

U.S. Genome Variation Estimates

Table 2 - Genes Available, By Pathway

These candidate genes have a role in numerous cellular and physiologic pathways, as well as in multiple diseases and outcomes of public health importance. The pathways listed are not mutually exclusive. The inclusion of genes in certain pathways was based on information gathered from the [GeneCards database](#), the [KEGG GENES database](#), and selected publications, for ACE (1, 2), CAPN10 (3), and SERPINE1 (4-6).

The genes have a proposed or established association with the listed diseases and clinical outcomes, but the genetic associations are not limited to these diseases.

Gene symbol	Gene name	Chromosomal position of gene	Variant (dbSNP ID) [*]	Nucleotide change [†]	Amino acid change [†]	Cellular or physiological pathway(s) [‡]
<i>ABCB1</i>	ATP-binding cassette, sub-family B (MDR/TAP), member 1	7q21.1	rs1045642	Ex27-55T>C T3435C	Ile1145Ile	Xenobiotic metabolism
<i>ACE</i>	Angiotensin I converting enzyme (peptidyl-dipeptidase A) 1	17q23.3	rs4646994	289-bp Alu insertion/deletion in intron 16		Blood pressure regulation; Nutrient metabolism [§]
<i>ADH1B</i>	Alcohol dehydrogenase 1B (class I), beta polypeptide	4q21-q23	rs1229984	Ex3+23G>A	Arg48His	Nutrient metabolism; Xenobiotic metabolism
			rs17033	Ex9+77A>G		
			rs2066702	Ex9+5C>T	Arg370Cys	
<i>ADH1C</i>	Alcohol dehydrogenase 1C (class I), gamma polypeptide	4q21-q23	rs1693482	Ex6-14G>A	Arg272Gln	Nutrient metabolism; Xenobiotic metabolism
			rs698	Ex8-56A>G	Ile350Val	
<i>ADRB1</i>	Adrenergic, beta-1-, receptor	10q24-q26	rs1801252	Ex1+231A>G	Ser49Gly	Blood pressure regulation, Cardiac function; Nutrient metabolism
<i>ADRB2</i>	Adrenergic, beta-2-, receptor, surface	5q31-q32	rs1042713	Ex1+265G>A	Gly16Arg	Blood pressure regulation, Cardiac function; Nutrient metabolism
			rs1042714	Ex1+298G>C	Glu27Gln	
<i>ADRB3</i>	Adrenergic, beta-3-, receptor	8p12-p11.2	rs4994	Ex1+387T>C	Trp64Arg	Nutrient metabolism
<i>ALAD</i>	Aminolevulinate, delta-, dehydratase	9q33.1	rs1800435	Ex4+13G>C G177C	Lys68Asn Lys88Asn	Nutrient metabolism; Xenobiotic metabolism
<i>B9D2</i> [¶]	B9 protein domain 2	19q13.2	rs1800468	Ex4-262G>A -800G>A of TGFB1		Unknown
			rs1800469	308bp 3' of STP C>T -509C>T of TGFB1		

<i>CAPN10</i>	Calpain 10	2q37.3	rs3792267	IVS3-176G>A		Apoptosis; Nutrient metabolism
<i>CAT</i>	Catalase	11p13	rs769214	-843A>G		Metabolism of free radicals/Oxidative stress; Nutrient metabolism
<i>CBS</i>	Cystathionine-beta-synthase	21q22.3	no rs number	844ins68 (68-bp insertion in exon 8)		Nutrient metabolism
<i>CCL5</i>	Chemokine (C-C motif) ligand 5	17q11.2-q12	rs2280788	-95C>G		Cell migration/motility; Immunity and inflammation
<i>CCR2</i>	Chemokine (C-C motif) receptor 2	3p21.31	rs1799864	Ex2+241G>A	Val64Ile	Cell migration/motility; Immunity and inflammation
<i>CXCL12</i>	Chemokine (C-X-C motif) ligand 12 (stromal cell-derived factor 1)	10q11.1	rs169097	Ex5+709C>T		Cell migration/motility; Immunity and inflammation
<i>CYP1A1</i>	Cytochrome P450, family 1, subfamily A, polypeptide 1	15q22-q24	rs2472299	-17961C>T		Nutrient metabolism; Xenobiotic metabolism
			rs2606345	IVS1+606G>T		
<i>CYP1A2</i>	Cytochrome P450, family 1, subfamily A, polypeptide 2	15q24	rs11854147	5341bp 3' of STP C>T		Nutrient metabolism; Xenobiotic metabolism
			rs2069514	-3859G>A		
			rs4886406	9773bp 3' of STP T>G		
<i>CYP1B1</i>	Cytochrome P450, family 1, subfamily B, polypeptide 1	2p21	rs1056836	Ex3+251G>C	Val432Leu	Nutrient metabolism; Xenobiotic metabolism
			rs1056837	Ex3+304T>C	Asp449Asp	
			rs162557	-2919C>T		
<i>CYP2A6</i>	Cytochrome P450, family 2, subfamily A, polypeptide 6	19q13.2	rs1801272	Ex3-15T>A	Leu160His	Nutrient metabolism; Xenobiotic metabolism
<i>CYP2C19</i>	Cytochrome P450, family 2, subfamily C, polypeptide 19	10q24.1-q24.3	rs4986893	Ex4-7G>A	Trp212Stp	Nutrient metabolism; Xenobiotic metabolism
			rs4986894	-97T>C		
<i>CYP2C9</i>	Cytochrome P450, family 2, subfamily C, polypeptide 9	10q24	rs1057910	Ex7-75A>C	Ile359Leu	Nutrient metabolism; Xenobiotic metabolism
<i>CYP2E1</i>	Cytochrome P450, family 2, subfamily E, polypeptide 1	10q24.3-qter	rs2031920	-1054C>T -1053C>T		Nutrient metabolism; Xenobiotic metabolism
<i>CYP3A4</i>	Cytochrome P450, family 3, subfamily A, polypeptide 4	7q21.1	rs2740574	-391A>G		Nutrient metabolism; Xenobiotic metabolism
<i>F2</i>	Coagulation factor II (thrombin)	11p11-q12	rs1799963	Ex14-1G>A		Cell migration/motility; Hemostasis
<i>F5</i>	Coagulation factor V (proaccelerin, labile factor)	1q23	rs6025	Ex10-11G>A	Arg534Gln	Hemostasis
<i>FAM82A**</i>	Family with sequence similarity 82, member A	2p22.2	rs163086	IVS10-1363C>T		Unknown
<i>FCGR2A</i>	Fc fragment of IgG, low affinity IIa, receptor (CD32)	1q23	rs1801274	Ex4-120A>G	His166Arg His167Arg	Immunity and inflammation

<i>FGB</i>	Fibrinogen beta chain	4q28	rs1800790	-462G>A		Cellular adhesion; Hemostasis
<i>IL10</i>	Interleukin 10	1q31-q32	rs1800871	-853C>T -819C>T		Apoptosis, Cell cycle, Cellular growth and differentiation; Immunity and inflammation
			rs1800872	-626C>A -592C>A		
			rs1800896	-1116A>G -1082A>G		
<i>IL1B</i>	Interleukin 1, beta	2q14	rs1143623	-2022G>C		Apoptosis, Cellular growth and differentiation; Immunity and inflammation
<i>IL4</i>	Interleukin 4	5q31.1	rs2243248	-1098T>G		Apoptosis, Cellular growth and differentiation; Immunity and inflammation
			rs2243250	-588C>T -524C>T -590C>T		
			rs2243270	IVS2-1297A>G		
<i>IL4R</i>	Interleukin 4 receptor	16p11.2-12.1	rs1801275	Ex12+828A>G	Gln576Arg	Apoptosis, Cellular growth and differentiation; Immunity and inflammation
			rs1805015	Ex12+608T>C	Ser503Pro	
<i>ITGA2</i>	Integrin, alpha 2 (CD49B, alpha 2 subunit of VLA-2 receptor)	5q23-q31	rs1126643	Ex7-21C>T	Phe253Phe	Cellular adhesion, Cell migration/motility; Hemostasis
<i>ITGB3</i>	Integrin, beta 3 (platelet glycoprotein IIIa, antigen CD61)	17q21.32	rs5918	Ex3+11T>C	Leu59Pro	Apoptosis; Cellular adhesion, Cell migration/motility; Hemostasis
<i>MBL2</i>	Mannose-binding lectin (protein C) 2, soluble (opsonic defect)	10q11.2-q21	rs11003125	-618G>C -550G>C		Immunity and inflammation
			rs1800450	Ex1-27G>A	Gly54Asp	
			rs1800451	Ex1-18G>A	Gly57Glu	
			rs5030737	Ex1-34C>T	Arg52Cys	
			rs7096206	-289G<C -221G>C		
<i>MTHFR</i>	5,10-methylenetetrahydrofolate reductase (NADPH)	1p36.3	rs1801131	Ex8-62A>C 1298A>C	Glu429Ala	Nutrient metabolism
			rs1801133	Ex5+79C>T 677C>T	Ala222Val	
			rs2066470	Ex2-120C>T	Pro39Pro	
<i>MTRR</i>	5-methyltetrahydrofolate-homocysteine methyltransferase reductase	5p15.3-p15.2	rs1801394	Ex2-64A>G	Ile22Met Ile49Met	Nutrient metabolism
	N-acetyltransferase 2 (arylamine		rs1041983	Ex2+288C>T	Tyr94Tyr	
			rs1208	Ex2-367A>G	Lys268Arg	

NAT2	N-acetyltransferase)	8p22	rs1799930	Ex2-580G>A	Arg197Gln	Xenobiotic metabolism
			rs1801279	Ex2+197G>A	Arg64Gln	
			rs1801280	Ex2+347T>C	Ile114Thr	
NOS2A	Nitric oxide synthase 2A (inducible, hepatocytes)	17q11.2-q12	rs1800482	G>C in promoter		Blood pressure regulation; Immunity and inflammation; Metabolism of free radicals/Oxidative stress; Nutrient metabolism
			rs9282799	-2892C>T -1173C>T		
NOS3	Nitric oxide synthase 3 (endothelial cell)	7q36	rs1799983	Ex8-63G>T	Glu298Asp	Blood pressure regulation, Cardiac function; Hemostasis; Metabolism of free radicals/Oxidative stress; Nutrient metabolism
			rs2070744	IVS1-762T>C -786T>C		
NQO1	NAD(P)H dehydrogenase, quinone 1	16q22.1	rs10517	Ex6-457C>T		Nutrient metabolism; Xenobiotic metabolism
			rs1800566	Ex6+40C>T 609C>T	Pro187Ser Pro149Ser Pro153Ser	
			rs34755915	IVS3+20G>A		
			rs689452	IVS1-27C>G		
			rs689453	Ex2+65G>A	Glu24Glu	
OGG1	8-oxoguanine DNA glycosylase	3p26.2	rs1052133	Ex6-315C>G	Ser326Cys Pro332Ala	DNA Repair
PON1	Paraoxonase 1	7q21.3	rs662	Ex6+78A>G	Gln192Arg	Metabolism of free radicals/Oxidative stress; Xenobiotic metabolism
			rs854560	Ex3+18T>A	Leu55Met	
PPARG	Peroxisome proliferator-activated receptor gamma	3p25	rs1801282	Ex4-49C>G	Pro12Ala	Cellular growth and differentiation; Immunity and inflammation; Nutrient metabolism
SERPINE1	Serpin peptidase inhibitor, clade E (nexin, plasminogen activator inhibitor type 1), member 1	7q21.3-q22	rs1799762 rs1799768 rs1799889	4G/5G insertion/deletion in promoter		Cellular adhesion, Cell migration/motility; Hemostasis; Nutrient metabolism
TGFB1	Transforming growth factor, beta 1	19q13.1	rs1982073	Ex1-327C>T	Pro10Leu	Apoptosis, Cell cycle, Cellular growth and differentiation; Immunity and inflammation
TLR4	Toll-like receptor 4	9q32-q33	rs4986790	Ex4+636A>G	Asp299Gly	Immunity and inflammation
TNF	Tumor necrosis factor (TNF superfamily, member 2)	6p21.3	rs1800629	-487G>A -308G>A		Apoptosis, Cell cycle, Cellular growth and differentiation; Immunity and inflammation; Nutrient metabolism
			rs1800750	-555G>A		
			rs361525	-417G>A -238G>A		
	Vitamin D (1,25-		rs2239185	IVS8-3968C>T		Cellular growth and differentiation; Immunity and inflammation; Nutrient

VDR	dihydroxyvitamin D3) receptor	12q13.11	rs731236	Ex11+32T>C	Ile352Ile (TaqI variant)	metabolism
XRCC1	X-ray repair complementing defective repair in Chinese hamster cells 1	19q13.2	rs1001581	IVS2-216G>A		DNA Repair
			rs1799782	Ex6-22C>T	Arg194Trp	
			rs25486	IVS9-59A>G		
			rs25487	Ex10-4G>A	Arg399Gln	
			rs25489	Ex9+16G>A	Arg280His	

HUGO Gene Nomenclature Committee. All chromosomal positions are from Entrez Gene.* Unique identifier in the Entrez SNP (single nucleotide polymorphism) database at NCBI.

† Nucleotide and amino acid change information are from the SNP500 Cancer database and dbSNP. Alternate designations and additional locus information are given, if available.

‡ Inclusion of genes in pathways is based on information gathered from the GeneCards database, the KEGG GENES database, and selected publications for *ACE* (1, 2), *CAPN10* (3), and *SERPINE1* (4-6).

§ Nutrient metabolism encompasses the intracellular and physiological metabolism of nucleotides, amino acids and proteins, carbohydrates (e.g., glucose), lipids, cofactors, vitamins, and hormones (e.g., estrogen).

¶ *B9D2* is a protein that lies partially within the *TGFB1* promoter. Although within *B9D2*, these two SNPs are believed to be promoter polymorphisms of *TGFB1*.

** *FAM82A* is a hypothetical protein that lies immediately 5' of *CYP1B1*. This variant may be an intronic polymorphism of *FAM82A* or a *CYP1B1* variant that lies 3' of the gene

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