#	***DISEASE***
1	17-beta-hydroxysteroid dehydrogenase defciency, type III
2	3-Hydroxy-3-methylglutaryl-CoA lyase deficiency
3	3-hydroxyacyl-CoA dehydrogenase defciency
4	3-hydroxyisobutryl-CoA hydrolase deficiency
5	3-Ketothiolase deficiency
6	3-methylglutaconic aciduria
7	46, XX sex reversal 4 46XY sex reversal 3
8	Aarskog-Scott syndrome
9	ABCD syndrome
10	Achalasia-addisonianism-alacrima syndrome
11	Achondrogenesis, type IB
12	Acyl-CoA dehydrogenase-9 (ACAD9) Deficiency
13	Adenosine deaminase deficiency
14	Adrenal insufficiency, congenital, with 46XY sex reversal, partial or complete
15	Adrenocortical insufficiency
16	Adrenoleukodystrophy, X-linked
17	Afibrinogenemia, congenital
18	Agammaglobulinemia, X-linked 1 Agammaglobulinemia and isolated hormone deficiency
19	Aicardi-Goutieres syndrome
20	Allan-Herndon-Dudley syndrome
21	Alpers-Huttenlocher syndrome
22	Alpha thalassemia
23	Alpha-mannosidosis
24	Alpha-methylacyl-CoA racemase deficiency
25	Alport syndrome
26	Alstrom syndrome
27	Amish infantile epilepsy syndrome
28	Amyotrophic lateral sclerosis
29	Anauxetic dysplasia
30	Andermann syndrome
31	Androgen insensitivity syndrome
32	Angelman syndrome
33	Antley-Bixler syndrome with genital anomalies and disordered steroidogenesis
34	Aplastic anemia
35	Apparent mineralocorticoid excess
36	Argininosuccinate lyase deficiency
37	Aromatase deficiency
38	Aromatic L-amino acid decarboxylase deficiency
39	Arterial calcification, generalized, of infancy, 1
40	Arthrogryposis, renal dysfunction, and cholestasis 1

41	Arts syndrome
42	Asperger syndrome susceptibility, X-linked
43	Ataxia neuropathy spectrum
44	Ataxia with isolated vitamin E deficiency
45	Ataxia, early-onset, with oculomotor apraxia and hypoalbuminemia
46	Ataxia-telangiectasia
47	Atelosteogenesis II
48	Autism susceptibility, X-linked
49	Autoimmune Lymphoproliferative Syndrome
50	Autoimmune polyendocrinopathy syndrome
51	Autosomal Recessive Spastic Ataxia of Charlevoix-Saguenay
52	Bannayan-Riley-Ruvalcaba syndrome
53	Bardet-Biedl syndrome
54	Barth syndrome
55	Bartter syndrome
56	Beta thalassemia
57	Bethlem myopathy 1
58	Bile acid synthesis defect
59	Biotinidase deficiency
60	Björnstad syndrome
61	Bloom syndrome
62	Bone mineral density variation QTL, osteoporosis
63	Brittle cornea syndrome 1
64	Caffey disease
65	Canavan disease
66	Carbamoylphosphate synthetase I deficiency
67	Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency
68	Carnitine palmitoyltransferase deficiency
69	Carnitine-acylcarnitine translocase deficiency
70	Carpenter syndrome
71	Cartilage-hair hypoplasia
72	Cataract 40, X-linked
73	Cerebellar hypoplasia and mental retardation with or without quadrupedal locomotion 1
74	Cerebral dysgenesis, neuropathy, ichthyosis, and palmoplantar keratoderma syndrome
75	Cerebrooculofacioskeletal syndrome 1
76	Cerebrotendinous xanthomatosis
77	Charcot-Marie-Tooth disease
78	Chediak-Higashi syndrome
79	Cholestasis
80	Chondrodysplasia punctata type 1, X-linked
81	Chondrodysplasia, Blomstrand type
82	Citrullinemia

83	COACH syndrome
84	Cockayne syndrome type B
85	Coenzyme Q10 deficiency, primary
86	Coffin-Lowry syndrome
87	Cohen syndrome
88	Cold-induced sweating syndrome 1
89	Combined cellular and humoral immune defects with granulomas
90	Combined oxidative phosphorylation deficiency
91	Combined pituitary hormone deficiency
92	Combined SAP deficiency
93	Congenital adrenal hyperplasia
94	Congenital amegakaryocytic thrombocytopenia
95	Congenital disorder of glycosylation
96	Congenital heart defects, nonsyndromic, 1, X-linked
97	Congenital ichthyosis
98	Congenital insensitivity to pain with anhidrosis
99	Corneal endothelial dystrophy
100	Costeff syndrome
101	Cowden syndrome 1
102	Craniofrontonasal dysplasia
103	Creatine deficiency syndrome
104	Cutis laxa
105	Cystic fibrosis
106	Cystinosis
107	D-bifunctional protein deficiency
108	De Sanc- tis-Cacchione syndrome
109	Deafness, X-linked 1 recessive
110	Dejerine-Sottas disease
111	Dent disease
112	Desmosterolosis
113	Diabetes mellitus, type I, susceptibility to
114	Diarrhea 4, malabsorptive, congenital
115	Diastrophic dysplasia
116	Dihydrolipoamide dehydrogenase deficiency
117	Dihydropyrimidine dehydrogenase deficiency
118	Donnai-Barrow syndrome
119	Duchenne muscular dystrophy
120	Dyskeratosis congenita, X-linked
121	Dystonia 27
122	Ectodermal dysplasia
123	Ehlers-Danlos syndrome
124	Eiken syndrome
125	Ellis-van Creveld syndrome
126	Encephalopathy, acute, infection-induced (herpes-specific)
127	Encephalopathy, neonatal severe

128	Epidermolysis bullosa dystrophica, AR Epidermolysis bullosa pruriginosa
129	Epidermolysis bullosa simplex with pyloric atresia
130	Epidermolysis bullosa, junctional
131	Epidermolysis bullosa, lethal acantholytic
132	Epilepsy, progressive myoclonic
133	Epileptic encephalopathy, early infantile
134	Ethylmalonic encephalopathy
135	Exudative vitreoretinopathy 2, X-linked
136	Fabry disease
137	Failure of tooth eruption, primary
138	Familial dysautonomia (HSAN3)
139	Familial hyperinsulinism
140	Familial Mediterranean fever
141	Fanconi anemia group C
142	Fetal akinesia deformation sequence
143	Fraser syndrome
144	Fucosidosis
145	Fuhrmann syndrome
146	Fumarase deficiency
147	Galactokinase deficiency
148	Galactosemia
149	Gallbladder disease 1
150	Gastric cancer, somatic
151	Gaucher disease
152	Geleophysic dysplasia 1
153	Gillessen-Kaesbach-Nishimura syndrome
154	Glioma susceptibility 2
155	Glucose-6-phosphate dehydrogenase deficiency
156	Glutaric aciduria
157	Glutathione synthetase deficiency
158	Glycine encephalopathy
159	Glycogen storage disease
160	GRACILE syndrome
161	Greenberg skeletal dysplasia
162	Griscelli syndrome
163	Guanidinoacetate methyltransferase deficiency
164	H. pylori infection, susceptibility to
165	Hemochromatosis
166	Hemophagocytic lymphohistiocytosis, familial
167	Hemophilia
168	Hepatic venoocclusive disease with immunodeficienc
169	Hepatitis B virus infection, susceptibility to
170	Hereditary fructose intolerance

171	Hermansky-Pudlak syndrome
172	Heterotaxy, visceral, 1, X-linked
173	HIV1 infection, resistance to
174	Holocarboxylase synthetase deficiency
175	Homocystinuria due to cystathionine beta-synthase deficiency
176	HPRT-related gout
177	HSD10 mitochondrial disease
178	Hyper IgM syndrome, X-linked
179	Hyper-IgD syndrome
180	Hyper-IgE recurrent infection syndrome, autosomal recessive
181	Hyperornithinemia-hyperammonemia-homocitrullinemia syndrome (Triple H syndrome)
182	Hypogonadotropic hypogonadism 7 without anosmia
183	Hypohidrotic ectodermal dysplasia
184	Hypomagnesemia 5, renal, with ocular involvement
185	Hypoparathyroidism-retardation-dysmorphism syndrome
186	Hypophosphatasia
187	Hypophosphatemic rickets
188	Hypothryoidism, congenital, nongoitrous 4
189	Hypotrichosis, congenital, with juvenile macular dystrophy
190	Ichthyosis, leukocyte vacuoles, alopecia, and sclerosing cholangitis
191	IFAP syndrome with or without BRESHECK syndrome
192	Immunodeficiency
193	Immunodeficiency-centromeric instability-facial anomalies syndrome 1
194	Immunodysregulation, polyendocrinopathy, and enteropathy, X-linked
195	Incontinentia pigmenti
196	Infantile neuroaxonal dystrophy
197	Invasive pneumococcal disease
198	Isovaleric acidemia
199	Johanson-Blizzard syndrome
200	Joubert syndrome
201	Kahrizi syndrome
202	Krabbe disease
203	L1 syndrome
204	Lacticacidemia due to PDX1 deficiency
205	Lathosterolosis
206	Leber congenital amaurosis
207	Leigh syndrome

208	Leigh syndrome due to cytochrome c oxidase deficiency
209	Leigh syndrome due to mitochondrial complex I deficiency
210	Leigh syndrome due to mitochondrial COX4 deficiency
211	Leigh syndrome with Complex IV deficiency
212	Leprechaunism
213	Lesch-Nyhan syndrome;
214	Lethal congenital contracture syndrome
215	Leukemia, acute myeloid
216	Leukocyte adhesion deficiency, type III
217	Leukodystrophy, hypomyelinating
218	Leydig cell hypoplasia;
219	Lhermitte-Duclos syndrome
220	LIG4 syndrome
221	Lipoid congenital adrenal hyperplasia
222	Lissencephaly
223	Long-chain 3-hydroxyacyl-CoA dehydrogenase (LCHAD) deficiency
224	Lowe syndrome
225	Lujan-Fryns syndrome
226	Lung cancer, susceptibility to
227	Luteinizing hormone resistance
228	Lymphoma, non-Hodgkin
229	Lymphoproliferative syndrome
230	Macrocephaly/autism syndrome
231	Macroglobulinemia, Waldenstrom, somatic
232	Macular degeneration, age-related, 3; AD
233	Mandibuloacral dysplasia
234	Maple syrup urine disease
235	Marinesco-Sjogren syndrome
236	Martsolf syndrome
237	Meckel syndrome
238	Medium-chain acyl-CoA dehydrogenase (MCAD) deficiency
239	MEDNIK syndrome
240	Megalencephalic leukoencephalopathy with subcortical cysts
241	Meningioma
242	Menkes disease
243	Mental retardation and microcephaly with pontine and cerebellar hypoplasia Mental retardation, with or without nystagmus
244	Mental retardation, autosomal recessive
245	Mental retardation, X-linked
246	Metachromatic leukodystrophy

247	Metaphyseal dysplasia without hypotrichosis
248	Methylmalonic acidemia
249	Methylmalonic aciduria
250	Mevalonic aciduria
251	Microphthalmia, syndromic
252	Microvascular complications of diabetes
253	Mitochondrial complex I deficiency
254	Mitochondrial complex III deficiency, nuclear
255	Mitochondrial complex IV deficiency
256	Mitochondrial DNA depletion syndrome
257	Mitochondrial neurogastrointestinal encephalopathy (MNGIE) disease
258	Mitochondrial trifunctional protein deficiency
259	Mohr-Tranebjaerg syndrome
260	Molybdenum cofactor deficiency
261	Mowat-Wilson syndrome
262	Mucolipidosis
263	Mucopolysaccharidosis
264	Mulibrey nanism
265	Multiple epiphyseal dysplasia
266	Multiple pterygium syndrome
267	Muscular dystrophy
268	Muscular dystrophy-dystroglycanopathy
269	Muscular dystrophy-dystroglycanopathy, FKRP-related
270	Myasthenic syndrome
271	Mycobacterium tuberculosis, susceptibility to
272	Myocerebrohepa- topathy syndrome
273	Myopathy, tubular aggregate
274	Myopia 6
275	Myosclerosis, congenital
276	Myotubular myopathy, X-linked
277	N-acetylglutamate synthase deficiency
278	Nance-Horan syndrome
279	Nemaline myopathy
280	Nephrolithiasis, type I
281	Nephronophthisis
282	Nephrotic syndrome
283	Neurodegeneration due to cerebral folate transport deficiency
284	Neurodegeneration with brain iron accumulation 1 HARP syndrome
285	Neuronal ceroid lipofuscinosis
286	Neuronopathy, distal hereditary motor, type VI
287	Neuropathy, congenital hypomyelinating
288	Neuropathy, hereditary, with or without age-related macular degeneration

289	Neuropathy, inflammatory demyelinating Neuropathy, recurrent, with pressure palsies
290	Neutropenia, severe congenital
291	Niemann-Pick disease
292	Nijmegen breakage syndrome
293	Non-syndromic hearing loss
294	Norrie disease
295	Odon- toonychodermal dysplasia
296	Ohdo syndrome, X-linked
297	Omenn syndrome
298	Opitz GBBB syndrome, type I
299	Ornithine transcarbamylase deficiency
300	Osteogenesis imperfecta
301	Osteopetrosis
302	Paget disease of bone 5, juvenile-onset
303	Peroxisomal acyl-CoA oxidase deficiency
304	Peroxisome biogenesis disorder
305	Phenylalanine hydroxylase deficiency (Phenylketonuria)
306	Phosphoribosylpyrophosphate synthetase superactivity
307	Pierson syndrome
308	Pitt-Hopkins syndrome
309	Plasminogen deficiency
310	Polycystic kidney disease, PKHD1-related
311	Polycystic liver disease 3 with or without kidney cysts
312	Pompe disease
313	Pontocerebellar hypoplasia
314	Porphyria, congenital erythropoietic
315	Premature ovarian failure 7
316	Primary lateral sclerosis
317	Progressive external ophthalmoplegia
318	Properdin deficiency, X-linked
319	Propionic acidemia, PCCA-related
320	Prostate cancer, somatic
321	Proteinuria, low molecular weight, with hypercalciuric nephrocalcinosis
322	Pseudohypoaldosteronism, type I
323	Pseudovaginal perineoscrotal hypospadias
324	PTEN hamartoma tumor syndrome
325	Pulmonary surfactant dysfunction
326	Pyogenic bacterial infections, recurrent, due to MYD88 deficiency
327	Pyridoxamine 5'-phosphate oxidase deficiency
328	Pyridoxine-dependent epilepsy
329	Pyruvate carboxylase deficiency
330	Pyruvate dehydrogenase E1-alpha deficiency

331	Pyruvate dehydrogenase phosphatase deficiency
332	Pyruvate kinase deficiency
333	Raine syndrome
334	Renal-hepatic-pancreatic dysplasia 1
335	Renpenning syndrome
336	Restrictive dermopathy, lethal
337	Retinitis pigmentosa
338	Rett syndrome, congenital variant
339	Rhizomelic chondrodysplasia punctata
340	Rickets, vitamin D-resistant
341	Roberts syndrome
342	Rosenberg-Chutorian syndrome
343	Roussy-Levy syndrome
344	Salla disease
345	Sandhoff disease
346	SC phocomelia syndrome
347	Schneckenbecken dysplasia
348	Schopf-Schulz-Passarge syndrome
349	Schwartz-Jampel syndrome, type 1
350	Seckel syndrome 1
351	Segawa syndrome
352	Senior-Løken
353	Senior-Loken syndrome
354	Septooptic dysplasia
355	Severe combined immunodeficiency (SCID)
356	Short-rib thoracic dysplasia with or without polydactyly
357	Shwachman-Diamond syndrome
358	Sialic acid storage disorder
359	Sialidosis, type I, II
360	Sickle cell disease
361	Simpson-Golabi-Behmel syndrome
362	Sjögren-Larsson syndrome
363	Smith-Lemli-Opitz syndrome
364	Smith-Magenis syndrome
365	Sotos syndrome 1
366	Spastic paralysis, infantile onset ascending
367	Spastic paraplegia 2, X-linked
368	Spermatogenic failure 8
369	Spinal muscular atrophy
370	Spondylocostal dysostosis 1, autosomal recessive
371	Squamous cell carcinoma, burn scar-related, somatic
372	Stormorken syndrome
373	Striatonigral degeneration, infantile
374	Stuve-Wiedemann syndrome
375	Succinic semialdehyde dehydrogenase deficiency
376	Succinyl CoA:3-oxoacid CoA transferase deficiency

377	Sudden infant death with dysgenesis of the testes syndrome
378	Sulfite oxidase deficiency
379	Systemic primary carnitine deficiency
380	Tay-Sachs disease
381	T-cell immunodeficiency, congenital alopecia, and nail dystrophy
382	Tetra-amelia syndrome 1
383	Thrombo- cytopenia, X-linked
384	Thrombotic thrombocytopenic purpura
385	Trichothiodystrophy 3, photosensitive
386	Trifunctional protein deficiency
387	Tuberculosis infection, protection against Tuberculosis, susceptibility to
388	Tyrosinemia
389	Ullrich congenital muscular dystrophy
390	Ulna and fibula, absence of, with severe limb deficiency
391	Usher syndrome
392	VACTERL association, X-linked
393	VATER association with macrocephaly and ventriculomegaly
394	Very long-chain acyl-CoA dehydrogenase (VLCAD) deficiency
395	Vitamin D-dependent rickets
396	Waardenburg syndrome
397	Waisman syndrome
398	Warburg micro syndrome 1
399	Weyers acrofacial dysostosis
400	Wilms tumor, somatic
401	Wilson disease
402	Wiskott-Aldrich syndrome
403	Wolcott-Rallison syndrome
404	Wrinkly skin syndrome
405	Xeroderma pigmentosum
406	Zellweger syndrome