

Supplemental Table 1. List of candidate genes analyzed in this study, grouped by indication (disease or pharmacology). Marker SNPs are all located in transcribed regions of the mature mRNA, or a splice variant. For some genes more than one marker SNP and tissue were used.

| Name | Symbol | Indication | Tissue (number of heterozygous subjects) | Marker SNP | % het | Replicates (n) or assay validation |
|--|---------|---|---|---------------------------|-------|--|
| Cardiovascular disease candidate genes | | | | | | |
| Angiotensin I converting enzyme | ACE | Blood pressure, CAD | Heart (31) | rs4309 (exon 9) | 51 | 6 |
| | | | Small bowel (32) | | 37 | 4 |
| | | | Liver (10) | rs4343 (exon 17) | 40 | 6 |
| | | | Heart (29) | rs4586 (exon 2) | 45 | 4 |
| Monocyte chemoattractant protein 1 | CCL2 | CAD, SLE, inflammation, infection | Monocytes (26) | rs13900 (3'UTR) | 46 | 4 |
| | | | | | | |
| Superoxide dismutase, mitochondrial | SOD2 | Oxidative stress, cancer | Heart (34) | rs4880 (exon 2) | 52 | 4 |
| | | | | rs5746092 (5'UTR) | 38 | 4 |
| Nicotinamide <i>N</i> -methyltransferase | NNMT | Homocysteine levels | Liver (19) Small bowel (20) | rs4646335 (exon 1) | 33 | 2 |
| Lipoprotein lipase | LPL | Triglyceride, cholesterol metabolism | Heart (15) | rs1059611 (exon 10) | 23 | 3 |
| 3-hydroxy-3-methylglutaryl-CoA reductase | HMGCR | Cholesterol synthesis, direct target (statins) | Liver (8) | rs12916 (exon 19) | 23 | 1 |
| | | | B-lymph (18) | | 20 | 1 |
| Colony-stimulating factor, macrophage-specific | CSF1 | Inflammation, endothelial function | Monocytes (18) | rs333970 (exon 6) | 43 | 4 |
| Prostaglandin D2 synthase | PTGDS | CAD | Heart (30) | rs6926 (exon 7) | 46 | 2 |
| Hypoxia-inducible factor 1, alpha subunit | HIF1A | Oxidative stress | Heart (18) | rs2057482 (exon 15) | 28 | 2 |
| Nitric oxide synthase, endothelial | NOS3 | Oxidative stress, vascular function | Heart (22) | rs1799983 (exon 8) | 34 | 4 |
| FMS-related tyrosine kinase | FLT1 | Endothelial function | Heart (19) | rs2296189 (exon 23) | 29 | 2 |
| L-type voltage-dependent calcium channel alpha subunit 1C | CACNA1C | Blood pressure | Heart (37) | rs1544514 (exon 4) | 27 | 6 |
| | | | | rs216008 (exon 30) | 40 | 6 |
| Adrenergic receptor beta2 | ADRB2 | Blood pressure | Heart (22) | rs1042719 (exon1) | 46 | 2 |
| | | | | rs11739136 (exon 3) | 12 | 6 |
| | | | | rs2656842 (exon 4) | 46 | 6 |
| BK channel beta1 subunit | KCNMB1 | Blood pressure | Heart (30) | rs2656841 (exon 4) | 47 | 6 |
| | | | | | | |
| Vitamin K epoxide reductase | VKORC1 | Warfarin target | Liver (29) Heart (26) B-lymph (12) | rs7294 (exon 3, 3'UTR) | 40 | 6 |
| Gamma-glutamyl carboxylase | GGCX | Blood coagulation | Liver (23) | rs699664 (exon 8) | 40 | 2 |
| Cholesteryl ester transfer protein | CETP | HDL cholesterol remodeling | Liver (29) | rs5884 (exon 14) | 47 | 6 |
| Heme oxygenase 1 | HMOX1 | Heme catabolism | Heart (5) | rs11555832 (3'UTR) | 8 | 1 |

| Drug metabolism candidate genes | | | | | | |
|---|---------|---------------------|-----------------------------------|--------------------------|----|---|
| Multidrug resistance polypeptide 1, MDR1 | ABCB1 | Drug transporter | Liver (22) | rs1045642 (exon 26) | 50 | 6 |
| | | | | rs1128503 (exon 12) | 44 | 6 |
| | | | | rs2032582 (exon 21) | 47 | 6 |
| Cytochrome P450, subfamily 2D, polypeptide 6 | CYP2D6 | Drug metabolism | Liver (26) | rs1065852 (exon 1) | 26 | 6 |
| | | | | rs1058164 (exon 3) | 32 | 6 |
| | | | | rs16947 (exon 6) | 37 | 6 |
| Cytochrome P450, subfamily 2C, polypeptide 9 | CYP2C9 | Drug metabolism | Liver (19) | rs1135840 (exon 9) | 30 | 6 |
| | | | | rs9332242 (exon 9) | 11 | 4 |
| Human peptide transporter 1 | SLC15A1 | Transporter | Small bowel (21) | rs1057911 (exon 9) | 9 | 4 |
| | | | | rs1339067 (exon 17) | 42 | 4 |
| Human peptide transporter 2 | SLC15A2 | Transporter | Kidney (8) | rs1143670 (exon 14) | 45 | 4 |
| | | | | rs1143671 (exon 15) | 45 | 4 |
| CNS disorder candidate genes | | | | | | |
| Dopamine D2 receptor | DRD2 | CNS | Prefrontal cortex, all lobes (30) | rs6277 (exon 7) | 44 | 6 |
| | | | | rs6275 (exon 7) | 33 | 4 |
| | | | | rs6279 (3' UTR) | 31 | 6 |
| Acetylcholine nicotinic receptor subunit a4 | CHRNA4 | CNS | Striatum (13) | rs1044393 (exon 5) | 20 | 4 |
| | | | | rs1044397 (exon 5) | 40 | 4 |
| | | | | rs2236196 (3' UTR) | 44 | 4 |
| mu Opioid receptor | OPRM1 | CNS | All lobes, Pons (8) | rs1799971 (exon 1) | 8 | 6 |
| Serotonin receptor 2A | HTR2A | CNS | Prefrontal cortex (16) | rs6313 (exon 1) | 15 | 2 |
| Brain-derived neurotrophic factor | BDNF | CNS | All lobes (9) | rs6265 (exon 2) | 35 | 4 |
| Serotonin transporter | SLC6A4 | CNS | Pons (29) | rs1042173 (3'UTR) | 60 | 3 |
| Tryptophan hydroxylase 2 | TPH2 | CNS | Pons (27) | rs7305115 (exon 7) | 38 | 4 |
| | | | | rs4290270 (exon 9) | 46 | 2 |
| Norepinephrine transporter | SLC6A2 | CNS | Pons (18) | rs5569 (exon 9) | 38 | 2 |
| Dysbindin 1 | DTNBP1 | CNS | Pons (9) | rs1047631(3'UTR) | 15 | 2 |
| Neuregulin 1 | NRG1 | CNS | Pons (8) | SNP8NRG433E1006 (exon 1) | 17 | 2 |
| Serotonin receptor 1B | HTR1B | CNS | Pons (16) | rs6296 (exon 1) | 33 | |
| Monoamine oxidase A | MAOA | CNS | Prefrontal cortex (19) | rs1137070 (exon 14) | 47 | 3 |
| | | | | rs6323 (exon 8) | 50 | 3 |
| Dopamine D3 receptor | DRD3 | CNS | Prefrontal cortex (21) | rs6280 (exon 2) | 38 | 3 |
| Estrogen receptor | ESR1 | CNS | Prefrontal cortex (46) | rs3798577 (3'UTR) | 44 | 3 |
| Dopamine transporter | SLC6A3 | CNS | Striatum (21) | rs6347 (exon 9) | 54 | 3 |
| Catechol-O-methyltransferase | COMT | CNS | Prefrontal cortex (10) | rs4633 (exon 3) | 65 | 1 |
| Diamine oxidase | DAO | CNS | Prefrontal cortex (21) | rs2070588 (5'UTR) | 37 | 3 |
| Glucocorticoid receptor | NR3C1 | CNS/stress hormonal | Prefrontal cortex (20) | rs6196 (exon 8) | 19 | 3 |
| Nad(p)h:menadiione oxidoreductase 1, dioxin-inducible 2 | NQO2 | CNS | B-lymph (9) | rs1143684 (exon 3) | 30 | 1 |

Supplemental Table 2. Oligonucleotide primers used for PCR amplification and SNaPshot primer extension reactions. The reverse PCR primer also served for gene specific cDNA synthesis.

| Symbol | Marker SNP(s) | Forward PCR primer | Reverse PCR primer | Extension primer |
|----------------|---------------|---|---|--|
| ACE | rs4309 | 5'TGAGATGGGCCATATACAGTACTAC3' | 5'CCCGACGCAGGGAGAC3' | 5'CTGCAGTACAAGGATCTGCC3' |
| | rs4343 | 5'CCCTTACAAGCAGAGGTGAGCTAA 3'(DNA); 5'ACCACCTACAGCGTGGCC3' (cDNA) | 5'CATGCCCATAACAGGTCTTCATATT3' | 5'GACGAATGTGATGGCCAC3' |
| MCP1 (CCL2) | rs4586 | 5'ATGCAATCAATGCCCCAGTC3' | 5'GCGAGCCTCTGCACTGAGAT3' | 5'AGATCTTCTATTGGTGAAGTTATA3' |
| | rs13900 | 5'CAACCCAAGAATCTGCAGCTAA3' | 5'GGCATAATGTTTCACATCAACAAAC3' | 5'TAGCTTTCCCCAGACACC3' |
| SOD2 | rs4880 | 5'GGTTGTTACAGTAGGCCG3' | 5'CAGCAGGCAGCTGGCT3' | 5'GAGCCCAGATACCCCAAA3' |
| | rs5746092 | 5'TTGCGGCGCAGCTGG3' | 5'CTGAAGCCGCTGCCGAA3' | 5'GGGCCTTAAGAAAGCGC3' |
| NNMT | rs4646335 | 5'GTCCTGTCTCTGAACTTTGGG3' | 5'GAGCTGTATGCAATGCTTGCC3' | 5'ATTGTAGACCAGAGGGAGCACT3' |
| LPL | rs1059611 | 5'TAAAGCAGCACATAGCACTGG3' | 5'GCAGATAGCCACAATGACCTT3' | 5'CCTTTCCAATATGTACAAGCTCC3' |
| HMGCR | rs12916 | 5'GCAAATATAAGCTGGGAAAAAGTTT3' | 5'AATTAAC TACAAAATCAGGAGTTTCATCAG 3' | 5'AAATCCATTTTCAACTGGCAGG3' |
| CSF1 | rs333970 | 5'TTCCTCTCAGCATCTTCTCCAC3' | 5'GGCAGATGGATGGTCTGTC3' | 5'GCCGGCAGATGTAAGTGTAC3' |
| PTGDS | rs6926 | 5'GGGCTGAAGCTGGGATC3' | 5'CTGACTTGCTTCCGGAGTTT3' | 5'CTCCCCGCCAAAGCA3' |
| HIF1A | rs2057482 | 5'CATTCTTTTTTTGGACACTGGT3' | 5'CAAGTTTGTGCAGTATTGTAGCCA3' | 5'ATGTAGAAAATATAAATAGACTGCTTTA GGTA3' |
| NOS3 | rs1799983 | 5'GAAACGGTCGCTTCGACGT3' | 5'GGCAGAAGGAAGAGTTCTGGG3' | 5'AGTAACCTTGAACCTTGGTGCAGGCC CCAGATGA3' |
| FLT1 | rs2296189 | 5'AATACTCCGTAAGACCACAGTC3' | 5'ACTCGACTTCTCTGAAATGGA3' | 5'GCTGTAGATTTTGTCAAAGATAGATTC3' |
| CACNA1C | rs1544514 | 5'GAACGAGTGAATATCTCTTTCTCATAA3' | 5'GCGGAGGTAGGCATTGGG3' | 5'CATAATTTTACGGTGAAGC3' |
| | rs216008 | 5'CCAGAGCTGCCTGTTCAAAAAT3' | 5'ATGAGCTTCAGGATCATCTCCAC3' | 5'TGCTCTTCACTGGCCTCTT3' |
| ADRB2 | rs1042719 | 5'TTCCAGGAGCTTCTGTGCCT3' | 5'GCCGTTGCTGGAGTAGCC3' | 5'TCTTCTTTGAAGGCCTATGG3' |
| KCNMB1 | rs11739136 | 5'CAGGAATCCAAGTGCCACC3' | 5'CCACAGGCATGGTACTGG3' | 5'CCAACATCAGGGACCAGGAG3' |
| | rs2656842 | 5'AAGTAGAGCCATCCATCCATGC3' | 5'GATTGGACTGGAAGAGTGGG3' | 5'CTGCTCCCCACTTGCAG3' |
| VKORC1 | rs7294 | 5'TTTTCTAACTCGCCCGCT3' | 5'TGGGTGTAAAAAGAGCGAGC3' | 5'CCTCTCCTGCCATACCC3' |
| GGCX | rs699664 | 5'GAGTGGCCTCGGAAGCTG3' | 5'GGAACACTGGGCTGAGGG3' | 5'GGTGTCTACTGCCCCC3' |
| CETP | rs5884 | 5'TCACCATGGGCATTTGATT3' | 5'CCACAGCGGT GATCATTGAC3' | 5'TGAGAGCAGCTCCGAGTCC3' |
| HMOX1 | Rs11555832 | 5'GGAGTTTTGAGACAGCTGCC3' | 5'CTGCAGCAGAGCCTGGAAG3' | 5'CTGCAGCAGAGCCTGGAAG3' |
| ABCB1 | rs1045642 | 5'CCTATGGAGACAACAGCCGG3' | 5'GGCATGTATGTTGGCCTCCT3' | 5'CTCCTTTGCTGCCCTCAC3' |
| | rs1128503 | 5'TTTCTCACTCGTCTGGTAGATCTT3' | 5'ACTGTTTCCAACCAGGGCC3' | 5'CTCTGCACCTTCAGGTTCAAG3' |
| | rs2032582 | 5'TTGTGAAATGAAATGTTGTCTGG3' | 5'CAATCATATTTAGTTTACTCACCTTCC3' | 5'TGAAAGATAAGAAAGAACTAGAAGGT3' |
| | rs2656841 | 5'AAGTAGAGCCATCCATCCATGC3' | 5'GATTGGACTGGAAGAGTGGG3' | 5'GCAGGTGGAGAAGGCATTG3' |
| CYP2D6 | rs769258 | 5'TGTGTCCAGAGGAGCCCAT3' | 5'GGCTCACCAGGAAAGCAAA3' | 5'ACCGCCCGCCTGTGCCCATCA3' |
| | rs1058164 | 5'TGTGTCCAGAGGAGCCCAT3' | 5'GGCTCACCAGGAAAGCAAA3' | 5'CGAGCAGAGGCGCTTCTCCGT3' |
| | rs16947 | 5'TGTGTCCAGAGGAGCCCAT3' | 5'GGCTCACCAGGAAAGCAAA3' | 5'AGCTTCAATGATGAGAACCTG3' |
| | rs1135840 | 5'GGCCAGCCACCATG3' | 5'GCACAGCACAAAGCTCATAGGG3' | 5'GTGCTTTGCTTTCTGCTGTA3' |
| CYP2C9 | rs9332242 | 5'GGATTTGTGTGGGAGAAGCC3' | 5'TGAAACATAGGAACTCTCC3' | 5'AATGCCTTTTCTACC3' |
| CYP2C9 | rs2017319 | 5'GGATTTGTGTGGGAGAAGCC3' | 5'TGAAACATAGGAACTCTCC3' | 5'GGCGGCACAGAGGCAAA3' |
| SLC15A1 | rs1339067 | 5'ACATTTCTTCTCCTGGATCACCA3' | 5'ACACTAGAAGCGTGTGGCGTT3' | F: 5'CTGGATCACCAGTCACTGC3' |

Running Title: Polymorphisms affecting RNA

| | | | | |
|---------|---------------------|---------------------------------|--------------------------------|--|
| | | | | R: 5'CTGCTTGAAGTCGTCAGTTAC3' |
| SLC15A2 | rs1143670 | 5'AGGAAAATGGCTGTTGGTATGATC3' | 5'CGCAACTGCAAATGCCAG3' | 5'GCTGTTGGTATGATCCTAGC3' |
| | rs1143671 | 5'GAAATGGCCCCAGCCC3' | 5'CATCTGCCAGATTCAAGACTTGTAG3' | 5'AACCTCCTGGGGACCTG3' |
| DRD2 | rs6277 | 5'CCAGCTGACTCTCCCCGAC3' | 5'GCATGCCATTCTTCTCTGG3' | 5'CGATCACATGTCGTGAACTGACTG ACTGGTTTGGCGGGGCTGTC3' |
| | rs6275 | 5'CCAGCTGACTCTCCCCGAC3' | 5'GCATGCCATTCTTCTCTGG3' | 5'GGAGTGCTGTGGAGACC3' |
| | rs6279 | 5'AGCCTGAGTCAGGGCCC3' | 5'ACCGCCTGCTCCACG3' | 5'CCCAGAGGCTGAGTTTTCT3' |
| CHRNA4 | rs1044393 | 5' TGAACATGCACAGCCGC3' | 5' CGAAGGCATAGGTGATGTCC3' | 5'TTGTAGGTGCCACGGC3' |
| | rs1044397 | 5' CACATGCAAGAAGGAGCCCT3' | 5' GGTGGTCTGCAATGTACTGGA3' | 5'CCGCAGCACAAAGC3' |
| | rs2236196 | 5' CCCTCTCCTAGCGAAGCAGAT3' | 5' GGTCTTGAGCCTCTCGGG3' | 5'CTAGCGAAGCAGATTGGAGC3' |
| OPRM1 | rs1799971 | 5'CCGTCAGTACCATGGACAGC3' | 5'GAGTACGCCAAGGCATCAGT3' | F: 5'ACTGATCGACTTGTCCACTTA GATGGC3' |
| | | | | R: 5'ACTGACTGACTGACCATGGGTC GGACAGGT3' |
| HTR2A | rs6313 | 5'GACACCAGGCTCTACAGTAATGACTTT3' | 5'TGTCCAGTTAAATGCATCAGAAGTG3' | 5'ACTGACTGACTGGAACCTTGACT CATCAGAAGTGTAGCTTCTCC3' |
| BDNF | rs6265 | 5'GCTTGACATCATTGGCTGACA3' | 5'CTGGTCCTCATCCAACAGCTC3' | 5'AACCTTGAACCTTGGGGCTGACT3' |
| SLC6A4 | rs1042173 | 5'TATCTGTTTGCTTCTAAAGGTTTC3' | 5'TGGACACACTATTTTTCATTTAG | 5'GGTTCTAGTAGATTCCAGCAATAAAATT 3' |
| TPH2 | rs7305115 | 5'ACGAGACTTTCTGGCAGGACTG3' | 5'TTAATTCTCCAATGGAGGAAAGGA3' | 5'GATCCCCTCTACACCCC3' |
| | rs4290270 | 5'ACGAGACTTTCTGGCAGGACTG3' | 5'TTAATTCTCCAATGGAGGAAAGGA3' | 5'AAAGGAGTCTGCTCCATA3' |
| SLC6A2 | rs5569 | 5'ATGGGAGGCATGGAGGCTGTC3' | 5'CGAGAAGGAAAGTGCTGAAGGTGAC3' | 5'GGCATGGAGGCTGTCATCAC3' |
| DTNBP1 | rs1047631 | 5'ATCCAGTTTTGGCTGTATGC3' | 5'CTGTTCTTTAAGTTTCTCACACATT3' | 5'TGTTTTATAGAGTTCTTGATTTTTAC3' |
| NRG1 | SNP8NRG433 E1006 | 5'CTGCTGCCACTACTGCTGCTGCT3' | 5'CACCTTCCCTCGATCACCA3' | 5'TAGCACACCGAGGCC3' |
| HTR1B | rs6296 | 5'CCACGTCCTCGGTCACCTCTATTAAC3' | 5'CACAATAAAGGCTCCCAAAATGATCC3' | 5'TCGAAATCCGGATCTCCTGTGTATGT3' |
| MAOA | rs1137070 | 5'AAATGGTCTCGGAAGGTGA3' | 5'TTTGATTCAGGTTCTGTACCCAG3' | 5'GGAAGGTGACCGAGAAAGA3' |
| | rs6323 | 5'ACTTCAGACCAGAGCTTCCAGC3' | 5'ATGCACTTAATGACAGCTCCA3' | 5'GAGAAACCAGTTAATTCAGCG3' |
| DRD3 | rs6280 | 5'TCTGCCCCACAGGTGTAGTTC3' | 5'GGCATCTCTGAGCCAGCTG3' | 5'ATCTCTGAGCCAGCTGAGT3' |
| ESR1 | rs3798577 | 5'TGGTGTTCATTTAGCCCTGG3' | 5'AGCCACAACAATCCTGCACA3' | 5'GGCATGGAGCTGAACAGTAC3' |
| SLC6A3 | rs6347 | 5'TTCATCATCTACCCGGAAGCC3' | 5'GAAGAAGACCACGGCCCAG3' | 5'ACGCTCCCTCTGTCTC3' |
| COMT | rs4633 | 5'GTGACACCAAGGAGCAGCG3' | 5'TGTCAATGGCCTCCAGCAC3' | 5'AGCGCATCCTGAACCA3' |
| DAO | rs2070588 | 5'GACGGGACTGATAACAGCAGC3' | 5'CACAAGCATCCATTATCAA3' | 5'CATCCAAGTCTCCCAACT3' |
| NR3C1 | rs6196 | 5'GGCAGTCACTTTTGTGAAACAGA3' | 5'GAGTATTGAATCCCCGAGATGTTAG3' | 5'CAATCAGATACCAAAATTTCAA3' |
| NQO2 | rs1143684 | 5'GCTGCACCGTCACAGTGTCT3' | 5'TGATATCTTTGTCTGTGGCCCTC3' | 5'GTCTGATTTGTATGCCATGAAC3' |