- P -Point mutation, or any insertion/deletion entirely inside one gene
- D -Deletion of a gene or genes
- C - Whole chromosome extra, missing, or both (see chromosomal aberrations)
- T -Trinucleotide repeat disorders: gene is extended in length

common disorders

SNo	Disorder	Mutation	Chromosome
1	22q11.2 deletion syndrome	D	22q
2	Angelman syndrome	DCP	15
3	Canavan disease		17p
4	Charcot–Marie–Tooth disease		
5	Color blindness	P	X
6	Cri du chat	D	5
7	Cystic fibrosis	P	7q
8	Down syndrome	C	21
9	Duchenne muscular dystrophy	D	Xp
10	Haemochromatosis	P	6
11	Haemophilia	P	X
12	Klinefelter syndrome	C	X
13	Neurofibromatosis		17q/22q/?
14	Phenylketonuria	P	12q
15	Polycystic kidney disease	P	16 (PKD1) or 4 (PKD2)
16	Prader–Willi syndrome	DC	15
17	Sickle-cell disease	P	11p
18	Tay–Sachs disease	P	15
19	Turner syndrome	C	X

Full list

SNo	Disorder name	Mutation type	Chromosome
1	1p36 deletion syndrome	D	1p36
2	18p deletion syndrome	D	18p
3	21-hydroxylase deficiency		6p21.3
4	45,X Turner syndrome	C	X
5	47,XX,+21 Down syndrome	C	21
6	47,XXX triple X syndrome	C	X
7	47,XXY	C	X

	Klinefelter syndrome		
8	47,XY,+21 Down syndrome	C	21
9	47,XYY syndrome	C	Y
10	5-ALA dehydratase-deficient porphyria ALA dehydratase deficiency		
11	5-aminolaevulinic dehydratase deficiency porphyria ALA dehydratase deficiency		
12	5p deletion syndrome Cri du chat	D	5p
13	5p- syndrome Cri du chat	D	5p
14	A-T ataxia telangiectasia		
15	AAT alpha 1-antitrypsin deficiency		
16	Absence of vas deferens congenital absence of the vas deferens		
17	Absent vasa congenital absence of the vas deferens		
18	aceruloplasminemia		
19	ACG2 achondrogenesis type II		
20	ACH achondroplasia		
21	Achondrogenesis type II		
22	achondroplasia	substitution	4p16.3
23	Acid beta-glucosidase deficiency Gaucher disease type 1		
24	Acrocephalosyndactyly (Apert) Apert syndrome		
25	acrocephalosyndactyly, type V Pfeiffer syndrome		
26	Acrocephaly Apert syndrome		
27	Acute cerebral Gaucher's disease Gaucher disease type 2		
28	acute intermittent porphyria		
29	ACY2 deficiency Canavan disease		
30	AD Alzheimer's disease		
31	Adelaide-type craniosynostosis Muenke syndrome		
32	Adenomatous Polyposis Coli		

	familial adenomatous polyposis		
33	Adenomatous Polyposis of the Colon familial adenomatous polyposis		
34	ADP ALA dehydratase deficiency		
35	adenylosuccinate lyase deficiency		
36	Adrenal gland disorders 21-hydroxylase deficiency		
37	Adrenogenital syndrome 21-hydroxylase deficiency		
38	Adrenoleukodystrophy		
39	AIP acute intermittent porphyria		
40	AIS androgen insensitivity syndrome		
41	AKU alkaptonuria		
42	ALA dehydratase porphyria ALA dehydratase deficiency		
43	ALA-D porphyria ALA dehydratase deficiency		
44	ALA dehydratase deficiency		
45	Albinism		
46	Alcaptonuria alkaptonuria		
47	Alexander disease		
48	alkaptonuria		
49	Alkaptonuric ochronosis alkaptonuria		
50	alpha 1-antitrypsin deficiency		
51	alpha-1 proteinase inhibitor alpha 1-antitrypsin deficiency		14q32.1
52	alpha-1 related emphysema alpha 1-antitrypsin deficiency		14q32.1
53	Alpha-galactosidase A deficiency Fabry disease	P	Xq22.1
54	ALS amyotrophic lateral sclerosis		
55	Alström syndrome		
56	ALX Alexander disease		
57	Alzheimer's disease		
58	Amelogenesis imperfecta		
59	Amino levulinic acid dehydratase deficiency ALA dehydratase deficiency		

60	Aminoacylase 2 deficiency Canavan disease		
61	amyotrophic lateral sclerosis		
62	Anderson-Fabry disease Fabry disease	P	Xq22.1
63	androgen insensitivity syndrome		
64	Anemia		
65	Anemia, hereditary sideroblastic X-linked sideroblastic anemia		X
66	Anemia, sex-linked hypochromic sideroblastic X-linked sideroblastic anemia		X
67	Anemia, splenic, familial Gaucher disease		
68	Angelman syndrome		
69	Angiokeratoma Corporis Diffusum Fabry disease	P	Xq22.1
70	Angiokeratoma diffuse Fabry disease		
71	Angiomatosis retinae von Hippel–Lindau disease		
72	ANH1 X-linked sideroblastic anemia		X
73	APC resistance, Leiden type factor V Leiden thrombophilia		
74	Apert syndrome		
75	AR deficiency androgen insensitivity syndrome		
76	AR-CMT2 Charcot-Marie-Tooth disease, type 2		
77	Arachnodactyly Marfan syndrome		
78	ARNSHL Nonsyndromic deafness#autosomal recessive		
79	Arthro-ophthalmopathy, hereditary progressive Stickler syndrome#COL2A1		
80	Arthrochalasis multiplex congenita Ehlers–Danlos syndrome#arthrochalasia type		
81	AS Angelman syndrome		
82	Asp deficiency Canavan disease		
83	Aspa deficiency Canavan disease		
84	Aspartoacylase deficiency Canavan disease		

85	ataxia telangiectasia		
86	Autism-Dementia-Ataxia-Loss of Purposeful Hand Use syndrome Rett syndrome		
87	autosomal dominant juvenile ALS amyotrophic lateral sclerosis, type 4		
88	Autosomal dominant opitz G/BBB syndrome 22q11.2 deletion syndrome	D	22q
89	autosomal recessive form of juvenile ALS type 3 Amyotrophic lateral sclerosis#type 2		
90	Autosomal recessive nonsyndromic hearing loss Nonsyndromic deafness#autosomal recessive		
91	Autosomal Recessive Sensorineural Hearing Impairment and Goiter Pendred syndrome		
92	AxD Alexander disease		
93	Ayerza syndrome primary pulmonary hypertension		
94	B variant of the Hexosaminidase GM2 gangliosidosis Sandhoff disease		
95	BANF neurofibromatosis type II		
96	Beare-Stevenson cutis gyrata syndrome		10q26
97	Benign paroxysmal peritonitis Mediterranean fever, familial		
98	Benjamin syndrome		
99	beta-thalassemia		
100	BH4 Deficiency tetrahydrobiopterin deficiency		
101	Bilateral Acoustic Neurofibromatosis neurofibromatosis type II		
102	biotinidase deficiency		
103	bladder cancer		
104	Bleeding disorders factor V Leiden thrombophilia		
105	Bloch-Sulzberger syndrome incontinentia pigmenti		
106	Bloom syndrome		15q26.1
107	Bone diseases		
108	Bone marrow diseases X-linked sideroblastic anemia		
109	Bonnevie-Ullrich syndrome Turner syndrome		
110	Bourneville disease		

	tuberous sclerosis		
111	Bourneville phakomatosis tuberous sclerosis		
112	Brain diseases prion disease		
113	breast cancer		
114	Birt–Hogg–Dubé syndrome		17
115	Brittle bone disease osteogenesis imperfecta		
116	Broad Thumb-Hallux syndrome Rubinstein-Taybi syndrome		
117	Bronze Diabetes hemochromatosis		
118	Bronzed cirrhosis hemochromatosis		
119	Bulbospinal muscular atrophy, X-linked Kennedy's disease		
120	Burger-Grutz syndrome lipoprotein lipase deficiency, familial		
121	CADASIL syndrome	P	3
122	CGD Chronic granulomatous disorder		
123	Campomelic dysplasia	C	17q24.3-q25.1
124	Canavan disease		
125	Cancer		
126	Cancer Family syndrome hereditary nonpolyposis colorectal cancer		
127	Cancer of breast breast cancer		
128	Cancer of the bladder bladder cancer		
129	Carboxylase Deficiency, Multiple, Late-Onset biotinidase deficiency	P	3
130	Cardiomyopathy Noonan syndrome		
131	Cat cry syndrome Cri du chat		
132	CAVD congenital absence of the vas deferens		
133	Caylor cardiofacial syndrome 22q11.2 deletion syndrome	D	22q
134	CBAVD congenital absence of the vas deferens		
135	CEP congenital erythropoietic porphyria		
	Ceramide trihexosidase deficiency		

136	Fabry disease		X
137	Cerebelloretinal Angiomatosis, familial von Hippel-Lindau disease	P	3 (p26-p25)
138	Cerebral arteriopathy with subcortical infarcts and leukoencephalopathy CADASIL syndrome	P	3
139	Cerebral autosomal dominant ateriopathy with subcortical infarcts and leukoencephalopathy CADASIL syndrome	P	3
140	Cerebral sclerosis tuberous sclerosis		9 (q34), 16 (p13.3)
141	Cerebroatrophic Hyperammonemia Rett syndrome		X
142	Cerebroside Lipidosis syndrome Gaucher disease	P	1(q21)
143	CF cystic fibrosis	D (most common); or substitution	CFTR (7q31.2)
144	CH congenital hypothyroidism		
145	Charcot disease amyotrophic lateral sclerosis		
146	Charcot-Marie-Tooth disease		
147	Chondrodystrophia achondroplasia		
148	Chondrodystrophy syndrome achondroplasia		
149	Chondrodystrophy with sensorineural deafness otospondylomegaepiphyseal dysplasia		
150	Chondrogenesis imperfecta achondrogenesis, type II		
151	Choreoathetosis self-mutilation hyperuricemia syndrome Lesch-Nyhan syndrome	P	X
152	Classic Galactosemia galactosemia	P	9 (p13)
153	Classical Ehlers–Danlos syndrome Ehlers–Danlos syndrome#classical type		
154	Classical Phenylketonuria phenylketonuria		
155	Cleft lip and palate Stickler syndrome		
156	Cloverleaf skull with thanatophoric dwarfism Thanatophoric dysplasia#type 2		
157	CLS Coffin-Lowry syndrome		
158	CMT Charcot-Marie-Tooth disease		

159	Cockayne syndrome		
160	Coffin-Lowry syndrome		
161	collagenopathy, types II and XI		
162	Colon Cancer, familial Nonpolyposis hereditary nonpolyposis colorectal cancer		
163	Colon cancer, familial familial adenomatous polyposis		
164	Colorectal cancer		
165	Complete HPRT deficiency Lesch-Nyhan syndrome		
166	Complete hypoxanthine-guanine phosphoribosyltransferase deficiency Lesch-Nyhan syndrome		
167	Compression neuropathy hereditary neuropathy with liability to pressure palsies		
168	Congenital adrenal hyperplasia 21-hydroxylase deficiency		
169	congenital bilateral absence of vas deferens Congenital absence of the vas deferens		
170	Congenital erythropoietic porphyria		
171	Congenital heart disease		
172	Congenital hypomyelination Charcot-Marie-Tooth disease#Type 1 Charcot-Marie-Tooth disease#Type 4		
173	Congenital hypothyroidism		
174	Congenital methemoglobinemia Methemoglobinemia#Congenital methaemoglobinaemia		
175	Congenital osteosclerosis achondroplasia		
176	Congenital sideroblastic anaemia X-linked sideroblastic anemia		X
177	Connective tissue disease		
178	Conotruncal anomaly face syndrome 22q11.2 deletion syndrome	D	22q
179	Cooley's Anemia beta-thalassemia		
180	Copper storage disease Wilson's disease		13 (q14.3)
181	Copper transport disease Menkes disease		
182	Coproporphyria, hereditary hereditary coproporphyria		
183	Coproporphyrinogen oxidase deficiency hereditary coproporphyria		
184	Cowden syndrome		

185	CPO deficiency hereditary coproporphyria		
186	CPRO deficiency hereditary coproporphyria		
187	CPX deficiency hereditary coproporphyria		
188	Craniofacial dysarthrosis Crouzon syndrome		
189	Craniofacial Dysostosis Crouzon syndrome		
190	Cretinism congenital hypothyroidism		
191	Creutzfeldt-Jakob disease prion disease		
192	Cri du chat	D	5p
193	Crohn's disease, fibrostenosing	P	16q12
194	Crouzon syndrome		FGFR2 (10q25.3-q26)
195	Crouzon syndrome with acanthosis nigricans Crouzonodermoskeletal syndrome		
196	Crouzonodermoskeletal syndrome		
197	CS Cockayne syndrome Cowden syndrome		
198	Curschmann-Batten-Steinert syndrome myotonic dystrophy		
199	cutis gyrata syndrome of Beare-Stevenson Beare-Stevenson cutis gyrata syndrome		
200	D-glycerate dehydrogenase deficiency hyperoxaluria, primary		
201	Dappled metaphysis syndrome spondyloepimetaphyseal dysplasia, Strudwick type		
202	DAT - Dementia Alzheimer's type Alzheimer's disease		
203	Genetic hypercalciuria Dent's disease		Xp11.22
204	DBMD muscular dystrophy, Duchenne and Becker types		
205	Deafness with goiter Pendred syndrome		
206	Deafness-retinitis pigmentosa syndrome Usher syndrome		
207	Deficiency disease, Phenylalanine Hydroxylase phenylketonuria	P	12q
208	Degenerative nerve diseases		
	de Grouchy syndrome 1		

209	De Grouchy syndrome	D	18p
210	Dejerine-Sottas syndrome Charcot-Marie-Tooth disease		
211	Delta-aminolevulinate dehydratase deficiency porphyria ALA dehydratase deficiency		
212	Dementia CADASIL syndrome		
213	demyelinogenic leukodystrophy Alexander disease		
214	Dermatosparactic type of Ehlers–Danlos syndrome Ehlers–Danlos syndrome#dermatosparaxis type		
215	Dermatosparaxis Ehlers–Danlos syndrome#dermatosparaxis type		
216	developmental disabilities		
217	dHMN Amyotrophic lateral sclerosis#type 4		
218	DHMN-V distal spinal muscular atrophy, type V		
219	DHTR deficiency androgen insensitivity syndrome		X
220	Diffuse Globoid Body Sclerosis Krabbe disease		
221	Di George's syndrome	D	22q
222	Dihydrotestosterone receptor deficiency androgen insensitivity syndrome		X
223	distal spinal muscular atrophy, type V		
224	DM1 Myotonic dystrophy#type 1	T	19
225	DM2 Myotonic dystrophy#type 2	T	3
226	Down syndrome		21
227	DSMAV distal spinal muscular atrophy, type V		
228	DSN Charcot-Marie-Tooth disease#type 4		
229	DSS Charcot-Marie-Tooth disease, type 4		
230	Duchenne/Becker muscular dystrophy Muscular dystrophy, Duchenne and Becker type		
231	Dwarf, achondroplastic achondroplasia		3
232	Dwarf, thanatophoric thanatophoric dysplasia		
233	Dwarfism		
	Dwarfism-retinal atrophy-deafness syndrome		

234	Cockayne syndrome		
235	dysmyelinogenic leukodystrophy Alexander disease		
236	Dystrophia myotonica myotonic dystrophy	T	19
237	dystrophia retinae pigmentosa-dysostosis syndrome Usher syndrome		
238	Early-Onset familial alzheimer disease (EOFAD) Alzheimer disease#type 1 Alzheimer disease#type 3 Alzheimer disease#type 4		
239	EDS Ehlers–Danlos syndrome		
240	Ehlers–Danlos syndrome		
241	Ekman-Lobstein disease osteogenesis imperfecta		
242	Entrapment neuropathy hereditary neuropathy with liability to pressure palsies		
243	Epiloia tuberous sclerosis		
244	EPP erythropoietic protoporphyria		
245	Erythroblastic anemia beta-thalassemia		
246	Erythrohepatic protoporphyria erythropoietic protoporphyria		
247	Erythroid 5-aminolevulinate synthetase deficiency X-linked sideroblastic anemia		
248	Erythropoietic porphyria congenital erythropoietic porphyria		
249	erythropoietic protoporphyria		
250	Erythropoietic uroporphyria congenital erythropoietic porphyria		
251	Eye cancer retinoblastoma FA - Friedreich ataxia Friedreich's ataxia		
252	FA fanconi anemia		
253	Fabry disease	P	Xq22.1
254	Facial injuries and disorders		
255	factor V Leiden thrombophilia		
256	FALS amyotrophic lateral sclerosis		
257	familial acoustic neuroma neurofibromatosis type II		

258	familial adenomatous polyposis		
259	familial Alzheimer disease (FAD) Alzheimer's disease		
260	familial amyotrophic lateral sclerosis amyotrophic lateral sclerosis		
261	familial dysautonomia		
262	familial fat-induced hypertriglyceridemia lipoprotein lipase deficiency, familial		
263	familial hemochromatosis hemochromatosis		
264	familial LPL deficiency lipoprotein lipase deficiency, familial		
265	familial nonpolyposis colon cancer hereditary nonpolyposis colorectal cancer		
266	familial paroxysmal polyserositis Mediterranean fever, familial		
267	familial PCT porphyria cutanea tarda		
268	familial pressure-sensitive neuropathy hereditary neuropathy with liability to pressure palsies		
269	familial primary pulmonary hypertension (FPPH) primary pulmonary hypertension		
270	Familial Turner syndrome Noonan syndrome		
271	familial vascular leukoencephalopathy CADASIL syndrome		
272	FAP familial adenomatous polyposis		
273	FD familial dysautonomia		
274	Female pseudo-Turner syndrome Noonan syndrome		
275	Ferrochelatase deficiency erythropoietic protoporphyria		
276	ferroportin disease Haemochromatosis#type 4		
277	Fever Mediterranean fever, familial		
278	FG syndrome		
279	FGFR3-associated coronal synostosis Muenke syndrome		
280	Fibrinoid degeneration of astrocytes Alexander disease		
281	Fibrocystic disease of the pancreas cystic fibrosis		
	FMF		

282	Mediterranean fever, familial		
283	Folling disease phenylketonuria		
284	fra(X) syndrome fragile X syndrome		Xq27.3
285	fragile X syndrome		Xq27.3
286	Fragilitas ossium osteogenesis imperfecta		
287	FRAXA syndrome fragile X syndrome		Xq27.3
288	FRDA Friedreich's ataxia		
289	Friedreich's ataxia Friedreich's ataxia		
290	Friedreich's ataxia		
291	FXS fragile X syndrome		Xq27.3
292	G6PD deficiency		
293	Galactokinase deficiency disease galactosemia		
294	Galactose-1-phosphate uridyl-transferase deficiency disease galactosemia		
295	galactosemia		
296	Galactosylceramidase deficiency disease Krabbe disease		
297	Galactosylceramide lipidosis Krabbe disease		
298	galactosylcerebrosidase deficiency Krabbe disease		
299	galactosylsphingosine lipidosis Krabbe disease		
300	GALC deficiency Krabbe disease		
301	GALT deficiency galactosemia		
302	Gaucher disease		
303	Gaucher-like disease pseudo-Gaucher disease		
304	GBA deficiency Gaucher disease type 1		
305	GD Gaucher's disease		
306	Genetic brain disorders		
307	genetic emphysema alpha 1-antitrypsin deficiency		

308	genetic hemochromatosis hemochromatosis		
309	Giant cell hepatitis, neonatal Neonatal hemochromatosis		
310	GLA deficiency Fabry disease		
311	Glioblastoma, retinal retinoblastoma		
312	Glioma, retinal retinoblastoma		
313	globoid cell leukodystrophy (GCL, GLD) Krabbe disease		
314	globoid cell leukoencephalopathy Krabbe disease		
315	Glucocerebrosidase deficiency Gaucher disease		
316	Glucocerebrosidosis Gaucher disease		
317	Glucosyl cerebroside lipidosis Gaucher disease		
318	Glucosylceramidase deficiency Gaucher disease		
319	Glucosylceramide beta-glucosidase deficiency Gaucher disease		
320	Glucosylceramide lipidosis Gaucher disease		
321	Glyceric aciduria hyperoxaluria, primary		
322	Glycine encephalopathy Nonketotic hyperglycinemia		
323	Glycolic aciduria hyperoxaluria, primary		
324	GM2 gangliosidosis, type 1 Tay-Sachs disease		
325	Goiter-deafness syndrome Pendred syndrome		
326	Graefe-Usher syndrome Usher syndrome		
327	Gronblad-Strandberg syndrome pseudoxanthoma elasticum		
328	Guenther porphyria congenital erythropoietic porphyria		
329	Gunther disease congenital erythropoietic porphyria		
330	Haemochromatosis hemochromatosis		

331	Hallgren syndrome Usher syndrome		
332	Harlequin type ichthyosis		
333	Hb S disease sickle cell anemia		
334	HCH hypochondroplasia		
335	HCP hereditary coproporphyria		
336	Head and brain malformations		
337	Hearing disorders and deafness		
338	Hearing problems in children		
339	HEF2A hemochromatosis#type 2		
340	HEF2B hemochromatosis#type 2		
341	Hematoporphyria porphyria		
342	Heme synthetase deficiency erythropoietic protoporphyria		
343	Hemochromatoses hemochromatosis		
344	hemochromatosis		
345	hemoglobin M disease methemoglobinemia#beta-globin type		
346	Hemoglobin S disease sickle cell anemia		
347	hemophilia		
348	HEP hepatoerythropoietic porphyria		
349	hepatic AGT deficiency hyperoxaluria, primary		
350	hepatoerythropoietic porphyria		
351	Hepatolenticular degeneration syndrome Wilson disease		
352	Hereditary arthro-ophthalmopathy Stickler syndrome		
353	Hereditary coproporphyria	P	3q12
354	Hereditary dystopic lipidosis Fabry disease		
355	Hereditary hemochromatosis (HHC) hemochromatosis		
356	Hereditary hemorrhagic telangiectasia (HHT)		
357	Hereditary Inclusion Body Myopathy skeletal muscle regeneration		

358	Hereditary iron-loading anemia X-linked sideroblastic anemia		
359	Hereditary motor and sensory neuropathy Charcot-Marie-Tooth disease		
360	Hereditary motor neuronopathy spinal muscular atrophy		
361	Hereditary motor neuronopathy, type V distal spinal muscular atrophy, type V		
362	Hereditary multiple exostoses		
363	Hereditary nonpolyposis colorectal cancer	DNA mismatch repair dysfunction usually in MSH2 and MLH1 genes	usually chromosomes 2 and 3
364	Hereditary periodic fever syndrome Mediterranean fever, familial		
365	Hereditary Polyposis Coli familial adenomatous polyposis		
366	Hereditary pulmonary emphysema alpha 1-antitrypsin deficiency		
367	Hereditary resistance to activated protein C factor V Leiden thrombophilia		
368	Hereditary sensory and autonomic neuropathy type III familial dysautonomia		
369	Hereditary spastic paraplegia infantile-onset ascending hereditary spastic paralysis		
370	Hereditary spinal ataxia Friedreich's ataxia		
371	Hereditary spinal sclerosis Friedreich's ataxia		
372	Herrick's anemia sickle cell anemia		
373	Heterozygous OSMED Weissenbacher-Zweymüller syndrome		
374	Heterozygous otospondylomegaepiphyseal dysplasia Weissenbacher-Zweymüller syndrome		
375	HexA deficiency Tay-Sachs disease		
376	Hexosaminidase A deficiency Tay-Sachs disease		
377	Hexosaminidase alpha-subunit deficiency (variant B) Tay-Sachs disease		
378	HFE-associated hemochromatosis hemochromatosis		
379	HGPS Progeria		
380	Hippel-Lindau disease		

	von Hippel-Lindau disease		
381	HLAH hemochromatosis		
382	HMN V distal spinal muscular atrophy, type V		
383	HMSN Charcot-Marie-Tooth disease		
384	HNPCC hereditary nonpolyposis colorectal cancer		
385	HNPP hereditary neuropathy with liability to pressure palsies		
386	homocystinuria		
387	Homogentisic acid oxidase deficiency alkaptonuria		
388	Homogentisic acidura alkaptonuria		
389	Homozygous porphyria cutanea tarda hepatoerythropoietic porphyria		
390	HP1 hyperoxaluria, primary		
391	HP2 hyperoxaluria, primary		
392	HPA hyperphenylalaninemia		
393	HPRT - Hypoxanthine-guanine phosphoribosyltransferase deficiency Lesch-Nyhan syndrome		
394	HSAN type III familial dysautonomia		
395	HSAN3 familial dysautonomia		
396	HSN-III familial dysautonomia		
397	Human dermatosparaxis Ehlers–Danlos syndrome#dermatosparaxis type		
398	Huntington's disease	T	4p16.3
399	Hutchinson-Gilford progeria syndrome progeria		
400	Hyperandrogenism, nonclassic type, due to 21-hydroxylase deficiency 21-hydroxylase deficiency		
401	Hyperchylomicronemia, familial lipoprotein lipase deficiency, familial		
402	Hyperglycinemia with ketoacidosis and leukopenia propionic acidemia		
	Hyperlipoproteinemia type I		

403	lipoprotein lipase deficiency, familial		
404	hyperoxaluria, primary		
405	hyperphenylalaninaemia hyperphenylalaninemia		
406	hyperphenylalaninemia		
407	Hypochondrodysplasia hypochondroplasia		
408	Hypochondrogenesis		
409	Hypochondroplasia		4p16.3
410	Hypochromic anemia X-linked sideroblastic anemia		
411	Hypocupremia, congenital Menkes disease		
412	Hypoxanthine phosphoribosyltransferse (HPRT) deficiency Lesch-Nyhan syndrome		
413	IAHSP infantile-onset ascending hereditary spastic paralysis		
414	ICF syndrome Immunodeficiency, centromere instability and facial anomalies syndrome		20q11.2
415	Idiopathic hemochromatosis hemochromatosis, type 3		
416	Idiopathic neonatal hemochromatosis hemochromatosis, neonatal		
417	Idiopathic pulmonary hypertension primary pulmonary hypertension		
418	Immune system disorders X-linked severe combined immunodeficiency		
419	Incontinentia pigmenti	P	Xq28
420	Infantile cerebral Gaucher's disease Gaucher disease type 2		
421	Infantile Gaucher disease Gaucher disease type 2		
422	infantile-onset ascending hereditary spastic paralysis		
423	Infertility		
424	inherited emphysema alpha 1-antitrypsin deficiency		
425	Inherited human transmissible spongiform encephalopathies prion disease		
426	inherited tendency to pressure palsies hereditary neuropathy with liability to pressure palsies		
427	Insley-Astley syndrome otospondylomegaepiphyseal dysplasia		
428	Intermittent acute porphyria syndrome acute intermittent porphyria		

429	Intestinal polyposis-cutaneous pigmentation syndrome Peutz–Jeghers syndrome		
430	IP incontinentia pigmenti		
431	Iron storage disorder hemochromatosis		
432	Isodicentric 15 isodicentric 15	Inv dup	15q11-14
433	Isolated deafness nonsyndromic deafness		
434	Jackson-Weiss syndrome		
435	JH Haemochromatosis#type 2		
436	Joubert syndrome		
437	JPLS Juvenile Primary Lateral Sclerosis		ALS2
438	juvenile amyotrophic lateral sclerosis Amyotrophic lateral sclerosis#type 2		
439	Juvenile gout, choreoathetosis, mental retardation syndrome Lesch-Nyhan syndrome		
440	juvenile hyperuricemia syndrome Lesch-Nyhan syndrome		
441	JWS Jackson-Weiss syndrome		
442	KD X-linked spinal-bulbar muscle atrophy		
443	Kennedy disease X-linked spinal-bulbar muscle atrophy		
444	Kennedy spinal and bulbar muscular atrophy X-linked spinal-bulbar muscle atrophy		
445	Kerasin histiocytosis Gaucher disease		
446	Kerasin lipoidosis Gaucher disease		
447	Kerasin thesaurismosis Gaucher disease		
448	ketotic glycinemia propionic acidemia		
449	ketotic hyperglycinemia propionic acidemia		
450	Kidney diseases hyperoxaluria, primary		
451	Klinefelter syndrome		
452	Klinefelter syndrome Klinefelter syndrome		
453	Kniest dysplasia		

454	Krabbe disease		
455	Lacunar dementia CADASIL syndrome		
456	Langer-Saldino achondrogenesis achondrogenesis, type II		
457	Langer-Saldino dysplasia achondrogenesis, type II		
458	Late-onset Alzheimer disease Alzheimer disease#type 2		
459	Late-onset familial Alzheimer disease (AD2) Alzheimer disease#type 2		
460	late-onset Krabbe disease (LOKD) Krabbe disease		
461	Learning Disorders Learning disability		
462	Lentiginosis, perioral Peutz-Jeghers syndrome		
463	Lesch-Nyhan syndrome		
464	Leukodystrophies		
465	leukodystrophy with Rosenthal fibers Alexander disease		
466	Leukodystrophy, spongiform Canavan disease		
467	LFS Li-Fraumeni syndrome		
468	Li-Fraumeni syndrome		
469	Lipase D deficiency lipoprotein lipase deficiency, familial		
470	LIPD deficiency lipoprotein lipase deficiency, familial		
471	Lipidosis, cerebroside Gaucher disease		
472	Lipidosis, ganglioside, infantile Tay-Sachs disease		
473	Lipoid histiocytosis (kerasin type) Gaucher disease		
474	lipoprotein lipase deficiency, familial		
475	Liver diseases galactosemia		
476	Lou Gehrig disease amyotrophic lateral sclerosis		
477	Louis-Bar syndrome ataxia telangiectasia		
478	Lynch syndrome hereditary nonpolyposis colorectal cancer		

479	Lysyl-hydroxylase deficiency Ehlers–Danlos syndrome#kyphoscoliosis type		
480	Machado-Joseph disease Spinocerebellar ataxia#type 3		
481	Male breast cancer breast cancer		
482	Male genital disorders		
483	Male Turner syndrome Noonan syndrome		
484	Malignant neoplasm of breast breast cancer		
485	malignant tumor of breast breast cancer		
486	Malignant tumor of urinary bladder bladder cancer		
487	Mammary cancer breast cancer		
488	Marfan syndrome		15
489	Marker X syndrome fragile X syndrome		
490	Martin-Bell syndrome fragile X syndrome		
491	McCune–Albright syndrome		20 q13.2-13.3
492	McLeod syndrome		X
493	MEDNIK	D	AP1S1
494	Mediterranean Anemia beta-thalassemia		
495	Mediterranean fever, familial		
496	Mega-epiphyseal dwarfism otospondylomegaepiphyseal dysplasia		
497	Menkea syndrome Menkes disease		
498	Menkes disease		
499	Mental retardation with osteocartilaginous abnormalities Coffin-Lowry syndrome		
500	Metabolic disorders		
501	Metatropic dwarfism, type II Kniest dysplasia		
502	Metatropic dysplasia type II Kniest dysplasia		
503	Methemoglobinemia#beta-globin type		
504	methylmalonic acidemia		
505	MFS Marfan syndrome		
	MHAM		

506	Cowden syndrome		
507	MK Menkes disease		
508	Micro syndrome		2q21.3
509	Microcephaly	P	1q31 (ASPM)
510	MMA methylmalonic acidemia		
511	MNK Menkes disease		
512	Monosomy 1p36 syndrome 1p36 deletion syndrome	D	1p36
513	monosomy X Turner syndrome		
514	Motor neuron disease, amyotrophic lateral sclerosis amyotrophic lateral sclerosis		
515	Movement disorders		
516	Mowat-Wilson syndrome		
517	Mucopolysaccharidosis (MPS I)		
518	Mucoviscidosis cystic fibrosis		
519	Muenke syndrome		
520	Multi-Infarct dementia CADASIL syndrome		
521	Multiple carboxylase deficiency, late-onset biotinidase deficiency		
522	Multiple hamartoma syndrome Cowden syndrome		
523	Multiple neurofibromatosis neurofibromatosis		
524	Muscular dystrophy		
525	Muscular dystrophy, Duchenne and Becker type		
526	Myotonia atrophica myotonic dystrophy		
527	Myotonia dystrophica myotonic dystrophy		
528	myotonic dystrophy		
529	Myxedema, congenital congenital hypothyroidism		
530	Nance-Insley syndrome otospondylomegaepiphyseal dysplasia		
531	Nance-Sweeney chondrodysplasia otospondylomegaepiphyseal dysplasia		
532	NBIA1 pantothenate kinase-associated neurodegeneration		

533	Neill-Dingwall syndrome Cockayne syndrome		
534	Neuroblastoma, retinal retinoblastoma		
535	Neurodegeneration with brain iron accumulation type 1 pantothenate kinase-associated neurodegeneration		
536	Neurofibromatosis type I		17q11.2
537	Neurofibromatosis type II		
538	Neurologic diseases		
539	Neuromuscular disorders		
540	neuronopathy, distal hereditary motor, type V Distal spinal muscular atrophy#type V		
541	neuronopathy, distal hereditary motor, with pyramidal features Amyotrophic lateral sclerosis#type 4		
542	Niemann-Pick Niemann–Pick disease	NPA, NPB, NPC1, NPC2, Sphingomyelin phosphodiesterase 1	SMPD1
543	Noack syndrome Pfeiffer syndrome		
544	Nonketotic hyperglycinemia Glycine encephalopathy		
545	Non-neuronopathic Gaucher disease Gaucher disease type 1		
546	Non-phenylketonuric hyperphenylalaninemia tetrahydrobiopterin deficiency		
547	nonsyndromic deafness		
548	Noonan syndrome		
549	Norrbottnian Gaucher disease Gaucher disease type 3		
550	Ochronosis alkaptonuria		
551	Ochronotic arthritis alkaptonuria		
552	OI osteogenesis imperfecta		
553	Osler-Weber-Rendu disease Hereditary hemorrhagic telangiectasia		
554	OSMED otospondylomegaepiphyseal dysplasia		
555	osteogenesis imperfecta		
556	Osteopsathyrosis osteogenesis imperfecta		
557	Osteosclerosis congenita achondroplasia		

558	Oto-spondylo-megaepiphyseal dysplasia otospondylomegaepiphyseal dysplasia		
559	otospondylomegaepiphyseal dysplasia		
560	Oxalosis hyperoxaluria, primary		
561	Oxaluria, primary hyperoxaluria, primary		
562	pantothenate kinase-associated neurodegeneration		
563	Patau Syndrome (Trisomy 13)		
564	PBGD deficiency acute intermittent porphyria		
565	PCC deficiency propionic acidemia		
566	PCT porphyria cutanea tarda		
567	PDM Myotonic dystrophy#type 2		
568	Pendred syndrome		
569	Periodic disease Mediterranean fever, familial		
570	Periodic peritonitis Mediterranean fever, familial		
571	Periorificial lentiginosis syndrome Peutz-Jeghers syndrome		
572	Peripheral nerve disorders familial dysautonomia		
573	Peripheral neurofibromatosis neurofibromatosis type I		
574	Peroneal muscular atrophy Charcot-Marie-Tooth disease		
575	peroxisomal alanine:glyoxylate aminotransferase deficiency hyperoxaluria, primary		
576	Peutz-Jeghers syndrome		
577	Pfeiffer syndrome		
578	Phenylalanine hydroxylase deficiency disease phenylketonuria		
579	phenylketonuria		
580	Pheochromocytoma von Hippel-Lindau disease		
581	Pierre Robin syndrome with fetal chondrodysplasia Weissenbacher-Zweymüller syndrome		
582	Pigmentary cirrhosis hemochromatosis		
583	PJS Peutz-Jeghers syndrome		

584	PKAN pantothenate kinase-associated neurodegeneration		
585	PKU phenylketonuria		
586	Plumboporphyria ALA deficiency porphyria		
587	PMA Charcot-Marie-tooth disease		
588	Polycystic kidney disease	P	16 (PKD1) or 4 (PKD2)
589	polyostotic fibrous dysplasia McCune–Albright syndrome		20 q13.2-13.3
590	polyposis coli familial adenomatous polyposis		
591	polyposis, hamartomatous intestinal Peutz-Jeghers syndrome		
592	polyposis, intestinal, II Peutz-Jeghers syndrome		
593	polyps-and-spots syndrome Peutz-Jeghers syndrome		
594	Porphobilinogen synthase deficiency ALA deficiency porphyria		
595	porphyria		
596	porphyrin disorder porphyria		
597	PPH primary pulmonary hypertension		
598	PPOX deficiency variegate porphyria		
599	Prader-Labhart-Willi syndrome Prader-Willi syndrome		
600	Prader-Willi syndrome		
601	presenile and senile dementia Alzheimer's disease		
602	primary hemochromatosis hemochromatosis		
603	primary hyperuricemia syndrome Lesch-Nyhan syndrome		
604	primary pulmonary hypertension		
605	primary senile degenerative dementia Alzheimer's disease		
606	prion disease		
607	procollagen type EDS VII, mutant Ehlers–Danlos syndrome#arthrochalasia type		
608	progeria Hutchinson Gilford Progeria Syndrome		

609	Progeria-like syndrome Cockayne syndrome		
610	progeroid nanism Cockayne syndrome		
611	progressive chorea, chronic hereditary (Huntington) Huntington's disease		
612	progressive muscular atrophy spinal muscular atrophy		
613	progressively deforming osteogenesis imperfecta with normal sclerae Osteogenesis imperfecta#Type III		
614	PROMM Myotonic dystrophy#type 2		
615	propionic acidemia		
616	propionyl-CoA carboxylase deficiency propionic acidemia		
617	protein C deficiency		
618	protein S deficiency		
619	protoporphyria erythropoietic protoporphyria		
620	protoporphyrinogen oxidase deficiency variegate porphyria		
621	proximal myotonic dystrophy Myotonic dystrophy#type 2		
622	proximal myotonic myopathy Myotonic dystrophy#type 2		
623	pseudo-Gaucher disease		
624	pseudo-Ullrich-Turner syndrome Noonan syndrome		
625	pseudoxanthoma elasticum		
626	psychosine lipidosis Krabbe disease		
627	pulmonary arterial hypertension primary pulmonary hypertension		
628	pulmonary hypertension primary pulmonary hypertension		
629	PWS Prader-Willi syndrome		
630	PXE - pseudoxanthoma elasticum pseudoxanthoma elasticum		
631	Rb retinoblastoma		
632	Recklinghausen disease, nerve neurofibromatosis type I		
633	Recurrent polyserositis		

	Mediterranean fever, familial		
634	Retinal disorders		
635	Retinitis pigmentosa-deafness syndrome Usher syndrome		
636	Retinoblastoma		
637	Rett syndrome		
638	RFALS type 3 Amyotrophic lateral sclerosis#type 2		
639	Ricker syndrome Myotonic dystrophy#type 2		
640	Riley-Day syndrome familial dysautonomia		
641	Roussy-Levy syndrome Charcot-Marie-Tooth disease		
642	RSTS Rubinstein-Taybi syndrome		
643	RTS Rett syndrome Rubinstein-Taybi syndrome		
644	RTT Rett syndrome		
645	Rubinstein-Taybi syndrome		
646	Sack-Barabas syndrome Ehlers–Danlos syndrome, vascular type		
647	SADDAN		
648	sarcoma family syndrome of Li and Fraumeni Li-Fraumeni syndrome		
649	sarcoma, breast, leukemia, and adrenal gland (SBLA) syndrome Li-Fraumeni syndrome		
650	SBLA syndrome Li-Fraumeni syndrome		
651	SBMA X-linked spinal-bulbar muscle atrophy		
652	SCD sickle cell anemia		
653	Schwannoma, acoustic, bilateral neurofibromatosis type II		
654	Schwartz–Jampel syndrome		
655	SCIDX1 X-linked severe combined immunodeficiency		
656	sclerosis tuberosa tuberous sclerosis		
657	SDAT Alzheimer's disease		

658	SED congenita spondyloepiphyseal dysplasia congenita		
659	SED Strudwick spondyloepimetaphyseal dysplasia, Strudwick type		
660	SEDc spondyloepiphyseal dysplasia congenita		
661	SEMD, Strudwick type spondyloepimetaphyseal dysplasia, Strudwick type		
662	senile dementia Alzheimer disease#type 2		
663	severe achondroplasia with developmental delay and acanthosis nigricans SADDAN		
664	Shprintzen syndrome 22q11.2 deletion syndrome	D	22q
665	sickle cell anemia	D	18q
666	Siderius X-linked mental retardation syndrome	PD	Xp11.22
667	skeleton-skin-brain syndrome SADDAN		
668	Skin pigmentation disorders		
669	SMA spinal muscular atrophy		
670	SMED, Strudwick type spondyloepimetaphyseal dysplasia, Strudwick type		
671	SMED, type I spondyloepimetaphyseal dysplasia, Strudwick type		
672	Smith-Lemli-Opitz syndrome		
673	Smith Magenis Syndrome		
674	South-African genetic porphyria variegate porphyria		
675	spastic paralysis, infantile onset ascending infantile-onset ascending hereditary spastic paralysis		
676	Speech and communication disorders		
677	sphingolipidosis, Tay-Sachs Tay-Sachs disease		
678	spinal-bulbar muscular atrophy		
679	spinal muscular atrophy		
680	spinal muscular atrophy, distal type V Distal spinal muscular atrophy#type V		
681	spinal muscular atrophy, distal, with upper limb predominance Distal spinal muscular atrophy#type V		
682	spinocerebellar ataxia		
683	spondyloepimetaphyseal dysplasia, Strudwick type		
684	spondyloepiphyseal dysplasia congenita		

685	spondyloepiphyseal dysplasia collagenopathy, types II and XI		
686	spondylometaepiphyseal dysplasia congenita, Strudwick type spondyloepimetaphyseal dysplasia, Strudwick type		
687	spondylometaphyseal dysplasia (SMD) spondyloepimetaphyseal dysplasia, Strudwick type		
688	spondylometaphyseal dysplasia, Strudwick type spondyloepimetaphyseal dysplasia, Strudwick type		
689	spongy degeneration of central nervous system Canavan disease		
690	spongy degeneration of the brain Canavan disease		
691	spongy degeneration of white matter in infancy Canavan disease		
692	sporadic primary pulmonary hypertension primary pulmonary hypertension		
693	SSB syndrome SADDAN		
694	steely hair syndrome Menkes disease		
695	Steinert disease myotonic dystrophy		
696	Steinert myotonic dystrophy syndrome myotonic dystrophy		
697	Stickler syndrome		
698	stroke CADASIL syndrome		
699	Strudwick syndrome spondyloepimetaphyseal dysplasia, Strudwick type		
700	subacute neuronopathic Gaucher disease Gaucher disease type 3		
701	Swedish genetic porphyria acute intermittent porphyria		
702	Swedish porphyria acute intermittent porphyria		
703	Swiss cheese cartilage dysplasia Kniest dysplasia		
704	Tay-Sachs disease		
705	TD - thanatophoric dwarfism thanatophoric dysplasia		
706	TD with straight femurs and cloverleaf skull thanatophoric dysplasia#Type 2		
707	Telangiectasia, cerebello-oculocutaneous ataxia telangiectasia		
	Testicular feminization syndrome		

708	androgen insensitivity syndrome		
709	tetrahydrobiopterin deficiency		
710	TFM - testicular feminization syndrome androgen insensitivity syndrome		
711	thalassemia intermedia beta-thalassemia		
712	Thalassemia Major beta-thalassemia		
713	thanatophoric dysplasia		
714	Thrombophilia due to deficiency of cofactor for activated protein C, Leiden type factor V Leiden thrombophilia		
715	Thyroid disease		
716	Tomaculous neuropathy hereditary neuropathy with liability to pressure palsies		
717	Total HPRT deficiency Lesch-Nyhan syndrome		
718	Total hypoxanthine-guanine phosphoribosyl transferase deficiency Lesch-Nyhan syndrome		
719	Transmissible dementias prion disease		
720	Transmissible spongiform encephalopathies prion disease		
721	Treacher Collins syndrome		5q32-q33.1
722	Trias fragilitis ossium osteogenesis imperfecta#Type I		
723	triple X syndrome		
724	Triplo X syndrome triple X syndrome		
725	Trisomy 21 Down syndrome		
726	Trisomy X triple X syndrome		
727	Troisier-Hanot-Chauffard syndrome hemochromatosis		
728	TS Turner syndrome		
729	TSD Tay-Sachs disease		
730	TSEs prion disease		
731	tuberose sclerosis tuberous sclerosis		
732	tuberous sclerosis		

733	Turner syndrome		
734	Turner syndrome in female with X chromosome Noonan syndrome		
735	Turner's phenotype, karyotype normal Noonan syndrome		
736	Turner's syndrome Turner syndrome		
737	Turner-like syndrome Noonan syndrome		
738	Type 2 Gaucher disease Gaucher disease type 2		
739	Type 3 Gaucher disease Gaucher disease type 3		
740	UDP-galactose-4-epimerase deficiency disease galactosemia		
741	UDP glucose 4-epimerase deficiency disease galactosemia		
742	UDP glucose hexose-1-phosphate uridylyltransferase deficiency galactosemia		
743	Ullrich-Noonan syndrome Noonan syndrome		
744	Ullrich-Turner syndrome Turner syndrome		
745	Undifferentiated deafness nonsyndromic deafness		
746	UPS deficiency acute intermittent porphyria		
747	Urinary bladder cancer bladder cancer		
748	UROD deficiency porphyria cutanea tarda		
749	Uroporphyrinogen decarboxylase deficiency porphyria cutanea tarda		
750	Uroporphyrinogen synthase deficiency acute intermittent porphyria		
751	UROS deficiency congenital erythropoietic porphyria		
752	Usher syndrome		
753	UTP hexose-1-phosphate uridylyltransferase deficiency galactosemia		
754	Van Bogaert-Bertrand syndrome Canavan disease		
755	Van der Hoeve syndrome osteogenesis imperfecta#Type I		
756	variegate porphyria		

757	Velocardiofacial syndrome 22q11.2 deletion syndrome	D	22q
758	VHL syndrome von Hippel-Lindau disease		
759	Vision impairment and blindness Alström syndrome		
760	Von Bogaert-Bertrand disease Canavan disease		
761	von Hippel-Lindau disease		
762	Von Recklenhausen-Applebaum disease hemochromatosis		
763	von Recklinghausen disease neurofibromatosis type I		
764	VP variegate porphyria		
765	Vrolik disease osteogenesis imperfecta		
766	Waardenburg syndrome		
767	Warburg Sjo Fledelius Syndrome Micro syndrome		2q21.3
768	WD Wilson disease		
769	Weissenbacher-Zweymüller syndrome		
770	Williams Syndrome		
771	Wilson disease		
772	Wilson's disease Wilson disease		
773	Wolf–Hirschhorn syndrome	D	4p
774	Wolff Periodic disease Mediterranean fever, familial		
775	WZS Weissenbacher-Zweymüller syndrome		
776	Xeroderma pigmentosum	ERCC4	15
777	X-linked mental retardation and macroorchidism fragile X syndrome		
778	X-linked primary hyperuricemia Lesch-Nyhan syndrome		
779	X-linked severe combined immunodeficiency		
780	X-linked sideroblastic anemia		
781	X-linked spinal-bulbar muscle atrophy Kennedy's disease		
782	X-linked uric aciduria enzyme defect Lesch-Nyhan syndrome		
783	X-SCID X-linked severe combined immunodeficiency		

784	XLSA X-linked sideroblastic anemia		
785	XSCID X-linked severe combined immunodeficiency		
786	XXX syndrome triple X syndrome		
787	XXXX syndrome 48, XXXX		
788	XXXXX syndrome 49, XXXXX		
789	XXY syndrome Klinefelter syndrome		
790	XXY trisomy Klinefelter syndrome		
791	XYY karyotype 47,XYY syndrome		
792	XYY syndrome 47,XYY syndrome		
793	YY syndrome 47,XYY syndrome		