	0.928401	T	C	0.1	1.14(1.58,0.82)	0.362053
rs2950851	chr5:90399012	0.92841	A	T	0.1	1.14(1.58,0.82)	0.362102
rs56377639	chr5:90399338	0.928417	A	G	0.1	1.14(1.58,0.82)	0.362083
rs2973453	chr5:90399822	0.928432	T	C	0.1	1.14(1.58,0.82)	0.362102
rs2367280	chr5:90400483	0.945318	G	A	0.59	1.27(1.04,1.54)	0.00643972
rs3112484	chr5:90401221	0.928455	G	A	0.1	1.14(1.58,0.82)	0.361996
rs7731652	chr5:90401398	0.944784	C	G	0.59	1.26(1.04,1.53)	0.00723891
chr5:90403009:I	chr5:90403009	0.930214	A	AG	0.1	1.16(1.61,0.83)	0.31182
rs12109264	chr5:90404006	0.929845	T	C	0.1	1.16(1.61,0.83)	0.309873
chr5:90404285:I	chr5:90404285	0.975358	TG	T	0.79	1.36(1.07,1.73)	0.0130424
chr5:90404416:I	chr5:90404416	0.898126	C	CT	0.07	1.08(1.6,0.73)	0.649489
rs73189008	chr5:90404615	0.929398	T	A	0.1	1.16(1.61,0.83)	0.310632
rs10071291	chr5:90405621	0.898669	A	G	0.07	1.07(1.59,0.73)	0.676972
rs188703823	chr5:90405691	0.875341	T	C	0.02	1.26(2.77,0.58)	0.595753
rs3952745	chr5:90405835	0.928454	A	G	0.1	1.14(1.58,0.82)	0.361023
chr5:90406703:I	chr5:90406703	0.909012	T	TG	0.1	1.13(1.57,0.82)	0.372742
rs2973450	chr5:90406998	0.9169	A	G	0.1	1.12(1.56,0.81)	0.402007
rs4512153	chr5:90407022	0.945108	G	A	0.59	1.26(1.03,1.53)	0.00731232
rs79973531	chr5:90407299	0.90164	A	G	0.07	1.07(1.58,0.72)	0.712788
rs2973449	chr5:90407743	0.916913	T	C	0.1	1.12(1.56,0.81)	0.402139
rs2973448	chr5:90408139	0.916926	A	C	0.1	1.12(1.56,0.81)	0.402062
rs76394730	chr5:90408471	0.897182	A	G	0.07	1.05(1.54,0.71)	0.763911
rs2973447	chr5:90409031	0.919442	C	A	0.1	1.15(1.59,0.82)	0.347247
rs2950852	chr5:90409081	0.919884	C	A	0.1	1.15(1.6,0.82)	0.347615
chr5:90409360:D	chr5:90409360	0.900521	AGAGAGT	A	0.06	1.18(1.77,0.78)	0.433506
rs2367282	chr5:90409739	0.999419	G	A	0.62	1.33(1.09,1.63)	0.00353783
rs2950853	chr5:90410295	0.920454	G	A	0.1	1.15(1.6,0.82)	0.346982
rs7707023	chr5:90410424	1	C	T	0.62	1.33(1.09,1.63)	0.00356136
rs10942625	chr5:90410692	1	A	G	0.61	1.3(1.06,1.58)	0.0041008
rs13357230	chr5:90411072	0.853891	A	G	0.07	1.07(1.59,0.73)	0.681168
rs1852732	chr5:90412371	1	C	G	0.39	1.3(1.59,1.07)	0.00361807

SNP	Chr:Position(bp)	info	Risk Allele	Other allele	RAF	OR (95% CI)	P
rs2973446	chr5:90414407	0.823587	C	A	0.79	1.35(1.06,1.73)	0.00800254
rs2367284	chr5:90418032	0.831714	A	G	0.79	1.35(1.06,1.72)	0.00962349
rs113091354	chr5:90423350	0.828451	G	C	0.05	1.1(1.73,0.71)	0.695043
rs10514345	chr5:90424279	1	T	C	0.88	1.2(0.9,1.6)	0.324861
rs4244207	chr5:90425042	0.948899	T	C	0.17	1.22(1.57,0.94)	0.355792
chr5:90425081:D	chr5:90425081	0.994493	T	TA	0.89	1.11(0.82,1.5)	0.478029
rs13158036	chr5:90425951	0.983919	C	A	0.89	1.08(0.8,1.46)	0.644432
rs2950858	chr5:90426633	0.96671	A	G	0.15	1.21(1.58,0.93)	0.393645
rs13163651	chr5:90427048	1	T	C	0.89	1.1(0.81,1.49)	0.524114
rs13190101	chr5:90430477	0.999735	A	C	0.89	1.1(0.81,1.49)	0.522528
rs13158026	chr5:90431779	0.98768	T	C	0.89	1.08(0.8,1.46)	0.635615
rs13161782	chr5:90431872	0.983753	C	A	0.89	1.08(0.8,1.46)	0.634646
rs55736649	chr5:90431974	0.982845	A	G	0.25	1.11(1.38,0.89)	0.704538
rs13161956	chr5:90431982	0.983209	G	A	0.89	1.08(0.8,1.46)	0.646476
rs2367286	chr5:90432314	0.942342	A	G	0.1	1.01(1.38,0.73)	0.887928
rs60378516	chr5:90433852	0.827877	A	G	0.99	2.06(0.84,5.06)	0.163677
rs16869507	chr5:90434109	0.811262	C	T	0.03	2.31(4.66,1.14)	0.0030603
rs10055598	chr5:90436277	0.98754	T	G	0.25	1.13(1.41,0.91)	0.591897
rs6873467	chr5:90436372	0.972629	G	T	0.84	1.09(0.84,1.41)	0.774732
chr5:90438092:I	chr5:90438092	0.839177	CA	C	0.99	1.88(0.81,4.35)	0.218693
rs59763490	chr5:90438189	0.839199	A	G	0.99	1.88(0.81,4.34)	0.218923
rs6873773	chr5:90438473	1	T	C	0.88	1.06(0.8,1.42)	0.848616
rs13172451	chr5:90439812	0.995336	T	C	0.88	1.17(0.87,1.58)	0.26619
rs4458614	chr5:90441146	1	G	A	0.27	1.09(1.35,0.88)	0.776412
rs34302350	chr5:90441702	0.975459	T	C	0.86	1.16(0.88,1.52)	0.523865
rs73771197	chr5:90442505	0.83507	A	G	0.98	1.75(0.82,3.71)	0.288431
rs4331933	chr5:90443593	0.979877	G	A	0.28	1.1(1.36,0.89)	0.704575
rs4336388	chr5:90444207	0.90002	G	C	0.09	1.07(1.5,0.76)	0.849353
rs4563659	chr5:90444664	0.967703	G	A	0.25	1.14(1.42,0.91)	0.521862
rs142639870	chr5:90444665	0.848095	C	T	0.02	2.37(5.18,1.08)	0.00678831
rs79787775	chr5:90444735	0.823315	C	T	0.99	1.83(0.79,4.27)	0.249259
rs147286604	chr5:90445479	0.819938	A	G	0.99	1.77(0.79,3.95)	0.269518
chr5:90445828:I	chr5:90445828	0.885413	T	TTTC	0.25	1.1(1.38,0.89)	0.636762
rs77469944	chr5:90445889	0.818937	G	A	0.99	1.77(0.79,3.95)	0.270237
rs73771199	chr5:90446283	0.804699	T	C	0.99	1.88(0.8,4.44)	0.222837
rs11745446	chr5:90447132	0.933958	T	A	0.07	1.02(1.48,0.71)	0.94089
rs13176728	chr5:90447216	1	A	G	0.87	1.09(0.82,1.44)	0.476858
rs4540211	chr5:90448114	0.97777	T	C	0.22	1.14(1.44,0.9)	0.519175
rs7726023	chr5:90449031	0.944821	C	T	0.93	1.03(0.71,1.51)	0.767764
rs13158963	chr5:90449154	0.990144	A	G	0.88	1.11(0.82,1.48)	0.556616
rs13160072	chr5:90449807	1	A	G	0.89	1.15(0.85,1.55)	0.318739
rs55980700	chr5:90449932	0.961311	T	C	0.22	1.19(1.49,0.94)	0.265003
rs13184171	chr5:90450113	0.989691	C	T	0.88	1.11(0.83,1.49)	0.548957
rs13169196	chr5:90451039	0.969579	G	A	0.86	1.07(0.81,1.4)	0.681747
rs4916703	chr5:90452758	0.952367	C	T	0.22	1.07(1.35,0.85)	0.697822
rs4916847	chr5:90453191	0.942872	G	C	0.08	1(1.43,0.7)	0.919102
rs4916704	chr5:90453242	0.941126	T	C	0.11	1.1(1.49,0.82)	0.956441
rs6864663	chr5:90455872	0.963115	C	T	0.24	1.14(1.42,0.91)	0.365867
rs6864534	chr5:90455923	0.940133	A	C	0.09	1.04(1.45,0.74)	0.967217
rs10069710	chr5:90457064	0.821098	C	A	0.03	1.14(2.07,0.63)	0.704866
rs145981926	chr5:90457773	0.930329	G	C	0.02	1.17(2.43,0.56)	0.705751
rs4916848	chr5:90458126	0.965006	C	A	0.24	1.14(1.42,0.91)	0.423848
rs13179009	chr5:90458169	0.960338	C	G	0.87	1.13(0.86,1.5)	0.328
rs4916705	chr5:90458387	0.913053	G	A	0.09	1.03(1.44,0.74)	0.992061
rs10077486	chr5:90459091	0.945433	A	G	0.09	1.05(1.48,0.75)	0.967256

Supplementary Table 6. SNP data from the GPR98 locus. The table indicates the SNP, chromosome position, info, risk allele, other allele, RAF, OR, p-value for 1,344 SNPs in the vicinity of the top SNP for *GPR98*. Info is a metric is similar to the r-squared metrics reported

by other programs like MaCH and Beagle. This metric typically takes values between 0 and 1, where values near 1 indicate that a SNP has been imputed with high certainty.

SNVs	Chr	Position	Ref	Alt	Major Allele	Minor Allele Frequency	Classification	Coding Classification	Gene(s)	Transcript	Exon	HGVS Coding	HGVS Protein	HGVS Description
chr5_88718759	5	88014576	A	C	C	0.001259446	UTR3		MEF2C					g.88014576A>C
rs577761885	5	88014579	C	A	A	0.001259446	UTR3		MEF2C					g.88014579C>A
rs150173081	5	88014838	G	T	T	0.007594937	UTR3		MEF2C					g.88014838G>T
rs145654234	5	88015123	C	A	A	0.002525253	UTR3		MEF2C					g.88015123C>A
chr5_88719583	5	88015400	T	C	C	0.001262626	UTR3		MEF2C					g.88015400T>C
chr5_88719613	5	88015430	T	A	A	0.001259446	UTR3		MEF2C					g.88015430T>A
rs34316	5	88015545	A	C	C	0.431472081	UTR3		MEF2C					g.88015545A>C
chr5_88719889	5	88015706	G	A	A	0.001259446	UTR3		MEF2C					g.88015706G>A
rs34317	5	88016008	A	G	G	0.229113924	UTR3		MEF2C					g.88016008A>G
rs17550568	5	88016068	G	T	T	0.003807107	UTR3		MEF2C					g.88016068G>T
chr5_88721592	5	88017409	G	A	A	0.002518892	UTR3		MEF2C					g.88017409G>A
rs139432368	5	88017615	C	T	T	0.003778338	UTR3		MEF2C					g.88017615C>T
chr5_88722286	5	88018103	C	G	G	0.001259446	UTR3		MEF2C					g.88018103C>G
rs536118906	5	88018257	A	G	G	0.001262626	UTR3		MEF2C					g.88018257A>G
chr5_88760989	5	88056806	A	G	G	0.001259446	Coding	Nonsyn SNV	MEF2C	XM_005248516.1	4	c.395T>C	p.Leu132Pro	g.88056806A>G
rs201296342	5	88056886	G	A	A	0.001259446	Coding	Synonymous	MEF2C	XM_005248516.1	4	c.315C>T	p.=	g.88056886G>A
rs186648089	5	88057041	A	G	G	0.001259446	Coding	Synonymous	MEF2C	XM_005248513.1	4	c.363T>C	p.=	g.88057041A>G
chr5_88882979	5	88178796	T	G	G	0.001259446	UTR5		MEF2C					g.88178796T>G
chr5_88887529	5	88183346	T	A	A	0.001259446	UTR5		MEF2C					g.88183346T>A
rs553991524	5	88185024	G	A	A	0.001259446	UTR5		MEF2C					g.88185024G>A
rs72773826	5	88185126	G	A	A	0.003778338	UTR5		MEF2C					g.88185126G>A
chr5_90393726	5	89689543	G	A	A	0.001259446	UTR3		CETN3					g.89689543G>A
chr5_90394021	5	89689838	G	A	A	0.001259446	UTR3		CETN3					g.89689838G>A
chr5_90395830	5	89691647	C	T	T	0.001259446	UTR3		CETN3					g.89691647C>T
chr5_90396222	5	89692039	C	T	T	0.001259446	UTR3		CETN3					g.89692039C>T
chr5_90396266	5	89692083	G	A	A	0.001259446	UTR3		CETN3					g.89692083G>A
chr5_90396418	5	89692235	C	G	G	0.001259446	UTR3		CETN3					g.89692235C>G
chr5_90396485	5	89692302	T	G	G	0.001259446	Coding	Nonsyn SNV	CETN3	XM_005248408.1	5	c.540A>C	p.Gln180His	g.89692302T>G
rs200441218	5	89692333	G	A	A	0.001259446	Coding	Nonsyn SNV	CETN3	XM_005248408.1	5	c.509C>T	p.Pro170Leu	g.89692333G>A
rs4873	5	89703641	G	C	C	0.234848485	Coding	Nonsyn SNV	CETN3	NM_004365.2	2	c.28C>G	p.Leu10Val	g.89703641G>C
rs150819002	5	89754331	T	C	C	0.002518892	UTR3		MBLAC2					g.89754331T>C
rs536154708	5	89754592	C	G	G	0.001259446	UTR3		MBLAC2					g.89754592C>G
rs13361468	5	89754788	C	T	T	0.005037783	UTR3		MBLAC2					g.89754788C>T
chr5_90459036	5	89754853	T	A	A	0.001259446	UTR3		MBLAC2					g.89754853T>A
chr5_90459335	5	89755152	A	C	C	0.001262626	UTR3		MBLAC2					g.89755152A>C
rs114494714	5	89755667	G	A	A	0.007556675	UTR3		MBLAC2					g.89755667G>A
rs115475015	5	89755738	T	C	C	0.001262626	UTR3		MBLAC2					g.89755738T>C
rs33963892	5	89755758	G	A	A	0.005050505	UTR3		MBLAC2					g.89755758G>A
chr5_90460048	5	89755865	A	T	T	0.002525253	UTR3		MBLAC2					g.89755865A>T
rs572378077	5	89755997	T	C	C	0.001262626	UTR3		MBLAC2					g.89755997T>C
rs13355867	5	89756102	G	A	A	0.005050505	UTR3		MBLAC2					g.89756102G>A
chr5_90460357	5	89756174	C	T	T	0.001262626	UTR3		MBLAC2					g.89756174C>T
rs186909308	5	89756308	C	A	A	0.001259446	UTR3		MBLAC2					g.89756308C>A
rs573496938	5	89756350	A	C	C	0.001265823	UTR3		MBLAC2					g.89756350A>C

SNVs	Chr	Position	Ref	Alt	Major Allele	Minor Allele Frequency	Classification	Coding Classification	Gene(s)	Transcript	Exon	HGVS Coding	HGVS Protein	HGVS Description
chr5_90460580	5	89756397	T	A	A	0.001259446	UTR3		MBLAC2					g.89756397T>A
rs35408393	5	89757044	G	A	A	0.01010101	Coding	Synonymous	MBLAC2	NM_203406.1	2	c.780C>T	p.=	g.89757044G>A
rs577351111	5	89769717	C	G	G	0.001259446	Coding	Synonymous	MBLAC2	NM_203406.1	1	c.393G>C	p.=	g.89769717C>G
rs376146450	5	89769720	A	G	G	0.001259446	Coding	Synonymous	MBLAC2	NM_203406.1	1	c.390T>C	p.=	g.89769720A>G
rs9293544	5	89769834	A	G	G	0.005037783	Coding	Synonymous	MBLAC2	NM_203406.1	1	c.276T>C	p.=	g.89769834A>G
chr5_90474044	5	89769861	A	G	G	0.001259446	Coding	Synonymous	MBLAC2	NM_203406.1	1	c.249T>C	p.=	g.89769861A>G
rs142019986	5	89769974	A	T	T	0.001259446	Coding	Nonsyn SNV	MBLAC2	NM_203406.1	1	c.136T>A	p.Ser46Thr	g.89769974A>T
rs2115501	5	89769999	A	G	G	0.107323232	Coding	Synonymous	MBLAC2	NM_203406.1	1	c.111T>C	p.=	g.89769999A>G
chr5_90474281	5	89770098	C	G	G	0.001259446	Coding	Synonymous	MBLAC2	NM_203406.1	1	c.12G>C	p.=	g.89770098C>G
rs73771266	5	89770122	C	T	T	0.003778338	UTR5		MBLAC2, POLR3G					g.89770122C>T
chr5_90474307	5	89770124	A	C	C	0.001259446	UTR5		MBLAC2, POLR3G					g.89770124A>C
chr5_90474387	5	89770204	G	C	C	0.002518892	UTR5		MBLAC2					g.89770204G>C
rs537775001	5	89770205	G	T	T	0.001259446	UTR5		MBLAC2					g.89770205G>T
rs529623416	5	89770306	T	C	C	0.001259446	UTR5		MBLAC2					g.89770306T>C
rs145567337	5	89770481	A	G	G	0.001259446	UTR5		MBLAC2					g.89770481A>G
rs114749727	5	89770558	A	C	C	0.001259446	UTR5		MBLAC2					g.89770558A>C
rs2303801	5	89770768	A	G	G	0.313602015	UTR5		POLR3G					g.89770768A>G
chr5_90485670	5	89781487	A	C	C	0.001259446	Coding	Nonsyn SNV	POLR3G	XM_005248405.1	2	c.103A>C	p.Thr35Pro	g.89781487A>C
chr5_90488129	5	89783946	T	G	G	0.001259446	Coding	Nonsyn SNV	POLR3G	XM_005248405.1	3	c.247T>G	p.Tyr83Asp	g.89783946T>G
chr5_90502231	5	89798048	A	C	C	0.001262626	UTR3		POLR3G					g.89798048A>C
rs10942596	5	89798109	G	A	A	0.460759494	UTR3		POLR3G					g.89798109G>A
chr5_90512199	5	89808016	A	G	G	0.001259446	UTR3		POLR3G					g.89808016A>G
rs562163116	5	89808335	G	A	A	0.001259446	UTR3		POLR3G					g.89808335G>A
rs7724507	5	89808531	G	C	C	0.001259446	UTR3		POLR3G					g.89808531G>C
rs201028228	5	89808878	G	A	A	0.016372796	UTR3		POLR3G					g.89808878G>A
rs72781088	5	89808979	C	T	T	0.068181818	UTR3		POLR3G					g.89808979C>T
rs367938529	5	89809231	G	A	A	0.001259446	UTR3		POLR3G					g.89809231G>A
rs570484746	5	89809360	T	C	C	0.001259446	UTR3		POLR3G					g.89809360T>C
rs146704994	5	89809616	T	C	C	0.001259446	UTR3		POLR3G					g.89809616T>C
rs75418010	5	89809679	T	C	C	0.069269521	UTR3		POLR3G					g.89809679T>C
rs11743044	5	89809737	C	A	A	0.003778338	UTR3		POLR3G					g.89809737C>A
rs37480	5	89809782	G	A	A	0.146464646	UTR3		POLR3G					g.89809782G>A
rs78679223	5	89809841	G	T	T	0.002518892	UTR3		POLR3G					g.89809841G>T
rs1047787	5	89810077	A	G	G	0.372795969	UTR3		POLR3G					g.89810077A>G
rs62375060	5	89810208	T	C	C	0.099496222	UTR3		POLR3G					g.89810208T>C
rs187983689	5	89810220	A	G	G	0.001259446	UTR3		POLR3G					g.89810220A>G
rs13169686	5	89810287	G	T	T	0.102015113	UTR3		POLR3G					g.89810287G>T
rs76367689	5	89811745	C	A	A	0.002518892	UTR3		LYSMD3					g.89811745C>A
chr5_90515978	5	89811795	T	C	C	0.001259446	UTR3		LYSMD3					g.89811795T>C
rs6862171	5	89811823	T	C	C	0.002518892	UTR3		LYSMD3					g.89811823T>C
chr5_90516071	5	89811888	G	A	A	0.001259446	UTR3		LYSMD3					g.89811888G>A
chr5_90516267	5	89812084	C	G	G	0.001259446	UTR3		LYSMD3					g.89812084C>G
rs74545801	5	89812223	A	G	G	0.003778338	UTR3		LYSMD3					g.89812223A>G
chr5_90516609	5	89812426	C	T	T	0.001262626	UTR3		LYSMD3					g.89812426C>T

SNVs	Chr	Position	Ref	Alt	Major Allele	Minor Allele Frequency	Classification	Coding Classification	Gene(s)	Transcript	Exon	HGVS Coding	HGVS Protein	HGVS Description
rs78447998	5	89812486	T	A	A	0.001269036	UTR3		LYSMD3					g.89812486T>A
rs3087840	5	89812676	A	T	T	0.379441624	UTR3		LYSMD3					g.89812676A>T
rs529584936	5	89812709	G	A	A	0.003787879	UTR3		LYSMD3					g.89812709G>A
rs182294974	5	89813208	G	A	A	0.002518892	UTR3		LYSMD3					g.89813208G>A
rs78691109	5	89813370	T	A	A	0.003778338	UTR3		LYSMD3					g.89813370T>A
rs74926905	5	89813433	G	T	T	0.019035533	UTR3		LYSMD3					g.89813433G>T
rs564564428	5	89813458	G	A	A	0.001259446	UTR3		LYSMD3					g.89813458G>A
chr5_90517652	5	89813469	A	G	G	0.001265823	UTR3		LYSMD3					g.89813469A>G
rs571095224	5	89813480	G	A	A	0.001265823	UTR3		LYSMD3					g.89813480G>A
chr5_90517783	5	89813600	A	C	C	0.001262626	UTR3		LYSMD3					g.89813600A>C
chr5_90517935	5	89813752	G	A	A	0.001262626	UTR3		LYSMD3					g.89813752G>A
chr5_90517986	5	89813803	G	A	A	0.001259446	UTR3		LYSMD3					g.89813803G>A
rs10040584	5	89814000	G	T	T	0.001259446	UTR3		LYSMD3					g.89814000G>T
chr5_90518386	5	89814203	C	T	T	0.001259446	UTR3		LYSMD3					g.89814203C>T
rs147031026	5	89814271	T	C	C	0.002518892	UTR3		LYSMD3					g.89814271T>C
chr5_90518503	5	89814320	A	T	T	0.002518892	UTR3		LYSMD3					g.89814320A>T
rs192126274	5	89814788	T	A	A	0.001262626	Coding	Nonsyn SNV	LYSMD3	XM_005248421.1	3	c.769A>T	p.Thr257Ser	g.89814788T>A
rs200384766	5	89814965	T	G	G	0.001259446	Coding	Nonsyn SNV	LYSMD3	XM_005248421.1	3	c.592A>C	p.Thr198Pro	g.89814965T>G
rs78764058	5	89814977	A	T	T	0.002525253	Coding	Nonsyn SNV	LYSMD3	XM_005248421.1	3	c.580T>A	p.Leu194Met	g.89814977A>T
rs62375061	5	89815108	T	G	G	0.017632242	Coding	Nonsyn SNV	LYSMD3	XM_005248421.1	3	c.449A>C	p.Tyr150Ser	g.89815108T>G
rs563272792	5	89820870	T	G	G	0.001259446	Coding	Synonymous	LYSMD3	XM_005248421.1	2	c.237A>C	p.=	g.89820870T>G
rs10069050	5	89820984	T	C	C	0.473551637	Coding	Synonymous	LYSMD3	XM_005248421.1	2	c.123A>G	p.=	g.89820984T>C
chr5_90529116	5	89824933	T	G	G	0.002518892	UTR5		LYSMD3					g.89824933T>G
rs37238	5	89824976	C	T	T	0.473551637	UTR5		LYSMD3					g.89824976C>T
rs603015	5	89825035	A	G	G	0.380352645	UTR5		LYSMD3					g.89825035A>G
chr5_90529500	5	89825317	T	G	G	0.001259446	UTR5		LYSMD3					g.89825317T>G
rs538359608	5	89825350	A	C	C	0.005037783	UTR5		LYSMD3					g.89825350A>C
rs150816712	5	89854646	T	A	A	0.003778338	UTR5		GPR98					g.89854646T>A
rs116110048	5	89854666	C	G	G	0.002518892	UTR5		GPR98					g.89854666C>G
rs61753944	5	89913740	T	C	C	0.001259446	Coding	Synonymous	GPR98	NM_032119.3	3	c.327T>C	p.=	g.89913740T>C
rs41311333	5	89914925	G	T	T	0.031486146	Coding	Nonsyn SNV	GPR98	NM_032119.3	4	c.380G>T	p.Arg127Leu	g.89914925G>T
rs61745496	5	89920984	T	G	G	0.001259446	Coding	Nonsyn SNV	GPR98	NM_032119.3	6	c.596T>G	p.Ile199Ser	g.89920984T>G
rs41303344	5	89923101	A	G	G	0.010075567	Coding	Nonsyn SNV	GPR98	NM_032119.3	7	c.746A>G	p.Lys249Arg	g.89923101A>G
rs200197273	5	89923208	G	C	C	0.001259446	Coding	Nonsyn SNV	GPR98	NM_032119.3	7	c.853G>C	p.Gly285Arg	g.89923208G>C
rs201236317	5	89923388	A	C	C	0.001259446	Coding	Nonsyn SNV	GPR98	NM_032119.3	7	c.1033A>C	p.Lys345Gln	g.89923388A>C
rs115239207	5	89923411	A	G	G	0.002518892	Coding	Synonymous	GPR98	NM_032119.3	7	c.1056A>G	p.=	g.89923411A>G
rs186639101	5	89923441	G	A	A	0.001259446	Coding	Synonymous	GPR98	NM_032119.3	7	c.1086G>A	p.=	g.89923441G>A
rs61744480	5	89925039	C	A	A	0.001259446	Coding	Nonsyn SNV	GPR98	NM_032119.3	9	c.1522C>A	p.Leu508Ile	g.89925039C>A
chr5_90629226	5	89925043	G	A	A	0.001259446	Coding	Nonsyn SNV	GPR98	NM_032119.3	9	c.1526G>A	p.Arg509Gln	g.89925043G>A
rs6889939	5	89925169	C	T	T	0.003778338	Coding	Nonsyn SNV	GPR98	NM_032119.3	9	c.1652C>T	p.Ala551Val	g.89925169C>T
rs184127858	5	89925293	A	C	C	0.003778338	Coding	Synonymous	GPR98	NM_032119.3	9	c.1776A>C	p.=	g.89925293A>C
chr5_90635280	5	89931097	T	C	C	0.001259446	Coding	Nonsyn SNV	GPR98	NM_032119.3	10	c.2006T>C	p.Val669Ala	g.89931097T>C
rs200187681	5	89933548	T	A	A	0.001262626	Coding	Nonsyn SNV	GPR98	NM_032119.3	11	c.2023T>A	p.Phe675Ile	g.89933548T>A
rs201007778	5	89939702	T	C	C	0.001259446	Coding	Nonsyn SNV	GPR98	NM_032119.3	14	c.2636T>C	p.Met879Thr	g.89939702T>C

SNVs	Chr	Position	Ref	Alt	Major Allele	Minor Allele Frequency	Classification	Coding Classification	Gene(s)	Transcript	Exon	HGVS Coding	HGVS Protein	HGVS Description
chr5_90643905	5	89939722	A	C	C	0.001259446	Coding	Nonsyn SNV	GPR98	NM_032119.3	14	c.2656A>C	p.Thr886Pro	g.89939722A>C
rs368163419	5	89941860	G	A	A	0.002518892	Coding	Nonsyn SNV	GPR98	NM_032119.3	16	c.2974G>A	p.Ala992Thr	g.89941860G>A
rs950692	5	89943433	G	A	A	0.137279597	Coding	Synonymous	GPR98	NM_032119.3	17	c.3141G>A	p.=	g.89943433G>A
rs183447491	5	89943547	C	T	T	0.001259446	Coding	Synonymous	GPR98	NM_032119.3	17	c.3255C>T	p.=	g.89943547C>T
rs2366777	5	89943571	G	T	T	0.314861461	Coding	Nonsyn SNV	GPR98	NM_032119.3	17	c.3279G>T	p.Leu1093Phe	g.89943571G>T
rs148097083	5	89943581	A	G	G	0.005037783	Coding	Nonsyn SNV	GPR98	NM_032119.3	17	c.3289A>G	p.Ser1097Gly	g.89943581A>G
rs80069610	5	89947434	G	A	A	0.001259446	Coding	Synonymous	GPR98	NM_032119.3	18	c.3303G>A	p.=	g.89947434G>A
rs200945405	5	89948189	A	G	G	0.002518892	Coding	Nonsyn SNV	GPR98	NM_032119.3	19	c.3443A>G	p.Asp1148Gly	g.89948189A>G
rs530678993	5	89949276	G	T	T	0.001262626	Coding	Synonymous	GPR98	NM_032119.3	20	c.3885G>T	p.=	g.89949276G>T
rs372789540	5	89949346	T	C	C	0.001262626	Coding	Nonsyn SNV	GPR98	NM_032119.3	20	c.3955T>C	p.Trp1319Arg	g.89949346T>C
chr5_90653570	5	89949387	G	A	A	0.001262626	Coding	Synonymous	GPR98	NM_032119.3	20	c.3996G>A	p.=	g.89949387G>A
rs41305898	5	89949605	T	C	C	0.005050505	Coding	Nonsyn SNV	GPR98	NM_032119.3	20	c.4214T>C	p.Phe1405Ser	g.89949605T>C
rs61740119	5	89949651	G	A	A	0.003787879	Coding	Synonymous	GPR98	NM_032119.3	20	c.4260G>A	p.=	g.89949651G>A
rs17543819	5	89953849	T	C	C	0.074307305	Coding	Synonymous	GPR98	NM_032119.3	21	c.4506T>C	p.=	g.89953849T>C
rs200955930	5	89954009	A	G	G	0.001259446	Coding	Nonsyn SNV	GPR98	NM_032119.3	21	c.4666A>G	p.Lys1556Glu	g.89954009A>G
chr5_90672671	5	89968488	T	C	C	0.001259446	Coding	Synonymous	GPR98	NM_032119.3	22	c.4878T>C	p.=	g.89968488T>C
rs72782753	5	89969880	G	A	A	0.006297229	Coding	Nonsyn SNV	GPR98	NM_032119.3	23	c.4939G>A	p.Val1647Ile	g.89969880G>A
rs371831553	5	89971170	C	T	T	0.003778338	Coding	Synonymous	GPR98	NM_032119.3	24	c.5221C>T	p.=	g.89971170C>T
rs565188390	5	89971179	G	A	A	0.001259446	Coding	Nonsyn SNV	GPR98	NM_032119.3	24	c.5230G>A	p.Val1744Ile	g.89971179G>A
rs41303346	5	89971253	A	G	G	0.016372796	Coding	Synonymous	GPR98	NM_032119.3	24	c.5304A>G	p.=	g.89971253A>G
rs17544552	5	89979518	T	C	C	0.071969697	Coding	Nonsyn SNV	GPR98	NM_032119.3	28	c.5780T>C	p.Met1927Thr	g.89979518T>C
rs41302834	5	89979568	A	G	G	0.003787879	Coding	Nonsyn SNV	GPR98	NM_032119.3	28	c.5830A>G	p.Asn1944Asp	g.89979568A>G
rs4916684	5	89979589	G	A	A	0.319444444	Coding	Nonsyn SNV	GPR98	NM_032119.3	28	c.5851G>A	p.Val1951Ile	g.89979589G>A
chr5_90683793	5	89979610	G	T	T	0.001262626	Coding	Nonsyn SNV	GPR98	NM_032119.3	28	c.5872G>T	p.Ala1958Ser	g.89979610G>T
rs41303352	5	89979691	G	A	A	0.143939394	Coding	Nonsyn SNV	GPR98	NM_032119.3	28	c.5953G>A	p.Asp1985Asn	g.89979691G>A
rs4916685	5	89979698	T	C	C	0.339240506	Coding	Nonsyn SNV	GPR98	NM_032119.3	28	c.5960T>C	p.Leu1987Pro	g.89979698T>C
rs16868972	5	89979750	T	G	G	0.162878788	Coding	Nonsyn SNV	GPR98	NM_032119.3	28	c.6012T>G	p.Phe2004Leu	g.89979750T>G
chr5_90683965	5	89979782	T	A	A	0.002518892	Coding	Nonsyn SNV	GPR98	NM_032119.3	28	c.6044T>A	p.Phe2015Tyr	g.89979782T>A
rs41308846	5	89979871	A	G	G	0.005050505	Coding	Nonsyn SNV	GPR98	NM_032119.3	28	c.6133A>G	p.Arg2045Gly	g.89979871A>G
chr5_90684083	5	89979900	C	T	T	0.001262626	Coding	Synonymous	GPR98	NM_032119.3	28	c.6162C>T	p.=	g.89979900C>T
rs16868974	5	89981611	T	C	C	0.018891688	Coding	Nonsyn SNV	GPR98	NM_032119.3	29	c.6289T>C	p.Cys2097Arg	g.89981611T>C
rs186999408	5	89981639	T	C	C	0.001259446	Coding	Nonsyn SNV	GPR98	NM_032119.3	29	c.6317T>C	p.Val2106Ala	g.89981639T>C
rs200055351	5	89985795	C	T	T	0.002518892	Coding	Nonsyn SNV	GPR98	NM_032119.3	30	c.6608C>T	p.Ala2203Val	g.89985795C>T
rs10037067	5	89985882	G	A	A	0.341772152	Coding	Nonsyn SNV	GPR98	NM_032119.3	30	c.6695G>A	p.Cys2232Tyr	g.89985882G>A
chr5_90690075	5	89985892	G	T	T	0.001262626	Coding	Nonsyn SNV	GPR98	NM_032119.3	30	c.6705G>T	p.Leu2235Phe	g.89985892G>T
rs199571511	5	89986845	T	C	C	0.001259446	Coding	Nonsyn SNV	GPR98	NM_032119.3	31	c.6938T>C	p.Leu2313Pro	g.89986845T>C
rs2366926	5	89988504	G	A	A	0.337531486	Coding	Nonsyn SNV	GPR98	NM_032119.3	32	c.7034G>A	p.Ser2345Asn	g.89988504G>A
rs111033452	5	89989749	T	C	C	0.003778338	Coding	Synonymous	GPR98	NM_032119.3	33	c.7176T>C	p.=	g.89989749T>C
rs111033429	5	89989752	T	C	C	0.010075567	Coding	Synonymous	GPR98	NM_032119.3	33	c.7179T>C	p.=	g.89989752T>C
rs16876822	5	89989779	A	G	G	0.337121212	Coding	Synonymous	GPR98	NM_032119.3	33	c.7206A>G	p.=	g.89989779A>G
rs111033430	5	89989802	G	A	A	0.001262626	Coding	Nonsyn SNV	GPR98	NM_032119.3	33	c.7229G>A	p.Cys2410Tyr	g.89989802G>A
rs201733037	5	89990155	T	C	C	0.005050505	Coding	Nonsyn SNV	GPR98	NM_032119.3	33	c.7582T>C	p.Ser2528Pro	g.89990155T>C
chr5_90694429	5	89990246	T	C	C	0.001262626	Coding	Nonsyn SNV	GPR98	NM_032119.3	33	c.7673T>C	p.Leu2558Pro	g.89990246T>C
rs200241260	5	89990379	G	C	C	0.001262626	Coding	Synonymous	GPR98	NM_032119.3	33	c.7806G>C	p.=	g.89990379G>C

SNVs	Chr	Position	Ref	Alt	Major Allele	Minor Allele Frequency	Classification	Coding Classification	Gene(s)	Transcript	Exon	HGVS Coding	HGVS Protein	HGVS Description
rs79915053	5	89990386	G	T	T	0.001262626	Coding	Nonsyn SNV	GPR98	NM_032119.3	33	c.7813G>T	p.Val2605Leu	g.89990386G>T
rs201214794	5	89990447	A	G	G	0.003787879	Coding	Nonsyn SNV	GPR98	NM_032119.3	33	c.7874A>G	p.His2625Arg	g.89990447A>G
rs376318779	5	89992918	T	A	A	0.001262626	Coding	Nonsyn SNV	GPR98	NM_032119.3	34	c.8110T>A	p.Phe2704Ile	g.89992918T>A
rs201963060	5	89999487	G	A	A	0.002518892	Coding	Nonsyn SNV	GPR98	NM_032119.3	35	c.8161G>A	p.Val2721Ile	g.89999487G>A
rs16869016	5	90000210	T	C	C	0.059193955	Coding	Nonsyn SNV	GPR98	NM_032119.3	36	c.8291T>C	p.Leu2764Ser	g.90000210T>C
rs111033530	5	90001237	A	G	G	0.01511335	Coding	Nonsyn SNV	GPR98	NM_032119.3	37	c.8407A>G	p.Thr2803Ala	g.90001237A>G
rs6880570	5	90001368	G	T	T	0.147355164	Coding	Synonymous	GPR98	NM_032119.3	37	c.8538G>T	p.=	g.90001368G>T
rs201586455	5	90002172	C	A	A	0.002525253	Coding	Nonsyn SNV	GPR98	NM_032119.3	38	c.8691C>A	p.Asp2897Glu	g.90002172C>A
rs56329646	5	90012312	T	C	C	0.006297229	Coding	Synonymous	GPR98	NM_032119.3	43	c.9213T>C	p.=	g.90012312T>C
rs13157270	5	90012379	A	G	G	0.103274559	Coding	Nonsyn SNV	GPR98	NM_032119.3	43	c.9280A>G	p.Ile3094Val	g.90012379A>G
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chr5_90802803	5	90098620	A	G	G	0.001259446	Coding	Nonsyn SNV	GPR98	NM_032119.3	71	c.14582A>G	p.His4861Arg	g.90098620A>G
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chr5_91369355	5	90665172	T	G	G	0.001259446	UTR3		ARRDC3					g.90665172T>G
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rs539115449	5	90665460	G	A	A	0.001259446	UTR3		ARRDC3					g.90665460G>A

SNVs	Chr	Position	Ref	Alt	Major Allele	Minor Allele Frequency	Classification	Coding Classification	Gene(s)	Transcript	Exon	HGVS Coding	HGVS Protein	HGVS Description
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rs192055939	5	90665725	C	A	A	0.001259446	UTR3		ARRDC3					g.90665725C>A
rs74345264	5	90665777	C	T	T	0.045454545	UTR3		ARRDC3					g.90665777C>T
rs114355367	5	90665843	A	G	G	0.001262626	UTR3		ARRDC3					g.90665843A>G
rs567558348	5	90666184	T	C	C	0.001259446	UTR3		ARRDC3					g.90666184T>C
chr5_91370395	5	90666212	C	T	T	0.001259446	UTR3		ARRDC3					g.90666212C>T
rs575963529	5	90666494	C	T	T	0.001265823	UTR3		ARRDC3					g.90666494C>T
chr5_91371295	5	90667112	C	G	G	0.001262626	UTR3		ARRDC3					g.90667112C>G
chr5_91373780	5	90669597	C	T	T	0.001259446	Coding	Synonymous	ARRDC3	NM_020801.2	7	c.1092G>A	p.=	g.90669597C>T
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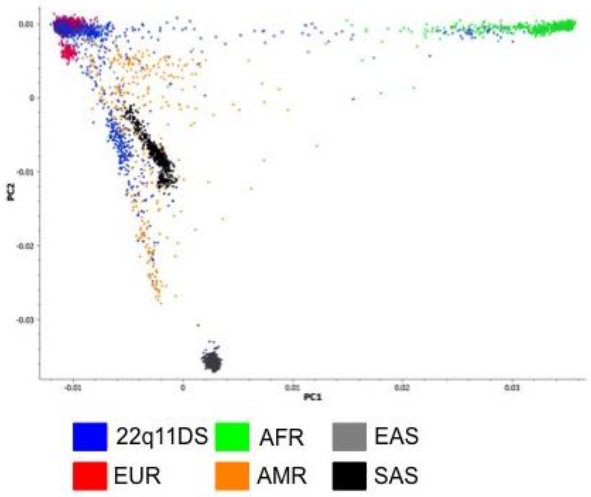
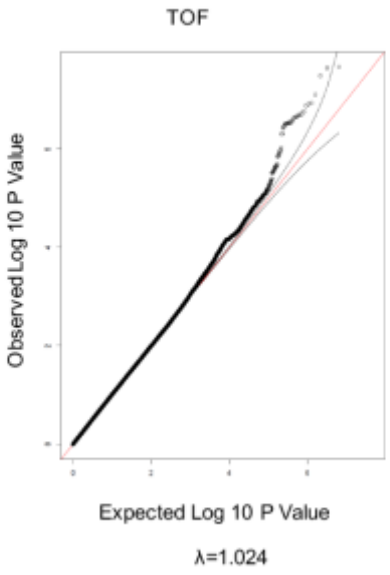
rs201823359	5	90678900	G	C	C	0.001259446	Coding	Nonsyn SNV	ARRDC3	NM_020801.2	1	c.10C>G	p.Arg4Gly	g.90678900G>C

Supplementary Table 7. Coding variants identified in the genes around 5q14.3 locus among 397 subjects with 22q11.2DS. Position is according to NCBI36/hg18 (March 2006) assembly, which was converted by CrossMap from GRCh38/hg38 to GRCh37/hg19.

Supplementary Figures



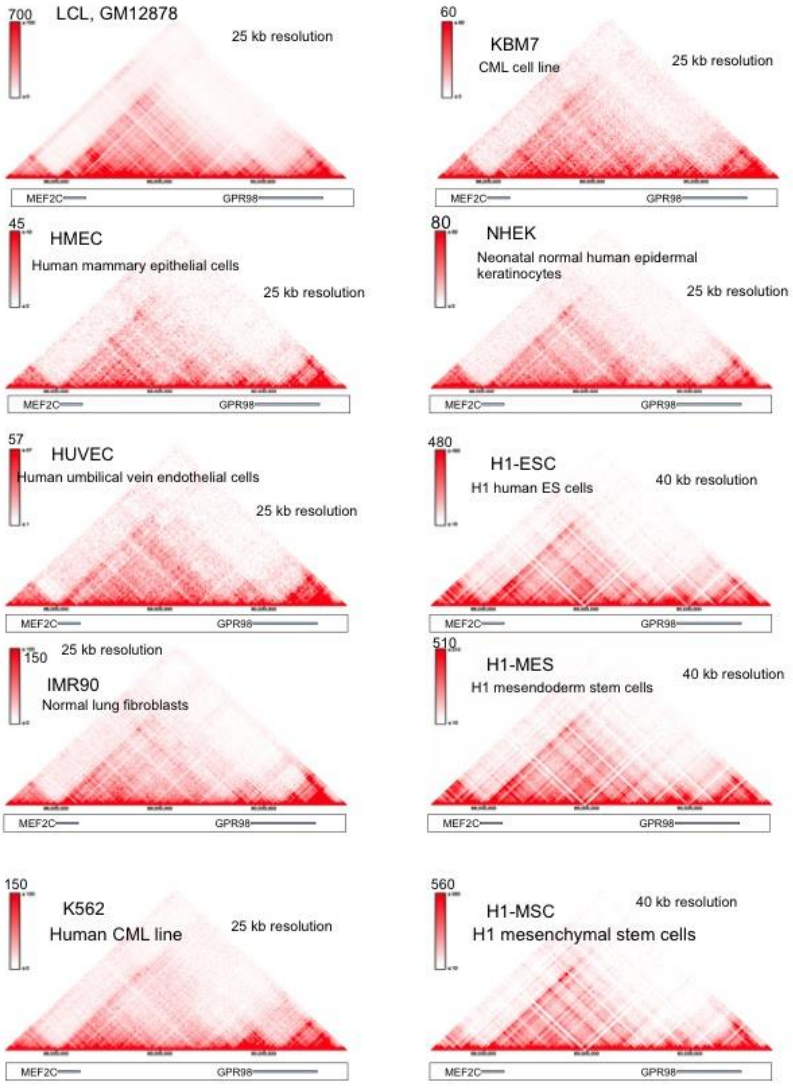
Supplementary Figure 1: Ethnicity of the 22q11.2DS cohort. The first two principal components (PCs) were calculated from linkage disequilibrium (LD) pruned common genotyped variants from 1,480 study subjects (blue dots) and 1000 Genomes Project population dataset to check the ethnicity of the cohort. The majority of the cohort clustered with the European (EUR, red) population, but some had African (AFR, green) or Asian ancestry (EAS, dark grey). The Chilean samples clustered with admixed American samples (AMR).

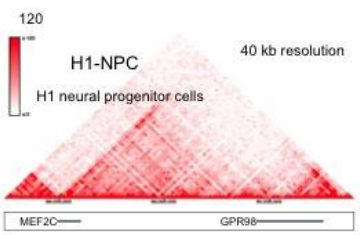


Supplementary Figure 2. Quantile-quantile (Q-Q) plots of GWAS of TOF. The plot is shown $\log_{10}$ transformed expected p-values plotted against $\log_{10}$ transformed observed p-values. Genomic inflation factor (lambda, λ) is defined as the median of the resulting chi-squared test statistics divided by the expected median of the chi-squared distribution. There was no obvious evidence that the observed test statistics deviated from the expected ($\lambda < 1.2$) with the following λ-value: TOF = 1.024, indicating that population stratification had negligible effects on the genetic analysis of these samples.

Supplementary Figure 3. LD matrix across of TOF GWAS region in 22q11.2DS (top) and 1000 Genome Project Caucasian cohorts (bottom). LD matrix was generated using Haploview, LD plot based on r^2. Key: $r^2 = 0$ is given in white, $0 < r^2 < 1$ is given in shades of grey and r2 = 1 is given in black. The pair-wise D' values are given in the boxes. Only the SNPs with MAF more than 0.05 were shown. Cohorts are indicated.

Hi-C data from http://www.3dgenome.org
Hg19; chr5:87500000-90800000

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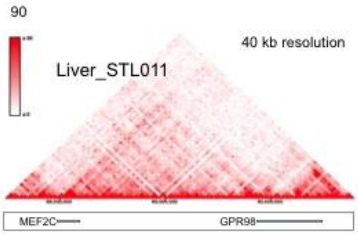
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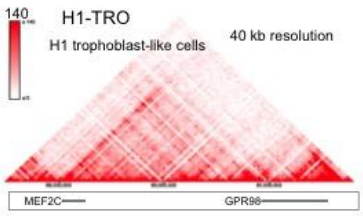
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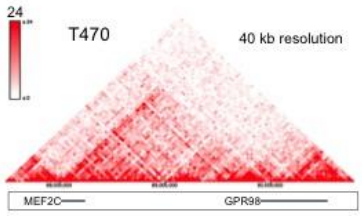
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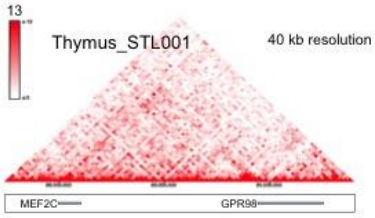
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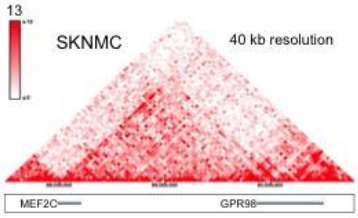
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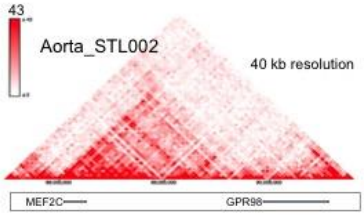
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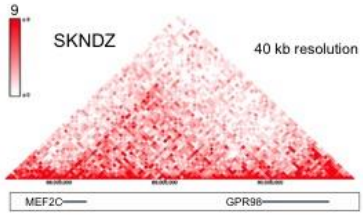
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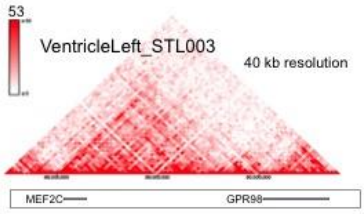
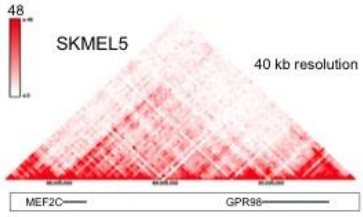
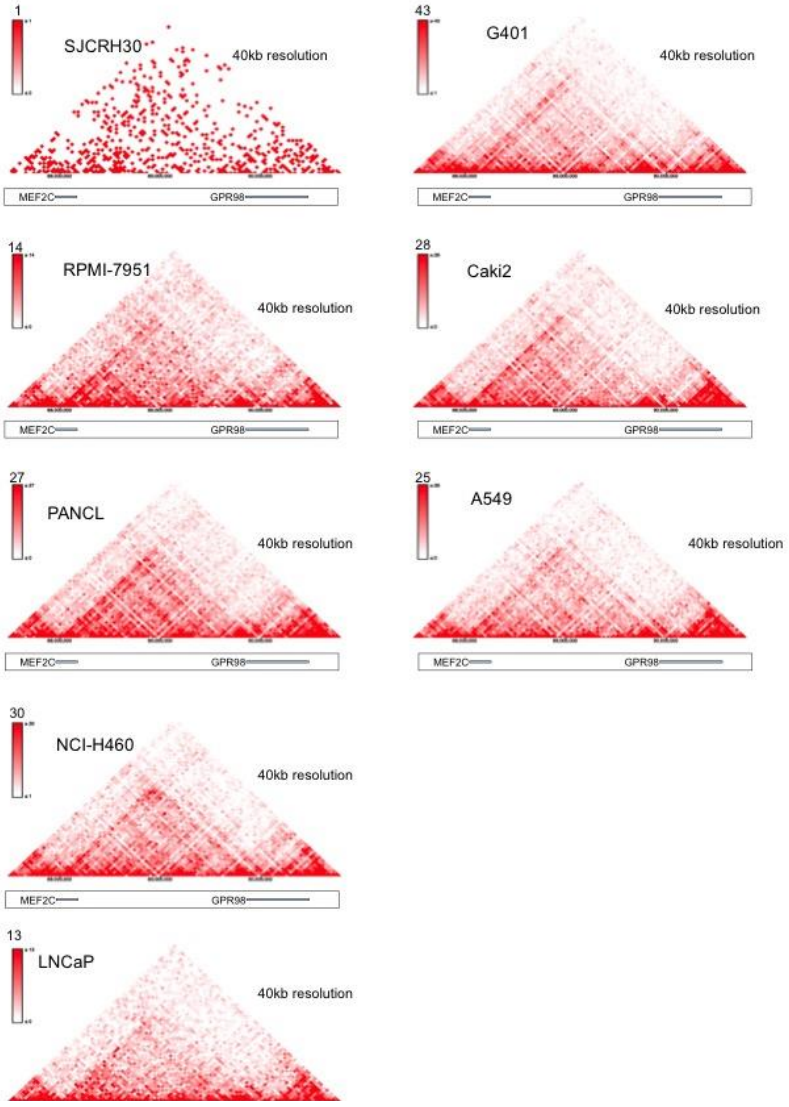
MEF2C
GPR98

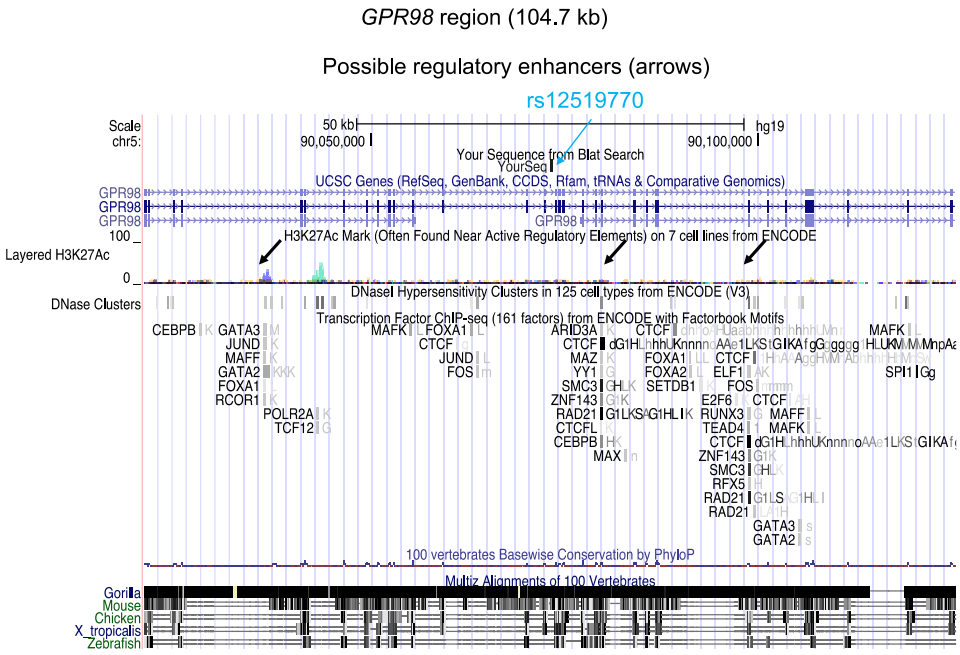

53
VentricleLeft_STL003
40 kb resolution
MEF2C
GPR98


48
SKMEL5
40 kb resolution
MEF2C
GPR98



Supplementary Figure 4. Hi-C data for the 5q14.3 locus. Chromatin conformation heat maps (Figure 3; chr5:87500000-90800000; hg19) in cell lines is shown (http://www.3dgenome.org).[1-3] The map resolution is indicated and it is either 25 kb or 40 kb. Contact matrices, representing the normalized number of contacts between a pair of loci are visualized as pixels. The pixels are colored with the minimum intensity being white and the maximum being red as indicated in the bar plot on the left. The maximum intensity is shown at the top of the bar. The *MEF2C* and

GPR98 gene loci are shown in horizontal boxes, with respect to the genomic interval in the heat map. Cell lines are described (http://www.3dgenome.org).[1-3]:



Supplementary Figure 5. Possible enhancers in the 104.7 kb GPR98 region. Snapshot from the UCSC Genome Browser (human; hg19) showing the 104.7 kb interval in the GPR98 locus with association to TOF. There are at least three different predicted enhancers (arrows) in the H3K27Ac mark track as indicated below for transcription factor binding sites identified by chromatin immunoprecipitation-sequence (ChIP-seq) that is obtained from the ENCODE project.

The evolutionary conservation track is shown below, where the exons of GPR98 show the highest conservation.

1. Dixon JR, Selvaraj S, Yue F, Kim A, Li Y, Shen Y, Hu M, Liu JS and Ren B. Topological domains in mammalian genomes identified by analysis of chromatin interactions. *Nature*. 2012;485:376-80.

2. Rao SS, Huntley MH, Durand NC, Stamenova EK, Bochkov ID, Robinson JT, Sanborn AL, Machol I, Omer AD, Lander ES and Aiden EL. A 3D map of the human genome at kilobase resolution reveals principles of chromatin looping. *Cell*. 2014;159:1665-80.

3. Dixon JR, Jung I, Selvaraj S, Shen Y, Antosiewicz-Bourget JE, Lee AY, Ye Z, Kim A, Rajagopal N, Xie W, Diao Y, Liang J, Zhao H, Lobanenkov VV, Ecker JR, Thomson JA and Ren B. Chromatin architecture reorganization during stem cell differentiation. *Nature*. 2015;518:331-6.