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Strand® Rare Disease Test

Indicative list of genes corresponding to rare diseases		
Sr. No.	**Disease/Broader term**	**Gene name**
1	17, 20-LYASE DEFICIENCY, ISOLATED	*CYP17A1*
2	17-ALPHA-HYDROXYLASE/17, 20-LYASE DEFICIENCY	*CYP17A1*
3	17-BETA HYDROXYSTEROID DEHYDROGENASE III DEFICIENCY	*HSD17B3*
4	3-HYDROXYACYL-COA DEHYDROGENASE DEFICIENCY	*HADH**
5	3-METHYLGLUTACONIC ACIDURIA	*AUH*, DNAJC19*
6	5-FLUOROURACIL TOXICITY	*DPYD*
7	ABCD SYNDROME	*EDNRB**
8	ACHALASIA-ADDISONIANISM-ALACRIMA SYNDROME	*AAAS*
9	ACHONDROGENESIS, TYPE IB	*SLC26A2*
10	ACYL-COA DEHYDROGENASE DEFICIENCY	*ACADL, ACADM, ACADVL**
11	ADRENAL HYPERPLASIA, CONGENITAL, DUE TO 21-HYDROXYLASE DEFICIENCY	*CYP21A2*
12	ADRENOLEUKODYSTROPHY	*ABCD1*, PEX1, PEX13*, PEX26*, PEX5*
13	AGAMMAGLOBULINEMIA	*BTK*
14	AGENESIS OF THE CORPUS CALLOSUM WITH PERIPHERAL NEUROPATHY	*SLC12A6*
15	AICARDI-GOUTIÈRES SYNDROME	*RNASEH2B, RNASEH2C, RNASEH2A, SAMHD1*
16	ALLAN-HERNDON-DUDLEY SYNDROME	*SLC16A2**
17	ALPERS DIFFUSE DEGENERATION OF CEREBRAL GRAY MATTER WITH HEPATIC CIRRHOSIS	*POLG*
18	ALPHA THALASSEMIA	*ATRX, HBA1*
19	ALPHA-METHYLACETOACETIC ACIDURIA	*ACAT1*
20	ALPHA-METHYLACYL-COA RACEMASE DEFICIENCY	*AMACR**
21	ALPORT SYNDROME	*COL4A4, COL4A3, COL4A5*
22	ALSTROM SYNDROME	*ALMS1*
23	AMEGAKARYOCYTIC THROMBOCYTOPENIA, CONGENITAL	*MPL*
24	AMYLOIDOSIS, HEREDITARY RENAL	*FGA*
25	ANAUXETIC DYSPLASIA	*RMRP*
26	ANDROGEN INSENSITIVITY; PARTIAL, WITH OR WITHOUT BREAST CANCER	*AR*
27	ANGELMAN SYNDROME	*UBE3A*
28	ANGELMAN SYNDROME-LIKE	*CDKL5*
29	ANTIBODY DEFICIENCY DUE TO ICOS DEFECT	*ICOS*
30	ANTLEY-BIXLER SYNDROME; DISORDERED STEROIDOGENESIS	*POR**
31	ARGININOSUCCINIC ACIDURIA	*ASL*
32	AROMATIC L-AMINO ACID DECARBOXYLASE DEFICIENCY	*DDC*
33	ARTERIAL CALCIFICATION, GENERALIZED, OF INFANCY	*ENPP1*
34	ARTHROGRYPOSIS	*GLE1, VPS33B*, VIPAR, VIPAS39*
35	ARTS SYNDROME	*PRPS1*
36	ATAXIA	*APTX, ATM*
37	ATELOSTEOGENESIS, TYPE II	*SLC26A2*
38	ATYPICAL MYCOBACTERIOSIS, FAMILIAL	*IFNGR1, IL12B, IL12RB1*, IKBKG, STAT1*
39	AUTOIMMUNE LYMPHOPROLIFERATIVE SYNDROME	*CASP10*, FAS*

*Genes having 85%-95% coverage. Note: Gene coverage varies from sample to sample in the NGS runs.

40	BANNAYAN-RILEY-RUVALCABA SYNDROME	*PTEN*
41	BARTTER SYNDROME	*KCNJ1, SLC12A1**
42	BCG AND SALMONELLA INFECTION, DISSEMINATED	*IFNGR1, IL12B*
43	BECKWITH-WIEDEMANN SYNDROME	*NSD1**
44	BETHLEM MYOPATHY	*COL6A1, COL6A2*, COL6A3*
45	BILE ACID SYNTHESIS DEFECT, CONGENITAL, 4	*AMACR**
46	BJORNSTAD SYNDROME	*BCS1L*
47	BLOOM SYNDROME	*BLM*
48	BLUE BABY SYNDROME	*HBB*
49	BRANCHED-CHAIN KETO ACID DEHYDROGENASE E1, BETA POLYPEPTIDE	*BCKDHB*
50	CANAVAN DISEASE	*ASPA*
51	CANDIDIASIS, FAMILIAL, 7	*STAT1*
52	CARBAMOYL PHOSPHATE SYNTHETASE I DEFICIENCY, HYPERAMMONEMIA DUE TO	*CPS1*
53	CARDIOENCEPHALOMYOPATHY	*COX15, SCO2**
54	CARDIOMYOPATHY	*FKTN**
55	CARNITINE DEFICIENCY, SYSTEMIC PRIMARY	*SLC22A5*
56	CARNITINE PALMITOYL TRANSFERASE DEFICIENCY	*CPT2, CPT1A*
57	CARNITINE-ACYLCARNITINE TRANSLOCASE DEFICIENCY	*SLC25A20**
58	CARPENTER SYNDROME	*RAB23*
59	CARTILAGE-HAIR HYPOPLASIA	*RMRP*
60	CEREBELLAR HYPOPLASIA AND MENTAL RETARDATION WITH OR WITHOUT QUADRUPEDAL LOCOMMOTION	*VLDLR*
61	CEREBRAL DYSGENESIS, NEUROPATHY, ICHTHYOSIS, AND PALMOPLANTAR KERATODERMA	*SNAP29*
62	CEREBROTENDINOUS XANTHOMATOSIS	*CYP27A1*
63	CHARCOT-MARIE-TOOTH DISEASE	*EGR2*, GDAP1, FGD4*, MPZ, PMP22*
64	CHEDIAK HIGASHI SYNDROME	*LYST*
65	CHILBLAIN LUPUS	*SAMHD1*
66	CHOLESTASIS	*ABCB4, ATP8B1, ABCB11*
67	CHONDRODYSPLASIA	*ARSE*, PTH1R*
68	CITRULLINEMIA, CLASSIC	*ASS1*
69	COCKAYNE SYNDROME	*ERCC8*
70	COENZYME Q10 DEFICIENCY	*ADCK3*, APTX, COQ2, COQ9, PDSS1, PDSS2*,*
71	COFFIN-LOWRY SYNDROME	*RPS6KA3*
72	COHEN SYNDROME	*VPS13B*
73	COLE DISEASE	*ENPP1*
74	COMBINED CELLULAR AND HUMORAL IMMUNE DEFECTS WITH GRANULOMAS	*RAG1,RAG2*
75	COMBINED OXIDATIVE PHOSPHORYLATION DEFICIENCY	*GFM1, MRPS22*, TUFM,*
76	COMBINED SAPOSIN DEFICIENCY	*PSAP*
77	COMPLEX I DEFICIENCY	*NDUFA1, NDUFA7, NDUFAF2, NDUFAF4, NDUFS3*, NDUFS5, NDUFV1*
78	COMPLEX IV DEFICIENCY	*COX10, COX15, COX6B1, FASTKD2, SCO1, SCO2**
79	CONGENITAL ADRENAL HYPERPLASIA	*CYP17A1*
80	CONGENITAL ADRENAL HYPOPLASIA	*NR0B1*
81	CONGENITAL AFIBRINOGENEMIA	*FGA*
82	CONGENITAL BILATERAL ABSENCE OF VAS DEFERENS	*CFTR*

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83	CONGENITAL DISORDER OF GLYCOSYLATION	*ALG1, ALG2, ALG3, ALG8*, ALG6, ALG12, B4GALT1, COG1, COG7, COG8*, DPM1, DOLK, DPAGT1*, MPI*, MGAT2, MOGS*, PMM2*, MPDU1*, SLC35A1, RFT1, SLC35C1**
84	CONGENITAL HEART DEFECTS, NONSYNDROMIC, 1, X-LINKED	*ZIC3**
85	CORNEAL DYSTROPHY AND PERCEPTIVE DEAFNESS	*SLC4A11*
86	CORPUS CALLOSUM, PARTIAL AGENESIS OF, X-LINKED	*L1CAM**
87	COWDEN SYNDROME	*PTEN*
88	CRANIOFRONTONASAL SYNDROME	*EFNB1**
89	CRASH SYNDROME	*L1CAM**
90	CREATINE DEFICIENCY SYNDROME, X-LINKED	*SLC6A8*
91	CRISPONI SYNDROME	*CRLF1**
92	CUTIS LAXA	*ATP6V0A2, EFEMP2*
93	CYSTIC FIBROSIS	*CFTR*
94	CYSTINOSIS	*CTNS**
95	D-BIFUNCTIONAL PROTEIN DEFICIENCY	*HSD17B4**
96	DE LA CHAPELLE DYSPLASIA	*SLC26A2*
97	DE SANCTIS-CACCHIONE SYNDROME	*XPA*
98	DEAFNESS	*GJB2, MYO7A, USH1C*
99	DEFICIENCY OF ACYL-COA DEHYDROGENASE FAMILY MEMBER 9	*ACAD9**
100	DEFICIENCY OF INTERLEUKIN-1-RECEPTOR ANTAGONIST	*IL1RN**
101	DENT DISEASE 1	*CLCN5*
102	DESMOSTEROLOSIS	*DHCR24**
103	DIABETES MELLITUS	*ABCC8, INSR*
104	DIASTROPHIC DYSPLASIA	*SLC26A2*
105	DIHYDROLIPOAMIDE DEHYDROGENASE DEFICIENCY	*DLD*
106	DIHYDROPYRIMIDINE DEHYDROGENASE DEFICIENCY	*DPYD*
107	DISORDERED STEROIDOGENESIS DUE TO CYTOCHROME P450 OXIDOREDUCTASE	*POR**
108	DONNAI-BARROW SYNDROME	*LRP2*
109	DONOHUE SYNDROME	*INSR*
110	DURSUN SYNDROME	*G6PC3*
111	DYSKERATOSIS CONGENITA	*DKC1*
112	DYSSEGMENTAL DYSPLASIA, SILVERMAN-HANDMAKER TYPE	*HSPG2*
113	ECTODERMAL DYSPLASIA SYNDROME	*EDA, IKBKG*
114	EHLERS-DANLOS SYNDROME	*COL1A2**
115	EIKEN SYNDROME	*PTH1R*
116	ELEJALDE DISEASE	*MYO5A*
117	ELLIS-VAN CREVELD SYNDROME	*EVC2, EVC**
118	ENCEPHALOPATHY	*ETHE1, COX10*
119	EPIDERMOLYSIS BULLOSA	*COL7A1, COL17A1, ITGA6, ITGB4, LAMB3, LAMC2, LAMA3, PLEC**
120	EPILEPSY	*PCDH19, ALDH7A1*, C25A22, CDKL5*
121	EPIPHYSEAL DYSPLASIA	*SLC26A2, EIF2AK3*
122	ESCOBAR SYNDROME	*CHRNG*
123	FABRY DISEASE	*GLA*
124	FAILURE OF TOOTH ERUPTION, PRIMARY	*PTH1R*
125	FAMILIAL HEMOPHAGOCYTIC LYMPHOHISTIOCYTOSIS	*UNC13D, STX11, STXBP2*
126	FAMILIAL MEDITERRANEAN FEVER	*MEFV**
127	FANCONI ANEMIA TYPE C	*FANCC*

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128	FATTY LIVER, ACUTE, OF PREGNANCY	*HADHA*
129	FEBRILE SEIZURES, FAMILIAL, 4	*GPR98*
130	FERTILE EUNUCH SYNDROME	*GNRHR*
131	FETAL AKINESIA DEFORMATION SEQUENCE	*RAPSN*
132	FIBROMATOSIS, JUVENILE HYALINE	*ANTXR2**
133	FIBULAR APLASIA OR HYPOPLASIA, FEMORAL BOWING	*WNT7A**
134	FRASER SYNDROME	*FRAS1, FREM2*
135	FRUCTOSE INTOLERANCE, HEREDITARY	*ALDOB*
136	FUCHS ENDOTHELIAL, 4; TYPE 2, AUTOSOMAL RECESSIVE	*SLC4A11*
137	FUCOSIDOSIS	*FUCA1*
138	FUKUYAMA CONGENITAL MUSCULAR DYSTROPHY	*FKTN**
139	FUMARASE DEFICIENCY	*FH*
140	GALACTOKINASE DEFICIENCY	*GALK1*
141	GALACTOSEMIA	*GALK1*
142	GALLBLADDER DISEASE 1	*ABCB4*
143	GAUCHER DISEASE	*GBA, PSAP*
144	GELEOPHYSIC DYSPLASIA	*ADAMTSL2*
145	GLUTARIC ACIDEMIA I	*GCDH**
146	GLUTATHIONE SYNTHETASE DEFICIENCY	*GSS*
147	GLYCINE ENCEPHALOPATHY	*AMT, GLDC**
148	GLYCOGEN STORAGE DISEASE	*AGL, GAA, GBE1, G6PC, PYGM*, SLC37A4,*
149	GM1 GANGLIOSIDOSIS	*GLB1**
150	GRISCELLI DISEASE	*BCS1L, MYO5A, RAB27A*
151	HARP SYNDROME	*PANK2*
152	HAWKINSINURIA	*HPD*
153	HEINZ BODY ANEMIAS, ALPHA-	*HBA1, HBB*
154	HEMATURIA, BENIGN FAMILIAL	*COL4A3*
155	HEMOCHROMATOSIS TYPE 2	*HFE2; HAMP*
156	HEMOGLOBIN H DISEASE, NONDELETIONAL	*HBA1*
157	HEMOPHILIA	*F8*, F9*
158	HEPATIC FAILURE, EARLY ONSET, AND NEUROLOGIC DISORDER	*SCO1*
159	HEPATIC VENOOCCLUSIVE DISEASE WITH IMMUNODEFICIENCY	*SP110*
160	HEREDITARY PERSISTENCE OF FETAL HEMOGLOBIN	*HBB*
161	HERMANSKY PUDLAK SYNDROME	*AP3B1, PLDN*
162	HERPETIC ENCEPHALITIS	*UNC93B1*, TLR3*
163	HETEROTAXY, VISCERAL, 1, X-LINKED	*ZIC3**
164	HOLOCARBOXYLASE SYNTHETASE DEFICIENCY	*HLCS**
165	HOMOCYSTINURIA	*CBS**
166	HOYERAAL-HREIDARSSON SYNDROME	*DKC1*
167	HURLER SYNDROME	*IDUA**
168	HYALINOSIS, INFANTILE SYSTEMIC	*ANTXR2**
169	HYDROCEPHALUS DUE TO CONGENITAL STENOSIS OF AQUEDUCT OF SYLVIUS;	*L1CAM**
170	HYDROPS-ECTOPIC CALCIFICATION-MOTH-EATEN SKELETAL DYSPLASIA	*LBR*
171	TRIFUNCTIONAL PROTEIN DEFICIENCY	*HADHA, HADHB*
172	HYPERANDROGENISM, NONCLASSIC TYPE, DUE TO 21-HYDROXYLASE DEFICIENCY	*CYP21A2*
173	HYPER-IGE RECURRENT INFECTION SYNDROME, AUTOSOMAL RECESSIVE	*DOCK8**
174	HYPERINSULINEMIC HYPOGLYCEMIA	*ABCC8, INSR, HADH**
175	HYPERORNITHINEMIA-HYPERAMMONEMIA-HOMOCITRULLINURIA SYNDROME	*SLC25A15*
176	HYPERTROPHIC NEUROPATHY OF DEJERINE-SOTTAS	*EGR2*, MPZ, PMP22*
177	HYPOCALCEMIC VITAMIN D-DEPENDENT RICKETS	*CYP27B1, VDR**

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178	HYPOGLYCEMIA OF INFANCY, LEUCINE-SENSITIVE	*ABCC8*
179	HYPOGONADOTROPIC HYPOGONADISM	*GNRHR*
180	HYPOPARATHYROIDISM-RETARDATION-DYSMORPHISM SYNDROME	*TBCE**
181	HYPOPHOSPHATASIA	*ALPL**
182	HYPOPHOSPHATEMIC OSTEOPENIA, AUTOSOMAL RECESSIVE	*DMP1*
183	HYPOPHOSPHATEMIC RICKETS	*ENPP1, CLCN5*
184	HYPOSPADIAS 1, X-LINKED	*AR*
185	HYPOTHYROIDISM, CONGENITAL, NONGOITROUS, 4	*TSHB*
186	ICHTHYOSIS	*ABCA12, CLDN1,*
187	IMMUNE DYSFUNCTION	*ORAI1, STIM1**
188	IMMUNODEFICIENCY	*CD3E, CD3G, CD247, CD40LG, CD19, CD3D*, DNMT3B, FOXN1*
189	INCONTINENTIA PIGMENTI, TYPE II	*IKBKG*
190	INFANTILE NEUROAXONAL DYSTROPHY	*2G6**
191	INFANTILE SIALIC ACID STORAGE DISORDER; SIALURIA, FINNISH TYPE	*SLC17A5*
192	INFERTILE MALE SYNDROME	*AR*
193	INSENSITIVITY TO PAIN, CONGENITAL, WITH ANHIDROSIS	*NTRK1*
194	INVASIVE PNEUMOCOCCAL DISEASE, RECURRENT ISOLATED, 2	*IKBKG*
195	JEUNE SYNDROME	*IFT80, DYNC2H1*
196	JOHANSON-BLIZZARD SYNDROME	*UBR1*
197	JOUBERT SYNDROME	*AHI1, CEP290, NPHP1*, TMEM67*
198	JUVENILE AMYOTROPHIC LATERAL SCLEROSIS 2	*ALS2*
199	KENNY-CAFFEY SYNDROME-1	*TBCE**
200	KERATOSIS FOLLICULARIS SPINULOSA DECALVANS, X-LINKED	*MBTPS2*
201	KRABBE DISEASE	*GALC*
202	LACTIC ACIDOSIS, FATAL INFANTILE (MTDNA DEPLETION)	*SUCLG1*
203	LATHOSTEROLOSIS	*SC5DL, SC5D*
204	LEIGH SYNDROME	*BCS1L, LRPPRC, PDHA1*, SURF1*
205	LEPRECHAUNISM	*INSR*
206	LESCH-NYHAN SYNDROME	*HPRT1*
207	LETHAL CONGENITAL CONTRACTURE SYNDROME	*ERBB3, GLE1*
208	LEUKOCYTE ADHESION DEFICIENCY TYPE III	*FERMT3*
209	LEUKODYSTROPHY	*FAM126A**
210	LHERMITTE-DUCLOS SYNDROME	*PTEN*
211	LIPOID CONGENITAL ADRENAL HYPERPLASIA	*CYP11A1, STAR*
212	LISSENCEPHALY	*DCX, RELN, TUBA1A*
213	LOWE OCULOCEREBRORENAL SYNDROME	*OCRL*
214	LUJAN-FRYNS SYNDROME	*MED12*
215	LYMPHOPROLIFERATIVE SYNDROME	*SH2D1A, XIAP**
216	MACROCEPHALY/AUTISM SYNDROME	*PTEN*
217	MANDIBULOACRAL DYSPLASIA	*ZMPSTE24*
218	MANNOSIDOSIS, ALPHA B, LYSOSOMAL	*MAN2B1*
219	MAPLE SYRUP URINE DISEASE	*BCKDHB*
220	MARINESCO-SJOGREN SYNDROME	*SIL1*
221	MARTSOLF SYNDROME	*RAB3GAP2*
222	MASA SYNDROME	*L1CAM**
223	MECKEL SYNDROME	*MKS1*, RPGRIP1L*
224	MEGALENCEPHALIC LEUKOENCEPHALOPATHY WITH SUBCORTICAL CYSTS	*MLC1**
225	MENKES DISEASE	*ATP7A*

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226	MENTAL RETARDATION	*ATRX, AFF2, AGTR2, ACSL4, ARHGEF6, BRWD3, CASK, CUL4B, DLG3*, GRIK2, HUWE1*, HSD17B10, IL1RAPL1, PRSS12, PAK3, FTSJ1, SLC9A6, UPF3B, OPHN1, RPS6KA3, NXF5, ST3GAL3*, NSUN2, SRD5A3, DOCK8*, UBE2A, RAB39BZNF41, TUSC3, TRAPPC9, ZNF711*, ZNF674, ZDHHC9*
227	METACHROMATIC LEUKODYSTROPHY	*ARSA*, PSAP*
228	METAPHYSEAL DYSPLASIA WITHOUT HYPOTRICHOSIS	*RMRP*
229	METHYLMALONIC ACIDURIA	*MUT, MMAA*
230	MEVALONIC ACIDURIA	*MVK**
231	MICROPHTHALMIA	*BCOR*,STRA6*
232	MITOCHONDRIAL COMPLEX III DEFICIENCY	*BCS1L, UQCRQ*
233	MITOCHONDRIAL DNA DEPLETION SYNDROME	*DGUOK*, POLG, RRM2B*, SUCLA2*
234	MITOCHONDRIAL NEUROGASTROINTESTINAL ENCEPHALOPATHY SYNDROME	*TYMP*
235	MITOCHONDRIAL RECESSIVE ATAXIA SYNDROME	*POLG*
236	MOHR-TRANEBJAERG SYNDROME	*TIMM8A*
237	MOLYBDENUM COFACTOR DEFICIENCY	*MOCS2, MOCS1*
238	MOWAT-WILSON SYNDROME	*ZEB2*
239	MSUD TYPE 2	*DBT*
240	MUCOLIPIDOSIS	*GNPTAB, MCOLN1*
241	MUCOPOLYSACCHARIDOSIS	*ARSB, GUSB, GLB1*, IDS**
242	MULIBREY NANISM	*TRIM37*
243	MULTIPLE ACYL-COA DEHYDROGENASE DEFICIENCY	*ETFDH*
244	MULTIPLE PTERYGIUM SYNDROME	*CHRNA1; CHRND; CHRNG*
245	MUSCLE-EYE-BRAIN DISEASE	*POMGNT1*
246	MUSCULAR DYSTROPHY	*COL6A1, COL6A2*, COL6A3, DMD, FKTN*, LAMA2, LARGE, LMN,*
247	MYASTHENIA GRAVIS, NEONATAL TRANSIENT	*CHRNA1; CHRND; CHRNG*
248	MYASTHENIC SYNDROME, CONGENITAL, ASSOCIATED WITH ACETYLCHOLINE RECEPTOR DEFICIENCY	*RAPSN*
249	MYD88 DEFICIENCY	*MYD88*
250	MYOPATHY	*NEB, MTM1, SEPN1*, STIM1**
251	MYOSCLEROSIS, CONGENITAL	*COL6A2**
252	N-ACETYLGLUTAMATE SYNTHASE DEFICIENCY	*NAGS**
253	NANCE-HORAN SYNDROME	*NHS**
254	NEPHROLITHIASIS, TYPE I	*CLCN5*
255	NEPHRONOPHTHISIS	*INVS, NPHP4, NPHP1**
256	NEPHROSIS	*NPHS1, NPHS2, PLCE1*
257	NEURAMINIDASE DEFICIENCY	*NEU1**
258	NEURODEGENERATION	*FOLR1, PANK2, PLA2G6**
259	NEURONAL CEROID LIPOFUSCINOSIS	*CLN3, CLN5, CTSD, CLN8, MFSD8, PPT1*, TPP1*
260	NEUROPATHY, CONGENITAL HYPOMYELINATING	*EGR2*, MPZ, IKBKAP*
261	NEUTROPENIA	*G6PC3, HAX1*
262	NEVO SYNDROME	*PLOD1**
263	NIEMANN-PICK DISEASE	*NPC1*, SMPD1**
264	NIJMEGEN BREAKAGE SYNDROME	*NBN*
265	NORRIE DISEASE	*NDP**

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266	OCCIPITAL HORN SYNDROME	*ATP7A*
267	ODONTOHYPOPHOSPHATASIA	*ALPL**
268	ODONTOONYCHODERMAL DYSPLASIA	*WNT10A**
269	OMENN SYNDROME	*DCLRE1C*, RAG1, RAG2*
270	OPITZ GBBB SYNDROME, X-LINKED	*MID1*
271	OPTICOACOUSTIC NERVE ATROPHY WITH DEMENTIA	*TIMM8A*
272	ORNITHINE TRANSCARBAMYLASE DEFICIENCY, HYPERAMMONEMIA DUE TO	*OTC*
273	OSSIFICATION OF POSTERIOR LONGITUDINAL LIGAMENT OF SPINE	*ENPP1*
274	OSTEOGENESIS IMPERFECTA	*CRTAP, COL1A2**
275	OSTEOPETROSIS	*CA2*, CLCN7, TCIRG1, OSTM1*
276	PAGET DISEASE, JUVENILE	*TNFRSF11B**
277	PARKINSON DISEASE 14	*PLA2G6**
278	PELGER-HUET ANOMALY	*LBR*
279	PELIZAEUS-MERZBACHER DISEASE; SPASTIC PARAPLEGIA 2, X-LINKED	*PLP1*
280	PERMANENT NEONATAL; TRANSIENT NEONATAL 2	*ABCC8*
281	PEROXISOMAL ACYL-COA OXIDASE DEFICIENCY	*ACOX1*
282	PEROXISOME BIOGENESIS DISORDER	*PEX5, PEX26*, PEX7*, PEX13**
283	PHENYLKETONURIA	*PAH*
284	PIERSON SYNDROME	*LAMB2*
285	PITUITARY DWARFISM	*HESX1, POU1F1*
286	PLASMINOGEN DEFICIENCY TYPE I	*PLG*
287	POLYCYSTIC KIDNEY DISEASE, AUTOSOMAL RECESSIVE	*PKHD1*
288	POLYGLUCOSAN BODY DISEASE, ADULT FORM	*GBE1*
289	PORPHYRIA, CONGENITAL ERYTHROPOIETIC	*UROS**
290	PRECOCIOUS PUBERTY, MALE-LIMITED	*LHCGR*
291	PRIMARY LATERAL SCLEROSIS, JUVENILE	*ALS2*
292	PROGRESSIVE EXTERNAL OPHTHALMOPLEGIA	*POLG*
293	PROGRESSIVE MYOCLONIC EPILEPSY	*CSTB, NHLRC1*
294	PROPIONIC ACIDEMIA	*PCCA**
295	PROTEINURIA, LOW MOLECULAR WEIGHT, WITH HYPERCALCIURIC NEPHROCALCINOSIS	*CLCN5*
296	PSEUDOHYPOALDOSTERONISM, TYPE I, AUTOSOMAL RECESSIVE	*SCNN1G*
297	PYRIDOXAMINE 5-PRIME-PHOSPHATE OXIDASE DEFICIENCY	*PNPO*
298	PYRUVATE CARBOXYLASE DEFICIENCY	*PC**
299	PYRUVATE DEHYDROGENASE E3-BINDING PROTEIN DEFICIENCY	*PDHX*
300	PYRUVATE DEHYDROGENASE PHOSPHATASE DEFICIENCY	*PDP1**
301	PYRUVATE KINASE DEFICIENCY OF RED CELLS	*PKLR*
302	RABSON-MENDENHALL SYNDROME	*INSR*
303	RAINE SYNDROME	*FAM20C**
304	RENAL-HEPATIC-PANCREATIC DYSPLASIA	*NPHP3*
305	RESPIRATORY DISTRESS SYNDROME IN PREMATURE INFANTS	*SFTPC, SFTPB*
306	RETINITIS PIGMENTOSA	*CLRN1, USH2A*
307	REYNOLDS SYNDROME	*LBR*
308	RHIZOMELIC CHONDRODYSPLASIA PUNCTATA	*PEX7, AGPS*
309	RIGID SPINE MUSCULAR DYSTROPHY 1	*SEPN1**
310	ROBERTS SYNDROME	*ESCO2*
311	ROUSSY-LEVY SYNDROME	*MPZ, PMP22, MPZ*
312	SALLA DISEASE	*SLC17A5*
313	SANDHOFF DISEASE	*HEXB*
314	SC PHOCOMELIA SYNDROME	*ESCO2*
315	SCHNECKENBECKEN DYSPLASIA	*SLC35D1**
316	SCHWARTZ-JAMPEL SYNDROME, TYPE 1	*HSPG2*

*Genes having 85%-95% coverage. Note: Gene coverage varies from sample to sample in the NGS runs.

317	SECKEL SYNDROME 1	*ATR*
318	SEGAWA SYNDROME, AUTOSOMAL RECESSIVE	*TH*
319	SENIOR-LOKEN SYNDROME	*IQCB1, NPHP1**
320	SEVERE COMBINED IMMUNODEFICIENCY	*ADA, IL2RG*, JAK3, LIG4, NHEJ1*, RAG1, RAG2,*
321	SHWACHMAN-DIAMOND SYNDROME	*SBDS*
322	SIMPSON-GOLABI-BEHMEL SYNDROME	*GPC3*, OFD1*
323	SJOGREN-LARSSON SYNDROME	*ALDH3A2*
324	SOTOS SYNDROME	*NSD1**
325	SPASTIC ATAXIA, CHARLEVOIX-SAGUENAY TYPE	*SACS*
326	SPINAL MUSCULAR ATROPHY	*AR*, ATP7A, PLEKHG5*, SMN1*, UBA1**
327	SPINOCEREBELLAR ATAXIA, AUTOSOMAL RECESSIVE 7	*TPP1*
328	SPONDYLOCOSTAL DYSOSTOSIS, AUTOSOMAL RECESSIVE 1	*DLL3**
329	STOCCO DOS SANTOS X-LINKED MENTAL RETARDATION SYNDROME	*SHROOM4**
330	STRIATONIGRAL DEGENERATION, INFANTILE	*NUP62*
331	STUVE-WIEDEMANN SYNDROME	*LIFR*
332	SUCCINIC SEMIALDEHYDE DEHYDROGENASE DEFICIENCY	*ALDH5A1*
333	SUCCINYL-COA:3-KETOACID COA TRANSFERASE DEFICIENCY	*OXCT1*
334	SUDDEN INFANT DEATH WITH DYSGENESIS OF THE TESTES SYNDROME	*TSPYL1*
335	SULFOCYSTEINURIA	*SUOX*
336	SURFACTANT METABOLISM DYSFUNCTION	*ABCA3, SFTPB*
337	TAY-SACHS DISEASE	*HEXA*
338	TETRA-AMELIA, AUTOSOMAL RECESSIVE	*WNT3*
339	THALASSEMIA MAJOR; SICKLE CELL ANEMIA	*HBB*
340	THROMBOCYTHEMIA 2	*MPL*
341	THROMBOSIS, HYPERHOMOCYSTEINEMIC	*CBS**
342	THROMBOTIC THROMBOCYTOPENIC PURPURA, CONGENITAL	*ADAMTS13*
343	TIGHT SKIN CONTRACTURE SYNDROME, LETHAL	*ZMPSTE24*
344	TOOTH AGENESIS, SELECTIVE, X-LINKED 1	*EDA*
345	TRICHOTHIODYSTROPHY, PHOTOSENSITIVE	*ERCC3,ECC2, GTF2H5*
346	TYROSINEMIA	*HPD, TAT, FAH*
347	ULNA AND FIBULA, ABSENCE OF, WITH SEVERE LIMB DEFICIENCY	*WNT7A**
348	USHER SYNDROME	*CLRN1, CDH23*, MYO7A, GPR98, USH1C, USH1G*, USH2A*
349	VACTERL ASSOCIATION, X-LINKED	*ZIC3**
350	VATER ASSOCIATION WITH MACROCEPHALY AND VENTRICULOMEGALY	*PTEN*
351	VITAMIN E, FAMILIAL ISOLATED DEFICIENCY OF	*TTPA*
352	WAARDENBURG-SHAH SYNDROME	*EDN3, EDNRB**
353	WALKER-WARBURG SYNDROME	*POMT1, POMT2**
354	WARBURG MICRO SYNDROME	*RAB3GAP2, RAB3GAP1*
355	WILSON DISEASE	*ATP7B**
356	WRINKLY SKIN SYNDROME	*ATP6V0A2*
357	XERODERMA PIGMENTOSUM	*DDB2, ERCC3, ERCC2, ERCC4, XPC, XPA*
358	X-LINKED ASPERGER SYNDROME-2	*NLGN4X**
359	ZELLWEGER SYNDROME	*PEX12, PEX1*

*Genes having 85%-95% coverage. Note: Gene coverage varies from sample to sample in the NGS runs.