STID: List of Disorders

11-beta-hydroxylase-deficient Congenital Adrenal Hyperplasia	Inclusion Body Myopathy 2
21-hydroxylase-deficient Congenital Adrenal Hyperplasia	Isovaleric Acidemia
6-pyruvoyl-tetrahydropterin Synthase Deficiency	Joubert Syndrome 2
ABCC8-related Hyperinsulinism	KCNJ11-related Familial Hyperinsulinism
Adenosine Deaminase Deficiency	Krabbe Disease
Alpha Thalassemia	LAMA2-related Muscular Dystrophy
Alpha-mannosidosis	Leigh Syndrome, French-Canadian Type
Alpha-sarcoglycanopathy	Lipoamide Dehydrogenase Deficiency
Alstrom Syndrome	Lipoid Congenital Adrenal Hyperplasia
AMT-related Glycine Encephalopathy	Lysosomal Acid Lipase Deficiency
Andermann Syndrome	Maple Syrup Urine Disease Type 1B
Argininemia	Maple Syrup Urine Disease Type Ia
Argininosuccinic Aciduria	Maple Syrup Urine Disease Type II
ARSACS	Medium Chain Acyl-CoA Dehydrogenase Deficiency
Aspartylglycosaminuria	Megalencephalic Leukoencephalopathy with Subcortical Cysts
Ataxia with Vitamin E Deficiency	Metachromatic Leukodystrophy
Ataxia-telangiectasia	Methylmalonic Acidemia, cblA Type
ATP7A-related Disorders	Methylmalonic Acidemia, cblB Type
Autosomal Recessive Osteopetrosis Type 1	Methylmalonic Aciduria and Homocystinuria, cblC Type
Bardet-Biedl Syndrome, BBS1-related	MKS1-related Disorders
Bardet-Biedl Syndrome, BBS10-related	Mucolipidosis III Gamma
Bardet-Biedl Syndrome, BBS12-related	Mucolipidosis IV
Bardet-Biedl Syndrome, BBS2-related	Mucopolysaccharidosis Type I
Beta-sarcoglycanopathy	Mucopolysaccharidosis Type II
Biotinidase Deficiency	Mucopolysaccharidosis Type IIIA
Bloom Syndrome	Mucopolysaccharidosis Type IIIB
Calpainopathy	Mucopolysaccharidosis Type IIIC
Canavan Disease	Muscle-eye-brain Disease
Carbamoylphosphate Synthetase I Deficiency	MUT-related Methylmalonic Acidemia
Carnitine Palmitoyltransferase IA Deficiency	MYO7A-related Disorders
Carnitine Palmitoyltransferase II Deficiency	NEB-related Nemaline Myopathy
Cartilage-hair Hypoplasia	Niemann-Pick Disease Type C
Cerebrotendinous Xanthomatosis	Niemann-Pick Disease Type C2
Citrullinemia Type 1	Niemann-Pick Disease, SMPD1-associated
CLN3-related Neuronal Ceroid Lipofuscinosis	Nijmegen Breakage Syndrome
CLN5-related Neuronal Ceroid Lipofuscinosis	Northern Epilepsy
CLN6-related Neuronal Ceroid Lipofuscinosis	Ornithine Transcarbamylase Deficiency
Cohen Syndrome	PCCA-related Propionic Acidemia
COL4A3-related Alport Syndrome	PCCB-related Propionic Acidemia
COL4A4-related Alport Syndrome	PCDH15-related Disorders
Congenital Disorder of Glycosylation Type Ia	Pendred Syndrome
Congenital Disorder of Glycosylation Type Ib	Peroxisome Biogenesis Disorder Type 3
Congenital Disorder of Glycosylation Type Ic	Peroxisome Biogenesis Disorder Type 4
Congenital Finnish Nephrosis	Peroxisome Biogenesis Disorder Type 5
Costeff Optic Atrophy Syndrome	Peroxisome Biogenesis Disorder Type 6
Cystic Fibrosis	PEX1-related Zellweger Syndrome Spectrum
Cystinosis	Phenylalanine Hydroxylase Deficiency
D-bifunctional Protein Deficiency	PKHD1-related Autosomal Recessive Polycystic Kidney Disease
Delta-sarcoglycanopathy	Polyglandular Autoimmune Syndrome Type 1
Dysferlinopathy	Pompe Disease
Dystrophinopathy (Including Duchenne/Becker Muscular Dystrophy)	PPT1-related Neuronal Ceroid Lipofuscinosis
ERCC6-related Disorders	Primary Carnitine Deficiency
ERCC8-related Disorders	Primary Hyperoxaluria Type 1
EVC-related Ellis-van Creveld Syndrome	Primary Hyperoxaluria Type 2
EVC2-related Ellis-van Creveld Syndrome	Primary Hyperoxaluria Type 3
Fabry Disease	PROP1-related Combined Pituitary Hormone Deficiency
Familial Dysautonomia	Pycnodysostosis
Familial Mediterranean Fever	Pyruvate Carboxylase Deficiency
Fanconi Anemia Complementation Group A	Rhizomelic Chondrodysplasia Punctata Type 1

Fanconi Anemia Type C	RTEL1-related Disorders
FKRP-related Disorders	Salla Disease
FKTN-related Disorders	Sandhoff Disease
Fragile X Syndrome	Segawa Syndrome
Galactokinase Deficiency	Short Chain Acyl-CoA Dehydrogenase Deficiency
Galactosemia	Sjogren-Larsson Syndrome
Gamma-sarcoglycanopathy	Smith-Lemli-Opitz Syndrome
Gaucher Disease	Spastic Paraplegia Type 15
GJB2-related DFNB1 Nonsyndromic Hearing Loss and Deafness	Spinal Muscular Atrophy
GLB1-related Disorders	Spondylothoracic Dysostosis
GLDC-related Glycine Encephalopathy	Steroid-resistant Nephrotic Syndrome
Glutaric Acidemia Type 1	Sulfate Transporter-related Osteochondrodysplasia
Glycogen Storage Disease Type Ia	TGM1-related Autosomal Recessive Congenital Ichthyosis
Glycogen Storage Disease Type Ib	TPP1-related Neuronal Ceroid Lipofuscinosis
Glycogen Storage Disease Type III	Tyrosinemia Type I
GNPTAB-related Disorders	Tyrosinemia Type II
GRACILE Syndrome	USH1C-related Disorders
HADHA-related Disorders	USH2A-related Disorders
Hb Beta Chain-related Hemoglobinopathy (Including BetaThalassemia and Sickle Cell Disease)	Usher Syndrome Type 3
Hereditary Fructose Intolerance	Very Long Chain Acyl-CoA Dehydrogenase Deficiency
Herlitz Junctional Epidermolysis Bullosa, LAMA3-related	Wilson Disease
Herlitz Junctional Epidermolysis Bullosa, LAMB3-related	X-linked Adrenoleukodystrophy
Herlitz Junctional Epidermolysis Bullosa, LAMC2-related	X-linked Alport Syndrome
Hexosaminidase A Deficiency (Including Tay-Sachs Disease)	X-linked Congenital Adrenal Hypoplasia
HMG-CoA Lyase Deficiency	X-linked Juvenile Retinoschisis
Holocarboxylase Synthetase Deficiency	X-linked Myotubular Myopathy
Homocystinuria Caused by Cystathionine Beta-synthase Deficiency	X-linked Severe Combined Immunodeficiency
Hydrolethalus Syndrome	Xeroderma Pigmentosum Group A
Hypophosphatasia, Autosomal Recessive	Xeroderma Pigmentosum Group C