Coordinates (allele) ***Gene, amino acid***	**Associated trait**
chr1:25589952(C) *RHCE, 226A*	RH E/e polymorphism
chr1:98121473(G) ***DPYD, 29R***	**Dihydropyrimidine dehydrogenase deficiency**
chr1:114179091(A) *PTPN22, 620W*	Diabetes mellitus, insulin-dependent, susceptibility to Hashimoto thyroiditis, susceptibility to, included; rheumatoid arthritis, susceptibility to, included; systemic lupus erythematosus, susceptibility to, included
~~chr1:154375026(G[A])~~ ~~*LMNA, 608G*~~	~~Hutchinson-Gilford progeria syndrome~~ ~~Restrictive dermopathy, lethal, included~~
chr1:155115542(C) ***NTRK1, 604H***	**Thyroid carcinoma, familial medullary**
chr1:155115570(G) ***NTRK1, 613G***	**Thyroid carcinoma, familial medullary**
chr1:194908856(G) *CFH, 62V*	Macular degeneration, age-related, 4, susceptibility to
chr1:194925860(C) *CFH, 402H*	Macular degeneration, age-related, 4, susceptibility to Basal laminar drusen, included; myocardial infarction, susceptibility to, included
~~chr2:38151842(C[T])~~ ~~*CYP1B1, 387E*~~*[K]*	~~Glaucoma 3, primary congenital, A~~
chr2:49043425(C) *FSHR, 680S*	Ovarian response to FSH stimulation Ovarian hyperstimulation syndrome, moderator of severity of, included
chr2:49044545(C) *FSHR, 307A*	Ovarian response to FSH stimulation
chr2:215943291(C) *FN1, 1974R*	Glomerulopathy with fibronectin deposits 2 [susceptibility to?]
chr3:115373505(C) *DRD3, 9G*	Schizophrenia, susceptibility to Essential tremor, susceptibility to, included
chr5:33987450(C) *SLC45A2, 374L*	Skin/hair/eye pigmentation 5, black/nonblack hair Skin/hair/eye pigmentation 5, dark/fair skin, included; skin/hair/eye pigmentation 5, dark/light eyes, included
chr5:35896825(T) ***IL7R, 66I***	**Severe combined immunodeficiency, autosomal recessive, T cell-negative, B cell-positive, NK cell-positive**
chr5:35906947(G) ***IL7R, 138V***	**Severe combined immunodeficiency, autosomal recessive, T cell-negative, B cell-positive, NK cell-positive**
chr5:74017026(T) ***HEXB, 62L***	**Sandhoff disease, infantile type**
chr5:147461148(A) *SPINK5, 420K*	Atopy, susceptibility to Asthma, susceptibility to, included; dermatitis, atopic, 6, susceptibility to, included
chr5:148186633(G) *ADRB2, 16G*	Obesity, susceptibility to, included Asthma, nocturnal, susceptibility to

Coordinates (allele) ***Gene, amino acid***	**Associated trait**
chr5:148186666(G) *ADRB2, 27E*	Obesity, susceptibility to Asthma, childhood, susceptibility to, included
chr6:32904663(A) *TAP2, 687**	Peptide transporter PSF2 polymorphism
chr6:32904729(T) *TAP2, 665T*	Peptide transporter PSF2 polymorphism
chr6:32908390(C) *TAP2, 379V*	Peptide transporter PSF2 polymorphism
chr6:38758606(T) *GLO1, 111E*	Autism, susceptibility to
chr6:46780902(A) *PLA2G7, 379V*	Asthma and atopy, susceptibility to
chr6:112489016(C) ***WISP3, 78R***	**Arthropathy, progressive pseudorheumatoid, of childhood**
chr6:149763383(G) *SUMO4, 55V*	Diabetes mellitus, insulin-dependent, 5 [susceptibility to]
chr6:160033862(A) *SOD2, 16V*	Superoxide dismutase 2 polymorphism
chr7:94872711(G) *PON2, 311S*	Paraoxonase 2 polymorphism
chr7:116986769(G) *CFTR, 470V*	CFTR polymorphism
chr7:122422409(A) *TAS2R16, 172N*	Alcohol dependence
chr7:141319174(G) *TAS2R38, 262A*	Phenylthiocarbamide tasting
chr7:150327044(T) *NOS3, 298D*	Coronary spasm, susceptibility to Alzheimer disease, late-onset, susceptibility to, included; hypertension, pregnancy-induced, susceptibility to, included; hypertension, resistant to conventional therapy, included; ischemic heart disease, susceptibility to, included; ischemic stroke, susceptibility to, included
chr9:135304807(G) ***ADAMTS13, 951G***	**Thrombotic thrombocytopenic purpura, congenital**
chr10:64085190(A) *ZNF365, 62T*	Uric acid nephrolithiasis, susceptibility to
chr10:75343107(T) *PLAU, 141L*	Alzheimer disease, late-onset, susceptibility to
chr10:115795046(G) *ADRB1, 389G*	Beta-1-adrenergic receptor polymorphism, gain-of-function Congestive heart failure, susceptibility to
chr10:115795047(G) *ADRB1, 389G*	Beta-1-adrenergic receptor polymorphism, gain-of-function Congestive heart failure, susceptibility to

Coordinates (allele) *Gene, amino acid*	Associated trait
chr11:5203410(A) *HBB, 146Y*	Hemoglobin Bologna-St. Orsola
chr11:5203412(A) *HBB, 146Y*	Hemoglobin Bologna-St. Orsola
chr11:5203453(T) *HBB, 132Q*	Hemoglobin K (Woolwich)
chr11:5203454(G) *HBB, 132Q*	Hemoglobin K (Woolwich)
chr11:5203485(T) *HBB, 121K*	Hemoglobin Egypt Hemoglobin O (Arab)
chr11:5203486(T) *HBB, 121K*	Hemoglobin Egypt Hemoglobin O (Arab)
chr11:5203487(T) *HBB, 121K*	Hemoglobin Egypt Hemoglobin O (Arab)
chr11:5204402(A) *HBB, 99V*	Hemoglobin Chemilly
chr11:5204408(A) *HBB, 97L*	Hemoglobin Wood
chr11:5204409(G) *HBB, 97L*	Hemoglobin Wood
chr11:5204415(C) *HBB, 95D*	Hemoglobin N, beta type
chr11:5204478(C) *HBB, 74D*	Hemoglobin Shepherds Bush
chr11:5204558(C) *HBB, 47G*	Hemoglobin Gavello
chr11:5204559(C) *HBB, 47G*	Hemoglobin Gavello
chr11:5204564(G) *HBB, 45S*	Hemoglobin Cheverly
chr11:5204605(C) *HBB, 31R*	Hemoglobin Hakkari
chr11:5204736(C) *HBB, 31R*	Hemoglobin Hakkari
chr11:5204737(T) *HBB, 31R*	Hemoglobin Hakkari
chr11:5204751(C) *HBB, 26G*	Hb Aubenas
chr11:5204752(C) *HBB, 26G*	Hb Aubenas

Coordinates (allele) ***Gene, amino acid***	**Associated trait**
chr11:5204757(A) *HBB, 24V*	Hemoglobin Savannah
chr11:5204758(C) *HBB, 24V*	Hemoglobin Savannah
chr11:5204805(T) *HBB, 8E*	Hemoglobin N (Timone)
chr11:5231166(T) *HBG2, 121K*	Hemoglobin F (Carlton)
chr11:17366148(T) *KCNJ11, 23K*	Diabetes mellitus, noninsulin-dependent, susceptibility to
chr11:17487669(G) ***USH1C, 608P***	**Deafness, neurosensory, autosomal recessive 18**
chr11:66084671(T) *ACTN3, 577**	Actinin, alpha-3 polymorphism ACTN3 deficiency; sprinting performance
chr12:9123535(T) *A2M, 1000I*	Alpha-2-macroglobulin polymorphism Alzheimer disease, susceptibility to, included
chr12:14884706(C) *ART4, 265D*	Dombrock blood group
chr12:110368991(T) *SH2B3, 262W*	Diabetes mellitus, insulin-dependent, susceptibility to Celiac disease, susceptibility to, 13, included
chr12:111833253(G) *OAS1, 162G*	Diabetes mellitus, type 1, susceptibility to
chr12:119921765(A) *HNF1A, 574S*	Hepatic adenoma [susceptibility to?]
chr12:120779718(T) ***HPD, 33T***	**Hawkinsinuria**
chr14:20010446(G) *NP, 51G*	Nucleoside phosphorylase polymorphism
chr15:46213776(A) *SLC24A5, 111T*	Skin/hair/eye pigmentation 4, fair/dark skin
chr16:167318(C) *HBA1, 113H*	Hemoglobin Twin Peaks
chr16:167319(A) *HBA1, 113H*	Hemoglobin Twin Peaks
chr16:167322(T) *HBA1, 114L*	Hemoglobin Nouakchott
chr16:167328(C) *HBA1, 116A*	Hemoglobin Ube-4
chr16:3647748(G) *DNASE1, 244R*	Systemic lupus erythematosus, susceptibility to

Coordinates (allele) ***Gene, amino acid***	**Associated trait**
chr16:55106002(C) ***BBS2, 70S***	**Bardet-Biedl syndrome 2**
chr16:87240737(A) *CYBA, 72Y*	CYBA polymorphism 242C-T
chr17:35133111(A) *ERBB2, 654I*	ERBB2 polymorphism
chr17:35133114(A) *ERBB2, 655I*	ERBB2 polymorphism
chr18:9107867(T) *NDUFV2, 29V*	Parkinson disease, susceptibility to
chr19:6664262(G) *C3, 314P*	C3 polymorphism, HAV 4-1 plus/minus type
chr21:33536120(T) *IFNAR2, 8F*	Hepatitis B virus, susceptibility to
chr21:33562658(A) *IL10RB, 47K*	Hepatitis B virus, susceptibility to
chr22:17281004(C) *PRODH, 521R*	Hyperprolinemia, type I Schizophrenia, susceptibility to, 4, included
chr22:29341610(G) *TCN2, 259R*	TCN2 polymorphism