KTEST

DANH SÁCH BỆNH ĐƯỢC SÀNG LỌC TRONG GÓI XÉT NGHIỆM "NGƯỜI MANG"

#	DISEASE	GENE	GHI CHÚ
1	17-beta-hydroxysteroid dehydrogenase defciency, type III	*HSD17B3*	
2	3-Hydroxy-3-methylglutaryl-CoA lyase deficiency	*HMGCL*	
3	3-hydroxyacyl-CoA dehydrogenase defciency	*HADH*	
4	3-hydroxyisobutryl-CoA hydrolase deficiency	*HIBCH*	
5	3-Ketothiolase deficiency	*ACAT1*	
6	3-methylglutaconic aciduria	*AUH, DNAJC19*	
7	46, XX sex reversal 4 46XY sex reversal 3	*NR5A1*	
8	Aarskog-Scott syndrome	*FGD1*	
9	ABCD syndrome	*EDNRB*	
10	Achalasia-addisonianism-alacrima syndrome	*AAAS*	
11	Achondrogenesis, type IB	*SLC26A2*	
12	Acyl-CoA dehydrogenase-9 (ACAD9) Deficiency	*ACAD9*	
13	Adenosine deaminase deficiency	*ADA*	
14	Adrenal insufficiency, congenital, with 46XY sex reversal, partial or complete	*CYP11A1*	
15	Adrenocortical insufficiency	*NR5A1*	
16	Adrenoleukodystrophy, X-linked	*ABCD1*	
17	Afibrinogenemia, congenital	*FGA*	
18	Agammaglobulinemia, X-linked 1 Agammaglobulinemia and isolated hormone deficiency	*BTK*	
19	Aicardi-Goutieres syndrome	*RNASEH2A, RNASEH2B, RNASEH2C, SAMHD1, TREX1*	
20	Allan-Herndon-Dudley syndrome	*SLC16A2*	
21	Alpers-Huttenlocher syndrome	*POLG*	
22	Alpha thalassemia	*HBA1, ATRX*	ATRX (Alpha thalassemia X-linked intellectual disability syndrome)
23	Alpha-mannosidosis	*MAN2B1*	
24	Alpha-methylacyl-CoA racemase deficiency	*AMACR*	
25	Alport syndrome	*COL4A3, COL4A4, COL4A5*	

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26	Alstrom syndrome	*ALMS1*	
27	Amish infantile epilepsy syndrome	*ST3GAL5*	
28	Amyotrophic lateral sclerosis	*ALS2*	
29	Anauxetic dysplasia	*RMRP*	
30	Andermann syndrome	*SLC12A6*	
31	Androgen insensitivity syndrome	*AR*	
32	Angelman syndrome	*UBE3A*	
33	Antley-Bixler syndrome with genital anomalies and disordered steroidogenesis	*POR*	
34	Aplastic anemia	*PRF1*	
35	Apparent mineralocorticoid excess	*HSD11B2*	
36	Argininosuccinate lyase deficiency	*ASL*	
37	Aromatase deficiency	*CYP19A1*	
38	Aromatic L-amino acid decarboxylase deficiency	*DDC*	
39	Arterial calcification, generalized, of infancy, 1	*ENPP1*	
40	Arthrogryposis, renal dysfunction, and cholestasis 1	*VPS33B, VIPAR*	
41	Arts syndrome	*PRPS1*	
42	Asperger syndrome susceptibility, X-linked	*NLGN4X*	
43	Ataxia neuropathy spectrum	*POLG*	
44	Ataxia with isolated vitamin E deficiency	*TTPA*	
45	Ataxia, early-onset, with oculomotor apraxia and hypoalbuminemia	*APTX*	
46	Ataxia-telangiectasia	*ATM*	
47	Atelosteogenesis II	*SLC26A2*	
48	Autism susceptibility, X-linked	*NLGN4X, RPL10*	
49	Autoimmune Lymphoproliferative Syndrome	*CASP10, FAS, FASLG*	
50	Autoimmune polyendocrinopathy syndrome	*AIRE*	
51	Autosomal Recessive Spastic Ataxia of Charlevoix-Saguenay	*SACS*	
52	Bannayan-Riley-Ruvalcaba syndrome	*PTEN*	
53	Bardet-Biedl syndrome	*CEP290, MKS1*	
54	Barth syndrome	*TAZ*	
55	Bartter syndrome	*SLC12A1, KCNJ1*	
56	Beta thalassemia	*HBB*	
57	Bethlem myopathy 1	*COL6A1, COL6A2, COL6A3*	
58	Bile acid synthesis defect	*AMACR*	
59	Biotinidase deficiency	*BTD*	

60	Björnstad syndrome	*BCS1L*	
61	Bloom syndrome	*BLM*	
62	Bone mineral density variation QTL, osteoporosis	*COL1A1*	
63	Brittle cornea syndrome 1	*ZNF469*	
64	Caffey disease	*COL1A1*	
65	Canavan disease	*ASPA*	
66	Carbamoylphosphate synthetase I deficiency	*CPS1*	
67	Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency	*COX15, SCO2*	
68	Carnitine palmitoyltransferase deficiency	*CPT1A, CPT2*	
69	Carnitine-acylcarnitine translocase deficiency	*SLC25A20*	
70	Carpenter syndrome	*RAB23*	
71	Cartilage-hair hypoplasia	*RMRP*	
72	Cataract 40, X-linked	*NHS*	
73	Cerebellar hypoplasia and mental retardation with or without quadrupedal locomotion 1	*VLDLR*	
74	Cerebral dysgenesis, neuropathy, ichthyosis, and palmoplantar keratoderma syndrome	*SNAP29*	
75	Cerebrooculofacioskeletal syndrome 1	*ERCC6*	
76	Cerebrotendinous xanthomatosis	*CYP27A1*	
77	Charcot-Marie-Tooth disease	*FGD4, GDAP1, LMNA, PMP22, PRPS1, PRX, SURF1*	
78	Chediak-Higashi syndrome	*LYST*	
79	Cholestasis	*ABCB4, ATP8B1*	
80	Chondrodysplasia punctata type 1, X-linked	*ARSE*	
81	Chondrodysplasia, Blomstrand type	*PTH1R*	
82	Citrullinemia	*ASS1*	
83	COACH syndrome	*RPGRIP1L, TMEM67*	
84	Cockayne syndrome type B	*ERCC6, ERCC8*	
85	Coenzyme Q10 deficiency, primary	*ADCK3, COQ2, COQ9, PDSS1, PDSS2*	
86	Coffin-Lowry syndrome	*RPS6KA3*	
87	Cohen syndrome	*VPS13B*	
88	Cold-induced sweating syndrome 1	*CRLF1*	
89	Combined cellular and humoral immune defects with granulomas	*RAG1, RAG2*	

90	Combined oxidative phosphorylation deficiency	*GFM1, MRPS16, TSFM, TUFM, MRPS22*	
91	Combined pituitary hormone deficiency	*POU1F1, PROP1, LHX3*	
92	Combined SAP deficiency	*PSAP*	
93	Congenital adrenal hyperplasia	*CYP11B1, CYP17A1, HSD3B2*	
94	Congenital amegakaryocytic thrombocytopenia	*MPL*	
95	Congenital disorder of glycosylation	*ALG1, COG1, COG7, COG8, DOLK, DPAGT1, DPM1, MGAT2, MOGS, MPDU1, MPI, PMM2, RFT1, SLC35A1, SLC35C1, SRD5A3, ALG12, ALG2, ALG3, ALG6, ALG8, ALG9, B4GALT1*	
96	Congenital heart defects, nonsyndromic, 1, X-linked	*ZIC3*	
97	Congenital ichthyosis	*ABCA12, TGM1*	
98	Congenital insensitivity to pain with anhidrosis	*NTRK1*	
99	Corneal endothelial dystrophy	*SLC4A11, TCF4*	
100	Costeff syndrome	*OPA3*	
101	Cowden syndrome 1	*PTEN*	
102	Craniofrontonasal dysplasia	*EFNB1*	
103	Creatine deficiency syndrome	*SLC6A8*	
104	Cutis laxa	*ATP6V0A2, EFEMP2, FBLN5*	
105	Cystic fibrosis	*CFTR*	
106	Cystinosis	*CTNS*	
107	D-bifunctional protein deficiency	*HSD17B4*	
108	De Sanc- tis-Cacchione syndrome	*ERCC6*	
109	Deafness, X-linked 1 recessive	*PRPS1*	
110	Dejerine-Sottas disease	*PRX, MPZ*	
111	Dent disease	*CLCN5, OCRL*	
112	Desmosterolosis	*DHCR24*	
113	Diabetes mellitus, type I, susceptibility to	*FOXP3*	
114	Diarrhea 4, malabsorptive, congenital	*NEUROG3*	
115	Diastrophic dysplasia	*SLC26A2*	
116	Dihydrolipoamide dehydrogenase deficiency	*DLD*	
117	Dihydropyrimidine dehydrogenase deficiency	*DPYD*	
118	Donnai-Barrow syndrome	*LRP2*	
119	Duchenne muscular dystrophy	*DMD*	
120	Dyskeratosis congenita, X-linked	*DKC1*	
121	Dystonia 27	*COL6A3*	
122	Ectodermal dysplasia	*IKBKG*	

123	Ehlers-Danlos syndrome	*COL1A1, PLOD1, COL1A2*	
124	Eiken syndrome	*PTH1R*	
125	Ellis-van Creveld syndrome	*EVC, EVC2*	
126	Encephalopathy, acute, infection-induced (herpes-specific)	*TLR3, UNC93B1*	
127	Encephalopathy, neonatal severe	*MECP2*	
128	Epidermolysis bullosa dystrophica, AR Epidermolysis bullosa pruriginosa	*COL7A1*	
129	Epidermolysis bullosa simplex with pyloric atresia	*PLEC*	
130	Epidermolysis bullosa, junctional	*COL17A1, ITGA6, ITGB4, LAMA3, LAMB3, LAMC2*	
131	Epidermolysis bullosa, lethal acantholytic	*DSP*	
132	Epilepsy, progressive myoclonic	*CSTB, EPM2A, NHLRC1*	
133	Epileptic encephalopathy, early infantile	*PCDH19, SLC25A22, ST3GAL3, ARHGEF9, ARX, CDKL5*	
134	Ethylmalonic encephalopathy	*ETHE1*	
135	Exudative vitreoretinopathy 2, X-linked	*NDP*	
136	Fabry disease	*GLA*	
137	Failure of tooth eruption, primary	*PTH1R*	
138	Familial dysautonomia (HSAN3)	*IKBKAP*	
139	Familial hyperinsulinism	*ABCC8, HADH*	
140	Familial Mediterranean fever	*MEFV*	
141	Fanconi anemia group C	*FANCC*	
142	Fetal akinesia deformation sequence	*DOK7, RAPSN*	
143	Fraser syndrome	*FRAS1, FREM2*	
144	Fucosidosis	*FUCA1*	
145	Fuhrmann syndrome	*WNT7A*	
146	Fumarase deficiency	*FH*	
147	Galactokinase deficiency	*GALK1*	
148	Galactosemia	*GALT*	
149	Gallbladder disease 1	*ABCB4*	
150	Gastric cancer, somatic	*CASP10, IL1RN*	IL1RN (Gastric cancer risk after H. pylori infection)
151	Gaucher disease	*PSAP, GBA*	
152	Geleophysic dysplasia 1	*ADAMTSL2*	
153	Gillessen-Kaesbach-Nishimura syndrome	*ALG9*	
154	Glioma susceptibility 2	*PTEN*	
155	Glucose-6-phosphate dehydrogenase deficiency	*G6PD*	
156	Glutaric aciduria	*ETFA, ETFB, ETFDH, GCDH*	
157	Glutathione synthetase deficiency	*GSS*	

158	Glycine encephalopathy	*AMT, GCSH, GLDC*	
159	Glycogen storage disease	*AGL, G6PC, GBE1, PYGM, SLC37A4*	
160	GRACILE syndrome	*BCS1L*	
161	Greenberg skeletal dysplasia	*LBR*	
162	Griscelli syndrome	*MYO5A, RAB27A*	
163	Guanidinoacetate methyltransferase deficiency	*GAMT*	
164	H. pylori infection, susceptibility to	*IFNGR1*	
165	Hemochromatosis	*HAMP, HFE2*	
166	Hemophagocytic lymphohistiocytosis, familial	*PRF1, STX11, STXBP2, UNC13D*	
167	Hemophilia	*F8, F9*	
168	Hepatic venoocclusive disease with immunodeficienc	*SP110*	
169	Hepatitis B virus infection, susceptibility to	*IFNGR1*	
170	Hereditary fructose intolerance	*ALDOB*	
171	Hermansky-Pudlak syndrome	*AP3B1, PLDN*	
172	Heterotaxy, visceral, 1, X-linked	*ZIC3*	
173	HIV1 infection, resistance to	*TLR3*	
174	Holocarboxylase synthetase deficiency	*HLCS*	
175	Homocystinuria due to cystathionine beta-synthase deficiency	*CBS*	
176	HPRT-related gout	*HPRT1*	
177	HSD10 mitochondrial disease	*HSD17B10, BCS1L*	
178	Hyper IgM syndrome, X-linked	*CD40LG*	
179	Hyper-IgD syndrome	*MVK*	
180	Hyper-IgE recurrent infection syndrome, autosomal recessive	*DOCK8*	
181	Hyperornithinemia-hyperammonemia-ho- mocitrullinemia syndrome (Triple H syndrome)	*SLC25A15*	
182	Hypogonadotropic hypogonadism 7 without anosmia	*GNRHR*	
183	Hypohidrotic ectodermal dysplasia	*EDA*	
184	Hypomagnesemia 5, renal, with ocular involvement	*CLDN19*	
185	Hypoparathyroidism-retardation-dysmorphism syndrome	*TBCE*	
186	Hypophosphatasia	*ALPL*	
187	Hypophosphatemic rickets	*CLCN5, DMP1, ENPP1*	
188	Hypothryoidism, congenital, nongoitrous 4	*TSHB*	

189	Hypotrichosis, congenital, with juvenile macular dystrophy	*CDH23*	
190	Ichthyosis, leukocyte vacuoles, alopecia, and sclerosing cholangitis	*CLDN1*	
191	IFAP syndrome with or without BRESHECK syndrome	*MBTPS2*	
192	Immunodeficiency	*CD19, CD247, CD3D, CD3E, CD3G, IFNGR1, IFNGR2, IKBKG, IL12B, IL12RB1, ORAI1, STAT1, STIM1, TYK2, ICOS*	
193	Immunodeficiency-centromeric instability-facial anomalies syndrome 1	*DNMT3B*	
194	Immunodysregulation, polyendocrinopathy, and enteropathy, X-linked	*FOXP3*	
195	Incontinentia pigmenti	*IKBKG*	
196	Infantile neuroaxonal dystrophy	*PLA2G6*	
197	Invasive pneumococcal disease	*IKBKG*	
198	Isovaleric acidemia	*IVD*	
199	Johanson-Blizzard syndrome	*UBR1*	
200	Joubert syndrome	*AHI1, CEP290, MKS1, NPHP1, OFD1, RPGRIP1L, TMEM67*	
201	Kahrizi syndrome	*SRD5A3*	
202	Krabbe disease	*GALC*	
203	L1 syndrome	*L1CAM*	
204	Lacticacidemia due to PDX1 deficiency	*PDHX*	
205	Lathosterolosis	*SC5DL*	
206	Leber congenital amaurosis	*CEP290*	
207	Leigh syndrome	*NDUFS7*	
208	Leigh syndrome due to cytochrome c oxidase deficiency	*COX15*	
209	Leigh syndrome due to mitochondrial complex I deficiency	*NDUFS3, NDUFS4, NDUFS6, NDUFS8*	
210	Leigh syndrome due to mitochondrial COX4 deficiency	*COX10, SURF1*	
211	Leigh syndrome with Complex IV deficiency	*LRPPRC*	
212	Leprechaunism	*INSR*	
213	Lesch-Nyhan syndrome;	*HPRT1*	
214	Lethal congenital contracture syndrome	*GLE1, ERBB3*	
215	Leukemia, acute myeloid	*NSD1*	
216	Leukocyte adhesion deficiency, type III	*FERMT3*	
217	Leukodystrophy, hypomyelinating	*FAM126A, GJC2*	

218	Leydig cell hypoplasia;	*LHCGR*	
219	Lhermitte-Duclos syndrome	*PTEN*	
220	LIG4 syndrome	*LIG4*	
221	Lipoid congenital adrenal hyperplasia	*STAR*	
222	Lissencephaly	*DCX, RELN, TUBA1A*	
223	Long-chain 3-hydroxyacyl-CoA dehydrogenase (LCHAD) deficiency	*HADHA*	
224	Lowe syndrome	*OCRL*	
225	Lujan-Fryns syndrome	*MED12*	
226	Lung cancer, susceptibility to	*FASLG*	
227	Luteinizing hormone resistance	*LHCGR*	
228	Lymphoma, non-Hodgkin	*CASP10, PRF1*	
229	Lymphoproliferative syndrome	*SH2D1A, XIAP*	
230	Macrocephaly/autism syndrome	*PTEN*	
231	Macroglobulinemia, Waldenstrom, somatic	*MYD88*	
232	Macular degeneration, age-related, 3; AD	*FBLN5*	
233	Mandibuloacral dysplasia	*LMNA, ZMPSTE24*	
234	Maple syrup urine disease	*BCKDHA, BCKDHB, DBT*	
235	Marinesco-Sjogren syndrome	*SIL1*	
236	Martsolf syndrome	*RAB3GAP2*	
237	Meckel syndrome	*CEP290, MKS1, RPGRIP1L, TMEM67*	
238	Medium-chain acyl-CoA dehydrogenase (MCAD) deficiency	*ACADM*	
239	MEDNIK syndrome	*AP1S1*	
240	Megalencephalic leukoencephalopathy with subcortical cysts	*MLC1*	
241	Meningioma	*PTEN*	
242	Menkes disease	*ATP7A*	
243	Mental retardation and microcephaly with pontine and cerebellar hypoplasia Mental retardation, with or without nystagmus	*CASK*	
244	Mental retardation, autosomal recessive	*GRIK2, NSUN2, PRSS12, TUSC3, TRAPPC9, ST3GAL5*	
245	Mental retardation, X-linked	*ACSL4, AFF2, AP1S2, ARHGEF6, BRWD3, CUL4B, DLG3, FGD1, FTSJ1, GDI1, HUWE1, IL1RAPL1, KDM5C, MECP2, NLGN4X, OPHN1, PAK3, RAB39B, RPL10, RPS6KA3, SHROOM4, SLC9A6, SOX3, SYP, UBE3A, UPF3B, ZDHHC9, ZNF711*	
246	Metachromatic leukodystrophy	*ARSA, PSAP*	

247	Metaphyseal dysplasia without hypotrichosis	*RMRP, PTH1R*	
248	Methylmalonic acidemia	*MUT*	
249	Methylmalonic aciduria	*MMAA, MMAB, MMACHC*	MMACHC (Methylmalonic aciduria and homocystin-uria)
250	Mevalonic aciduria	*MUT, MVK*	
251	Microphthalmia, syndromic	*BCOR, STRA6*	
252	Microvascular complications of diabetes	*IL1RN*	
253	Mitochondrial complex I deficiency	*NDUFA1, NDUFAF2, NDUFAF4, NDUFV1, NDUFS3, NDUFS4, NDUFS6, NDUFS8*	
254	Mitochondrial complex III deficiency, nuclear	*UQCRB, UQCRQ, BCS1L*	
255	Mitochondrial complex IV deficiency	*COX6B1, FASTKD2, COX10, SCO1*	
256	Mitochondrial DNA depletion syndrome	*C10orf2, DGUOK, RRM2B, SUCLA2, SUCLG1, TK2*	
257	Mitochondrial neurogastrointestinal encephalopathy (MNGIE) disease	*TYMP, BCS1L*	
258	Mitochondrial trifunctional protein deficiency	*HADHA, HADHB*	
259	Mohr-Tranebjaerg syndrome	*TIMM8A*	
260	Molybdenum cofactor deficiency	*MOCS1, MOCS2*	
261	Mowat-Wilson syndrome	*ZEB2*	
262	Mucolipidosis	*GNPTAB, MCOLN1*	
263	Mucopolysaccharidosis	*ARSB, GLB1, GUSB, HGSNAT, IDS, IDUA, NAGLU, SGSH*	
264	Mulibrey nanism	*TRIM37*	
265	Multiple epiphyseal dysplasia	*SLC26A2*	
266	Multiple pterygium syndrome	*CHRNA1, CHRND, CHRNG*	
267	Muscular dystrophy	*LAMA2, SEPN1*	
268	Muscular dystrophy-dystroglycanopathy	*POMGNT1, POMT1, POMT2, LARGE*	
269	Muscular dystrophy-dystroglycanopathy, FKRP-related	*FKRP, FKTN*	
270	Myasthenic syndrome	*ALG2, CHRNA1, CHRND, DOK7, RAPSN, DPAGT1*	
271	Mycobacterium tuberculosis, susceptibility to	*SP110*	
272	Myocerebrohepa- topathy syndrome	*POLG*	
273	Myopathy, tubular aggregate	*ORAI1, STIM1*	
274	Myopia 6	*SCO2*	Nearsightedness
275	Myosclerosis, congenital	*COL6A2*	
276	Myotubular myopathy, X-linked	*MTM1*	

277	N-acetylglutamate synthase deficiency	*NAGS*	
278	Nance-Horan syndrome	*NHS*	
279	Nemaline myopathy	*NEB*	
280	Nephrolithiasis, type I	*CLCN5*	
281	Nephronophthisis	*NPHP1, TMEM67, INVS*	
282	Nephrotic syndrome	*NPHS1, NPHS2, PLCE1, LAMB2*	
283	Neurodegeneration due to cerebral folate transport deficiency	*FOLR1*	
284	Neurodegeneration with brain iron accumulation 1 HARP syndrome	*PANK2*	
285	Neuronal ceroid lipofuscinosis	*CLN3, CLN5, CLN6, CLN8, TPP1, MFSD8, PPT1, CTSD, CTSF*	
286	Neuronopathy, distal hereditary motor, type VI	*IGHMBP2*	
287	Neuropathy, congenital hypomyelinating	*EGR2*	
288	Neuropathy, hereditary, with or without age-related macular degeneration	*FBLN5*	
289	Neuropathy, inflammatory demyelinating Neuropathy, recurrent, with pressure palsies	*PMP22*	
290	Neutropenia, severe congenital	*G6PC3, WAS, HAX1*	
291	Niemann-Pick disease	*NPC1, NPC2, SMPD1*	
292	Nijmegen breakage syndrome	*NBN*	
293	Non-syndromic hearing loss	*MYO7A, PRPS1, USH1C, GJB2*	
294	Norrie disease	*NDP*	
295	Odon- toonychodermal dysplasia	*WNT10A*	
296	Ohdo syndrome, X-linked	*MED12*	
297	Omenn syndrome	*DCLRE1C, RAG1, RAG2*	
298	Opitz GBBB syndrome, type I	*MID1*	
299	Ornithine transcarbamylase deficiency	*OTC*	
300	Osteogenesis imperfecta	*COL1A1, LEPRE1, CRTAP*	
301	Osteopetrosis	*CA2, CLCN7, OSTM1, TCIRG1*	
302	Paget disease of bone 5, juvenile-onset	*TNFRSF11B*	
303	Peroxisomal acyl-CoA oxidase deficiency	*ACOX1*	
304	Peroxisome biogenesis disorder	*PEX5, PEX13*	
305	Phenylalanine hydroxylase deficiency (Phenylketonuria)	*PAH*	
306	Phosphoribosylpyrophosphate synthetase superactivity	*PRPS1*	
307	Pierson syndrome	*LAMB2*	

308	Pitt-Hopkins syndrome	*TCF4*	
309	Plasminogen deficiency	*PLG*	
310	Polycystic kidney disease, PKHD1-related	*PKHD1*	
311	Polycystic liver disease 3 with or without kidney cysts	*ALG8*	
312	Pompe disease	*GAA*	
313	Pontocerebellar hypoplasia	*TSEN54*	
314	Porphyria, congenital erythropoietic	*UROS*	
315	Premature ovarian failure 7	*NR5A1*	
316	Primary lateral sclerosis	*ALS2*	
317	Progressive external ophthalmoplegia	*POLG, RRM2B*	
318	Properdin deficiency, X-linked	*CFP*	
319	Propionic acidemia, PCCA-related	*PCCA, PCCB*	
320	Prostate cancer, somatic	*PTEN*	
321	Proteinuria, low molecular weight, with hypercalciuric nephrocalcinosis	*CLCN5*	
322	Pseudohypoaldosteronism, type I	*SCNN1A, SCNN1B, SCNN1G*	
323	Pseudovaginal perineoscrotal hypospadias	*SRD5A2*	
324	PTEN hamartoma tumor syndrome	*PTEN*	
325	Pulmonary surfactant dysfunction	*ABCA3, SFTPB, SFTPC, ACADL*	
326	Pyogenic bacterial infections, recurrent, due to MYD88 deficiency	*MYD88*	
327	Pyridoxamine 5'-phosphate oxidase deficiency	*PNPO*	
328	Pyridoxine-dependent epilepsy	*ALDH7A1*	
329	Pyruvate carboxylase deficiency	*PC*	
330	Pyruvate dehydrogenase E1-alpha deficiency	*PDHA1*	
331	Pyruvate dehydrogenase phosphatase deficiency	*PDP1*	
332	Pyruvate kinase deficiency	*PKLR*	
333	Raine syndrome	*FAM20C*	
334	Renal-hepatic-pancreatic dysplasia 1	*NPHP3*	
335	Renpenning syndrome	*PQBP1*	
336	Restrictive dermopathy, lethal	*ZMPSTE24*	
337	Retinitis pigmentosa	*POMGNT1, CLRN1*	
338	Rett syndrome, congenital variant	*FOXG1*	
339	Rhizomelic chondrodysplasia punctata	*AGPS, PEX7*	
340	Rickets, vitamin D-resistant	*VDR*	
341	Roberts syndrome	*ESCO2*	
342	Rosenberg-Chutorian syndrome	*PRPS1*	
343	Roussy-Levy syndrome	*PMP22*	

344	Salla disease	*SLC17A5*	
345	Sandhoff disease	*HEXB*	
346	SC phocomelia syndrome	*ESCO2*	
347	Schneckenbecken dysplasia	*SLC35D1*	
348	Schopf-Schulz-Passarge syndrome	*WNT10A*	
349	Schwartz-Jampel syndrome, type 1	*HSPG2*	
350	Seckel syndrome 1	*ATR*	
351	Segawa syndrome	*TH*	
352	Senior-Løken	*CEP290, NPHP1*	
353	Senior-Loken syndrome	*NPHP4, IQCB1*	
354	Septooptic dysplasia	*HESX1*	
355	Severe combined immunodeficiency (SCID)	*IL2RG, NHEJ1, DCLRE1C, JAK3*	
356	Short-rib thoracic dysplasia with or without polydactyly	*DYNC2H1, IFT80*	
357	Shwachman-Diamond syndrome	*SBDS*	
358	Sialic acid storage disorder	*SLC17A5*	
359	Sialidosis, type I, II	*NEU1*	
360	Sickle cell disease	*HBB*	
361	Simpson-Golabi-Behmel syndrome	*OFD1, GPC3*	
362	Sjögren-Larsson syndrome	*ALDH3A2*	
363	Smith-Lemli-Opitz syndrome	*DHCR7*	
364	Smith-Magenis syndrome	*SMS*	
365	Sotos syndrome 1	*NSD1*	
366	Spastic paralysis, infantile onset ascending	*ALS2*	
367	Spastic paraplegia 2, X-linked	*PLP1*	
368	Spermatogenic failure 8	*NR5A1*	
369	Spinal muscular atrophy	*PLEKHG5, SMN1, UBA1*	
370	Spondylocostal dysostosis 1, autosomal recessive	*DLL3*	
371	Squamous cell carcinoma, burn scar-related, somatic	*FAS*	
372	Stormorken syndrome	*STIM1*	
373	Striatonigral degeneration, infantile	*NUP62*	
374	Stuve-Wiedemann syndrome	*LIFR*	
375	Succinic semialdehyde dehydrogenase deficiency	*ALDH5A1*	
376	Succinyl CoA:3-oxoacid CoA transferase deficiency	*OXCT1*	
377	Sudden infant death with dysgenesis of the testes syndrome	*TSPYL1*	
378	Sulfite oxidase deficiency	*SUOX*	
379	Systemic primary carnitine deficiency	*SLC22A5*	

380	Tay-Sachs disease	*HEXA*	
381	T-cell immunodeficiency, congenital alopecia, and nail dystrophy	*FOXN1*	
382	Tetra-amelia syndrome 1	*WNT3*	
383	Thrombo- cytopenia, X-linked	*WAS*	
384	Thrombotic thrombocytopenic purpura	*ADAMTS13*	
385	Trichothiodystrophy 3, photosensitive	*GTF2H5*	
386	Trifunctional protein deficiency	*HADHB*	
387	Tuberculosis infection, protection against Tuberculosis, susceptibility to	*IFNGR1*	
388	Tyrosinemia	*FAH, TAT, HPD*	
389	Ullrich congenital muscular dystrophy	*COL6A1, COL6A2, COL6A3*	
390	Ulna and fibula, absence of, with severe limb deficiency	*WNT7A*	
391	Usher syndrome	*GPR98, MYO7A, USH1C, USH1G, USH2A, CDH23, CLRN1*	
392	VACTERL association, X-linked	*ZIC3*	
393	VATER association with macrocephaly and ventriculomegaly	*PTEN*	
394	Very long-chain acyl-CoA dehydrogenase (VLCAD) deficiency	*ACADVL*	
395	Vitamin D-dependent rickets	*CYP27B1*	
396	Waardenburg syndrome	*EDN3, EDNRB*	
397	Waisman syndrome	*RAB39B*	
398	Warburg micro syndrome 1	*RAB3GAP1*	
399	Weyers acrofacial dysostosis	*EVC, EVC2*	
400	Wilms tumor, somatic	*GPC3*	
401	Wilson disease	*ATP7B*	
402	Wiskott-Aldrich syndrome	*WAS*	
403	Wolcott-Rallison syndrome	*EIF2AK3*	
404	Wrinkly skin syndrome	*ATP6V0A2*	
405	Xeroderma pigmentosum	*XPA, ERCC3, XPC, ERCC2, DDB2, ERCC4, ERCC5*	
406	Zellweger syndrome	*PEX1, PEX10, PEX12, PEX26, PEX13*	