OMIM #	NAME	GENE	DISEASE TYPE
302045	CARDIOMYOPATHY, DILATED, 3B; CMD3B	DMD	cardiac
208000	ARTERIAL CALCIFICATION, GENERALIZED, OF INFANCY; GACI	ENPP1	cardiac
241550	HYPOPLASTIC LEFT HEART SYNDROME	GJA1	cardiac
220400	JERVELL AND LANGE-NIELSEN SYNDROME 1; JLNS1	KCNQ1	cardiac
255960	MYXOMA, INTRACARDIAC	PRKAR1A	cardiac
302060	BARTH SYNDROME; BTHS	TAZ	cardiac
300069	CARDIOMYOPATHY, DILATED, 3A; CMD3A	TAZ	cardiac
611705	MYOPATHY, EARLY-ONSET, WITH FATAL CARDIOMYOPATHY	TTN	cardiac
242500	ICHTHYOSIS CONGENITA, HARLEQUIN FETUS	ABCA12	cutaneous
228600	FIBROMATOSIS, JUVENILE HYALINE	ANTXR2	cutaneous
219200	CUTIS LAXA, AR, II	ATP6V0A2	cutaneous
607626	ICHTHYOSIS, LEUKOCYTE VACUOLES, ALOPECIA, AND SCLEROSING CHOLANGITIS	CLDN1	cutaneous
226650	EPIDERMOLYSIS BULLOSA, JUNCTIONAL, NON-HERLITZ	COL17A1	cutaneous
225320	EHLERS-DANLOS SYNDROME, AR, CARDIAC VALVULAR	COL1A2	cutaneous
226600	EPIDERMOLYSIS BULLOSA DYSTROPHICA, AR; RDEB	COL7A1	cutaneous
278740	XERODERMA PIGMENTOSUM, COMPLEMENTATION GROUP E	DDB2	cutaneous
607655	SKIN FRAGILITY-WOOLLY HAIR SYNDROME	DSP	cutaneous
609638	EPIDERMOLYSIS BULLOSA, LETHAL ACANTHOLYTIC	DSP	cutaneous
305100	ECTODERMAL DYSPLASIA, HYPOHIDROTIC, XLR; XHED	EDA	cutaneous
277580	WAARDENBURG-SHAH SYNDROME	EDN3	cutaneous
277580	WAARDENBURG-SHAH SYNDROME	EDNRB	cutaneous
600501	ABCD SYNDROME	EDNRB	cutaneous
219100	CUTIS LAXA, AR, I	EFEMP2	cutaneous
278730	XERODERMA PIGMENTOSUM, COMPLEMENTATION GROUP D; XPD	ERCC2	cutaneous
601675	TRICHOTHIODYSTROPHY, PHOTOSENSITIVE; TTDP	ERCC2	cutaneous
601675	TRICHOTHIODYSTROPHY, PHOTOSENSITIVE; TTDP	ERCC3	cutaneous
610651	XERODERMA PIGMENTOSUM, COMPLEMENTATION GROUP B; XPB	ERCC3	cutaneous
278760	XERODERMA PIGMENTOSUM, COMPLEMENTATION GROUP F; XPF	ERCC4	cutaneous
278780	XERODERMA PIGMENTOSUM, COMPLEMENTATION GROUP G; XPG	ERCC5	cutaneous
278800	DE SANCTIS-CACCHIONE SYNDROME	ERCC6	cutaneous
219100	CUTIS LAXA, AR, I	FBLN5	cutaneous
601675	TRICHOTHIODYSTROPHY, PHOTOSENSITIVE; TTDP	GTF2H5	cutaneous
226730	EPIDERMOLYSIS BULLOSA JUNCTIONALIS WITH PYLORIC ATRESIA	ITGA6	cutaneous
226730	EPIDERMOLYSIS BULLOSA JUNCTIONALIS WITH PYLORIC ATRESIA	ITGB4	cutaneous
226650	EPIDERMOLYSIS BULLOSA, JUNCTIONAL, NON-HERLITZ	ITGB4	cutaneous
245660	LARYNGOONYCHOCUTANEOUS SYNDROME; LOCS	LAMA3	cutaneous
226650	EPIDERMOLYSIS BULLOSA, JUNCTIONAL, NON-HERLITZ	LAMA3	cutaneous
226700	EPIDERMOLYSIS BULLOSA, JUNCTIONAL, HERLITZ	LAMA3	cutaneous
226650	EPIDERMOLYSIS BULLOSA, JUNCTIONAL, NON-HERLITZ	LAMB3	cutaneous
226700	EPIDERMOLYSIS BULLOSA, JUNCTIONAL, HERLITZ	LAMB3	cutaneous
226650	EPIDERMOLYSIS BULLOSA, JUNCTIONAL, NON-HERLITZ	LAMC2	cutaneous
226700	EPIDERMOLYSIS BULLOSA, JUNCTIONAL, HERLITZ	LAMC2	cutaneous
275210	TIGHT SKIN CONTRACTURE SYNDROME, LETHAL	LMNA	cutaneous
226670	EPIDERMOLYSIS BULLOSA SIMPLEX WITH MUSCULAR DYSTROPHY	PLEC1	cutaneous
277580	WAARDENBURG-SHAH SYNDROME	SOX10	cutaneous
601706	YEMENITE DEAF-BLIND HYPOPIGMENTATION SYNDROME	SOX10	cutaneous
242300	ICHTHYOSIS, LAMELLAR, 1; LI1	TGM1	cutaneous
263700	PORPHYRIA, CONGENITAL ERYTHROPOIETIC	UROS	cutaneous
257980	ODONTOONYCHODERMAL DYSPLASIA; OODD	WNT10A	cutaneous
278800	DE SANCTIS-CACCHIONE SYNDROME	XPA	cutaneous
278700	XERODERMA PIGMENTOSUM, COMPLEMENTATION GROUP A; XPA	XPA	cutaneous
275210	TIGHT SKIN CONTRACTURE SYNDROME, LETHAL	ZMPSTE24	cutaneous
229200	BRITTLE CORNEA SYNDROME; BCS	ZNF469	cutaneous
220290	DEAFNESS, AR 1A	GJB2	deafness
600060	DEAFNESS, NEUROSENSORY, AR 2; DFNB2	MYO7A	deafness
302950	CHONDRODYSPLASIA PUNCTATA 1, XLR RECESSIVE; CDPX1	ARSE	developmental
300215	LISSENCEPHALY, XLR, 2 LISX2	ARX	developmental
605039	C-LIKE SYNDROME	CD96	developmental
211750	C SYNDROME	CD96	developmental
253290	MULTIPLE PTERYGIUM SYNDROME, LETHAL	CHRNA1	developmental
253290	MULTIPLE PTERYGIUM SYNDROME, LETHAL	CHRND	developmental
253290	MULTIPLE PTERYGIUM SYNDROME, LETHAL	CHRNG	developmental
265000	MULTIPLE PTERYGIUM SYNDROME, ESCOBAR	CHRNG	developmental
601378	CRISPONI SYNDROME	CRLF1	developmental

602398	DESMOSTEROLOSIS	DHCR24	developmental
270400	SMITH-LEMLI-OPITZ SYNDROME; SLOS	DHCR7	developmental
607598	ICOS DEFICIENCY; LCCS2	ERBB3	developmental
214150	CEREBROOCULOFACIOSKELETAL SYNDROME 1; COFS1	ERCC6	developmental
268300	ROBERTS SYNDROME; RBS	ESCO2	developmental
314390	VACTERL ASSOCIATION WITH HYDROCEPHALUS, XLR	FANCB	developmental
219000	FRASER SYNDROME	FRAS1	developmental
219000	FRASER SYNDROME	FREM2	developmental
190685	DOWN SYNDROME	GATA1	developmental
253310	LETHAL CONGENITAL CONTRACTURE SYNDROME 1; LCCS1	GLE1	developmental
200990	ACROCALLOSAL SYNDROME; ACLS	GLI3	developmental
236680	HYDROLETHALUS SYNDROME 1	HYLS1	developmental
300472	CORPUS CALLOSUM, AGENESIS OF, WITH MENTAL RETARDATION, OCULAR COLOBOMA,	IGBP1	developmental
222448	DONNAI-BARROW SYNDROME	LRP2	developmental
249000	MECKEL SYNDROME, 1; MKS1	MKS1	developmental
310600	NORRIE DISEASE; ND	NDP	developmental
300018	DOSAGE-SENSITIVE SEX REVERSAL; DSS	NR0B1	developmental
309000	LOWE OCULOCEREBRORENAL SYNDROME; OCRL	OCRL	developmental
300209	SIMPSON-GOLABI-BEHMEL SYNDROME, 2	OFD1	developmental
215100	RHIZOMELIC CHONDRODYSPLASIA PUNCTATA, 1; RCDP1	PEX7	developmental
601451	NEVO SYNDROME	PLOD1	developmental
309500	RENPENNING SYNDROME 1; RENS1	PQBP1	developmental
201000	CARPENTER SYNDROME	RAB23	developmental
600118	WARBURG MICRO SYNDROME; WARBM	RAB3GAP1	developmental
208150	FETAL AKINESIA DEATION SEQUENCE; FADS	RAPSN	developmental
257320	LISSENCEPHALY 2; LIS2	RELN	developmental
611561	MECKEL SYNDROME, 5; MKS5	RPGRIP1L	developmental
222600	DIASTROPHIC DYSPLASIA	SLC26A2	developmental
256050	ATELOSTEOGENESIS, II; AOII	SLC26A2	developmental
601186	MICROPHTHALMIA, SYNDROMIC 9; MCOPS9	STRA6	developmental
311150	OPTICOACOUSTIC NERVE ATROPHY WITH DEMENTIA	TIMM8A	developmental
253250	MULIBREY NANISM	TRIM37	developmental
243800	JOHANSON-BLIZZARD SYNDROME; JBS	UBR1	developmental
208085	ARTHROGRYPOSIS, RENAL DYSFUNCTION, AND CHOLESTASIS	VPS33B	developmental
273395	TETRA-AMELIA, AR	WNT3	developmental
306955	HETEROTAXY, VISCERAL, 1, XLR; HTX1	ZIC3	developmental
608612	MANDIBULOACRAL DYSPLASIA WITH B LIPODYSTROPHY; MADB	ZMPSTE24	developmental
231550	ACHALASIA-ADDISONIANISM-ALACRIMA SYNDROME; AAA	AAAS	endocrine
308370	INFERTILE MALE SYNDROME	AR	endocrine
201710	LIPOID CONGENITAL ADRENAL HYPERPLASIA	CYP11A1	endocrine
201910	ADRENAL HYPERPLASIA, CONGENITAL, DUE TO 21-HYDROXYLASE DEFICIENCY	CYP21A2	endocrine
264700	VITAMIN D-DEPENDENT RICKETS, I	CYP27B1	endocrine
262600	PITUITARY DWARFISM III	HESX1	endocrine
270450	INSULIN-LIKE GROWTH FACTOR I, RESISTANCE TO	IGF1	endocrine
246200	DONOHUE SYNDROME	INSR	endocrine
262600	PITUITARY DWARFISM III	LHX3	endocrine
262600	PITUITARY DWARFISM III	POU1F1	endocrine
262600	PITUITARY DWARFISM III	PROP1	endocrine
274600	PENDRED SYNDROME; PDS	SLC26A4	endocrine
201710	LIPOID CONGENITAL ADRENAL HYPERPLASIA	STAR	endocrine
244460	KENNY-CAFFEY SYNDROME, 1; KCS	TBCE	endocrine
275100	HYPOTHYROIDISM, CONGENITAL, NONGOITROUS, 4; CHNG4	TSHB	endocrine
300048	INTESTINAL PSEUDOOBSTRUCTION, NEURONAL, CHRONIC IDIOPATHIC, XLR	FLNA	gastroenterologic
608456	COLORECTAL ADENOMATOUS POLYPOSIS, AR	MUTYH	gastroenterologic
610370	DIARRHEA 4, MALABSORPTIVE, CONGENITAL	NEUROG3	gastroenterologic
274150	THROMBOTIC THROMBOCYTOPENIC PURPURA, CONGENITAL; TTP	ADAMTS13	hematologic
301040	α-THALASSEMIA/MENTAL RETARDATION SYNDROME,NONDELETION , XLR ATRX	ATRX	hematologic
612416	FACTOR XI DEFICIENCY	F11	hematologic
188055	THROMBOPHILIA DUE TO ACTIVATED PROTEIN C RESISTANCE	F5	hematologic
202400	AFIBRINOGENEMIA, CONGENITAL	FGA	hematologic
202400	AFIBRINOGENEMIA, CONGENITAL	FGB	hematologic
202400	AFIBRINOGENEMIA, CONGENITAL	FGG	hematologic
266130	GLUTATHIONE SYNTHETASE DEFICIENCY	GSS	hematologic
602390	HEMOCHROMATOSIS, JUVENILE; JH	HAMP	hematologic
141800	HEMOGLOBIN--ALPHA LOCUS 1; HBA1	HBA1	hematologic

141900	HEMOGLOBIN--BETA LOCUS; HBB	HBB	hematologic
603903	SICKLE CELL ANEMIA	HBB	hematologic
602390	HEMOCHROMATOSIS, JUVENILE; JH	HFE2	hematologic
604498	AMEGAKARYOCYTIC THROMBOCYTOPENIA, CONGENITAL; CAMT	MPL	hematologic
266200	PYRUVATE KINASE DEFICIENCY OF RED CELLS	PKLR	hematologic
217090	PLASMINOGEN DEFICIENCY, I	PLG	hematologic
612304	THROMBOPHILIA, HEREDITARY, DUE TO PROTEIN C DEFICIENCY, AUTOSOMAL	PROC	hematologic
260400	SHWACHMAN-DIAMOND SYNDROME; SDS	SBDS	hematologic
604250	HEMOCHROMATOSIS, 3	TFR2	hematologic
215600	CIRRHOSIS, FAMILIAL	KRT18	hepatic
215600	CIRRHOSIS, FAMILIAL	KRT8	hepatic
107400	PROTEASE INHIBITOR 1; PI	SERPINA1	hepatic
102700	SEVERE COMBINED IMMUNODEFICIENCY, AR, T CELL-NEGATIVE,	ADA	immunodeficiency
240300	AUTOIMMUNE POLYENDOCRINE SYNDROME, I; APS1	AIRE	immunodeficiency
208900	ATAXIA-TELANGIECTASIA; AT	ATM	immunodeficiency
210900	BLOOM SYNDROME; BLM	BLM	immunodeficiency
300755	AGAMMAGLOBULINEMIA, XLR XLA	BTK	immunodeficiency
308230	IMMUNODEFICIENCY WITH HYPER-IgM, 1; HIGM1	CD40LG	immunodeficiency
312060	PROPERDIN DEFICIENCY, XLR	CFP	immunodeficiency
603554	OMENN SYNDROME	DCLRE1C	immunodeficiency
300240	HOYERAAL-HREIDARSSON SYNDROME; HHS	DKC1	immunodeficiency
242860	IMMUNODEFICIENCY-CENTROMERIC INSTABILITY-FACIAL ANOMALIES SYNDROME	DNMT3B	immunodeficiency
601705	T-CELL IMMUNODEFICIENCY, CONGENITAL ALOPECIA, AND NAIL DYSTROPHY	FOXN1	immunodeficiency
304790	IMMUNODYSREGULATION, POLYENDOCRINOPATHY, AND ENTEROPATHY, XLR	FOXP3	immunodeficiency
300291	ECTODERMAL DYSPLASIA, HYPOHIDROTIC, WITH IMMUNE DEFICIENCY	IKBKG	immunodeficiency
300301	OSTEOPETROSIS, LYMPHEDEMA, ECTODERMAL DYSPLASIA, ANHIDROSIS, IMMUNODEFICIENCY, OLEDAID	IKBKG	immunodeficiency
300400	SEVERE COMBINED IMMUNODEFICIENCY, XLR; SCIDX1	IL2RG	immunodeficiency
312863	COMBINED IMMUNODEFICIENCY, XLR; CIDX	IL2RG	immunodeficiency
600802	SEVERE COMBINED IMMUNODEFICIENCY, AR, T CELL-NEGATIVE, B CELL-POSITIVE, NK CELL NEGATIVE	JAK3	immunodeficiency
214500	CHEDIAK-HIGASHI SYNDROME; CHS	LYST	immunodeficiency
249100	FAMILIAL MEDITERRANEAN FEVER; FMF	MEFV	immunodeficiency
251260	NIJMEGEN BREAKAGE SYNDROME	NBN	immunodeficiency
607624	GRISCELLI SYNDROME, 2; GS2	RAB27A	immunodeficiency
601457	SEVERE COMBINED IMMUNODEFICIENCY, AR, T CELL-NEGATIVE,	RAG1	immunodeficiency
603554	OMENN SYNDROME	RAG1	immunodeficiency
601457	SEVERE COMBINED IMMUNODEFICIENCY, AR, T CELL-NEGATIVE,	RAG2	immunodeficiency
603554	OMENN SYNDROME	RAG2	immunodeficiency
250250	CARTILAGE-HAIR HYPOPLASIA; CHH	RMRP	immunodeficiency
308240	LYMPHOPROLIFERATIVE SYNDROME, XLR, 1; XLP1	SH2D1A	immunodeficiency
235550	HEPATIC VENOOCCLUSIVE DISEASE WITH IMMUNODEFICIENCY; VODI	SP110	immunodeficiency
301000	WISKOTT-ALDRICH SYNDROME; WAS	WAS	immunodeficiency
601847	CHOLESTASIS, PROGRESSIVE FAMILIAL INTRAHEPATIC 2; PFIC2	ABCB11	metabolic
611126	ACYL-CoA DEHYDROGENASE FAMILY, MEMBER 9, DEFICIENCY OF	ACAD9	metabolic
201460	ACYL-CoA DEHYDROGENASE, LONG-CHAIN, DEFICIENCY OF	ACADL	metabolic
201450	ACYL-CoA DEHYDROGENASE, MEDIUM-CHAIN, DEFICIENCY OF	ACADM	metabolic
201470	ACYL-CoA DEHYDROGENASE, SHORT-CHAIN, DEFICIENCY OF	ACADS	metabolic
610006	2-METHYLBUTYRYL-CoA DEHYDROGENASE DEFICIENCY	ACADSB	metabolic
201475	ACYL-CoA DEHYDROGENASE, VERY LONG-CHAIN, DEFICIENCY OF	ACADVL	metabolic
264470	PEROXISOMAL ACYL-CoA OXIDASE DEFICIENCY	ACOX1	metabolic
208400	ASPARTYLGLUCOSAMINURIA	AGA	metabolic
232400	GLYCOGEN STORAGE DISEASE III	AGL	metabolic
600121	RHIZOMELIC CHONDRODYSPLASIA PUNCTATA, 3; RCDP3	AGPS	metabolic
259900	HYPEROXALURIA, PRIMARY, I	AGXT	metabolic
271980	SUCCINIC SEMIALDEHYDE DEHYDROGENASE DEFICIENCY	ALDH5A1	metabolic
229600	FRUCTOSE INTOLERANCE, HEREDITARY	ALDOB	metabolic
608540	CONGENITAL DISORDER OF GLYCOSYLATION, Ik; CDG1K	ALG1	metabolic
603147	CONGENITAL DISORDER OF GLYCOSYLATION, Ic; CDG1C	ALG6	metabolic
214950	BILE ACID SYNTHESIS DEFECT, CONGENITAL, 4	AMACR	metabolic
605899	GLYCINE ENCEPHALOPATHY; GCE	AMT	metabolic
253200	MUCOPOLYSACCHARIDOSIS VI	ARSB	metabolic
207900	ARGININOSUCCINIC ACIDURIA	ASL	metabolic
271900	CANAVAN DISEASE	ASPA	metabolic
215700	CITRULLINEMIA, CLASSIC	ASS1	metabolic
608688	AICAR TRANSYLASE/IMP CYCLOHYDROLASE, DEFICIENCY OF	ATIC	metabolic
277900	WILSON DISEASE	ATP7B	metabolic

211600	CHOLESTASIS, PROGRESSIVE FAMILIAL INTRAHEPATIC 1; PFIC1	ATP8B1	metabolic
250950	3-METHYLGLUTACONIC ACIDURIA, I	AUH	metabolic
607091	CONGENITAL DISORDER OF GLYCOSYLATION, IId; CDG2D	B4GALT1	metabolic
248600	MAPLE SYRUP URINE DISEASE Ia	BCKDHA	metabolic
248600	MAPLE SYRUP URINE DISEASE, CLASSIC, IB	BCKDHB	metabolic
124000	MITOCHONDRIAL COMPLEX III DEFICIENCY	BCS1L	metabolic
603358	GRACILE SYNDROME	BCS1L	metabolic
253260	BIOTINIDASE DEFICIENCY	BTD	metabolic
236200	HOMOCYSTINURIA	CBS	metabolic
237300	CARBAMOYL PHOSPHATE SYNTHETASE I DEFICIENCY, HYPERAMMONEMIA	CPS1	metabolic
255120	CARNITINE PALMITOYLTRANSFERASE I DEFICIENCY	CPT1A	metabolic
255110	CARNITINE PALMITOYLTRANSFERASE II DEFICIENCY, LATE-ONSET	CPT2	metabolic
600649	CARNITINE PALMITOYLTRANSFERASE II DEFICIENCY, INFANTILE	CPT2	metabolic
608836	CARNITINE PALMITOYLTRANSFERASE II DEFICIENCY, LETHAL NEONATAL	CPT2	metabolic
219750	CYSTINOSIS, ADULT NONNEPHROPATHIC	CTNS	metabolic
219800	CYSTINOSIS, NEPHROPATHIC; CTNS	CTNS	metabolic
219900	CYSTINOSIS, LATE-ONSET JUVENILE OR ADOLESCENT NEPHROPATHIC	CTNS	metabolic
265800	PYCNODYSOSTOSIS	CTSK	metabolic
213700	CEREBROTENDINOUS XANTHOMATOSIS	CYP27A1	metabolic
600721	D-2-HYDROXYGLUTARIC ACIDURIA	D2HGDH	metabolic
608643	AROMATIC L-AMINO ACID DECARBOXYLASE DEFICIENCY	DDC	metabolic
248600	MAPLE SYRUP URINE DISEASE III	DLD	metabolic
610198	3-METHYLGLUTACONIC ACIDURIA, V	DNAJC19	metabolic
610768	CONGENITAL DISORDER OF GLYCOSYLATION, Im; CDG1M	DOLK	metabolic
608093	CONGENITAL DISORDER OF GLYCOSYLATION, Ij; CDG1J	DPAGT1	metabolic
608799	CONGENITAL DISORDER OF GLYCOSYLATION, Ie; CDG1E	DPM1	metabolic
274270	DIHYDROPYRIMIDINE DEHYDROGENASE; DPYD	DPYD	metabolic
231680	MULTIPLE ACYL-CoA DEHYDROGENASE DEFICIENCY; MADD	ETFA	metabolic
231680	MULTIPLE ACYL-CoA DEHYDROGENASE DEFICIENCY; MADD	ETFB	metabolic
231680	MULTIPLE ACYL-CoA DEHYDROGENASE DEFICIENCY; MADD	ETFDH	metabolic
602473	ENCEPHALOPATHY, ETHYLMALONIC	ETHE1	metabolic
276700	TYROSINEMIA, I	FAH	metabolic
606812	FUMARASE DEFICIENCY	FH	metabolic
230000	FUCOSIDOSIS	FUCA1	metabolic
232200	GLYCOGEN STORAGE DISEASE I	G6PC3	metabolic
305900	GLUCOSE-6-PHOSPHATE DEHYDROGENASE; G6PD	G6PD	metabolic
232300	GLYCOGEN STORAGE DISEASE II	GAA	metabolic
245200	KRABBE DISEASE	GALC	metabolic
230400	GALACTOSEMIA	GALT	metabolic
230800	GAUCHER DISEASE, I	GBA	metabolic
230900	GAUCHER DISEASE, II	GBA	metabolic
231000	GAUCHER DISEASE, III	GBA	metabolic
608013	GAUCHER DISEASE, PERINATAL LETHAL	GBA	metabolic
232500	GLYCOGEN STORAGE DISEASE IV	GBE1	metabolic
231670	GLUTARIC ACIDEMIA I	GCDH	metabolic
605899	GLYCINE ENCEPHALOPATHY; GCE	GCSH	metabolic
609060	COMBINED OXIDATIVE PHOSPHORYLATION DEFICIENCY 1; COXPD1	GFM1	metabolic
301500	FABRY DISEASE	GLA	metabolic
230600	GM1-GANGLIOSIDOSIS, II	GLB1	metabolic
605899	GLYCINE ENCEPHALOPATHY; GCE	GLDC	metabolic
252500	MUCOLIPIDOSIS II ALPHA/BETA	GNPTAB	metabolic
252600	MUCOLIPIDOSIS III ALPHA/BETA	GNPTAB	metabolic
253230	MUCOPOLYSACCHARIDOSIS VIII	GNS	metabolic
260000	HYPEROXALURIA, PRIMARY, II	GRHPR	metabolic
253220	MUCOPOLYSACCHARIDOSIS VII	GUSB	metabolic
231530	3-HYDROXYACYL-CoA DEHYDROGENASE DEFICIENCY	HADH	metabolic
609016	LONG-CHAIN 3-HYDROXYACYL-CoA DEHYDROGENASE DEFICIENCY	HADHA	metabolic
609015	TRIFUNCTIONAL PROTEIN DEFICIENCY	HADHA	metabolic
609015	TRIFUNCTIONAL PROTEIN DEFICIENCY	HADHB	metabolic
235200	HEMOCHROMATOSIS; HFE	HFE	metabolic
235200	HEMOCHROMATOSIS; HFE	HFE2	metabolic
203500	ALKAPTONURIA	HGD	metabolic
252930	MUCOPOLYSACCHARIDOSIS IIIC	HGSNAT	metabolic
250620	BETA-HYDROXYISOBUTYRYL CoA DEACYLASE, DEFICIENCY OF	HIBCH	metabolic
246450	3-HYDROXY-3-METHYLGLUTARYL-CoA LYASE DEFICIENCY	HMGCL	metabolic

261515	D-BIFUNCTIONAL PROTEIN DEFICIENCY	HSD17B4	metabolic
607014	HURLER SYNDROME	IDUA	metabolic
243500	ISOVALERIC ACIDEMIA; IVA	IVD	metabolic
220111	LEIGH SYNDROME, FRENCH-CANADIAN ; LSFC	LRPPRC	metabolic
248500	MANNOSIDOSIS, ALPHA B, LYSOSOMAL	MAN2B1	metabolic
210210	3-METHYLCROTONYL-CoA CARBOXYLASE 2 DEFICIENCY	MCCC2	metabolic
252650	MUCOLIPIDOSIS IV	MCOLN1	metabolic
212066	CONGENITAL DISORDER OF GLYCOSYLATION, IIa; CDG2A	MGAT2	metabolic
251110	METHYLMALONIC ACIDURIA, cblB	MMAB	metabolic
277400	METHYLMALONIC ACIDURIA AND HOMOCYSTINURIA, cblC	MMACHC	metabolic
606056	CONGENITAL DISORDER OF GLYCOSYLATION, IIb; CDG2B	MOGS	metabolic
602579	CONGENITAL DISORDER OF GLYCOSYLATION, Ib; CDG1B	MPI	metabolic
610498	COMBINED OXIDATIVE PHOSPHORYLATION DEFICIENCY 2; COXPD2	MRPS16	metabolic
611719	COMBINED OXIDATIVE PHOSPHORYLATION DEFICIENCY 5; COXPD5	MRPS22	metabolic
236250	HOMOCYSTINURIA DUE TO DEFICIENCY OF N(5,10)-METHYLENETETRAHYDROFOLATE	MTHFR	metabolic
200100	ABETALIPOPROTEINEMIA; ABL	MTTP	metabolic
251000	METHYLMALONIC ACIDURIA DUE TO METHYLMALONYL-CoA MUTASE DEFICIENCY	MUT	metabolic
610377	MEVALONIC ACIDURIA	MVK	metabolic
609241	SCHINDLER DISEASE, I	NAGA	metabolic
237310	N-ACETYLGLUTAMATE SYNTHASE DEFICIENCY	NAGS	metabolic
256550	NEURAMINIDASE DEFICIENCY	NEU1	metabolic
257220	NIEMANN-PICK DISEASE, C1; NPC1	NPC1	metabolic
607625	NIEMANN-PICK DISEASE, C2	NPC2	metabolic
258501	3-METHYLGLUTACONIC ACIDURIA, III	OPA3	metabolic
311250	ORNITHINE TRANSCARBAMYLASE DEFICIENCY, HYPERAMMONEMIA DUE TO	OTC	metabolic
261600	PHENYLKETONURIA; PKU	PAH	metabolic
266150	PYRUVATE CARBOXYLASE DEFICIENCY	PC	metabolic
245349	PYRUVATE DEHYDROGENASE E3-BINDING PROTEIN DEFICIENCY	PDHX	metabolic
608782	PYRUVATE DEHYDROGENASE PHOSPHATASE DEFICIENCY	PDP1	metabolic
212065	CONGENITAL DISORDER OF GLYCOSYLATION, Ia; CDG1A	PMM2	metabolic
610090	PYRIDOXAMINE 5-PRIME-PHOSPHATE OXIDASE DEFICIENCY	PNPO	metabolic
261740	GLYCOGEN STORAGE DISEASE OF HEART, LETHAL CONGENITAL	PRKAG2	metabolic
611721	COMBINED SAPOSIN DEFICIENCY	PSAP	metabolic
610992	PHOSPHOSERINE AMINOTRANSFERASE DEFICIENCY	PSAT1	metabolic
607330	LATHOSTEROLOSIS	SC5DL	metabolic
604377	CARDIOENCEPHALOMYOPATHY, FATAL INFANTILE, DUE TO CYTOCHROME c OXIDASE	SCO2	metabolic
265120	SURFACTANT METABOLISM DYSFUNCTION, PULMONARY, 1; SMDP1	SFTPB	metabolic
252900	MUCOPOLYSACCHARIDOSIS IIIA	SGSH	metabolic
604369	SIALURIA, FINNISH	SLC17A5	metabolic
269920	INFANTILE SIALIC ACID STORAGE DISORDER	SLC17A5	metabolic
238970	HYPERORNITHINEMIA-HYPERAMMONEMIA-HOMOCITRULLINURIA SYNDROME	SLC25A15	metabolic
603585	CONGENITAL DISORDER OF GLYCOSYLATION, IIf; CDG2F	SLC35A1	metabolic
266265	CONGENITAL DISORDER OF GLYCOSYLATION, IIc; CDG2C	SLC35C1	metabolic
232220	GLYCOGEN STORAGE DISEASE Ib	SLC37A4	metabolic
232240	GLYCOGEN STORAGE DISEASE Ic	SLC37A4	metabolic
607616	NIEMANN-PICK DISEASE, B	SMPD1	metabolic
257200	NIEMANN-PICK DISEASE, A	SMPD1	metabolic
245400	LACTIC ACIDOSIS, FATAL INFANTILE	SUCLG1	metabolic
272300	SULFOCYSTEINURIA	SUOX	metabolic
605407	SEGAWA SYNDROME, AR	TH	metabolic
610505	COMBINED OXIDATIVE PHOSPHORYLATION DEFICIENCY 3; COXPD3	TSFM	metabolic
124000	MITOCHONDRIAL COMPLEX III DEFICIENCY	UQCRB	metabolic
124000	MITOCHONDRIAL COMPLEX III DEFICIENCY	UQCRQ	metabolic
203800	ALSTROM SYNDROME; ALMS	ALMS1	neurodegenerative
300100	ADRENOLEUKODYSTROPHY; ALD	ABCD1	neurological
225410	EHLERS-DANLOS SYNDROME, VII, AR	ADAMTS2	neurological
608629	JOUBERT SYNDROME 3; JBTS3	AHI1	neurological
270200	SJOGREN-LARSSON SYNDROME; SLS	ALDH3A2	neurological
205100	AMYOTROPHIC LATERAL SCLEROSIS 2, JUVENILE; ALS2	ALS2	neurological
606353	PRIMARY LATERAL SCLEROSIS, JUVENILE; PLSJ	ALS2	neurological
208920	ATAXIA, EARLY-ONSET, WITH OCULOMOTOR APRAXIA AND HYPOALBUMINEMIA;	APTX	neurological
607426	COENZYME Q10 DEFICIENCY	APTX	neurological
250100	METACHROMATIC LEUKODYSTROPHY	ARSA	neurological
308350	EPILEPTIC ENCEPHALOPATHY, EARLY INFANTILE, 1	ARX	neurological
300004	CORPUS CALLOSUM, AGENESIS OF, WITH ABNORMAL GENITALIA	ARX	neurological

309400	MENKES DISEASE	ATP7A	neurological
251880	MITOCHONDRIAL DNA DEPLETION SYNDROME, HEPATOCEREBRAL	C10ORF2	neurological
271245	INFANTILE-ONSET SPINOCEREBELLAR ATAXIA; IOSCA	C10ORF2	neurological
607426	COENZYME Q10 DEFICIENCY	CABC1	neurological
300672	EPILEPTIC ENCEPHALOPATHY, EARLY INFANTILE, 2	CDKL5	neurological
610188	JOUBERT SYNDROME 5; JBTS5	CEP290	neurological
204200	CEROID LIPOFUSCINOSIS, NEURONAL, 3; CLN3	CLN3	neurological
256731	CEROID LIPOFUSCINOSIS, NEURONAL, 5; CLN5	CLN5	neurological
601780	CEROID LIPOFUSCINOSIS, NEURONAL, 6; CLN6	CLN6	neurological
600143	CEROID LIPOFUSCINOSIS, NEURONAL, 8; CLN8	CLN8	neurological
610003	CEROID LIPOFUSCINOSIS, NEURONAL, 8, NORTHERN EPILEPSY	CLN8	neurological
607426	COENZYME Q10 DEFICIENCY	COQ2	neurological
254800	MYOCLONIC EPILEPSY OF UNVERRICHT AND LUNDBORG	CSTB	neurological
610127	CEROID LIPOFUSCINOSIS, NEURONAL, 10; CLN10	CTSD	neurological
300067	LISSENCEPHALY, XLR, 1; LISX1	DCX	neurological
251880	MITOCHONDRIAL DNA DEPLETION SYNDROME, HEPATOCEREBRAL	DGUOK	neurological
310200	MUSCULAR DYSTROPHY, DUCHENNE ; DMD	DMD	neurological
145900	HYPERTROPHIC NEUROPATHY OF DEJERINE-SOTTAS. CMT3, CMT4F	EGR2	neurological
605253	NEUROPATHY, HYPOMYELINATING/CHARCOT-MARIE-TOOTH DISEASE, 4E	EGR2	neurological
254780	MYOCLONIC EPILEPSY OF LAFORA	EPM2A	neurological
133540	COCKAYNE SYNDROME, B; CSB	ERCC6	neurological
216400	COCKAYNE SYNDROME, A; CSA	ERCC8	neurological
610532	LEUKODYSTROPHY, HYPOMYELINATING, 5	FAM126A	neurological
609311	CHARCOT-MARIE-TOOTH DISEASE, 4H; CMT4H	FGD4	neurological
606612	MUSCULAR DYSTROPHY, CONGENITAL, 1C; MDC1C	FKRP	neurological
253280	MUSCLE-EYE-BRAIN DISEASE; MEB	FKRP	neurological
253800	FUKUYAMA CONGENITAL MUSCULAR DYSTROPHY; FCMD	FKTN	neurological
608804	LEUKODYSTROPHY, HYPOMYELINATING, 2	GJC2	neurological
230500	GM1-GANGLIOSIDOSIS, I	GLB1	neurological
600737	INCLUSION BODY MYOPATHY 2, AR; IBM2	GNE	neurological
272800	TAY-SACHS DISEASE; TSD	HEXA	neurological
268800	SANDHOFF DISEASE	HEXB	neurological
300322	LESCH-NYHAN SYNDROME; LNS	HPRT1	neurological
300220	MENTAL RETARDATION, XLR, SYNDROMIC 10; MRXS10	HSD17B10	neurological
604320	SPINAL MUSCULAR ATROPHY, DISTAL, AR, 1; DSMA1	IGHMBP2	neurological
223900	NEUROPATHY, HEREDITARY SENSORY AND AUTONOMIC, III; HSAN3	IKBKAP	neurological
611726	EPILEPSY, PROGRESSIVE MYOCLONIC 3; EPM3	KCTD7	neurological
307000	HYDROCEPHALUS DUE TO CONGENITAL STENOSIS OF AQUEDUCT OF SYLVIUS; HSAS	L1CAM	neurological
304100	CORPUS CALLOSUM, PARTIAL AGENESIS OF, XLR	L1CAM	neurological
303350	MASA SYNDROME	L1CAM	neurological
607855	MUSCULAR DYSTROPHY, CONGENITAL MEROSIN-DEFICIENT, 1A; MDC1A	LAMA2	neurological
606369	EPILEPTIC ENCEPHALOPATHY, LENNOX-GASTAUT	MAPK10	neurological
105830	ANGELMAN SYNDROME AS	MECP2	neurological
300673	ENCEPHALOPATHY, NEONATAL SEVERE, DUE TO MECP2 MUTATIONS	MECP2	neurological
312750	RETT SYNDROME; RTT	MECP2	neurological
309520	LUJAN-FRYNS SYNDROME	MED12	neurological
610951	CEROID LIPOFUSCINOSIS, NEURONAL, 7; CLN7	MFSD8	neurological
604004	MEGALENCEPHALIC LEUKOENCEPHALOPATHY WITH SUBCORTICAL CYSTS; MLC	MLC1	neurological
252150	MOLYBDENUM COFACTOR DEFICIENCY	MOCS1	neurological
252150	MOLYBDENUM COFACTOR DEFICIENCY	MOCS2	neurological
256810	NAVAJO NEUROHEPATOPATHY; NN	MPV17	neurological
251880	MITOCHONDRIAL DNA DEPLETION SYNDROME, HEPATOCEREBRAL	MPV17	neurological
145900	HYPERTROPHIC NEUROPATHY OF DEJERINE-SOTTAS. CMT3, CMT4F	MPZ	neurological
605253	NEUROPATHY, HYPOMYELINATING/CHARCOT-MARIE-TOOTH DISEASE, 4E	MPZ	neurological
310400	MYOTUBULAR MYOPATHY 1; MTM1	MTM1	neurological
214450	GRISCELLI SYNDROME, 1; GS1	MYO5A	neurological
256710	ELEJALDE DISEASE	MYO5A	neurological
256030	NEMALINE MYOPATHY 2; NEM2	NEB	neurological
254780	MYOCLONIC EPILEPSY OF LAFORA	NHLRC1	neurological
609583	JOUBERT SYNDROME 4; JBTS4	NPHP1	neurological
256800	INSENSITIVITY TO PAIN, CONGENITAL, WITH ANHIDROSIS; CIPA	NTRK1	neurological
271930	STRIATONIGRAL DEGENERATION, INFANTILE; SNDI	NUP62	neurological
206700	ANIRIDIA, CEREBELLAR ATAXIA, AND MENTAL DEFICIENCY	PAX6	neurological
308930	LEIGH SYNDROME, XLR	PDHA1	neurological
607426	COENZYME Q10 DEFICIENCY	PDSS1	neurological

607426	COENZYME Q10 DEFICIENCY	PDSS2	neurological
256600	NEUROAXONAL DYSTROPHY, INFANTILE; INAD1	PLA2G6	neurological
611067	SPINAL MUSCULAR ATROPHY, DISTAL, AR, 4; DSMA4	PLEKHG5	neurological
312080	PELIZAEUS-MERZBACHER DISEASE; PMD	PLP1	neurological
312920	SPASTIC PARAPLEGIA 2, XLR; SPG2	PLP1	neurological
145900	HYPERTROPHIC NEUROPATHY OF DEJERINE-SOTTAS. CMT3, CMT4F	PMP22	neurological
203700	ALPERS DIFFUSE DEGENERATION OF CEREBRAL GRAY MATTER WITH HEPATIC CIRRHOSIS	POLG	neurological
253280	MUSCLE-EYE-BRAIN DISEASE; MEB	POMGNT1	neurological
236670	WALKER-WARBURG SYNDROME; WWS	POMT1	neurological
236670	WALKER-WARBURG SYNDROME; WWS	POMT2	neurological
256730	CEROID LIPOFUSCINOSIS, NEURONAL, 1; CLN1	PPT1	neurological
301835	ARTS SYNDROME; ARTS	PRPS1	neurological
145900	HYPERTROPHIC NEUROPATHY OF DEJERINE-SOTTAS. CMT3, CMT4F	PRX	neurological
249900	METACHROMATIC LEUKODYSTROPHY DUE TO SAPOSIN B DEFICIENCY	PSAP	neurological
611722	KRABBE DISEASE, ATYPICAL, DUE TO SAPOSIN A DEFICIENCY	PSAP	neurological
212720	MARTSOLF SYNDROME	RAB3GAP2	neurological
270550	SPASTIC ATAXIA, CHARLEVOIX-SAGUENAY ; SACS	SACS	neurological
602771	RIGID SPINE MUSCULAR DYSTROPHY 1; RSMD1	SEPN1	neurological
608099	MUSCULAR DYSTROPHY, LIMB-GIRDLE, 2D; LGMD2D	SGCA	neurological
248800	Marinesco-Sjogren Syndrome	SIL1	neurological
218000	AGENESIS OF THE CORPUS CALLOSUM WITH PERIPHERAL NEUROPATHY; ACCPN	SLC12A6	neurological
300523	ALLAN-HERNDON-DUDLEY SYNDROME AHDS	SLC16A2	neurological
609304	EPILEPTIC ENCEPHALOPATHY, EARLY INFANTILE, 3	SLC25A22	neurological
300352	CREATINE DEFICIENCY SYNDROME, XLR	SLC6A8	neurological
300243	MENTAL RETARDATION, XLR, SYNDROMIC, CHRISTIANSON	SLC9A6	neurological
253550	SPINAL MUSCULAR ATROPHY, II; SMA2	SMN1	neurological
253300	SPINAL MUSCULAR ATROPHY, I; SMA1	SMN1	neurological
253400	SPINAL MUSCULAR ATROPHY, III; SMA3	SMN1	neurological
609528	CEREBRAL DYSGENESIS, NEUROPATHY, ICHTHYOSIS, PALMOPLANTAR KERATODERMA	SNAP29	neurological
609056	AMISH INFANTILE EPILEPSY SYNDROME	ST3GAL5	neurological
612164	EPILEPTIC ENCEPHALOPATHY, EARLY INFANTILE, 4	STXBP1	neurological
241410	HYPOPARATHYROIDISM-RETARDATION-DYSMORPHISM SYNDROME; HRD	TBCE	neurological
609560	MITOCHONDRIAL DNA DEPLETION SYNDROME, MYOPATHIC	TK2	neurological
610688	JOUBERT SYNDROME 6; JBTS6	TMEM67	neurological
605355	NEMALINE MYOPATHY 5; NEM5	TNNT1	neurological
204500	CEROID LIPOFUSCINOSIS, NEURONAL, 2; CLN2	TPP1	neurological
225750	AICARDI-GOUTIERES SYNDROME 1; AGS1	TREX1	neurological
225753	PONTOCEREBELLAR HYPOPLASIA 4; PCH4	TSEN54	neurological
277470	PONTOCEREBELLAR HYPOPLASIA 2A; PCH2A	TSEN54	neurological
277460	VITAMIN E, FAMILIAL ISOLATED DEFICIENCY OF; VED	TTPA	neurological
301830	SPINAL MUSCULAR ATROPHY, XLR 2; SMAX2	UBA1	neurological
224050	CEREBELLAR HYPOPLASIA AND MENTAL RETARDATION WITH/WITHOUT QUADRUPEDAL	VLDLR	neurological
216550	COHEN SYNDROME; COH1	VPS13B	neurological
102770	MYOADENYLATE DEAMINASE DEFICIENCY, MYOPATHY DUE TO	AMPD1	neuromuscular
608840	MUSCULAR DYSTROPHY, CONGENITAL, 1D	LARGE	neuromuscular
601067	USHER SYNDROME, ID; USH1D	CDH23	ocular
303100	CHOROIDEREMIA; CHM	CHM	ocular
276902	USHER SYNDROME, III; USH3	CLRN1	ocular
262300	ACHROMATOPSIA 3; ACHM3	CNGB3	ocular
605472	USHER SYNDROME, IIC; USH2C	GPR98	ocular
276900	USHER SYNDROME, I	MYO7A	ocular
602083	USHER SYNDROME, IF; USH1F	PCDH15	ocular
312700	RETINOSCHISIS 1, XLR, JUVENILE; RS1	RS1	ocular
217400	CORNEAL DYSTROPHY AND PERCEPTIVE DEAFNESS	SLC4A11	ocular
276904	USHER SYNDROME, IC; USH1C	USH1C	ocular
606943	USHER SYNDROME, IG; USH1G	USH1G	ocular
276901	USHER SYNDROME, IIA; USH2A	USH2A	ocular
267430	RENAL TUBULAR DYSGENESIS; RTD	ACE	renal
267430	RENAL TUBULAR DYSGENESIS; RTD	AGT	renal
267430	RENAL TUBULAR DYSGENESIS; RTD	AGTR1	renal
248190	HYPOMAGNESEMIA, RENAL, WITH OCULAR INVOLVEMENT	CLDN19	renal
203780	ALPORT SYNDROME, AR	COL4A3	renal
203780	ALPORT SYNDROME, AR	COL4A4	renal
602088	NEPHRONOPHTHISIS 2; NPHP2	INVS	renal
609254	SENIOR-LOKEN SYNDROME 5; SLSN5	IQCB1	renal

241200	BARTTER SYNDROME, ANTENATAL, 2	KCNJ1	renal
609049	PIERSON SYNDROME	LAMB2	renal
256100	NEPHRONOPHTHISIS 1; NPHP1	NPHP1	renal
266900	SENIOR-LOKEN SYNDROME 1; SLSN1	NPHP1	renal
208540	RENAL-HEPATIC-PANCREATIC DYSPLASIA; RHPD	NPHP3	renal
606966	NEPHRONOPHTHISIS 4; NPHP4	NPHP4	renal
256300	NEPHROSIS 1, CONGENITAL, FINNISH ; NPHS1	NPHS1	renal
263200	POLYCYSTIC KIDNEY DISEASE, AR; ARPKD	PKHD1	renal
610725	NEPHROTIC SYNDROME, 3; NPHS3	PLCE1	renal
606407	HYPOTONIA-CYSTINURIA SYNDROME	PREPL	renal
267430	RENAL TUBULAR DYSGENESIS; RTD	REN	renal
264350	PSEUDOHYPOALDOSTERONISM, I, AR; PHA1	SCNN1A	renal
264350	PSEUDOHYPOALDOSTERONISM, I, AR; PHA1	SCNN1B	renal
264350	PSEUDOHYPOALDOSTERONISM, I, AR; PHA1	SCNN1G	renal
601678	BARTTER SYNDROME, ANTENATAL, 1	SLC12A1	renal
606407	HYPOTONIA-CYSTINURIA SYNDROME	SLC3A1	renal
256370	NEPHROTIC SYNDROME, EARLY-ONSET, WITH DIFFUSE MESANGIAL SCLEROSIS	WT1	renal
265450	PULMONARY VENOOCCLUSIVE DISEASE; PVOD	BMPR2	respiratory
219700	CYSTIC FIBROSIS; CF	CFTR	respiratory
265380	NEWBORN PULMONARY HYPERTENSION, FAMILIAL PERSISTENT	CPS1	respiratory
267450	RESPIRATORY DISTRESS SYNDROME IN PREMATURE INFANTS	SFTPA1	respiratory
267450	RESPIRATORY DISTRESS SYNDROME IN PREMATURE INFANTS	SFTPB	respiratory
267450	RESPIRATORY DISTRESS SYNDROME IN PREMATURE INFANTS	SFTPC	respiratory
265100	PULMONARY ALVEOLAR MICROLITHIASIS	SLC34A2	respiratory
608800	SUDDEN INFANT DEATH WITH DYSGENESIS OF THE TESTES SYNDROME; SIDDT	TSPYL1	respiratory
231050	GELEOPHYSIC DYSPLASIA	ADAMTSL2	skeletal
241510	HYPOPHOSPHATASIA, CHILDHOOD	ALPL	skeletal
236490	HYALINOSIS, INFANTILE SYSTEMIC	ANTXR2	skeletal
210600	SECKEL SYNDROME 1	ATR	skeletal
259730	OSTEOPETROSIS, AR 3; OPTB3	CA2	skeletal
215150	OTOSPONDYLOMEGAEPIPHYSEAL DYSPLASIA; OSMED	COL11A2	skeletal
215150	OTOSPONDYLOMEGAEPIPHYSEAL DYSPLASIA; OSMED	COL2A1	skeletal
610854	OSTEOGENESIS IMPERFECTA, IIB	CRTAP	skeletal
277300	SPONDYLOCOSTAL DYSOSTOSIS, AR 1; SCDO1	DLL3	skeletal
241520	HYPOPHOSPHATEMIC RICKETS, AR	DMP1	skeletal
226980	EPIPHYSEAL DYSPLASIA, MULTIPLE, WITH EARLY-ONSET DIABETES MELLITUS	EIF2AK3	skeletal
259775	RAINE SYNDROME; RNS	FAM20C	skeletal
207410	ANTLEY-BIXLER SYNDROME; ABS	FGFR2	skeletal
224410	DYSSEGMENTAL DYSPLASIA, SILVERMAN-HANDMAKER ; DDSH	HSPG2	skeletal
215140	HYDROPS-ECTOPIC CALCIFICATION-MOTH-EATEN SKELETAL DYSPLASIA	LBR	skeletal
610915	OSTEOGENESIS IMPERFECTA, VIII	LEPRE1	skeletal
601559	STUVE-WIEDEMANN SYNDROME	LIFR	skeletal
259770	OSTEOPOROSIS-PSEUDOGLIOMA SYNDROME; OPPG	LRP5	skeletal
259720	OSTEOPETROSIS, AR 5; OPTB5	OSTM1	skeletal
215045	CHONDRODYSPLASIA, BLOMSTRAND ; BOCD	PTH1R	skeletal
607095	ANAUXETIC DYSPLASIA	RMRP	skeletal
600972	ACHONDROGENESIS, IB; ACG1B	SLC26A2	skeletal
269250	SCHNECKENBECKEN DYSPLASIA	SLC35D1	skeletal
259700	OSTEOPETROSIS, AR 1; OPTB1	TCIRG1	skeletal
239000	PAGET DISEASE, JUVENILE	TNFRSF11B	skeletal
277440	VITAMIN D-DEPENDENT RICKETS, II	VDR	skeletal
228930	FIBULAR APLASIA OR HYPOPLASIA, FEMORAL BOWING AND POLY-, SYN-, AND	WNT7A	skeletal
276820	ULNA AND FIBULA, ABSENCE OF, WITH SEVERE LIMB DEFICIENCY	WNT7A	skeletal