Table S1. Diseases and genes included in the ECS panel.

No.	Disease	Gene	Inheritance pattern
1	Wilson Disease	*ATP7B*	AR
2	Primary Carnitine Deficiency	*SLC22A5*	AR
3	Phenylketonuria	*PAH*	AR
4	Hyperphenylalaninemia, BH4-deficient, A	*PTS*	AR
5	MUT-Related Methylmalonic Acidemia	*MMUT*	AR
6	MMAA-Related Methylmalonic Acidemia	*MMAA*	AR
7	MMAB-Related Methylmalonic Acidemia	*MMAB*	AR
8	Methylmalonic Aciduria and Homocystinuria cblC type	*MMACHC*	AR
9	Methylmalonic Aciduria and Homocystinuria cblD type	*MMADHC*	AR
10	Homocystinuria-Megaloblastic Anemia cblE type	*MTRR*	AR
11	Homocystinuria-Megaloblastic Anemia cblG type	*MTR*	AR
12	MCEE-Related Methylmalonic Acidemia	*MCEE*	AR
13	Homocystinuria Due to Cystathionine Beta-Synthase Deficiency	*CBS*	AR
14	Glutaric Acidemia I	*GCDH*	AR
15	Glutaric acidemia IIA	*ETFA*	AR
16	Glutaric acidemia IIB	*ETFB*	AR
17	Glutaric acidemia IIC	*ETFDH*	AR
18	Acyl-CoA Dehydrogenase Deficiency, Medium-Chain	*ACADM*	AR
19	Acyl-CoA Dehydrogenase Deficiency, Very Long-Chain	*ACADVL*	AR
20	Citrullinemia	*ASS1*	AR
21	Isovaleric Acidemia	*IVD*	AR
22	Propionic Acidemia	*PCCA*, *PCCB*	AR
23	Glycogen Storage Disease Type Ia	*G6PC*	AR
24	Glycogen Storage Disease Type Ib	*SLC37A4*	AR
25	Glycogen Storage Disease Type Ic	*SLC37A4*	AR
26	Glycogen Storage Disease Type II	*GAA*	AR
27	Glycogen Storage Disease type IV	*GBE1*	AR
28	Niemann-Pick Disease Type A	*SMPD1*	AR
29	Niemann-Pick Disease Type B	*SMPD1*	AR
30	Niemann-Pick Disease Type C1	*NPC1*	AR
31	Niemann-Pick Disease Type C2	*NPC2*	AR
32	Maple Syrup Urine Disease Type 1A	*BCKDHA*	AR
33	Maple Syrup Urine Disease Type 1B	*BCKDHB*	AR
34	Maple Syrup Urine Disease, type 2	*DBT*	AR
35	Maple Syrup Urine Disease Type 3	*DLD*	AR
36	Hurler Syndrome	*IDUA*	AR
37	Hurler-Scheie Syndrome	*IDUA*	AR
38	Mucopolysaccharidosis type V	*IDUA*	AR
39	Mucopolysaccharidosis II	*IDS*	XL

40	Mucopolysaccharidosis Type IIIA	*SGSH*	AR
41	Mucopolysaccharidosis Type IIIB	*NAGLU*	AR
42	Mucopolysaccharidosis type IIIC	*HGSNAT*	AR
43	Mucopolysaccharidosis type IIID	*GNS*	AR
44	Mucopolysaccharidosis type IVA	*GALNS*	AR
45	Mucopolysaccharidosis type IVB	*GLB1*	AR
46	Mucopolysaccharidosis type VI	*ARSB*	AR
47	Tyrosinemia Type 1	*FAH*	AR
48	Fabry Disease	*GLA*	XL
49	Biotinidase Deficiency	*BTD*	AR
50	Holocarboxylase Synthetase Deficiency	*HLCS*	AR
51	Hyperornithinemia-Hyperammonemia-Homocitrullinuria Syndrome	*SLC25A15*	AR
52	Carbamoylphosphate Synthetase I Deficiency	*CPS1*	AR
53	Ornithine Transcarbamylase Deficiency	*OTC*	XL
54	Argininosuccinic aciduria	*ASL*	AR
55	Glycine encephalopathy	*AMT*, *GLDC*	AR
56	3-Hydroxy-3-Methylglutaryl-CoA Synthase 2 Deficiency	*HMGCS2*	AR
57	Congenital Disorders of Glycosylation Ia	*PMM2*	AR
58	Peroxisome biogenesis disorder 1A(Zellweger)	*PEX1*	AR
59	Krabbe Disease	*GALC*	AR
60	Familial Hyperinsulinemic Hypoglycemia 2	*KCNJ11*	AR
61	Familial Hyperinsulinemic Hypoglycemia 4	*HADH*	AR
62	Hypophosphatasia, infantile	*ALPL*	AR
63	Hypophosphatasia, childhood	*ALPL*	AR
64	Metachromatic Leukodystrophy due to Arylsulfatase A	*ARSA*	AR
65	Galactosemia	*GALT*	AR
66	Alpha-Mannosidosis	*MAN2B1*	AR
67	Beta-Ketothiolase Deficiency	*ACAT1*	AR
68	Adenosine Deaminase Deficiency	*ADA*	AR
69	Sitosterolemia	*ABCG5, ABCG8*	AR
70	Molybdenum Cofactor Deficiency A	*MOCS1*	AR
71	Hereditary Fructose Intolerance	*ALDOB*	AR
72	Tay-Sachs Disease	*HEXA*	AR
73	Smith-Lemli-Opitz syndrome	*DHCR7*	AR
74	Duchenne Muscular Dystrophy	*DMD*	XL
75	Spinal Muscular Atrophy	*SMN1*	AR
76	Joubert Syndrome 2	*TMEM216*	AR
77	Joubert Syndrome 3	*AHI1*	AR
78	Joubert Syndrome 5	*CEP290*	AR
79	Joubert Syndrome 6	*TMEM67*	AR
80	Joubert Syndrome 9	*CC2D2A*	AR

81	Joubert Syndrome 17	*CPLANE1*	AR
82	X-Linked Centronuclear Myopathy	*MTM1*	XL
83	Neuronal Ceroid-Lipofuscinoses 1	*PPT1*	AR
84	Neuronal Ceroid-Lipofuscinoses 2	*TPP1*	AR
85	Neuronal Ceroid-Lipofuscinoses 3	*CLN3*	AR
86	Neuronal Ceroid-Lipofuscinoses 4A	*CLN6*	AR
87	Neuronal Ceroid-Lipofuscinoses 5	*CLN5*	AR
88	Neuronal Ceroid-Lipofuscinoses 6	*CLN6*	AR
89	Neuronal Ceroid-Lipofuscinoses 7	*MFSD8*	AR
90	Limb-Girdle Muscular Dystrophy type 2A	*CAPN3*	AR
91	Limb-Girdle Muscular Dystrophy type 2B	*DYSF*	AR
92	Limb-Girdle Muscular Dystrophy type 2C	*SGCG*	AR
93	Limb-Girdle Muscular Dystrophy type 2D	*SGCA*	AR
94	Megalencephalic Leukoencephalopathy with Subcortical Cysts 1	*MLC1*	AR
95	Canavan Disease	*ASPA*	AR
96	Autosomal Recessive Osteopetrosis 1	*TCIRG1*	AR
97	Oculocutaneous Albinism Type 1	*TYR*	AR
98	Oculocutaneous Albinism Type 2	*OCA2*	AR
99	Oculocutaneous Albinism Type 3	*TYRP1*	AR
100	Oculocutaneous Albinism Type 4	*SLC45A2*	AR
101	Oculocutaneous Albinism Type 6	*SLC24A5*	AR
102	Oculocutaneous Albinism Type 7	*LRMDA*	AR
103	X-Linked Ocular Albinism	*GPR143*	XL
104	Hermansky-Pudlak Syndrome 1	*HPS1*	AR
105	Hermansky-Pudlak Syndrome 3	*HPS3*	AR
106	Autosomal Recessive Congenital Ichthyosis 1	*TGM1*	AR
107	Autosomal Recessive Congenital Ichthyosis 4A	*ABCA12*	AR
108	Autosomal Recessive Congenital Ichthyosis 4B	*ABCA12*	AR
109	Netherton syndrome	*SPINK5*	AR
110	Sjögren-Larsson syndrome	*ALDH3A2*	AR
111	LAMA3-Related Junctional Epidermolysis Bullosa	*LAMA3*	AR
112	LAMB3-Related Junctional Epidermolysis Bullosa	*LAMB3*	AR
113	LAMC2-Related Junctional Epidermolysis Bullosa	*LAMC2*	AR
114	Non-Herlitz type Junctional Epidermolysis Bullosa	*COL17A1*	AR
115	Autosomal Recessive Epidermolysis Bullosa Dystrophica	*COL7A1*	AR
116	Hemophilia B	*F9*	XL
117	Alpha (α)-thalassemia	*HBA1*, *HBA2*	AR
118	Beta (β)-thalassemia	*HBB*	AR
119	Sickle Cell Anemia	*HBB*	AR

120	Fanconi anemia, complementation group A	*FANCA*	AR
121	Fanconi anemia, complementation group C	*FANCC*	AR
122	Fanconi anemia, complementation group D2	*FANCD2*	AR
123	Fanconi anemia, complementation group G	*FANCG*	AR
124	Fanconi anemia, complementation group I	*FANCI*	AR
125	Hemophagocytic lymphohistiocytosis, familial, 2	*PRF1*	AR
126	Hemophagocytic lymphohistiocytosis, familial, 3	*UNC13D*	AR
127	Hemophagocytic lymphohistiocytosis, familial, 4	*STX11*	AR
128	Hemophagocytic lymphohistiocytosis, familial, 5	*STXBP2*	AR
129	Omenn syndrome	*RAG1*, *RAG2*	AR
130	Severe combined immunodeficiency, B cell-negative	*RAG1*, *RAG2*	AR
131	X-Linked Severe Combined Immunodeficiency	*IL2RG*	XL
132	X-Linked Adrenal Hypoplasia Congenita	*NR0B1*	XL
133	Progressive Familial Intrahepatic Cholestasis 2	*ABCB11*	AR
134	Progressive Familial Intrahepatic Cholestasis 3	*ABCB4*	AR
135	Progressive Familial Intrahepatic Cholestasis 4	*TJP2*	AR
136	Alport syndrome 2, autosomal recessive	*COL4A3*, *COL4A4*	AR
137	Nephrotic syndrome, type 1	*NPHS1*	AR
138	Nephronophthisis 3	*NPHP3*	AR
139	Nephronophthisis 11	*TMEM67*	AR
140	Nephropathic Cystinosis	*CTNS*	AR
141	Cystic Fibrosis	*CFTR*	AR
142	Autosomal Recessive Deafness 1A	*GJB2*	AR
143	Autosomal Recessive Deafness 4, with Enlarged Vestibular Aqueduct	*SLC26A4*	AR
144	Wolfram Syndrome 1	*WFS1*	AR
145	Ellis-van Creveld Syndrome	*EVC2*	AR
146	Osteoporosis-Pseudoglioma Syndrome	*LRP5*	AR
147	Immunodeficiency-Centromeric instability-Facial anomalies syndrome 1	*DNMT3B*	AR
148	Meckel Syndrome 2	*TMEM216*	AR
149	Meckel Syndrome 3	*TMEM67*	AR
150	Meckel Syndrome 4	*CEP290*	AR
151	X-Linked Hypohidrotic Ectodermal Dysplasia	*EDA*	XL
152	COACH Syndrome	*TMEM67*, *CC2D2A*	AR