



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	<i>CYP17A1</i>
2	17-ALPHA-HYDROXYLASE/17, 20-LYASE DEFICIENCY	<i>CYP17A1</i>
3	17-BETA HYDROXYSTEROID DEHYDROGENASE III DEFICIENCY	<i>HSD17B3</i>
4	3-HYDROXYACYL-COA DEHYDROGENASE DEFICIENCY	<i>HADH*</i>
5	3-METHYLGLUTACONIC ACIDURIA	<i>AUH*, DNAJC19</i>
6	5-FLUOROURACIL TOXICITY	<i>DPYD</i>
7	ABCD SYNDROME	<i>EDNRB*</i>
8	ACHALASIA-ADDISONIANISM-ALACRIMA SYNDROME	<i>AAAS</i>
9	ACHONDROGENESIS, TYPE IB	<i>SLC26A2</i>
10	ACYL-COA DEHYDROGENASE DEFICIENCY	<i>ACADL, ACADM, ACADVL*</i>
11	ADRENAL HYPERPLASIA, CONGENITAL, DUE TO 21-HYDROXYLASE DEFICIENCY	<i>CYP21A2</i>
12	ADRENOLEUKODYSTROPHY	<i>ABCD1*, PEX1, PEX13*, PEX26*, PEX5</i>
13	AGAMMAGLOBULINEMIA	<i>BTK</i>
14	AGENESIS OF THE CORPUS CALLOSUM WITH PERIPHERAL NEUROPATHY	<i>SLC12A6</i>
15	AICARDI-GOUTIÈRES SYNDROME	<i>RNASEH2B, RNASEH2C, RNASEH2A, SAMHD1</i>
16	ALLAN-HERNDON-DUDLEY SYNDROME	<i>SLC16A2*</i>
17	ALPERS DIFFUSE DEGENERATION OF CEREBRAL GRAY MATTER WITH HEPATIC CIRRHOSIS	<i>POLG</i>
18	ALPHA THALASSEMIA	<i>ATRX, HBA1</i>
19	ALPHA-METHYLACETOACETIC ACIDURIA	<i>ACAT1</i>
20	ALPHA-METHYLACYL-COA RACEMASE DEFICIENCY	<i>AMACR*</i>
21	ALPORT SYNDROME	<i>COL4A4, COL4A3, COL4A5</i>
22	ALSTROM SYNDROME	<i>ALMS1</i>
23	AMEGAKARYOCYTIC THROMBOCYTOPENIA, CONGENITAL	<i>MPL</i>
24	AMYLOIDOSIS, HEREDITARY RENAL	<i>FGA</i>
25	ANAUXETIC DYSPLASIA	<i>RMRP</i>
26	ANDROGEN INSENSITIVITY; PARTIAL, WITH OR WITHOUT BREAST CANCER	<i>AR</i>
27	ANGELMAN SYNDROME	<i>UBE3A</i>
28	ANGELMAN SYNDROME-LIKE	<i>CDKL5</i>
29	ANTIBODY DEFICIENCY DUE TO ICOS DEFECT	<i>ICOS</i>
30	ANTLEY-BIXLER SYNDROME; DISORDERED STEROIDOGENESIS	<i>POR*</i>
31	ARGININOSUCCINIC ACIDURIA	<i>ASL</i>
32	AROMATIC L-AMINO ACID DECARBOXYLASE DEFICIENCY	<i>DDC</i>
33	ARTERIAL CALCIFICATION, GENERALIZED, OF INFANCY	<i>ENPP1</i>
34	ARTHROGRYPOSIS	<i>GLE1, VPS33B*, VIPAR, VIPAS39</i>
35	ARTS SYNDROME	<i>PRPS1</i>
36	ATAXIA	<i>APTX, ATM</i>
37	ATELOSTEOGENESIS, TYPE II	<i>SLC26A2</i>
38	ATYPICAL MYCOBACTERIOSIS, FAMILIAL	<i>IFNGR1, IL12B, IL12RB1*, IKBK, STAT1</i>
39	AUTOIMMUNE LYMPHOPROLIFERATIVE SYNDROME	<i>CASP10*, FAS</i>

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

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

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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	CHRNA7
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	GELEOPHYSC 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 SYLVIVUS;	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	<i>ABCC8</i>
179	HYPOGONADOTROPIC HYPOGONADISM	<i>GNRHR</i>
180	HYPOPARATHYROIDISM-RETARDATION-DYSMORPHISM SYNDROME	<i>TBCE*</i>
181	HYPOPHOSPHATASIA	<i>ALPL*</i>
182	HYPOPHOSPHATEMIC OSTEOPENIA, AUTOSOMAL RECESSIVE	<i>DMP1</i>
183	HYPOPHOSPHATEMIC RICKETS	<i>ENPP1, CLCN5</i>
184	HYPOSPADIAS 1, X-LINKED	<i>AR</i>
185	HYPOTHYROIDISM, CONGENITAL, NONGOITROUS, 4	<i>TSHB</i>
186	ICHTHYOSIS	<i>ABCA12, CLDN1,</i>
187	IMMUNE DYSFUNCTION	<i>ORAI1, STIM1*</i>
188	IMMUNODEFICIENCY	<i>CD3E, CD3G, CD247, CD40LG, CD19, CD3D*, DNMT3B, FOXP1</i>
189	INCONTINENTIA PIGMENTI, TYPE II	<i>IKBKG</i>
190	INFANTILE NEUROAXONAL DYSTROPHY	<i>2G6*</i>
191	INFANTILE SIALIC ACID STORAGE DISORDER; SIALURIA, FINNISH TYPE	<i>SLC17A5</i>
192	INFERTILE MALE SYNDROME	<i>AR</i>
193	INSENSITIVITY TO PAIN, CONGENITAL, WITH ANHIDROSIS	<i>NTRK1</i>
194	INVASIVE PNEUMOCOCCAL DISEASE, RECURRENT ISOLATED, 2	<i>IKBKG</i>
195	JEUNE SYNDROME	<i>IFT80, DYNC2H1</i>
196	JOHANSON-BLIZZARD SYNDROME	<i>UBR1</i>
197	JOUBERT SYNDROME	<i>AHI1, CEP290, NPHP1*, TMEM67</i>
198	JUVENILE AMYOTROPHIC LATERAL SCLEROSIS 2	<i>ALS2</i>
199	KENNY-CAFFEY SYNDROME-1	<i>TBCE*</i>
200	KERATOSIS FOLLICULARIS SPINULOSA DECALVANS, X-LINKED	<i>MBTPS2</i>
201	KRABBE DISEASE	<i>GALC</i>
202	LACTIC ACIDOSIS, FATAL INFANTILE (MTDNA DEPLETION)	<i>SUCLG1</i>
203	LATHOSTEROLOSIS	<i>SC5DL, SC5D</i>
204	LEIGH SYNDROME	<i>BCS1L, LRPPRC, PDHA1*, SURF1</i>
205	LEPRECHAUNISM	<i>INSR</i>
206	LESCH-NYHAN SYNDROME	<i>HPRT1</i>
207	LETHAL CONGENITAL CONTRACTURE SYNDROME	<i>ERBB3, GLE1</i>
208	LEUKOCYTE ADHESION DEFICIENCY TYPE III	<i>FERMT3</i>
209	LEUKODYSTROPHY	<i>FAM126A*</i>
210	LHERMITTE-DUCLOS SYNDROME	<i>PTEN</i>
211	LIPOID CONGENITAL ADRENAL HYPERPLASIA	<i>CYP11A1, STAR</i>
212	LISSENCEPHALY	<i>DCX, RELN, TUBA1A</i>
213	LOWE OCULOCEREBRORENAL SYNDROME	<i>OCRL</i>
214	LUJAN-FRYNS SYNDROME	<i>MED12</i>
215	LYMPHOPROLIFERATIVE SYNDROME	<i>SH2D1A, XIAP*</i>
216	MACROCEPHALY/AUTISM SYNDROME	<i>PTEN</i>
217	MANDIBULOACRAL DYSPLASIA	<i>ZMPSTE24</i>
218	MANNOSIDOSIS, ALPHA B, LYSOSOMAL	<i>MAN2B1</i>
219	MAPLE SYRUP URINE DISEASE	<i>BCKDHB</i>
220	MARINESCO-SJOGREN SYNDROME	<i>SIL1</i>
221	MARTSOLF SYNDROME	<i>RAB3GAP2</i>
222	MASA SYNDROME	<i>L1CAM*</i>
223	MECKEL SYNDROME	<i>MKS1*, RRGRI1L</i>
224	MEGALENCEPHALIC LEUKOENCEPHALOPATHY WITH SUBCORTICAL CYSTS	<i>MLC1*</i>
225	MENKES DISEASE	<i>ATP7A</i>

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

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

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

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

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