Count	Disease Name
1	ABCC8-related Hyperinsulinism
2	Medium Chain Acyl-CoA Dehydrogenase Deficiency
3	Short Chain Acyl-CoA Dehydrogenase Deficiency
4	Very Long Chain Acyl-CoA Dehydrogenase Deficiency
5	Aspartylglycosaminuria
6	Glycogen Storage Disease Type III
7	Primary Hyperoxaluria Type 1
8	Polyglandular Autoimmune Syndrome Type 1
9	Sjogren-Larsson Syndrome
10	Hereditary Fructose Intolerance
11	Hypophosphatasia, Autosomal Recessive
12	Metachromatic Leukodystrophy
13	Canavan Disease
14	Citrullinemia Type 1
15	Ataxia-telangiectasia
16	Wilson Disease
17	Bardet-Biedl Syndrome, BBS1-related
18	Bardet-Biedl Syndrome, BBS10-related
19	Maple Syrup Urine Disease Type 1B
20	GRACILE Syndrome
21	Bloom Syndrome
22	Biotinidase Deficiency
23	Homocystinuria Caused by Cystathionine Beta-synthase Deficiency
24	Cystic Fibrosis
25	CLN3-related Neuronal Ceroid Lipofuscinosis
26	CLN5-related Neuronal Ceroid Lipofuscinosis
27	Northern Epilepsy
28	Usher Syndrome Type 3
29	Carnitine Palmitoyltransferase IA Deficiency
30	Carnitine Palmitoyltransferase II Deficiency
31	Cystinosis
32	Pycnodysostosis
33	21-hydroxylase-deficient Congenital Adrenal Hyperplasia
34	Smith-Lemli-Opitz Syndrome
35	Lipoamide Dehydrogenase Deficiency
36	Tyrosinemia Type I
37	Fanconi Anemia Type C
38	FKTN-related Disorders
39	Fragile X Syndrome
40	Glycogen Storage Disease Type Ia
41	Pompe Disease
42	Krabbe Disease
43	Galactosemia
44	Gaucher Disease
45	Glutaric Acidemia Type 1
46	GJB2-related DFNB1 Nonsyndromic Hearing Loss and Deafness
47	Inclusion Body Myopathy 2
48	Primary Hyperoxaluria Type 2
49	HADHA-related Disorders
50	Alpha Thalassemia
51	Hb Beta Chain-related Hemoglobinopathy (Including Beta Thalassemia and Sickle Cell Disease)
52	Hexosaminidase A Deficiency (Including Tay-Sachs Disease)
53	D-bifunctional Protein Deficiency
54	Mucopolysaccharidosis Type I
55	Familial Dysautonomia
56	Isovaleric Acidemia
57	Herlitz Junctional Epidermolysis Bullosa, LAMA3-related
58	Herlitz Junctional Epidermolysis Bullosa, LAMB3-related
59	Herlitz Junctional Epidermolysis Bullosa, LAMC2-related
60	Alpha-mannosidosis
61	Mucolipidosis IV
62	Familial Mediterranean Fever
63	Megalencephalic Leukoencephalopathy with Subcortical Cysts
64	Congenital Disorder of Glycosylation Type Ib
65	Nijmegen Breakage Syndrome
66	NEB-related Nemaline Myopathy
67	Niemann-Pick Disease Type C
68	Congenital Finnish Nephrosis
69	Steroid-resistant Nephrotic Syndrome
70	Costeff Optic Atrophy Syndrome
71	Phenylalanine Hydroxylase Deficiency
72	PCDH15-related Disorders
73	PEX1-related Zellweger Syndrome Spectrum
74	Rhizomelic Chondrodysplasia Punctata Type 1
75	PKHD1-related Autosomal Recessive Polycystic Kidney Disease
76	Congenital Disorder of Glycosylation Type Ia
77	Muscle-eye-brain Disease
78	PPT1-related Neuronal Ceroid Lipofuscinosis
79	PROP1-related Combined Pituitary Hormone Deficiency
80	Glycogen Storage Disease Type V
81	Cartilage-hair Hypoplasia
82	X-linked Juvenile Retinoschisis
83	ARSACS
84	Alpha-1-Antitrypsin Deficiency
85	Alpha-sarcoglycanopathy
86	Beta-sarcoglycanopathy
87	Andermann Syndrome
88	Salla Disease
89	Primary Carnitine Deficiency
90	Sulfate Transporter-related Osteochondrodysplasia
91	Pendred Syndrome
92	Glycogen Storage Disease Type Ib
93	Spinal Muscular Atrophy
94	Niemann-Pick Disease, SMPD1-associated
95	Segawa Syndrome
96	Joubert Syndrome 2
97	TPP1-related Neuronal Ceroid Lipofuscinosis
98	Ataxia with Vitamin E Deficiency
99	Cohen Syndrome