

SUPPLEMENTAL MATERIAL

Online Methods:

Cohorts and samples studied

The Family Blood Pressure Program (FBPP), funded by the National Heart, Lung, and Blood Institute (NHBLI), is a large family-based sample of 17,129 subjects; each ascertained through a proband with clinical HTN or elevated BP. The aims were to identify genes influencing BP levels, HTN and its cardiovascular complications in Americans of European, African, Hispanic and Asian ancestry. The families were ascertained through four networks, GenNet, GENOA, HyperGEN and SAPHIRE, and included European American (EA), African American (AA), Hispanic (HA) and Asian American (AsA) subjects. The FBPP study includes data on over 200 BP and HTN relevant variables by collecting data on 17,129 individuals through 21,600 physical examinations and blood samples. SBP and DBP were measured using an automated oscillometric BP measurement device with a consistent protocol across networks while PP and MAP were calculated as $PP=SBP-DBP$ and $MAP=(2DBP+SBP)/3$. Samples with missing SBP, DBP, BMI, gender or age were not included in this analysis.

The CLUE study includes 2 large cohorts of volunteers from Washington County in Maryland, who were ascertained and sampled in 1974 and 1989 by the George W. Comstock Center for Public Health Research and Prevention in Hagerstown, Maryland, and was funded by the National Cancer Institute. CLUE I (Campaign Against Cancer and Stroke, 1974) included 26,147 participants (23,951 of whom were local residents). Fifteen mL of blood was drawn from each individual along with information on health history and BP. CLUE II (Campaign Against Cancer and Heart Disease, 1989) was an expansion of CLUE I with some participant overlap. Twenty mL of blood was collected from 32,894 individuals (25,076 were local residents) with a brief health history and an extended questionnaire. About 30% of adult residents of Washington County, MD participated in the study. The Odyssey cohort consisted of 8,394 individuals who participated in both CLUE I and II. SBP and DBP were measured by mercury filled sphygmomanometers, with a subset of 384 Odyssey samples measured by aneroid sphygmomanometers; PP and MAP were calculated as in the FBPP study. Samples with missing SBP, DBP, BMI, gender or age were not included in the analysis.

ARIC (Atherosclerosis Risk In Communities) study is a population-based, prospective-cohort cardiovascular disease study of 15,792 persons aged 45-64 years at baseline (1987-89) randomly chosen from four US communities: Forsyth County, NC (12% African American (AA)); Jackson, MI (100% AA); suburban Minneapolis, MN (<1% AA); and Washington County, MD (<1% AA). Cohort members completed four clinic examinations in 1987-89, 1990-92, 1993-95, and 1996-98 with data on health status, selected risk factors, family medical history, employment, educational status, diet and physical activity; a blood sample of over 20 mL was also collected. Sitting SBP/DBP was measured with a random zero sphygmomanometer while PP and MAP were calculated as in the FBPP. Samples with missing SBP, DBP, BMI, gender or age were not included in the analysis.

DNA sequencing of PCR products using bidirectional dideoxy Sanger chemistry was supported by the RS&G (Resequencing and Genotyping Service) program

(<http://rsng.nhlbi.nih.gov/scripts/index.cfm>) of the NHLBI and was performed at the J. Craig Venter Institute, Rockville, MD (JCVI, who sequenced *HSD11B2*, *NR3C2*, *SCNN1A*, *SCNN1B*, *SCNN1G*, and *WNK1*) and at the University of Washington, Seattle, WA (UW, who sequenced *AGT*, *CYP11B1*, *CYP17A1*, *NR3C1* and *WNK4*).

DNA sequencing protocol:

Briefly, 5'-M13 tailed-gene specific PCR primers were designed to cover the target region with amplicon size ranging from 350-800 bp and with a minimum of 100 bp overlap between adjacent amplicons, resulting in double-stranded coverage of all targeted regions. Overlapping amplicons were used to validate gene-specific primer sequences in independent experiments and rule out the possibility of allele-specific PCR amplification. All primer sequences were compared to the whole genome assembly to verify uniqueness against pseudogenes and gene families. Designed amplicons were separated into two categories based on their calculated melting temperatures (T_m). Standard GC content and high GC content amplicons were processed separately (at JCVI) and following temperature gradient optimization of small-scale reactions to determine optimal thermal cycling conditions (at UW).

JCVI protocols: PCR amplifications were performed in 384-well plates in a volume of 10 μ L. The standard GC content PCR protocol was comprised of 3.0 μ L of 0.4 μ M mixed forward and reverse primers, 3.0 μ L of DNA (1.67ng/ μ L) and 0.05 μ L (0.25 units) of AmpliTaq Gold® DNA polymerase (Applied Biosystems). The high GC PCR protocol was comprised of 3.0 μ L of 1.2 μ M mixed forward and reverse primers, 3.0 μ L of DNA (10.0ng/ μ L) and 0.075 μ L (0.375 units) of AmpliTaq Gold® DNA polymerase (Applied Biosystems). All PCR amplifications were performed on dual 384-well GeneAmp® PCR System 9700 thermal cyclers (Applied Biosystems) under the following program: 96°C for 5 minutes (1X); 94°C for 30 seconds, 60°C for 45 seconds, 72°C for 45 seconds (40X); 72°C for 10 minutes (1X); 10°C final hold. PCR products were cleaned using Shrimp Alkaline Phosphatase and Exonuclease I (SAP/Exo mix) (USB Corporation) by adding 5.0 μ L of SAP/Exo mix directly to the 10 μ L volume PCR products in 384-well plates. For standard GC PCR products the SAP/Exo mix was comprised of 0.5 μ L of SAP (1 unit) and 0.1 μ L of exonuclease I (0.5 unit). For high GC PCR products the SAP/Exo mix was comprised of 0.5 μ L of SAP (1 unit) and 0.175 μ L of exonuclease I (1.76 units). Using dual 384-well GeneAmp® PCR System 9700 thermal cyclers (Applied Biosystems), digestions were carried out at two temperatures, 37°C followed by 47°C (standard reaction) or 50°C (high GC reaction), with a final heat inactivation step at 72°C.

Bi-directional sequencing reactions were performed in 384-well plates using a 1/64th BigDye™ Terminator v3.1 chemistry (Applied Biosystems) and 1.5 μ L of cleaned PCR products in dual 384-well GeneAmp® PCR System 9700 thermal cyclers (Applied Biosystems). Reaction products were precipitated using a sodium acetate/ethanol mixture followed by a final ethanol rinse and air-drying. Beckman-Coulter FX robots and Cartesian Technologies Pixsys 4200 nanoliter liquid handling systems were employed in setting up PCR, PCR clean-up, sequencing reactions and precipitation. Immediately before sequencing, the precipitated sequencing reactions were

resuspended in 10.0µL of 0.75mM EDTA using a µFill™ (Bio-Tek™ Instruments) and shaken using a Microplate Shaker (Union Scientific™ Corporation). Chromatograms were generated from sequence reaction on an Applied Biosystems ABI 3730XL capillary sequencer. Data flow was tracked by using a custom-designed LIMS system.

UW protocols: Production level PCR amplifications were performed in 96-well plates in a volume of 7 µl comprising 0.2 µl each of 7 µM forward and reverse primers, 2.8 µl DNA (5 ng/µl), and 0.4 µl Elongase Enzyme (Invitrogen) or iProof polymerase (Bio-Rad) per well. Following evaluation by 1% agarose gel electrophoresis, reactions were diluted four to six fold in ddH₂O. Dilution of the products eliminated the need for any purification of the PCR products prior to sequencing.

Sequencing reactions were performed in MJ Tetrad PTC 225 thermal cyclers in 384-well format by using 5% BDT v3.1 sequencing chemistry (ABI). Reaction products were precipitated in ethanol with CleanSeq magnetic beads (Agencourt). Perkin Elmer Minitrak, Multiprobe, and Evolution P3 robots were used to automate liquid handling in the setup of PCR, sequencing reactions and precipitation reactions. Reaction products were air dried and diluted to 30 µl with ddH₂O. Chromatograms were generated from sequence reaction on an Applied Biosystems ABI 3730XL capillary sequencer. Data flow was tracked by using a custom-designed LIMS system.

Variant discovery and analysis:

All chromatograms were initially base- and quality-called by using ABI KB (on 3730xl sequencer), filtered using DSPTrace software, base- and quality-called by TraceTuner with custom calibration for 3730xl at JCVI, while all chromatograms were based-called by using Phred. They were then assembled into contigs by using Phrap and scanned for SNP and INDELS with JCVI Resequencing Analysis software and by PolyPhred, version 6.02 [1] to identify polymorphic sites. Data quality was monitored and assessed at multiple production checkpoints using numerous methods, such as removing low-quality sequence (Phred score < 30), and low signal-to-noise ratio sequences (SNR < 20), resulting in reads averaging > 450 bp with an average Phred score of 40. Following all assembly of all chromatograms onto an initial reference sequence putative polymorphic sites were selectively reviewed by sequencing analysts using Consed [2]. Individual polymorphic sites in regions with lower quality data, ambiguous base calls, deviations from Hardy-Weinberg equilibrium or those identified using laboratory quality control tools were reviewed to eliminate potential false positive positions. Variations were formatted and submitted to dbSNP for assignment of ss and rs identification numbers.

Sequence quality

In order to assess sequence quality we computed sequencing accuracy using 4 duplicated samples and base calls in the overlapping amplicons. On comparing the DNA sequences of the 4 individuals who were sequenced in duplicate, 303 differences were detected from a total of 479,995bp (error rate of 0.063% per sample). On comparing the base calls in all overlapping reads, 16,021 differences were detected

from a total of 6,673,015bp (estimated error rate of 0.24% per sample). Thus, overall data quality is very high.

Genotyping methods

The Sequenom pool genotyping primers were tested for their genotyping accuracy on 179 HapMap and known variant allele carriers in the respective FBPP samples. For each reaction pool, we used ~10-20ng of genomic DNA per sample and followed the Sequenom standard protocol. We used a Tecan Genesis liquid handler and Tecan Gemini software (Tecan Trading AG, Switzerland) to make 384-well DNA template and dispense PCR master mix into each well. We genotyped rs2681472 (*ATP2B1*) using Taqman technology and a pre-designed SNP assay from Applied Biosystems Inc. (Foster City, CA). For this assay, we used ~5-10ng of genomic DNA per sample following a standard protocol for 384-well plate reactions.

Genotypes were called using the automated Sequenom and Taqman software (TYPER Suite 4.0.2.0 and SDS 2.2p1). We manually called genotypes whenever the software failed to process the intensity signals automatically. Based on replicate samples, and by comparing our genotypes for 179 HapMap samples with those in the HapMap Project, we obtained a concordance rate >99.9%. Samples with <90% completeness of genotypes and variants with <90% completeness of samples were excluded from further analysis.

Supplemental References for Methods:

- 1 Stephens M, Sloan JS, Robertson PD, Scheet P, Nickerson DA. Automating sequence-based detection and genotyping of SNPs from diploid samples. *Nat. Genet.* 2006;38:1457-1462.
- 2 Gordon D, Abagian C, Green P. Consed: a graphical tool for sequence finishing. *Genome Res.* 1998;8:195-202.

Online Table Legends:

Online Table I: Hypertension genes selected for resequencing. For each gene, we include the chromosomal position (hg19), the nature and inheritance of known disease mutations, and the associated hypertension/blood pressure trait phenotypes. (^aLOF/GOF: loss/gain of function; ^bAR/AD: autosomal recessive/dominant; ^cAH: Adrenal Hyperplasia, AME: Apparent Mineralocorticoid Excess; EH: Essential Hypertension, GR: Glucocorticoid Receptor deficiency, LS: Liddle Syndrome, PHA: Pseudo-hypoaldosteronism, EO: Early Onset hypertension with severe exacerbation in pregnancy; RTD: Renal Tubular Dysgenesis.)

Online Table II: List of all known disease causing coding mutations from the Human Gene Mutation Database (HGMD) at the 11 genes selected for resequencing.

Online Table III: Summary of demographic and phenotypic information for ARIC, CLUE and FBPP samples. For each study we provide sample size, % female, % hypertensive, % medicated, observed BP values and residuals after correction for covariates (see Methods) for individuals by European (EA) and African (AA) ancestry.

Online Table IV: Genetic association results of pooled non-coding variants by gene and population. The numbers of variants (k) are provided; P-values with $FDR \leq 0.05$ are highlighted in yellow.

Online Table V: Genetic association results of 6 imputed *CYP17A1* variants for SBP LTA in ARIC EAs; chromosomal position, rsID, alleles (A1, A2), MAF, effect size in mmHg, standard error (SE) and corresponding P-values are shown. All entries with $FDR \leq 0.05$ are highlighted in yellow.

Online Table VI: Complete genetic association results of 27 replicated GWAS variants in CLUE and FBPP samples; chromosomal position, rsID, minor allele, MAF and corresponding P-values are shown. All entries with $FDR \leq 0.05$ are highlighted in yellow.

Online Table VII: Complete genetic association results of 11 rare deleterious variants in CLUE and FBPP samples; chromosomal position, rsID, minor allele, MAF and corresponding P-values are shown. All entries with $FDR \leq 0.05$ are highlighted in yellow.

Online Table VIII: Power calculations. Statistical power is shown as a function of the variant allele frequency (MAF), the variants' effect on blood pressure in standard deviation units (σ) and sample size. All power values $< 10\%$ are designated by * and values above 80% are highlighted in yellow.

Online Table IX: List of 12 disease causing HGMD mutations in 69 of the total of 560 GenNet individuals. For each variant, we provide the corresponding chromosomal location, reference and alternate alleles (A1/A2) and residues (Res1/Res2), strand, rsID, minor allele frequency in EA and AA, phyloP score and disease association. Note that all variants are non-synonymous except one exonic splicing variant marked as *.

Online Table X: Allelic effects of variants in 5 mutation classes; for each class, the number of variants found in studied cohorts (# variants), number of variants in each class that are found in EVS (# in EVS), median absolute effect size of variant's residuals and standard error (average across all individuals with the rare alleles, τ and SE_{τ}), τ value and standard error in mmHg (τ_{mmHg} SE_{mmHg}), median minor allele frequency when found in EVS (q) and other wise (!) are provided.

Online Table I.

<i>Gene Name</i>	<i>Chromosomal Position (hg19)</i>	<i>Mutation type^a</i>	<i>Mode of Inheritance^b</i>	<i>Associated Disease^c</i>	<i>Trait</i>
<i>AGT</i>	chr1:230,836,298-230,851,926	LOF	AR	EH, RTD	Hypo + Hypertension
<i>CYP11B1</i>	chr8:143,951,810-143,961,548	LOF	AR	AH IV	Hypertension
<i>CYP17A1</i>	chr10:104,590,010-104,597,876	LOF	AR	AH V	Hypertension
<i>HSD11B2</i>	chr16:67,462,017-67,474,452	LOF	AR	AME	Hypertension
<i>NR3C1</i>	chr5:142,655,537-142,817,077	LOF	AD	GR	Hypertension
<i>NR3C2</i>	chr4:148,996,918-149,366,699	LOF/GOF	AD	PHA I/EO	Hypo/Hypertension
<i>SCNN1A</i>	chr12:6,453,015-6,487,715	LOF	AR	PHA I	Hypertension
<i>SCNN1B</i>	chr16:23,310,632-23,395,619	GOF/LOF	AD/AR	LS/PHA I	Hyper/Hypotension
<i>SCNN1G</i>	chr16:23,191,040-23,231,200	GOF/LOF	AD/AR	LS/PHA I	Hypertension
<i>WNK1</i>	chr12:859,732-1,020,958	GOF	AD	PHA II	Hypertension
<i>WNK4</i>	chr17:40,930,733-40,951,031	LOF	AD	PHA II	Hypertension

Online Table II.

<i>chr</i>	<i>bp</i>	<i>A1</i>	<i>A2</i>	<i>strand</i>	<i>dbSNP</i>	<i>HGVS_cdna</i>	<i>HGVS_protein</i>	<i>gene</i>	<i>disease</i>
1	230841679	C	T	-	NA	NM_000029.3:c.1124G-A	NP_000020.1:p.R375Q	<i>AGT</i>	Renal tubular dysgenesis
1	230845755	T	C	-	rs56073403	NM_000029.3:c.842A-G	NP_000020.1:p.Y281C	<i>AGT</i>	Hypertension
1	230845866	A	C	-	rs5041	NM_000029.3:c.731T-G	NP_000020.1:p.L244R	<i>AGT</i>	Hypertension
1	230845872	G	A	-	NA	NM_000029.3:c.725C-T	NP_000020.1:p.T242I	<i>AGT</i>	Hypertension
1	230845993	G	A	-	NA	NM_000029.3:c.604C-T	NP_000020.1:p.Q202X	<i>AGT</i>	Renal tubular dysgenesis
1	230846470	G	A	-	rs41271499	NM_000029.3:c.127C-T	NP_000020.1:p.L43F	<i>AGT</i>	Preeclampsia
10	104590499	C	T	-	NA	NM_000102.3:c.1487G-A	NP_000093.1:p.R496H	<i>CYP17A1</i>	17,20-lyase deficiency
10	104590500	G	A	-	NA	NM_000102.3:c.1486C-T	NP_000093.1:p.R496C	<i>CYP17A1</i>	Steroid-17 alpha-hydroxylase deficiency
10	104590592	A	G	-	NA	NM_000102.3:c.1394T-C	NP_000093.1:p.L465P	<i>CYP17A1</i>	Steroid-17 alpha-hydroxylase deficiency
10	104590605	G	A	-	NA	NM_000102.3:c.1381C-T	NP_000093.1:p.Q461X	<i>CYP17A1</i>	Steroid-17 alpha-hydroxylase deficiency
10	104590628	A	G	-	NA	NM_000102.3:c.1358T-C	NP_000093.1:p.F453S	<i>CYP17A1</i>	17-alpha-hydroxylase/17,20-lyase deficiency
10	104590641	G	A	-	NA	NM_000102.3:c.1345C-T	NP_000093.1:p.R449C	<i>CYP17A1</i>	17-alpha-hydroxylase/17,20-lyase deficiency
10	104590662	A	G	-	NA	NM_000102.3:c.1324T-C	NP_000093.1:p.C442R	<i>CYP17A1</i>	17-alpha-hydroxylase/17,20-lyase deficiency
10	104590667	C	T	-	NA	NM_000102.3:c.1319G-A	NP_000093.1:p.R440H	<i>CYP17A1</i>	Steroid-17 alpha-hydroxylase deficiency
10	104590668	G	A	-	NA	NM_000102.3:c.1318C-T	NP_000093.1:p.R440C	<i>CYP17A1</i>	Steroid-17 alpha-hydroxylase deficiency
10	104590685	G	A	-	NA	NM_000102.3:c.1301C-T	NP_000093.1:p.P434L	<i>CYP17A1</i>	17-alpha-hydroxylase/17,20-lyase deficiency
10	104590703	G	A	-	NA	NM_000102.3:c.1283C-T	NP_000093.1:p.P428L	<i>CYP17A1</i>	17-alpha-hydroxylase/17,20-lyase deficiency
10	104590736	A	C	-	NA	NM_000102.3:c.1250T-G	NP_000093.1:p.F417C	<i>CYP17A1</i>	17,20-lyase deficiency
10	104590739	C	T	-	NA	NM_000102.3:c.1247G-A	NP_000093.1:p.R416H	<i>CYP17A1</i>	Steroid-17 alpha-hydroxylase deficiency
10	104590740	G	A	-	NA	NM_000102.3:c.1246C-T	NP_000093.1:p.R416C	<i>CYP17A1</i>	Steroid-17 alpha-hydroxylase deficiency
10	104591282	G	C	-	NA	NM_000102.3:c.1226C-G	NP_000093.1:p.P409R	<i>CYP17A1</i>	Steroid-17 alpha-hydroxylase deficiency
10	104591292	A	G	-	NA	NM_000102.3:c.1216T-C	NP_000093.1:p.W406R	<i>CYP17A1</i>	Steroid-17 alpha-hydroxylase deficiency
10	104591315	G	A	-	NA	NM_000102.3:c.1193C-T	NP_000093.1:p.A398V	<i>CYP17A1</i>	17-alpha-hydroxylase/17,20-lyase deficiency
10	104591346	T	A	-	NA	NM_000102.3:c.1162A-T	NP_000093.1:p.K388X	<i>CYP17A1</i>	17-alpha-hydroxylase/17,20-lyase deficiency
10	104592289	T	A	-	NA	NM_000102.3:c.1118A-T	NP_000093.1:p.H373L	<i>CYP17A1</i>	Steroid-17 alpha-hydroxylase deficiency
10	104592290	G	C	-	NA	NM_000102.3:c.1117C-G	NP_000093.1:p.H373D	<i>CYP17A1</i>	17-alpha-hydroxylase/17,20-lyase

10	104592290	G	T	-	NA	NM_000102.3:c.1117C-A	NP_000093.1:p.H373N	<i>CYP17A1</i>	deficiency 17-alpha-hydroxylase/17,20-lyase deficiency
10	104592322	C	T	-	NA	NM_000102.3:c.1085G-A	NP_000093.1:p.R362H	<i>CYP17A1</i>	17-alpha-hydroxylase/17,20-lyase deficiency
10	104592323	G	A	-	NA	NM_000102.3:c.1084C-T	NP_000093.1:p.R362C	<i>CYP17A1</i>	Steroid-17 alpha-hydroxylase deficiency
10	104592334	C	T	-	NA	NM_000102.3:c.1073G-A	NP_000093.1:p.R358Q	<i>CYP17A1</i>	Steroid-17 alpha-hydroxylase deficiency
10	104592335	G	A	-	NA	NM_000102.3:c.1072C-T	NP_000093.1:p.R358X	<i>CYP17A1</i>	17-alpha-hydroxylase/17,20-lyase deficiency
10	104592344	C	T	-	NA	NM_000102.3:c.1063G-A	NP_000093.1:p.A355T	<i>CYP17A1</i>	17-alpha-hydroxylase/17,20-lyase deficiency
10	104592367	C	T	-	rs61754278	NM_000102.3:c.1040G-A	NP_000093.1:p.R347H	<i>CYP17A1</i>	Steroid-17 alpha-hydroxylase deficiency
10	104592368	G	A	-	NA	NM_000102.3:c.1039C-T	NP_000093.1:p.R347C	<i>CYP17A1</i>	17,20-lyase deficiency
10	104592383	G	T	-	NA	NM_000102.3:c.1024C-A	NP_000093.1:p.P342T	<i>CYP17A1</i>	Pseudohermaphroditism
10	104592412	A	G	-	NA	NM_000102.3:c.995T-C	NP_000093.1:p.I332T	<i>CYP17A1</i>	17-alpha-hydroxylase/17,20-lyase deficiency
10	104592420	G	C	-	NA	NM_000102.3:c.987C-G	NP_000093.1:p.Y329X	<i>CYP17A1</i>	Steroid-17 alpha-hydroxylase deficiency
10	104592422	A	C	-	NA	NM_000102.3:c.985T-G	NP_000093.1:p.Y329D	<i>CYP17A1</i>	Steroid-17 alpha-hydroxylase deficiency
10	104592805	T	C	-	NA	NM_000102.3:c.914A-G	NP_000093.1:p.E305G	<i>CYP17A1</i>	17,20-lyase deficiency
10	104592815	C	G	-	NA	NM_000102.3:c.904G-C	NP_000093.1:p.A302P	<i>CYP17A1</i>	Steroid-17 alpha-hydroxylase deficiency
10	104592856	G	T	-	NA	NM_000102.3:c.863C-A	NP_000093.1:p.S288X	<i>CYP17A1</i>	Steroid-17 alpha-hydroxylase deficiency
10	104593830	C	T	-	NA	NM_000102.3:c.716G-A	NP_000093.1:p.R239Q	<i>CYP17A1</i>	Steroid-17 alpha-hydroxylase deficiency
10	104593831	G	A	-	NA	NM_000102.3:c.715C-T	NP_000093.1:p.R239X	<i>CYP17A1</i>	Pseudohermaphroditism
10	104593863	G	A	-	NA	NM_000102.3:c.683C-T	NP_000093.1:p.T228I	<i>CYP17A1</i>	Steroid-17 alpha-hydroxylase deficiency ?
10	104594582	A	G	-	NA	NM_000102.3:c.626T-C	NP_000093.1:p.L209P	<i>CYP17A1</i>	17-alpha-hydroxylase/17,20-lyase deficiency
10	104594607	A	T	-	NA	NM_000102.3:c.601T-A	NP_000093.1:p.Y201N	<i>CYP17A1</i>	17-alpha-hydroxylase/17,20-lyase deficiency
10	104594628	C	A	-	NA	NM_000102.3:c.580G-T	NP_000093.1:p.E194X	<i>CYP17A1</i>	Steroid-17 alpha-hydroxylase deficiency
10	104594675	A	T	-	NA	NM_000102.3:c.533T-A	NP_000093.1:p.V178D	<i>CYP17A1</i>	Steroid-17 alpha-hydroxylase deficiency
10	104594679	T	C	-	NA	NM_000102.3:c.529A-G	NP_000093.1:p.N177D	<i>CYP17A1</i>	17,20-lyase deficiency
10	104594687	G	T	-	NA	NM_000102.3:c.521C-A	NP_000093.1:p.A174E	<i>CYP17A1</i>	Steroid-17 alpha-hydroxylase deficiency
10	104595073	C	T	-	NA	NM_000102.3:c.374G-A	NP_000093.1:p.R125Q	<i>CYP17A1</i>	Steroid-17 alpha-hydroxylase deficiency
10	104595074	G	A	-	NA	NM_000102.3:c.373C-T	NP_000093.1:p.R125X	<i>CYP17A1</i>	17-alpha-hydroxylase/17,20-lyase deficiency
10	104595100	T	A	-	NA	NM_000102.3:c.347A-T	NP_000093.1:p.D116V	<i>CYP17A1</i>	17-alpha-hydroxylase/17,20-lyase deficiency

10	104595107	A	C	-	NA	NM_000102.3:c.340T-G	NP_000093.1:p.F114V	<i>CYP17A1</i>	17-alpha-hydroxylase/17,20-lyase deficiency
10	104595116	C	T	-	NA	NM_000102.3:c.331G-A	NP_000093.1:p.G111S	<i>CYP17A1</i>	17-alpha-hydroxylase/17,20-lyase deficiency
10	104595119	T	A	-	NA	NM_000102.3:c.328A-T	NP_000093.1:p.K110X	<i>CYP17A1</i>	17-alpha-hydroxylase/17,20-lyase deficiency
10	104595131	A	G	-	NA	NM_000102.3:c.316T-C	NP_000093.1:p.S106P	<i>CYP17A1</i>	Steroid-17 alpha-hydroxylase deficiency
10	104596832	C	T	-	NA	NM_000102.3:c.287G-A	NP_000093.1:p.R96Q	<i>CYP17A1</i>	17-alpha-hydroxylase/17,20-lyase deficiency
10	104596833	G	A	-	NA	NM_000102.3:c.286C-T	NP_000093.1:p.R96W	<i>CYP17A1</i>	17-alpha-hydroxylase/17,20-lyase deficiency
10	104596841	A	C	-	NA	NM_000102.3:c.278T-G	NP_000093.1:p.F93C	<i>CYP17A1</i>	17-alpha-hydroxylase/17,20-lyase deficiency
10	104596850	C	T	-	NA	NM_000102.3:c.269G-A	NP_000093.1:p.G90D	<i>CYP17A1</i>	Steroid-17 alpha-hydroxylase deficiency
10	104596874	G	T	-	NA	NM_000102.3:c.245C-A	NP_000093.1:p.A82D	<i>CYP17A1</i>	17-alpha-hydroxylase/17,20-lyase deficiency
10	104596928	T	G	-	NA	NM_000102.3:c.191A-C	NP_000093.1:p.Y64S	<i>CYP17A1</i>	17-alpha-hydroxylase/17,20-lyase deficiency
10	104597038	G	T	-	NA	NM_000102.3:c.81C-A	NP_000093.1:p.Y27X	<i>CYP17A1</i>	17-alpha-hydroxylase/17,20-lyase deficiency
10	104597057	C	T	-	rs61754263	NM_000102.3:c.62G-A	NP_000093.1:p.R21K	<i>CYP17A1</i>	17-alpha-hydroxylase/17,20-lyase deficiency
10	104597068	C	T	-	NA	NM_000102.3:c.51G-A	NP_000093.1:p.W17X	<i>CYP17A1</i>	Steroid-17 alpha-hydroxylase deficiency
10	104597116	C	G	-	NA	NM_000102.3:c.3G-C	NP_000093.1:p.M1I	<i>CYP17A1</i>	Steroid-17 alpha-hydroxylase deficiency
10	104597117	A	G	-	NA	NM_000102.3:c.2T-C	NP_000093.1:p.M1T	<i>CYP17A1</i>	Steroid-17 alpha-hydroxylase deficiency
12	6457364	G	A	-	NA	NM_001038.5:c.1685C-T	NP_001029.1:p.S562L	<i>SCNN1A</i>	Pseudohypoaldosteronism 1
12	6458147	G	A	-	NA	NM_001038.5:c.1522C-T	NP_001029.1:p.R508X	<i>SCNN1A</i>	Pseudohypoaldosteronism 1
12	6458353	G	A	-	NA	NM_001038.5:c.1474C-T	NP_001029.1:p.R492X	<i>SCNN1A</i>	Pseudohypoaldosteronism 1
12	6458548	C	T	-	NA	NM_001038.5:c.1384G-A	NP_001029.1:p.V462I	<i>SCNN1A</i>	Bronchiectasis
12	6464943	C	A	-	NA	NM_001038.5:c.979G-T	NP_001029.1:p.G327C	<i>SCNN1A</i>	Pseudohypoaldosteronism 1
12	6471365	A	G	-	NA	NM_001038.5:c.727T-C	NP_001029.1:p.S243P	<i>SCNN1A</i>	Pseudohypoaldosteronism 1
12	6472752	G	A	-	rs55797039	NM_001038.5:c.541C-T	NP_001029.1:p.R181W	<i>SCNN1A</i>	Cystic fibrosis, non-classic
12	6483552	C	T	-	NA	NM_001038.5:c.398G-A	NP_001029.1:p.C133Y	<i>SCNN1A</i>	Pseudohypoaldosteronism 1
12	6483610	C	T	-	NA	NM_001038.5:c.340G-A	NP_001029.1:p.V114I	<i>SCNN1A</i>	Cystic fibrosis, non-classic
12	6483767	G	T	-	NA	NM_001038.5:c.183C-A	NP_001029.1:p.F61L	<i>SCNN1A</i>	Cystic fibrosis, non-classic
12	6483784	G	A	-	NA	NM_001038.5:c.166C-T	NP_001029.1:p.R56X	<i>SCNN1A</i>	Pseudohypoaldosteronism 1
16	23200921	G	A	+	rs5736	NM_001039.3:c.547G-A	NP_001030.2:p.G183S	<i>SCNN1G</i>	Bronchiectasis
16	23200963	G	A	+	rs5738	NM_001039.3:c.589G-A	NP_001030.2:p.E197K	<i>SCNN1G</i>	Bronchiectasis

16	23224022	C	T	+	NA	NM_001039.3:c.1318C-T	NP_001030.2:p.R440X	SCNN1G	Pseudohypoaldosteronism 1
16	23226429	A	G	+	rs148985177	NM_001039.3:c.1589A-G	NP_001030.2:p.N530S	SCNN1G	Liddle syndrome
16	23226539	C	T	+	NA	NM_001039.3:c.1699C-T	NP_001030.2:p.Q567X	SCNN1G	Liddle syndrome
16	23226558	G	A	+	NA	NM_001039.3:c.1718G-A	NP_001030.2:p.W573X	SCNN1G	Pseudohypoaldosteronism 1
16	23226564	G	A	+	NA	NM_001039.3:c.1724G-A	NP_001030.2:p.W575X	SCNN1G	Pseudohypoaldosteronism 1
16	23226573	C	T	+	NA	NM_001039.3:c.1733C-T	NP_001030.2:p.A578V	SCNN1G	Hypertension
16	23226647	C	T	+	NA	NM_001039.3:c.1807C-T	NP_001030.2:p.P603S	SCNN1G	Hypertension
16	23226667	G	C	+	NA	NM_001039.3:c.1827G-C	NP_001030.2:p.L609F	SCNN1G	Hypertension
16	23360029	G	A	+	NA	NM_000336.2:c.109G-A	NP_000327.2:p.G37S	SCNN1B	Pseudohypoaldosteronism 1
16	23360165	C	G	+	rs35731153	NM_000336.2:c.245C-G	NP_000327.2:p.S82C	SCNN1B	Cystic fibrosis, non-classic
16	23366671	C	T	+	NA	NM_000336.2:c.637C-T	NP_000327.2:p.Q213X	SCNN1B	Pseudohypoaldosteronism 1
16	23379200	C	T	+	NA	NM_000336.2:c.800C-T	NP_000327.2:p.P267L	SCNN1B	Cystic fibrosis, non-classic
16	23379263	A	G	+	NA	NM_000336.2:c.863A-G	NP_000327.2:p.N288S	SCNN1B	Bronchiectasis
16	23379280	G	A	+	rs72654338	NM_000336.2:c.880G-A	NP_000327.2:p.G294S	SCNN1B	Cystic fibrosis, non-classic
16	23382781	G	A	+	rs61759921	NM_000336.2:c.1042G-A	NP_000327.2:p.V348M	SCNN1B	Cystic fibrosis, non-classic
16	23383157	C	A	+	NA	NM_000336.2:c.1105C-A	NP_000327.2:p.P369T	SCNN1B	Bronchiectasis ?
16	23391814	G	A	+	NA	NM_000336.2:c.1615G-A	NP_000327.2:p.E539K	SCNN1B	Cystic fibrosis, non-classic
16	23391887	G	A	+	rs149868979	NM_000336.2:c.1688G-A	NP_000327.2:p.R563Q	SCNN1B	Hypertension
16	23391895	C	T	+	NA	NM_000336.2:c.1696C-T	NP_000327.2:p.R566X	SCNN1B	Liddle syndrome
16	23391970	C	T	+	NA	NM_000336.2:c.1771C-T	NP_000327.2:p.Q591X	SCNN1B	Liddle syndrome
16	23391973	C	T	+	NA	NM_000336.2:c.1774C-T	NP_000327.2:p.P592S	SCNN1B	Hypertension
16	23392048	C	T	+	NA	NM_000336.2:c.1849C-T	NP_000327.2:p.P617S	SCNN1B	Liddle syndrome
16	23392049	C	A	+	NA	NM_000336.2:c.1850C-A	NP_000327.2:p.P617H	SCNN1B	Liddle syndrome
16	23392049	C	T	+	NA	NM_000336.2:c.1850C-T	NP_000327.2:p.P617L	SCNN1B	Liddle syndrome
16	23392051	C	T	+	NA	NM_000336.2:c.1852C-T	NP_000327.2:p.P618S	SCNN1B	Liddle syndrome
16	23392052	C	A	+	NA	NM_000336.2:c.1853C-A	NP_000327.2:p.P618H	SCNN1B	Liddle syndrome
16	23392052	C	G	+	NA	NM_000336.2:c.1853C-G	NP_000327.2:p.P618R	SCNN1B	Liddle syndrome
16	23392052	C	T	+	NA	NM_000336.2:c.1853C-T	NP_000327.2:p.P618L	SCNN1B	Liddle syndrome
16	23392057	T	C	+	NA	NM_000336.2:c.1858T-C	NP_000327.2:p.Y620H	SCNN1B	Liddle syndrome
16	23392093	G	A	+	NA	NM_000336.2:c.1894G-A	NP_000327.2:p.E632K	SCNN1B	Hypertension
16	67469696	A	T	+	NA	NM_000196.3:c.431A-T	NP_000187.3:p.D144V	HSD11B2	Hypertension, hypokalemic
16	67469917	T	G	+	NA	NM_000196.3:c.536T-G	NP_000187.3:p.L179R	HSD11B2	Apparent mineralocorticoid excess
16	67469920	C	T	+	NA	NM_000196.3:c.539C-T	NP_000187.3:p.S180F	HSD11B2	Apparent mineralocorticoid excess
16	67469935	T	C	+	NA	NM_000196.3:c.554T-C	NP_000187.3:p.F185S	HSD11B2	Hypertension, hypokalemic
16	67469937	C	T	+	NA	NM_000196.3:c.556C-T	NP_000187.3:p.R186C	HSD11B2	Apparent mineralocorticoid excess
16	67470003	C	T	+	NA	NM_000196.3:c.622C-T	NP_000187.3:p.R208C	HSD11B2	Apparent mineralocorticoid excess

16	67470004	G	A	+	rs28934592	NM_000196.3:c.623G-A	NP_000187.3:p.R208H	<i>HSD11B2</i>	Apparent mineralocorticoid excess
16	67470018	C	T	+	NA	NM_000196.3:c.637C-T	NP_000187.3:p.R213C	<i>HSD11B2</i>	Apparent mineralocorticoid excess
16	67470043	C	T	+	NA	NM_000196.3:c.662C-T	NP_000187.3:p.A221V	<i>HSD11B2</i>	Apparent mineralocorticoid excess
16	67470154	G	A	+	NA	NM_000196.3:c.667G-A	NP_000187.3:p.D223N	<i>HSD11B2</i>	Apparent mineralocorticoid excess
16	67470167	C	T	+	NA	NM_000196.3:c.680C-T	NP_000187.3:p.P227L	<i>HSD11B2</i>	Hypertension
16	67470182	A	G	+	NA	NM_000196.3:c.695A-G	NP_000187.3:p.Y232C	<i>HSD11B2</i>	Apparent mineralocorticoid excess
16	67470197	C	T	+	NA	NM_000196.3:c.710C-T	NP_000187.3:p.A237V	<i>HSD11B2</i>	Apparent mineralocorticoid excess
16	67470217	G	A	+	NA	NM_000196.3:c.730G-A	NP_000187.3:p.D244N	<i>HSD11B2</i>	Apparent mineralocorticoid excess
16	67470236	T	G	+	NA	NM_000196.3:c.749T-G	NP_000187.3:p.L250R	<i>HSD11B2</i>	Apparent mineralocorticoid excess
16	67470523	C	T	+	rs28934594	NM_000196.3:c.835C-T	NP_000187.3:p.R279C	<i>HSD11B2</i>	Apparent mineralocorticoid excess
16	67470671	C	T	+	NA	NM_000196.3:c.983C-T	NP_000187.3:p.A328V	<i>HSD11B2</i>	Apparent mineralocorticoid excess
16	67470697	C	T	+	NA	NM_000196.3:c.1009C-T	NP_000187.3:p.R337C	<i>HSD11B2</i>	Apparent mineralocorticoid excess
16	67470700	T	C	+	NA	NM_000196.3:c.1012T-C	NP_000187.3:p.Y338H	<i>HSD11B2</i>	Apparent mineralocorticoid excess
16	67470763	C	T	+	NA	NM_000196.3:c.1075C-T	NP_000187.3:p.R359W	<i>HSD11B2</i>	Apparent mineralocorticoid excess
16	67470808	C	T	+	NA	NM_000196.3:c.1120C-T	NP_000187.3:p.R374X	<i>HSD11B2</i>	Hypertension
16	67470815	T	C	+	NA	NM_000196.3:c.1127T-C	NP_000187.3:p.L376P	<i>HSD11B2</i>	Apparent mineralocorticoid excess
17	40939342	G	A	+	rs55997156	NM_032387.4:c.1523G-A	NP_115763.2:p.R508H	<i>WNK4</i>	Hypertension in pregnancy ?
17	40939417	T	C	+	rs112963672	NM_032387.4:c.1598T-C	NP_115763.2:p.L533P	<i>WNK4</i>	Hypertension in pregnancy ?
17	40939455	A	G	+	NA	NM_032387.4:c.1636A-G	NP_115763.2:p.M546V	<i>WNK4</i>	Hypertension ?
17	40939485	C	A	+	rs56003090	NM_032387.4:c.1666C-A	NP_115763.2:p.P556T	<i>WNK4</i>	Hypertension ?
17	40939498	A	G	+	NA	NM_032387.4:c.1679A-G	NP_115763.2:p.E560G	<i>WNK4</i>	Pseudohypoaldosteronism 2
17	40939501	C	T	+	NA	NM_032387.4:c.1682C-T	NP_115763.2:p.P561L	<i>WNK4</i>	Pseudohypoaldosteronism 2
17	40939503	G	A	+	NA	NM_032387.4:c.1684G-A	NP_115763.2:p.E562K	<i>WNK4</i>	Pseudohypoaldosteronism 2
17	40939509	G	C	+	NA	NM_032387.4:c.1690G-C	NP_115763.2:p.D564H	<i>WNK4</i>	Pseudohypoaldosteronism 2
17	40939510	A	C	+	NA	NM_032387.4:c.1691A-C	NP_115763.2:p.D564A	<i>WNK4</i>	Pseudohypoaldosteronism 2
17	40939512	C	G	+	NA	NM_032387.4:c.1693C-G	NP_115763.2:p.Q565E	<i>WNK4</i>	Pseudohypoaldosteronism 2
17	40948214	A	G	+	NA	NM_032387.4:c.3505A-G	NP_115763.2:p.K1169E	<i>WNK4</i>	Pseudohypoaldosteronism 2
17	40948226	C	A	+	NA	NM_032387.4:c.3517C-A	NP_115763.2:p.P1173T	<i>WNK4</i>	Hypertension ?
17	40948262	C	T	+	NA	NM_032387.4:c.3553C-T	NP_115763.2:p.R1185C	<i>WNK4</i>	Pseudohypoaldosteronism 2
4	149002514	A	G	-	NA	NM_000901.4:c.2936T-C	NP_000892.2:p.L979P	<i>NR3C2</i>	Pseudohypoaldosteronism 1
4	149002535	T	C	-	NA	NM_000901.4:c.2915A-G	NP_000892.2:p.E972G	<i>NR3C2</i>	Pseudohypoaldosteronism 1
4	149002551	G	A	-	NA	NM_000901.4:c.2899C-T	NP_000892.2:p.Q967X	<i>NR3C2</i>	Pseudohypoaldosteronism 1
4	149002611	G	A	-	NA	NM_000901.4:c.2839C-T	NP_000892.2:p.R947X	<i>NR3C2</i>	Pseudohypoaldosteronism 1
4	149002637	A	C	-	NA	NM_000901.4:c.2813T-G	NP_000892.2:p.L938R	<i>NR3C2</i>	Pseudohypoaldosteronism 1
4	149035283	A	G	-	NA	NM_000901.4:c.2771T-C	NP_000892.2:p.L924P	<i>NR3C2</i>	Pseudohypoaldosteronism 1
4	149041320	A	G	-	NA	NM_000901.4:c.2630T-C	NP_000892.2:p.L877P	<i>NR3C2</i>	Pseudohypoaldosteronism 1

4	149041369	G	A	-	NA	NM_000901.4:c.2581C-T	NP_000892.2:p.R861X	NR3C2	Pseudohypoaldosteronism 1
4	149041407	A	G	-	NA	NM_000901.4:c.2543T-C	NP_000892.2:p.L848P	NR3C2	Pseudohypoaldosteronism 1
4	149041423	A	G	-	NA	NM_000901.4:c.2527T-C	NP_000892.2:p.S843P	NR3C2	Pseudohypoaldosteronism 1
4	149073677	G	A	-	NA	NM_000901.4:c.2453C-T	NP_000892.2:p.S818L	NR3C2	Pseudohypoaldosteronism 1
4	149073685	G	T	-	NA	NM_000901.4:c.2445C-A	NP_000892.2:p.S815R	NR3C2	Pseudohypoaldosteronism 1
4	149073701	G	A	-	rs41511344	NM_000901.4:c.2429C-T	NP_000892.2:p.S810L	NR3C2	Hypertension, early onset exacerbated in pregnancy
4	149073717	A	G	-	NA	NM_000901.4:c.2413T-C	NP_000892.2:p.S805P	NR3C2	Pseudohypoaldosteronism 1
4	149075740	T	C	-	NA	NM_000901.4:c.2327A-G	NP_000892.2:p.Q776R	NR3C2	Pseudohypoaldosteronism 1
4	149075757	G	T	-	NA	NM_000901.4:c.2310C-A	NP_000892.2:p.N770K	NR3C2	Pseudohypoaldosteronism 1
4	149075792	G	A	-	NA	NM_000901.4:c.2275C-T	NP_000892.2:p.P759S	NR3C2	Pseudohypoaldosteronism 1
4	149076043	G	C	-	NA	NM_000901.4:c.2024C-G	NP_000892.2:p.S675X	NR3C2	Pseudohypoaldosteronism 1
4	149076047	T	A	-	NA	NM_000901.4:c.2020A-T	NP_000892.2:p.K674X	NR3C2	Pseudohypoaldosteronism 1
4	149076050	G	A	-	NA	NM_000901.4:c.2017C-T	NP_000892.2:p.R673X	NR3C2	Pseudohypoaldosteronism 1
4	149115934	T	G	-	NA	NM_000901.4:c.1977A-C	NP_000892.2:p.R659S	NR3C2	Pseudohypoaldosteronism 1
4	149115957	G	A	-	NA	NM_000901.4:c.1954C-T	NP_000892.2:p.R652X	NR3C2	Pseudohypoaldosteronism 1
4	149115976	G	T	-	NA	NM_000901.4:c.1935C-A	NP_000892.2:p.C645X	NR3C2	Pseudohypoaldosteronism 1
4	149115977	C	G	-	NA	NM_000901.4:c.1934G-C	NP_000892.2:p.C645S	NR3C2	Pseudohypoaldosteronism 1
4	149181130	C	T	-	NA	NM_000901.4:c.1897G-A	NP_000892.2:p.G633R	NR3C2	Pseudohypoaldosteronism 1
4	149181210	C	G	-	NA	NM_000901.4:c.1817G-C	NP_000892.2:p.C606S	NR3C2	Pseudohypoaldosteronism 1
4	149181259	G	A	-	NA	NM_000901.4:c.1768C-T	NP_000892.2:p.R590X	NR3C2	Pseudohypoaldosteronism 1
4	149356334	C	T	-	NA	NM_000901.4:c.1679G-A	NP_000892.2:p.W560X	NR3C2	Pseudohypoaldosteronism 1
4	149356404	G	A	-	NA	NM_000901.4:c.1609C-T	NP_000892.2:p.R537X	NR3C2	Pseudohypoaldosteronism 1
4	149356705	A	T	-	NA	NM_000901.4:c.1308T-A	NP_000892.2:p.C436X	NR3C2	Pseudohypoaldosteronism 1
4	149356984	G	T	-	NA	NM_000901.4:c.1029C-A	NP_000892.2:p.Y343X	NR3C2	Pseudohypoaldosteronism 1
4	149357525	G	C	-	NA	NM_000901.4:c.488C-G	NP_000892.2:p.S163X	NR3C2	Pseudohypoaldosteronism 1
4	149357611	A	T	-	NA	NM_000901.4:c.402T-A	NP_000892.2:p.Y134X	NR3C2	Pseudohypoaldosteronism 1
5	142661470	A	G	-	NA	NM_001018077.1:c.2318T-C	NP_001018087.1:p.L773P	NR3C1	Glucocorticoid receptor deficiency
5	142661547	A	C	-	NA	NM_001018077.1:c.2241T-G	NP_001018087.1:p.I747M	NR3C1	Glucocorticoid receptor deficiency
5	142661579	A	G	-	NA	NM_001018077.1:c.2209T-C	NP_001018087.1:p.F737L	NR3C1	Glucocorticoid receptor deficiency
5	142661603	C	T	-	NA	NM_001018077.1:c.2185G-A	NP_001018087.1:p.V729I	NR3C1	Glucocorticoid receptor deficiency
5	142662173	C	T	-	NA	NM_001018077.1:c.2141G-A	NP_001018087.1:p.R714Q	NR3C1	Glucocorticoid receptor deficiency

5	142662279	C	T	-	NA	NM_001018077.1:c.2035G-A	NP_001018087.1:p.G679S	NR3C1	Glucocorticoid receptor deficiency
5	142675126	T	A	-	NA	NM_001018077.1:c.1922A-T	NP_001018087.1:p.D641V	NR3C1	Glucocorticoid receptor deficiency
5	142680085	A	G	-	NA	NM_001018077.1:c.1712T-C	NP_001018087.1:p.V571A	NR3C1	Glucocorticoid receptor deficiency
5	142680121	A	T	-	NA	NM_001018077.1:c.1676T-A	NP_001018087.1:p.I559N	NR3C1	Glucocorticoid receptor deficiency
5	142680130	G	A	-	NA	NM_001018077.1:c.1667C-T	NP_001018087.1:p.T556I	NR3C1	Glucocorticoid resistance
5	142689700	C	T	-	NA	NM_001018077.1:c.1430G-A	NP_001018087.1:p.R477H	NR3C1	Glucocorticoid receptor deficiency
5	142689725	G	A	-	NA	NM_001018077.1:c.1405C-T	NP_001018087.1:p.R469X	NR3C1	Glucocorticoid receptor deficiency
5	142693717	C	G	-	NA	NM_001018077.1:c.1201G-C	NP_001018087.1:p.D401H	NR3C1	Increased glucocorticoid sensitivity
8	143955835	A	G	-	NA	NM_000497.3:c.1466T-C	NP_000488.3:p.L489S	CYP11B1	Adrenal hyperplasia
8	143956389	A	G	-	NA	NM_000497.3:c.1382T-C	NP_000488.3:p.L461P	CYP11B1	Adrenal hyperplasia
8	143956411	G	A	-	NA	NM_000497.3:c.1360C-T	NP_000488.3:p.R454C	CYP11B1	Steroid-11 beta-hydroxylase deficiency
8	143956413	C	T	-	NA	NM_000497.3:c.1358G-A	NP_000488.3:p.R453Q	CYP11B1	Adrenal hyperplasia
8	143956428	C	T	-	rs28934586	NM_000497.3:c.1343G-A	NP_000488.3:p.R448H	CYP11B1	Steroid-11 beta-hydroxylase deficiency
8	143956429	G	A	-	NA	NM_000497.3:c.1342C-T	NP_000488.3:p.R448C	CYP11B1	Adrenal hyperplasia
8	143956434	C	A	-	NA	NM_000497.3:c.1337G-T	NP_000488.3:p.G446V	CYP11B1	Steroid-11 beta-hydroxylase deficiency
8	143956440	C	T	-	NA	NM_000497.3:c.1331G-A	NP_000488.3:p.G444D	CYP11B1	Steroid-11 beta-hydroxylase deficiency
8	143956449	A	C	-	NA	NM_000497.3:c.1322T-G	NP_000488.3:p.V441G	CYP11B1	Steroid-11 beta-hydroxylase deficiency
8	143956491	C	T	-	NA	NM_000497.3:c.1280G-A	NP_000488.3:p.R427H	CYP11B1	Adrenal hyperplasia
8	143956502	A	C	-	NA	NM_000497.3:c.1269T-G	NP_000488.3:p.Y423X	CYP11B1	Steroid-11 beta-hydroxylase deficiency
8	143956693	G	A	-	rs4541	NM_000497.3:c.1157C-T	NP_000488.3:p.A386V	CYP11B1	Adrenal hyperplasia
8	143956699	C	T	-	NA	NM_000497.3:c.1151G-A	NP_000488.3:p.R384Q	CYP11B1	Steroid-11 beta-hydroxylase deficiency
8	143956700	G	C	-	NA	NM_000497.3:c.1150C-G	NP_000488.3:p.R384G	CYP11B1	Steroid-11 beta-hydroxylase deficiency
8	143956714	C	A	-	NA	NM_000497.3:c.1136G-T	NP_000488.3:p.G379V	CYP11B1	Steroid-11 beta-hydroxylase deficiency
8	143957128	C	T	-	NA	NM_000497.3:c.1121G-A	NP_000488.3:p.R374Q	CYP11B1	Steroid-11 beta-hydroxylase deficiency
8	143957137	T	C	-	NA	NM_000497.3:c.1112A-G	NP_000488.3:p.E371G	CYP11B1	Adrenal hyperplasia
8	143957146	G	T	-	NA	NM_000497.3:c.1103C-A	NP_000488.3:p.A368D	CYP11B1	Steroid-11 beta-hydroxylase deficiency
8	143957183	G	A	-	rs146124466	NM_000497.3:c.1066C-T	NP_000488.3:p.Q356X	CYP11B1	Steroid-11 beta-hydroxylase deficiency
8	143957228	G	T	-	NA	NM_000497.3:c.1021C-A	NP_000488.3:p.R341S	CYP11B1	Adrenal hyperplasia
8	143957237	G	A	-	NA	NM_000497.3:c.1012C-T	NP_000488.3:p.Q338X	CYP11B1	Steroid-11 beta-hydroxylase deficiency

8	143957257	G	A	-	NA	NM_000497.3:c.992C-T	NP_000488.3:p.A331V	<i>CYP11B1</i>	Adrenal hyperplasia
8	143957288	A	C	-	NA	NM_000497.3:c.961T-G	NP_000488.3:p.F321V	<i>CYP11B1</i>	Adrenal hyperplasia
8	143957293	G	A	-	NA	NM_000497.3:c.956C-T	NP_000488.3:p.T319M	<i>CYP11B1</i>	Steroid-11 beta-hydroxylase deficiency
8	143957658	G	A	-	NA	NM_000497.3:c.953C-T	NP_000488.3:p.T318M	<i>CYP11B1</i>	Steroid-11 beta-hydroxylase deficiency
8	143957658	G	C	-	NA	NM_000497.3:c.953C-G	NP_000488.3:p.T318R	<i>CYP11B1</i>	Steroid-11 beta-hydroxylase deficiency
8	143957659	T	G	-	NA	NM_000497.3:c.952A-C	NP_000488.3:p.T318P	<i>CYP11B1</i>	Steroid-11 beta-hydroxylase deficiency
8	143957671	C	G	-	NA	NM_000497.3:c.940G-C	NP_000488.3:p.G314R	<i>CYP11B1</i>	Steroid-11 beta-hydroxylase deficiency
8	143957694	G	A	-	NA	NM_000497.3:c.917C-T	NP_000488.3:p.A306V	<i>CYP11B1</i>	Steroid-11 beta-hydroxylase deficiency
8	143957715	A	G	-	NA	NM_000497.3:c.896T-C	NP_000488.3:p.L299P	<i>CYP11B1</i>	Steroid-11 beta-hydroxylase deficiency
8	143957811	C	T	-	NA	NM_000497.3:c.800G-A	NP_000488.3:p.G267D	<i>CYP11B1</i>	Adrenal hyperplasia
8	143958098	C	G	-	NA	NM_000497.3:c.799G-C	NP_000488.3:p.G267R	<i>CYP11B1</i>	Adrenal hyperplasia
8	143958104	G	A	-	NA	NM_000497.3:c.793C-T	NP_000488.3:p.Q265X	<i>CYP11B1</i>	Adrenal hyperplasia
8	143958121	G	T	-	NA	NM_000497.3:c.776C-A	NP_000488.3:p.A259D	<i>CYP11B1</i>	Steroid-11 beta-hydroxylase deficiency
8	143958157	C	T	-	NA	NM_000497.3:c.740G-A	NP_000488.3:p.W247X	<i>CYP11B1</i>	Adrenal hyperplasia
8	143958514	T	A	-	NA	NM_000497.3:c.520A-T	NP_000488.3:p.K174X	<i>CYP11B1</i>	Steroid-11 beta-hydroxylase deficiency
8	143958540	G	T	-	NA	NM_000497.3:c.494C-A	NP_000488.3:p.A165D	<i>CYP11B1</i>	Steroid-11 beta-hydroxylase deficiency
8	143958558	G	A	-	NA	NM_000497.3:c.476C-T	NP_000488.3:p.P159L	<i>CYP11B1</i>	Steroid-11 beta-hydroxylase deficiency, non-classic
8	143958613	G	A	-	NA	NM_000497.3:c.421C-T	NP_000488.3:p.R141X	<i>CYP11B1</i>	Steroid-11 beta-hydroxylase deficiency
8	143958637	T	G	-	rs104894067	NM_000497.3:c.397A-C	NP_000488.3:p.N133H	<i>CYP11B1</i>	Steroid-11 beta-hydroxylase deficiency
8	143960458	C	T	-	NA	NM_000497.3:c.385G-A	NP_000488.3:p.V129M	<i>CYP11B1</i>	Adrenal hyperplasia
8	143960495	C	G	-	NA	NM_000497.3:c.348G-C	NP_000488.3:p.W116C	<i>CYP11B1</i>	Steroid-11 beta-hydroxylase deficiency
8	143960496	C	T	-	NA	NM_000497.3:c.347G-A	NP_000488.3:p.W116X	<i>CYP11B1</i>	Steroid-11 beta-hydroxylase deficiency
8	143960497	A	C	-	NA	NM_000497.3:c.346T-G	NP_000488.3:p.W116G	<i>CYP11B1</i>	Steroid-11 beta-hydroxylase deficiency
8	143960562	G	A	-	NA	NM_000497.3:c.281C-T	NP_000488.3:p.P94L	<i>CYP11B1</i>	Steroid-11 beta-hydroxylase deficiency
8	143960579	C	T	-	NA	NM_000497.3:c.264G-A	NP_000488.3:p.M88I	<i>CYP11B1</i>	Steroid-11 beta-hydroxylase deficiency, non-classic
8	143961102	C	T	-	rs4534	NM_000497.3:c.128G-A	NP_000488.3:p.R43Q	<i>CYP11B1</i>	Adrenal hyperplasia
8	143961106	G	A	-	NA	NM_000497.3:c.124C-T	NP_000488.3:p.P42S	<i>CYP11B1</i>	Steroid-11 beta-hydroxylase deficiency
8	143961175	G	A	-	NA	NM_000497.3:c.55C-T	NP_000488.3:p.Q19X	<i>CYP11B1</i>	Steroid-11 beta-hydroxylase deficiency

Online Table III.

<i>Studies</i>	<i>N</i>	<i>% Female</i>	<i>Age in years</i> <i>Mean ± SD (Range)</i>	<i>% Hypertensive</i>	<i>% HTN medicated</i>
ARIC EA	9,747	53.0	54.3 ± 5.7 (44 – 66)	27.3	19.7
ARIC AA	3,207	62.6	53.3 ± 5.8 (44 – 66)	55.4	40.4
CLUE EA	6,591	63.7	55.0 ± 13.5 (28 – 95)	48.0	24.2
FBPP EA	3,659	53.2	50.4 ± 15.6 (11 – 95)	43.2	28.0
FBPP AA	2,862	62.3	44.4 ± 13.9 (12 – 88)	49.5	28.9

<i>Studies</i>	<i>SBP (mmHg)</i> <i>Mean ± SD (Range)</i>	<i>DBP (mmHg)</i> <i>Mean ± SD (Range)</i>	<i>σ_{residuals} (mmHg)</i>	
			<i>SBP</i>	<i>DBP</i>
ARIC EA	121.4 ± 19.5 (61 – 221)	73.6 ± 11.5 (12 – 139)	17.9	11.0
ARIC AA	134.6 ± 23.2 (73 – 257)	83.9 ± 13.7 (34 – 152)	17.7	9.4
CLUE EA	128.7 ± 17.1 (80 – 220)	79.5 ± 9.3 (50 – 134)	17.2	10.0
FBPP EA	121.2 ± 29.4 (68 – 240)	68.9 ± 10.6 (37 – 114)	18.9	10.0
FBPP AA	127.8 ± 21.5 (72 – 221)	74.8 ± 12.7 (39 – 139)	21.3	12.2

Online Table IV.

<i>Gene</i>	EA			AA		
	<i>SBP</i>	<i>DBP</i>	<i>k</i>	<i>SBP</i>	<i>DBP</i>	<i>k</i>
All	0.007	0.004	1,106	0.066	0.064	1,667
<i>AGT</i>	0.004	0.121	59	0.666	0.675	113
<i>CYP11B1</i>	0.022	0.180	52	0.115	0.370	86
<i>CYP17A1</i>	0.026	0.098	19	0.017	0.310	30
<i>HSD11B2</i>	0.010	0.016	52	0.147	0.883	63
<i>NR3C1</i>	0.377	0.703	91	0.030	0.040	163
<i>NR3C2</i>	4.0×10^{-4}	0.058	325	0.024	0.555	494
<i>SCNN1A</i>	0.044	0.242	64	0.305	0.583	136
<i>SCNN1B</i>	0.129	0.172	63	0.057	0.539	109
<i>SCNN1G</i>	1.9×10^{-4}	0.123	131	0.195	0.447	120
<i>WNK1</i>	0.381	0.172	216	0.005	9.8×10^{-4}	281
<i>WNK4</i>	0.376	0.307	34	0.009	0.336	72

Online Table V.

<i>Chr</i>	<i>BP</i>	<i>rsID</i>	<i>A1</i>	<i>A2</i>	<i>MAF (%)</i>	<i>Effect (mmHg)</i>	<i>SE</i>	<i>P-value</i>
10	104598995	chr10:104598995	A	G	9.58	-1.715	0.4167	3.87x10 ⁻⁵
10	104594507	rs1004467	G	A	9.54	-1.759	0.4183	2.64x10 ⁻⁵
10	104604916	rs11191416	G	T	9.59	-1.713	0.4163	3.90x10 ⁻⁵
10	104591393	rs17115100	T	G	9.54	-1.759	0.4183	2.64x10 ⁻⁵
10	104595849	rs3824755	C	G	9.58	-1.738	0.4167	3.05x10 ⁻⁵
10	104604563	rs7912206	T	C	9.58	-1.715	0.4167	3.87x10 ⁻⁵

Online Table VI.

Locus	rsID	Chr	hg19pos	Strand	Common Allele	Rare Allele	CLUE			FBPP_EA			FBPP_AA		
							MAF (%)	SBP P-value	DBP P-value	MAF (%)	SBP P-value	DBP P-value	MAF (%)	SBP P-value	DBP P-value
<i>MOV10</i>	rs2932538	1	113127062	-	C	T	24.9	0.151	0.242	24.3	0.839	0.970	18.1	0.655	0.358
<i>HAAO</i>	rs7585445	2	43083070	+	T	C	26.6	0.735	0.411	27.3	0.960	0.966	47.0	0.258	0.790
<i>SLC4A7</i>	rs13082711	3	27537909	+	T	C	21.9	0.433	0.284	22.9	0.204	0.123	6.0	0.559	0.204
<i>ULK4</i>	rs3774372	3	41877414	+	T	C	17.0	0.814	0.055	16.3	0.951	0.116	19.8	0.272	0.575
<i>MDS1</i>	rs419076	3	169100894	+	C	T	49.5	0.928	0.474	48.0	0.255	0.901	50.1	0.564	0.340
<i>SLC39A8</i>	rs13107325	4	103326864	+	C	T	7.1	0.119	0.051	6.7	0.946	0.824	1.1	0.818	0.977
<i>GUCY1A3</i>	rs13139571	4	156783668	+	C	A	24.8	0.153	0.402	22.5	0.756	0.839	14.5	0.098	0.282
<i>C5orf23</i> <i>NPR3</i>	rs1173771	5	32815028	-	C	T	39.0	0.186	0.155	39.4	0.715	0.636	23.1	0.056	0.207
<i>EBF1</i>	rs11953630	5	157845402	+	C	T	38.3	0.682	0.922	36.4	0.294	0.325	20.2	0.393	0.847
<i>HFE</i> - missense	rs1799945	6	26091179	+	C	G	15.0	0.191	0.089	15.9	0.061	0.290	2.9	0.363	0.834
<i>HFE</i>	rs1800562	6	26093141	+	G	A	6.3	0.890	0.860	6.4	0.332	0.217	0.9	0.978	0.512
<i>BAT3</i>	rs805303	6	31616366	-	C	T	35.8	0.762	0.392	36.8	0.154	0.733	59.5	0.731	0.661
<i>GRIFIN</i>	rs2969070	7	2319260	+	A	G	36.6	0.567	0.900	38.0	0.845	0.817	7.6	0.129	0.246
<i>NRCAM</i>	rs2193241	7	107753620	+	T	C	41.8	0.206	0.859	41.0	0.819	0.426	70.5	0.151	0.103
<i>C8ORF38</i>	rs16917233	8	96044967	+	A	C	14.3	0.695	0.241	14.4	0.636	0.633	14.8	0.604	0.562
<i>CACNB2</i>	rs4373814	10	18419972	+	G	C	43.8	0.541	0.041	42.6	0.245	0.329	56.7	0.119	0.202
<i>PLCE1</i>	rs932764	10	95895940	+	A	G	44.6	0.522	0.132	43.3	0.300	0.523	21.5	0.972	0.399
<i>ADM</i>	rs7129220	11	10350538	+	G	A	11.5	0.841	0.848	10.8	0.552	0.644	6.8	0.042	0.697
<i>TMEM133</i>	rs604723	11	100610546	+	C	T	26.3	0.477	0.481	28.8	0.365	0.420	4.3	0.857	0.440
<i>ATP2B1</i>	rs1681472	12	90008709	-	T	C	17.1	5.18E-04	1.96E-04	16.0	0.023	0.006	10.1	0.182	0.175
<i>TBX3</i>	rs11067335	12	115390155	+	C	T	30.1	0.411	0.418	31.5	0.733	0.496	34.5	0.405	0.784
<i>FES</i>	rs2521501	15	91437388	-	T	A	32.2	1.69E-05	8.05E-04	32.4	0.478	0.357	22.5	0.360	0.172
<i>SLC12A3</i>	rs11648751	16	56937012	+	C	T	11.8	0.376	0.397	11.1	0.272	0.567	17.4	0.442	0.672
<i>GOSR2</i>	rs17608766	17	45013271	+	T	C	13.9	0.016	0.064	13.4	0.922	0.425	2.1	0.582	0.185
<i>ZNF652</i>	rs12940887	17	47402807	+	C	T	37.0	0.938	0.814	38.1	0.459	0.066	8.1	0.981	0.355
<i>JAG1</i>	rs1327235	20	10969030	+	A	G	45.6	0.230	0.125	47.5	0.472	0.013	53.2	0.855	0.469

<i>C20orf174</i> <i>ACAT1</i> <i>EDN3</i>	rs6015450	20	57751117	+	A	G	12.7	0.278	0.237	13.4	0.022	0.253	20.7	0.245	0.513
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Online Table VII.

BP trait	Rare variant	Chr	Alleles	CLUE			FBPP (EA)			FBPP (AA)		
				MAF (%)	Effect	P-value	MAF (%)	Effect	P-value	MAF (%)	Effect	P-value
SBP	rs17755373	12	T	1.00	0.019	0.825	1.40	0.099	0.314	0.30	0.231	0.364
DBP	rs17755373	12	T	1.00	0.155	0.072	1.40	0.041	0.675	0.30	0.302	0.234
PP	rs17755373	12	T	1.00	0.099	0.251	1.40	0.101	0.304	0.30	0.082	0.748
MAP	rs17755373	12	T	1.00	0.099	0.248	1.40	0.074	0.451	0.30	0.298	0.239
SBP	rs2286007	12	T	7.80	0.008	0.814	7.20	0.042	0.354	0.90	-0.03	0.83
DBP	rs2286007	12	T	7.80	0.009	0.79	7.20	0.031	0.488	0.90	0.052	0.712
PP	rs2286007	12	T	7.80	0.004	0.911	7.20	0.033	0.464	0.90	0.001	0.996
MAP	rs2286007	12	T	7.80	0.009	0.785	7.20	0.025	0.583	0.90	0.007	0.963
SBP	rs2681472*	12	C	17.10	0.081	5.18x10⁻⁴	16.00	0.073	0.023	10.10	0.059	0.182
DBP	rs2681472*	12	C	17.10	0.087	1.96x10⁻⁴	16.00	0.087	0.006	10.10	0.059	0.175
PP	rs2681472*	12	C	17.10	0.042	0.07	16.00	0.034	0.281	10.10	0.035	0.433
MAP	rs2681472*	12	C	17.10	0.091	9.44x10⁻⁵	16.00	0.081	0.012	10.10	0.055	0.214
SBP	rs56030257	17	C	0.10	0.074	0.825	0.10	0.212	0.636	0.00	-	-
DBP	rs56030257	17	C	0.10	0.076	0.819	0.10	0.014	0.975	0.00	-	-
PP	rs56030257	17	C	0.10	0.164	0.623	0.10	0.269	0.548	0.00	-	-
MAP	rs56030257	17	C	0.10	0.008	0.982	0.10	0.062	0.889	0.00	-	-
SBP	rs5742912	12	G	2.40	0.022	0.695	1.70	0.156	0.094	0.30	0.103	0.66
DBP	rs5742912	12	G	2.40	0.093	0.101	1.70	0.087	0.35	0.30	0.106	0.651
PP	rs5742912	12	G	2.40	0.045	0.433	1.70	0.145	0.122	0.30	0.23	0.328
MAP	rs5742912	12	G	2.40	0.065	0.249	1.70	0.112	0.235	0.30	0.11	0.637
SBP	rs72646501	16	A	0.20	0.292	0.182	0.10	0.098	0.758	1.80	0.135	0.198
DBP	rs72646501	16	A	0.20	0.3	0.169	0.10	0.179	0.573	1.80	0.165	0.115
PP	rs72646501	16	A	0.20	0.162	0.458	0.10	0.003	0.992	1.80	0.323	0.002
MAP	rs72646501	16	A	0.20	0.321	0.142	0.10	0.2	0.528	1.80	0.073	0.487
SBP	rs72650764	12	G	0.03	0.625	0.377	0.01	0.755	0.451	0.03	0.121	0.866
DBP	rs72650764	12	G	0.03	1.109	0.117	0.01	0.881	0.379	0.03	1.088	0.128
PP	rs72650764	12	G	0.03	0.029	0.967	0.01	0.375	0.708	0.03	0.706	0.328
MAP	rs72650764	12	G	0.03	0.958	0.175	0.01	0.856	0.392	0.03	0.749	0.294
SBP	rs72647528	16	A	0.10	0.157	0.53	0.01	0.395	0.43	0.00	-	-
DBP	rs72647528	16	A	0.10	0.05	0.842	0.01	0.476	0.342	0.00	-	-
PP	rs72647528	16	A	0.10	0.259	0.301	0.01	0.186	0.71	0.00	-	-

	rs72647528											
MAP		16	A	0.10	-0.05	0.843	0.01	0.407	0.415	0.00	-	-
SBP	rs72647542	16	T	0.00	-	-	0.10	0.656	0.143	0.00	-	-
DBP	rs72647542	16	T	0.00	-	-	0.10	0.607	0.175	0.00	-	-
PP	rs72647542	16	T	0.00	-	-	0.10	0.437	0.329	0.00	-	-
MAP	rs72647542	16	T	0.00	-	-	0.10	0.877	0.05	0.00	-	-
	rs72645625											
SBP		4	C	0.00	-	-	0.04	0.954	0.099	0.03	0.273	0.704
DBP	rs72645625	4	C	0.00	-	-	0.04	0.593	0.305	0.03	0.116	0.871
	rs72645625											
PP		4	C	0.00	-	-	0.04	0.838	0.147	0.03	0.478	0.508
	rs72645625											
MAP		4	C	0.00	-	-	0.04	0.725	0.21	0.03	0.017	0.981
	rs72657550											
SBP		12	G	0.00	-	-	0.04	0.363	0.717	0.00	-	-
DBP	rs72657550	12	G	0.00	-	-	0.04	0.178	0.859	0.00	-	-
	rs72657550											
PP		12	G	0.00	-	-	0.04	0.602	0.548	0.00	-	-
	rs72657550											
MAP		12	G	0.00	-	-	0.04	0.852	0.394	0.00	-	-
	rs72654338											
SBP		16	A	0.00	-	-	0.06	0.702	0.321	0.05	0.046	0.937
DBP	rs72654338	16	A	0.00	-	-	0.06	1.043	0.141	0.05	1.112	0.057
	rs72654338											
PP		16	A	0.00	-	-	0.06	0.193	0.785	0.05	0.961	0.103
MAP	rs72654338	16	A	0.00	-	-	0.06	0.811	0.252	0.05	0.806	0.167

Online Table VIII.

		<i>Effect Size (σ)</i>					
(A)	<i>N=3,000</i>	0.01	0.05	0.25	0.50	1.00	2.00
MAF (%)	0.05	*	*	*	*	14	74
	0.10	*	*	*	*	36	98
	1	*	*	19	85	100	100
	5	*	*	92	100	100	100
	10	*	*	100	100	100	100
	20	*	10	100	100	100	100
(B)	<i>N=7,000</i>						
MAF (%)	0.05	*	*	*	*	44	99
	0.10	*	*	*	17	82	100
	1	*	*	55	100	100	100
	5	*	*	100	100	100	100
	10	*	15	100	100	100	100
	20	*	33	100	100	100	100

Online Table IX.

<i>Chromosomal coordinate (hg19)</i>	<i>Gene</i>	<i>Strand</i>	<i>A1</i>	<i>A2</i>	<i>Res 1</i>	<i>Res 2</i>	<i>rsID</i>	<i>EA MAF (%)</i>	<i>AA MAF (%)</i>	<i>phyloP score</i>	<i>Associated Disease</i>
chr1:23084586 6	<i>AGT</i>	-	A	C	L	R	rs5041	0.00	0.61	1.76	Hypertension
chr8:14395669 3	<i>CYP11B</i> 1	-	G	A	A	V	rs4541	1.12	3.85	1.51	Adrenal hyperplasia
chr8:14396110 2	<i>CYP11B</i> 1	-	C	T	R	Q	rs4534	1.07	1.25	0.44	Adrenal hyperplasia
chr10:1045923 67	<i>CYP17A</i> 1	-	C	T	R	H	rs61754278	0.00	0.36	3.98	Steroid-17 alpha-hydroxylase deficiency
chr10:1045970 57	<i>CYP17A</i> 1	-	C	T	R	K	rs61754263	0.36	0.19	-0.39	17-alpha-hydroxylase/17,20-lyase deficiency
chr10:1045971 16	<i>CYP17A</i> 1	-	C	G	M	I		0.00	0.19	5.06	Steroid-17 alpha-hydroxylase deficiency
chr16:2320092 1	<i>SCNN1G</i>	+	G	A	G	S	rs5736	0.00	2.90	0.62	Bronchiectasis
chr16:2320096 3	<i>SCNN1G</i>	+	G	A	E	K	rs5738	1.44	0.36	0.97	Bronchiectasis
chr16:2336016 5	<i>SCNN1B</i>	+	C	G	S	C	rs35731153	0.18	0.00	4.29	Cystic fibrosis, non-classic
chr16:2337928 0*	<i>SCNN1B</i>	+	G	A	G	S	rs72654338	0.19	0.00	3.99	Cystic fibrosis, non-classic
chr16:6747000 4	<i>HSD11B</i> 2	+	G	A	R	H	rs28934592	0.00	0.38	5.17	Apparent mineralocorticoid excess
chr17:4093934 2	<i>WNK4</i>	+	G	A	R	H	rs55997156	0.18	0.00	3.30	Pseudohypoaldosteronism 2

Online Table X.

			EA+AA				
Mutation Class		# Variants	T	T_{mmHg}	SE_T	SE_{mmHg}	# In EVS
(a)	<i>HGMD DMs (111 syndromic patients)</i>	91	3.568	57.1	0.340	5.4	8
(b)	<i>HGMD DMs (69 GenNet)</i>	12	0.728	11.6	0.415	6.6	11
(c)	<i>RS&G SNVs (560 GenNet)</i>	2,379	0.550	8.8	0.019	0.3	808
(d)	<i>Deleterious RS&G SNVs (~13K CLUE+FBPP)</i>	11	0.109	1.7	0.154	2.5	8
(e)	<i>ICBP SNPs (~13K CLUE+FBPP)</i>	27	0.014	0.2	0.003	0.0	-

		EA						AA					
Class	# Variants	T	T_{mmHg}	SE_T	SE_{mmHg}	# In EVS	q (%)	T	T_{mmHg}	SE_T	SE_{mmHg}	# In EVS	q (%)
(a)	91	-	-	-	-	8	0.01	-	-	-	-	8	0.03
(b)	12	0.11 7	1.9	0.49 2	7.9	9	0.86	0.33 8	5.4	0.16 6	2.7	8	2.32
(c)	2,379	0.51 5	8.2	0.01 8	0.3	419	0.05	0.51 3	8.2	0.01 9	0.3	664	1.23
(d)	11	0.15 2	2.4	0.16 0	2.6	8	0.09	0.17 8	2.8	0.04 3	0.7	4	0.43
(e)	27	0.01 2	0.2	0.00 3	0.0	-	(!)	0.02 7	0.4	0.00 8	0.1	-	(!)

Online Figure Legend:

Online Figure 1: Distribution of variants found by RS&G and the 1000 Genomes Project (TGP) for each region that was targeted for resequencing and cumulatively across all genes in EA and AA samples.

Online Figure I.

