List of *human* genes screened entirely or partly by DHPLC (last updated December 15, 2003)

Gene	*MIM#*	*Disease*	*Reference*
1C7	109170	Immune response	Ribas et al. [2001]
ABCA4 (ABCR)	601691	Macular degeneration	Rivera et al. [2000] Gerth et al. [2002] Rudolph et al. [2002] Cremonesi et al. [2003b] Schmidt et al. [2003]
ABCB1	171050	Parkinson's disease	Furuno et al. [2002]
ABCG1	603076	Bipolar affective disorder	Kirov et al. [2001]
ABCG2 (BCRP)	603756	Drug efflux transporter	Backstrom et al. [2003]
ADRB2	109690	Asthma, chronic obstructive pulmonary disease, congestive heart failure	Yoshida et al. [2002] Lynch et al. [2002]
AGXT	259900	Hyperoxaluria type 1	Pirulli et al. [2001]
AHRR	606517	Micropenis	Fujita et al. [2002]
AIPL1	604392	Leber congenital amaurosis, retinitis pigmentosa	Hanein et al. [2002] Kaliq et al.[2003]
AKT1	164730	TypeII diabetes	Matsubara et al. [2001]
ALAD	125270	Lead poisoning	Niu et al. [2001]
ALAP	145500	Essential hypertension	Yamamoto et al. [2002]
ALG3	601110	Congenital disorder of glycosylation	Schollen et al. [2002]
ALG6	604566	Congenital disorder of glycosylation	Schollen et al. [2002]
ALS2	205100	Amyotrophic lateral sclerosis 2	Hadano et al. [2001]
APC	175100	Adenomatous polyposis, desmoid tumor	Wu et al. [2001] Eccles et al. [2001] Al-Tassan et al. [2002] Gavert et al. [2002] Jones et al. [2002] Smith et al. [2002] Young et al. [2002] Emmerson et al. [2003] Heinritz et al [2003] Mihalatos et al. [2003]
APCS	104770	Type 2 diabetes mellitus	Wolford et al. [2003]
APM1	605441	Type 2 diabetes	Vasseur et al. [2002]
APOD	107740	Lipid metabolism	Desai et a. [2002]
APOH	138700	Regulation of ß2-glycoprotein I expression	Mehdi et al. [2003]
AR	313700	Micropenis	Ishii et al. [2001] Muroya et al. [2001]
ARIX (PHOX2A)	602753	Congenital fibrosis of the extraocular muscles (CFEOM2)	Nakano et al. [2001]
ARPKD	263200	Recessive polycystic kidney disease	Furu et al. [2003]
ARX	300382	X-linked mental retardation	Bienvenu et al. [2002]
ASAH	228000	Prostate cancer	Seelan et al. [2000]

ATM	208900	Ataxia-telangiectasia Hereditary breast and ovarian cancer Sporadic breast cancer	Thorstenson et al. [2000, 2001, 2003] Teraoka et al. [2001] Atencio et al. [2001] Iannuzzi et al. [2002] Offit et al. [2002] Bernstein et al. 2003a, b] Bernstein et al. [2003] Mitui et al. [2003] Pause et al. [2003]
ATP1A2	182340	Familial hemiplegic migraine type 2	De Fusco et al. [2003]
ATP2A2	108740	Darier's disease	Jacobsen et al. [1999, 2001b] Jones et al. [2002]
ATP6V1B1	192132	Autosomal recessive distal renal tubular acidosis with hearing loss	Stover et al. [2002]
ATP6V0A4	605239	Autosomal recessive distal renal tubular acidosis with hearing loss	Stover et al. [2002]
ATP7B	606882	Wilson's disease	Weirich et al. [2002a]
ATRX	300032	Myelodysplastic syndrome	Steensma et al. [2003]
AXIN1	603816	Hepatocellular carcinoma, hepatoblastoma	Taniguchi et al. [2002a]
AXIN2	604025	Colorectal cancer, hepatocellular carcinoma, hepatoblastoma	Liu et al. [2000a] Taniguchi et al. [2002a]
BACE	604252	Alzheimer disease	Cruts et al. [2001]
BACH1	605882	Early-onset breast cancer	Cantor et al. [2001]
BAX	600040	Tumor progression and genomic instability	Bacon et al. [2001]
BCKDE1A	248600	Maple Syrup urine disease	Kessler et al. [2002]
BCL10	603517	Endometrial cancer	Cohn et al. [2000]
BHD	135150	Birt-Hogg-Dube syndrome	Nickerson et al. [2002]
BMPR1A	601299	Juvenile intestinal polyposis	Kim et al. [2003a]
BMPR2	600799	Familial primary pulmonary hypertension.	Deng et al. [2000] Humbert et al. [2002] Rindermann et al. [2003]
BRAF	164757	Melanoma	Meyer et al. [2003a]

BRCA1	113705	Hereditary breast and ovarian cancer	Wagner et al. [1998, 1999b] Arnold et al. [1999, 2002] Gross et al. [1999, 2000] Kiechle et al. [2000] Shiri-Sverdlov et al. [2000] Garcia-Closas et al. [2001] Moller et al. [2001] Tomka et al. [2001] Andrulis et al. [2002] Arnold et al. [2002] Kang et al. [2002] Meindl et al. [2002] Muhr et al. [2002] Pfeiffer et al. [2002] Sevilla et al. [2002] Deng et al. [2003b] Ginolhac et al. [2003] Meyer et al. [2003b] Rajkumar et al. [2003]
BRCA2	600185	Hereditary breast and ovarian cancer	Wagner et al. [1999a, 1999b] Gross et al. [2000] Shiri-Sverdlov et al. [2000] Garcia-Closas et al. [2001] Kang et al. [2002] Meindl et al. [2002] Muhr et al. [2002] Real et al. [2002] Boettger et al. [2003] Meyer et al. [2003b] Rajkumar et al. [2003]
BRUNOL3	602538	Heart and thymus developmental defects	Lichtner et al. [2002]
C4B	120820	Immunodeficiency	Jaatinen et al. [2003]
CACNA1A (CACNL1A4)	601011	Familial hemiplegic migraine (FHM) and episodic ataxia type-2 (EA-2)	Ophoff et al. [1996]
CAPN-3	114240	Calpainopathy	De Paula et al. [2002]
CASR	601199	Sporadic isolated hypoparathyroidism	Hendy et al. [2003]
CAST	114090	Alzheimer disease	Nakayama et al. [2002]
CCM1	604214	Familial cerebral cavernous malformation	Dupre et al. [2003]
CCND1	168461	Nasopharyngeal carcinoma	Deng et al. [2002]
CCR5	601373	AIDS	Martin et al. [1998]
CD2BP1	604416	PAPA syndrome, familial recurrent arthritis	Wise et al. [2002]
CDH1	192090	Hereditary prostate cancer; somatic mutations in ductal and lobular breast cancer, gastric cancer	Salahshor et al. [2001] Jonsson et al. [2002] Lei et al. [2002] Fricke et al. [2003]
CEA	109770	Colorectal cancer	Zimmer and Thomas [2001]

CFTR (ABCC7)	602421	Cystic fibrosis, idiopathic chronic pancreatitis	Liu et al. [1998] Jones et al. [1999] Le Marechal et al. [2001a] Audrézet et al. [2002] Elahi et al. [2002] Ferec et al. [2002] Ravnik-Glavac et al. [2002] Scotet et al. [2002] Cremonesi et al. [2003b] Girardet et al [2003]
CHAC	200150	Chorea-acanthcytosis	Dobson-Stone et al. [2002]
CHAT	118490	Late-onset Alzheimer's disease	Harold et al. [2003]
CHEK2	114480	Breast cancer, prostate cancer	Dong et al. [2003] Offit et al. [2003] Rajkumar et al. [2003]
CHRNA3	118503	Megacystis-microcolon-hypoperistalsis syndrome (MMIHS)	Lev-Lehman et al. [2001]
CHRNA4	118504	Attention deficit/hyperactivity disorder	Todd et al. [2003]
CHRNB4	118509	Megacystis-microcolon-hypoperistalsis syndrome (MMIHS)	Lev-Lehman et al. [2001]
CHX10	142993	Microphthalmia, anophthalmia, coloboma	Morrison et al. [2002]
CLCN1	118425	Myotonia	Wu et al. [2002]
CLIC5	254770	Myoclonic epilepsy	Suzuki et al. [2002]
CLN2	204500	Late-infantile neuronal ceroid lipofuscinosis	Lam et al. [2001b]
CNOT7	604913	Colorectal cancer	Flanagan et al. [2003]
COL17A1	113811	Epidermolysis bullosa	Pfendner et al. [2003]
COL3A1	120180	Ehlers-Danlos syndrome type IV	Giunta and Steinmann [2000]
COL4A3	120070	Alport syndrome	Van Der Loop et al. [2000]
COL7A1	120120	Epidermolysis bullosa	Pfendner et al. [2003]
COPG2	604355	Autism	Bonora et al. [2002]
CPA1	114850	Autism	Bonora et al. [2002]
CPA5	209850	Autism	Bonora et al. [2002]
CRB1	604210	Leber congenital amaurosis, retinitis pigmentosa	Hanein et al. [2002] Kaliq et a. [2003]
CRP	123260	Type 2 diabetes mellitus	Wolford et al. [2003]
CRX	602225	Leber congenital amaurosis	Hanein et al. [2002]
CTNNB1	116806	Uveal melanoma, hepatocellular carcinoma, hepatoblastoma	Edmunds et al. [2002] Taniguchi et al. [2002a]
CUX2	NA	Bipolar disorder	Jacobsen et al. [2001a]
CYP2A6	122720	Drug metabolism	Pitarque et al. [2001]
CYP2B6	605059	Drug metabolism	Zanger et al. [2002]
CX26	121011	Sensorineural deafness	Pallares-Ruiz et al. [2001]
DBC2		Breast cancer	Hamaguchi et al. [2002]
DBY	400010	Male infertility and evolution	Sun et al. [1999] Shen et al. [2000]
DDC (AADC)	107930	Schizophrenia and bipolar disorder	Speight et al. [2000]
DGSC	192430	Schizophrenia	Williams et al. [2002b]
DGS1	601755	Schizophrenia	Williams et al. [2002b]
DISC1	605210	Schizophrenia	Devon et al. [2001a]
DISC2	606271	Schizophrenia	Devon et al. [2001a]

DMD	310200	Duchenne Muscular Dystrophy	Bennett et al. [2001] Tuffery-Giraud et al. [2003]
DNMT3B	602900	De novo DNA methylation	Okano et al. [1999]
DPM1	603503	Congenital disorder of glycosylation	Schollen et al. [2002]
DPYD	274270	5-fluorouracil toxicity	Ezzeldin et al. [2002] Fischer et al. [2003] Gross et al. [2002, 2003]
DRD2	126450	Attention-deficit hyperactivity disorder (ADHD)	Todd & Lobos [2002]
DRD3	126451	Schizophrenia	Anney et al. [2002]
EGR2	129010	Charcot-Marie-Tooth disease	Takashima et al. [2000, 2001]
ELA2 (NE)	130130	Lung cancer	Taniguchi et al. [2002b]
ELN	130160	Intracranial aneurysm	Hofer et al. [2003]
ELOVL4	605512	Stargardt-like macular dystrophy and other macular dystrophy phenotypes	Ayyagari et al. [2001] Bernstein et al. [2001] Zhang et al. [2001]
ESR	133430	Prostate cancer	Cancel-Tassin et al. [2003]
ESR2	601663	Isoforms hERß530 and hERß549	Xu et al. [2003]
EXT1 *EXT2*	133700 133701	Multiple exostoses	Dobson-Stone et al. [2000] Li et al. [2002]
F11 (Factor XI)	264900	Factor XI deficiency	Mitchell et al. [2003]
F8C (Factor VIII)	306700	Hemophilia A	Oldenburg et al. [2001] Frusconi et al. [2002] Bogdanova et al. [2002] Bicocchi et al.[2003]
F9 (Factor IX)	306900	Hemophilia B	Castaldo et al. [2003]
FAM10A4	606796	B-cell chronic lymphocytic leukemia (BCLL)	Sossey-Alaoui et al. [2002a]
FANCA	227650	Fanconi anemia group A	Rischewski and Schneppenheim [2001]
FBLN1	135820	Vitreoretinal dystrophy	Weigell-Weber et al. [2003]
FBLN5	604580	Inherited cutis laxa	Markova et al. [2003]
FBN1	134797	Marfan syndrome and related connective tissue disorders	Liu et al. [1997c] Schrijver et al. [1999, 2002a, 2002b] Halliday et al. [2002] Matyas et al. [2002]
FGFR2	176943	Syndromic craniosynostosis	Kan et al. [2002]
FGFR3	134934	Skeletal dysplasia	Hyland et al. [2003]
FHIT	601153	Peutz-Jeghers syndrome	Zhao et al. [2003]
FLNA	300017	Diverse congenital malformations	Robertson et al [2003]
FLT3	136351	Acute myeloid leukemia	Bianchini et al. [2003]
FOXP2	602081	Autism and specific language impairment	Newbury et al. [2002]
FRAXA	309550	Fragile X syndrome	Brightwell et al. [2002]
FRAXE	309548	Fragile X syndrome	Brightwell et al. [2002]
FTH1	134770	Hyperferritinemia	Cremonesi et al. [2003a]
FTL	134790	Hereditary hyperferritinaemia cataract syndrome	Cremonesi et al. [2003b, 2003c]
G6PT1	602671	Glycogen storage disease 1 non-A	Lam et al. [2000a] Santer et al. [2000]

GABRG2	137164	Myoclonic epilepsy	Gennaro et al. [2003] Madia et al. [2003] Nakayama et al. [2003]
GAD2	138275	Type 1 diabetes	Johnson et al. [2002]
GCK	138079	Type II diabetes mellitus, sudden infant death syndrome, Maturity Onset Diabetes of the Young (MODY)	Boutin et al. [2001] Burchell et al. [2002] Pruhova et al. [2003]
GCLC	254770	Myoclonic epilepsy	Suzuki et al. [2002]
γ-GCS	NA	Lung cancer	Yang et al. [2002]
GFAP	137780	Alexander disease	Gorospe et al. [2002]
GJB1	304040	Charcot-Marie-Tooth disease	Takashima et al. [2000, 2001]
GJB2	121011	Hereditary hearing loss	Kelsell et al. [2000] Weigell-Weber et al. [2000] Lin et al. [2001] Rickard et al. [2001] Pallares-Ruiz et al. [2002] Gurtler et al. [2003]
GJB3	603324	Erythrokeratoderma variabilis, hereditary hearing loss	Kelsell et al. [2000] Wilgoss et al. [1999] Mhatre et al. [2003b]
GJB4	605425	Erythrokeratoderma variabilis	Richard et al. [2003]
GNA11	139313	Congestive heart failure	Lynch et al. [2002]
GNAQ	600998	Congestive heart failure	Lynch et al. [2002]
GNAS	139320	Congestive heart failure, Albright hereditary osteodystrophy	Lynch et al. [2002] Rickard & Wilson [2003]
GPR75	NA	Macular degeneration	Sauer et al. [2001]
GREAT	219050	Cryptorchidism	Gorlov et al. [2002]
GRIN1	138249	Schizophrenia	Williams et al. [2002a]
GRIN2A	138253	Schizophrenia	Williams et al. [2002a
GRIN2B	138252	Schizophrenia	Williams et al. [2002a]
GRIN2C	138254	Schizophrenia	Williams et al. [2002a]
GRIN2D	602717	Schizophrenia	Williams et al. [2002a]
GRM5	604102	Schizophrenia	Devon et al. [2001b]
GSC	138890	Hemifacial microsomia	Kelberman et al. [2001]
GSC1	192430	Schizophrenia	Williams et al. [2002b]
GSTP1	134660	Lung cancer	Yang et al. [2002]
GSTM1	138350	Lung cancer	Yang et al. [2002]
GSTT1	600436	Lung cancer	Yang et al. [2002]
GUCY2D	600179	Leber congenital amaurosis, retinitis pigmentosa	Hanein et al. [2002] Khaliq et al. [2003]
GZMB	123910	Apoptosis	McIlroy et al. [2003]
HBB	141900	ß-Thalassemia	Colosimo et al. [2002a] Webster et al. [2002] Su et al. [2003a] Wu et al. [2003] Yip et al. [2003]
HFE	235200	Hereditary hemochromatosis	Le Gac et al. [2001] Liang et al. [2001] Pissard et al. [2002] Toomajian&Kreitman [2002] Fruchon et al. [2003]

HLA-A	142800	Transplantation	Etokebe et al. [2003]
HMBS	176000	Acute intermittent porphyria	Lam et al. [2001c]
HPRP3	601850	Autosomal dominant retinitis pigmentosa	Chakarova et al. [2002]
HRAS1	190020	Congestive heart failure	Lynch et al. [2002]
HSPA6 (HSP70B')	140555	Prostate cancer	Hecker et al. [2000]
HSPCAL4	140576	Autoimmune disease	Passarino et al. [2003]
HSPCB	140572	Autoimmune disease	Passarino et al. [2003]
HUT2	601611	Hypertension	Ranade et al. [2001]
HYMAI	606546	Transient neonatal diabetes mellitus	Mackay et al. [2002]
IFNGR1	107470	Mycobacterium avium-intracellulare pulmonary disease	Huang et al. [1998]
IKBKAP	603722	Familial dysautonomia	Lehavi et al. [2003]
IL4	147780	Asthma	Kabesch et al. [2003]
IL8	146930	Susceptibility to viral bronchiolitis	Hull et al. [2001]
IL10	124092	Immunity	D'Alfonso et al. [2000, 2002a]
IL13	147683	IL-13 SNPs and total serum IgE levels	Graves et al. [2000]
INK4A	600160	Melanoma	Orlow et al. [2001]
INNSR	147671	Type-2 diabetes mellitus	Wolford et al. [2001b]
ITGA6	147556	Epidermolysis bullosa	Pfendner et al. [2003]
ITGB4	147557	Epidermolysis bullosa	Pfendner et al. [2003]
ITGB7	147559	Inflammatory bowel disease	Van Heel et al. [2001]
JAG1	118450	Alagille syndrome	Heritage et al. [2002]
JAM3	606871	Hypoplastic left heart	Phillips et al. [2002]
JRK	603210	Childhood absence epilepsy	Morita et al. [1999]
K9	144200	Epidermolytic palmoplantar keratoderma	Rugg et al. [2002]
KCNE1	176261	Congenital long QT syndrome (LQTS)	Jongbloed et al. [2002]
KCNE2	603796	Congenital long QT syndrome (LQTS)	Jongbloed et al. [2002]
KCNH2	152427	Congenital long QT syndrome (LQTS)	Jongbloed et al. [2002]
KCNJ10	602208	Type 2 diabetes	Farook et al. [2002]
KCNQ1	192500	Congenital long QT syndrome (LQTS)	Jongbloed et al. [2002]
KCNN3	602983	Schizophrenia	Bowen et al. [2001]
KIAA0057	254770	Myoclonic epilepsy	Suzuki et al. [2002]
KIT	606764	Gastrointestinal stromal tumors (GISTs)	Corless et al. [2002]
K-ras	601599	Colorectal cancer	Smith et al. [2002] Hunt et al. [2002]
KRT1	139350	Palmoplantar keratoderma	Terron-Kwiatkowski et al. [2002]
KvLQT1	220400	Jervell and Lange-Nielson syndrome (JLSN1)	Wang et al. [2002b]
LAMA2	156225	Congenital muscular dystrophy	Tezak et al. [2003]
LAMA3	600805	Epidermolysis bullosa	Pfendner et al. [2003]
LAMB3	150310	Epidermolysis bullosa	Pfendner et al. [2003]
LAMC2	150292	Epidermolysis bullosa	Pfendner et al. [2003]
LDLR	143890	Familial hypercholesterolaemia	Bunn et al. [2002] Bodamer et al. [2002]
LHCGR	152790	Leydig cell hypoplasia	Richter-Unruh et al. [2002]
LIPC	151670	Coronary artery disease	Su et al. [2002b, 2003b]

LMNA	150330	Familial partial lipodystrophy, Type II diabetes mellitus, autosomal dominant dilated cardiomyopathy	Speckman et al. [2000] Wolford et al. [2001a] Arbustini et al. [2002] Spinarova et al. [2003] Taylor et al. [2003]
LPL	238600	Coronary atherosclerotic heart disease	Su et al. [2000, 2002b]
LRP5	603506	Diabetes mellitus	Twells et al. [2003]
LRRC1	254770	Myoclonic epilepsy	Suzuki et al. [2002]
MAG	159460	Multiple sclerosis	D'Alfonso et al. [2002b]
MAPK1	176948	Congestive heart failure	Lynch et al. [2002]
MAPT	157140	Pick's disease, primary progressive aphasia	Pickering-Brown et al. [2000] Sobrido et al. [2003]
MC4R	155541	Early-onset obesity	Miraglia Del Giudice et al. [2002]
MCCD1	NA	Polymorphism analysis	Semple et al. [2003]
MECP2	300005	Rett syndrome, infantile autism	Buyse et al. [2000] Lam et al. [2000b] Orrico et al. [2000] Girard et al. [2001] Hoffbuhr et al. [2001] Nicolao et al. [2001] Beyer et al. [2002] Yaron et al. [2002] Thistlethwaite et al. [2003]
MEN1	131100	Multiple endocrine neoplasia type 1	Park et al. [2003]
MEST	601029	Autism	Bonora et al. [2002]
MET	164860	Papillary renal carcinomas, familial gastric cancer	Nickerson et al. [2000, 2001] Lindor et al. [2001]
MIDI	300000	Opitz syndrome	Winter et al. [2003]
MIF	153620	Systemic-onset juvenile idiopathic arthritis	Donn et a. [2001, 2002]
MLC1 (KIAA0027)	605908	Megalencephalic leukoencephalopathy with subcortical cysts, schizophrenia	Bettecken et al. [2002] Devaney et al. [2002] Rubie et al. [2003]
MLH1	120436	Hereditary nonpolyposis colorectal cancer, endometrial carcinoma	Pawar et al. [2000] Harvey et al. [2000] Holinski-Feder et al., [2001] Kurzawski et al. [2002] Baldinu et al. [2002] Young et al. [2002] Deng et al. [2003a] Isidro et al. [2003] Wang et al. [2003a, 2003b] Wei et al. [2003]
MLH3	604395	Colorectal cancer	Lipkin et al. [2001] Liu et al. [2003]
MMP2	120360	Lung cancer	Yu et al. [2002]
MPI	154550	Congenital disorder of glycosylation	Schollen et al. [2002]
MPDU1	604041	Congenital disorder of glycosylation	Schollen et al. [2002]
MPP4	606575	Retinitis pigmentosa	Conte et al. [2002]
MPZ	159440	Charcot-Marie-Tooth disease	Takashima et al. [2000, 2001]

MSH2	120435	Hereditary nonpolyposis colorectal cancer, endometrial carcinoma	Pawar et al. [2000] Harvey et al. [2000] Holinski-Feder et al. [2001] Kurzawski et al. [2002] Baldinu et al. [2002] Young et al. [2002] Foulkes et al. [2003] Isidro et al. [2003] Wang et al. [2003a. 2003b] Wei et al. [2003]
mtDNA		Mutations in various mitochondrial diseases	McAndrew et al. [2000] van den Bosch et al. [2000] Bayat et al. [2002] Christodoulou et al. [2002] Liu et al. [2002a] Walter et al. [2002] Conley et al. [2003] Danielson et al. [2003] LaBerge et al. [2003] Romano et al. [2003]
MTM1	310400	Myotubular myopathy	Flex et al. [2002] Biancalana et al. [2003]
MTMR2	603557	Charcot-Marie-Tooth disease type 4B.	Bolino et al. [2000, 2001] Nelis et al. [2002]
MUTYH	604933	Hereditary nonpolyposis colon cancer	Al-Tassan et al. [2002] Jones et al. [2002]
MYH7	160760	Familial hypertrophic cardiomyopathy (HCM)	Blair et al. [2002] Ackerman et al. [2002] Van Driest et al. [2002a]
MYH9	160775	MYHIIA syndrome, hereditary macro-thrombocytopenia and progressive deafness	Heath et al. [2001] Mhatre et al. [2003a]
MYOC	601652	Primary open angle glaucoma	Cobb et al. [2002] Challa et al. [2002] Jansson et al. [2003] Melki et al. [2003a, 2003b]
MYO18B		Lung cancer	Nishioka et al. [2002]
NAB1	600800	Peripheral neuropathy	Venken et al. [2002]
NAB2	602381	Peripheral neuropathy	Venken et al. [2002]
NBS1	602667	Acute lymphoblastic leukemia (ALL), colorectal carcinoma	Varon et al. [2001, 2002, 2003]
NCSTN	605254	Alzheimer disease	Dermaut et al. [2002]
NDUFV1	161015	Mitochondrial complex I deficiency	Benit et al. [2001b]
NDUFV2	600532	Mitochondrial complex I deficiency, bipolar disorder	Benit et al. [2003] Washizuka et al. [2003]
NDUFS1	157655	Mitochondrial complex I deficiency	Benit et al. [2001b]
NEFL	162280	Charcot-Marie-Tooth disease	Jordanova et al. [2003]
NF1	162200	Neurofibromatosis type 1	O'Donovan et al. [1998] Han et al. [2001] De Luca et al. [2002] Luc et al. [2003] Upadhyaya et al. [2003]

NF2	607379	Neurofibromatosis type 2, meningioma	Szijan et al. [2003]
Notch3	600276	Subcortical ischemic strike and vascular dementia	Escary et al. [2000]
NPHS2	604766	Late-onset focal segmental glomerulosclerosis	Tsukaguchi et al. [2002]
NR3C1	138040	Atherosclerosis	Cotton and Bray, 2001 Ye et al. [2003]
NRAS	164790	Acute myeloid leukemia	Bowen et al. [2003]
NTF3	162660	Schizophrenia	Hattori et al. [2002]
NTS	162650	Schizophrenia	Austin et al. [2000b]
NTSR1	162651	Schizophrenia	Austin et al. [2000a]
OA1	300500	X-linked ocular albinism	Hegde et al. [2002]
OCRL	309000	Lowe syndrome	Lin et al. [2000]
OLR1	602601	Alzheimer disease	Luedecking-Zimmer et al. [2002]
OPRM1	600018	Heroin addiction, idiopathic generalized epilepsy	Shi et al. [2002] Wilkie et al. [2002]
OR2H3 (FAT11)	600578	HLA-linked olfactory receptor genes	Eklund et al. [2000]
OTX1	600036	Dyslexia	Francks et al. [2002]
P14 (ARF)	600160	Uveal melanoma	Edmunds et al. [2002]
P15 (INK4B)	600160	Uveal melanoma	Edmunds et al. [2002]
P16 (INK4A)	600160	Uveal melanoma	Edmunds et al. [2002]
PAH	261600	Phenylketonuria	Brautigam et al. [2003]
PAI-1	173360	Coronary artery disease	Wang et al. [2003c]
PAX6	106210	Microphthalmia, anophthalmia, coloboma	Malandrini et al. [2001] Morrison et al. [2002]
PBX1	176310	Diabetes mellitus type 2	Thameem et al. [2001]
PCDH8	603580	Schizophrenia	Bray et al. [2002]
PDGFRB	173410	Hypereosinophilic syndrome	Pardanani et al. [2003]
PKD1	601313	Autosomal dominant polycystic kidney disease	Mizoguchi et al. [2001] Rossetti et al. [2002]
PKD2	173910	Autosomal dominant polycystic kidney disease	Rossetti et al. [2002]
PKHD1	606702	Autosomal recessive polycystic kidney disease	Onuchic et al. [2002] Ward et al. [2002] Rossetti et al. [2003]
PKHDL1	263200	Autosomal recessive polycystic kidney disease	Hogan et al. [2003]
PLAGL1 (ZAC)	603044	Transient neonatal diabetes mellitus	Mackay et al. [2002]
PMM2	601785	Carbohydrate-deficient glycoprotein syndrome type 1A	Erlandson et al. [2000, 2001] Schollen et al. [2002]
PMP22	601097	Charcot-Marie-Tooth disease	Takashima et al. [2000, 2001]
PPARA	170998	Familial combined hyperlipidemia	Eurlings et al. [2002]
PRX	605725	Dejerine-Sottas neuropathy	Boerkoel et al. [2001]
PPOX	600923	Variegate porphyria	Whatley et al. [1999] Christiansen et al. [2001] Lam et al. [2001a]
PRKAG2	602743	Familial hypertrophic cardiomyopathy (HCM), sporadic Wolff-Parkinson-White symdrome	Blair et al. [2001] Vaughan et al. [2003]
PRKCSH	177060	Autosomal dominant polycystic liver disease	Li et al. [2003]
PRL	176760	Multiple sclerosis, systemic lupus erythematosus	Mellai et al. [2003]

PRLR	176761	Multiple sclerosis, systemic lupus erythematosus	Mellai et al. [2003]
PROC	176860	Thromboembolism	Taliani et al. [2001]
PRPF31	600138	Autosomal dominant retinitis pigmentosa	Vithana et al. [2001]
PRSS1	276000	Hereditary pancreatitis, idiopathic chronic pancreatitis	Le Marechal et al. [2001b] Audrézet et al. [2002]
PRSS16	607169	Autoimmunity	Lie et al. [2002]
PS1	104311	Alzheimer disease	Athan et al. [2001] Xu et al. [2002]
PTCH2 (Patched 2)	603673	Neuroblastoma, basal cell nevus syndrome	Jogi et al. [2000] Lam et al. [2002a]
PTEN/MMAC1	601728	Glioblastoma, endometrial carcinoma	Liu et al. [1997b, 1998] Marsh et al. [2001] Baldinu et al. [2002]
PTPN11	176876	Noonan syndrome, LEOPARD syndrome, cardiofaciocutaneous syndrome	Ion et al. [2002] Kosaki et al. [2002] Legius et al. [2002] Tartaglia et al. [2002, 2003] Musante et al. [2003] Schollen et al. [2003]
PTPRC	151460	Immunodeficiency	Stanton et al. [2003]
RAB3A	179490	Congestive heart failure	Lynch et al. [2002]
RAB4	179511	Congestive heart failure	Lynch et al. [2002]
RAB5C	604037	Congestive heart failure	Lynch et al. [2002]
RAD	179503	Congestive heart failure	Lynch et al. [2002]
RAGE (AGER)	600214	Diabetes mellitus type 2	Hudson et al. [2001]
RASSF1A	605082	Medulloblastoma	Lusher et al. [2002]
RESISTIN	605565	Polycystic ovary syndrome	Urbanek et al. [2003]
RET	164761	Multiple endocrine neoplasia type 2	Marsh et al. [2001]
RNASEL	180435	Prostate cancer	Rennert et al. [2002]
RPE65	180069	Leber congenital amaurosis	Hanein et al. [2002]
RPGR	312610	Retinitis pigmentosa	Koonekoop et al. [2003]
RPGRIP1	605446	Leber congenital amaurosis	Gerber et al. [2001] Hanein et al. [2002]
RYR1	180901	Malignant hyperthermia	McWilliams et al. [2002] Tammaro et al. [2003]
RYR2	180902	Arrhythmogenic right ventricular cardiomyopathy type 2 (ARVD2); familial polymorphic ventricular tachycardia	Tiso et al. [2001] Laitinen et al. [2001]
SCFR	164920	Hypereosinophilic syndrome	Pardanani et al. [2003]
SCN1A	182389	Myoclonic epilepsy, familial febrile seizures	Claes et al. [2001] Malacarne et al. [2002] Gennaro et al. [2003]
SCN5A	601144	Brugada syndrome; sudden unexplained nocturnal death syndrome (SUNDS)	Vatta et al. [2002a, 2000b] Valdivia et al. [2002] Mok et al. [2003]
SEMA4F	603706	Dyslexia	Francks et al. [2002]
SHOX	312865	Leri-Weill dyschondrosteosis	Ross et al. [2002]
SHP	604630	Increassed birth weight	Hung et al. [2003]
SIL	181590	Holoprosencephaly	Karkera et al. [2002]
SIX3	603714	Microphthalmia, anophthalmia, coloboma	Morrison et al. [2002]

SLC26A4	274600	Dendred's syndrome	Prasad et al. [2002]
SLC6A4	182138	Worldwide screen for DNA variation, association with anxiety	Glatt et al. [2001] Hu et al. [2002]
SLC11A1 (NRAMP1)	600266	Mycobacterium avium-intracellulare pulmonary disease	Huang et al. [1998]
SLC11A3	604653	Hyperferritinaemia	Cazzola et al. [2002]
SLC12A6 (KCC3A)	604878	Agenesis of the corpus callosum with peripheral neuropathy	Howard et al. [2002]
SLC18A2	193001	Worldwide screen for DNA variation	Glatt et al. [2001]
SLC18A3	600336	Late-onset Alzheimer's disease	Harold et al. [2003]
SLC22A2 (OCT2)	602608	Renal transport of xenobiotics	Leabman et al. [2003]
SLC25A1	190315	Schizophrenia	Williams et al. [2002b]
SLC26A4	274600	Dendred's syndrome	Prasad et al. [2002]
SLC39A1 (ZIRTL)	604740	Haemochromatosis	Sebastiani et al. [2003]
SLC6A4	182138	Worldwide screen for DNA variation, association with anxiety	Glatt et l. [2001] Hu et al. [2002]
SMAD4	607010	Uveal melanoma	Edmunds et al. [2002]
SMARCB1 (SNF5)	601607	Various cancers	Sevenet et al. [1999]
SMN1	600354	Spinal muscular atrophy	Mazzei et al. [2002] Sutomo et al. [2002]
SNX3	605930	Microcephaly, microphthalmia, ectrodactyly, prognathism (MMEP) phenotype	Vervoort et al. [2002]
SORBS1	605264	Obesity and type-2 diabetes	Lin et al. [2002]
SOX2	184429	Anophthalmia	Fantes et al. [2003]
SPINK1	167790	Idiopathic chronic pancreatitis	Audrézet et al. [2002]
SPINK5	605010	Netherton syndrome	Bitoun et al. [2002]
SRD5A2	607306	Micropenis	Sasaki et al. [2003]
ST7	600833	Cancer	Brown et al. [2002]
Stk22A1	192430	Schizophrenia	Williams et al. [2002b]
SUOX	272300	Sulfocysteinuria	Lam et al. [2002b]
TCF1 (HNF-1α)	142410	Type II diabetes mellitus, Maturity Onset Diabetes of the Young (MODY)	Boutin et al. [2001] Pruhova et al. [2003]
TCF14 (HNF4A)	600281	Maturity Onset Diabetes of the Young (MODY)	Pruhova et al. [2003]
TCRB	186930	Polymorphism and haplotypes	Donaldson et al. [2002]
TCRγ	186970	Acute lymphoblastic leukemia	Zur Stadt et al. [2001]
TCRD	186810	Acute lymphoblastic leukemia	Zur Stadt et al. [2003]
TDGF1	187395	Holoprosencephaly	De la Cruz et al. [2002]
TFP1	152310	Venous thrombosis	Amini-Nekoo et al. [2001]
TGFBR2	190182	Uveal melanoma	Edmunds et al. [2002]
TGM2	190196	Maturity-onset diabetes (MODY)	Bernassola et al. [2002]
TMEFF2 (HPP1)	605734	Colorectal cancer	Young et al [2002]
TNF	191160	Graft versus host disease	Wang et al. [2002a]
TNFRSF1A	191190	Tumor necrosis factor receptor-associated periodic syndrome	Dode et al. [2002]
TNFRSF1B	191191	Familial combined hyperlipidemia	Geurts et al. [2000]
TNFRSF6	134637	Follicle center lymphoma	Do et al. [2003]
TNNT2	191045	Hypertrophic cardiomyopathy	Ackerman et al. [2002] Van Driest et al. [2002a]

TP53 (p53)	191170	Various cancers	Gross et al. [2001] Keller et al. [2001] Quintanilla-Martinez et al. [2001] Edmunds et al. [2002] Geoerger et al. [2002] Leonard et al. [2002] Lingle et al. [2002] Narayanaswami et al. [2002] Smith et al. [2002] Tomizawa et al. [2002] Breton et al. [2003] Fricke et al. [2003] Temam et al. [2003]
TP73 (p73)	601990	Various cancers	Mai et al. [1998] Yokomizo et al. [1999a, b] Liu et al. [2000b]
TPM1	191010	Cardiomyopathy	Van Driest et al. [2002a, 2002b]
TPMT	187680	Drug metabolism	Hall et al. [2001] Schaeffeler et al. [2001]
TRX-1	187700	Various human tumor cell lines	Berggren and Powis [2001]
TSAP6		Prostate cancer	Porkka et al. [2003]
TSC1	605284	Mutations in tuberous sclerosis-1	Jones et al. [1999, 2000, 2001] Benit et al. [2000, 2001a] Dabora et al. [2001] Roberts et al. [2001] Emmerson et al. [2002, 2003] Franz et al. [2002]
TSC2	191092	Mutations in tuberous sclerosis-2	Choy et al. [1999] Jones et al. [1999, 2000, 2001] Dabora et al. [2001] Antonarakis et al. [2002] Emmerson et al. [2002, 2003] Franz et al. [2002] Roberts et al. [2002]
T-STAR	600131	Childhood absence epilepsy (CAE)	Sugimoto et al. [2001]
TTID	604103	Limb Girdle Muscular Dystrophy 1A	Hauser et al. [2000]
TWIST	601622	Craniosynostosis	Elanko et al. [2001]
UBE3A	601623	Angelman syndrome	Bercovich et al. [2000]
UGT1 (UGT1A1)	191740	Gilbert syndrome	Pirulli et al. [2000]
UNC93A		Epithelial ovarian cancer	Liu et al. [2002b]
USH1C	605242	Usher syndrome type 1C	Bitner-Glindzicz et al. [2000] Blaydon et al. [2003]
USH3	276902	Usher syndrome type III	Fields et al. [2002]
USP9Y (DFFRY)	400005	Male infertility and evolution	Sun et al. [1999] Shen et al. [2000]
VEGF	192240	Amyotrophic lateral sclerosis, DiGeorge syndrome	Lambrechts et al. [2003] Stalmans et al [2003]

VHL	193300	Hippel-Lindau disease	Klein et al. [2001] Marsh et al. [2001] Menegatti et al. [2001] Turner et al. [2002] Weirich et al. [2002b]
VMD2	153700	Best's vitelliform macular dystrophy	Marchant et al. [2002]
WASF3	605068	Neuroblastoma	Sossey-Alaoui et al. [2002b]
WFS1	606201	Wolfram syndrome	Colosimo et al. [2002b] Colosimo et al. [2003]
WRN	277700	Werner syndrome	Passarino et al. [2001]
XRCC9 (FANCG)	602956	Fanconi anemia	Auerbach et al. [2003]
ZNF74	194543	Schizophrenia	Williams et al. [2000]

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