Classifications of Disorders

updated 1/2020

Type of Disorder	Disorders	Abbreviation
Amino Acid Disorders	Argininemia, Arginase Deficiency	ARG
	Arginosuccinic Aciduria	ASA
	Benign Hyperphenylalaninemia	PHE
	Biopterin Defect in Cofactor Biosynthesis	BIOPT (BS)
	Biopterin Defect in Cofactor Regeneration	BIOPT (Reg)
	Carbamoyltransferase Deficiency, Carbamoyl Phosphate Synthetase I Deficiency	CPS
	Citrullinemia Type I	CIT I
	Citrullinemia Type II	CIT II
	Homocystinuria	HCY
	Homocystinuria due to MTHFR (5,10-methylenetetrahydrofolate reductasedeficiency, Remethylation Defect)	RMD
	Hypermethionemia	MET
	Maple Syrup Urine Disease	MSUD
	Ornithine Transcarbamylase Deficiency	OTC
	Phenylketonuria (Classic)	PKU
	Pyruvate Carboxylase Deficiency	PC
	Tyrosinemia Type I	TYR I
	Tyrosinemia Type II	TYR II
	Tyrosinemia Type III	TYR III
Disorders of Galactose Metabolism	Galactoepimerase Deficiency	GALE
	Galactokinase deficiency	GALK
	Galactosemia (Classical)	GALT
Endocrine Disorders	Congenital Adrenal Hyperplasia	CAH
	Congenital Hypothyroidism	CH
Fatty Acid Oxidation Disorders	2,4 Dienoyl CoA Reductase Deficiency	DE RED
	Carnitine Palmitoyl Transferase Deficiency Type 1	CPT I
	Carnitine Palmitoyl Transferase Deficiency Type 2	CPT II
	Carnitine Uptake Defect	CUD
	Carnitine/Acylcarnitine Translocase Deficiency	CACT
	Formiminoglutamic Acidemia, Glutamate Formiminotransferase Deficiency (FIGLU)	FIGLU
	Glutaric Acidemia Type 2	GA I
	Isobutyryl-CoA dehydrogenase deficiency	IBG
	Long Chain Hydroxyacyl-CoA Dehydrogenase Deficiency	LCHAD
	Medium Chain Acyl-CoA Dehydrogenase Deficiency	MCAD
	Medium/Short Chain 3-Hydroxyacyl-CoA Dehydrogenase Deficiency	M/SCHAD
	Short Chain Acyl-CoA Dehydrogenase Deficiency	SCAD
	Trifunctional Protein Deficiency	TFP
	Very Long Chain Acyl-CoA Dehydrogenase Deficiency	VLCAD
Hemoglobin Disorders	Alpha-Thalassemia (Bart's Hb)	Hemoglobin Bart's
	Hemoglobinopathies (Various other)	Hgb Var
	Other Hemoglobinopathies	Variant Hemoglobins
	S, Beta-Thalassemia	Hemoglobin Sβ° Thal
	S, βeta-thalassemia (Sickle Beta Thalassemia)	Sβ-Thal

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Type of Disorder	Disorders	Abbreviation
	S,C Disease	Hemoglobin SC
	S,S Disease (Sickle Cell Disease)	Hemoglobin S
Hemoglobin Traits	Hemoglobin C Trait (Carrier)	Hgb FAC
	Hemoglobin D Trait (Carrier)	Hgb FAD
	Hemoglobin E Trait (Carrier)	Hgb FAE
	Hemoglobin S Trait (Carrier)	Hgb FAS
Immune Disorders	Severe Combined Immunodeficiency Syndrome	SCID
	T-cell related lymphocyte deficiencies	T-Cell Lymphopenias
Metabolic Disorder of Biotin Recycling	Biotinidase Deficiency	BIO
Organic Acid Disorders	2-Methyl 3 Hydroxy Butyric Aciduria	2M3HBA
	2-Methyl Butyryl-CoA Dehydrogenase Deficiency	2MBG
	3-Hydroxy-3-Methylglutaryl CoA Lyase Deficiency	HMG
	3-Methylcrotonyl CoA Carboxylase Deficiency	3MCC
	3-Methylglutaconic Aciduria	3MGA
	Beta-Ketothiolase Deficiency	BKT
	Ethylmalonic Encephalopathy	EME
	Glutaric Acidemia Type 1	GA I
	Isovaleric Acidemia	IVA
	Malonic Aciduria	MAL
	Methylmalonic Acidemia	MMA
	Methylmalonic Acidemia caused by cobalamin A or cobalamin B deficiencies	Cbl A, B
	Methylmalonic Acidemia with Homocystinuria (Cbl C, D)	Cbl C, D
	Multiple CoA Carboxylase Deficiency	MCD
	Propionic Acidemia	PPA
Other Disorders	Critical Congenital Heart Disease	CCHD
	Cystic Fibrosis	CF
	Hearing Loss	Hearing
Peroxisomal Disorders	X-Linked Adrenoleukodystrophy	ALD
Neuromuscular Disorders	Spinal Muscular Atrophy	SMA