Carrier Screening Invitae Comprehensive Panel

555 enfermedades analizadas.

1. 17-beta hydroxysteroid dehydrogenase 3 deficiency (HSD17B3)
2. 2-methyl-3-hydroxybutyric aciduria (HSD17B10)
3. 3-hydroxy-3-methylglutarayl-CoA lyase deficiency (HMGCL)
4. ABCA3-related conditions (ABCA3)
5. ABCA4-related conditions (ABCA4)
6. ABCB11-related conditions (ABCB11)
7. ABCC8-related conditions (ABCC8)
8. Abetalipoproteinemia (MTTP)
9. Achromatopsia (CNGB3)
10. Acrodermatitis enteropathica (SLC39A4)
11. Adenosine deaminase deficiency (ADA)
12. ADGRV1-related conditions (ADGRV1)
13. Agenesis of the corpus callosum with peripheral neuropathy (SLC12A6)
14. AHI1-related conditions (AHI1)
15. Aicardi-Goutieres syndrome 2 (RNASEH2B)
16. Aicardi-Goutieres syndrome 3 (RNASEH2C)
17. Aicardi-Goutieres syndrome 4 (RNASEH2A)
18. Aicardi-Goutieres syndrome 5 (SAMHD1)
19. AIPL1-related conditions (AIPL1)
20. Aldosterone synthase deficiency (CYP11B2)
21. ALG13-related conditions (ALG13)
22. Alpha-mannosidosis (MAN2B1)
23. Alpha-N-acetylgalactosaminidase deficiency (NAGA)
24. Alpha-thalassemia (HBA1/HBA2)
25. Alpha-thalassemia X-linked intellectual disability syndrome (ARTX)
26. Alport syndrome (COL4A3)
27. Alport syndrome (COL4A4)
28. Alport syndrome (COL4A5)
29. Alström syndrome (ALMS1)
30. Androgen insensitivity syndrome (AR)
31. Arginase deficiency (ARG1)
32. Arginine:glycine amidinotransferase deficiency (GATM)
33. Argininosuccinate lyase deficiency (ASL)
34. ARL6-related conditions (ARL6)
35. Aromatase deficiency (CYP19A1)
36. ARX-related conditions (ARX)
37. Asparagine synthetase deficiency (ASNS)
38. Aspartylglucosaminuria (AGA)
39. Ataxia with vitamin E deficiency (TTPA)
40. Ataxia-telangiectasia-like disorder (MRE11)
41. ATM-related cancers (ATM)
42. ATP7A-related conditions (ATP7A)

43. ATP8B1-related conditions (ATP8B1)
44. Atransferrinemia (TF)
45. Autoimmune polyendocrinopathy with candidiasis and ectodermal dysplasia (AIRE)
46. Autosomal recessive congenital ichthyosis (ABCA12)
47. Autosomal recessive congenital ichthyosis (TGM1)
48. Autosomal recessive spastic ataxia of Charlevoix-Saguenay (SACS)
49. AVPR2-related conditions (AVPR2)
50. Bardet-Biedl syndrome (BBS10)
51. Bardet-Biedl syndrome (BBS12)
52. Bardet-Biedl syndrome (BBS7)
53. Bardet-Biedl syndrome (BBS9)
54. Barth syndrome (TAZ)
55. Bartter syndrome type 1 (SLC12A1)
56. Bartter syndrome type 2 (KCNJ1)
57. BBS1-related conditions (BBS1)
58. BBS2-related conditions (BBS2)
59. BBS4-related conditions (BBS4)
60. BBS5-related conditions (BBS5)
61. BCS1L-related conditions (BCS1L)
62. Beta-ketothiolase deficiency (ACAT1)
63. Beta-mannosidosis (MANBA)
64. Biopterin-deficient hyperphenylalaninemia (PCBD1)
65. Biopterin-deficient hyperphenylalaninemia (PTS)
66. Biopterin-deficient hyperphenylalaninemia (QDPR)
67. Biotin-responsive basal ganglia disease (SLC19A3)
68. Bloom syndrome (BLM)
69. BRIP1-related conditions (BRIP1)
70. Brittle cornea syndrome (PRDM5)
71. Brittle cornea syndrome (ZNF469)
72. BSND-related conditions (BSND)
73. Canavan disease (ASPA)
74. Carbamoyl phosphate synthetase I deficiency (CPS1)
75. Cardioencephalomyopathy (SCO2)
76. Carnitine palmitoyltransferase I deficiency (CPT1A)
77. Carnitine palmitoyltransferase II deficiency (CPT2)
78. Carnitine-acylcarnitine translocase deficiency (SLC25A20)
79. Carpenter syndrome (RAB23)
80. Cartilage-hair hypoplasia-anauxetic dysplasia spectrum disorders (RMRP)
81. Catecholaminergic polymorphic ventricular tachycardia (CASQ2)
82. CC2D2A-related conditions (CC2D2A)
83. CDH23-related conditions (CDH23)
84. CEP290-related conditions (CEP290)
85. Cerebellar ataxia, intellectual disability, and dysequilibrium syndrome 1 (VLDLR)
86. Cerebral dysgenesis, neuropathy, ichthyosis, and keratoderma (SNAP29)
87. Cerebrotendinous xanthomatosis (CYP27A1)
88. CERKL-related conditions (CERKL)

89. CFTR-related conditions (CFTR)
90. Charcot-Marie-Tooth disease type 1X (GJB1)
91. Charcot-Marie-Tooth disease type 4D (NDRG1)
92. Chediak-Higashi syndrome (LYST)
93. Childhood-onset dystonia with optic atrophy and basal ganglia abnormalities (MECR)
94. Chorea-acanthocytosis (VPS13A)
95. Choroideremia (CHM)
96. Chronic granulomatous disease (CYBA)
97. Chronic granulomatous disease (CYBB)
98. Chronic granulomatous disease (NCF2)
99. Citrin deficiency (SLC25A13)
100. Citrullinemia type 1 (ASS1)
101. CLN3-related conditions (CLN3)
102. CLRN1-related conditions (CLRN1)
103. Cobalamin C deficiency (MMACHC)
104. Cobalamin D deficiency (MMADHC)
105. Cobalamin F deficiency (LMBRD1)
106. Cobalamin X deficiency (HCFC1)
107. Cockayne syndrome A (ERCC8)
108. Cockayne syndrome B (ERCC6)
109. Cohen syndrome (VPS13B)
110. COL11A2-related conditions (COL11A2)
111. COL17A1-related conditions (COL17A1)
112. Combined immunodeficiency due to IKBKB deficiency (IKBKB)
113. Combined malonic and methylmalonic aciduria (ACSF3)
114. Combined oxidative phosphorylation deficiency 1 (GFM1)
115. Combined oxidative phosphorylation deficiency 3 (TSFM)
116. Combined pituitary hormone deficiency (LHX3)
117. Combined pituitary hormone deficiency (POU1F1)
118. Combined pituitary hormone deficiency (PROP1)
119. Congenital adrenal hyperplasia due to 21-hydroxylase deficiency (CYP21A2)
120. Congenital adrenal hyperplasia due to 3-beta-hydroxysteroid dehydrogenase deficiency (HSD3B2)
121. Congenital adrenal insufficiency (CYP11A1)
122. Congenital amegakaryocytic thrombocytopenia (MPL)
123. Congenital chronic diarrhea (DGAT1)
124. Congenital disorder of glycosylation (SLC35A3)
125. Congenital disorder of glycosylation type Ia (PMM2)
126. Congenital disorder of glycosylation type Ib (MPI)
127. Congenital disorder of glycosylation type Ic (ALG6)
128. Congenital disorder of glycosylation type Ik (ALG1)
129. Congenital disorder of glycosylation type Iv (NGLY1)
130. Congenital dyserythropoietic anemia type II (SEC23B)
131. Congenital hydrocephalus-1 (CCDC88C)
132. Congenital hypothyroidism (TSHB)
133. Congenital insensitivity to pain with anhidrosis (NTRK1)

134. Congenital myasthenic syndrome (CHAT)
135. Congenital myasthenic syndrome (CHRNE)
136. Congenital nephrotic syndrome type 1 (NPHS1)
137. Congenital nephrotic syndrome type 2 (NPHS2)
138. Congenital secretory chloride diarrhea (SLC26A3)
139. Corneal dystrophy and perceptive deafness (SLC4A11)
140. CRB1-related conditions (CRB1)
141. CTSC-related conditions (CTSC)
142. CYP17A1-related conditions (CYP17A1)
143. CYP1B1-related conditions (CYP1B1)
144. CYP7B1-related conditions (CYP7B1)
145. Cystinosis (CTNS)
146. Cytochrome P450 oxidoreductase deficiency (POR)
147. Desbuquois dysplasia type 1 (CANT1)
148. Developmental and epileptic encephalopathy (CAD)
149. DGUOK-related conditions (DGUOK)
150. DHDDS-related conditions (DHDDS)
151. Dihydrolipoamide dehydrogenase deficiency (DLD)
152. Distal renal tubular acidosis with deafness (ATP6V1B1)
153. DMD-related conditions (DMD)
154. DOK7-related conditions (DOK7)
155. Donnai-Barrow syndrome (LRP2)
156. Dubin-Johnson syndrome (ABCC2)
157. DUOX2-related conditions (DUOX2)
158. DYNC2H1-related conditions (DYNC2H1)
159. DYSF-related conditions (DYSF)
160. Dyskeratosis congenita spectrum disorders (DKC1)
161. Dyskeratosis congenita spectrum disorders (RTEL1)
162. Dyskeratosis congenita spectrum disorders (TERT)
163. Dystrophic epidermolysis bullosa (COL7A1)
164. EDA-related conditions (EDA)
165. Ehlers-Danlos syndrome, dermatosparaxis type (ADAMTS2)
166. Ehlers-Danlos syndrome, kyphoscoliotic type (PLOD1)
167. Ellis-van Creveld syndrome (EVC)
168. Emery-Dreifuss muscular dystrophy (EMD)
169. Epimerase deficiency galactosemia (GALE)
170. ERCC2-related conditions (ERCC2)
171. Ethylmalonic encephalopathy (ETHE1)
172. EVC2-related conditions (EVC2)
173. Fabry disease (GLA)
174. Factor IX deficiency (hemophilia B) (F9)
175. Familial chylomicronemia syndrome (LPL)
176. Familial dysautonomia (ELP1)
177. Familial hemophagocytic lymphohistiocytosis type 2 (PRF1)
178. Familial hemophagocytic lymphohistiocytosis type 3 (UNC13D)
179. Familial hemophagocytic lymphohistiocytosis type 4 (STX11)

180. Familial hemophagocytic lymphohistiocytosis type 5 (STXBP2)
181. Familial hyperaldosteronism type I (CYP11B1)
182. Familial hypercholesterolemia (LDLR)
183. Familial hypercholesterolemia (LDLRAP1)
184. Fanconi anemia type A (FANCA)
185. Fanconi anemia type B (FANCB)
186. Fanconi anemia type C (FANCC)
187. Fanconi anemia type D2 (FANCD2)
188. Fanconi anemia type E (FANCE)
189. Fanconi anemia type G (FANCG)
190. Fanconi anemia type I (FANCI)
191. Fanconi anemia type L (FANCL)
192. FHL1-related conditions (FHL1)
193. FKBP10-related conditions (FKBP10)
194. FMR1-related conditions including fragile X syndrome (FMR1)
195. Foveal hypoplasia (SLC38A8)
196. Fraser syndrome (FRAS1)
197. Fraser syndrome (FREM2)
198. Fraser syndrome (GRIP1)
199. Fructose-1,6-bisphosphatase deficiency (FBP1)
200. Fucosidosis (FUCA1)
201. Galactokinase deficiency galactosemia (GALK1)
202. Galactosemia (GALT)
203. Galactosialidosis (CTSA)
204. GBA-related conditions (GBA)
205. GBE1-related conditions (GBE1)
206. GCH1-related conditions (GCH1)
207. GDF5-related conditions (GDF5)
208. Geme oxygenase 1 deficiency (HMOX1)
209. Gemolytic anemia, CD59-mediated (CD59)
210. Gereditary hemochromatosis type 2 (HAMP)
211. Gereditary hemochromatosis type 2 (HJV)
212. Gereditary hemochromatosis type 3 (TFR2)
213. Gereditary leiomyomatosis and renal cell cancer (FH)
214. Geroderma osteodysplastica (GORAB)
215. GHR-related conditions (GHR)
216. Gitelman syndrome (SLC12A3)
217. GJB2-related conditions (GJB2)
218. GLB1-related conditions (GLB1)
219. GLE1-related conditions (GLE1)
220. Glutaric acidemia type I (GCDH)
221. Glutaric acidemia type IIA (ETFA)
222. Glutaric acidemia type IIB (ETFB)
223. Glutaric acidemia type IIC (ETFDH)
224. Glutathione synthetase deficiency (GSS)
225. Glycine encephalopathy (AMT)

226. Glycine encephalopathy (GLDC)
227. Glycogen storage disease type Ia (G6PC)
228. Glycogen storage disease type II (Pompe disease) (GAA)
229. Glycogen storage disease type III (AGL)
230. Glycogen storage disease type IXb (PHKB)
231. Glycogen storage disease type IXc (PHKG2)
232. Glycogen storage disease type V (PYGM)
233. Glycogen storage disease type VII (PFKM)
234. GM3 synthase deficiency (ST3GAL5)
235. GNE-related conditions (GNE)
236. GNPTAB-related conditions (GNPTAB)
237. Golocarboxylase synthetase deficiency (HLCS)
238. Guanidinoacetate methyltransferase deficiency (GAMT)
239. GUCY2D-related conditions (GUCY2D)
240. Gyrate atrophy of the choroid and retina (OAT)
241. HADHA-related conditions (HADHA)
242. HBB-related hemoglobinopathies (HBB)
243. Hereditary fructose intolerance (ALDOB)
244. Hermansky-Pudlak syndrome type 1 (HPS1)
245. Hermansky-Pudlak syndrome type 3 (HPS3)
246. Hermansky-Pudlak syndrome type 4 (HPS4)
247. Hermansky-Pudlak syndrome type 5 (HPS5)
248. Hermansky-Pudlak syndrome type 6 (HPS6)
249. Hermansky-Pudlak syndrome type 8 (BLOC1S3)
250. Hermansky-Pudlak syndrome type 9 (BLOC1S6)
251. HGSNAT-related conditions (HGSNAT)
252. Homocystinuria due to cobalamin E deficiency (MTRR)
253. Homocystinuria due to cobalamin G deficiency (MTR)
254. Homocystinuria due to cystathionine beta-synthase deficiency (CBS)
255. Homocystinuria due to MTHFR deficiency (MTHFR)
256. HPRT1-related conditions (HPRT1)
257. HSD17B4-related conditions (HSD17B4)
258. Hydrolethalus syndrome type 1 (HYLS1)
259. Hyper-IgM immunodeficiency (CD40)
260. Hyperornithinemia-hyperammonemia-homocitrullinuria syndrome (SLC25A15)
261. Hyperphosphatemic familial tumoral calcinosis (GALNT3)
262. Hypomyelinating leukodystrophy-12 (VPS11)
263. Hypophosphatasia (ALPL)
264. Ichthyosis prematurity syndrome (SLC27A4)
265. IGHMBP2-related conditions (IGHMBP2)
266. Imerslund-Gräsbeck syndrome (AMN)
267. Immunodeficiency-centromeric instability-facial anomalies syndrome 1 (DNMT3B)
268. Immunodeficiency-centromeric instability-facial anomalies syndrome 2 (ZBTB24)
269. Isolated ectopia lentis (ADAMTSL4)
270. Isovaleric acidemia (IVD)
271. ITGB3-related conditions (ITGB3)

272. Johanson-Blizzard syndrome (UBR1)
273. Joubert syndrome and related disorders (MKS1)
274. Joubert syndrome and related disorders (RPGRIP1L)
275. Joubert syndrome and related disorders (TMEM216)
276. Junctional epidermolysis bullosa (LAMC2)
277. Junctional epidermolysis bullosa with pyloric atresia (ITGA6)
278. Junctional epidermolysis bullosa with pyloric atresia (ITGB4)
279. KCNJ11-related conditions (KCNJ11)
280. Krabbe disease (GALC)
281. L1 syndrome (L1CAM)
282. LAMA2-related muscular dystrophy (LAMA2)
283. LAMA3-related conditions (LAMA3)
284. LAMB3-related conditions (LAMB3)
285. Leber congenital amaurosis 5 (LCA5)
286. Leukoencephalopathy with vanishing white matter (EIF2B1)
287. Leukoencephalopathy with vanishing white matter (EIF2B2)
288. Leukoencephalopathy with vanishing white matter (EIF2B3)
289. Leukoencephalopathy with vanishing white matter (EIF2B4)
290. Leukoencephalopathy with vanishing white matter (EIF2B5)
291. LIG4 syndrome (LIG4)
292. Limb-girdle muscular dystrophy (CAPN3)
293. Limb-girdle muscular dystrophy type 2 (SGCD)
294. Limb-girdle muscular dystrophy type 2C (SGCG)
295. Limb-girdle muscular dystrophy type 2D (SGCA)
296. Limb-girdle muscular dystrophy type 2E (SGCB)
297. Lipoid congenital adrenal hyperplasia (STAR)
298. LRAT-related conditions (LRAT)
299. Lysinuric protein intolerance (SLC7A7)
300. Lysosomal acid lipase deficiency (LIPA(
301. Major histocompatibility complex class II deficiency (CIITA)
302. Malonyl-CoA decarboxylase deficiency (MLYCD)
303. Maple syrup urine disease type 1A (BCKDHA)
304. Maple syrup urine disease type 1B (BCKDHB)
305. Maple syrup urine disease type 2 (DBT)
306. MECP2-related conditions (MECP2)
307. Medium-chain acyl-CoA dehydrogenase deficiency (ACADM)
308. Medium/short-chain 3-hydroxyacyl-CoA dehydrogenase deficiency (HADH)
309. MEDNIK syndrome (AP1S1)
310. Megalencephalic leukoencephalopathy with subcortical cysts 1 (MLC1)
311. Metabolic crises with rhabdomyolysis, cardiac arrhythmias and neurodegeneration (TANGO2)
312. Metachromatic leukodystrophy (ARSA)
313. Methylmalonic acidemia (MCEE)
314. Methylmalonic acidemia (MMAA)
315. Methylmalonic acidemia (MMAB)
316. Methylmalonic acidemia (MUT)

317. MFSD8-related conditions (MFSD8)
318. Microcephalic osteodysplastic primordial dwarfism type II (PCNT)
319. Microcephaly, postnatal progressive, with seizures and brain atrophy (MED17)
320. Mitochondrial complex I deficiency 1 (NDUFS4)
321. Mitochondrial complex I deficiency 10 (NDUFAF2)
322. Mitochondrial complex I deficiency 16 (NDUFAF5)
323. Mitochondrial complex I deficiency 19 (FOXRED1)
324. Mitochondrial complex I deficiency 20/ACAD9 deficiency (ACAD9)
325. Mitochondrial complex I deficiency 3 (NDUFS7)
326. Mitochondrial complex I deficiency 4 (NDUFV1)
327. Mitochondrial complex I deficiency 9 (NDUFS6)
328. Mitochondrial complex IV deficiency / Leigh syndrome, French Canadian type (LRPPRC)
329. Mitochondrial complex IV deficiency 12 (PET100)
330. Mitochondrial complex IV deficiency 6 (COX15)
331. Mitochondrial DNA depletion syndrome-2 (TK2)
332. Mitochondrial neurogastrointestinal encephalomyopathy (TYMP)
333. Mitochondrial trifunctional protein deficiency (HADHB)
334. MKKS-related conditions (MKKS)
335. Molybdenum cofactor deficiency (MOCS1)
336. Molybdenum cofactor deficiency (MOCS2)
337. MPV17-related conditions (MPV17)
338. Mucolipidosis type III gamma (GNPTG)
339. Mucolipidosis type IV (MCOLN1)
340. Mucopolysaccharidosis type I (IDUA)
341. Mucopolysaccharidosis type II (IDS)
342. Mucopolysaccharidosis type IIIA (SGSH)
343. Mucopolysaccharidosis type IIIB (NAGLU)
344. Mucopolysaccharidosis type IIID (GNS)
345. Mucopolysaccharidosis type IVA (GALNS)
346. Mucopolysaccharidosis type IX (HYAL1)
347. Mucopolysaccharidosis type VI (ARSB)
348. Mucopolysaccharidosis type VII (GUSB)
349. Mulibrey nanism (TRIM37)
350. Multiple pterygium syndrome (CHRNG)
351. Multiple sulfatase deficiency (SUMF1)
352. Muscular dystrophy-dystroglycanopathy (FKRP)
353. Muscular dystrophy-dystroglycanopathy (FKTN)
354. Muscular dystrophy-dystroglycanopathy (LARGE1)
355. Muscular dystrophy-dystroglycanopathy (POMT1)
356. Muscular dystrophy-dystroglycanopathy (POMT2)
357. Muscular dystrophy-dystroglycanopathy (RXYLT1)
358. MUSK-related conditions (MUSK)
359. MVK-related conditions (MVK)
360. Myopathy, lactic acidosis, and sideroblastic anemia 1 (PUS1)
361. Myotonia congenita (CLCN1)
362. N-acetylglutamate synthase deficiency (NAGS)

363. NBN-related cancers (NBN)
364. Nemaline myopathy 2 (NEM)
365. Nephrogenic diabetes insipidus (AQP2)
366. Nephronophthisis (INVS)
367. Nephronophthisis (NPHP1)
368. Neuronal ceroid lipofuscinosis type 1 (PPT1)
369. Neuronal ceroid lipofuscinosis type 10 (CTSD)
370. Neuronal ceroid lipofuscinosis type 2 (TPP1)
371. Neuronal ceroid lipofuscinosis type 5 (CLN5)
372. Neuronal ceroid lipofuscinosis type 6 (CLN6)
373. Neuronal ceroid lipofuscinosis type 8 (CLN8)
374. Niemann-Pick disease type C (NPC1)
375. Niemann-Pick disease type C (NPC2)
376. Niemann-Pick disease types A and B (SMPD1)
377. Nonsyndromic deafness (LOXHD1)
378. Nonsyndromic deafness (MYO15A)
379. Nonsyndromic deafness (MYO15A)
380. Nonsyndromic deafness (OTOA)
381. Nonsyndromic deafness (SYNE4)
382. Nonsyndromic deafness (TMC1)
383. Nonsyndromic deafness (TMPRSS3)
384. Nonsyndromic intellectual disability (CC2D1A)
385. NR0B1-related conditions (NR0B1)
386. NR2E3-related conditions (NR2E3)
387. NSMCE3 deficiency (NSMCE3)
388. OCRL-related conditions (OCRL)
389. Oculocutaneous albinism type 2 OCA2)
390. Oculocutaneous albinism type 3 (TYRP1)
391. Oculocutaneous albinism type 4 (SLC45A2)
392. Oculocutaneous albinism types 1A and 1B (TYR)
393. OPA3-related conditions (OPA3)
394. Opitz GBBB syndrome (MID1)
395. Ornithine transcarbamylase deficiency (OTC)
396. Osteogenesis imperfecta (BMP1)
397. Osteogenesis imperfecta (CRTAP)
398. Osteogenesis imperfecta (P3H1)
399. Osteopetrosis (TCIRG1)
400. OSTM1 deficiency associated osteopetrosis (OSTM1)
401. OTOF-related conditions (OTOF)
402. Pantothenate kinase-associated neurodegeneration (PANK2)
403. Parkinson disease 15 (FBXO7)
404. PCDH15-related conditions (PCDH15)
405. Peroxisomal acyl-CoA oxidase deficiency (ACOX1)
406. PEX5-related conditions (PEX5)
407. PEX7-related conditions (PEX7)
408. PGM3-congenital disorder of glycosylation (PGM3)

409. Phenylalanine hydroxylase deficiency (PAH)
410. Phosphoglycerate dehydrogenase deficiency (PHGDH)
411. PIGN-congenital disorder of glycosylation (PIGN)
412. PJVK-related conditions (DFNB59 aka PJVK)
413. PLA2G6-related conditions (PLA2G6)
414. PLEKHG5-related conditions (PLEKHG5)
415. PLP1-related conditions (PLP1)
416. POLG-related conditions (POLG)
417. Polycystic kidney disease (PKHD1)
418. Polymicrogyria (ADGRG1)
419. POMGNT1-related conditions (POMGNT1)
420. Pontocerebellar hypoplasia (TSEN54)
421. Pontocerebellar hypoplasia type 1B (EXOSC3)
422. Pontocerebellar hypoplasia type 2D (SEPSECS)
423. Pontocerebellar hypoplasia type 2E (VPS53)
424. Pontocerebellar hypoplasia type 6 (RARS2)
425. Primary carnitine deficiency (SLC22A5)
426. Primary ciliary dyskinesia (CCDC103)
427. Primary ciliary dyskinesia (CCDC39)
428. Primary ciliary dyskinesia (DNAH11)
429. Primary ciliary dyskinesia (DNAH5)
430. Primary ciliary dyskinesia (DNAI1)
431. Primary ciliary dyskinesia (DNAI2)
432. Primary hyperoxaluria type 1 (AGXT)
433. Primary hyperoxaluria type 2 (GRHPR)
434. Primary hyperoxaluria type 3 (HOGA1)
435. Primary microcephaly (MCPH1)
436. Progressive early-onset encepahlopathy with brain atrophy and thin corpus callosum (PEBAT) (TBCD)
437. Progressive familial intrahepatic cholestasis 3 (ABCB4)
438. Progressive pseudorheumatoid dysplasia (WISP3)
439. Prolidase deficiency (PEPD)
440. Propionic acidemia (PCCA)
441. Propionic acidemia (PCCB)
442. PRPS1-related conditions (PRPS1)
443. PSAP-related conditions (PSAP)
444. Pycnodysostosis (CTSK)
445. Pyridoxal 5′-phosphate-dependent epilepsy (PNPO)
446. Pyridoxine-dependent epilepsy (ALDH7A1)
447. Pyruvate carboxylase deficiency (PC)
448. Pyruvate dehydrogenase complex deficiency (PDHA1)
449. Pyruvate dehydrogenase complex deficiency (PDHB)
450. RAPSN-related conditions (RAPSN)
451. RDH12-related conditions (RDH12)
452. Refsum disease (PHYH)
453. Retinitis pigmentosa 2 (RP2)

454. Retinitis pigmentosa 25 (EYS)
455. Retinitis pigmentosa 28 (FAM161A)
456. Retinitis pigmentosa 36 (GNPAT)
457. Retinitis pigmentosa 62 (MAK)
458. Rhizomelic chondrodysplasia punctata type 2 (GNPAT)
459. Rhizomelic chondrodysplasia punctata type 3 (AGPS)
460. RLBP1-related conditions (RLBP1)
461. Roberts syndrome (ESCO2)
462. RPE65-related conditions (RPE65)
463. RYR1-related conditions (RYR1)
464. SAMD9-related conditions (SAMD9)
465. Sandhoff disease (HEXB)
466. Schimke immuno-osseous dysplasia (SMARCAL1)
467. Seckel syndrome (CEP152)
468. Sepiapterin reductase deficiency (SPR)
469. Severe combined immunodeficiency due to CD3-delta deficiency (CD3D)
470. Severe combined immunodeficiency due to CD3-epsilon deficiency (CD3E)
471. Severe combined immunodeficiency due to CD45 deficiency (PTPRC)
472. Severe combined immunodeficiency due to DCLRE1C (Artemis) deficiency (DCLRE1C)
473. Severe combined immunodeficiency due to FOXN1 deficiency (FOXN1)
474. Severe combined immunodeficiency due to IL7R-alpha deficiency (IL7R)
475. Severe combined immunodeficiency due to JAK3 deficiency (JAK3)
476. Severe combined immunodeficiency due to RAG1 deficiency (RAG1)
477. Severe combined immunodeficiency due to RAG2 deficiency (RAG2)
478. Severe congenital neutropenia due to G6PC3 deficiency (G6PC3)
479. Severe congenital neutropenia due to HAX1 deficiency (HAX1)
480. Severe congenital neutropenia type 5 (VPS45)
481. Sialic acid storage diseases (SLC17A5)
482. Sialidosis (NEU1)
483. Sjögren-Larsson syndrome (ALDH3A2)
484. SLC26A2-related conditions (SLC26A2)
485. SLC26A4-related conditions (SLC26A4)
486. SLC37A4-related conditions (SLC37A4)
487. Smith-Lemli-Opitz syndrome (DHCR7)
488. Spastic paraplegia type 15 (ZFYVE26)
489. Spastic paraplegia type 49 (TECPR2)
490. Spastic tetraplegia, thin corpus callosum, and progressive microcephaly (SLC1A4)
491. SPG11-related conditions (SPG11)
492. Spinal muscular atrophy*
493. Spinocerebellar ataxia (ANO10)
494. Spondylocostal dysostosis (DLL3)
495. Spondylocostal dysostosis (MESP2)
496. Steel syndrome (COL27A1)
497. Steroid 5-alpha-reductase deficiency (SRD5A2)
498. Stüve-Wiedemann syndrome (LIFR)
499. Sulfite oxidase deficiency (SUOX)

500. SURF1-related conditions (SURF1)
501. Tay-Sachs disease (HEXA)
502. TBCE-related conditions (TBCE)
503. Thiamine-responsive megaloblastic anemia (SLC19A2)
504. Thyroid dyshormonogenesis (SLC5A5)
505. Thyroid dyshormonogenesis (TG)
506. Thyroid dyshormonogenesis (TPO)
507. TMEM67-related conditions (TMEM67)
508. Transcobalamin II deficiency (TCN2)
509. Transient infantile liver failure (TRMU)
510. TREX1-related conditions (TREX1)
511. Trichohepatoenteric syndrome (SKIV2L)
512. Trichohepatoenteric syndrome (TTC37)
513. TRIM32-related conditions (TRIM32)
514. Trimethylaminuria (FMO3)
515. Triple A syndrome (AAAS)
516. TSHR-related conditions (TSHR)
517. TULP1-related conditions (TULP1)
518. Tyrosine hydroxylase deficiency (TH)
519. Tyrosinemia type I (FAH)
520. Tyrosinemia type II (TAT)
521. Tyrosinemia type III (HPD)
522. USH1C-related conditions (USH1C)
523. USH2A-related conditions (USH2A)
524. Very long-chain acyl-CoA dehydrogenase deficiency (ACADVL)
525. Vici syndrome (EPG5)
526. Vitamin D-dependent rickets type 1A (CYP27B1)
527. Vitamin D-dependent rickets type 2A (VDR)
528. VRK1-related conditions (VRK1)
529. VSX2-related conditions (VSX2)
530. Warsaw syndrome (DDX11)
531. WAS-related conditions (WAS)
532. Werner syndrome (WRN)
533. Wilson disease (ATP7B)
534. WNT10A-related conditions (WNT10A)
535. Wolcott-Rallison syndrome (EIF2AK3)
536. Woodhouse-Sakati syndrome (DCAF17)
537. X-linked adrenoleukodystrophy (ABCD1)
538. X-linked agammaglobulinemia (BTK)
539. X-linked chondrodysplasia punctata type 1 (ARSE)
540. X-linked creatine transporter deficiency (SLC6A8)
541. X-linked hyper-IgM immunodeficiency (CD40LG)
542. X-linked juvenile retinoschisis (RS1)
543. X-linked myotubular myopathy (MTM1)
544. X-linked severe combined immunodeficiency (IL2RG)
545. Xeroderma pigmentosum complementation group A (XPA)

546. Xeroderma pigmentosum complementation group C (XPC)
547. Xeroderma pigmentosum, variant type (POLH)
548. Zellweger spectrum disorder (PEX1)
549. Zellweger spectrum disorder (PEX10)
550. Zellweger spectrum disorder (PEX12)
551. Zellweger spectrum disorder (PEX13)
552. Zellweger spectrum disorder (PEX16)
553. Zellweger spectrum disorder (PEX2)
554. Zellweger spectrum disorder (PEX26)
555. Zellweger spectrum disorder (PEX6)