疾患遺伝子一覧

疾患			遺伝子				文献
疾患名	MIM	遺伝形式	染色体座	遺伝子記号	遺伝子名	MIM	
脳血管障害							
アミロイドアンギオパチー・アミロイドーシス							
Amyloidosis VI(Cerebroarterial amyloidosis, Icelandic type)	#105150	AD	20p11.2	*CST3*	Cystatin C	*604312	*Proc Natl Acad Sci* **83**:2974–2978, 1986 *Acta Neurol Scand* **76**:102–114, 1987 *FEBS Let* **216**:229–233, 1987
Amyloidosis VII(Leptomeningeal amyloidosis)	#105210	AD	18q11.2–q12.1	*TTR*	Transthyretin(prealbumin, amyloidosis type I)	+176300	*Neurology* **47**:1562–1567, 1996 *Am J Path* **148**:361–366, 1996
Hereditary cerebral hemorrhage with amyloidosis, Dutch type(HCHWAD)	#609065	AD	21q21.3–22.05	*APP*	Amyloid beta(A4)precursor protein	*104760	*Science* **248**:1124–1126, 1990
凝固線溶系異常							
Hemophilia A(HEMA)	+306700	XR	Xq28	*F8*	Coagulation factor VIII, procoagulant component	+306700	*Nature* **315**:427–430, 1985 *New Engl J Med* **313**:842–848, 1985
Hemophilia B(HEMB)	#306900	XR	Xq27.1–q27.2	*F9*	Coagulation factor IX	*300746	*Nature* **303**:181–182, 1983
von Willebrand disease(VWD)	+193400	AD	12p13.3	*VWF*	von Willebrand factor	+193400	*J Clin Invest* **79**:1459–1465, 1987
Factor V deficiency	#227400	AR	1q23	*F5*	Coagulatio factor V(proaccelerin, labile factor)	*612309	*Nature* **369**:64–67, 1994
Hereditary thrombophilia due to protein C deficiency, autosomal dominant	#176860	AD	2q13–q14	*PROC*	Protein C(inactivator of coagulation factors Va and VIIIa)	*612283	*Proc Natl Acad Sci* **84**:2829–2832, 1987
Hereditary thrombophilia due to protein S deficiency, autosomal dominant	#612336	AD	3p11.1–q11.2	*PROS1*	Protein S(alpha)	*176880	*Blood* **73**:479–483, 1989
Plasminogen deficiency, type I	#217090	AR	6q26	*PLG*	Plasminogen	*173350	*Proc Natl Acad Sci* **88**:115–119, 1991
Plaminogen activator inhibitor 1(PAI1) deficiency	*173360	AD	7q21.3–q22	*SERPINE1*	Serpin peptidase inhibitor, clade E (nexin, plasminogen activator inhibitor type 1), member 1	*173360	*New Engl J Med* **327**:1729–1733, 1992
Antithromnin III deficiency	+107300	AD	1q23–q25	*SERPINC1*	Serpin peptidase inhibitor, clade C (antithrombin), member 1	+107300	*J Biol Chem* **261**:1174–1176, 1986 *Nucleic Acids Res* **14**:2408, 1986
血管異常							
Cerebral cavernous malformations 1(CCM1)	#116860	AD	7q11.2–q21	*KRIT1*	KRIT1, ankyrin repeat containing	*604214	*Nat Genet* **23**:189–193, 1999
Cerebral cavernous malformations 2(CCM2)	#603284	AD	7p13	*CCM2*	Cerebral cavernous malformations 2	*607929	*Am J Hum Genet* **73**:1459–1464, 2003
Cerebral cavernous malformations 3(CCM3)	#603285	AD	3p26.1	*PDCD10*	Programmed cell death 10	*609118	*Am J Hum Genet* **76**:42–51, 2005
CADASIL・その他							
Brain small vessel disease with hemorrhage	#607595	AD	13q34	*COL4A1*	Collagen, type IV, alpha-1	*120130	*New Engl J Med* **354**:1489–1496, 2006
Hereditary angiopathy with nephropathy, aneurysms, and muscle cramps (HANAC)	#611773	AD	13q34	*COL4A1*	Collagen, type IV, alpha-1	*120130	*New Engl J Med* **357**:2687–2695, 2007
Cerebral arteriopathy, autosomal dominant, with subcortical infarcts and leukoencephalopathy(CADASIL)	#125310	AD	19p13.2–p13.1	*NOTHC3*	Notch homolog 3(*Drosophila*)	*600276	*Nature* **383**:707–710, 1996
Cerebral autosomal recessive arteriopathy with subcortical infarcts and leukoencephalopathy(CARASIL)	#600142	AR	10q25.3–q26.2	*HTRA1*	HtrA serine peptidase 1	*602194	*New Engl J Med* **360**:1729–1739, 2009
痴呆性疾患							
Alzheimer disease 1(AD1)	#104300	AD	21q21.3–22.05	*APP*	Amyloid beta(A4)precursor protein	*104760	*Nature* **349**:704–706, 1991
Alzheimer disease 3(AD3)	#607822	AD	14q24.3	*PSEN1*	Presenilin 1	+104311	*Nature* **375**:754–760, 1995
Alzheimer disease 4(AD4)	#606889	AD	1q31–q42	*PSEN2*	Presenelin 2(Alzheimer disease 4)	+600759	*Science* **269**:973–977, 1995 *Nature* **376**:775–778, 1995
Frontotemporal dementia(FTD)	#600274	AD	17q21.1	*MAPT*	Microtubule-associated protein tau	+157140	*Nature* **393**:702–705, 1998 *Proc Natl Acad Sci* **95**:7737–7741, 1998 *Ann Neurol* **43**:815–825, 1998
Frontotemporal lobar degenetation with ubiquitin-positive inclusions(FTLDU)	#607485	AD	17q21.32	*GRN*	Granulin	*138945	*Nature* **442**:916–919,920–924, 2006
Frontotemporal dementia, chromosome 3-linked(FTD3)	#600795	AD	3q11.2	*CHMP2B*	Chromatin-modifying protein 2B	+609512	*Nat Genet* **37**:806–808, 2005
Encephalopathy, familial, with neuroserpin inclusion bodies(FENIB)	#604218	AD	3q26	*SERPINI1*	Serpin peptidase inhibitor, clade I (neuroserpin), member 1	*602445	*Nature* **401**:376–379, 1999
Familial British dementia(FBD)	#176500	AD	13q14	*ITM2B*	Integral membrane protein 2B	*603904	*Nature* **399**:776–781, 1999
Prion diseases							
Familial Creutzfeldt-Jakob disease(CJD)	#123400	AD	20pter–p12	*PRNP*	Prion protein	*176640	*Lancet* **I**:51–52, 1989
Gerstmann-Sträussler disease(GSD)	#137440	AD	20pter–p12	*PRNP*	Prion protein	*176640	*Nature* **338**:342–345, 1989
Huntington disease-like 1(HDL1)	#603218	AD	20pter–p12	*PRNP*	Prion protein	*176640	*Am J Hum Genet* **69**:1385–1388, 2001 *Brain* **122**:2375–2386, 1999
Familial fatal insomnia(FFI)	#600072	AD	20pter–p12	*PRNP*	Prion protein	*176640	*New Engl J Med* **326**:444–449, 1992
Opticoacoustic nerve atrophy with dementia(Jensen syndrome)	#311150	XR	Xq22	*TIMM8A*	Traslocase of inner mitochindrial membrane 8 homolog A(yeast)	*300356	*Am J Hum Genet* **61**(suppl.): A349, 1997
錐体外路疾患							
Huntington disease(HD)	+143100	AD	4p16.3	*HTT*	Huntingtin	+143100	*Cell* **72**:971–983, 1993
Huntington disease-like 2(HDL2)	#606438	AD	16q24.3	*JPH3*	Junctophilin 3	*605268	*Nat Genet* **29**:377–378, 2001 *Ann Neurol* **50**:373–380, 2001
Benign hereditary chorea(BHC)	#118700	AD	14q13	*NKX2-1*	NK2 homeobox 1	*600635	*Hum Mol Genet* **11**:971–979, 2002 *J Clin Invvest* **109**:475–480, 2002
Choreoacanthocytosis(CHAC)	#200150	AR	9q21	VPS13A	Vacuolar protein sorting 13 homolog A(*S. cerecisiae*)	*605978	*Nat Genet* **28**:119–120, 2001 *Nat Genet* **28**:121–122, 2001
Adult-onset basal ganglia disease (neuroferritinopathy)	#606159	AD	19q13.3	*FTL*	Ferritin, light polypeptide	*134790	*Nat Genet* **28**:350–354, 2001
Striatonigral degeneration, infantile (SNDI)	#271930	AR	19q13.32–q13.31	*NUP62*	Nucleoporin 62kDa	*605815	*Ann Neurol* **60**:214–222, 2006
Neurodegeneratin with brain iron accumulation 1(NMIA1; Hallervorden-Spatz disease)	#234200	AR	20p13–p12.3	*PANK2*	Pantothenate kinase 2	*606157	*Nat Genet* **28**:345–349, 2001
Hypoprebetalipoproteinemia, acatntocytosis, retinitis pigmentosa, and pallidal degeneration(HARP syndrome)	#607236	AR	20p13–p12.3	*PANK2*	Pantothenate kinase 2	*606157	*Neurology* **58**:1673–1674, 2002
Neuroaxonal dystrophy, infantile(INAD1)	#256600	AR	22q13.1	*PLA2G6*	Phospholipase A2, group VI (cytosplic, calcium-independent)	*603604	*Nat Genet* **38**:752–754, 2006
Neurodegeneration with brain iron accumulation 2(NBIA2)	#610217	AR	22q13.1	*PLA2G6*	Phospholipase A2, group VI (cytosplic, calcium-independent)	*603604	*Nat Genet* **38**:752–754, 2006

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Karak syndrome	#608395	AR	22q13.1	*PLA2G6*	Phospholipase A2, group VI (cytosplic, calcium-independent)	*603604	*Nat Genet* **38**：752-754, 2006
Torsion dystonia 1, autosomal domonant (DYT1, Dystonia musculorum deformans 1)	#128100	AD	9q34	*TOR1A*	torsin family 1, member A (torsin A)	*605204	*Nat Genet* **17**：40-48, 1997
Myoclonic dystonia (DYT11)	#159900	AD	7q21	*SGCE*	Sarcoglycan, epsilon	*604149	*Nat Genet* **29**：66-69, 2001
Dystonia 12 (DYT12)	#128235	AD	19q12-q13.2	*ATP1A3*	ATPase, Na^+/K^+ transporting, alpha 3 polypeptide	*182350	*Neuron* **43**：169-175, 2004
Myoclonus-dystonia syndrome	#159900	AD	11q23.1	*DRD2*	Dopamine receptor D2	*126450	*Proc Natl Acad Sci* **96**：5173-5176, 1999
Torsion dystonia 3, X-linked (DYT3)	#314250	XR	Xq13	*TAF1*	TAF1 RNA polymerase II, TATA box binding protein (TBP)-associated factor, 250kDa	*313650	*Am J Hum Genet* **80**：393-406, 2007
Dystonia 16 (DYT16)	#612067	AR	2q31.3	*PRKRA*	Protein kinase, interferon-inducible double stranded DNA dependent activator	*603424	*Lancet Neurol* **7**：207-215, 2008
Dystonia 18 (DYT18)	#612126	AD	1p35-p31.3	*SLC2A1*	Solute carrier family 2 (facilitated glucose transporter), member1	*138140	*J Clin Invvest* **118**：2157-2168, 2008
Juvenile-onset dystonia	#607371	AD	7p22-p12	*ACTB*	actin, beta	*102630	*Ann Neurol* **52**：465-476, 2002
McLeod syndrome	+314850	XR	Xp21.2-p21.1	*XK*	X-linked Kx blood group (McLeod syndrome)	+314850	*Cell* **77**：869-880, 1994
Mohr-Tranebjaerg syndrome (MTS)	#304700	XR	Xq22	*TIMM8A*	Traslocase of inner mitochindrial membrane 8 homolog A (yeast)	*300356	*Nat Genet* **14**：177-180, 1996
Hypogonadism, alopetia, daibetes mellitus, mental retardation, and extrapyramidal syndrome (Woodhouse-Sakati syndrome)	#241080	AR	2q22.3-q35	*C2orf37*	Chromosome 2 open reading frame 37	*612515	*Am J Hum Genet* **83**：681-691, 2008
Paroxysmal nonkinesigenic dyskinesia 1 (PNKD1)	#118800	AD	2q35	*PNKD*	Paroxymal nonkinesigenic dyskinesia ; Myofibrillogenesis regulator 1 (MR1 ; KIAA1184)	*609023	*Hum Mol Genet* **13**：3161-3170, 2004
Gilles de la Tourette syndrome	#137580	AD	13q31	*SLITRK1*	SLIT and NTRK-like family, member 1	*609678	*Science* **310**：317-320, 2005
Familial Parkinson disease type 1 (PARK1, PD1)	#168601	AD	4q21	*SNCA*	Synuclein, alpha (non A4 component of amyloid precursor)	*163890	*Science* **276**：2045-2047, 1997
Familial Parkinson disease type 4 (PARK4, PD4)	#605543	AD	4q21	*SNCA*	Synuclein, alpha (non A4 component of amyloid precursor)	*163890	*Science* **302**：841, 2003
Parkinson disease 2, autosomal recessive juvenile (PARK2)	#600116	AR	6q25.2-q27	*PARK2*	Parkinson disease (autosomal recessive, juvenile) 2, parkin	*602544	*Nature* **392**：605-608, 1998
Parkinson disease 5 (PARK5)	+191342	AD	4p14	*UCHL1*	Ubiquitin carboxyl-terminal esterase L1 (ubiquitin thiolesterase)	+191342	*Nature* **395**：451-452, 1998
Parkinson disease 6, autosomal recessive early-onset (PARK6)	#605909	AR	1p36.2	*PINK1*	PTEN induced putative kinase 1	*608309	*Science* **304**：1158-1160, 2004
Parkinson disease 7, autosomal recessive early-onset (PARK7)	*606324	AR	1p36	*PARK7*	Parkinson disease (autosomal recessive, early onset) 7	*602533	*Science* **299**：256-259, 2003
Parkinson disease 8 (PARK8)	#607060	AD	12q12	*LRRK2*	Leucine-rich repeat kinase	*609007	*Neuron* **44**：595-600, 2004
Kufor-Rakeb syndrome (PARK9)	*606693	AR	1p36	*ATP13A2*	ATPase type 13A2	*610513	*Nat Genet* **38**：1184-1191, 2006
Parkinson disease 13 (PARK13)	*610297	AD	2p12	*HTRA2*	HtrA serine peptidase 2	*606441	*Hum Mol Genet* **14**：2099-2111, 2005
常染色体優性脊髄小脳運動失調症							
Spinocerebellar ataxia 1 (SCA1)	#164400	AD	6p23	*ATXN1*	Ataxin 1	*601556	*Nat Genet* **4**：221-226, 1993
Spinocerebellar ataxia 2 (SCA2)	#183090	AD	12q24	*ATXN2*	Ataxin 2	*601517	*Nat Genet* **14**：269-276, 1996 *Nat Genet* **14**：277-284, 1996 *Nat Genet* **14**：285-291, 1996
Machado-Joseph disease (MJD, Spinocerebellar ataxia 3, SCA3)	#109150	AD	14q32.1	*ATXN3*	Ataxin 3	*607047	*Nat Genet* **8**：221-228, 1994
Spinocerebellar ataxia 5 (SCA5)	#600224	AD	11q13	*SPTBN2*	Spectrin, beta, nonerythrocytic, 2	*604985	*Nat Genet* **38**：184-190, 2006
Spinocerebellar ataxia 6 (SCA6)	#183086	AD	19p13	*CACNA1A*	Calcium channel, voltage-dependent, P/Q type, alpha 1A subunit	*601011	*Nat Genet* **15**：62-69, 1997
Spinocerebellar ataxia 7 (SCA7)	#164500	AD	3p21.1-p12	*ATXN7*	Ataxin 7	*607640	*Nat Genet* **17**：65-70, 1997
Spinocerebellar ataxia 8 (SCA8)	#608768	AD	13q21	*ATXN8OS*	ATXN8 opposite strand (non-coding)	*603680	*Nat Genet* **21**：379-384, 1999
Spinocerebellar ataxia 10 (SCA10)	#603516	AD	22q13-qter	*ATXN10*	Ataxin 10	*611150	*Nat Genet* **26**：191-194, 2000
Spinocerebellar ataxia 11 (SCA11)	#604432	AD	15q15.2	*TTBK2*	Tau tubulin kinase 2	*611695	*Nat Genet* **39**：1434-1436, 2007
Spinocerebellar ataxia 12 (SCA12)	#604326	AD	5q31-q33	*PPP2R2B*	Protein phosphatase 2 (formerly 2A), regulatory subunit B, beta isoform	*604325	*Nat Genet* **23**：391-392, 1999
Spinocerebellar ataxia 13 (SCA13)	#605259	AD	19q13.3-q13.4	*KCNC3*	Potassium voltage-gated channel, Shaw-related subfamily, member 3	*176264	*Nat Genet* **38**：447-45, 2006
Spinocerebellar ataxia 14 (SCA14)	#605361	AD	19q13.4	*PRKCG*	Protein kinase C, gamma	*176980	*Am J Hum Genet* **73**：839-849, 2003
Spinocerebellar ataxia 15 (SCA15)	#606658	AD	3p26-p25	*ITPR1*	Inositol 1, 4, 5-triphosphate receptor, type 1	*147265	*PLoS Genet* **3**：e108, 2007
Spinocerebellar ataxia 17 (SCA17)	#607136	AD	6q27	*TBP*	TATA box binding protein	*600075	*Hum Mol Genet* **8**：2047-2053, 1999 *Eur J Hum Genet* **9**：150-164, 2001 *Hum Mol Genet* **10**：1441-1448, 2001 *Brain* **124**：1939-1947, 2001
Spinocerebellar ataxia 27 (SCA27)	#609307	AD	13q34	*FGF14*	Fibroblast growth factor 14	*601515	*Am J Hum Genet* **72**：191-199, 2003
Dentatorubral-pallidoluysian atrophy (DRPLA)	#125370	AD	12p13.31	*ATN1*	Atrophin 1	*607462	*Nat Genet* **6**：9-13, 1994 *Nat Genet* **6**：14-18, 1994
Cerebellar atrophy, ataxia, and mental retardation	*600702	AD	12q13	*SCN8A*	Sodium channel, voltage gated, type VIII, alpha subunit	*600702	*J Med Genet* **43**：527-530, 2006
常染色体性劣性脊髄小脳運動失調症							
Friedreich ataxia (FRDA)	#229300	AR	9q13	*FXN*	Frataxin	*606829	*Science* **271**：1423-1427, 1996
Ataxia, early-onset, with oculomotor apraxia and hypoalbunemia (EAOH)	#208920	AR	9p13.3	*APTX*	Aprataxin	*606350	*Nat Genet* **29**：184-188, 2001 *Nat Genet* **29**：189-193, 2001
Spinocerebellar ataxia, autosomal recessive 1 (SCAR1 ; Ataxia-oculomotor apraxia 2, AOA2)	#606002	AR	9q34	*SETX*	Senataxin	*608465	*Nat Genet* **36**：205-312, 2004
Spinocerebellar ataxia, autosomal recessive 8 (SCAR8)	#610743	AR	6q25	*SYNE1*	Spectrin repeat containong, nuclear envelope 1	*608441	*Nat Genet* **39**：80-85, 2007
Spinocerebellar ataxia, autosomal recessive 9 (SCAR9)	#612016	AR	1q42.2	*CABC1*	Chaperone, ABC1 activity of bc1 complex homolog (*S. pombe*)	*696980	*Am J Hum Genet* **82**：623-630, 2008 *Am J Hum Genet* **82**：661-672, 2008
Ataxia-telangiectasia (AT, Louis-Bar syndrome)	#208900	AR	11q22.3	*ATM*	Ataxia telangiectasia mutated	*607585	*Science* **268**：1749-1753, 1995

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Ataxia-telangiectasia-like disorder (ATLD)	#604391	AR	11q21	*MRE11A*	MRE11 meiotic recombination 11 homolog A (*S. cerevisiae*)	*600814	*Cell* **99** : 577–587, 1999
Spinocerebellar ataxia, autosomal recessive, with axonal neuropathy (SCAN1)	#607250	AR	14q31–q32	*TDP1*	Tyrosyl-DNA phosphodiesterase 1	*607198	*Nat Genet* **32** : 267–272, 2002
Cerebellar ataxia, Cayman type (ATCAY)	#601238	AR	19p13.3	*ATCAY*	Ataxia, cerebellar, Cayman type (caytaxin)	*608179	*Nat Genet* **35** : 264–269, 2003
Marinesco-Sjögren syndrome (MSS)	#248800	AR	5q31	*SIL1*	SIL1 homolog, endoplasmic reticulum chaperone (*S. cerevisiae*)	*608005	*Nat Genet* **37** : 1309–1311, 2005 *Nat Genet* **37** : 1312–1314, 2005
ビタミン・脂質代謝異常							
Familial isolated deficiency of vitamin E (VED)	#277460	AR	8q13.1–q13.3	*TTPA*	Tocopherol (alpha) transfer protein	*600415	*Nat Genet* **9** : 56–62, 1995
Abetalipoproteinemia (ABL ; Bassen-Kornzweig syndrome)	#200100	AR	4q22–q24	*MTTP*	Microsomal triglyceride transfer protein	*157147	*Nature* **365** : 65–69, 1993 *Hum Mol Genet* **2** : 2109–2116, 1993
Hypobetalipoproteinemia, normotriglyceridemic	+107730	AD	2p24	*APOB*	Apolipoprotein B [including Ag (x) antigen]	+107730	*Biochem Biophys Res Commun* **148** : 279–285, 1987
Hereditary megaloblastic anaemia 1 (MGA1)	#261100	AR	10p12.1	*CUBN*	Cubilin (intrinsic factor–cobalamin receptor)	*602997	*Nat Genet* **21** : 309–313, 1999
Hereditary megaloblastic anaemia 1 (MGA1)	#261100	AR	14q32	*AMN*	Amnionless homolog (mouse)	*605799	*Nat Genet* **33** : 426–429, 2003
Transcobalamin II deficiency	+275350	AR	22q11.2–qter	*TCN2*	Transcobalamin II ; macrocytic anemia	+275350	*Biochem Buophys Res Commun* **204** : 1111–1118, 1994 *Hum Mol Genet* **3** : 1835–1840, 1994
その他							
Fragile X tremor/ataxia syndrome (FXTAS)	#300623	XR	Xq27.3	*FMR1*	Fragile X mental retardation 1	*309550	*Neurology* **57** : 127–130, 2001
ARTS syndrome	#301835	XR	Xq22–q24	*PRPS1*	Phosphoribosylpyrophosphate synthetase 1	*311850	*Am J Hum Genet* **81** : 507–518, 2007
先天性眼振							
Nystagmus 1, Congenital, X-linked (NYS1)	#310700	XD	Xq26.2–q27	*FRMD7*	FERM domain ontaining 7	*300628	*Nat Genet* **38** : 1242–1244, 2006
家族性痙性対麻痺							
Spastic paraplegia 1, X-linked (SPG1)	#303350	XR	Xq28	*L1CAM*	L1 cell adhesion molecule	*308840	*Nat Genet* **7** : 402–407, 1994
Spastic paraplegia 2, X-linked (SPG2)	#312920	XR	Xq22	*PLP1*	Proteolipid protein 1	*300401	*Nat Genet* **6** : 257–262, 1994
Spastic paraplegia 3A, autosomal dominant (SPG3A)	#182600	AD	14q11–q21	*ATL1*	Atlastin GTPase 1	*606439	*Nat Genet* **29** : 326–331, 2001
Spastic paraplegia 4, autosomal dominant (SPG4)	#182601	AD	2p22–p21	*SPAST*	Spastin	*604277	*Nat Genet* **23** : 296–303, 1999
Spastic paraplegia 5A, autosomal recessive (SPG5A)	#270800	AR	8q21.3	*CYP7B1*	Cytochrome P450, family 7, subfamily B, polypeptide 1	+603711	*Am J Hum Genet* **82** : 510–515, 2008
Spastic paraplegia 6, autosomal dominant (SPG6)	#600363	AD	15q11.1	*NIPA1*	Non imprinted in Prader–Willi/Angelman syndrome 1	*608145	*Am J Hum Genet* **73** : 967–961, 2003
Spastic paraplegia 7, autosomal recessive (SPG7)	#607259	AR	16q24.3	*SPG7*	Spastic paraplegia 7 (pure and complicated autosomal recessive)	*602783	*Cell* **93** : 973–983, 1998
Spastic paraplegia 8, autosomal dominant (SPG8)	#603563	AD	8q24.13	*KIAA0196*	KIAA0196	*610657	*Am J Hum Genet* **80** : 152–161, 2007
Spastic paraplegia 10, autosomal dominant (SPG10)	#604187	AD	12q13	*KIF5A*	Kinesin family member 5A	*602821	*Am J Hum Genet* **71** : 1189–1194, 2002
Spastic paraplegia 11, autosomal recessive (SPG11)	#604360	AR	15q21.1	*SPG11*	Spastic paraplegia 11 (autosomal recessive)	*610844	*Nat Genet* **39** : 366–372, 2007
Spastic paraplegia 13, autosomal dominant (SPG13)	#605280	AD	2q24	*HSPD1*	Heat shock 60kDa protein 1 (chaperonin)	*118190	*Am J Hum Genet* **70** : 1328–1332, 2002
Spastic paraplegia 15, autosomal recessive (SPG15)	#270700	AR	14q24.1	*ZFYVE26*	Zinc finger, FYVE domain containing 26	*612012	*Am J Hum Genet* **82** : 992–1002, 2008
Spastic paraplegia 17 (Silver syndrome)	#270685	AD	11q12–q14	*BSCL2*	Bernardinelli-Seip congenital lipodystrophy 2 (seipin)	*606158	*Nat Genet* **36** : 271–276, 2004
Spastic paraplegia 20, autosomal recessive (SPG20 ; Troyer syndrome)	#275900	AR	13q12.3	*SPG20*	Spatic paraplegia 20, spastin (Troyer syndrome)	*607111	*Nat Genet* **31** : 347–348, 2002
Mast syndrome (Spastic paraplegia 21, autosomal recessive ; SPG21)	#248900	AR	15q21–q22	*SPG21*	Spastic paraplegia 21 (autosomal recessive, Mast syndrome)	*608181	*Am J Hum Genet* **73** : 1147–1156, 2003
Allan-Herndon-Dudley syndrome (Spastic paraplegia 22, X-linked ; SPG22)	#300523	XD	Xq13.2	*SLC16A2*	Solute carrier family 16, member 2 (monocarboxylic acid transporter 8)	*300095	*Thyroid* **13** : 672, 2003 *Lancet* **364** : 1435–1437, 2004 *Am J Hum Genet* **74** : 168–175, 2004
Spastic paraplegia 31, autosomal dominant (SPG31)	#610250	AD	2p11.2	*REEP1*	Receptor accessory protein 1	*609139	*Am J Hum Genet* **79** : 365–369, 2006
Spastic paraplegia 33, autosomal dominant (SPG33)	#610244	AD	10q24.2	*ZFYVE27*	Zinc finger FYVE domain containing protein 27	*610243	*Am J Hum Genet* **79** : 351–357, 2006
Spastic paraplegia 39, autosomal recessive (SPG39)	#612020	AR	19p13.3	*PNPLA6*	Patatin-like phospholipase domain containing 6	*603197	*Am J Hum Genet* **82** : 780–785, 2008
Spastic paraplegia 42, autosomal dominant (SPG42)	#612539	AD	3q25.31	*SLC33A1*	Solute carrier family 33 (acetyl-CoA transporter), member 1	*603690	*Am J Hum Genet* **83** : 752–759, 2008
Infantile-onset ascending hereditary spastic paralysis (IAHSP)	#607225	AR	2q33	*ALS2*	Amyotrophic lateral sclerosis 2 (juvenile)	*606352	*Am J Hum Genet* **71** : 518–527, 2002
Spastic ataxia, Charlevoix-Saguenay type (SACS)	#270550	AR	13q12	*SACS*	Spastic ataxia of Charlevoix-Saguenay (sacsin)	*604490	*Nat Genet* **24** : 120–125, 2000
Sjögren-Larsson syndrome (SLS)	#270200	AR	17p11.2	*ALDH3A2*	Aldehyde dehydrogenase 3 family, member A2	*609523	*Nat Genet* **12** : 52–57, 1996
運動ニューロン疾患							
家族性筋萎縮性側索硬化症							
Amyotrophic lateral sclerosis 1 (ALS1)	#105400	AD	21q22.1	*SOD1*	Superoxide dismutase 1, soluble	*147450	*Nature* **362** : 59–62, 1993
Amyotrophic lateral sclerosis 2, juvenile (ALS2)	#205100	AR	2q33	*ALS2*	Amyotrophic lateral sclerosis 2 (juvenile)	*606352	*Nat Genet* **29** : 160–165, 2001 *Nat Genet* **29** : 166–173, 2001
Amyotrophic lateral sclerosis 4, juvenile (ALS4)	#602433	AD	9q34	*SETX*	Senataxin	*608465	*Am J Hum Genet* **74** 1128–1135, 2004
Amyotrophic lateral sclerosis 6 (ALS6)	%608030	AD	16q11.2	*FUS*	Fusion [involved in t (12 ; 16) in malignant liposarcoma]	*137070	*Science* **323** : 1205–1208, 2009 *Science* **323** : 1208–1211, 2009
Amyotrophic lateral sclerosis 8 (ALS8) Incl. Spinal muscular atrophy, proximal, adult, autosomal (Finkel type)	#608627 #182980	AD	20q13.3	*VAPB*	VAMP (vesicle-associated membrane protein)-associated protein B and C	*605704	*Am J Hum Genet* **75** : 822–831, 2004
Amyotrophic lateral sclerosis 9 (ALS9)	#611895	AD	14q11.2	*ANG*	Angiogenin, ribonuclease, RNase A family, 5	*105850	*Nat Genet* **38** : 411–413, 2006

疾患			遺伝子				文献
疾患名	MIM	遺伝形式	染色体座	遺伝子記号	遺伝子名	MIM	
Amyotrophic lateral sclerosis 10(ALS10)	#612069	AD	1p36.2	*TARDBP*	TAR DNA binding protein	*605078	*Science* **319** : 1668–1672, 2008 *Nat Genet* **40** : 572–574, 2008
脊髄性筋萎縮症・遠位型運動ニューロノパチー/ニュロパチー							
Spinal muscular atrophy							*Cell* **80** : 155–165, 1995 *Nat Genet* **11** : 335–337, 1995 *Lancet* **346** : 316–317, 1995
Spinal muscular atrophy, type I(SMA1, Werdnig–Hoffmann disease)	#253300	AR	5q12.2–q13.3	*SMN1*	Survival of motor neuron 1, telomeric	*600354	
Spinal muscular atrophy, type II (SMA2, Intermediate type)	#253550	AR	5q12.2–q13.3	*SMN1*	Survival of motor neuron 1, telomeric	*600354	
Spinal muscular atrophy, type III (SMA3, Kugelberg–Welander syndrome)	#253400	AR	5q12.2–q13.3	*SMN1*	Survival of motor neuron 1, telomeric	*600354	
Spinal muscular atrophy, type IV (SMA4)	#271150	AR	5q12.2–q13.3	*SMN1*	Survival of motor neuron 1, telomeric	*600354	
Spinal and bulbar muscular atrophy, X–linked 1(SMAX1, SBMA)	#313200	XR	Xq11–q12	*AR*	Androgen receptor	*313700	*Nature* 352 : **77**–79, 1991
Spinal muscular atrophy, distal, autosomal recessive, 1(DSMA1 ; SMARD1 ; HMN6)	#604320	AR	11q13.2–q13.4	*IGHMBP2*	Immunoglobulin mu binding protein 2	*600502	*Nat Genet* **29** : 75–77, 2001
Spinal muscular atrophy, distal, autosomal reseccive, 4(DSMA4)	#611067	AR	1p36	*PLEKHG5*	Pleckstrin homology domain containing, family G(with RhoGef domain)member 5	*611101	*Am J Hum Genet* **8** : 67–76, 2007
Neuronopathy, distal hereditary motor, type IIA(HMN2A)	#158590	AD	12q24.3	*HSPB8*	Heat shock 22kDa protein 8	*608014	*Nat Genet* **36** : 597–601, 2004
Neuronopathy, distal hereditary motor, type V(HMN5 ; DSMAV)	#600794	AD	7p15	*GARS*	Glycyl–tRNA synthetase	*600287	*Am J Hum Genet* **72** : 1293–1299, 2003
Neuronopathy, distal hereditary motor, type VIIB(HMN7B)	#607641	AD	2p13	*DCTN1*	Dynactin 1(p150, glued homolog, *Drosophila*)	*601143	*Nat Genet* **33** : 455–456, 2003
遺伝性ニューロパチー							
遺伝性運動感覚ニューロパチー(Charcot–Marie–Tooth 病)							
Charcot–Marie–Tooth disease, demyelinating, type 1A(CMT1A)	#118220	AD	17p11.2	*PMP22*	Peripheral myelin protein 22	*601097	*Cell* **66** : 219–232, 1991
Charcot–Marie–Tooth disease, demyelinating, type 1B(CMT1B)	#118200	AD	1q22–q23	*MPZ*	Myelin protein zero	*159440	*Nat Genet* **5** : 31–34, 1993
Charcot–Marie–Tooth disease, demyelinating, type 1C(CMT1C)	#601098	AD	16p13.3–p12	*LITAF*	Lipopolysaccaride–induced tumor necrosis factor–alpha factor	*603795	*Neurology* **60** : 22–26, 2003
Charcot–Marie–Tooth disease, demyelinating, type 1D(CMT1D)	#607678	AD	10q21.1–q22.1	*EGR2*	Early growth response 2	*129010	*Nat Genet* **18** : 53–55, 1998
Charcot–Marie–Tooth disease and deafness(CMT1E)	#118300	AD	17p11.2	*PMP22*	Peripheral myelin protein 22	*601097	*Am J Hum Genet* **64** : 1580–1593, 1999
Charcot–Marie–Tooth disease, demyelinating, type 1F(CMT1F)	#607734	AD	8p21	*NEFL*	Neurofilament, light chain	*162280	*Brain* **126** : 590–597, 2003
Peripheral demyelinating neuropathy, central dysmyelination, Waardenburg syndrome, and Hirschsprung disease (PCWH)	#609136	AD	22q13	*SOX10*	SRY(sex–determining region Y)–box 10	*602229	*Ann Neurol* **46** : 313–318, 1999
Charcot–Marie–Tooth disease, X–linked, 1 (CMTX1)	#302800	XR	Xq13.1	*GJB1*	Gap junction protein, beta 1, 32kDa	*304040	*Science* **262** : 2039–2042, 1993
Charcot–Marie–Tooth disease, X–linked recessive, 5(CMTX5)	#311070	XR	Xq22–q24	*PRPS1*	Phosphoribosylpyrophosphate synthetase 1	*311850	*Am J Hum Genet* **81** : 552–558, 2007
Charcot–Marie–Tooth disease, dominant intermediate B(CMTDIB ; DI–CMTB ; CMTDI1)	#606482	AD	19p12–p13.2	*DNM2*	Dynamin 2	*602378	*Nat Genet* **37** : 289–294, 2005
Charcot–Marie–Tooth disease, dominant intermediate C(CMTDIC ; DI–CMTC)	#608323	AD	1p35	*YARS*	Tyrosyl–tRNA synthetase	*603623	*Nat Genet* **38** : 197–202, 2006
Charcot–Marie–Tooth disease, dominant intermediate D(CMTDID)	#607791	AD	1q22–q23	*MPZ*	Myelin protein zero	*159440	*J Neurol Neurosurg Psychiat* **67** : 174–179, 1999
Charcot–Marie–Tooth disease, recessive intermediate A(CMTRIA ; RI–CMTA)	#608340	AR	8q21	*GDAP1*	Ganglioside–induced differentiation–associated protein 1	*606598	*Neurology* **59** : 1865–1872, 2002
Charcot–Marie–Tooth disease, axonal, type 2A1(CMT2A1)	#118210	AD	1p36.2	*KIF1B*	Kinesin family member 1B	*605995	*Cell* **105** : 587–597, 2001
Charcot–Marie–Tooth disease, axonal, type 2A2(CMT2A2)	#609260	AD	1p36.2	*MFN2*	Mitofusion 2	*608507	*Nat Genet* **36** : 449–451, 2004
Charcot–Marie–Tooth disease, axonal, type 2B(CMT2B)	#600882	AD	3q21	*RAB7*	RAB7, member RAS oncogene family	*602298	*Am J Hum Genet* **72** : 722–727, 2003
Charcot–Marie–Tooth disease, axonal, type 2B1(CMT2B1)	#605588	AR	1q21.1	*LMNA*	Lamin A/C	*150330	*Am J Hum Genet* **70** : 726–736, 2002
Charcot–Marie–Tooth disease, axonal, type 2D(CMT2D)	#601472	AD	7p15	*GARS*	Glycyl–tRNA synthetase	*600287	*Am J Hum Genet* **72** : 1293–1299, 2003
Charcot–Marie–Tooth disease, axonal, type 2E(CMT2E)	#607084	AD	8p21	*NEFL*	Neurofilament, light chain	*162280	*Am J Hum Genet* **67** : 37–46, 2000
Charcot–Marie–Tooth disease, axonal, type 2F(CMT2F)	#606595	AD	7q11–q21	*HSPB1*	Heat–shock 27–kD protein 1	*602195	*Nat Genet* **36** : 602–606, 2004
Charcot–Marie–Tooth disease, axonal, type 2I(CMT2I)	#607677	AD	1q22–q23	*MPZ*	Myelin protein zero	*159440	*Neurology* **50** : 1397–1401, 1998
Charcot–Marie–Tooth disease, axonal, type 2J(CMT2J)	#607736	AD	1q22–q23	*MPZ*	Myelin protein zero	*159440	*Brain* **122** : 281–290, 1999
Charcot–Marie–Tooth disease, axonal, type 2K(CMT2K)	#607831	AR	8q21	*GDAP1*	Ganglioside–induced differentiation–associated protein 1	*606598	*Arch Neurol* **60** : 598–604, 2003
Charcot–Marie–Tooth disease, axonal, type 2L(CMT2L)	#608673	AD	12q24.3	*HSPB8*	Heat shock 22kDa protein 8	*608014	*Hum Genet* **116** : 222–224, 2005
Hypertrophic neuropathy of Dejerine–Sottas(HMSN3)	#145900	AD	17p11.2	*PMP22*	Peripheral myelin protein 22	*601097	*Nat Genet* **5** : 269–273, 1993
Hypertrophic neuropathy of Dejerine–Sottas(HMSN3)	#145900	AD	1q22	*MPZ*	Myelin protein zero(Charcot–Marie–Tooth neuropathy 1B)	*159440	*Nat Genet* **5** : 266–268, 1993
Hypertrophic neuropathy of Dejerine–Sottas(HMSN3)	#145900	AR	19q13.13–13.2	*PRX*	Periaxin	*605725	*Am J Hum Genet* **68** : 325–333, 2001

疾患			遺伝子				文献
疾患名	MIM	遺伝形式	染色体座	遺伝子記号	遺伝子名	MIM	
Hypertrophic neuropathy of Dejerine-Sottas (HMSN3)	#145900	AD	10q21.1-q22.1	*EGR2*	Early growth response 2	*129010	*Neurology* **52**: 1827-1832, 1999
Charcot-Marie-Tooth disease, type 4A (CMT4A)	#214400	AR	8q21	*GDAP1*	Ganglioside-induced differentiation-associated protein 1	*606598	*Nat Genet* **30**: 21-22, 2002
Charcot-Marie-Tooth disease, type 4B1 (CMT4B1)	#601382	AR	11q22	*MTMR2*	Myotubularin related protein 2	*603557	*Nat Genet* **25**: 17-19, 2000
Charcot-Marie-Tooth disease, type 4B2 (CMT4B2)	*604563	AR	11p15	*SBF2*	SET binding factor 2	*607697	*Am J Hum Genet* **72**: 1141-1153, 2003 *Hum Mol Genet* **12**: 349-356, 2003
Charcot-Marie-Tooth disease, type 4C (CMT4C)	#601596	AR	5q32	*SH3TC2*	SH3 domain and tetratricopeptide repeats 2	*608206	*Am J Hum Genet* **73**: 1106-1119, 2003
Charcot-Marie-Tooth disease, type 4D (CMT4D; Hereditary motor and sensory neuropathy, Lom type)	#601455	AR	8q24.3	*NDRG1*	N-myc downstream regulated 1	*605262	*Am J Hum Genet* **67**: 47-58, 2000
Charcot-Marie-Tooth disease, type 4F (CMT4F)	#145900	AR	19q13.13-13.2	*PRX*	Periaxin	*605725	*Hum Mol Genet* **10**: 415-421, 2001
Charcot-Marie-Tooth disease, type 4H (CMT4H)	#609311	AR	12p11.2	*FGD4*	FYVE, RhoGEF, and PH doamin-containing protein 4	*611104	*Am J Hum Genet* **81**: 1-16 & 158-164, 2007
Charcot-Marie-Tooth disease, type 4J (CMT4J)	#611228	AR	6q21	*FIG4*	FIG4 homolog (*S. cerevisiae*)	*609390	*Nature* **448**: 68-72, 2007
Slowed nerve conduction velocity, autosomal dominant	#608236	AD	8p23	*ARHGEF10*	RHO guanine nucleotide exchange factor (GEF) 10	*608136	*Am J Hum Genet* **73**: 926-932, 2003
Neuropathy, congenital hypomyelinating (CHN)	#605253	AD	1q22-q23	*MPZ*	Myelin protein zero	*159440	*Neuron* **17**: 451-460, 1996
Neuropathy, congenital hypomyelinating (CHN)	#605253	AR	10q21.1-q22.1	*EGR2*	Early growth response 2	*129010	*Nat Genet* **18**: 53-55, 1998
Giant axonal neuropathy 1 (GAN1)	#256850	AR	16q24.1	*GAN*	Gigaxonin	*605379	*Nat Genet* **26**: 370-374, 2000
46, XY gonadal dysgenesis, complete or partial, DHH-related	#233420	AR	12q13.1	*DHH*	Desert hedgehog homolog (*Drosophila*)	*605423	*Am J Hum Genet* **67**: 1302-1305, 2000
Hereditary neuropathy with liability to pressure palsies (HNPP)	#162500	AD	17p11.2	*PMP22*	Peripheral myelin protein 22	*601097	*Cell* **72**: 143-151, 1993
Peripheral neuropathy and sensorineural hearing impairment		AD	1p35.1	*GJB3*	Gap junction protein, beta 3, 31kDa (connexin 31)	+603324	*Hum Mol Genet* **10**: 947-952
Congenital cataracts facial dysmorphism neuropathy syndrome (CCFDN)	#604168	AR	18q23-qter	*CTDP1*	CTD (carboxy-terminal domain, RNA polymerase II, polypeptide A) phosphatase, subunit 1	*604927	*Nat Genet* **35**: 185-189, 2003
Navajo neurohepatopathy (NN)	#256810	AR	2p23-p21	*MPV17*	MpV17 mitochondrial inner membrane protein	*137960	*Nat Genet* **38**: 570-575, 2006
Indifference to pain, congenital, autosomal recessive	#243000	AR	2q24	*SCN9A*	Sodium channel, voltage-gated, type IX, alpha subunit	*603415	*Nature* **444**: 894-898, 2006
Paroxysmal extreme pain disorder	#167400	AR	2q24	*SCN9A*	Sodium channel, voltage-gated, type IX, alpha subunit	*603415	*Neuron* **52**: 767-774, 2006
遺伝性感覚・自律神経ニューロパチー							
Neuropathy, hereditary sensory and autonomic, type I (HSAN1; HSN1)	#162400	AD	9q22	*SPTLC1*	Serine palmitoyltransferase, long chain base subunit 1	*605712	*Nat Genet* **27**: 261-262, 2001
Neuropathy, hereditary sensory and autonomic, type II (HSAN2; HSN2)	#201300	AR	12p13.33	*WNK1*	WNK lysine deficient protein kinase 1	*605232	*Am J Hum Genet* **74**: 1064-1073, 2004
Neuropathy, hereditary sensory and autonomic, type III (HSAN3; Riley-Day syndrome, DYS)	#223900	AR	9q31	*IKBKAP*	Inhibitor of kappa light chain polypeptide gene enhancer in B-cells, kinase complex-associated protein	*603722	*Am J Hum Genet* **68**: 598-605, 2001
Insensitivity to pain, congenital, with anhidrosis (CIPA; HSAN4)	#256800	AR	1q21-q22	*NTRK1*	Neurotrophic tyrosine kinase, receptor, type 1	*191315	*Nat Genet* **13**: 485-488, 1996
Neuropathy, hereditary sensory and autonomic, type V (HSAN5)	#608654	AR	1p13.1	*NGF*	Nerve growth factor (beta polypeptide)	*162030	*Hum Mol Genet* **13**: 799-805, 2004
アミロイド・ニューロパチー							
Amyloidosis I	+176300	AD	18q11.2-q12.1	*TTR*	Transthyretin (prealbumin, amyloidosis type I)	+176300	*Biochem Biophy Res Commun* **123**: 921-928, 1984
Amyloid polyneuropathy-nephropathy, Iowa type (Amyloidosis, van Allen type, Amyloidosis IV, formerly)	*107680	AD	11q23	*APOA1*	Apolipoprotein A-I,	*107680	*Genomics* **8**: 318-323, 1990
Amyloidosis, Finnish type (Amyloidosis V, Lattice corneal dystrophy, type II)	#105120	AD	9q34	*GSN*	Gelsolin (amyloidosis, Finnish type)	*137350	*New Engl J Med* **325**: 1780-1785, 1991
Amyloidosis, primary cutaneous	#105250	AD	5p13.1	*OSMR*	Oncostatin M receptor	*601743	*Am J Hum Genet* **82**: 73-80, 2008
その他							
Amyotrophy, hereditary neuralgic (HNA)	#162100	AD	17q25	*SEPT9*	Septin 9	*604061	*Nat Genet* **37**: 1044-1046, 2005
筋肉疾患							
筋ジストロフィー関連疾患							
Muscular dystrophy, Duchenne types (DMD)	#310200	XR	Xp21.2	*DMD*	Dystrophin	*300377	*Nature* **323**: 646-650, 1986
Muscular dystrophy, Becker type (BMD)	#300376	XR	Xp21.2	*DMD*	Dystrophin	*300377	
Muscular dystrophy, limb-girdle, type 1A (LGMD1A)	#159000	AD	5q31	*MYOT*	Myotilin	*604103	*Hum Mol Genet* **9**: 2141-2147, 2000
Muscular dystrophy, limb-girdle, type 1B (LGMD1B)	#159001	AD	1q21.2	*LMNA*	Lamin A/C	*150330	*Hum Mol Genet* **9**: 1453-1459, 2000
Muscular dystrophy, limb-girdle, type 1C (LGMD1C)	#607801	AD	3p25	*CAV3*	Caveolin 3	*601253	*Nature Genet* **18**: 365-368, 1998 *Hum Mol Genet* **7**: 871-877, 1998
Muscular dystrophy, limb-girdle, type 2A (LGMD2A)	#253600	AR	15q15.1-q21.1	*CAPN3*	Calpain 3, (p94)	*114240	*Cell* **81**: 27-40, 1995
Muscular dystrophy, limb-girdle, type 2B (LGMD2B)	#253601	AR	2p13.3-p13.1	*DYSF*	Dysferlin, limb girdle muscular dystrophy 2B (autosomal recessive)	*603009	*Nat Genet* **20**: 37-42, 1998
Muscular dystrophy, limb-girdle, type 2C (LGMD2C)	#253700	AR	13q12	*SGCG*	Sarcoglycan, gamma (35kDa dystrophin-associated glycoprotein)	*608896	*Science* **270**: 819-821, 1995
Muscular dystrophy, limb-girdle, type 2D (LGMD2D)	#608099	AR	17q12-q21.33	*SGCA*	Sarcoglycan, alpha (50kDa dystrophin-associated glycoprotein)	*600119	*Cell* **78**: 625-633, 1994
Muscular dystrophy, limb-girdle, type 2E (LGMD2E)	#604286	AR	4q12	*SGCB*	Sarcoglycan, beta (43kDa dystrophin-associated glycoprotein)	*600900	*Nat Genet* **11**: 257-265, 1995 *Nat Genet* **11**: 266-273, 1995

疾患			遺伝子				文献
疾患名	MIM	遺伝形式	染色体座	遺伝子記号	遺伝子名	MIM	
Muscular dystrophy, limb-girdle, type 2F (LGMD2F)	#601287	AR	5q33	*SGCD*	Sarcoglycan, delta(35kDa dystrophin-associated glycoprotein)	*601411	*Nat Genet* **14**:195-198, 1996
Muscular dystrophy, limb-girdle, type 2G (LGMD2G)	#601954	AR	17q12	*TCAP*	Titin-cap(telethonin)	*604488	*Nat Genet* **24**:163-166, 2000
Muscular dystrophy, limb-girdle, type 2H (LGMD2H)	#254110	AR	9q31-q34.1	*TRIM32*	Tripartite motof-containong 32	*602290	*Am J Hum Genet* **70**:663-672, 2002
Muscular dystrophy, limb-girdle, type 2I (LGMD2I)	#607155	AR	19q13.3	*FKRP*	Fukutin-related protein	*606596	*Hum Mol Genet* **10**:2851-2859, 2001
Muscular dystrophy, limb-girdle, type 2J (LGMD2J)	#608807	AR	2q31	*TTN*	Titin	+188840	*Am J Hum Genet* **71**:492-500, 2002
Muscular dystrophy, limb-girdle, type 2K (LGMD2K)	#609308	AR	9q34.1	*POMT1*	Protein-O-mannosyltransferase 1	*607423	*Neuromusc Disord* **15**:271-275, 2005
Muscular dystrophy, limb-girdle, type 2M (LGMD2M)	#611588	AR	9q31	*FCMD*	Fukutin	*607440	*Ann Neurol* **60**:603-610, 2006
Miyoshi(三好)myopathy(MM)	#254130	AR	2p13.3-p13.1	*DYSF*	Dysferlin, limb girdle muscular dystrophy 2B(autosomal recessive)	*603009	*Nat Genet* **20**:31-36, 1998
Oculopharyngeal muscular dystrophy (OPMD)	#164300	AD	14q11.2-q13	*PABPN1*	Poly(A)binding protein, nuclear 1	*602279	*Nat Genet* **18**:164-167, 1998
Emery-Dreifuss muscular dystrophy, X-linked(EDMD)	#310300	XR	Xq28	*EMD*	Emerin	*300384	*Nat Genet* **8**:323-327, 1994
Emery-Dreifuss muscular dystrophy, autosomal dominant(EDMD2)	#181350	AD	1q21.2-q21.3	*LMNA*	Lamin A/C	*150330	*Nat Genet* **21**:285-288, 1999
Emery-Dreifuss muscular dystrophy, autosomal recessive(EDMD3)	#604929	AR	1q21.2-q21.3	*LMNA*	Lamin A/C	*150330	*Am J Hum Genet* **66**:1407-1412, 2000
Epidermolysis bullosa simplex and limb-girdle muscular dystrophy(MD-EBS)	#226670	AR	8q24	*PLEC1*	Plectin 1, intermediate filament binding protein, 500kDa	*601282	*Nat Genet* **13**:450-457, 1996 *Hum Mol Genet* **5**:1539-1546, 1996
Facioscapulohumeral muscular dystrophy 1A(FSHMD1A)	#158900	AD	4q35	*D4Z4*	(*DUX4*; Double homeobox protein 4)	*606009	*Nat Genet* **2**:26-30, 1992 *Hum Mol Genet* **3**:1287-1295, 1994
Muscular dystrophy, congenital merosin-deficient, 1A(MDC1A)	#607855	AR	6q22-q23	*LAMA2*	Laminin, alpha 2	*156225	*Nat Genet* **11**:216-218, 1995
Muscular dystrophy, congenital, 1C (MDC1C)	#606612	AR	19q13.3	*FKRP*	Fukutin-related protein	*606596	*Am J Hum Genet* **69**:1198-1209, 2001
Muscular dystrophy, congenital, type 1D (MDC1D)	#608840	AR	22q12.3-q13.1	*LARGE*	Like-glycosyltransferase	*603590	*Hum Mol Genet* **12**:2853-2861, 2003
Rigid spine muscular dystrophy 1 (RSMD1)	#602771	AR	1p36.13	*SEPN1*	Selenoprotein N, 1	*606210	*Nat Genet* **29**:17-18, 2001
Fukuyama(福山)congenital muscular dystrophy(FCMD)	#253800	AR	9q31	*FCMD*	Fukutin	*607440	*Nature* **394**:388-392, 1998
Walker-Warburg syndrome(WWS)	#236670	AR	9q34.1	*POMT1*	Protein-O-mannosyltransferase 1	*607423	*Am J Hum Genet* **71**:1033-1043, 2002
Muscle-eye-brain disease(MEB)	#253280	AR	1p32-p33	*POMGNT1*	Protein O-mannose beta-1, 2-N-acetylglucosamyltransferase	*606822	*Dev Cell* **1**:7171-724, 2001
Hereditary rippling muscle disease (RMD)	#606072	AD	3p25	*CAV3*	Caveolin 3	+601253	*Nat Genet* **28**:218-219, 2001
Tibial muscular dystrophy, tardive (TMD)	#600334	AD	2q24.3	*TTN*	Titin	+188840	*Am J Hum Genet* **71**:492-500, 2002
先天性ミオパチー・その他のミオパチー							
Muscle hypertrophy	+601788	AD	2q32.1	*MSTN*	Myostatin	+601788	*New Engl J Med* **350**:2682-2688, 2004
Congenital myopathy		AR	12q13	*ITGA7*	Integrin, alpha 7	*600536	*Nat Genet* **19**:94-97, 1998
Bethlem myopathy	#158810	AD	21q22.3	*COL6A1*	Collagen, type VI, alpha 1	*120220	*Nat Genet* **14**:113-115, 1996
Bethlem myopathy	#158810	AD	21q22.3	*COL6A2*	Collagen, type VI, alpha 2	*120240	*Nat Genet* **14**:113-115, 1996
Bethlem myopathy	#158810	AD	2q37	*COL6A3*	Collagen, type VI, alpha 3	*120250	*Hum Mol Genet* **7**:807-812, 1998
Ullrich congenital muscular dystrophy (UCMD)	#254090	AR	21q22.3	*COL6A1*	Collagen, type VI, alpha 1	*120220	*Am J Hum Genet* **73**:355-369, 2003
Ullrich congenital muscular dystrophy (UCMD)	#254090	AR	21q22.3	*COL6A2*	Collagen, type VI, alpha 2	*120240	*Proc Natl Acd Sci* **98**:7516-7521, 2001 *Ann Neurol* **50**:261-265, 2001
Ullrich congenital muscular dystrophy (UCMD)	#254090	AR	2q37	*COL6A3*	Collagen, type VI, alpha 3	*120250	*Am J Hum Genet* **70**:1446-1458, 2002
Barth syndrome(BTHS)	#302060	XR	Xq28	*TAZ*	Tafazzin(cardiomyopathy, dilated 3A(X-linked); endocardial fibloelastosis 2; Barth syndrome)	*300394	*Nat Genet* **13**:385-399, 1996
Nemaline myopathy, 1(NEM1)	#609284	AD	1q22-q23	*TPM3*	Toropmyosin 3	*191030	*Nat Genet* **9**:75-79, 1995
Nemaline myopathy, 2(NEM2)	#256030	AR	2q22	*NEB*	Nebulin	*161650	*Proc Natl Acd Sci* **96**:2305-2310, 1999
Nemaline myopathy, 3(NEM3)	#161800	AD	1q42.1	*ACTA1*	Actin, alpha 1, skeletal muscle	+102610	*Nat Genet* **23**:208-212, 1999
Nemaline myopathy, 4(NEM4, CAP myopathy)	#609285	AD	9p13.2-p13.1	*TPM2*	Tropomyosin 2(beta)	*190990	*Neuromusc Disord* **12**:151-158, 2002
Nemaline myopathy, 5(NEM5)	#605355	AR	19q13.4	*TNNT1*	Troponin T1(skeletal, slow)	*191041	*Am J Hum Genet* **67**:814-821, 2000
Nemaline myopathy, 7(NEM7)	#610687	AR	14q12	*CFL2*	Cofilin 2(muscle)	*601443	*Am J Hum Genet* **80**:162-167, 2007
Myopathy, congenital, with fiber-type disproportion(CFTD)	#255310	AR	1q42.1	*ACTA1*	Actin, alpha 1, skeletal muscle	+102610	*Ann Neurol* **56**:689-694, 2004
		AR	1p36.13	*SEPN1*	Selenoprotein N, 1	*606210	*Ann Neurol* **59**:546-552, 2006
Central core disease of muscle(CCD)	#117000	AD	19q13.1	*RYR1*	Ryanodine receptor 1(skeletal)	*180901	*Nat Genet* **5**:46-50, 1993 *Nat Genet* **5**:51-55, 1993
Myotubular myopathy 1(MTM1)	#310400	XR	Xq28	*MTM1*	Myotubularin 1	*310415	*Nat Genet* **13**:175-182, 1996
Myopathy, centronuclear, autosomal dominant	#160150	AD	19p13.2	*DNM2*	Dynamin 2	*602378	*Nat Genet* **37**:1207-1209, 2005
Myopathy, centronuclar, autosomal recessive	#255200	AR	2q14	*BIN1*	Bridging Integrator 1 (Amphiphysin 2)	*601248	*Nat Genet* **39**:1134-1139, 2007
Myopathy, myofibrillar, desmin-related	#601419	AD/AR	2q35	*DES*	Desmin	*125660	*Nat Genet* **19**:402-403, 1998
Alpha-b crystallinopathy	#601419	AD	11q22.3-q23.1	*CRYAB*	Crystallin, alpha B	+123590	*Nat Genet* **20**:92-95, 1998
Filaminopathy, autosomal dominant	#609524	AD	7q32.1	*FLNC*	Filamin C	*102565	*Am J Hum Genet* **77**:297-304, 2005
Myoneurogastrointestinal encephalomyopathy syndrome (MNGIE)	#603041	AR	22q13	*TYMP*	thymidine phosphorylase	*131222	*Science* **283**:689-692, 1999
Hereditary inclusion body myopathy 2 (IBM2)	#600737	AR	9p12-p13	*GNE*	Glucosamine(UDP-N-acetyl)-2-epimerase/N-acetylmannosamine kinase	*603824	*Nat Genet* **29**:83-87, 2001
Inclusion body myopathy 3, autosomal dominant(IBM3)	#605637	AD	17p13.1	*MYH2*	Myosin, heavy chain 2, skeletal muscle, adult	*160740	*Proc Natl Acad Sci* **97**:14614-14619, 2000
Inclusion body myopathy with early-onset Paget disease and frontotemporal dementia(IBMPED)	#167320	AD	9p21.1-p12	*VCP*	Valosin-containing protein	*601023	*Nat Genet* **36**:377-381, 2004

疾患			遺伝子				文献
疾患名	MIM	遺伝形式	染色体座	遺伝子記号	遺伝子名	MIM	
Myopathy, myosin storage	#608358	AD	14q12	*MYH7*	Myosin, heavy polypeptide 7, cardiac muscle, beta	+160760	*Ann Neurol* **54**：494–500, 2003 *Neurology* **61**：1519–1523, 2003
悪性高熱症候群							
Malignant hyperthermia susceptibility 1（MHS1）	#145600	AD	19q13.1	*RYR1*	Ryanodine receptor 1（skeletal）	*180901	*Genomics* **11**：751–755,1 991
Malignant hyperthermia susceptibility 5（MHS5）	#601887	AD	1q32	*CACNA1S*	Calcium channel, voltage–dependent, L type, alpha 1S subunit	*114208	*Am J Hum Genet* **60**：1316–1325, 1997
筋強直（ミオトニー）性疾患・その他							
Dystrophia myotonica（DM, Steinert disease）	#160900	AD	19q13.2–q13.3	*DMPK*	Dystrophia myotonica–protein kinase	*605377	*Nature* **355**：545–546, 1992 *Nature* **355**：547–548, 1992 *Nature* **355**：548–551, 1992 *Science* **255**：1256–1258, 1992 *Cell* **68**：799–808, 1992
Myotonic dystrophy type 2（DM2）	#602668	AD	3q13.3–q24	*CNBP*	CCHC–type zinc finger, nucleic acid binding protein	*116955	*Science* **293**：864–867, 2001
Autosomal dominant myotonia congenita（Thomsen's disease）	#160800	AD	7q35	*CLCN1*	Chloride channel 1, skeletal muscle（Thomsen disease, autosomal dominant）	*118425	*Nat Genet* **3**：305–309, 1993
Autosomal recessive myotonia congenita（MCR, Becker disease, Generalized myotonia）	#255700	AR	7q35	*CLCN1*	Chloride channel 1, skeletal muscle（Thomsen disease, autosomal dominant）	*118425	*Science* **257**：797–800, 1992
Paramyotonia congenita of von Eulenburg（PMC）	#168300	AD	17q23.1–q25.3	*SCN4A*	Sodium channel, voltage–gated, type IV, alpha polypeptide	+603967	*Nat Genet* **2**：148–152, 1992
Schwartz–Jampel syndrome, type 1（SJS1, chondrodystrophic myotonia）	#255800	AR	1p36.1–p35	*HSPG2*	Heparan sulfate proteoglycan 2（perlecan）	*142461	*Nat Genet* **26**：480–483, 2000
Schwartz–Jampel syndrome, type 2（SJS2）/Stüve–Wiedermann syndrome（SWS）	#601559	AR	5p13.1	*LIFR*	Leukemia inhibitory factor receptor alpha	*151443	*J Hum Genet* **74**：298–305, 2004
Crisponi syndrome	#601378	AR	19p12	*CRLF1*	Cytokine receptor–like factor 1	*604237	*Am J Hum Genet* **80**：971–981, 2007
先天性筋無力症							
Slow–channel congenital myasthenic syndrome（SCCMG, Cingenital myasthenic syndrome, type II）	#601462	AD	2q24–q32	*CHRNA1*	Cholinergic receptor, nicotinic, alpha polypeptide 1（muscle）	*100690	*Neuron* **15**：229–239, 1995
Slow–channel congenital myasthenic syndrome（SCCMG, Cingenital myasthenic syndrome, type II）	#601462	AD	17p12–p11	*CHRNB1*	Cholinergic receptor, nicotinic, beta polypeptide 1（muscle）	*100710	*Ann Neurol* **39**：712–723, 1996 *Hum Mol Genet* **5**：1217–1227, 1996
Slow–channel congenital mysthenic syndrome（SCCMS）	#601462	AR	2q33–q34	*CHRND*	Cholinergic receptor, nicotinic, delta polypeptide	*100720	*Ann Neurol* **51**：102–112, 2002
Slow–channel congenital myasthenic syndrome（SCCMG, Cingenital myasthenic syndrome, type II）	#601462	AD	17p13–p12	*CHRNE*	Cholinergic receptor, nicotinic, epsilon polypeptide	*100725	*Proc Natl Acad Sci* **92**：758–762, 1995
Fast–channel myasthenic syndrome and arthrogryposis multiplex congenita	#608930	AR	2q33–q34	*CHRND*	Cholinergic receptor, nicotinic, delta polypeptide	*100720	*J Clin Invest* **108**：125–130. 2001
Endplate acetylcholinesterase deficiency（EAD, Congenital myasthenic syndrome, type Ic）	#603034	AR	3p24.2	*COLQ*	Collagen–like tail subunit（single strand of homotrimer）of asymmetric acethylcholinesterase	*603033	*Am J Hum Genet* **63**：967–975, 1998
Congenital myasthenic syndrome caused by sodium channel SCN4A	+603967	AR/AD	17q23.1–q25.3	*SCN4A*	Sodium channel, voltage–gated, type IV, alpha subunit	+603967	*Proc Natl Acad Sci* **100**：7377–7382, 2003
Congenital myasthenic syndrome	#608931	AR	9q31.3–q32	*MUSK*	Muscle, skeletal, receptor tyrosine kinase	*601296	*Hum Mol Genet* **13**：3229–3240, 2004
Myasthenia, limb–girdle, familial	#254300	AR	4p16.2	*DOK7*	Docking protein 7	*610285	*Science* **313**：1975–1978, 2006
眼筋ミオパチー・眼筋麻痺							
Congenital fibrosis of the extraocular muscles type1（CFEOM1）	#135700	AD	12q12	KIF21A	Kinesin family member 21A	*608283	Nat Genet **35**：318–321,2003
Congenital fibrosis of the extraocular muscles type 2（CFEOM2）	#602078	AR	11q13.2	*PHOX2A*	Paired–like（aristaless）homeobox 2a	*602753	*Nat Genet* **29**：315–320, 2001
Progressive external ophthalmoplegia（PEO）	#609286	AD	10q24	*C10orf2*	Chromosome 10 open reading frame 2	*606075	*Nat Genet* **28**：223–231, 2001
Progressive external ophthalmoplegia（PEO）	#157640	AD, AR	15q24	*POLG*	Polymerase（DNA directed）, gamma	*174763	*Nat Genet* **28**：211–212, 2001
Duane–radial ray syndrome（DRRS, Okihiro syndrome）	#607323	AD	20q13.13–q13.2	*SALL4*	Sal–like 4（*Drosophila*）	*607343	*Am J Hum Genet* **71**：1195–1199, 2002 *Hum Mol Genet* **11**：2979–2987, 2002
発作性・周期性疾患							
周期性四肢麻痺							
Periodic paralysis I（Hypokalemic periodic paralysis, HOKPP）	#170400	AD	1q32	*CACNA1S*	Calcium channel, voltage–dependent, L type, alpha 1S subunit	*114208	*Cell* **77**：863–868, 1994
Hypokalrmic periodic paralysis（HOKPP）	#170400	AD	11q13–q14	*KCNE3*	Potassium voltage–gated channel, Isk–related family, member 3	*604433	*Cell* **104**：217–231, 2001
Hypokalemic periodic paralysis type 2	#170400	AD	17q23.1–q25.3	*SCN4A*	Sodium channel, voltage–gated, type IV, alpha subunit	+603967	*Neurology* **53**：1932–1936, 1999
Periodic paralysis II（Hyperkalemic periodic paralysis, HYPP, Gamstorp disease）	#170500	AD	17q23.1–q25.3	*SCN4A*	Sodium channel, voltage–gated, type IV, alpha subunit	+603967	*Cell* **67**：1021–1027, 1991 *Nature* **354**：387–389, 1991
Andersen's syndrome	#170390	AD	17q23	*KCNJ2*	Potassium inwardly–rectifying channel, subfamily J, number 2	*600681	*Cell* **105**：511–519, 2001
Alternating hemiplegia of childhood	#104290	AD	1q21–q23	*ATP1A2*	ATPase, Na^+/K^+ transporting, alpha 2（+）polypeptide	*182340	*Ann Neurol* **55**：884–887, 2004
片（偏）頭痛							
Familial hemiplegic migraine 1（FHM1, MHP1）	#141500	AD	19p13	*CACNA1A*	Calcium channel, voltage–dependent, P/Q type, alpha 1A subunit	*601011	*Cell* **87**：543–552, 1996
Familial hemiplegic migraine 2（FHM2）	#602481	AD	1q21–q23	*ATP1A2*	ATPase, Na^+/K^+ transporting, alpha 2（+）polypeptide	*182340	*Nat Genet* **33**：192–196, 2003
Familial hemiplegic migraine 3（FHM3）	#609634	AD	2q24	*SCN1A*	Sodium channel, voltage–gated, type I, alpha subunit	*182389	*Lancet* **366**：371–377, 2005
周期性（発作性）失調症							
Episodic ataxia, type 2（EA2）	#108500	AD	19p13	*CACNA1A*	Calcium channel, voltage–dependent, P/Q type, alpha 1A subunit	*601011	*Cell* **87**：543–552, 1996

疾患			遺伝子				文献
疾患名	MIM	遺伝形式	染色体座	遺伝子記号	遺伝子名	MIM	
Myokymia with periodic ataxia(Episodic ataxia type 1, EA1)	#160120	AD	12p13	*KCNA1*	Porassium voltage-gated channel, shaker-related subfamily, member 1(episodic ataxia with myokymia)	*176260	*Nat Genet* **8**:136-140, 1994
驚愕病							
Kok disease(hyperekplexia; startle disease)	#149400	AD	5q32	*GLRA1*	Glycine receptor, alpha 1	*138491	*Nat Genet* **5**:351-358, 1993
Hyperkplexia	#149400	AD	4q31.3	*GLRB*	Glycine receptor, beta	*138492	*Hum Mol Genet* **11**:853-860, 2002
Hyperekplexia, hereditary	#149400	AD	11p15.2-p15.1	*SLC6A5*	Solute carrier family 6, member 5	*604159	*Nat Genet* **38**:801-806, 2006
てんかん							
Nocturnal frontal lobe epilepsy, type 1 (ENFL1)	#600513	AD	20q13.2-q13.3	*CHRNA4*	Cholinergic receptor, nicotinic, alpha 4	*118504	*Nat Genet* **11**:201-203, 1995
Autosomal dominant nocturnal frontal lobe epilepsy type 3(ENFL3)	#605375	AD	1p21	*CHRNB2*	Cholinergic receptor, nicotinic, bata 2(neuronal)	*118507	*Nat Genet* **26**:275-276, 2000
Benign neonatal epilepsy 1(EBN1)	#121200	AD	20q13.3	*KCNQ2*	Potassium voltage-gated channel, KQT-like subfamily, member 2	*602235	*Science* **279**:403-406, 1998 *Nat Genet* **18**:25-29, 1998
Benign neonatal epilepsy 2(EBN2)	#121201	AD	8q24	*KCNQ3*	Potassium voltage-gated channel, KQT-like subfamily, member 3	*602232	*Nat Genet* **18**:53-55, 1998
Familial febrile convulsions 4(FEB4)	#604352	AD	5q14	*GPR98*	G protein-coupled receptor 98	*602851	*Ann Neurol* **52**:654-657, 2002
Myokymia and neonatal epilepsy	#606437	AD	20q13.3	*KCNQ2*	Potassium voltage-gated channel, KQT-like subfamily, member 2	*602235	*Proc Natl Acad Sci* **98**:12272-12277, 2001
Generalized epilepsy with febrile seizures plus(GEFS$^+$)	#604233	AD	19q13	*SCN1B*	Sodium channel, voltage-gated, type I, beta	*600235	*Nat Genet* **19**:366-370, 1998
Generalized epilepsy with febrile seizures plus, type 2(GEFSP2)	#604233	AD	2q24	*SCN1A*	Sodium channel, voltage-gated, type I, alpha subunit	*182389	*Nat Genet* **24**:343-456, 2000
Generalized epilepsy with febrile seizures plus(GEFS$^+$type 3)/Childhood absence epilepsy and febrile seizures	#604233	AD	5q34	*GABRG2*	Gamma-aminobutylic acid (GABA)A receptor, gamma 2	*137164	*Nat Genet* **28**:46-48, 2001 *Nat Genet* **28**:49-52, 2001
Febrile seizures associated with afebrile seizures	#607745	AD	2q22-q23	*SCN2A1*	Sodium channel, voltage-gated, type II, alpha 1 polypeptide	*182390	*Proc Natl Acad Sci* **98**:6384-6389, 2001
Autosomal dominant partial epilepsy with auditory features	#600512	AD	10q24	*LGI1*	Leucine-rich, glioma inactivated 1	*604619	*Nat Genet* **30**:335-341, 2002 *Hum Mol Genet* **11**:1119-1128, 2002
Epilepsy, childhood absence, 1(ECA1)	%600131	AD	8q24				
Epilepsy, juvenile absence, 1(JCA1)	#607631	AD					
Epilepsy, juvenile myoclonic(JME)	#606904	AD	3q26-qter	*CLCN2*	Chloride channel 2	*600570	*Nat Genet* **33**:527-532, 2003(retracted)
Epilepsy with grand mal seizures on awakening(EGMA)	#607628	AD	3q26-qter	*CLCN2*	Chloride channel 2	*600570	*Nat Genet* **41**:954-955, 2009
Generalized epilepsy and paroxysmal dyskinesia	#609446	AD	10q22.3	*KCNMA1*	Potassium channel, calcium-activated, large conductance, subfamily M, alpha member 1	*600150	*Nat Genet* **37**:733-738, 2005
Amish infantile epilepsy syndrome	#609056	AR	2p11.2	*SIAT9*	Sialyltransferase 9	*604402	*Nat Genet* **36**:1225-1229, 2004
Epileptic encephalopathy, early infantile, 1	#308350	XR	Xp22.13	*ARX*	Aristaless related homeobox	*300382	*Nat Genet* **30**:441-445, 2002 *Hum Mol Genet* **11**:981-991, 2002 *Neurology* **59**:348-356, 2002
Myoclonus epilepsy of Unverricht and Lundborg(EPM1)	#254800	AR	21q22.3	*CSTB*	Cystatin B(stefin B)	*601145	*Science* **271**:1731-1734, 1996
Progressive myoclonus epilepsy 2 (EPM2A, Lafora disease)	#254780	AR	6q24	*EPM2A*	Epilepsy, progressive myoclonus 2, Lafora disease(laforin)	*607566	*Nat Genet* **20**:358-361, 1998
Lafora disease	#254780	AR	6p22.3	*NHLRC1*	NHL repeat containing 1	*608072	*Nat Genet* **35**:125-127, 2003
Epilepsy, progressive myoclonic(EPM3)	#611726	AR	7q11.21	*KCTD7*	Potassium channel tetramerisation domain containing 7	*611725	*Ann Neurol* **61**:579-586, 2007
Myoclonic epilepsy, juvenile, 1(EJM1)	#254770	AR	6p12-p11	*EFHC1*	EF hand domain(C-terminal)-containing 1	*608815	*Nat Genet* **36**:842-849, 2004
Juvenile myoclonic epilepsy	#606904	AD	2q22-q23	*CACNB4*	Calcium channel, voltage-dependent, beta 4 subunit	+601949	*Am J Hum Genet* **66**:1531-1639, 2000
Juvenile myoclonic epilepsy	#606904	AD	5q34-q35	*GABRA1*	Gamma-aminobutyric acid (GABA)A receptor, alpha 1	*137160	*Nat Genet* **31**:184-189, 2002
その他							
Familial Mediterranean fever(MEFV)	#249100	AR	16p13	*MEFV*	Mediterranean fever	*608107	*Cell* **90**:797-807, 1997 *Nat Genet* **17**:25-31, 1997
視床下部・自律神経障害・睡眠障害							
Wolfram syndrome(WFS, Diabetes inspidus and mellitus with optic atrophy and deafness)	#222300	AR	4p16.1	*WFS1*	Wolfram syndrome 1(wolframin)	*606201	*Nat Genet* **20**:143-148, 1998 *Hum Mol Genet* **7**:2021-2028, 1998
Wolfram syndrome 2	#604928	AR	4q22-q24	*CISD2*	CDGSH iron sulfur domain protein 2	*611507	*Am J Hum Genet* **81**:673-684, 2007
Kallmann syndrome 1(KAL1)	+308700	XR	Xp22.3	*KAL1*	Kallmann syndrome 1 sequence	+308700	*New Engl J Med* **326**:1752-1755, 1992 *Proc Nat Acad Sci* **89**:8190-8194, 1992
Kallmann syndrome 2(KAL2)	#147950	AD	8p11.2-p11.1	*FGFR1*	Fibroblast growth factor receptor 1(fms-related tyrosine kinase 2, Pfeiffer syndrome)	*136350	*Nat Genet* **33**:463-465, 2003
Familial advanced sleep phase syndrome (FASPS)	#604348	AD	2q37.3	*PER2*	Period homolog 2(Drosophila) [KIAA0347]	*603426	*Science* **291**:1040-1043, 2001 *Am J Hum Genet* **68**:753-758, 2001
Congenital failure of autonomic control (Ondine cruse, Congenital central hypoventilation syndrome, CCHS)	#209880	AR	20q13.2-q13.3	*EDN3*	Endothelin 3	*131242	*Nat Genet* **13**:395-396, 1996
Congenital failure of autonomic control (Ondine cruse; Congenital central hypoventilation syndrome, CCHS)	#209880	AR	10q11.2	*RET*	Ret proto-oncogene	+164761	*Am J Hum Genet* **62**:715-717, 1998
Congenital failure of autonomic control (Ondine cruse; Congenital central hypoventilation syndrome, CCHS)	#209880	AR	5p13.1-p12	*GDNF*	Glial cell derived neurotrophic factor	*600837	*Am J Hum Genet* **62**:715-717, 1998
Congenital central hypoventilation syndrome	#209880	*de novo*	4p12	*PHOX2B*	Paired-like homeobox 2B	*603851	*Nat Genet* **33**:459-461, 2003
Pitt-Hopkins Syndrome	#610954	AR	18q21.1	*TCF4*	Transcription Factor 4	*602272	*Am J Hum Genet* **80**:988-993, 2007 *Am J Hum Genet* **80**:994-1001, 2007

疾患			遺伝子				文献
疾患名	MIM	遺伝形式	染色体座	遺伝子記号	遺伝子名	MIM	
Hirschsprung disease associated with microcephaly, mental retardation, hypertelorism, subcutaneous cleft palate and short stature (Mowat–Wilson syndrome)	#235730	*de novo*	2q22	*SIP1*	Survival of motor neuron protein interracting protein 1	*602595	*Nat Genet* **27** : 369–370, 2001
Triple A syndrome	#231550	AR	12q13	*AAAS*	AAAS gene	*605378	*Poc Natl Acd Sci* **103** : 2298–2303, 2006
X-linked chronic idiopathic intestinal pseudo-obstruction with central nervous system involvement	#300048	XR	Xq28	*FLNA*	Filamin A	*300017	*Am J Hum Genet* **80** : 751–758, 2007
中枢神経系の発生異常・奇形							
Pituitary anomalies and holoprosencephaly-like features (HPE9)	#610829	AD	2q14	*GLI2*	GLI-Kruppel family member GLI2	*165230	*Pro Natl Acad Sci* **100** : 13424–13429, 2003
Polymicrogyria, bilateral frontoparietal	#606854	AR	16q13	*GPR56*	G protein-coupled receptor 56	*604110	*Science* **303** : 2033–2036, 2004
Schizencephaly	#269160	*de novo*/AD	10q26.1	*EMX2*	*empty spiracle* (*Drosophila*) homolog 2	*600035	*Nat Genet* **12** : 94–96, 1996
Holoprosencephaly 2, alobar or semilobar (HPE2)	#157170	*de novo*/AD	2p21	*SIX3*	*sine oculis* homeobox (*Drosophila*) homolog 3	*603714	*Nat Genet* **22** : 196–198, 1999
Holoprosencephaly 3 (HPE3)	#142945	AD	7q36	*SHH*	*sonic hedgehog* (*Drosophila*) homolog	*600725	*Nat Genet* **14** : 353–356, 1996 *Nat Genet* **14** : 357–360, 1996
Holoprosencephaly 4 (HPE4)	#142946	*de novo*	18p11.3	*TGIF*	TGFB-induced factor (TALE family homeobox)	*602630	*Nat Genet* **25** : 205–208, 2000
Holoprosencephaly 5 (HPE5)	#609637	AD	13q32	*ZIC2*	Zic family member 2 (*odd-paired Drosophila* homolog)	*603073	*Nat Genet* **20** : 180–183, 1998
Miller–Dieker lissencephaly syndrome (MDLS)	#247200	AR	17p13.3	*PAFAH1B1*	Platelet-activating factor acetylhydrolase, isoform Ib, alpha subunit 45kD	*601545	*Nature* **364** : 717-721, 1993 *Nature* **370** : 216–218, 1994
Norman–Roberts type lissencephaly syndrome	#257320	AR	7q22	*RELN*	Reelin	*600514	*Nat Genet* **26** : 93–96, 2000
Lissencephaly, X-linked, 2 (LISX2)	#300215	XR	Xp22.13	*ARX*	Aristaless related homeobax	*300382	*Nat Genet* **32** : 359–369, 2002
Corpus callosum, agenesis of, with abnormal genitalia	#300004	XR	Xp22.13	*ARX*	Aristaless related homeobax	*300382	*Hum Mutat* **23** : 147–159, 2004
X-linked lissencephaly/Double cortex syndrome	#300067	XD	Xq22.3–q23	*DCX*	Doublecortex, lissencephaly, X-linked (doublecortin)	*300121	*Cell* **92** : 51–61, 1998 *Cell* **92** : 63–72, 1998
Familial porencephaly	#175780	AD	13q34	*COL4A1*	Collagen, type IV, alpha-1	*120130	*Science* **308** : 1167–1171, 2005
Autosomal recessive periventricular heterotopia with microcephaly	#608097	AR	20q13.13	*ARFGEF2*	ADP-ribosylation factor guanine nucleotide-exchange factor 2 (brefeldin A-inhibited)	*605371	*Nat Genet* **36** : 69–76, 2004
Periventricular heterotopia	#300049	XD	Xq28	*FLNA*	Filamin A, alpha (actin-binding protein-280)	*300017	*Neuron* **21** : 1315–1325, 1998
Heterotopia, periventricular, autosomal recessive	#608097	AR	20q13.13	*ARFGEF2*	ADP-ribosylation factor guanine nucleotide exchange factor 2	*605371	*Nat Genet* **36** : 69–76, 2004
Cortical dysplasia-focal epilepsy syndrome	#610042	AR	7q35–q36	*CNTNAP2*	Contactin-associater protein-like 2	*604569	*New Engl J Med* **354** : 1370–1377, 2006
Hydrocephalus due to congenital stenosis of aqueduct of Sylvius (HSAS1)	#307000	XR	Xq28	*L1CAM*	L1 cell adhesion molecule	*308840	*Nat Genet* **2** : 107–112, 1992
Knobloch syndrome (KNO)	#267750	AR	21q22.3	*COL18A1*	Collagen, type XVIII, alpha 1	*120328	*Hum Mol Genet* **9** : 2051–2058, 2000
Sotos syndrome	#117550	Sporadic	5q35	*NSD1*	Nuclear receptor binding SET domain protein 1	*606681	*Nat Genet* **30** : 365–366, 2002
Primary autosomal recessive microcephaly 1 (MCPH1)	#251200	AR	8p23	*MCPH1*	Microcephaly, primary autosomal recessive 1	*607117	*Am J Hum Genet* **71** : 136–142, 2002
Primary autosomal recessive microcephaly 5 (MCPH5)	#608716	AR	1q31	*ASPM*	Asp (abnormal spindle)-like, microcephaly associated (*Drosophila*)	*605481	*Nat Genet* **32** : 316–320, 2002
Seckel syndrome (SCKL1)	#210600	AR	3q22.1–q24	*ATR*	Ataxia telangiectasia and Rad3 related	*601215	*Nat Genet* **33** : 497–501, 2003
Bosley–Salih–Alorainy syndrome	#601536	AR	7p15.3	*HOXA1*	Homeobox A1	*142955	*Nat Genet* **37** : 1035–1037, 2005
Joubert syndrome 3	#608629	AR	6q23.3	*AHI1*	Abelson helper integration site 1	*608894	*Nat Genet* **36** : 1008–1013, 2004
Joubert syndrome 5	#610188	AR	12q21.3	*CEP290*	Centrosomal Protein, 290-kD	*610142	*Nat Genet* **38** : 623–625, 2006 *Nat Genet* **38** : 671–684, 2006
Joubert syndrome 7	#611560	AR	16q12.2	*RPGRIP1L*	RPGRIP1-Like	*610937	*Nat Genet* **39** : 875–881, 2007 *Nat Genet* **39** : 882–888, 2007
Pontocerebellar hypoplasia, type 6	#611523	AR	6q16.1	*RARS2*	Arginyl-tRNA synthetase 2	*611524	*Am J Hum Genet* **81** : 857–862, 2007
Dandy–Walker syndrome	#220200	*de novo*	3q24	*ZIC1*	Zinc finger protein of cerebellum, 1	*600470	*Nat Genet* **36** : 1053–1055, 2004
Dandy–Walker syndrome	#220200	*de novo*	3q24	*ZIC4*	Zinc finger protein of cerebellum, 4	*608948	*Nat Genet* **36** : 1053–1055, 2004
Diabetes mellitus, permanent neonatal, with cerebellar agenesis	#609069	AR	10p13–p12.1	*PTF1A*	Pancreas transcription factor 1, alpha subunit	*607194	*Nat Genet* **36** : 1301–1305, 2004
Warburg Micro syndrome 1	#600118	AR	2q21.3	*RAB3GAP*	RAB3 GTPase-activating protein	*602536	*Nat Genet* **37** : 221–223, 2005
Feingold syndrome	#164280	AD	2q24.1	*MYCN*	v-myc avian myelocytomatosis viral-related oncogene, neuroblastoma-derived	*164840	*Nat Genet* **37** : 465–467, 2005
Cerebral dysgenesis, neuropathy, ichthyosis, and palmoplantar keratoderma syndrome	#609528	AR	22q11.2	*SNAP29*	Synaptosomal-associated protein, 29-kD	*604202	*Am J Hum Genet* **77** : 242–251, 2005
Meckel syndrome, type 3	#607361	AR	8q21.13–q22.1	*TMEM67*	Transmembrane Protein 67	*609884	*Nat Genet* **38** : 191–196, 2006
Meckel syndrome, type 5	#611561	AR	16q12.2	*RPGRIP1L*	RPGRIP1-Like	*610937	*Nat Genet* **39** : 875–881, 2007
Opitz–Kaveggia syndrome	#305450	XR	Xq12–q21.31	*MED12*	Mediator of RNA polymerase II transcription, subunit 12, *S. Cerevisiae*, homolog of	*300188	*Nat Genet* **39** : 451–453, 2007
頭蓋・顔面・脊柱・その他の形成異常							
Johanson–Blizzard syndrome	#243800	AR	15q15–q21.1	*UBR1*	Ubiquitin-protein ligase E3 component N-recognin 1	*605981	*Nat Genet* **37** : 1345–1350, 2005
Noonan syndrome 4	#610733	AD	2p22–p21	*SOS1*	*son of sevenless, Drosophila*, homolog 1	*182530	*Nat Genet* **39** : 70–74, 2007 *Nat Genet* **39** : 75–79, 2007
Costello syndrome	#218040	AD	11p15.5	*HRAS*	v-HS-ras harvey rat sarcoma viral oncogene homolog	*190020	*Nat Genet* **37** : 1038–1040, 2005
Cardio-facio-cutaneous syndrome	#115150	AD	7q34	*BRAF*	v-raf murine sarcoma viral oncogene homolog B1	*164757	*Nat Genet* **38** : 294–296, 2006
Cardio-facio-cutaneous syndrome	#115150	AD	12p12.1	*KRAS2*	v-KI-ras2 Kirsten rat sarcoma 2 viral oncogene homolog	*190070	*Nat Genet* **38** : 294–296, 2006
Cornelia de Lange syndrome	#122470	AD	5p13.1	*NIPBL*	Nippled-B-like	*608667	*Nat Genet* **36** : 631–635, 2004 *Nat Genet* **36** : 636–641, 2004

疾患			遺伝子				文献
疾患名	MIM	遺伝形式	染色体座	遺伝子記号	遺伝子名	MIM	
Cornelia de Lange syndrome, X-linked	#300590	XR	Xp11.22-p11.21	*SMC1A*	Structural maintenance of chromosomes 1A	*300040	*Nat Genet* **38**：528-530, 2006
Microphthalmia with associated anomalies 2	#300166	XD	Xp11.4-p21.2	*BCOR*	BCL-6-interacting corepressor	*300485	*Nat Genet* **36**：411-416, 2004
Oculofaciocardiodental syndrome	#300166	XD	Xp11.4	*BCOR*	BCL-6-interacting corepressor	*300485	*Nat Genet* **36**：411-416, 2004
Vertical talus, congenital	#192950	AD	2q31-q32	*HOXD10*	Homeobox D10	+142984	*Am J Hum Genet* **75**：92-96,2004
Loeys-Dietz syndrome	#609192	AD	3q22	*TGFBR2*	Transforming growth factor-beta receptor, type 2	+190182	*Nat Genet* **37**：275-281, 2005
Loeys-Dietz syndrome	#609192	AD	9q33-q34	*TGFBR1*	Transforming growth factor-beta receptor, type 1	*190181	*Nat Genet* **37**：275-281, 2005
Hydrolethalus syndrome	#236680	AR	11q23-q25	*HYLS1*	Hydrolethalus syndrome 1	*610693	*Hum Mol Genet* **14**：1475-1488, 2005
Cleidocranial dysplasia（CCD）	#119600	AD	6p21	*RUNX2*	Runt-related transcription factor 2	*600211	*Cell* **89**：773-779, 1997
Crouzon syndrome（Craniofacial dysostosis type I, CFD1）	#123500	AD	10q26	*FGFR2*	Fibroblast growth factor receptor 2	*176943	*Nat Genet* **8**：98-103, 1994
Jackson-Weiss syndrome	#123150	AD	10q26	*FGFR2*	Fibroblast growth factor receptor 2	*176943	*Nat Genet* **8**：275-279, 1994
Apert syndrome（Acrocephalosyndactyly, type I, ACS1）	#101200	AD	10q26	*FGFR2*	Fibroblast growth factor receptor 2	*176943	*Nat Genet* **9**：165-172, 1995
Saethre-Chotzen syndrome（SCS, Acrocephalosyndactyly, type III, ACS3）	#101400	AD	7p21	*TWIST*	*twist* homolog 1（*Drosophila*）	*601622	*Nat Genet* **15**：36-41, 1997
Saethre-Chotzen syndrome（SCS, Acrocephalosyndactyly, type III, ACS3）	#101400	AD	10q26	*FGFR2*	Fibroblast growth factor receptor 2	*176943	*Am J Hum Genet* **62**：1370-1380, 1998
Pfeiffer syndrome（Acrocephalosyndactyly, type V, ACS5）	#101600	AD	8p11.2-p11.1	*FGFR1*	Fibroblast growth factor receptor 1	*136350	*Nat Genet* **8**：269-274, 1994
Pfeiffer syndrome（Acrocephalosyndactyly, type V, ACS5）	#101600	AD	10q26	*FGFR2*	Fibroblast growth factor receptor 2	*176943	*Nat Genet* **9**：173-176, 1995
Antley-Bixler syndrome	#207410	AR	7q11.2	*POR*	P450（cytochrome）oxidorecuctase	*124015	*Nat Genet* **36**：228-230, 2004
Beare-Stevenson cutis gyrata syndrome	#123790	AD	10q26	*FGFR2*	Fibroblast growth factor receptor 2	*176943	*Nat Genet* **13**：492-494, 1996
Greig cephalopolysyndactyly syndrome（GCPS）	#175700	AD	7p13	*GLI3*	GLI-Kruppel family member GLI3（Greig cephalosyndactyly syndrome）	*165240	*Hum Genet* **87**：452-456, 1991
Formina parietalia permagna（Parietal foramina 1）（FPP/PFM1）	#168500	AD	5q34-q35	*MSX2*	*msh*（*Drosophila*）homeo box homolog 2	*123101	*Nat Genet* **24**：387-390, 2000 *Hum Mol Genet* **9**：1251-1255, 2000
Symmetric parietal foramina（PFM2）	#609597	AD	11p11-p12	*ALX4*	Aristaless-like homeobox 4	*605420	*Nat Genet* **27**：17-18, 2001
Rubinstein syndrome（Rubinstein-Taybi syndrome）	#180849	AD	16p13.3	*CREBBP*	CREB binding protein（Rubinstein-Taybi syndrome）	*600140	*Nature* **376**：348-351, 1995
Sacral agenesis syndrome（Currarino triad, SCRA1）	#176450	AD	7q36	*HLXB9*	Homeo box HB9	*142994	*Nat Genet* **20**：358-361, 1998
Opitz syndrome（X-linked Opitz G/BBB syndrome）	#300000	XR	Xp22	*MID1*	Midline 1（Opitz/BBB syndrome）	*300552	*Nat Genet* **17**：285-291, 1997
Pelger-Huët anomaly（PHA）	#169400	AD	1q42.1	*LBR*	Lamin B receptor	*600024	*Nat Genet* **31**：410-414, 2002
Hypoparathyroidism-retardation-dysmorphism syndrome（HRD, Sanjad-Sakati syndorme）	#241410	AR	1q42-q43	*TBCE*	Tubulin-specific chaperone e	*604934	*Nat Genet* **32**：448-452, 2002
Kenny-Caffey syndrome type 1（AR-KC）	#244460	AR	1q42-q43	*TBCE*	Tubulin-specific chaperone e	*604934	*Nat Genet* **32**：448-452, 2002
Fraser syndrome（cryptophthalmos with other malformayions）	#219000	AR	4q21	*FRAS1*	Fraser syndrome 1	*607830	*Nat Genet* **34**：203-208, 2003
Oculodentodigital dysplasia（ODDD）	#164200	AD	6q21-q23.2	*GJA1*	Gap junction protein, alpha 1, 43kDa（connexin 43）	*121014	*Am J Hum Genet* **72**：408-418, 2003
Dyggve-Melchior-Clausen dysplasia（DMC）	#223800	AR	18q21.1	*DYM*	dymeclin	*607461	*Am J Hum Genet* **72**：419-428, 2003
Smith-McCort dysplasia（SMC）	#607326	AR	18q21.1	*DYM*	dymeclin	*607461	*Am J Hum Genet* **72**：419-428, 2003
Macrocephaly, multiple lipomas, and hemangiomata（Bannayan-Zonana syndrome, Bannayan-Riley-Ruvalcaba syndrome）	#153480	AD	10q23.3	*PTEN*	Phosphate and tensin homolog（mutated in multiple advanced cancers 1）	*601728	*Cancer Res* **58**：2724-2726, 1998
Proteus syndrome	%176920	*de novo*	10q23.3	*PTEN*	Phosphate and tensin homolog（mutated in multiple advanced cancers 1）	*601728	*J Med Genet* **39**：937-940, 2002
VACTERL association with hydrocephalus	#276950	?AR	10q23.3	*PTEN*	Phosphate and tensin homolog（mutated in multiple advanced cancers 1）	*601728	*J Med Genet* **38**：820-823, 2001
Multiple pterygium syndrome, Escobar variant	#265000	AR	2q33-q34	*CHRNG*	Cholinergic receptor, nicotinic, gamma polypeptide	*100730	*Am J Hum Genet* **79**：303-312, 2006
Peters-plus syndrome	#261540	AR	13q12.3	*B3GALTL*	UDP-GAL：beta-GlcNAc beta-1, 3-galactosyltransferase-like	*610308	*Am J Hum Genet* **79**：562-566, 2006
Microphthalmia, syndromic 7（MCOPS7）	#309801	XR	Xp22	*HCCS*	Holocytochrome c synthase（cytochrome c heme-lyase）	*300056	*Am J Hum Genet* **79**：878-889, 2006
Chromosome 22q13.3 deletion syndrome	#606232	AD	22q13.3	*SHANK3*	SH3 and multiple ankyrin repeat domain 3	*606230	*Nat Genet* **39**：25-27, 2007
Donnai-Barrow syndrome	#222448	AR	2q24-q31	*LRP2*	Low density lipoprotein receptor-related protein 2	*600073	*Nat Genet* **39**：957-959, 2007
母斑症							
Neurofibromatosis, type I（NF1）	+162200	AD	17q11.2	*NF1*	Neurofibromin 1（neurofibromatosis, von Recklinghausen disease, Watson disease）	+162200	*Cell* **62**：599-608, 1990 *Science* **249**：181-186, 1990
Neurofibromatosis, type 1-like syndrome	#611431	AD	15q13.2	*SPRED1*	Sprouty-related EVH1 domain-containing protein 1	*609291	*Nat Genet* **39**：1120-1126, 2007
Neurpfibromatosis, type II（NF2）	#101000	AD	22q12.2	*NF2*	Neurofibromin 2（bilateral acoustic neuroma）	*607379	*Nature* **363**：515-521, 1993
Tuberous sclerosis 1（TSC1）	#191100	AD	9q34	*TSC1*	Tuberous sclerosis 1	*605284	*Science* **277**：805-808, 1997
Tuberous sclerosis 2（TSC2）	#191100	AD	16p13.3	*TSC2*	Tuberous sclerosis 2	*191092	*Cell* **75**：1305-1315, 1993
von Hippel-Lindau syndrome	#193300	AD	3p25.3	*VHL*	von Hippel-Lindau syndrome	*608537	*Science* **260**：1317-1320, 1993
腫瘍・過誤腫							
Pallister-Hall syndrome（PHS）	#146510	AD	7p13	*GLI3*	GLI-Kruppel family member GLI3（Greig cephalosyndactyly syndrome）	*165240	*Nat Genet* **15**：266-268, 1997
Glioblastoma multiforme	#137800		10q25.3-q26.1	*DMBT1*	Deleted in malignant brain tumors 1	*601969	*Nat Genet* **17**：32-39, 1997
Glioblastoma multiforme	#137800		10q23.3	*PTEN*	Phosphate and tensin homolog（mutated in multiple advanced cancers 1）	*601728	*Science* **275**：1943-1947, 1997 *Nat Genet* **15**：356-362, 1997
Cowden disease（CD）/Lhermitte-Duclos syndrome	#158350	AD	10q23.3	*PTEN*	Phosphate and tensin homolog（mutated in multiple advanced cancers 1）	*601728	*Nat Genet* **16**：64-67, 1997

疾患			遺伝子				文献
疾患名	MIM	遺伝形式	染色体座	遺伝子記号	遺伝子名	MIM	
Medulloblastoma	#155255		10q24-q25	*SUFU*	Suppressor of fused homolog (*Drosophila*)	*607035	*Nat Genet* **31** : 306-310, 2002
Neuroblastoma	#256700		1p36.3-p36.2	*NBL1*	Neuroblastoma, suppression of tumorigenecity 1	*600613	*Oncogene* **9** : 2785-2791, 1994
Neuroblastoma	#256700		4p12	*PHOX2B*	Paired-like homeobox 2B	*603851	*Am J Hum Genet* **74** : 761-764, 2004
Familial nonchromaffin paragangliomas 1 (Familial glomus tumors 1) (PGL1)	#168000	AD	11q23	*SDHD*	Succinate dehydrogenase complex, subunit D, integral membrane protein	*602690	*Science* **287** : 848-851, 2000
Familial nonchromaffin paragangliomas 3 (Familial glomus tumors 3) (PGL3)	#605373	AD	1q13	*SDHC*	Succinate dehydrogenase complex, subunit C, integral membrane protein, 15-KD	*602413	*Nat Genet* **26** : 268-270, 2000
Familial Schwannomatosis	#162091	AD	22q12.2	*SMARCB1*	SWI/SNF-related, matrix-associated, actin-dependent regulator of chromatin, subfamily B, member 1	*601607	*Am J Hum Genet* **80** : 805-810, 2007
精神発達遅滞・自閉症							
Autosomal recessive mental retardation	#249500	AR	4q25-q26	*PRSS12*	Protease, serine, 12 (neurotrypsin, motopsin)	*606709	*Science* **298** : 1779-1781, 2002
Mental Retardation, Autosomal reseccive 6	#611092	AR	6q21	*GRIK2*	Glutamate receptor, ionotropic, kainate 2	*138244	*Am J Hum Genet* **81** : 792-798, 2007
Bardet-Biedl syndrome type 1 (BBS1)	#209900	AR	11q13	*BBS1*	Bardet-Biedl syndrome 1	*209901	*Nat Genet* **31** : 435-438, 2002
Bardet-Biedl syndrome type 2 (BBS2)	#209900	AR	16q21	*BBS2*	Bardet-Biedl syndrome 2	*606151	*Hum Mol Genet* **10** : 865-874, 2001
Bardet-Biedl syndrome 3	#209900	AR	3p12-q13	*ARL6*	ADP-ribosylation factor-like 6	*608845	*Nat Genet* **36** : 989-993, 2004
Bardet-Biedl syndrome 5	#209900	AR	2q31	*BBS5*	Bardet-Biedl syndrome 5	*603650	*Cell* **117** : 19-31, 2004
Bardet-Biedl syndrome type 7 (BBS7)	#209900	AR	4q27	*BBS7*	Bardet-Biedl syndrome 7	*607590	*Am J Hum Genet* **72** : 650-658, 2003
Bardet-Biedl syndrome type 8 (BBS8)	#209900	AR	14q32.1	*TTC8*	Tetratricopeptide repeat domain 8	*608132	*Nature* **425** : 628-633, 2003
Bardet-Biedl Syndrome 9	#209900	AR	7p14	*BBS9*	Bardet-Biedl syndrome 9	*607968	*Am J Hum Genet* **77** : 1021-1033, 2005
Bardet-Biedl syndrome 10	#209900	AR	12q21.2	*BBS10*	Bardet-Biedl syndrome 10	*610148	*Nat Genet* **38** : 521-524, 2006
Bardet-Biedl syndrome 11	#209900	AR	9q31-q34.1	*TRIM32*	Tripartite motif-containing protein 32	*602290	*Proc Natl Acad Sci* **103** : 6287-6292, 2006
Berdet-Biedl Syndrome 12	#209900	AR	4q27	*BBS12*	Bardet-Biedl syndrome 12	*610683	*Am J Hum Genet* **80** : 1-11, 2007
Cohen syndrome	#216550	AR	8q22-q23	*COH1*	Cohen syndrome 1	*607817	*Am J Hum Genet* **72** : 1359-1369, 2003
Timothy syndrome	#601005	AD	12p13.3	*CACNA1C*	Calcium channel, voltage-dependent, L type, alpha-1C subunit	*114205	*Cell* **119** : 19-31, 2004
Martsolf syndrome	#212720	AR	1q41	*RAB3GAP2*	RAB3 GTPase-activating protein, noncatalytic subunit	*609275	*Am J Hum Genet* **78** : 702-707, 2006
Autism, X-linked, susceptibility to, 1	#300425	XR	Xq13.1	*NLGN3*	Neuroligin 3	*300336	*Nat Genet* **34** : 27-29, 2003
Autism, X-linked, susceptibility to, 2	#300495	XR	Xp22.33	*NLGN4*	Neuroligin 4	*300427	*Am J Hum Genet* **74** : 552-557, 2004
Coffin-Lowry syndrome (CLS)	#303600	XD	Xp22.2-p22.1	*RPS6KA3*	Ribosomal protein S6 kinase, 90kD, polypeptide 3	*300075	*Nature* **384** : 567-570, 1996
X-linked non-specific mental retardation (MRX19)	*300075	XR	Xp22	*RPS6KA3*	Ribosomal protein S6 kinase, 90kD, polypeptide 3	*300075	*Nat Genet* **22** : 13-14, 1999
Mental retardation, X-linked 59	#300630	XR	Xp22	*AP1S2*	Adaptor-related protein complex 1, sigma-2 subunit	*300629	*Am J Hum Genet* **79** : 1119-1124, 2006
X-linked nonspecific mental retardation (MRX)		XR	Xp22.3	*VCX*	Variable charge, X chromosome	*300229	*Am J Hum Genet* **67** : 563-573, 2000
X-linked non-specific mental retardation (MRX34)	#300143	XR	Xp22.1-p21.3	*IL1RAPL1*	Interleukin 1 receptor accessory protein-like 1	*300206	*Nat Genet* **23** : 25-31, 1999
Mental retardation, X-linked, with or without seizures, ARX-related (MRXARA)	#300419	XR	Xp22.13	*ARX*	Aristaless related homeobax	*300382	*Hum Mol Genet* **11** : 981-991, 2002
Partington X-linked mental retardation syndrome (PRTS)	#309510	XR	Xp22.13	*ARX*	Aristaless related homeobax	*300382	*Nat Genet* **30** : 441-445, 2002
Mental Retardation, X-linked, 93	#300659	XR	Xq13	*BRWD3*	Bromodomain-and WD repeat domain-containing protein 3	*300553	*Am J Hum Genet* **8** : 367-374, 2007
Monoamine oxidase A deficiency (Brunner syndrome)	+309850	XR	Xp11.23	*MAOA*	Monoamine oxidase A	+309850	*Science* **262** : 578-580, 1993
Renpenning syndrome 1 (RENS1)	#309500	XR	Xp11.23	*PQBP1*	Polyglutamine binding protein 1	+300463	*Nat Genet* **35** : 313-315, 2003
Mental retardation, X-linked 9 (MRX9)	#309549	XR	Xp11.23	*FTSJ1*	FTSJ homolog 1	*300499	*Am J Hum Genet* **75** : 305-309, 2004
Mental retardation, X-linked, JARID1C-related	#300534	XR	Xp11.22-p11.21	*KDM5C*	lysine (K)-specific demethylase 5C	*314690	*Am J Hum Genet* **76** : 227-236, 2005
Mental retardation, X-linked 92	+300573	XR	Xp11	*ZNF674*	Zinc finger protein 674	+300573	*Am J Hum Genet* **78** : 265-278, 2006
X-linked nonspecific mental retardation 58 (MRX58)	#300210	XR	Xq11.4	*TM4SF2*	Transmembrane 4 superfamily member 2	*300096	*Nat Genet* **24** : 167-170, 2000
X-linked non-specific mental retardation 60 (MRX60)	#300486	XR	Xq12	*OPHN1*	Oligophrenin 1	*300127	*Nature* **392** : 923-926, 1998
X-linked alpha-thalassemia/mental retardation syndrome (ATRX)	#301040	XR	Xq13	*ATRX*	Alpha thalassemia/mental retardation syndrome, X-linked (RAD54 homolog, *S. cerevixiae*)	*300032	*Cell* **80** : 837-845, 1995
X-linked mental retardation with growth retardation, deafness, and microgenitalism (Juberg-Marsidi syndrome, JMS)	#309580	XR	Xq13	*ATRX*	Alpha thalassemia/mental retardation syndrome, X-linked (RAD54 homolog, *S. cerevixiae*)	*300032	*Nat Genet* **12** : 385-360, 1996
Mental retardation, X-linked nonsyndromic, DLG3-related	+300189	XR	Xq13.1	*DLG3*	Discs large, drosophila, homolog of, 3	+300189	*Am J Hum Genet* **75** : 318-324, 2004
X-linked non-specific mental retardation (MRX30)	#300558	XR	Xq22	*PAK3*	p21 (CDKN1A)-activated kinase 3	*300142	*Nat Genet* **20** : 25-30, 1998
Nonsyndromic X-linked mental retardation	+300034	XR	Xq22-q23	*AGTR2*	Angiotensin II receptor, type 2	+300034	*Science* **296** : 2401-2403, 2002
Nonsyndromic X-linked mental retardation (MRX63)	#300387	XR	Xq22.3-q23	*ACSL4*	Acyl-CoA synthetase long-chain member 4	*300157	*Nat Genet* **30** : 436-440, 2002
Mental retardation, X-linked, syndromic, UBE2A-related	*312180	XR	Xq24-q25	*UBE2A*	Ubiquitin-conjugating enzyme E2A	*312180	*Am J Hum Genet* **79** : 549-555, 2006
Mental Retardation, X-linked, syndromic 14	#300676	XR	Xq25-q26	*UPF3B*	UPF3, yeast, homolog of, B	*300298	*Nat Genet* **39** : 1127-1133, 2007
X-linked nonspecific mental retardation (MRX46)	#300436	XR	Xq26	*ARHGEF6*	Rac/Cdc42 guanine exchange factor (GEF) 6	*300267	*Nat Genet* **26** : 247-250, 2000
Börjeson-Forssman-Lehmann syndrome	#301900	XR	Xq26	*PHF6*	PHD finger protein 6	*300414	*Nat Genet* **32** : 661-665, 2002
Mental retardation, X-linked, ZDHH9-related	+300646	XR	Xq26.1	*ZDHHC9*	Zinc finger, DHHC-type containing 9	+300646	*Am J Hum Genet* **80** : 982-987, 2007
Lujan-Fryns syndrome	#309520	XR	Xq13	*MED12*	Mediator complex subunit 12	*300188	*J Med Genet* **44** : 472-477, 2007

疾患			遺伝子				文献
疾患名	MIM	遺伝形式	染色体座	遺伝子記号	遺伝子名	MIM	
X-linked mental retardation with isolated growth hormone deficiency (MRGH)	#300123	XR	Xq26.3	*SOX3*	SRY (sex determining region Y)-box 3	*313430	*Am J Hum Genet* **71** : 1450-1455, 2002
MASA (mental retardation, aphasia, shuffling gait, and adducted thumbs) syndrome	#303350	XR	Xq28	*L1CAM*	L1 cell adhesion molecule [hydrocephalus, stenosis of aqueduct of Sylvius 1, MASA (mental retardation, aphasia, shuffling gait and adduced thumbs) syndrome, spastic paraplegia 1]	*308840	*Nat Genet* **7** : 402-407, 1994 *Nat Genet* **7** : 408-413, 1994
Fragile X syndrome (Martin-Bell syndrome)	#300624	XR	Xq27.3	*FMR1*	Fragile X mental retardation 1	*309550	*Science* **252** : 1179-1181, 1991 *Science* **252** : 1097-1102, 1991 *Cell* **67** : 1047-1058, 1991
X-linked mental retardation associated with fragile site FRAXE	+309548	XR	Xq28	*FMR2*	Fragile X mental retardation 2	+309548	*Cell* **74** : 127-134, 1993
X-linked non-specific mental retardation (MRX41, MRX48)	+300104	XR	Xq28	*GDI1*	GDP dissociation inhibitor 1	+300104	*Nat Genet* **19** : 134-139, 1998
Rett syndrome (RTT)	#312750	XD	Xq28	*MECP2*	Methyl-CpG-binding protein 2 (Rett syndrome)	*300005	*Nat Genet* **23** : 185-188, 1999
X-linked mental retardation and progressive spasticity		XR	Xq28	*MECP2*	Methyl-CpG-binding protein 2 (Rett syndrome)	*300005	*Am J Hum Genet* **67** : 982-985, 2000
Mental retardation with psychosis, pyramidal sign and macroorchidism (PPM-X syndrome)	#300055	XR	Xq28	*MECP2*	Methyl CpG binding protein 2	*300005	*Am J Hum Genet* **70** : 1034-1037, 2002
Rett syndrome, atypical, CDKL5-related	#312750	*de novo*	Xp22	*CDKL5*	Cyclin-dependent kinase-like 5	*300203	*Am J Hum Genet* **75** : 1079-1093, 2004 *Am J Hum Genet* **75** : 1149-1154, 2004
X-linked mental retardation with seizures	#300352	XR	Xq28	*SLC6A8*	Solute carrier family 6 (neurotransmitter transporter, creatine), member 8	*300036	*Am J Hum Genet* **70** : 1349-1356, 2002
言語発達障害							
Speech and language disorder (Developmental verbal dyspraxia, SPCH1)	#602081	AD	7q31	*FOXP2*	Forkhead box P2	*605317	*Nature* **413** : 519-523, 2001
代謝異常							
糖原病・糖代謝異常							
Fructose-1, 6-bisphosphatse deficiency	#229700	AR	9q22.2-q22.3	*FBP1*	Fructose-1, 6-bisphosphatase 1	*611570	*Biochem Biophys Res Commun* **210** : 797-804, 1995
Glycogen storage disease II (Pompe disease)	#232300	AR	17q25.2-q25.3	*GAA*	Glucosidase, alpha ; acid (Pompe disease, glycogen storage disease type II)	*606800	*Biochem J* **272** : 493-497, 1990
Glycogen storage disease III (Cori disease)	#232400	AR	1p21	*AGL*	Amylo-1, 6-glucosidase, 4-alpha-glucotransferase	*610860	*J Clin Invest* **98** : 352-357, 1996 *Biochem Biophys Res Commun* **224** : 493-499, 1996
Glycogen storage disease V (McArdle's disease)	#232600	AR	11q12-q13.2	*PYGM*	Phophorylase, glycogen ; muscle (McArdle syndrome, glycogen storage disease type V)	*608455	*New Engl J Med* **329** : 241-245, 1993
Glycogen storage disease VII [Tarui (垂井) disease]	#232800	AR	12q13.3	*PFKM*	Phosphofructokinase, muscle	*610681	*J Biol Chem* **265** : 9392-9395, 1990
Deficiency of M subunit of phosphoglycerate mutase (Glycogen storage disease X)	#261670	AR	7p13-p12.3	*PGAM2*	Phosphoglycerate mutase 2 (muscle)	*612931	*Am J Hum Genet* **52** : 472-477, 1993
Glycogen storage disease XIII (GSD13)	#612932	AR	17pter-p12	*ENO3*	Enolase 3, (beta, muscle)	*131370	*Ann Neurol* **50** : 202-207, 2001
Phosphoglycerate kinase 1 deficiency	#300653	XR	Xq13	*PGK1*	Phosphoglycerate kinase 1	*311800	*Blood* **79** : 1582-1585, 1992 *Blood* **77** : 1348-1352, 1991
Glycogen storage disease XI (GSD11 ; Lactate dehydrogenase A deficiency)	#612933	AR	11p15.4	*LDHA*	Lactate dehydrogenase A	*150000	*Biochem Biophys Res Commun* **168** : 677-682, 1990
Autosomal recessive phosphorylase kinase deficiency of liver and muscle	#261750	AR	16q12-q13	*PHKB*	Phosphorylase kinase, beta	*172490	*Hum Mol Genet* **6** : 1109-1115, 1997
Pyruvate dehydrogenase E2 deficiency	#245348	AR	11q23.1	*DLAT*	Dihydrolipoamide S-acetyltransferase	*608770	*Ann Neurol* **58** : 234-241, 2005
Pyruvate dehydrogenase E3-binding protein deficiency	#245349	AR	11p13	*PDHX*	Pyruvare dehydrogenase complex, component X	*608769	*Am J Hum Genet* **61** : 1318-1326, 1997
Pyruvare carboxylase deficiency (Leigh necrotizing encephalopathy)	#266150	AR	11q13.4-q13.5	*PC*	Pyruvate carboxylase	*608786	*Am J Hum Genet* **62** : 1312-1319, 1998
Pyruvate dehydrogenase deficiency	#312170	XR	Xp22.2-p22.1	*PDHA1*	Pyruvate dehydrogenase (lipoamide) alpha 1	*300502	*Am J Hum Genet* **44** : 358-364, 1989
Succinic semialdehyde dehydrogenase deficiency	#271980	AR	6p22	*ALDH5A1*	Aldehyde dehydrogenase 5 family, member A1	*610045	*Am J Hum Genet* **63** : 399-408, 1998
アミノ酸・ビタミン代謝異常							
Phenylketonuria (PKU)	#261600	AR	12q24.1	*PAH*	Phenyalanine hydroxylase	*612349	*Biochemistry* **25** : 743-749, 1986
Phenylketonuria II (PKU2)	#261630	AR	4p15.31	*QDPR*	Quinoid dihydropteridine reductase	*612676	*Am J Hum Genet* **47** : 279-285, 1990
Phenylketonuria III	#261640	AR	11q22.3-q23.2	*PTS*	6-Pyruvoyltetrahydropterin synthase	*612719	*Am J Hum Genet* **54** : 782-792, 1994
Hyperphenylalaninemia with primapterinuria	#264070	AR	10q22	*PCBD1*	6-Pyruvoyl-tetrahydropterin synthase/dimerization cofactor of hepatocyte nuclear factor 1 alpha (TCF1)	*126090	*Am J Hum Genet* **53** : 768-774, 1993
GTP cyclohydolase I deficiency (Hyperphenylalanemia with neopterin deficiency)	#233910	AR	14q22.1-q22.2	*GCH1*	GTP cyclohydrolase 1	*600225	*J Pediat* **126** : 401-405, 1995 *J Biol Chem* **270** : 10062-10071, 1995
DOPA-responsive dystonia (DRD, 瀬川病)	#128230	AD	14q22.1-q22.2	*GCH1*	GTP cyclohydrolase 1	*600225	*Nat Genet* **8** : 236-242, 1994
Segawa syndrome, autosomal recessive	#605407	AR	11p15.5	*TH*	tyrosine hydroxylase	*191290	*Hum Genet* **95** : 123-125, 1995
Dystonia, DOPA-responsive, due to sepiapterin reductase deficiency	+182125	AR	2p14-p12	*SPR*	Sepiapterin reductase (7, 8-dihydrobiopterin : NADP + oxidoreductase)	+182125	*Am J Hum Genet* **69** : 269-277, 2001
Dopamine beta-hydroxylase deficiency, congenital	#223360	AR	9q34	*DBH*	Dopamine beta-hydroxylase (dopamine beta-monooxygenase)	*609312	*Am J Med Genet* **108** : 140-147, 2002
Aromatic L-amino acid decarboxylase deficiency	#608643	AR	7p11	*DDC*	Dopa decarboxylase (aromatic L-amino acid decarboxylase)	*107930	*J Inherit Metab Dis* **21** : 4, 1998

疾患			遺伝子				文献
疾患名	MIM	遺伝形式	染色体座	遺伝子記号	遺伝子名	MIM	
Homocystinuria due to deficiency N(5, 10)-methylenetetrahydrofolate reductase activity(MTHFR deficiency)	#236250	AR	1p36.3	*MTHFR*	5, 10-methylenetetrahydroforate reductase(NADPH)	*607093	*Nat Genet* **7** : 195-200, 1994
Sulfocysteinuria	#272300	AR	12q13.13	*SUOX*	Sulfite oxidase	*606887	*Cell* **91** : 973-983, 1997
Methionine adenosyl transferase deficiency	#250850	AD, AR	10q22	*MAT1A*	Methionine adenosyltransferase I, alpha	*610550	*J Clin Invest* **95** : 1943-1947, 1995
Tyrosinemia, type I	+276700	AR	15q23-q25	*FAH*	Fumarylacetoacetate hydrolase (fumarylacetoacetase)	+276700	*J Clin Invest* **90** : 1185-1192, 1992
Tyrosine transaminase deficiency (Tyrosinemia, type II)	+276600	AR	16q22.1-q22.3	*TAT*	Tyrosine aminotransferase	+276600	*Hum Genet* **77** : 352-358, 1987
Homocystinuria	+236200	AR	21q22.3	*CBS*	Cystathionine-beta-synthase	+236200	*Hum Mutat* **1** : 113-123, 1992
Methylmalonic aciduria and homocystinuria, cblC type	#277400	AR	1p34.1	*MMACHC*	Methylmalonic aciduria (cobalamin deficiency) cblC type, with homocystinuria	*609831	*Nat Genet* **38** : 93-100, 2006
Methylmalonic aciduria and homocystinuria, cblD type	#277410	AR	2q23.2	*MMADHC*	Methylmalonic aciduria (cobalamin deficiency) cblD type, with homocystinuria	*611935	*New Engl J Med* **358** : 1454-1464, 2008
Methylcobalamin deficiency, cbl E type	#236270	AR	5p15.3-p15.2	*MTRR*	5-methyltetrahydrofolate-homocysteine methyltransferase reductase	*602568	*Proc Nat Acad Sci* **95** : 3059-3064, 1998
Methylcobalamin deficiency, cbl G type	#250940	AR	1q43	*MTR*	5-methyltetrahydrofolate-homocysteine methyltransferase	*156570	*Hum Mol Genet* **5** : 1867-1874, 1996 *Hum Mol Genet* **5** : 1859-1865, 1996
Isolated nonketotic hyperglycemia, type I	#605899	AR	9p22	*GLDC*	Glycine dehydrogenase (carboxylating ; glycine decarboxylase, glycine cleavage system protein P)	*238300	*Biochem Biophys Res Commun* **174** : 1176-1182, 1991 *Mol Biol Med* **8** : 65-79, 1991
Isolated nonketotic hyperglycemia, type II	#605899	AR	3p21.2-p21.1	*AMT*	Aminomethyltransferase (glycine cleavage system protein T)	*238310	*Hum Genet* **93** : 655-658, 1994
Nonketotic hyperglycenemia(NKH) / Glycine encephalopathy(GCE)	#605899		16q24	*GCSH*	Glycine cleavage system protein H(aminomethyl carrier)	*238330	*Am J Hum Genet* **48** : 351-361, 1991
Transient neonatal hyperglicinemia (TNH)	#605899	AD	9q22	*GLDC*	Glycine dehydrogenase (decarboxylating, glycine decarboxylase, glycine cleavage system protein P)	*238300	*Ann Neurol* **52** : 643-646, 2002
Transient neonatal hyperglycinemia (TNH)		AD	16q24	*GCSH*	Glycine cleavage system protein H(aminomethyl carrier)	*238330	*Ann Neurol* **52** : 643-646, 2002
Dimethylglycine dehydrogenase deficiency(DMGDHD)	#605850	AR	5q12.2-q12.3	*DMGDH*	Dimethylglycine dehydrogenase precursor	*605849	*Am J Hum Genet* **68** : 839-847, 2001
Hyperammonemia due to carbamoylphosphate synthetase I (CPS1) deficiency	#237300	AR	2q35	*CPS1*	Carbamoyl-phosphate synthetase 1, mitochondrial	*608307	*J Clin Invest* **91** : 1884-1887, 1993
Phosphoglycerate dehydrogenase deficiency	#601815	AR	1q12	*PHGDH*	Phosphoglycerate dehydrogenase	*606879	*Am J Hum Genet* **67** : 1389-1399, 2000
Hyperammonemia due to ornithine transcarbamylase deficiency(OTC)	#311250	XR	Xp21.1	*OTC*	Ornithine carbamoyltransferase	*300461	*J Clin Invest* **82** : 1353-1358, 1988
Argininosuccinic aciduria	#207900	AR	7cen-q11.2	*ASL*	Argininosuccinate lyase	*608310	*Proc Natl Acad Sci* **87** : 9625-96259, 1990 *Am J Hum Genet* **47** (suppl.) : A169, 1990
Argininemia	#207800	AR	6q23	*ARG1*	Arginase, liver	*608313	*J Clin Invest* **86** : 347-350, 1990
Arginine : glycine amidinotransferase deficiency	#612718	AR	15q15.3	*GATM*	Glycine amidinotransferase(L-arginine : glycine amidinotransferase)	*602360	*Am J Hum Genet* **69** : 1127-1133, 2001
N-acetylglutamate synthase deficiency	#237310	AR	17q21.31	*(NAGS)*	N-acetylglutamate synthase	*608300	*Ann Neurol* **52** : 845-849, 2002
Maple syrup urine disease, type IA (MSUD IA)	#248600	AR	19q13.1-q13.2	*BCKDHA*	Branched chain keto acid dehydrogenase E1, alpha polypeptide(maple syrup urine disease)	*608348	*J Clin Invest* **83** : 1425-1429, 1989
Maple syrup urine disease, type IB (MSUD IB)	#248600	AR	6p22-p21	*BCKDHB*	Branched chain keto acid dehydrogenase E1, beta polypeptide(maple syrup urin disease)	*248611	*J Clin Invest* **87** : 1862-1866, 1991
Maple syrup urine disease, type II (MSUD, type II)	#248600	AR	1p31	*DBT*	Dihydrolipoamide branched chain transacylase E2	*248610	*Biochem Biophys Res Commun* **174** : 804-809, 1991 *J Clin Invest* **87** : 1207-1211, 1991
Lactic acidosis due to lipoamide dehydrogenase deficiency(Maple syrup urine disease, type III)	#248600	AR	7q31-q32	*DLD*	Dihydrolipoamide dehydrogenase	*238331	*Proc Natl Acd Sci* **90** : 5186-5190, 1999
Isovaleric acidemia(IVA)	#243500	AR	15q14-q15	*IVD*	Isovaleryl Coenzyme A dehydrogenase	*607036	*Am J Hum Genet* **49** : 147-157, 1991
Propionic acidemia, type I(Ketotic hyperglycinemia, type I)	#606054	AR	13q32	*PCCA*	Propionyl Coenzyme A carboxylase, alpha polypeptide	*232000	*Hum Genet* **101** : 93-96, 1997
Propionic acidemia, type II(Ketotic hyperglycinemia, type II)	#606054	AR	3q21-q22	*PCCB*	Propionyl Coenzyme A carboxylase, beta polypeptide	*232050	*Genomics* **8** : 249-254, 1990
Methylmalonic aciduria due to methylmalonic CoA mutase deficiency	#251000	AR	6p21	*MUT*	Methylmalonyl Coenzyme A mutase	*609058	*Proc Natl Acad Sci* **87** : 3147-3150, 1990
Methylmalonyl-CoA epimerase deficiency	#251120	AR	2p13.3	*MCEE*	Methylmalonyl CoA epimerase	*608419	*Hum Mutat* **27** : 640-643, 2006
3-Methylcrotonylglycinuria I (complementation group A)	#210200	AR	3q25-q27	*MCCC1*	Methylcrotonyl-Coenzyme A carboxylase 1(alpha)	*609010	*Am J Hum Genet* **68** : 334-346, 2001 *Hum Mol Genet* **10** : 1299-1306, 2001 *J Clin Invest* **107** : 495-504, 2001
3-Methylcrotonylglycinuria II (complementation group B)	#210210	AR	5q12-q13	*MCCC2*	Methylcrotonyl-Coenzyme A carboxylase 2(beta)	*609014	*Am J Hum Genet* **68** : 334-346, 2001 *Hum Mol Genet* **10** : 1299-1306, 2001 *J Clin Invest* **107** : 495-504, 2001
3-Methylglutaconic aciduria, type I	#250950	AR	9q22	*AUH*	AU RNA binding protein/enoyl-Coenzyme A hydratase	*600529	*Am J Hum Genet* **71** : 1463-1466, 2002
Familial hyperlysinemia	#238700	AR	7q31.3	*AASS*	Alpha-aminoadipic semialdehyde synthetase	*605133	*Am J Hum Genet* **66** : 1736-1743, 2000
Congenital glutamine deficiency	#610015	AR	1q31	*GLUL*	Glutamate-ammonia ligase	*138290	*New Engl J Med* **353** : 1926-1933, 2005
Pyroglutamic aciduria(5-Oxoprolinuria)	#266130	AR	20q11.2	*GSS*	Glutathione synthetase	*601002	*Nat Genet* **14** : 361-365, 1996
Malonyl-CoA carboxylase(MLYCD) deficiency	#248360	AR	16q24	*MLYCD*	Malonyl-Coenzyme A decarboxylase	*606761	*Am J Hum Genet* **65** : 318-326, 1999

疾患			遺伝子				文献
疾患名	MIM	遺伝形式	染色体座	遺伝子記号	遺伝子名	MIM	
Holocarboxylase synthetase deficiency	#253270	AR	21q22.1	*HLCS*	Holocarboxylase synthetase〔biotin-[proprionyl-Coenzyme A-carboxylase(ATP-hydrolysing)]ligase〕	*609018	*Nat Genet* **8**:122-128, 1994
Biotinidase deficiency(Late-onset multiple carboxylase deficiency)	#253260	AR	3p25	*BTD*	Biotinidase	*609019	*Nat Genet* **11**:96-98, 1995
有機酸代謝異常							
Encephalopathy, ethylmalonic	#602473	AR	19q13.32	*ETHE1*	ETHE1 gene	*608451	*Am J Hum Genet* **74**:239-252, 2004
3-Hydroxy-3-methylglutaryl-coenzyme A lyase deficiency	+246450	AR	1pter-p33	*HMGCL*	3-hydroxymethyl-3-methylglutaryl-Coenzyme A lyase(hydroxymethylglutaric aciduria)	+246450	*Am J Hum Genet* **51**(suppl.):A173, 1992
Glutaric acidemia I(Glutaryl-CoA dehydrogenase deficiency)	#231670	AR	19p13.2	*GCDH*	Glutaryl-Coenzyme A dehydrogenase	*608801	*Am J Hum Genet* **51**(suppl.):A165, 1992
AMACR deficiency	+604489	AR	5p13.2-q11.1	*AMACR*	Alpha-methylacyl-CoA racemase	+604489	*Nat Genet* **24**:188-191, 2000
Alpha-methylacetoacetic aciduria (Mitochondrial acetoacetyl-CoA thiolase deficiency)	#203750	AR	11q22.3-q23.1	*ACAT1*	Acetyl-Coenzyme A acetyltransferase 1(acetoacetyl Coenzyme A thiolase)	*607809	*Biochem Biophys Res Commun* **179**:124-129, 1991
Short chain acyl-CoA dehydrogenase (SCADH)deficiency	#201470	AR	12q22-qter	*ACADS*	Acyl-Coenzyme A dehydrogenase, C-2 to C-3 short chain	*606885	*Am J Hum Genet* **45**(suppl.):A208, 1989
Medium-chain acyl-CoA dehydrogenase (MCADH)deficiency	#201450	AR	1p31	*ACADM*	Acyl-Coenzyme A dehydrogenase, C-4 to C12 straight chain	*607008	*Biochem Biophys Res Commun* **171**:498-506, 1990
Deficiency of long chain acyl-CoA dehydrogenease(LCAD)	#201460	AR	2q34-q35	*ACADL*	Acyl-Coenzyme A dehydrogenase, long chain	*609576	*Proc Natl Acad Sci* **92**:841-845, 1995
Trifunctional protein deficiency, type 1 (Long-chain 3-hydroxyacyl-CoA dehydrogenase(LCHAD)deficiency)	#609015	AR	2p23	*HADHA*	Hydroxyacyl-Coenzyme A dehydrogenase/3-ketoacyl-Coenzyme A thiolase/enoyl-Coenzyme A hydratase (trifunctional protein), alpha subunit	*600890	*Biochem Biophys Acta* **1215**:347-350, 1994
Trifunctional protein deficiency, type 2 (Long-chain 3-hydroxyacyl-CoA dehydrogenase(LCHAD)deficiency)	#609015	AR	2p23	*HADHB*	Hydroxyacyl-Coenzyme A dehydrogenase/3-ketoacyl-Coenzyme A thiolase/enoyl-Coenzyme A hydratase (trifunctional protein), beta subunit	*143450	*Am J Hum Genet* **58**:979-988, 1996
Peroxisomal acyl-CoA oxidase deficiency	#264470	AR	17q25	*ACOX1*	Acyl-Coenzyme A oxidase 1, palmitoyl	*609751	*J Clin Invest* **94**:526-531, 1994
Carnitine palmytoyltransferase I deficiency	#255120	AR	11q13	*CPT1A*	Carnitine palmytoyltransferase 1A(liver)	*600528	*J Clin Invest* **102**:527-531, 1998
Carnitine palmitoyltransferase II(CPT II) deficiency	#255110	AR	1p32	*CPT2*	Carnitine palmytoyltransferase II	*600650	*Poc Natl Acad Sci* **89**:8429-8433, 1992
Carnitine-acylcarnitine translocase (CACT)deficiency	+212138	AR	3p21.31	*SLC25A20*	Solute carrier family 25(carnitine/acylcarnitine translocase), member 20	+212138	*Am J Hum Genet* **61**:1239-1245, 1997
Refsum disease(Heredopathia atactica polyneuritiformis)	#266500	AR	10pter-p11.2	*PHYH*	Phytanoyl-CoA hydroxylase (Refsum disease)	*602026	*Nat Genet* **17**:185-189, 1997 *Nat Genet* **17**:190-193, 1997
胆汁酸・ステロール・ステロイド代謝異常							
2-Methylbutyrylglycinuria	#610006	AR	10q25-q26	*ACADSB*	Acyl-Coenzyme A dehydrogenase, short/branched chain	*600301	*Am J Hum Genet* **67**:1095-1103, 2000 *Pediat Res* **47**:830-833, 2000
Deficiency of very long chain acyl-CoA dehydrogenease(VLCAD)	#201475	AR	17p11.2-p11.13	*ACADVL*	Acyl-Coenzyme A dehydrogenase, very long chain	*609575	*Proc Natl Acad Sci* **92**:10496-10500, 1995 *Am J Hum Genet* **57**:273-283, 1995
Cerebrotendinous xanthomatosis(CTX)	#213700	AR	2q33-qter	*CYP27A1*	Cytochrome P450, subfamily XXVIIA(stroid 27-hydroxylase, cebrotendinous xanthomatosis), polypeptide 1	*606530	*J Biol Chem* **266**:7779-7783, 1991
Smith-Lemli-Opitz syndrome(SLOS)	#270400	AR	11q12-q13	*DHCR7*	7-dehydrocholesterol reductase	*602858	*Am J Hum Genet* **63**:55-62, 1998 *Am J Hum Genet* **63**:329-338, 1998
Lathosterolosis	#607330	AR	11q23.3	*SC5DL*	Sterol-C5-desaturase(ERG3 delta-5-desaturase homolog, fungal)-like	*602286	*Am J Hum Genet* **71**:952-958, 2002
Peroxisomal D-bifunctional protein deficiency	#261515	AR	5q2	*HSD17B4*	Hydroxysteroid(17-beta) dehydrogenase 4	*601860	*Am J Hum Genet* **61**:1153-1162, 1997
核酸代謝異常							
Lesch-Nyhan syndrome(LNS)	#300322	XR	Xq26-q27.2	*HPRT1*	Hypoxanthine phosphoribosyltransferase 1	*308000	*J Clin Invest* **72**:767-772, 1983
Adenylosuccinate lyase(ADSL)deficiency	#103050	AR	22q13.1	*ADSL*	Adenylosuccinate lyase	*608222	*Nat Genet* **1**:59-63, 1992
Myopathy due to myoadenylate deaminase deficiency	+102770	AD	1p21-p13	*AMPD1*	Adnosine monophosphate deaminase 1(isoform M)	+102770	*Proc Natl Acad Sci* **89**:6457-6461, 1992
Dihydropyrimidinase deficiency	+222748	AR	8q22	*DPYS*	Dihydropyrimidinase	+222748	*Am J Hum Genet* **63**:717-726, 1998
Beta-ureidopropionase deficiency	+606673	AR	22q11.2	*UPB1*	Beta-ureidopropionase	+606673	*Hum Mol Genet* **13**:2793-2801, 2004
ポルフィリア							
Acute intermittent porphyria(AIP)	#176000	AD	11q23.2-qter	*HMBS*	Hydrosymethylbilane synthase	*609806	*Nucleic Acids Res* **17**:6637-6647, 1989
Porphyria variegata	#176200	AD	1q22	*PPOX*	Protoporphyrinogen oxidase	*600923	*Hum Mol Genet* **5**:407-410, 1996 *Nat Genet* **13**:95-97, 1996
Coproporphyria	#121300	AD	3q12	*CPOX*	Coproporphyrinogen oxidase	*612732	*Hum Mol Genet* **3**:477-480, 1994 *Hum Mol Genet* **3**:1325-1330, 1994 *Hum Mol Genet* **3**:1807-1810, 1994
Delta-aminolevulinate dehydratase (ALAD)deficiency	#612740	AR	9q34	*ALAD*	Aminolevulinate, delta-, dehydratase	*125270	*Biochem Biophys Res Commun* **172**:237-242, 1990
金属代謝関連疾患							
Aceruloplasminemia	#604290	AR	3q23-q24	*CP*	Ceruloplasmin(ferroxidase)	*117700	*Biochem Biophys Res Cimmun* **217**:89-95, 1995 *Nat Genet* **9**:267-272, 1995 *Proc Natl Acad Sci* **92**:2539-2543, 1995
Sulfocysteinuria(Sulfite oxidase deficiency)	#272300	AR	12q13.13	*SUOX*	Sulfite oxidase	*606887	*Cell* **91**:973-983, 1997

疾患			遺伝子				文献
疾患名	MIM	遺伝形式	染色体座	遺伝子記号	遺伝子名	MIM	
Molybdenum cofactor deficiency (MOCOD)	#252150	AR	6p21.3	*MOCS1*	Molybdenum cofactor synthesis 1	*603707	*Nat Genet* **20** : 51–53, 1998
Molybdenum cofactor deficiency type C	#252150	AR	14q23.3	*GPHN*	Gephyrin	*603930	*Am J Hum Genet* **68** : 208–213.2001
トランスポーター障害							
Hypomagnesemia, secondary hypocalcemia (HOMG)	#602014	AR	9q12	*TRPM6*	Transient receptor potential cation channel, subfamily M, member 6	*607009	*Nat Genet* **31** : 166–170, 2002 *Nat Genet* **31** : 171–174, 2002
Brody myopathy	#601003	AR	16p12.1	*ATP2A1*	ATPase, Ca^{++} transporting, cardiac muscle, fast twitch 1	*108730	*Nat Genet* **14** : 461–464, 1996
Menkes syndrome	#309400	XR	Xq12–q13	*ATP7A*	ATPase, Cu^{++} transporting, alpha polypeptide (Menkes syndrome)	*300011	*Nat Genet* **3** : 7–13, 1993 *Nat Genet* **3** : 14–19, 1993 *Nat Genet* **3** : 20–25, 1993
Occipital horn syndrome	#304150	XR	Xq12–q13	*ATP7A*	ATPase, Cu^{++} transporting, alpha polypeptide (Menkes syndrome)	*300011	*Nat Genet* **8** : 195–202, 1993
Wilson disease	#277900	AR	13q14.3–q21.1	*ATP7B*	ATPase, Cu^{++} transporting, beta polypeptide	*606882	*Nat Genet* **5** : 327–337, 1993 *Nat Genet* **5** : 344–350, 1993
Kufor–Rakeb syndrome	#606693	AR	1p36	*ATP13A2*	ATPase, type 13A2	*610513	*Nat Genet* **38** : 1184–1191, 2006
Tangier disease (HDLDT1, high density lipoprotein deficiency, Tangier type 1)	#205400	AR	9q22–q31	*ABCA1*	ATP–binding cassette, sub–family A (ABC1), member 1	*600046	*Nat Genet* **22** : 336–345, 1999 *Nat Genet* **22** : 347–351, 1999 *Nat Genet* **22** : 352–355, 1999
Sideroblastic anemia and spinocerebellar ataxia (ASAT, XLSA/A)	#301310	XR	Xq13	*ABCB7*	ATP–binding cassette, sub–family B (MDR/TAP), member 7	*300135	*Hum Mol Genet* **8** : 743–749, 1999
Adrenoleukodystrophy (ALD)	#300100	XR	Xq28	*ABCD1*	ATP–binding cassette, sub–family D (ALD), member 1	*300371	*Nature* **361** : 726–730, 1993
Glucose transport defect, blood–brain barrier	#606777	AD	1p35–p31.3	*SLC2A1*	Solute carrier family 2 (facilitated glucose transporter), member1	*138140	*Nat Genet* **18** : 188–191, 1998
Fanconii–Bickel syndrome (FBK)	#227810	AR	3q36.1–q26.3	*SLC2A2*	Solute carrier family 2 (facilitated glucose transporter), member 2	*138160	*Diabetologia* **37** : 420–427, 1994
Hartnup disorder	#234500	AR	5p15	*SLC6A19*	Solute carrier family 6 (Neurotransmitter transporter), member 19	*608893	*Nat Genet* **36** : 999–1002, 2004 *Nat Genet* **36** : 1003–1007, 2004
Lysinuric protein intolerance (LPI)	#222700	AR	14q11.2	*SLC7A7*	Solute carrier family 7 (cationic amino acid transporter, y^+ system), member 7	*603593	*Nat Genet* **21** : 293–296, 1999 *Nat Genet* **21** : 297–301, 1999
Agenesis of corpus callosum with neuropathy (ACCPN, Charlevoix disease)	#218000	AR	15q13–q14	*SLC12A6*	Solute carrier family 12 (potassium/chloride transporters), member 6	*604878	*Nat Genet* **32** : 384–392, 2002
Sialic acid storage diseases (SASD)		AR	6q14–q15	*SLC17A5*	Solute carrier family 17 (anion/sugar transporter), member 5	*604322	*Nat Genet* **23** : 462–465, 1999
Infantile sialic acid storage disease (ISSD)	#269920						
Salla disease (sialuria, Finish type)	#604369						
Basal ganglia disease, biotin–responsive	#607483	AR	2q36.3	*SLC19A3*	Solute carrier family 19, member 3	*606152	*Am J Hum Genet* **77** : 16–26, 2005
Amish type microcephaly (MCPHA)	#607196	AR	17q25.3	*SLC25A19*	Solute carrier family 25 (mitochondrial deoxynucleotide carrier), member 19	*606521	*Nat Genet* **32** : 175–179, 2002
Primary systemic carnitine deficiency (SCD/CDSP)	#212140	AR	5q31	*SLC22A5*	Solute carrier family 22 (organic cation transporter), member 5	*603377	*Nat Genet* **21** : 91–94, 1999 *Hum Mol Genet* **8** : 655–660, 1999
Adult–onset type II citrullinemia (CTLN2)	#603471	AR	7q21.3	*SLC25A13*	Solute carrier family 25, member 13 (citrin)	*603859	*Nat Genet* **22** : 159–163, 1999
Hyperornithinaemia–hyperammonaemia–homocitrullinuria (HHH) syndrome	#238970	AR	13q14	*SLC25A15*	Solute carrier family 25 (mitochondrial carrier, ornithine transporter), member 15	*603861	*Nat Genet* **22** : 151–158, 1999
X–linked creatine deficiency	#300352	XR	Xq28	*SLC6A8*	Solute carrier family 6 (neurotransmitter transporter, creatine), member 8	*300036	*Am J Hum Genet* **68** : 1497–1500, 2001
ミトコンドリア脳筋症・エネルギー代謝							
ACAD 9 deficiency	#611126	*de novo*	3q26	*ACAD9*	Acyl–CoA dehydrogenase family, member 9	*611103	*Am J Hum Genet* **81** : 87–103, 2007
Coenzyme Q10 deficiency	#607426	AR	10q12.1	*PDSS1*	Prenyl diphosphate synthase, subunit 1	*607429	*J Clin Invest* **117** : 765–772, 2007
Coenzyme Q10 deficiency	#607426	AR	6q21	*PDSS2*	Prenyl diphosphate synthase, subunit 2	*610564	*Am J Hum Genet* **79** : 1125–1129, 2006
Leigh syndrome, French–Canadian type	#220111	AR	2p21	*LRPPRC*	Leucine–rich PPR–motif containing	*607544	*Pro Natl Acad Sci* **100** : 605–610, 2003
Combined oxidative phosphorylation deficiency	#609060	AR	3q25	*GFM1*	Mitochondrial elongation factor G1	*606639	*N Engl J Med* **351** : 2080–2086, 2004
Combined oxidative phosphorylation deficiency 4	#610678	AR	16p11.2	*TUFM*	Tu translation elongation factor, mitochondrial	*602389	*Am J Hum Genet* **80** : 44–58, 2007
Leukoencephalopathy with brain stem and spinal cord involvement and Lactate elevation	#611105	AR	1q25.1	*DARS2*	Aspartyl–tRNA synthetase, mitochondrial	*610956	*Nat Genet* **39** : 534–539, 2007
Mitochondrial DNA–depletion syndrome (MDS)	#251880	AR	2p13	*DGUOK*	Deoxyguanosine kinase	*601465	*Nat Genet* **29** : 337–341, 2001
Mitochondrial DNA depletion myopathy	#609560	AR	16q22	*TK2*	Thymidine kinase–2, mitochondrial	*188250	*Nat Genet* **29** : 342–344, 2001
Lactic acidosis, fatal infantile	#245400	AR	2p11.3	*SUCLG1*	Succinate–CoA ligase, alpha subunit	*611224	*Am J Hum Genet* **81** : 383–387, 2007
Mitochondrial DNA depletion syndrome, encephalopathic form, with methylmalonic aciduria	#612073	AR	13q12.2–q13	*SUCLA2*	Succinate–CoA ligase, ADP–forming, beta subunit	*603921	*Am J Hum Genet* **76** : 108–1086, 2005
Glutaric aciduria IIA (Deficiency of electron transfer flavoprotein, alpha polypeptide)	#231680	AR	15q23–q25	*ETFA*	Electron–transfer–flavoprotein, alpha polypeptide (glutaric aciduria II)	*608503	*Am J Hum Genet* **49** : 575–580, 1991
Glutaric aciduria IIB (Deficiency of electron transfer flavoprotein, beta polypeptide)	#231680	AR	19q13.3–q13.4	*ETFB*	Electron–transfer–flavoprotein, beta polypeptide	*130410	*Hum Mol Genet* **3** : 429–435, 1994
Glutaric aciduria IIC (Electron transfer flavoprotein : ubiquinone oxidoreductase deficiency)	#231680	AR	4q32–q35	*ETFDH*	Electron–trasferring–flavoprotein dehydrogenase	*231675	*Clin Res* **41** : 271A, 1993

疾患			遺伝子				文献
疾患名	MIM	遺伝形式	染色体座	遺伝子記号	遺伝子名	MIM	
Mitochondrial complex I deficiency	#252010	AR	11q13	*NDUFV1*	NADH dehydrogenase (ubiquinone) flavoprotein 1, 51kD	*161015	*Nat Genet* **21**: 260–261, 1999
Complex I deficiency with cardiomyopathy and encephalomyopathy	#252010	AR	1q23	*NDUFS2*	NADH dehydrogenase (ubiquinone) Fe–S protein 2, 49kD (NADH–coenzyme Q reductase)	*602985	*Ann Neurol* **49**: 195–201, 2001
Mitochondrial complex I deficiency	#252010	AR	5q11.1	*NDUFS4*	NADH dehydrogenase (ubiquinone) Fe–S protein 4, 18kD (NADH–coenzyme Q reductase)	*602694	*Am J Hum Genet* **62**: 262–268, 1998
Leigh syndrome due to complex I deficiency	#256000	AR	19p13	*NDUFS7*	NADH dehydrogenase (ubiquinone) Fe–S protein 7, 20kD (NADH–coenzyme Q reductase)	*601825	*Am J Hum Genet* **64**: 1505–1510, 1999
Leigh syndrome due to complex I deficiency	#256000	AR	11q13	*NDUFS8*	NADH dehydrogenase (ubiquinone) Fe–S protein 8, 23kD (NADH–coenzyme Q reductase)	*602141	*Am J Hum Genet* **63**: 1598–1608, 1998
Leigh syndrome due to complex II deficiency	#256000	AR	5p15	*SDHA*	Succinate dehydrogenase complex, subunit A, flavoprotein (Fp)	*600857	*Nat Genet* **11**: 144–149, 1995
Complex III deficiency with tubulopathy, encephalopathy and liver failure	#124000	AR	2q33	*BCS1L*	BCS1-like (yeast)	*603647	*Nat Genet* **29**: 57–60, 2001
Cytochrome c oxidase deficiency with neonatal-onset hepatic failure and encephalopathy	#220110	AR	17p13.1	*SCO1*	SCO (cytochrome oxidase deficient, yeast) homolog 1	*603644	*Am J Hum Genet* **67**: 1104–1109, 2000
Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency	#604377	AR	22q13	*SCO2*	SCO (cytochrome oxidase deficient, yeast) homolog 2	*604272	*Nat Genet* **23**: 333–337, 1999
Leigh syndrome due to cytochrome c oxidase deficiency	#256000	AR	9q33–q34	*SURF1*	Surfeit 1	*185620	*Nat Genet* **20**: 337–343, 1998 *Am J Hum Genet* **63**: 1609–1621, 1998
Isolated leukodystrophy		AR	9q33–q34	*SURF1*	Surfeit 1	*185620	*Ann Neurol* **49**: 797–800, 2001
Deficiency of mitochindrial respiratory chain complex IV (Cytochrome c oxidase deficiency)	#220110	AR	17p12	*COX10*	COX10 (yeast) homolog, cytochrome c oxidase assembly protein (heme A : farnesyltransferase)	*602125	*Hum Mol Genet* **9**: 1245–1249, 2000
Leber optic atrophy and dystonia	#500001		mtDNA	*MTND6*	Complex I, subunit ND6	*516006	*Proc Natl Acad Sci* **91**: 6206–6210, 1994
			mtDNA	*MTND4*	Complex I, subunit ND4	*516003	*Am J Hum Genet* **58**: 703–711, 1996
			mtDNA	*MTND1*	Complex I, subunit ND1	*516000	*Arch Neurol* **64**: 890–893, 2007 *Neurogenetics* **4**: 199–205, 2003
Cytochrome c oxidase II deficiency			mtDNA	*MTCO2*	Complex IV, cytochrome c oxidase subunit II	*516040	*Am J Hum Genet* **65**: 1030–1039, 1999
Cytochrome c oxidase deficiency	*516050		mtDNA	*MTCO3*	Complex IV, cytochrome c oxidase subunit III	*516050	*Nat Genet* **12**: 410–416, 1996
Leigh syndrome due to ATP synthase (complex V) 6 deficinecy/ Neuronal muscle weakness, ataxia and retinitis pigmentosa (NARP)	+516060 #551500		mtDNA	*MTATP6*	ATP synthase F0 subunit 6	*516060	*Am J Hum Genet* **46**: 428–422, 1990
Mitochondrial myopathy, encephalopathy, lactic acidosis, and stroke-like episodes (MELAS)	#540000		mtDNA	*MTTL1*	tRNA leucine 1 (UUA/G)	*590050	*Nature* **348**: 651–653, 1990 *Biochem Biophys Res Commun* **173**: 816–822, 1990
Myoclonic epilepsy associated with ragged-red fibers (MERRF)	#545000		mtDNA	*MTTK*	tRNA lysine	*590060	*Cell* **61**: 931–937, 1990 *Biochem Int* **21**: 789–796, 1990
	#545000		mtDNA	*MTTL1*	tRNA leucine 1 (UUA/G)	*590050	*J Clin Invest* **92**: 2906–2915, 1993
Mitochondrial encephalomyopathy due to complex III deficiency			mtDNA	*MTCYB*	Cytochrome b of complex III	*516020	*Am J Hum Genet* **67**: 1400–1410, 2000
Kearns–Sayre syndrome (KSS), Chronic progressive ophthalmoplegia (CPEO) with myopathy	#530000		mtDNA				*Lancet* **I**: 885, 1988
ペルオキシソーム形成障害							
A 群							
Zellweger syndrome	#214100						
Infantile Refsum disease	#266510						
Neonatal adrenoleukodystrophy	#202370						
Peroxisome biogenesis disorder complementation group 1 [E]	+602136	AR	7q21.2	*PEX1*	Peroxisome biogenesis factor 1	+602136	*Nat Genet* **17**: 445–448, 1997 *Nat Genet* **17**: 449–452, 1997
Peroxisome biogenesis disorder complementation group 2	#601539	AR	12p13.3	*PEX5*	peroxisomal biogenesis factor 5	*600414	*Nat Genet* **9**: 115–125, 1995
Peroxisome biogenesis disorder complementation group 3	+601758	AR	17q21.1	*PEX12*	Peroxisome biogenesis factor 12	+601758	*Nat Genet* **15**: 385–388, 1997
Peroxisome biogenesis disorder complementation group 4 [C]	+601498	AR	6p21.1	*PEX6*	Peroxisomal biogenesis factor 6	+601498	*Am J Hum Genet* **59**: 1210–1220, 1996 *EMBO J* **15**: 2914–2923, 1996
Peroxisome biogenesis disorder complementation group 7 [B]	+602859	AR	1p36.32	*PEX10*	Peroxisome biogenesis factor 10	+602859	*Am J Hum Genet* **63**: 1622–1630, 1998
Peroxisome biogenesis disorder complementation group 9 [D]	+603360	AR	11p	*PEX16*	Peroxisome biogenesis factor 16	+603360	*Am J Hum Genet* **63**: 1622–1630, 1998
Peroxisome biogenesis disorder complementation group 10 [F]	+170993	AR	8q21.1	*PXMP3*	Peroxisomal membrane protein 3, 35kD (Zellweger syndrome)	+170993	*Science* **255**: 1132–1134, 1992
Peroxisome biogenesis disorder, complementation group G (Zellweger syndrome, complementation group G)	#214100	AR	6q23–q24	*PEX3*	Peroxisome biogenesis factor 3	*603164	*Hum Mol Genet* **9**: 1995–1999, 2000 *Am J Hum Genet* **67**: 967–975, 2000 *Am J Hum Genet* **67**: 976–981, 2000
B 群							
Rhizomeric chondrodysplasia punctata type 1 (RCDP1)	#215100	AR	6q22–q24	*PEX7*	Peroxisomal biogenesis factor 7	*601757	
Peroxisome biogenesis disorder complementation group 11	+601757	AR	6q22–q24	*PEX7*	Peroxisomal biogenesis factor 7	+601757	*Nat Genet* **15**: 369–376, 1997 *Nat Genet* **15**: 377–380, 1997 *Nat Genet* **15**: 381–384, 1997
Refsum disease	#266500	AR	6q22–q24	*PEX7*	Peroxisome biogenesis factor 7	*601757	*Am J Hum Genet* **72**: 471–477, 2003
リピドーシス	#266500	AR	10p13	*PHYH*	phytanoyl-CoA 2-hydroxylase	*602026	*Nat Genet* **17**: 190–193, 1997
Gaucher disease, type I	#230800	AR	1q21	*GBA*	Glucosidase, beta ; acid (including glucosylceramidase)	*606463	*New Engl J Med* **316**: 570–575, 1987

疾患			遺伝子				文献
疾患名	MIM	遺伝形式	染色体座	遺伝子記号	遺伝子名	MIM	
Gaucher disease, type II	#230900	AR	1q21	*GBA*	Glucosidase, beta ; acid (including glucosylceramidase)	*606463	
Gaucher disease, type III	#231000	AR	1q21	*GBA*	Glucosidase, beta ; acid (including glucosylceramidase)	*606463	
Niemann-Pick disease, types A, B and E (Sphigomyelin lipidosis)	#257200	AR	11p15.4-p15.1	*NPC1*	Sphingomyelin phosphodiesterase 1, acid lysosomal (acid sphingomyelinase)	*607623	*Proc Natl Acad Sci* **88** : 3748-3752, 1991 *Biochem Biophys Res Commun* **179** : 1187-1191, 1991 *J Clin Invwst* **88** : 806-810, 1991
Niemann-Pick disease, type C1 (NPC1)	#257220	AR	18q11-q12	*NPC1*	Niemann-Pick disease, type C1	*607623	*Science* **277** : 228-231, 1997
Niemann-Pick disease, type C2 (NPC2)	#607625	AR	14q24.3	*NPC2*	Niemann-Pick disease, type C2	*601015	*Science* **290** : 2298-2301, 2000
Frontal lobe atrophy due to a mutauon in NPC2	#607625	AR	14q24.3	*NPC2*	Niemann-Pick disease, type C2	*601015	*Ann Neurol* **52** : 743-749, 2002
Fabry disease (Angiokeratoma corporis diffusum)	#301500	XR	Xq21.3-q22	*GLA*	Galactosidase, alpha	*300644	*J Clin Invest* **83** : 1390-1399, 1989
Beata-galactosidase-1 (GLB1) deficiency	#230500	AR	3p21.33	*GLB1*	Galactosidase, beta 1	*611458	*Am J Hum Genet* **49** : 566-574, 1991 *Am J Hum Genet* **49** : 435-442, 1991 *Am J Hum Genet* **49** : 1091-1093, 1991
GM1 gangliosidosis, type I	#230500						
Morquio syndrome B	#253010						
GM1 gangliosidosis, type II	#230600						
GM1 gangliosidosis, type III	#230650						
Tay-Sachs disease (TSD ; GM2 gangliosisdosis type I)	#272800	AR	15q24.1	*HEXA*	Hexosaminidase A (alpha polypeptide)	*606869	*Scienece* **232** : 1646-1648, 1986
Tay-Sachs disease, AB variant (GM2 gangliosidosis type AB)	+272750	AR	5q31.3-q33.1	*GM2A*	GM2 ganglioside activator protein	+272750	*FEFS Lett* **290** : 1-3, 1991
Sandhoff disease (GM2 gangliosidosis type II)	#268800	AR	5q13.3	*HEXB*	Hexosaminidase B (beta polypeptide)	*606873	*J Biol Chem* **264** : 5155-5158, 1989 *Hum Genet* **81** : 287-288, 1989
白質ジストロフィー・白質脳症							
Metachromatic leukodystrophy (MLD)	#250100	AR	22q13.31-qter	*ARSA*	Arylsulfatase A	*607574	*Proc Natl Acad Sci* **86** : 9436-9440, 1989
Metachromatic leukodystrophy due to saposin B deficiency	#249900	AR	10q22.1	*PSAP*	Prosaposin	*176801	*Proc Natl Acad Sci* **87** : 2541-2544, 1990
Krabbe disease (Globoid cell leukodystrophy)	#245200	AR	14q31	*GALC*	Galactosylceramidase (Krabbe disease)	*606890	*Biochem Biophys Res Commun* **198** : 485-491, 1994
Canavan disease	#271900	AR	17p13.3	*ASPA*	Aspartoacylase (aminoacylase 2, Canavan disease)	*608034	*Nat Genet* **5** : 118-123, 1993
Alexander disease	#203450	AR	17q21	*GFAP*	Glial fibrillary acidic protein	*137780	*Nat Genet* **27** : 117-120, 2001
Pelizaeus-Merzbacher disease (PMD)	#312080	XR	Xq22	*PLP1*	Proteolipid protein 1	*300401	*Am J Hum Genet* **45** : 435-442, 1989 *Am J Hum Genet* **45** (Suppl) : A169, 1989 *Proc Natl Acad Sci* **86** : 8128-8131, 1989
Leukoencephalopathy with vanishing white matter (VWM)	#603896	AR	12q24.3	*EIF2B1*	Eukaryotic translation initiation factor 2B, subunit 1 alpha, 26kD	*606686	*Ann Neurol* **51** : 264-270, 2002
Leukoencephalopathy with vanishing white matter (VWM)	#603896	AR	14q24.3	*EIF2B2*	Eukaryotic translation initiation factor 2B, subunit 2 beta, 39kD	*606454	*Nat Genet* **29** : 383-388, 2001
Leukoencephalopathy with vanishing white matter (VWM)	#603896	AR	1p34.1	*EIF2B3*	Eukaryotic translation initiation factor 2B, subunit 3 gamma	*606273	*Ann Neurol* **51** : 264-270, 2002
Leukoencephalopathy with vanishing white matter (VWM)	#603896	AR	2p23.3	*EIF2B4*	Eukaryotic translation initiation factor 2B, subunit 4 delta	*606687	*Ann Neurol* **51** : 264-270, 2002
Leukoencephalopathy with vanishing white matter (VWM)	#603896	AR	3q27.3	*EIF2B5*	Eukaryotic translation initiation factor 2B, subunit 5 epsilon, 82kD	*603945	*Nat Genet* **29** : 383-388, 2001
Cree leukoencephalopathy (CLE)	#603896	AR	3q27.3	*EIF2B5*	Eukaryotic translation initiation factor 2B, subunit 5 epsilon, 82kDa	*603945	*Ann Neurol* **52** : 506-510, 2002
Megalencephalic leukoencephalopathy with subcortical cysts (MLC)	#604004	AR	22q13.33	*MLC1*	Megalencephalic leukoencephalopathy with subcortical cysts 1	*605908	*Am J Hum Genet* **68** : 831-838, 2001
Polycystic lipomembranous osteodysplasia with sclerosing leukoencephalopathy (PLOSL)	#221770	AR	19q13.1	*TYROBP*	TYRO protein tyrosine kinase-binding protein	*604142	*Nat Genet* **25** : 357-361, 2000
Ribose 5-phosphate isomerase deficiency	#608611	AR	2p11.2	*RPIA*	Ribose 5-phosphate isomerase A (ribose 5-phospate epimerase)	*180430	*Am J Hum Genet* **74** : 745-751, 2004
Leukodystrophy, demyelinating, adult-onset, autosomal dominant	#169500	AD	5q23.3-q31.1	*LMNB1*	Lamin B1	*150340	*Nat Genet* **38** : 1114-1123, 2006
Leukencephalopathy with dystonia and motor neuropathy, SCPx-deficient	*184755	AR	1p32	*SCP2*	Sterol carrier protein 2	*184755	*Am J Hum Genet* **78** : 1046-1052, 2006
Aicardi-Goutieres syndrome 1 (AGS1)	#225750	AR	3p21.3-p21.2	*TREX1*	Three prime repair exonuclease 1	*606609	*Nat Genet* **38** : 917-920, 2006
Aicardi-Goutieres syndrome 2 (AGS2)	#610181	AR	13q14-q21	*RNASEH2B*	Ribonuclease H2, subunit B	*610326	*Nat Genet* **38** : 910-916, 2006
Aicardi-Goutieres syndrome 3 (AGS3)	#610329	AR	11q13.2	*RNASEH2C*	Ribonuclease H2, subunit C	*610330	*Nat Genet* **38** : 910-916, 2006
Aicardi-Goutieres syndrome 4 (AGS4)	#610333	AR	19p13.13	*RNASEH2A*	Ribonuclease H2, subunit A	*606034	*Nat Genet* **38** : 910-916, 2006
Aicardi-Goutieres syndrome 5 (AGS5)	#610905	AD	3p21.3-p21.2	*TREX1*	Three prime repair exonuclease 1	*606609	*Am J Hum Genet* **80** : 811-815, 2007
Retinal vasculopathy with cerebral leukodystrophy	#192315	AD	3p21.3-p21.2	*TREX1*	Three prime repair exonuclease 1	*606609	*Nat Genet* **39** : 1068-1070, 2007
Leukodystrophy, dysmyelinating, and spastic paraparesis with or without dystonia	#612443	AR	16q23	*FA2H*	Fatty acid 2-hydroxylase	*611026	*Am J Hum Genet* **83** : 643-648, 2008
Leukodystrophy, hypomyelinating, 5	#610532	AR	7p15.3	*FAM126A*	Family with sequence similarity 126, member A	*610531	*Nat Genet* **38** : 1111-1113, 2006
ムコリピドーシスその他							
Mucolipidosis IV (MLIV)	#252650	AR	19p13.3-p13.2	*MCOLN1*	Mucolipin 1	*605248	*Nat Genet* **26** : 120-123, 2000 *Hum Mol Genet* **9** : 2471-2478, 2000 *Am J Hum Genet* **67** : 1110-1120, 2000
Neuraminidase deficiency	#256550	AR	6p21	*NEU1*	Neuraminidase 1 (lysosomal sialidase)	*608272	*Nat Genet* **15** : 316-320, 1997
Neuraminidase deficiency with beta-galactosidase deficiency	+256540	AR	20q13.12	*CTSA*	cathepsin A	+256540	*Proc Jpn Acad* **66B** : 217-222, 1990
Fucosidosis	#230000	AR	1p34	*FUCA1*	Fucosidase, alpha-L-1, tissue	*612280	*J Mol Neurosci* **1** : 177-180, 1989
Mannosidosis, alpha B, lysosomal	#248500	AR	19cen-q13.1	*MAN2B1*	Mannosidase, alpha, class 2B, member 1	*609458	*Hum Mol Genet* **6** : 717-726, 1997
Mannosidosis, beta A, lysosomal	#248510	AR	4q22-q25	*MANBA*	Mannosidase, beta A, lysosomal	*609489	*Hum Mol Genet* **7** : 75-83, 1998

疾患			遺伝子				文献
疾患名	MIM	遺伝形式	染色体座	遺伝子記号	遺伝子名	MIM	
Aspartylglucosaminuria	+208400	AR	4q34.3	*AGA*	Aspartylglucosaminidase	+208400	*Proc Natl Acad Sci* **88**：11222–11226, 1991 *J Biol Chem* **266**：12105–12113, 1991 *Genomics* **11**：206–211, 1991
Schindler disease, type I	#609241	AR	22q13–qter	*NAGA*	N–acetylgalactosaminidase, alpha–	*104170	*J Clin Invest* **86**：1752–1756, 1990
酸性ムコ多糖症							
Hurler syndrome	#607014	AR	4p16.3	*IDUA*	Iduronidase, alpha–L	*252800	*Hum Mutat* **1**：103–108, 1992 *Hum Mutat* **1**：333–339, 1992
Mucopolysaccharidosis type II	+309900	XR	Xq27.3–q28	*IDS*	Iduronate 2–sulfatase（Hunter syndrome）	+309900	*Hum Genet* **87**：205–206, 1991
Mucopolysaccharidosis type IIIA	#252900	AR	17q25.3	*SGSH*	N–sulfoglucosamine sulfohydrolase（sulfamidase）	*605270	*Nat Genet* **11**：465–467, 1995
copolysaccharidosis type IIIB	#252920	AR	17q11–q21	*NAGLU*	N–acetylglucosaminidase, alpha–（Sanfilippo disease IIIB）	*609701	*Am J Hum Genet* **57**（suppl）：A185, 1995 *Proc Natl Acad Sci* **93**：6101–6105, 1996.
Mucopolysaccharidosis type IIIC	#252930	AR	8p11.1	*HGSNAT*	Heparan–alpha–glucosaminide N–acetyltransferase	*610453	*Am J Hum Genet* **79**：738–744, 2006 *Am J Hum Genet* **79**：807–819, 2006
Morquio syndrome A	#253000	AR	16q24.3	*GALNS*	Galactosamine（N–acetyl）–6–sulfate sulfatase（Morquio syndrome, mucopolysacchridosis IVA）	*612222	*J Clin Invest* **90**：1049–1053, 1992
Mucopolysaccharidosis type VI	#253200	AR	5q11–q13	*ARSB*	Arylsulfatase B	*611542	*J Biol Chem* **266**：21386–21391, 1991
Mucopolysaccharidosis type VII	#253220	AR	7q11.21	*GUSB*	Glucuronidase, beta	*611499	*Am J Hum Genet* **48**：89–96, 1991
グリコシレーション異常							
Congenital disorder of glycosylation, type Ia（CDG1A）	#212065	AR	16p13	*PMM2*	Phosphomannomutase 2	*601785	*Nat Genet* **15**：88–92, 1997
Congenital disorder of glycosylation, type Ib（CDG1B）	#602579	AR	15q22–qter	*MPI*	Mannose phosphate isomerase	*154550	*Am J Hum Genet* **62**：1535–1539, 1998 *J Clin Invest* **101**：1414–1420, 1998
Congenital disorder of glycosylation, type Ic（CDG1C）	#603147	AR	1p31.3	*ALG6*	Asparagine–linked glycosylation 6, alpha–1, 3–glucosyltransferase homolog（*S. cerevisiae*）［Dolichyl pyrophosphate $Man_9GlcNAc_2$ a1, 3–glucosyltransferase］	*604566	*Proc Natl Acad Sci* **96**：6982–6987, 1999
Congenital disorder of glycosylation, type Id（CDG1D）	#601110	AR	3q27.3	*ALG3*	Asparagine–linked glycosylation 3 homolog（yeast, alpha–1, 3–mannosyltransferase）	*608750	*EMBO J* **18**：6816–6822, 1999
Congenital disorder of glycosylation, type Ie（CDG1E）	#608799	AR	20q13.1	*DPM1*	Dolichyl–phosphate mannosyltrasferase polypeptide 1, catalytic subunit	*603503	*J Clin Invest* **105**：191–198, 2000 *J Clin Invest* **105**：223–239, 2000
Congenital disorder of glycosylation, type If（CDG1F）	#609180	AR	17p13.1–p12	*MPDU1*	Mannose–P–dolichol utilization defect 1	*604041	*J Clin Invest* **108**：1613–1619, 2001 *J Clin Invest* **108**：1687–1695, 2001
Congenital disorder of glycosylation, type Ik（CDG1K）	#608540	AR	16p13.3	*ALG1*	Asparagine–linked glycosylation 1 homolog（yeast, beta–1, 4–mannosyltransferase）	*605907	*Am J Hum Genet* **74**：472–481, 2004 *Am J Hum Genet* **74**：545–551, 2004 *Hum Mol Genet* **13**：535–542, 2004
Congenital disorder of glycosylation, type IIa（CDG2A）	#212066	AR	14q21	*MGAT2*	Mannosyl（alpha–1, 6–）–glycoproyein beta–1, 2–N–acetylglucosaminyltransferase	*602616	*Am J Hum Genet* **59**：810–817, 1996
Congenital disorder of glycosylation, type IIb（CDG2B）	#606056	AR	2p13.1	*MOGS*	mannosyl–oligosaccharide glucosidase	*601336	*Am J Hum Genet* **66**：1744–1756, 2000
Congenital disorder of glycosylation, type IIc（CDG2C）	#266265	AR	11p11.2	*SLC35C1*	Solute carrier family 35, member C1	*605881	*Nat Genet* **28**：73–76, 2001
Congenital disorder of glycosylation, type IId（CDG2D）	#607091	AR	9p13	*B4GALT1*	UDP–Gal：beta GlcNAc beta 1, 4–galactosyltransferase, polypeptide 1	*137060	*J Clin Invest* **109**：725–733, 2002
Congenital disorder of glycosylation, type IIh（CDG2H）	#611182	AR	16q22.1	*COG8*	Component of oligomeric Golgi complex 8	*606979	*Hum Molec Genet* **16**：717–730, 2007 *Hum Molec Genet* **16**：731–741, 2007
神経セロイドリポフスチン蓄積症							
Ceroid lipofuscinosis, neuronal, 1（CLN1）	#256730	AR	1p32	*PPT1*	Palmytoyl–protein thioesterase 1（ceroid–lipofuscinosis, neuronal 1, infantile）	*600722	*Nature* **376**：584–587, 1995
Ceroid lipofuscinosis, neuronal, 2（CLN2）	#204500	AR	11p15	*TPP1*	Ceroid–lipofuscinosis, neuronal 2, late infantile（Jansky–Bielschowsky disease）	*607998	*Science* **277**：1802–1805, 1997
Ceroid lipofuscinosis, neuronal, 3（CLN3）	#204200	AR	16p12	*CLN3*	Ceroid–lipofuscinosis, neuronal 3, juvenile（Batten, Spielmeyer–Vogt disease）	*607042	*Cell* **82**：949–957, 1995
Ceroid lipofuscinosis, neuronal, 5（CLN5）	#256731	AR	13q21.1–q32	*CLN5*	Ceroid–lipofuscinosis, neuronal 5	*276731	*Nat Genet* **19**：286–288, 1998
Ceroid lipofuscinosis, neuronal, 6（CLN6）	#601780	AR	15q23	*CLN6*	Ceroid–lipofuscinosis, neuronal, late infantile, variant	*606725	*Am J Hum Genet* **70**：324–335, 2002 *Am J Hum Genet* **70**：537–542, 2002
Ceroid lipofuscinosis, neuronal, 7（CLN7）	#610951	AR	4q28.1–q28.2	*MFSD8*	Major facilitator superfamily domain–containing protein 8	*611124	*Am J Hum Genet* **81**：136–146, 2007
Ceroid lipofuscinosis, neuronal, 8（CLN8）	#600143	AR	8p23	*CLN8*	Ceroid–lipofuscinosis, neuronal 8（epilepsy, progressive, with mental retardation）	*607837	*Nat Genet* **23**：233–236, 1999
DNA 合成・修復障害							
Xeroderma pigmentosum, complementation group A（XPA）	#278700	AR	9q22.3	*XPA*	Xeroderma pigmentosum, compementation group A	*611153	*Nature* **348**：73–76, 1990
Xeroderma pigmentosum, complementation group B（XPB）	#610651	AD	2q21	*ERCC3*	Excision repair cross–complementing rodent repair deficiency, complementation group 3（xeroderma pigmentosum B complementing）	*133510	*Cell* **62**：777–791, 1990
Xeroderma pigmentosum, complementation group D（XPD）	#278730	AR	19q13.2–q13.3	*ERCC2*	Excision repair cross–complementing rodent repair deficiency, complementation group 2 D）	*126340	*Nat Genet* **7**：189–194, 1994 *Hum Mol Genet* **3**：1783–1788, 1994

疾患			遺伝子				文献
疾患名	MIM	遺伝形式	染色体座	遺伝子記号	遺伝子名	MIM	
Xeroderma pigmentosum, complementation group G (XPG)	#278780	AR	13q22-q34	*ERCC5*	Excision repair cross-complementing rodent repair deficiency, complementation group 5 pigmentosum, complementaion group G (Cockayne syndrome)	*133530	*Hum Mol Genet* **3** : 963-967, 1994
Cockayne syndrome, type A (CSA)	#216400	AR	5q12.1	*ERCC8*	Excision repair cross-complementing rodent repair deficiency, complementation group 8	*609412	*Cell* **82** : 555-564, 1995
Cockayne syndrome, type B (CSB)	#133540	AR	10q11	*ERCC6*	Excision repair cross-complementing rodent repair deficiency, complementation group 6	+609413	*Cell* **71** : 939-953, 1992
Nijmegen breakege syndrome (Ataxia-telangiectasia variant VI)	#251260	AR	8q21-q24	*NBN*	Nibrin	*602667	*Cell* **93** : 467-476, 1998 *Cell* **93** : 477-486, 1998 *Nat Genet* **19** : 179-181, 1998
Werner syndrome	#277700	AR	8p12	*WRN*	Werner syndrome, RecQ helicase-like	*604611	*Science* **272** : 258-262, 1996
			1q22	*LMNA*	Lamin A/C	*150330	*Lancet* **362** : 416-417, 2003
Hutchinson-Gilford progeria syndrome (HGPS)	#176670	AR	1q22	*LMNA*	Lamin A/C	*150330	*Nature* **423** : 293-298, 2003.
その他							
Chediak-Higashi syndrome (CHS1)	#214500	AR	1q42.1-q42.2	*LYST*	Lysosomal trafficking regulator	*606897	*Nature* **382** : 262-265, 1996 *Nat Genet* **14** : 307-311, 1996
Chronic infantile neurological cutaneous and articular (CINCA) syndrome	#607115	AD/*de novo*	1q44	*NLRP3*	NLR family, pyrin domain containing 3	*606416	*Am J Hum Genet* **71** : 198-203, 2002
Trimethylaminuria (fish-odour syndrome)	#602079	AR	1q24.3	*FMO3*	Flavin containing monooxygenase 3	*136132	*Nat Genet* **17** : 491-494, 1997
Cold-induced sweating syndrome (CISS)	#272430	AR	19p12	*CRLF1*	Cytokine receptor-like factor 1	*604237	*Am J Hum Genet* **72** : 375-383, 2003
Angioedema, hereditary (HAE)	#106100	AD	11q11-q13.1	*SERPING1*	Serpin peptidase inhibitor, clade G (C1 inhibitor), member 1	*606860	*J Biol Chem* **264** : 3066-3071, 1989 *J Clin Invest* **83** : 1888-1893, 1989
Angioedema, hereditary, type III (HAE III)	#610618	AD	5q33-qter	*F12*	Coagulation factor XII (Hageman factor)	*610619	*Biochem Biophys Res Commun* **343** : 1286-1289, 2006 *Am J Hum Genet* **79** : 1098-1104, 2006.
Incontinentia pigmenti, type II (IP2)	#308310	XD	Xq28	IKBKG	Inhibitor of kappa light polypeptide gene enhancer in B-cells, kinase gamma	*300248	*Nature* **405** : 466-472, 2000

1. 疾患名：疾患名は原則として“Mendelian Inheritance in Man”に準拠。“Mendelian Inheritance in Man”は Online Mendelian Inheritance in Man™ (OMIM™) として公開されている [C02]。
2. MIM：“Menderian Inheritance in Man”での番号。
3. 遺伝形式：AD：常染色体優性，AR：常染色体劣性，XD：X 連鎖優性，XR：X 連鎖劣性，*de novo*：新生突然変異
4. 遺伝子：遺伝子記号及び遺伝子名は，HUGO Gene Nomenclature Committee [C01] に準拠。ヒトの遺伝子は大文字イタリックで記載するのが正規の記載法。