InterGenetics
DIAGNOSTIC GENETIC CENTER

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Progenomis®

	GENE SYMBOL	DISEASE	OMIM ID
1	AAAS	ACHALASIA-ADDISONIANISM-ALACRIMA SYNDROME	231550
2	ABCA12	ICHTHYOSIS CONGENITA, HARLEQUIN FETUS TYPE	242500
3	ABCA3	SURFACTANT METABOLISM DYSFUNCTION, PULMONARY, 3	610921
4	ABCA4	STARGARDT DISEASE	248200
5	ABCB11	CHOLESTASIS, PROGRESSIVE FAMILIAL INTRAHEPATIC 2	601847
6	ABCB4	CHOLESTASIS, PROGRESSIVE FAMILIAL INTRAHEPATIC	602347
7	ABCB7	ANEMIA, SIDEROBLASTIC, WITH ATAXIA, X-LINKED	301310
8	ABCC8	HYPERINSULINEMIC HYPOGLYCEMIA, FAMILIAL, 1	256450
9	ABCD1	ADRENOLEUKODYSTROPHY	300100
10	ACAD9	DEFICIENCY OF ACYL-CoA DEHYDROGENASE FAMILY MEMBER 9	611126
11	ACADL	ACYL-CoA DEHYDROGENASE, LONG-CHAIN, DEFICIENCY OF	201475
12	ACADM	ACYL-CoA DEHYDROGENASE, MEDIUM-CHAIN, DEFICIENCY OF	201450
13	ACADS	ACYL-CoA DEHYDROGENASE, SHORT-CHAIN, DEFICIENCY OF	201470
14	ACADSB	2-METHYLBUTYRYLGLYCINURIA	610006
15	ACADVL	ACYL-CoA DEHYDROGENASE, VERY LONG-CHAIN, DEFICIENCY OF	201475
16	ACAT1	ALPHA-METHYLACETOACETIC ACIDURIA	203750
17	ACE	RENAL TUBULAR DYSGENESIS	267430
18	ACOX1	PEROXISOMAL ACYL-CoA OXIDASE DEFICIENCY	264470
19	ACSL4	MENTAL RETARDATION, X-LINKED 68	300387
20	ADA	SEVERE COMBINED IMMUNODEFICIENCY, AUT REC, T CELL-NEGATIVE,	102700
21	ADAMTS13	THROMBOTIC THROMBOCYTOPENIC PURPURA, CONGENITAL	274150
22	ADAMTS2	EHLERS-DANLOS SYNDROME, TYPE VIIC	225410
23	ADAMTSL2	GELEOPHYSIC DYSPLASIA	231050
24	AFF2	MENTAL RETARDATION X-LINKED ASSOCIATED WITH FRAGILE SITE	309548
25	AGA	ASPARTYLGLUCOSAMINURIA	208400
26	AGL	GLYCOGEN STORAGE DISEASE III	232400
27	AGPS	RHIZOMELIC CHONDRODYSPLASIA PUNCTATA, TYPE 3	600121
28	AGT	RENAL TUBULAR DYSGENESIS	267430
29	AGTR1	RENAL TUBULAR DYSGENESIS	267430
30	AGTR2	MENTAL RETARDATION X-LINKED 88	300034
31	AGXT	HYPEROXALURIA, PRIMARY, TYPE 1	259900
32	AHI1	JOUBERT SYNDROME 3	608629
33	AIFM1	COMBINED OXIDATIVE PHOSPHORYLATION DEFICIENCY 6, X-LINKED	300816
34	AIRE	AUTOIMMUNE POLYENDOCRINE SYNDROME TYPE I	240300
35	ALDH3A2	SJOGREN-LARSSON SYNDROME	270200
36	ALDH5A1	SUCCINIC SEMIALDEHYDE DEHYDROGENASE DEFICIENCY	271980
37	ALDH7A1	EPILEPSY, PYRIDOXINE-DEPENDENT	266100
38	ALDOB	FRUCTOSE INTOLERANCE, HEREDITARY	229600
39	ALG1	CONGENITAL DISORDER OF GLYCOSYLATION, TYPE Ik	608540
40	ALG12	CONGENITAL DISORDER OF GLYCOSYLATION TYPE Ig	607143
41	ALG2	CONGENITAL DISORDER OF GLYCOSYLATION TYPE Ii	607906
42	ALG3	CONGENITAL DISORDER OF GLYCOSYLATION, TYPE Id	601110
43	ALG6	CONGENITAL DISORDER OF GLYCOSYLATION TYPE Ic	603147
44	ALG8	CONGENITAL DISORDER OF GLYCOSYLATION, TYPE Ih	608104
45	ALG9	CONGENITAL DISORDER OF GLYCOSYLATION, TYPE Il	608776
46	ALMS1	ALSTROM SYNDROME	203800
47	ALPL	HYPOPHOSPHATASIA, CHILDHOOD	241510
48	ALS2	PRIMARY LATERAL SCLEROSIS, JUVENILE	606353
49	AMACR	BILE ACID SYNTHESIS DEFECT, CONGENITAL, 4	214950
50	AMPD1	MYOPATHY DUE TO MYOADENYLATE DEAMINASE DEFICIENCY	615511
51	AMT	GLYCINE ENCEPHALOPATHY	605899
52	ANTXR2	FIBROMATOSIS, JUVENILE HYALINE	228600
53	AP1S2	MENTAL RETARDATION, X-LINKED 59	300630
54	AP3B1	HERMANSKY-PUDLAK SYNDROME 2	608233
55	APTX	COENZYME Q10 DEFICIENCY	607426
56	AR	INFERTILE MALE SYNDROME	308370
57	ARHGEF6	MENTAL RETARDATION, X-LINKED 46	300436
58	ARHGEF9	HYPEREKPLEXIA AND EPILEPSY	300607
59	ARSA	METACHROMATIC LEUKODYSTROPHY	250100
60	ARSB	MUCOPOLYSACCHARIDOSIS TYPE VI	253200
61	ARSE	CHONDRODYSPLASIA PUNCTATA 1, X-LINKED RECESSIVE	302950
62	ARX	EPILEPTIC ENCEPHALOPATHY, EARLY INFANTILE, 1	308350

63	ASL	ARGININOSUCCINIC ACIDURIA	207900
64	ASPA	CANAVAN DISEASE	271900
65	ASS1	CITRULLINEMIA, CLASSIC	215700
66	ATIC	AICA-RIBOSIDURIA DUE TO ATIC DEFICIENCY	608688
67	ATM	ATAXIA-TELANGIECTASIA	208900
68	ATP2B3	SPINOCEREBELLAR ATAXIA, X-LINKED 1	302500
69	ATP6AP2	MENTAL RETARDATION, X-LINKED, SYNDROMIC, HEDERA TYPE	300423
70	ATP6V0A2	CUTIS LAXA, AUTOSOMAL RECESSIVE, TYPE II	219200
71	ATP7A	MENKES DISEASE	309400
72	ATP7B	WILSON DISEASE	277900
73	ATP8B1	CHOLESTASIS, PROGRESSIVE FAMILIAL INTRAHEPATIC 1	211600
74	ATR	SECKEL SYNDROME 1	210600
75	ATRX	α-THALASSEMIA/MENTAL RETARDATION SYNDROME,NONDELETION TYPE, X-LINKED	301040
76	AUH	3-METHYLGLUTACONIC ACIDURIA, TYPE I	250950
77	AVPR2	DIABETES INSIPIDUS, NEPHROGENIC, X-LINKED	304800
78	B3GAT3	MULTIPLE JOINT DISLOCATIONS, SHORT STATURE, CRANIOFACIAL DYSMORPHISM, AND CONGEI	245600
79	B4GALT1	CONGENITAL DISORDER OF GLYCOSYLATION TYPE IId	607091
80	BCKDHA	MAPLE SYRUP URINE DISEASE Type Ia	248600
81	BCKDHB	BRANCHED-CHAIN KETO ACID DEHYDROGENASE E1, BETA POLYPEPTIDE	248611
82	BCOR	MICROPHTHALMIA, SYNDROMIC	300166
83	BCS1L	MITOCHONDRIAL COMPLEX III DEFICIENCY	124000
84	BLM	BLOOM SYNDROME	210900
85	BLOC1S6	HERMANSKY-PUDLAK SYNDROME 9	604310
86	BMPR2	PULMONARY HYPERTENSION, PRIMARY, 1	178600
87	BRWD3	MENTAL RETARDATION, X-LINKED 93	300659
88	BTD	BIOTINIDASE DEFICIENCY	253260
89	BTK	AGAMMAGLOBULINEMIA, X-LINKED XLA	300755
90	C10orf2	INFANTILE-ONSET SPINOCEREBELLAR ATAXIA	271245
91	CA2	OSTEOPETROSIS, AUTOSOMAL RECESSIVE 3	259730
92	CABC1	COENZYME Q10 DEFICIENCY	607426
93	CACNA1F	NIGHT BLINDNESS, CONGENITAL STATIONARY, TYPE 2A, X-LINKED	300071
94	CASK	MENTAL RETARDATION AND MICROCEPHALY WITH PONTINE AND CEREBELLAR HYPOPLASIA	300749
95	CBS	HOMOCYSTINURIA	236200
96	CD19	IMMUNODEFICIENCY, COMMON VARIABLE, 3	613493
97	CD3D	SCID, AUT REC, T CELL-NEGATIVE, B CELL+, NK CELL+	186790
98	CD3E	IMMUNODEFICIENCY DUE TO DEFECT IN CD3-EPSILON	186830
99	CD3G	IMMUNODEFICIENCY DUE TO DEFECT IN CD3-GAMMA	186740
100	CD3Z	IMMUNODEFICIENCY DUE TO DEFECT IN CD3-ZETA	186780
101	CD40LG	IMMUNODEFICIENCY WITH HYPER-IgM, TYPE 1	308230
102	CD96	C SYNDROME	211750
103	CDH23	USHER SYNDROME, TYPE ID	601067
104	CDKL5	EPILEPTIC ENCEPHALOPATHY, EARLY INFANTILE, 2	300672
105	CEP120	SHORT-RIB THORACIC DYSPLASIA 13 WITH OR WITHOUT POLYDACTYLY	613446
106	CEP290	JOUBERT SYNDROME 5	610188
107	CFP	PROPERDIN DEFICIENCY, X-LINKED	312060
108	CFTR	CYSTIC FIBROSIS	219700
109	CHM	CHOROIDEREMIA, X-LINKED	303100
110	CHRDL1	MEGALOCORNEA, X-LINKED	309300
111	CHRNA1	MULTIPLE PTERYGIUM SYNDROME, LETHAL TYPE	253290
112	CHRND	MULTIPLE PTERYGIUM SYNDROME, LETHAL TYPE	253290
113	CHRNG	MULTIPLE PTERYGIUM SYNDROME, LETHAL TYPE	253290
114	CKAP2L	FILIPPI SYNDROME	272440
115	CLCN5	DENT DISEASE 1	300009
116	CLCN7	OSTEOPETROSIS, AUTOSOMAL RECESSIVE 4	611490
117	CLDN1	ICHTHYOSIS, LEUKOCYTE VACUOLES, ALOPECIA, AND SCLEROSING CHOLANGITIS	607626
118	CLDN19	HYPOMAGNESEMIA, RENAL, WITH OCULAR INVOLVEMENT	248190
119	CLIC2	MENTAL RETARDATION, X-LINKED, SYNDROMIC 32	300886
120	CLN3	NEURONAL CEROID LIPOFUSCINOSIS 3	204200
121	CLN5	NEURONAL CEROID LIPOFUSCINOSIS 5	256731
122	CLN6	CEROID LIPOFUSCINOSIS, NEURONAL, 6	601780
123	CLN8	CEROID LIPOFUSCINOSIS, NEURONAL, 8	600143
124	CLRN1	USHER SYNDROME TYPE 3A	276902
125	CNGB3	ACHROMATOPSIA 3	262300
126	COG1	CONGENITAL DISORDER OF GLYCOSYLATION TYPE Iig	611209
127	COG7	CONGENITAL DISORDER OF GLYCOSYLATION, TYPE Iie	608779
128	COG8	CONGENITAL DISORDER OF GLYCOSYLATION TYPE Iih	611182
129	COL11A2	OTOSPONDYLOMEGAEPIPHYSEAL DYSPLASIA	215150
130	COL17A1	EPIDERMOLYSIS BULLOSA, JUNCTIONAL, NON-HERLITZ TYPE	226650
131	COL1A2	OSTEOGENESIS IMPERFECTA, TYPE II	166210

132	COL4A3	ALPORT SYNDROME, AUTOSOMAL RECESSIVE	203780
133	COL4A4	ALPORT SYNDROME, AUTOSOMAL RECESSIVE	203780
134	COL4A5	ALPORT SYNDROME, X-LINKED	301050
135	COL4A6	DEAFNESS, X-LINKED 6	300914
136	COL7A1	EPIDERMOLYSIS BULLOSA DYSTROPHICA, AUTOSOMAL RECESSIVE	226600
137	COQ2	COENZYME Q10 DEFICIENCY	607426
138	COQ9	COENZYME Q10 DEFICIENCY	607426
139	COX10	COMPLEX IV DEFICIENCY	220110
140	COX15	COMPLEX IV DEFICIENCY	220110
141	COX6B1	COMPLEX IV DEFICIENCY	220110
142	CPS1	CARBAMOYL PHOSPHATE SYNTHETASE I DEFICIENCY, HYPERAMMONEMIA DUE TO	237300
143	CPT1A	CARNITINE PALMITOYLTRANSFERASE I DEFICIENCY	255120
144	CPT2	CARNITINE PALMITOYLTRANSFERASE II DEFICIENCY, LETHAL NEONATAL	608836
145	CRLF1	CRISPONI SYNDROME	601378
146	CRTAP	OSTEOGENESIS IMPERFECTA, TYPE IIB	610854
147	CSF2RA	SURFACTANT METABOLISM DYSFUNCTION, PULMONARY, 4, X-LINKED	300770
148	CSTB	MYOCLONIC EPILEPSY OF UNVERRICHT AND LUNDBORG	254800
149	CTNS	CYSTINOSIS, NEPHROPATHIC	219800
150	CTSD	CEROID LIPOFUSCINOSIS, NEURONAL, 10	610127
151	CTSK	PYCNODYSOSTOSIS	265800
152	CUL4B	MENTAL RETARDATION X-LINKED WITH BRACHYDACTYLY AND MACROGLOSSIA	300639
153	CYBB	GRANULOMATOUS DISEASE, CHRONIC, X-LINKED	306400
154	CYP11A1	LIPOID CONGENITAL ADRENAL HYPERPLASIA	201710
155	CYP11B1	CONGENITAL ADRENAL HYPERPLASIA	202010
156	CYP17A1	CONGENITAL ADRENAL HYPERPLASIA	202110
157	CYP21A2	ADRENAL HYPERPLASIA, CONGENITAL, DUE TO 21-HYDROXYLASE DEFICIENCY	201910
158	CYP27A1	CEREBROTENDINOUS XANTHOMATOSIS	213700
159	CYP27B1	VITAMIN D-DEPENDENT osteopenia, TYPE I	264700
160	D2HGDH	D-2-HYDROXYGLUTARIC ACIDURIA 1	600721
161	DBT	MSUD type 2	248610
162	DCLRE1C	OMENN SYNDROME	603554
163	DCX	LISSENCEPHALY, X-LINKED, 1	300067
164	DDB2	XERODERMA PIGMENTOSUM, COMPLEMENTATION GROUP E	278740
165	DDC	AROMATIC L-AMINO ACID DECARBOXYLASE DEFICIENCY	608643
166	DGUOK	MITOCHONDRIAL DNA DEPLETION SYNDROME, HEPATOCEREBRAL FORM	251880
167	DHCR24	DESMOSTEROLOSIS	602398
168	DHCR7	SMITH-LEMLI-OPITZ SYNDROME	270400
169	DKC1	HOYERAAL-HREIDARSSON SYNDROME	300240
170	DLAT	PYRUVATE DEHYDROGENASE E2 DEFICIENCY	245348
171	DLD	DIHYDROLIPOAMIDE DEHYDROGENASE DEFICIENCY	238331
172	DLG3	MENTAL RETARDATION X-LINKED 90	300189
173	DLL3	SPONDYLOCOSTAL DYSOSTOSIS 1, AUTOSOMAL RECESSIVE	277300
174	DMD	MUSCULAR DYSTROPHY, DUCHENNE TYPE	310200
175	DMP1	HYPOPHOSPHATEMIC OSTEOPENIA, AUTOSOMAL RECESSIVE	241520
176	DNAJC19	3-METHYLGLUTACONIC ACIDURIA, TYPE V	610198
177	DNMT3B	IMMUNODEFICIENCY-CENTROMERIC INSTABILITY-FACIAL ANOMALIES SYNDROME	242860
178	DOCK8	HYPER-IgE RECURRENT INFECTION SYNDROME, AUTOSOMAL RECESSIVE	243700
179	DOLK	CONGENITAL DISORDER OF GLYCOSYLATION TYPE Im	610768
180	DPAGT1	CONGENITAL DISORDER OF GLYCOSYLATION, TYPE Ij	608093
181	DPM1	CONGENITAL DISORDER OF GLYCOSYLATION, TYPE Ie	608799
182	DPYD	DIHYDROPYRIMIDINE DEHYDROGENASE	274270
183	DSP	EPIDERMOLYSIS BULLOSA, LETHAL ACANTHOLYTIC	609638
184	DYNC2H1	ASPHYXIATING THORACIC DYSTROPHY 3	613091
185	EDA	ECTODERMAL DYSPLASIA, HYPOHIDROTIC, X-LINKED	305100
186	EDN3	WAARDENBURG-SHAH SYNDROME	277580
187	EDNRB	ABCD SYNDROME	600501
188	EFEMP2	CUTIS LAXA, AUTOSOMAL RECESSIVE, TYPE I	219100
189	EFNB1	CRANIOFRONTONASAL SYNDROME	304110
190	EGR2	HYPERTROPHIC NEUROPATHY OF DEJERINE-SOTTAS	145900
191	EIF2AK3	EPIPHYSEAL DYSPLASIA, MULTIPLE, WITH EARLY-ONSET DIABETES MELLITUS	226980
192	EMD	EMERY-DREIFUSS MUSCULAR DYSTROPHY, X-LINKED	310300
193	ENPP1	ARTERIAL CALCIFICATION, GENERALIZED, OF INFANCY	208000
194	EPM2A	MYOCLONIC EPILEPSY OF LAFORA	254780
195	ERBB3	LETHAL CONGENITAL CONTRACTURE SYNDROME 2	607598
196	ERCC2	TRICHOTHIODYSTROPHY, PHOTOSENSITIVE	601675
197	ERCC3	XERODERMA PIGMENTOSUM, COMPLEMENTATION GROUP B	610651
198	ERCC4	XERODERMA PIGMENTOSUM, COMPLEMENTATION GROUP F	278760
199	ERCC5	XERODERMA PIGMENTOSUM, COMPLEMENTATION GROUP G	278780
200	ERCC6	CEREBROOCULOFACIOSKELETAL SYNDROME 1	214150

201	ERCC8	COCKAYNE SYNDROME, TYPE A	216400
202	ESCO2	ROBERTS SYNDROME	268300
203	ETFA	MULTIPLE ACYL-CoA DEHYDROGENASE DEFICIENCY	231680
204	ETFB	MULTIPLE ACYL-CoA DEHYDROGENASE DEFICIENCY	231680
205	ETFDH	MULTIPLE ACYL-CoA DEHYDROGENASE DEFICIENCY	231680
206	ETHE1	ENCEPHALOPATHY, ETHYLMALONIC	602473
207	EVC	ELLIS-VAN CREVELD SYNDROME	225500
208	EVC2	ELLIS-VAN CREVELD SYNDROME	607261
209	F11	FACTOR XI DEFICIENCY	612416
210	F5	FACTOR V DEFICIENCY	227400
211	F8	HEMOPHILIA A	306700
212	F9	HEMOPHILIA B	300746
213	FAH	TYROSINEMIA, TYPE I	276700
214	FAM126A	LEUKODYSTROPHY, HYPOMYELINATING, 5	610532
215	FAM20C	RAINE SYNDROME	259775
216	FANCA	FANCONI ANEMIA, COMPLEMENTATION GROUP A	227650
217	FANCB	FANCONI ANEMIA, COMPLEMENTATION GROUP B	300514
218	FANCC	FANCONI ANEMIA, COMPLEMENTATION GROUP C	227645
219	FANCD1	FANCONI ANEMIA, COMPLEMENTATION GROUP D1	605724
220	FANCD2	FANCONI ANEMIA, COMPLEMENTATION GROUP D2	227646
221	FANCE	FANCONI ANEMIA, COMPLEMENTATION GROUP E	600901
222	FANCF	FANCONI ANEMIA, COMPLEMENTATION GROUP F	603467
223	FANCG	FANCONI ANEMIA, COMPLEMENTATION GROUP G	614082
224	FANCI	FANCONI ANEMIA, COMPLEMENTATION GROUP I	609053
225	FANCJ	FANCONI ANEMIA, COMPLEMENTATION GROUP J	609054
226	FANCL	FANCONI ANEMIA, COMPLEMENTATION GROUP L	614083
227	FANCM	FANCONI ANEMIA, COMPLEMENTATION GROUP M	614087
228	FANCN	FANCONI ANEMIA, COMPLEMENTATION GROUP N	610832
229	FANCO	FANCONI ANEMIA, COMPLEMENTATION GROUP O	613390
230	FANCP	FANCONI ANEMIA, COMPLEMENTATION GROUP P	613951
231	FASTKD2	Complex IV deficiency	220110
232	FBLN5	CUTIS LAXA, AUTOSOMAL RECESSIVE, TYPE I	219100
233	FERMT3	LEUKOCYTE ADHESION DEFICIENCY TYPE III	612840
234	FGA	AFIBRINOGENEMIA, CONGENITAL	202400
235	FGB	AFIBRINOGENEMIA, CONGENITAL	202400
236	FGD1	FACIOGENITAL DYSPLASIA	305400
237	FGD4	CHARCOT-MARIE-TOOTH DISEASE, TYPE 4H	609311
238	FGF16	METACARPAL 4-5 FUSION, X-LINKED	309630
239	FGG	AFIBRINOGENEMIA, CONGENITAL	202400
240	FH	FUMARASE DEFICIENCY	606812
241	FKRP	MUSCULAR DYSTROPHY, CONGENITAL, 1C	606612
242	FKTN	FUKUYAMA CONGENITAL MUSCULAR DYSTROPHY	253800
243	FLNA	CARDIAC VALVULAR DYSPLASIA, X-LINKED	314400
244	FOLR1	NEURODEGENERATION DUE TO CEREBRAL FOLATE TRANSPORT DEFICIENCY	613068
245	FOXN1	T-CELL IMMUNODEFICIENCY, CONGENITAL ALOPECIA, AND NAIL DYSTROPHY	601705
246	FOXP3	IMMUNODYSREGULATION, POLYENDOCRINOPATHY, AND ENTEROPATHY, X-LINKED	304790
247	FRAS1	FRASER SYNDROME	219000
248	FREM2	FRASER SYNDROME	219000
249	FTSJ1	MENTAL RETARDATION, X-LINKED 9	309549
250	FUCA1	FUCOSIDOSIS	230000
251	G6PC3	GLYCOGEN STORAGE DISEASE I	232200
252	G6PD	GLUCOSE-6-PHOSPHATE DEHYDROGENASE	305900
253	GAA	GLYCOGEN STORAGE DISEASE II	232300
254	GALC	KRABBE DISEASE	245200
255	GALK1	GALACTOKINASE DEFICIENCY	230200
256	GALT	GALACTOSEMIA	230400
257	GAMT	GUANIDINOACETATE METHYLTRANSFERASE DEFICIENCY	612736
258	GATA1	ANEMIA, X-LINKED, WITH OR WITHOUT NEUTROPENIA AND/OR PLATELET ABNORMALITIES	300835
259	GBA	GAUCHER DISEASE	608013
260	GBE1	GLYCOGEN STORAGE DISEASE IV	232500
261	GCDH	GLUTARIC ACIDEMIA I	231670
262	GCSH	GLYCINE ENCEPHALOPATHY	605899
263	GDAP1	CHARCOT-MARIE-TOOTH DISEASE TYPE 4A	214400
264	GDI1	MENTAL RETARDATION, X-LINKED 41, 48	309541
265	GFM1	COMBINED OXIDATIVE PHOSPHORYLATION DEFICIENCY 1	609060
266	GJA1	OCULODENTODIGITAL DYSPLASIA, AUTOSOMAL RECESSIVE	257850
267	GJB2	CONGENITAL DEAFNESS WITH KERATOPACHYDERMIA & CONSTRICTIONS OF FINGERS & TOES	124500
268	GJC2	LEUKODYSTROPHY, HYPOMYELINATING, 2	608804
269	GLA	FABRY DISEASE	301500

270	GLB1	GM1-GANGLIOSIDOSIS, TYPE I	230500
271	GLDC	GLYCINE ENCEPHALOPATHY	605899
272	GLE1	LETHAL CONGENITAL CONTRACTURE SYNDROME 1	253310
273	GNE	INCLUSION BODY MYOPATHY 2, AUTOSOMAL RECESSIVE	600737
274	GNPTAB	MUCOLIPIDOSIS II ALPHA/BETA	252500
275	GNRHR	HYPOGONADOTROPIC HYPOGONADISM	146110
276	GNS	MUCOPOLYSACCHARIDOSIS, TYPE IIID	252940
277	GPC3	SIMPSON-GOLABI-BEHMEL SYNDROME, TYPE 1	312870
278	GPR143	NYSTAGMUS, 6, CONGENITAL, X-LINKED	300814
279	GPR179	NIGHT BLINDNESS, CONGENITAL STATIONARY, TYPE 1E	614565
280	GPR98	USHER SYNDROME, TYPE IIC	605472
281	GRHPR	HYPEROXALURIA, PRIMARY, TYPE II	260000
282	GRIA3	MENTAL RETARDATION, X-LINKED 94	300699
283	GRIK2	MENTAL RETARDATION AUTOSOMAL RECESSIVE 6	611092
284	GSS	GLUTATHIONE SYNTHETASE DEFICIENCY	266130
285	GTF2H5	TRICHOTHIODYSTROPHY, PHOTOSENSITIVE	601675
286	GUSB	MUCOPOLYSACCHARIDOSIS TYPE VII SLY SYNDROME	253220
287	HADH	3-HYDROXYACYL-CoA DEHYDROGENASE DEFICIENCY	231530
288	HADHA	TRIFUNCTIONAL PROTEIN DEFICIENCY	609015
289	HADHB	TRIFUNCTIONAL PROTEIN DEFICIENCY	609015
290	HAMP	HEMOCHROMATOSIS, JUVENILE, TYPE 2B	602390
291	HAX1	NEUTROPENIA, SEVERE CONGENITAL, AUTOSOMAL RECESSIVE 3	610738
292	HBA1	ALPHA THALASSEMIA	141800
293	HBB	THALASSEMIA MAJOR	141900
294	HCFC1	MENTAL RETARDATION, X-LINKED 3	309451
295	HESX1	PITUITARY DWARFISM III	262600
296	HEXA	TAY-SACHS DISEASE	272800
297	HEXB	SANDHOFF DISEASE	268800
298	HFE	HEMOCHROMATOSIS, TYPE 1	235200
299	HFE2	HEMOCHROMATOSIS, TYPE 2	602390
300	HGD	ALKAPTONURIA	203500
301	HGSNAT	MUCOPOLYSACCHARIDOSIS TYPE IIIC (Sanfilippo type c)	252930
302	HIBCH	BETA-HYDROXYISOBUTYRYL CoA DEACYLASE, DEFICIENCY OF	250620
303	HJV	HEMOCHROMATOSIS, JUVENILE, TYPE 2A	602390
304	HLCS	HOLOCARBOXYLASE SYNTHETASE DEFICIENCY	253270
305	HMGCL	3-HYDROXY-3-METHYLGLUTARYL-CoA LYASE DEFICIENCY	246450
306	HPRT1	LESCH-NYHAN SYNDROME	300322
307	HSD11B2	CORTISOL 11-BETA-KETOREDUCTASE DEFICIENCY	218030
308	HSD17B10	MENTAL RETARDATION, X-LINKED, SYNDROMIC 10	300220
309	HSD17B3	17-@BETA HYDROXYSTEROID DEHYDROGENASE III DEFICIENCY	605573
310	HSD17B4	D-BIFUNCTIONAL PROTEIN DEFICIENCY	261515
311	HSD3B2	3-BETA-HYDROXYSTEROID DEHYDROGENASE DEFICIENCY TYPE II	201810
312	HSPG2	DYSSEGMENTAL DYSPLASIA, SILVERMAN-HANDMAKER TYPE	224410
313	HUWE1	MENTAL RETARDATION X-LINKED SYNDROMIC TURNER TYPE	300706
314	HYLS1	HYDROLETHALUS SYNDROME	236680
315	ICOS	ANTIBODY DEFICIENCY DUE TO ICOS DEFECT	607594
316	IDS	MUCOPOLYSACCHARIDOSIS TYPE II	309900
317	IDUA	HURLER SYNDROME	607014
318	IFNGR1	ATYPICAL MYCOBACTERIOSIS, FAMILIAL	209950
319	IFNGR2	ATYPICAL MYCOBACTERIOSIS, FAMILIAL	209950
320	IFT80	ASPHYXIATING THORACIC DYSTROPHY 2	611263
321	IFT140	SHORT-RIB THORACIC DYSPLASIA 9 WITH OR WITHOUT POLYDACTYLY	266920
322	IFT172	SHORT-RIB THORACIC DYSPLASIA 10 WITH OR WITHOUT POLYDACTYLY; RETINITIS PIGMENTOSA	615630
323	IGBP1	CORPUS CALLOSUM, AGENESIS OF, WITH MENTAL RETARDATION, OCULAR COLOBOMA, AND M	300472
324	IGF1	INSULIN-LIKE GROWTH FACTOR I DEFICIENCY	608747
325	IGHMBP2	SPINAL MUSCULAR ATROPHY, DISTAL, AUTOSOMAL RECESSIVE, 1	604320
326	IGSF1	HYPOTHYROIDISM, CENTRAL, AND TESTICULAR ENLARGEMENT, X-LINKED	300888
327	IKBKAP	NEUROPATHY, HEREDITARY SENSORY AND AUTONOMIC, TYPE III	223900
328	IKBKG	ECTODERMAL DYSPLASIA, HYPOHIDROTIC, WITH IMMUNE DEFICIENCY	300291
329	IL12B	ATYPICAL MYCOBACTERIOSIS, FAMILIAL	209950
330	IL12RB1	ATYPICAL MYCOBACTERIOSIS, FAMILIAL	209950
331	IL1RAPL1	MENTAL RETARDATION, X-LINKED 21	300143
332	IL1RN	DEFICIENCY OF INTERLEUKIN 1 RECEPTOR ANTAGONIST	612852
333	IL2RG	SEVERE COMBINED IMMUNODEFICIENCY, X-LINKED	300400
334	INSR	DONOHUE SYNDROME	246200
335	INVS	NEPHRONOPHTHISIS 2	602088
336	IQCB1	SENIOR-LOKEN SYNDROME 5	609254
337	IQSEC2	MENTAL RETARDATION, X-LINKED 1	309530
338	ITGA6	EPIDERMOLYSIS BULLOSA JUNCTIONALIS WITH PYLORIC ATRESIA	226730

339	ITGB4	EPIDERMOLYSIS BULLOSA, JUNCTIONAL, NON-HERLITZ TYPE	226650
340	IVD	ISOVALERIC ACIDEMIA	243500
341	JAK3	SEVERE COMBINED IMMUNODEFICIENCY, AUT REC, T CELL—, B CELL+, NK CELL—	600802
342	KAL1	HYPOGONADOTROPIC HYPOGONADISM 1 WITH ANOSMIA, X-LINKED	308700
343	KCNJ1	BARTTER SYNDROME, ANTENATAL, TYPE 2	241200
344	KCNQ1	JERVELL AND LANGE-NIELSEN SYNDROME 1	220400
345	KCTD7	EPILEPSY, PROGRESSIVE MYOCLONIC 3, WITH INTRACELLULAR INCLUSIONS	611726
346	KDM5C	MENTAL RETARDATION, X-LINKED, SYNDROMIC	314690
347	KIAA0586	JOUBERT SYNDROME 23; SHORT-RIB THORACIC DYSPLASIA 14 WITH POLYDACTYLY	616490
348	KIAA2022	MENTAL RETARDATION, X-LINKED 98	300912
349	KRT18	CIRRHOSIS, FAMILIAL	215600
350	KRT8	CIRRHOSIS, FAMILIAL	215600
351	L1CAM	CORPUS CALLOSUM, PARTIAL AGENESIS OF, X-LINKED	304100
352	LAMA2	MUSCULAR DYSTROPHY, CONGENITAL MEROSIN-DEFICIENT, 1A	607855
353	LAMA3	EPIDERMOLYSIS BULLOSA, JUNCTIONAL, NON-HERLITZ TYPE	226650
354	LAMB2	PIERSON SYNDROME	609049
355	LAMB3	EPIDERMOLYSIS BULLOSA, JUNCTIONAL, HERLITZ TYPE	226700
356	LAMC2	EPIDERMOLYSIS BULLOSA, JUNCTIONAL, HERLITZ TYPE	226700
357	LARGE	MUSCULAR DYSTROPHY, CONGENITAL, TYPE 1D	608840
358	LBR	HYDROPS-ECTOPIC CALCIFICATION-MOTH-EATEN SKELETAL DYSPLASIA	215140
359	LEPRE1	OSTEOGENESIS IMPERFECTA, TYPE VIII	610915
360	LHCGR	PRECOCIOUS PUBERTY, MALE-LIMITED	176410
361	LHX3	PITUITARY HORMONE DEFICIENCY, COMBINED, 3	221750
362	LIFR	STUVE-WIEDEMANN SYNDROME	601559
363	LIG4	SEVERE COMBINED IMMUNODEFICIENCY WITH SENSITIVITY TO IONIZING RADIATION	602450
364	LMNA	EMERY-DREIFUSS MUSCULAR DYSTROPHY, ATYPICAL, AUTOSOMAL RECESSIVE	181350
365	LRP2	DONNAI-BARROW SYNDROME	222448
366	LRP5	OSTEOPOROSIS-PSEUDOGLIOMA SYNDROME	259770
367	LRPPRC	LEIGH SYNDROME, FRENCH-CANADIAN TYPE	220111
368	LYST	CHEDIAK HIGASHI SYNDROME	214500
369	MAGT1	IMMUNODEFICIENCY, X-LINKED, WITH MAGNESIUM DEFECT, EPSTEIN-BARR VIRUS INFECTION, /	300853
370	MAMLD1	HYPOSPADIAS 2, X-LINKED	300758
371	MAN2B1	MANNOSIDOSIS, ALPHA B, LYSOSOMAL	248500
372	MAOA	BRUNNER SYNDROME, X-LINKED	300615
373	MBTPS2	ICHTHYOSIS FOLLICULARIS, ATRICHIA, AND PHOTOPHOBIA SYNDROME	308205
374	MCCC2	3-METHYLCROTONYL-CoA CARBOXYLASE 2 DEFICIENCY	210210
375	MCOLN1	MUCOLIPIDOSIS IV	252650
376	MECP2	RETT SYNDROME	312750
377	MED12	LUJAN-FRYNS SYNDROME	309520
378	MEFV	FAMILIAL MEDITERRANEAN FEVER	249100
379	MFSD8	CEROID LIPOFUSCINOSIS, NEURONAL, 7	610951
380	MGAT2	CONGENITAL DISORDER OF GLYCOSYLATIO, TYPE IIa	212066
381	MID1	OPITZ GBBB SYNDROME, X-LINKED	300000
382	MITF	WAARDENBURG SYNDROME, TYPE 2A	193510
383	MKS1	MECKEL SYNDROME TYPE 1	249000
384	MLC1	MEGALENCEPHALIC LEUKOENCEPHALOPATHY WITH SUBCORTICAL CYSTS	604004
385	MMAA	METHYLMALONIC ACIDURIA, cblA TYPE	251100
386	MMAB	METHYLMALONIC ACIDURIA, cblB TYPE	251110
387	MMACHC	METHYLMALONIC ACIDURIA AND HOMOCYSTINURIA, cblC TYPE	277400
388	MOCS1	MOLYBDENUM COFACTOR DEFICIENCY	252150
389	MOCS2	MOLYBDENUM COFACTOR DEFICIENCY	252150
390	MOGS	CONGENITAL DISORDER OF GLYCOSYLATION TYPE IIb	606056
391	MPDU1	CONGENITAL DISORDER OF GLYCOSYLATION, TYPE If	609180
392	MPI	CONGENITAL DISORDER OF GLYCOSYLATION, TYPE Ib	602579
393	MPL	AMEGAKARYOCYTIC THROMBOCYTOPENIA, CONGENITAL	604498
394	MPV17	MITOCHONDRIAL DNA DEPLETION SYNDROME, HEPATOCEREBRAL FORM	251880
395	MPZ	HYPERTROPHIC NEUROPATHY OF DEJERINE-SOTTAS	145900
396	MRPS16	COMBINED OXIDATIVE PHOSPHORYLATION DEFICIENCY 2	610498
397	MRPS22	COMBINED OXIDATIVE PHOSPHORYLATION DEFICIENCY 5	611719
398	MTHFR	HOMOCYSTINURIA DUE TO MTHFR DEFICIENCY	236250
399	MTM1	MYOTUBULAR MYOPATHY 1	310400
400	MUT	METHYLMALONIC ACIDURIA DUE TO METHYLMALONYL-CoA MUTASE DEFICIENCY	251000
401	MUTYH	FAMILIAL ADENOMATOUS POLYPOSIS 2	608456
402	MVK	MEVALONIC ACIDURIA	610377
403	MYD88D	MYD88 DEFICIENCY	612260
404	MYO5A	GRISCELLI SYNDROME, TYPE 1	214450
405	MYO7A	USHER SYNDROME, TYPE I	276900
406	NAA10	OGDEN SYNDROME, X-LINKED	300855
407	NAGA	SCHINDLER DISEASE, TYPE I	609241

408	NAGS	N-ACETYLGLUTAMATE SYNTHASE DEFICIENCY	237310
409	NBN	NIJMEGEN BREAKAGE SYNDROME	251260
410	NDP	NORRIE DISEASE	310600
411	NDUFA1	COMPLEX I DEFICIENCY	252010
412	NDUFA7	COMPLEX I DEFICIENCY	252010
413	NDUFAF2	COMPLEX I DEFICIENCY	252010
414	NDUFAF4	COMPLEX I DEFICIENCY	252010
415	NDUFS3	COMPLEX I DEFICIENCY	252010
416	NDUFS4	COMPLEX I DEFICIENCY	252010
417	NDUFS5	COMPLEX I DEFICIENCY	252010
418	NDUFS6	COMPLEX I DEFICIENCY	252010
419	NDUFS7	COMPLEX I DEFICIENCY	252010
420	NDUFS8	COMPLEX I DEFICIENCY	252010
421	NDUFV1	COMPLEX I DEFICIENCY	252010
422	NEB	NEMALINE MYOPATHY 2	256030
423	NEK1	SHORT-RIB THORACIC DYSPLASIA 6 WITH OR WITHOUT POLYDACTYLY	604588
424	NEU1	NEURAMINIDASE DEFICIENCY	256550
425	NEUROG3	DIARRHEA 4, MALABSORPTIVE, CONGENITAL	610370
426	NHEJ1	SCID W MICROCEPHALY, GROWTH RETARDATION, & SENS TO IONIZING RADIATION	611291
427	NHLRC1	MYOCLONIC EPILEPSY OF LAFORA	254780
428	NHS	NANCE-HORAN SYNDROME	302350
429	NLGN4X	MENTAL RETARDATION, X-LINKED	300495
430	NPC1	NIEMANN-PICK DISEASE, TYPE C1	257220
431	NPC2	NIEMANN-PICK DISEASE, TYPE C2	607625
432	NPHP1	NEPHRONOPHTHISIS 1	256100
433	NPHP3	RENAL-HEPATIC-PANCREATIC DYSPLASIA	208540
434	NPHP4	NEPHRONOPHTHISIS 4	606966
435	NPHS1	NEPHROSIS 1, CONGENITAL, FINNISH TYPE	256300
436	NPHS2	NEPHROTIC SYNDROME, TYPE 2	600995
437	NR0B1	CONGENITAL ADRENAL HYPOPLASIA	300200
438	NR5A1	GONADAL DYSGENESIS WITH ADRENAL FAILURE	612965
439	NSUN2	MENTAL RETARDATION, AUTOSOMAL RECESSIVE 5	610916
440	NTRK1	INSENSITIVITY TO PAIN, CONGENITAL, WITH ANHIDROSIS	256800
441	NUP62	STRIATONIGRAL DEGENERATION, INFANTILE	271930
442	NYX	NIGHT BLINDNESS, CONGENITAL STATIONARY, TYPE 1A, X-LINKED	310500
443	OCRL	LOWE OCULOCEREBRORENAL SYNDROME	309000
444	OFD1	SIMPSON-GOLABI-BEHMEL SYNDROME, TYPE 2	300209
445	OPA3	3-METHYLGLUTACONIC ACIDURIA, TYPE III	258501
446	OPHN1	MENTAL RETARDATION, XLR, W CEREBELLAR HYPOPLASIA & DISTINCTIVE FACIAL APPEARANCE	300486
447	OPN1LW	COLORBLINDNESS, PROTAN, X-LINKED	303900
448	OPN1MW	COLORBLINDNESS, DEUTAN, X-LINKED	303800
449	ORAI1	IMMUNE DYSFUNCTION WITH T-CELL INACTIVATION DUE TO CALCIUM ENTRY DEFECT 1	612782
450	OSTM1	OSTEOPETROSIS, AUTOSOMAL RECESSIVE 5	259720
451	OTC	ORNITHINE TRANSCARBAMYLASE DEFICIENCY, HYPERAMMONEMIA DUE TO	311250
452	PAH	PHENYLKETONURIA	261600
453	PAK3	MENTAL RETARDATION, X-LINKED 30	300558
454	PANK2	NEURODEGENERATION WITH BRAIN IRON ACCUMULATION 1 (Hallervorden-Spatz)	234200
455	PAX6	ANIRIDIA	106210
456	PC	PYRUVATE CARBOXYLASE DEFICIENCY	266150
457	PCCA	PROPIONIC ACIDEMIA	606054
458	PCCB	PROPIONIC ACIDEMIA	606054
459	PCDH15	DEAFNESS, AUTOSOMAL RECESSIVE 23	609533
460	PCDH19	EPILEPSY, FEMALE-RESTRICTED, WITH MENTAL RETARDATION	300088
461	PDHA1	LEIGH SYNDROME, X-LINKED	308930
462	PDHX	PYRUVATE DEHYDROGENASE E3-BINDING PROTEIN DEFICIENCY	245349
463	PDP1	PYRUVATE DEHYDROGENASE PHOSPHATASE DEFICIENCY	608782
464	PDSS1	COENZYME Q10 DEFICIENCY	607426
465	PDSS2	COENZYME Q10 DEFICIENCY	607426
466	PEX1	ZELLWEGER SYNDROME	214100
467	PEX10	ADRENOLEUKODYSTROPHY, AUTOSOMAL NEONATAL FORM | PEX10	202370
468	PEX12	ZELLWEGER SYNDROME	214100
469	PEX13	ADRENOLEUKODYSTROPHY, AUTOSOMAL NEONATAL FORM | PEX13	202370
470	PEX26	ADRENOLEUKODYSTROPHY, AUTOSOMAL NEONATAL FORM | PEX26	202370
471	PEX5	ADRENOLEUKODYSTROPHY, AUTOSOMAL NEONATAL FORM | PEX5	202370
472	PEX7	RHIZOMELIC CHONDRODYSPLASIA PUNCTATA TYPE 1	215100
473	PGK1	PHOSPHOGLYCERATE KINASE 1 DEFICIENCY, X-LINKED	300653
474	PHF6	BORJESON-FORSSMAN-LEHMANN SYNDROME, X-LINKED	301900
475	PHKA1	GLYCOGEN STORAGE DISEASE, TYPE Ixd, X-LINKED	300559
476	PHKA2	GLYCOGEN STORAGE DISEASE, TYPE Ixa1, X-LINKED	306000

477	PIGA	MULTIPLE CONGENITAL ANOMALIES-HYPOTONIA-SEIZURES SYNDROME 2	300868
478	PIGN	MULTIPLE CONGENITAL ANOMALIES-HYPOTONIA-SEIZURES SYNDROME 1	614080
479	PKHD1	POLYCYSTIC KIDNEY DISEASE, AUTOSOMAL RECESSIVE	263200
480	PKLR	PYRUVATE KINASE DEFICIENCY OF RED CELLS	266200
481	PLA2G6	INFANTILE NEUROAXONAL DYSTROPHY	256600
482	PLCE1	NEPHROTIC SYNDROME, TYPE 3	610725
492	POLG	ALPERS DIFFUSE DEGENERATION OF CEREBRAL GRAY MATTER WITH HEPATIC CIRRHOSIS	203700
493	POMGNT1	MUSCLE-EYE-BRAIN DISEASE	253280
494	POMT1	WALKER-WARBURG SYNDROME	236670
495	POMT2	WALKER-WARBURG SYNDROME	236670
496	POR	ANTLEY-BIXLER SYNDROME	201750
497	POU1F1	PITUITARY DWARFISM III	262600
498	POU3F4	DEAFNESS, X-LINKED 2	304400
499	PPT1	NEURONAL CEROID LIPOFUSCINOSIS 1	256730
500	PQBP1	RENPENNING SYNDROME 1	309500
501	PRF1	HEMOPHAGOCYTIC LYMPHOHISTIOCYTOSIS, FAMILIAL, 2	603553
502	PRKAG2	GLYCOGEN STORAGE DISEASE OF HEART, LETHAL CONGENITAL	261740
503	PROC	THROMBOPHILIA DUE TO PROTEIN C DEFICIENCY, AUTOSOMAL RECESSIVE	612304
504	PROP1	PITUITARY DWARFISM III	262600
505	PRPS1	ARTS SYNDROME	301835
506	PRSS12	MENTAL RETARDATION, AUTOSOMAL RECESSIVE 1	249500
507	PRX	HYPERTROPHIC NEUROPATHY OF DEJERINE-SOTTAS	145900
508	PSAP	METACHROMATIC LEUKODYSTROPHY DUE TO SAPOSIN B DEFICIENCY	249900
509	PSAT1	PHOSPHOSERINE AMINOTRANSFERASE DEFICIENCY	610992
510	PTH1R	CHONDRODYSPLASIA, BLOMSTRAND TYPE	215045
511	RAB23	CARPENTER SYNDROME	201000
512	RAB27A	GRISCELLI SYNDROME, TYPE 2	607624
513	RAB39B	MENTAL RETARDATION X-LINKED 72	300271
514	RAB3GAP1	WARBURG MICRO SYNDROME	600118
515	RAB3GAP2	MARTSOLF SYNDROME	212720
516	RAB40AL	MENTAL RETARDATION, X-LINKED, SYNDROMIC, MARTIN-PROBST TYPE	300519
517	RAG1	SEVERE COMBINED IMMUNODEFICIENCY, AUTOSOMAL RECESSIVE, T CELL-NEGATIVE,	601457
518	RAG2	OMENN SYNDROME	603554
519	RAPSN	FETAL AKINESIA DEFORMATION SEQUENCE	208150
520	RBM10	TARP SYNDROME, X-LINKED	311900
521	RELN	LISSENCEPHALY 2	257320
522	RFT1	CONGENITAL DISORDER OF GLYCOSYLATION, TYPE In	612015
523	RMRP	ANAUXETIC DYSPLASIA	607095
524	RNASEH2A	AICARDI-GOUTIERES SYNDROME 4	610333
525	RNASEH2B	AICARDI-GOUTIERES SYNDROME 2	610181
526	RNASEH2C	AICARDI-GOUTIERES SYNDROME 3	610329
527	RP2	RETINITIS PIGMENTOSA 2; RP2	300757
528	RPGR	RETINITIS PIGMENTOSA 3, X-LINKED	300029
529	RPGRIP1L	MECKEL SYNDROME, TYPE 5	611561
530	RRM2B	MITOCHONDRIAL DNA DEPLETION SYNDROME, ENCEPHALOMYOPATHIC, WITH RENAL TUBULOI	612075
531	RS1	RETINOSCHISIS 1, X-LINKED, JUVENILE	312700
532	SACS	SPASTIC ATAXIA, CHARLEVOIX-SAGUENAY TYPE	270550
533	SAMDH1	AICARDI-GOUTIERES SYNDROME 5	612952
534	SAT1	KERATOSIS FOLLICULARIS SPINULOSA DECALVANS, X-LINKED	308800
535	SBDS	SHWACHMAN-DIAMOND SYNDROME	260400
536	SC5DL	LATHOSTEROLOSIS	607330
537	SCNN1A	PSEUDOHYPOALDOSTERONISM, TYPE I, AUTOSOMAL RECESSIVE	264350
538	SCNN1B	PSEUDOHYPOALDOSTERONISM, TYPE I, AUTOSOMAL RECESSIVE	264350
539	SCNN1G	PSEUDOHYPOALDOSTERONISM, TYPE I, AUTOSOMAL RECESSIVE	264350
540	SCO1	COMPLEX IV DEFICIENCY	220110
541	SCO2	CARDIOENCEPHALOMYOPATHY, FATAL INFANTILE, DUE TO CYTOCHROME c OXIDASE	604377
542	SEPN1	RIGID SPINE MUSCULAR DYSTROPHY 1	602771
543	SFTPA1	PULMONARY FIBROSIS, IDIOPATHIC, SUSCEPTIBILITY TO	178500
544	SFTPB	RESPIRATORY DISTRESS SYNDROME IN PREMATURE INFANTS	267450
545	SFTPC	RESPIRATORY DISTRESS SYNDROME IN PREMATURE INFANTS	267450
546	SGCA	MUSCULAR DYSTROPHY, LIMB-GIRDLE, TYPE 2D	608099
547	SGSH	MUCOPOLYSACCHARIDOSIS TYPE IIIA	252900
548	SH2D1A	LYMPHOPROLIFERATIVE SYNDROME, X-LINKED, 1	308240
549	SHOX	LANGER MESOMELIC DYSPLASIA, X-LINKED	249700
550	SHROOM4	STOCCO DOS SANTOS X-LINKED MENTAL RETARDATION SYNDROME	300434
551	SIL1	MARINESCO-SJOGREN SYNDROME	248800
552	SLC12A1	BARTTER SYNDROME, ANTENATAL, TYPE 1	601678
553	SLC12A6	AGENESIS OF THE CORPUS CALLOSUM WITH PERIPHERAL NEUROPATHY	218000
554	SLC16A2	ALLAN-HERNDON-DUDLEY SYNDROME	300523

555	SLC17A5	INFANTILE SIALIC ACID STORAGE DISORDER	269920
556	SLC22A5	CARNITINE DEFICIENCY, SYSTEMIC PRIMARY	212140
557	SLC25A15	HYPERORNITHINEMIA-HYPERAMMONEMIA-HOMOCITRULLINURIA SYNDROME	238970
558	SLC25A20	CARNITINE-ACYLCARNITINE TRANSLOCASE DEFICIENCY	212138
559	SLC25A22	EPILEPTIC ENCEPHALOPATHY, EARLY INFANTILE, 3	609304
560	SLC26A2	DIASTROPHIC DYSPLASIA	222600
561	SLC26A4	PENDRED SYNDROME	274600
562	SLC34A2	PULMONARY ALVEOLAR MICROLITHIASIS	265100
563	SLC35A1	CONGENITAL DISORDER OF GLYCOSYLATION, TYPE Iif	603585
564	SLC35C1	CONGENITAL DISORDER OF GLYCOSYLATION TYPE IIc	266265
565	SLC35D1	SCHNECKENBECKEN DYSPLASIA	269250
566	SLC37A4	GLYCOGEN STORAGE DISEASE Ib	232220
567	SLC3A1	CYSTINURIA	220100
568	SLC4A11	CORNEAL DYSTROPHY AND PERCEPTIVE DEAFNESS	217400
569	SLC6A8	CREATINE DEFICIENCY SYNDROME, X-LINKED	300352
570	SLC9A6	MENTAL RETARDATION, X-LINKED ANGELMAN, SYNDROMIC, CHRISTIANSON	300243
571	SMC1A	CORNELIA DE LANGE SYNDROME 2, X-LINKED	300590
572	SMN1	SPINAL MUSCULAR ATROPHY TYPE I	253300
573	SMOC1	MICROPHTHALMIA AND LIMB ANOMALIES	206920
574	SMPD1	NIEMANN-PICK DISEASE, TYPE A	257200
575	SMS	MENTAL RETARDATION, X-LINKED, SNYDER-ROBINSON TYPE	309583
576	SNAP29	CEREBRAL DYSGENESIS, NEUROPATHY, ICHTHYOSIS, AND PALMOPLANTAR KERATODERMA	609528
577	SOX3	MENTAL RETARDATION, X-LINKED, WITH PANHYPOPITUITARISM	300123
578	SP110	HEPATIC VENOOCCLUSIVE DISEASE WITH IMMUNODEFICIENCY	235550
579	SRD5A2	PSEUDOVAGINAL PERINEOSCROTAL HYPOSPADIAS	264600
580	SRD5A3	KAHRIZI SYNDROME	611715
581	ST3GAL3	EPILEPTIC ENCEPHALOPATHY, EARLY INFANTILE	606494
582	ST3GAL5	AMISH INFANTILE EPILEPSY SYNDROME	609056
583	STAMBP	MICROCEPHALY-CAPILLARY MALFORMATION SYNDROME	614261
584	STAR	LIPOID CONGENITAL ADRENAL HYPERPLASIA	201710
585	STAT1	ATYPICAL MYCOBACTERIOSIS, FAMILIAL	209950
586	STIM1	IMMUNE DYSFUNCTION WITH T-CELL INACTIVATION DUE TO CALCIUM ENTRY DEFECT 2	612783
587	STRA6	MICROPHTHALMIA, SYNDROMIC 9 (Matthew-Wood syndrome)	601186
588	STS	ICHTHYOSIS, X-LINKED	308100
589	STX11	HEMOPHAGOCYTIC LYMPHOHISTIOCYTOSIS, FAMILIAL, 4	603552
590	STXBP2	HEMOPHAGOCYTIC LYMPHOHISTIOCYTOSIS, FAMILIAL, 5	613101
591	SUCLA2	MITOCHONDRIAL DNA DEPLETION SYNDROME, ENCEPHALOMYOPATHIC FORM	612073
592	SUCLG1	LACTIC ACIDOSIS, FATAL INFANTILE	245400
593	SUOX	SULFOCYSTEINURIA	272300
594	SURF1	LEIGH SYNDROME	256000
595	SYP	MENTAL RETARDATION X-LINKED SYP-RELATED	300802
596	TAF1	DYSTONIA-PARKINSONISM, X-LINKED	314250
597	TAZ	BARTH SYNDROME	302060
598	TBC1D20	WARBURG MICRO SYNDROME 4	615663
599	TBCE	HYPOPARATHYROIDISM-RETARDATION-DYSMORPHISM SYNDROME	241410
600	TBX22	CLEFT PALATE WITH ANKYLOGLOSSIA, X-LINKED	303400
601	TCIRG1	OSTEOPETROSIS, AUTOSOMAL RECESSIVE 1	259700
602	TCTN2	MECKEL SYNDROME, TYPE 8	613885
603	TFR2	HEMOCHROMATOSIS, TYPE 3	604250
604	TGM1	ICHTHYOSIS, LAMELLAR, 1	242300
605	TH	SEGAWA SYNDROME, AUTOSOMAL RECESSIVE	605407
606	TIMM8A	OPTICOACOUSTIC NERVE ATROPHY WITH DEMENTIA	311150
607	TK2	MITOCHONDRIAL DNA DEPLETION SYNDROME, MYOPATHIC FORM	609560
608	TLR3	HERPES SIMPLEX ENCEPHALITIS, SUSCEPTIBILITY TO, 2	613002
609	TMEM67	JOUBERT SYNDROME 6	610688
610	TNFRSF11B	PAGET DISEASE, JUVENILE	239000
611	TNNT1	NEMALINE MYOPATHY 5	605355
612	TPP1	NEURONAL CEROID LIPOFUSCINOSIS 2	204500
613	TRAPPC2	SPONDYLOEPIPHYSEAL DYSPLASIA TARDA	313400
614	TRAPPC9	MENTAL RETARDATION AUTOSOMAL RECESSIVE 13	613192
615	TREX1	AICARDI-GOUTIERES SYNDROME 1	225750
616	TRIM37	MULIBREY NANISM	253250
617	TSEN54	PONTOCEREBELLAR HYPOPLASIA TYPE 4	225753
618	TSFM	COMBINED OXIDATIVE PHOSPHORYLATION DEFICIENCY 3	610505
619	TSHB	HYPOTHYROIDISM, CONGENITAL, NONGOITROUS, 4	275100
620	TSPAN7	MENTAL RETARDATION, X-LINKED 58	300210
621	TSPYL1	SUDDEN INFANT DEATH WITH DYSGENESIS OF THE TESTES SYNDROME	608800
622	TTC21B	NEPHRONOPHTHISIS 12; SHORT-RIB THORACIC DYSPLASIA 4 WITH OR WITHOUT POLYDACTYLY	613820
623	TTN	CARDIOMYOPATHY, DILATED, 1G	604145

624	TTPA	VITAMIN E, FAMILIAL ISOLATED DEFICIENCY OF	277460
625	TUBA1a	LISSENCEPHALY 3	611603
626	TUFM	COMBINED OXIDATIVE PHOSPHORYLATION DEFICIENCY 4	610678
627	TUSC3	MENTAL RETARDATION AUTOSOMAL RECESSIVE 7	611093
628	TYK2	ATYPICAL MYCOBACTERIOSIS, FAMILIAL	209950
629	TYMP	MITOCHONDRIAL NEUROGASTROINTESTINAL ENCEPHALOPATHY SYNDROME	603041
630	TYR	ALBINISM, OCULOCUTANEOUS, TYPE IA	203100
631	UBA1	SPINAL MUSCULAR ATROPHY, X-LINKED 2	301830
632	UBE2A	MENTAL RETARDATION X-LINKED SYNDROMIC UBE2A-RELATED	312180
633	UBR1	JOHANSON-BLIZZARD SYNDROME	243800
634	UNC13D	HEMOPHAGOCYTIC LYMPHOHISTIOCYTOSIS, FAMILIAL, 3	608898
635	UNC93B1	HERPES SIMPLEX ENCEPHALITIS, SUSCEPTIBILITY TO, 1	610551
636	UPF3B	MENTAL RETARDATION, X-LINKED, SYNDROMIC 14	300676
637	UQCRB	MITOCHONDRIAL COMPLEX III DEFICIENCY	124000
638	UQCRQ	MITOCHONDRIAL COMPLEX III DEFICIENCY	124000
639	UROS	PORPHYRIA, CONGENITAL ERYTHROPOIETIC	263700
640	USH1C	USHER SYNDROME, TYPE IC	276904
641	USH1G	USHER SYNDROME, TYPE IG	606943
642	USH2A	USHER SYNDROME, TYPE IIA	276901
643	VAX1	MICROPHTHALMIA, SYNDROMIC 11	614402
644	VDR	VITAMIN D-DEPENDENT OSTEOPENIA, TYPE II	277440
645	VIPAR	ARTHROGRYPOSIS, RENAL DYSFUNCTION, AND CHOLESTASIS 2	613404
646	VLDLR	CEREBELLAR HYPOPLASIA AND MENTAL RETARDATION WITH OR WITHOUT QUADRUPEDAL	224050
647	VMA21	MYOPATHY, X-LINKED, WITH EXCESSIVE AUTOPHAGY	300913
648	VPS13B	COHEN SYNDROME	216550
649	VPS33B	ARTHROGRYPOSIS, RENAL DYSFUNCTION, AND CHOLESTASIS	208085
650	WAS	WISKOTT-ALDRICH SYNDROME	301000
651	WDR19	SENIOR-LOKEN SYNDROME 8; NEPHRONOPHTHISIS 13	616307
652	WDR34	SHORT-RIB THORACIC DYSPLASIA 11 WITH OR WITHOUT POLYDACTYLY	615633
653	WDR35	CRANIOECTODERMAL DYSPLASIA 2; SHORT-RIB THORACIC DYSPLASIA 7 WITH OR WITHOUT POL	613610
654	WDR60	SHORT-RIB THORACIC DYSPLASIA 8 WITH OR WITHOUT POLYDACTYLY	615503
655	WNT10A	ODONTOONYCHODERMAL DYSPLASIA	257980
656	WNT3	TETRA-AMELIA, AUTOSOMAL RECESSIVE	273395
657	WNT7A	SPONDYLOCOSTAL DYSOSTOSIS, AUTOSOMAL RECESSIVE 1	277300
658	XIAP	LYMPHOPROLIFERATIVE SYNDROME, X-LINKED, 2	300635
659	XPA	XERODERMA PIGMENTOSUM, COMPLEMENTATION GROUP A	278700
660	XPC	XERODERMA PIGMENTOSUM, COMPLEMENTATION GROUP C	278720
661	ZC4H2	WIEACKER-WOLFF SYNDROME, X-LINKED	314580
662	ZDHHC15	MENTAL RETARDATION, X-LINKED 91	300577
663	ZDHHC9	MENTAL RETARDATION X-LINKED SYNDROMIC ZDHHC9-RELATED	300799
664	ZIC3	HETEROTAXY, VISCERAL, 1, X-LINKED	306955
665	ZMPSTE24	MANDIBULOACRAL DYSPLASIA WITH TYPE B LIPODYSTROPHY	608612
666	ZNF41	MENTAL RETARDATION X-LINKED 89	314995
667	ZNF469	BRITTLE CORNEA SYNDROME (Ehlers-Danlos syndrome type VIB)	229200
668	ZNF674	MENTAL RETARDATION X-LINKED 92	300573
669	ZNF711	MENTAL RETARDATION X-LINKED ZNF711-RELATED	300803
670	ZNF81	MENTAL RETARDATION, X-LINKED 45	300498

NOTE: The applied methodology may not necessarily detect all the possible pathogenic mutations of each gene/disease. Generally, the analysis covers a large percentage (~95%) of mutations in exons-coding regions of the genes, while it does not generally detect mutations in non-coding regions/introns and exonic deletions or duplications >15-20 base-pairs.