Amplexa Genetics®

Analysis: Genes4Life

Test type: Carrier screening test

#	Condition	Gene
1	17-Beta-Hydroxysteroid Dehydrogenase Deficiency, Type III	*HSD17B3*
2	21-Hydroxylase-Deficient Congenital Adrenal Hyperplasi	*CYP21A2*
3	2-Methylbutyryl-CoA Dehydrogenase Deficiency	*ACADSB*
4	3-Hydroxy-3-Methylglutaryl-CoA Lyase Deficiency	*HMGCL*
5	3-Hydroxyacyl-CoA Dehydrogenase Deficiency	*HADH*
6	3-Hydroxyisobutryl-CoA Hydrolase Deficiency	*HIBCH*
7	3-Methylcrotonyl-CoA Carboxylase 1 Deficiency	*MCCC1*
8	3-Methylcrotonyl-CoA Carboxylase 2 Deficiency	*MCCC2*
	A	
9	Abetalipoproteinemia	*MTTP*
10	Achalasia-Addisonianism-Alacrimia Syndrome	*AAAS*
11	Achondrogenesis, Type 1B	*SLC26A2*
12	Achromatopsia CNGB3-Related	*CNGB3*
13	Acrodermatitis Enteropathica	*SLC39A4*
14	Acute Infantile Liver Failure, TRMU-Related	*TRMU*
15	Adenosine Deaminase Deficiency	*ADA*
16	Adrenal Hyperplasia	*HSD3B2*
17	Adrenal Hyperplasia V	*CYP17A1*
18	AdrenoleukoDystrophy	*ABCD1*
19	Aicardi-Goutieres syndrom (AGS)	*RNASEH2A*
20	Aicardi-Goutieres syndrom (AGS)	*RNASEH2B*
21	Aicardi-Goutieres syndrom (AGS)	*RNASEH2C*
22	Aicardi-Goutieres syndrom (AGS)	*SAMHD1*
23	Aicardi-Goutieres Syndrome 1	*TREX1*
24	AICA-Ribosiduria	*ATIC*
25	Albinism, Oculocutaneous, Type I	*TYR*
26	Albinism, Oculocutaneous, Type II	*OCA2*
27	Albinism, Oculocutaneous, Type III	*TYRP1*
28	Albinism, Oculocutaneous, Type IV	*SLC45A2*
29	Albinism, Oculocutaneous, Type VII	*LRMDA*
30	Alkaptonuria	*HGD*
31	Alpers Syndrome	*POLG*
32	Alpha thalassemia	*HBA1*
33	Alpha thalassemia	*HBA2*
34	Alpha-Mannosidosis	*MAN2B1*
35	Alpha-Methylacetoacetic Aciduria	*ACAT1*
36	Alpha-N-Acetylgalactosaminidase Deficiency, Type 1 (Schindler Disease)	*NAGA*
37	Alport Syndrome, COL4A3-Related	*COL4A3*
38	Alport Syndrome, COL4A4-Related	*COL4A4*

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#	Condition	Gene
39	Alport Syndrome, COL4A5-Related	*COL4A5*
40	Alstrom Syndrome	*ALMS1*
41	Amish Infantile Epilepsy Syndrome	*ST3GAL5*
42	Argininosuccinic Aciduria	*ASL*
43	Aromatic L-amino acid Decarboxylase Deficiency	*DDC*
44	Arthrogryposis, Mental Retardation and Seizures	SLC35A3
45	Arthrogryposis, Renal Dysfunction and Cholestasis 1	*VPS33B*
46	Asparagine Synthetase Deficiency	*ASNS*
47	Aspartylglycosaminuria	*AGA*
48	Ataxia	*TTPA*
49	Ataxia Telangiectasia	*ATM*
	B	
50	Bardet-Biedl Syndrome 1	*BBS1*
51	Bardet-Biedl Syndrome 10	*BBS10*
52	Bardet-Biedl Syndrome 11	*TRIM32*
53	Bardet-Biedl Syndrome 12	BBS12
54	Bardet-Biedl Syndrome 13 / Meckel-Gruber Syndrome 1 / Joubert Syndrome 28	*MKS1*
55	Bardet-Biedl Syndrome 16	*SDCCAG8*
56	Bardet-Biedl Syndrome 17	*LZTFL1*
57	Bardet-Biedl Syndrome 18	*BBIP1*
58	Bardet-Biedl Syndrome 19	*IFT27*
59	Bardet-Biedl Syndrome 2	BBS2
60	Bardet-Biedl Syndrome 20	*IFT172*
61	Bardet-Biedl syndrome 21 / Cone-Rod Dystrophy 16 / Retinitis Pigmentosa 64	C8orf37
62	Bardet-Biedl Syndrome 3	*ARL6*
63	Bardet-Biedl Syndrome 4	*BBS4*
64	Bardet-Biedl Syndrome 5	BBS5
65	Bardet-Biedl Syndrome 6	*MKKS*
66	Bardet-Biedl Syndrome 7	*BBS7*
67	Bardet-Biedl Syndrome 8	*TTC8*
68	Bardet-Biedl Syndrome 9	*BBS9*
69	Bartter Syndrome	*BSND*
70	Bernard-Soulier Syndrome, Type A1	*GP1BA*
71	Bernard-Soulier Syndrome, Type C	*GP9*
72	Beta-Thalassemia	*HBB*
73	Bile Acid Synthesis Defect, Type 4	*AMACR*
74	Biotinidase Deficiency	BTD
75	Blomstrand Chondrodysplasia	*PTH1R*
76	Bloom Syndrome	*BLM*

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#	Condition	Gene
77	Brittle Cornea Syndrome 1	*ZNF469*
	C	
78	Canavan Disease	*ASPA*
79	Carbamoyl-Phosphate Synthetase 1 Deficiency	*CPS1*
80	Carnitine Deficiency	SLC22A5
81	Carnitine Palmitoyltransferase Deficiency, Type 1A	*CPT1A*
82	Carnitine-acylcarnitine Translocase Deficiency	SLC25A20
83	Carpenter Syndrome	*RAB23*
84	Cartilage-Hair Hypoplasia	*RMRP*
85	Cerebellar Hypoplasia and Mental Retardation	*VLDLR*
86	Cerebellar Hypoplasia, Type 1	*EXOSC3*
87	Cerebral Creatine Deficiency Syndrome 2	*GAMT*
88	Cerebral Dysgenesis, Neuropathy, Ichthyosis and Palmoplantar Keratoderma Syndrome	SNAP29
89	Cerebrotendinous Xanthomatosis	*CYP27A1*
90	Ceroid Lipofuscinosis	MFSD8
91	Ceroid Lipofuscinosis	*CLN8*
92	Charcot-Marie-Tooth Disease, Type 2B1	*LMNA*
93	Charcot-Marie-Tooth Disease, Type 2EE	*MPV17*
94	Charcot-Marie-Tooth Disease, Type 2S	*IGHMBP2*
95	Charcot-Marie-Tooth Disease, Type 4F	PRX
96	Charcot-Marie-Tooth Disease, Type 4H	*FGD4*
97	Choreoacanthocytosis	*VPS13A*
98	Choroideremia	CHM
99	Chronic Granulomatous Disease 1	*NCF1*
100	Chronic Granulomatous Disease 4	*CYBA*
101	Ciliary Dyskinesia, Type 1	*DNAI1*
102	Ciliary Dyskinesia, Type 9	*DNAI2*
103	Citrin Deficiency	*SLC25A13*
104	Citrullinemia, Type 1	ASS1
105	Classical homocystinuria	*CBS*
106	Coenzyme Q10 Deficiency, Type 2	PDSS1
107	Coenzyme Q10 Deficiency, Type 4	*COQ8A*
108	Cohen Syndrome	*VPS13B*
109	Cold-induced Sweating Syndrome 1	*CRLF1*
110	Colobomatous Microphthalmia	STRA6
111	Combined Malonic and Methylmalonic Aciduria	*ACSF3*
112	Combined Oxidative Phosphorylation Deficiency 1	GFM1
113	Combined Oxidative Phosphorylation Deficiency 3	TSFM
114	Cone-Rod Dystrophy and Hearing Loss 2	CEP250

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#	Condition	Gene
115	Congenital Adrenal Hyperplasia, 11-b hydroxylase	*CYP11B1*
116	Congenital Amegakaryocytic Thrombocytopenia	*MPL*
117	Congenital Disorder of Glycosylation, Type 1a	*PMM2*
118	Congenital Disorder of Glycosylation, Type Ib	*MPI*
119	Congenital Disorder of Glycosylation, Type Ic	*ALG6*
120	Congenital Disorder of Glycosylation, Type Ie	*DPM1*
121	Congenital Disorder of Glycosylation, Type II	*B4GALT1*
122	Congenital Disorder of Glycosylation, Type Iia	*MGAT2*
123	Congenital Disorder of Glycosylation, Type IIb	*MOGS*
124	Congenital Disorder of Glycosylation, Type Iic	*SLC35C1*
125	Congenital Disorder of Glycosylation, Type Iif	*SLC35A1*
126	Congenital Disorder of Glycosylation, Type Ik	*ALG1*
127	Congenital Erythropoietic Porphyria	*UROS*
128	Congenital Hypothyroidism	*PAX8*
129	Congenital Hypothyroidism	*TSHR*
130	Congenital Insensitivity to pain with Anhidrosis	*NTRK1*
131	Congenital Myasthenic Syndrome 4B	*CHRNE*
132	Congenital Non-Bullous Ichthyosiform Erythroderma	*ABCA12*
133	Congenital Thrombotic Thrombocytopenic Purpura	*ADAMTS13*
134	Corneal Endothelial Dystrophy	*SLC4A11*
135	Corpus Callosum Agenesis-Neuronopathy Syndrome	*SLC12A6*
136	Costeff Syndrome	*OPA3*
137	CPT II Deficiency, Infantile	*CPT2*
138	Cutis Laxa Classic, Type 2	*ATP6V0A2*
139	Cutis Laxa, Type IA	*FBLN5*
140	Cutis Laxa, Type IB	*EFEMP2*
141	Cystic Fibrosis	*CFTR*
142	Cystinosis	*CTNS*
143	Cystinuria	*SLC3A1*
	D	
144	Deafness 53	*COL11A2*
145	Deafness 77	*LOXHD1*
146	Dejerine-Sottas Disease	*PMP22*
147	Desmosterolosis	*DHCR24*
148	Dihydropyrimidine Dehydrogenase Deficiency	*DPYD*
149	Distal Renal Tubular Acidosis	*ATP6V1B1*
150	Donnai-Barrow Syndrome	*LRP2*
151	Dopa-Responsive Dystonia	*TH*
152	Duchenne Muscular Dystrophy	*DMD*

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#	Condition	Gene
153	Dysplasminogenemia	*PLG*
	E	
154	Ehlers-Danlos Syndrome, Cardiac Valvular Type	*COL1A2*
155	Ehlers-Danlos Syndrome, Dermatosparaxis Type	*ADAMTS2*
156	Ellis-van Creveld Syndrome	*EVC2*
157	Ellis-van Creveld Syndrome	*EVC*
158	Emery-Dreifuss Muscular Dystrophy	*FHL1*
159	Emphysema	*SERPINA1*
160	Epidermolysis Bullosa, Type 1	*LAMB3*
161	Epidermolysis Bullosa, Type 2	*LAMA3*
162	Epidermolysis Bullosa, Type 3	*LAMC2*
163	Epidermolysis Bullosa, Type 4	*COL17A1*
164	Epidermolysis Bullosa, Type 5	*ITGB4*
165	Epidermolysis Bullosa, Type 6	*ITGA6*
166	Ethylmalonic Encephalopathy	*ETHE1*
	F	
167	Fabry Disease	*GLA*
168	Factor IX Deficiency	*F9*
169	Factor VIII Deficiency	*F8*
170	Factor XI Deficiency	*F11*
171	Familial Chloride Diarrhea	*SLC26A3*
172	Familial Dysautonomia	*ELP1*
173	Familial Hyperinsulinemic Hypoglycemia	*ABCC8*
174	Familial Hyperinsulinism	*KCNJ11*
175	Familial Mediterranean Fever	*MEFV*
176	Fanconi Anemia, Group C	*FANCC*
177	Fanconi Anemia, Group G	*FANCG*
178	Fanconi Anemia, Troup A	*FANCA*
179	Fetal Akinesia Deformation Sequence	*RAPSN*
180	Fragile X Syndrome	*FMR1*
181	French Canadian Type of Leigh Syndrome	*LRPPRC*
182	Fucosidosis	*FUCA1*
183	Fuhrmann Syndrome	*WNT7A*
184	Fumarase Deficiency	*FH*
	G	
185	Galactokinase Deficiency, Type II	*GALK1*
186	Galactosemia	*GALT*
187	Gaucher Disease	*GBA*

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#	Condition	Gene
188	Gitelman Syndrome	*SLC12A3*
189	Glutaric Acidemia IIA	*ETFA*
190	Glutaric Acidemia IIC	*ETFDH*
191	Glutaric Acidemia, Type 1	*GCDH*
192	Glycine Encephalopathy	*GLDC*
193	Glycogen Storage Disease VII	*PFKM*
194	Glycogen Storage Disease, Type 1A	*G6PC1*
195	Glycogen Storage Disease, Type 2 (Pompe Disease)	*GAA*
196	Glycogen Storage Disease, Type 3	*AGL*
197	Glycogen Storage Disease, Type 4	*GBE1*
198	Glycogen Storage Disease, Type B	*SLC37A4*
199	GRACILE Syndrome	*BCS1L*
200	Greenberg Skeletal Dysplasia	*LBR*
201	Griscelli Syndrome, Type 1	*MYO5A*
202	Griscelli Syndrome, Type 2	*RAB27A*
203	Gyrate Atrophy of Choroid and Retina	*OAT*
	H	
204	Hemochromatosis, Type 2	*HJV*
205	Hemochromatosis, Type 3	*TFR2*
206	Hepatic Venoocclusive Disease with Immunodeficiency	*SP110*
207	Hereditary Fructose Intolerance	*ALDOB*
208	Hermansky-Pudlak Syndrome 1	*HPS1*
209	Holocarboxylase Synthetase Deficiency	*HLCS*
210	Homocystinuria	*MTHFR*
211	Homocystinuria-Megaloblastic Anemia	*MTRR*
212	Hunter Syndrome	*IDS*
213	Hurler syndrome	*IDUA*
214	Hyaline Fibromatosis Syndrome	*ANTXR2*
215	Hydrolethalus Syndrome	*HYLS1*
216	Hyperoxaluria, Type I	*AGXT*
217	Hyperoxaluria, Type II	*GRHPR*
218	Hyperoxaluria, Type III	*HOGA1*
219	Hyperphenylalaninemia	*PTS*
220	Hypoaldosteronism, CMO I Deficiency	*CYP11B2*
221	Hypohidrotic Ectodermal Dysplasia	*EDA*
222	Hypomyelinating leukodystrophy 12	*VPS11*
223	Hypophosphatasia	*ALPL*
	I	
224	Immunodeficiency-Centromeric Instability-Facial Anomalies Syndrome 1	*DNMT3B*

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#	Condition	Gene
225	Infantile Striatonigral Degeneration	*NUP62*
226	Insulin-Like Growth Factor I Deficiency	*IGF1*
227	Isovaleric Acidemia	*IVD*
	J	
228	Jervell and Lange-Nielsen Syndrome	*KCNQ1*
229	Joubert Syndrome 1	*INPP5E*
230	Joubert Syndrome 12	*KIF7*
231	Joubert Syndrome 13	*TCTN1*
232	Joubert Syndrome 14	*TMEM237*
233	Joubert Syndrome 15	*CEP41*
234	Joubert Syndrome 16	*TMEM138*
235	Joubert Syndrome 17	*CPLANE1*
236	Joubert Syndrome 18	*TCTN3*
237	Joubert Syndrome 2	*TMEM216*
238	Joubert Syndrome 20	*TMEM231*
239	Joubert Syndrome 21	*CSPP1*
240	Joubert Syndrome 22	*PDE6D*
241	Joubert Syndrome 23	*KIAA0586*
242	Joubert Syndrome 24	*TCTN2*
243	Joubert Syndrome 27	*B9D1*
244	Joubert Syndrome 3	*AHI1*
245	Joubert Syndrome 4	*NPHP1*
246	Joubert Syndrome 5	*CEP290*
247	Joubert Syndrome 6	*TMEM67*
248	Joubert Syndrome 7	*RPGRIP1L*
249	Joubert Syndrome 8	*ARL13B*
250	Joubert Syndrome 9	*CC2D2A*
251	Juvenile Amyotrophic Lateral Sclerosis 2	*ALS2*
252	Juvenile Hemochromatosis, Type 2	*HAMP*
253	Juvenile Neuronal Ceroid Lipofuscinose	*CLN3*
254	Juvenile Paget Disease	*TNFRSF11B*
255	Juvenile Retinoschisis	*RS1*
	K	
256	Kenny-Caffey Syndrome, Type 1	*TBCE*
257	Krabbe Disease	*GALC*
	L	
258	Lamellar Ichthyosis	*TGM1*
259	Late Infantile Neuronal Ceroid Lipofuscinoses	*CLN6*
260	Lathosterolosis	*SC5D*

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#	Condition	Gene
261	Leber Congenital Amaurosis 16	*KCNJ13*
262	Leber Congenital Amaurosis 2	*RPE65*
263	Leprechaunism	*INSR*
264	Lethal Congenital Contracture Syndrome 1	*GLE1*
265	Lethal Congenital Contracture Syndrome 2	*ERBB3*
266	Lethal Osteosclerotic Bone Dysplasia	*FAM20C*
267	Leukoencephalopathy with Vanishing White Matter	*EIF2B5*
268	Limb-Girdle Muscular Dystrophy, Type 1	*CAPN3*
269	Limb-Girdle Muscular Dystrophy, Type 17	*PLEC*
270	Limb-Girdle Muscular Dystrophy, Type 2	*DYSF*
271	Limb-Girdle Muscular Dystrophy, Type 23	*LAMA2*
272	Limb-Girdle Muscular Dystrophy, Type 3	*SGCA*
273	Limb-Girdle Muscular Dystrophy, Type 4	*SGCB*
274	Limb-Girdle Muscular Dystrophy, Type 5	*SGCG*
275	Limb-Girdle Muscular Dystrophy, Type 5C	*FKRP*
276	Limb-Girdle Muscular Dystrophy, Type C1	*POMT1*
277	Limb-Girdle Muscular Dystrophy, Type C3	*POMGNT1*
278	Lipoid Adrenal Hyperplasia	*STAR*
279	Lipoprotein Lipase Deficiency	*LPL*
280	Long-Chain Hydroxyacyl-CoA Dehydrogenase Deficiency	*HADHA*
281	Lysinuric Protein Intolerance	*SLC7A7*
	M	
282	Mandibuloacral Dysplasia	*ZMPSTE24*
283	Maple syrup urine Disease, Type 1A	*BCKDHA*
284	Maple syrup urine Disease, Type 1B	*BCKDHB*
285	Maple syrup urine Disease, Type II	*DBT*
286	Maple Syrup Urine Disease, Type III	*DLD*
287	Marinesco-Sjögren Syndrome	*SIL1*
288	Medium Chain Acyl-CoA Dehydrogenase Deficiency	*ACADM*
289	Megalencephalic Leukoencephalopathy 1	*MLC1*
290	Metachromatic Leukodystrophy	*ARSA*
291	Methylmalonic Aciduria, MMAB-Related	*MMAB*
292	Methylmalonic Aciduria, MMACHC-Related	*MMACHC*
293	Methylmalonic Aciduria, MMUT-Related	*MMUT*
294	Methylmalonic Aciduria, MMAA-Related	*MMAA*
295	Mitochondrial Complex I Deficiency, ACAD9-Related	*ACAD9*
296	Mitochondrial DNA depletion Syndrome 1, TYMP-Related	*TYMP*
297	Mitochondrial Trifunctional Protein Deficiency 2	*HADHB*
298	Molybdenum Cofactor Deficiency, Type A	*MOCS1*

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299	Mucolipidosis II/IIIA	*GNPTAB*
300	Mucolipidosis III gamma	*GNPTG*
301	Mucolipidosis, Type IV	*MCOLN1*
302	Mucopolysaccharidosis, Type IVB	*GLB1*
303	Mucopolysaccharidosis, Type IX	*HYAL1*
304	Mucopolysaccharidosis, Type VI	*ARSB*
305	Mucopolysaccharidosis, Type VII	*GUSB*
306	Mulibrey Nanism	*TRIM37*
307	Multiple Pterygium Syndrome	*CHRNA1*
308	Multiple Sulfatase Deficiency	*SUMF1*
309	Myasthenic Syndrome 13	*DPAGT1*
310	Myasthenic Syndrome 22	*PREPL*
311	Myoclonic Epilepsy of Lafora, Type 2A	*EPM2A*
312	Myoclonic Epilepsy of Lafora, Type 2B	*NHLRC1*
313	Myoclonic Epilepsy of Unverricht and Lundborg, Type 1A	*CSTB*
314	Myophosphorylase Deficiency	*PYGM*
	N	
315	N-Acetylglutamate Synthase Deficiency	*NAGS*
316	Nemaline Myopathy 2	*NEB*
317	Nemaline Myopathy 5	*TNNT1*
318	Neonatal Glycine Encephalopathy	*AMT*
319	Neonatal Glycine Encephalopathy	*GCSH*
320	Neonatal Ichthyosis-Sclerosing Cholangitis	*CLDN1*
321	Nephronophthisis 3	*NPHP3*
322	Nephrotic Syndrome, Type 1	*NPHS1*
323	Nephrotic Syndrome, Type 2	*NPHS2*
324	Neuronal ceroid lipofuscinosis, PPT1-Related	*PPT1*
325	Neuronal ceroid lipofuscinosis, TPP1-Related	*TPP1*
326	Neuronal ceroid-lipofuscinosis, CLN5-Related	*CLN5*
327	Neutropenia, Severe congenital 3	*HAX1*
328	Neutropenia, Severe congenital 4	*G6PC3*
329	Niemann-Pick Disease	*SMPD1*
330	Niemann-Pick Disease, Type C1	*NPC1*
331	Niemann-pick Disease, Type C2	*NPC2*
332	Nijmegen Breakage Syndrome	*NBN*
333	Nonaka Myopathy	*GNE*
334	Non-Syndromic Hearing Loss	*TMC1*
335	Non-Syndromic Hearing Loss, GJB2-Related	*GJB2*

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	O	
336	Odontoonychodermal Dysplasia	*WNT10A*
337	Omenn syndrome	*RAG2*
338	Omenn Syndrome	*RAG1*
339	Omenn Syndrome	*DCLRE1C*
340	Osteopetrosis	*CA2*
341	Osteopetrosis, Infantile Malignant	*TCIRG1*
	P	
342	Pendred Syndrome	*SLC26A4*
343	Peroxisomal Acyl-CoA Oxidase Deficiency	*ACOX1*
344	Peroxisome Biogenesis Disorder	*PEX7*
345	Perrault Syndrome 1	*HSD17B4*
346	Perrault Syndrome 5	*TWNK*
347	Phenylketonuria	*PAH*
348	Pierson Syndrome	*LAMB2*
349	Pituitary Hormone Deficiency 2	*PROP1*
350	Polycystic Kidney Disease 4	*PKHD1*
351	Polyglandular aAutoimmune Syndrome, Type 1	*AIRE*
352	Polymicrogyria	*ADGRG1*
353	Polyneuropathy, Hearing Loss, Ataxia, Retinitis Pigmentosa and Cataract, PHARC Disorder	*ABHD12*
354	Pontocerebellar Hypoplasia, Type 1	*RARS2*
355	Pontocerebellar Hypoplasia, Type 2	*TSEN54*
356	Postnatal Progressive Microcephaly	*MED17*
357	Primary Ciliary Dyskinesia, DNAH5-Related	*DNAH5*
358	Primary congenital glaucoma	*CYP1B1*
359	Progressive Cerebellocerebral Atrophy, Type 1	*SEPSECS*
360	Progressive Cerebellocerebral Atrophy, Type 2	*VPS53*
361	Progressive Familial Intrahepatic Cholestasis, Type 1	*ATP8B1*
362	Progressive Familial Intrahepatic Cholestasis, Type 2	*ABCB11*
363	Propionicacidemia	*PCCA*
364	Propionicacidemia	*PCCB*
365	Pseudohypoaldosteronism, Type IB	*SCNN1B*
366	Pseudohypoaldosteronism, Type IB1	*SCNN1A*
367	Pseudohypoaldosteronism, Type IB3	*SCNN1G*
368	Pycnodysostosis	*CTSK*
369	Pyridoxamine 5'-Phosphate Oxidase Deficiency	*PNPO*
370	Pyruvate Kinase Deficiency	*PKLR*
	R	
371	Renal Hypomagnesemia 5	*CLDN19*

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#	Condition	Gene
372	Retinitis Pigmentosa 12	*CRB1*
373	Retinitis Pigmentosa 25	*EYS*
374	Retinitis Pigmentosa 26	*CERKL*
375	Retinitis Pigmentosa 28	*FAM161A*
376	Retinitis Pigmentosa 59	*DHDDS*
377	Rhizomelic Chondrodysplasia Punctata, Type 3	*AGPS*
378	Roberts Syndrome	*ESCO2*
	S	
379	Salla Disease	*SLC17A5*
380	Sandhoff Disease	*HEXB*
381	Sanfilippo Syndrome A	*SGSH*
382	Sanfilippo Syndrome B	*NAGLU*
383	Sanfilippo Syndrome C	*HGSNAT*
384	Schimke Immunoosseous Dysplasia	*SMARCAL1*
385	Schneckenbecken Dysplasia	*SLC35D1*
386	Schwartz-Jampel Syndrome, Type 1	*HSPG2*
387	Seckel Syndrome	*ATR*
388	Senior-Loken Syndrome 4	*NPHP4*
389	Senior-Loken Syndrome 5	*IQCB1*
390	Short-Chain Acyl-CoA Dehydrogenase Deficiency	*ACADS*
391	Short-Rib Thoracic Dysplasia	*TTC21B*
392	Shwachman-Diamond Syndrome	*SBDS*
393	Sialidosis, Type I	*NEU1*
394	Sjogren-Larsson Syndrome	*ALDH3A2*
395	Skin Fragility-Woolly Hair Syndrome	*DSP*
396	Smith-Lemli-Opitz Syndrome	*DHCR7*
397	Spastic Ataxia of Charlevoix-Saguenay	*SACS*
398	Spinal Muscular Atrophy	*SMN1*
399	Spinal Muscular Atrophy Type 2	*SMN2*
400	Stargardt Disease 1	*ABCA4*
401	Steel Syndrome	*COL27A1*
402	Stuve-Wiedemann Syndrome / Schwartz-Jampel Syndrome, Type 2	*LIFR*
403	Succinate-CoA Ligase Deficiency	*SUCLA2*
404	Sudden infant death with dysgenesis of the testes Syndrome	*TSPYL1*
405	Sulfite Oxidase Deficiency	*SUOX*
	T	
406	Tay-Sachs Disease	*HEXA*
407	T-cell immunodeficiency, Congenital Alopecia and Nail Dystrophy	*FOXN1*
408	Tetra-Amelia Syndrome 1	*WNT3*

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Test type: Carrier screening test

#	Condition	Gene
409	Tyrosinemia, Type I	*FAH*
	U	
410	Usher Syndrome, Type 1	*USH1G*
411	Usher Syndrome, Type 1B	*MYO7A*
412	Usher Syndrome, Type 1C	*USH1C*
413	Usher Syndrome, Type 1D	*CDH23*
414	Usher Syndrome, Type 1F	*PCDH15*
415	Usher Syndrome, Type 2A	*USH2A*
416	Usher Syndrome, Type 2C	*ADGRV1*
417	Usher Syndrome, Type 2D	*WHRN*
418	Usher Syndrome, Type 3	*CLRN1*
419	Usher Syndrome, Type IJ	*CIB2*
	V	
420	Very-long Chain Acyl-CoA Dehydrogenase Deficiency	*ACADVL*
421	Vitamin D-dependent Rickets, Type I	*CYP27B1*
	W	
422	Walker-Warburg Syndrome	*FKTN*
423	Warsaw Breakage Syndrome	*DDX11*
424	Wilson Disease	*ATP7B*
425	Wolcott-Rallison Syndrome	*EIF2AK3*
426	Wolman Disease	*LIPA*
	Z	
427	Zellweger spectrum Disorders 1A	*PEX1*
428	Zellweger spectrum Disorders 4A	*PEX6*
429	Zellweger spectrum Disorders 5A	*PEX2*
430	Zellweger spectrum Disorders 6A	*PEX10*