

Recent identified disease genes with landmark references after January 2007.

Gene symbol	Disease ID	Disease name in OMIM	References
SORL1	98	{Alzheimer disease, pathogenesis, association with}, 104300 (3)	[1]
FAM83H	99	Amelogenesis imperfecta, type 3, 130900 (3)	[2]
IRAK3	153	{Asthma susceptibility 5}, 611064 (3)	[3]
CASP8	228	{Breast cancer, protection against}, 114480 (3)	[4]
AKT1	228	Breast cancer, somatic, 114480 (3)	[5]
HMMR	228	{Breast cancer, susceptibility to}, 114480 (3)	[6]
PALB2	228	{Breast cancer, susceptibility to}, 114480 (3)	[7]
BFSP1	277	Cataract, cortical, juvenile-onset, 611391 (3)	[8]
CHMP4B	277	Cataract, posterior polar, 3, 605387 (3)	[9]
FGD4	301	Charcot-Marie-Tooth disease, type 4H, 609311 (3)	[10]
FIG4	301	Charcot-Marie-Tooth disease, type 4J, 611228 (3)	[11]
PRPS1	301	Charcot-Marie-Tooth disease, X-linked recessive, 5, 311070 (3)	[12]
AKT1	346	Colorectal cancer, somatic, 114500 (3)	[5]
SMAD7	346	{Colorectal cancer, susceptibility to}, 114500 (3)	[13]
COG8	354	Congenital disorder of glycosylation, type IIh, 611182 (3)	[14]
TMEM15	354	Congenital disorder of glycosylation, type Im, 610768 (3)	[15]
UBIAD1	362	Corneal dystrophy, crystalline, of Schnyder, 121800 (3)	[16]
LRP6	365	Coronary artery disease, autosomal dominant 2, 610947 (3)	[17]
CCDC50	406	Deafness, autosomal dominant 44, 607453 (3)	[18]
RDX	406	Deafness, autosomal recessive, 24, 611022 (3)	[19]
ESRRB	406	Deafness, autosomal recessive 35, 608565 (3)	[20]
FGF3	406	Deafness, congenital with inner ear agenesis, microtia, and microdontia, 610706 (3)	[21]
WFS1	427	{Diabetes mellitus, noninsulin-dependent, association with}, 125853 (3)	[22]
CDKAL1	427	{Diabetes mellitus, noninsulin-dependent, susceptibility to}, 125853 (3)	[23]

IGF2BP2	427	{Diabetes mellitus, noninsulin-dependent, susceptibility to}, 125853 (3)	[23]
SLC30A8	427	{Diabetes mellitus, noninsulin-dependent, susceptibility to}, 135853 (3)	[24]
TAF1	462	Dystonia-Parkinsonism, X-linked, 314250 (3)	[25]
KCTD7	495	Epilepsy, progressive myoclonic 3, 611726 (3)	[26]
LCA5	873	Leber congenital amaurosis, type V, 604537 (3)	[27]
SCN4B	912	Long QT syndrome-10, 611819 (3)	[28]
AKAP9	912	Long QT syndrome-11, 611820 (3)	[29]
C3	937	{Macular degeneration, age-related, 9}, 611378 (3)	[30]
CX3CR1	937	{Macular degeneration, age-related, susceptibility to}, 602075 (3)	[31]
NCR3	940	{Malaria, mild, susceptibility to}, 609148 (3)	[32]
TIRAP	940	{Malaria, protection against}, 611162 (3)	[33]
FCGR2B	940	{Malaria, resistance to}, 611162 (3)	[34]
MBD5	990	Mental retardation, autosomal dominant 1, 156200 (3)	[35]
DOCK8	990	Mental retardation, autosomal dominant 2 (3)	[36]
GRIK2	990	Mental retardation, autosomal recessive, 6, 611092 (3)	[37]
DIP2B	990	Mental retardation, FRA12A type, 136630 (3)	[38]
CUL4B	990	Mental retardation-hypotonic facies syndrome, X-linked, 2, 300639 (3)	[39]
BRWD3	990	Mental retardation, X-linked 93, 300659 (3)	[40]
GRIA3	990	Mental retardation, X-linked 94, 300699 (3)	[41]
UPF3B	990	Mental retardation, X-linked, syndromic 14, 300676 (3)	[42]
BMP4	1012	Microphthalmia, syndromic 6, 607932 (3)	[43]
STRA6	1012	Microphthalmia, syndromic 9, 601186 (3)	[44]
C6orf66	1016	Mitochondrial complex I deficiency, 252010 (3)	[45]
LRP8	1054	{Myocardial infarction, susceptibility to}, 608446 (3)	[46]
FTO	1126	{Obesity, associated with}, 601665 (3)	[47]
TNFSF11	1161	Osteopetrosis, autosomal recessive 2, 259710 (3)	[48]

PLEKHM1	1161	Osteopetrosis, autosomal recessive 6, 611497 (3)	[49]
AKT1	1170	Ovarian cancer, somatic, 604370 (3)	[5]
HERC2	1227	[Skin/hair/eye pigmentation 1, blond/brown hair], 227220 (3)	[50]
TYR	1227	[Skin/hair/eye pigmentation 3, freckling], 601800 (3)	[50]
SLC24A2	1227	[Skin/hair/eye pigmentation 6, blond/brown hair], 210750 (3)	[50]
KITLG	1227	[Skin/hair/eye pigmentation 7, blond/brown hair], 611664 (3)	[50]
HNF1B	1272	{Prostate cancer, susceptibility to}, 176807 (3)	[51]
TOPORS	1316	Retinitis pigmentosa-31, 609923 (3)	[52]
STAT4	1324	{Rheumatoid arthritis, association with}, 180300 (3)	[53]
TREX1	1471	{Systemic lupus erythematosus, susceptibility to}, 152700 (3)	[54]
CR2	1471	{Systemic lupus erythematosus, susceptibility to, 9}, 610927 (3)	[55]
STAT4	1471	{Systemic lupus erythematosus, association with}, 152700 (3)	[53]
BANK1	1471	{Systemic lupus erythematosus, association with}, 152700 (3)	[56]
LARGE	1578	Walker-Warburg syndrome, 236670 (3)	[57]

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