Disorders Alphabetical by Disease

updated 1/2020

Disorders	Abbreviation	Classification	Recommended Uniform Screening Panel (RUSP) Classification
2,4 Dienoyl CoA Reductase Deficiency	DE RED	Fatty Acid Oxidation Disorder	Secondary Condition
2-Methyl 3 Hydroxy Butyric Aciduria	2M3HBA	Organic Acid Disorder	Secondary Condition
2-Methyl Butyryl-CoA Dehydrogenase Deficiency	2MBG	Organic Acid Disorder	Secondary Condition (called 2-Methylbutyrylglycinuria on RUSP)
3-Hydroxy-3-Methylglutaryl CoA Lyase Deficiency	HMG	Organic Acid Disorder	Core Condition
3-Methylcrotonyl CoA Carboxylase Deficiency	3MCC	Organic Acid Disorder	Core Condition
3-Methylglutaconic Aciduria	3MGA	Organic Acid Disorder	Secondary Condition
Alpha-Thalassemia (Bart's Hb)	Hemoglobin Bart's	Hemoglobin Disorder	Secondary Conditoin
Argininemia, Arginase Deficiency	ARG	Amino Acid Disorder	Secondary Condition
Arginosuccinic Aciduria	ASA	Amino Acid Disorder	Core Condition
Benign Hyperphenylalaninemia	PHE	Amino Acid Disorder	Secondary Condition
Beta-Ketothiolase Deficiency	BKT	Organic Acid Disorder	Core Condition
Biopterin Defect in Cofactor Biosynthesis	BIOPT (BS)	Amino Acid Disorder	Secondary Condition
Biopterin Defect in Cofactor Regeneration	BIOPT (Reg)	Amino Acid Disorder	Secondary Condition
Biotinidase Deficiency	BIO	Metabolic Disorder of Biotin Recycling	Core Condition
Carbamoyltransferase Deficiency, Carbamoyl Phosphate Synthetase I Deficiency	CPS	Amino Acid Disorder	Not on RUSP
Carnitine Palmitoyl Transferase Deficiency Type 1	CPT I	Fatty Acid Oxidation Disorder	Secondary Condition
Carnitine Palmitoyl Transferase Deficiency Type 2	CPT II	Fatty Acid Oxidation Disorder	Secondary Condition
Carnitine Uptake Defect	CUD	Fatty Acid Oxidation Disorder	Secondary Condition
Carnitine/Acylcarnitine Translocase Deficiency	CACT	Fatty Acid Oxidation Disorder	Secondary Condition
Citrullinemia Type I	CIT I	Amino Acid Disorder	Core Condition
Citrullinemia Type II	CIT II	Amino Acid Disorder	Secondary Condition
Congenital Adrenal Hyperplasia	CAH	Endocrine Disorder	Core Condition
Congenital Hypothyroidism	CH	Endocrine Disorder	Core Condition
Critical Congenital Heart Disease	CCHD	Other	Core Condition
Cystic Fibrosis	CF	Other	Core Condition
Ethylmalonic Encephalopathy	EME	Organic Acid Disorder	Not on RUSP
Formiminoglutamic Acidemia, Glutamate Formiminotransferase Deficiency (FIGLU)	FIGLU	Fatty Acid Oxidation Disorder	Not on RUSP
Galactoepimerase Deficiency	GALE	Disorder of Galactose Metabolism	Secondary Condition
Galactokinase deficiency	GALK	Disorder of Galactose Metabolism	Secondary Condition
Galactosemia (Classical)	GALT	Disorder of Galactose Metabolism	Core Condition
Glutaric Acidemia Type 1	GA I	Organic Acid Disorder	Core Condition
Glutaric Acidemia Type 2	GA II	Fatty Acid Oxidation Disorder	Secondary Condition
Hearing Loss	Hearing	Other	Core Condition
Hemoglobin C Trait (Carrier)	Hgb FAC	Hemoglobin Trait	Not on RUSP
Hemoglobin D Trait (Carrier)	Hgb FAD	Hemoglobin Trait	Not on RUSP
Hemoglobin E Trait (Carrier)	Hgb FAE	Hemoglobin Trait	Not on RUSP
Hemoglobin S Trait (Carrier)	Hgb FAS	Hemoglobin Trait	Not on RUSP
Hemoglobinopathies (Various other)	Hgb Var	Hemoglobin Disorder	Secondary Condition
Homocystinuria	HCY	Amino Acid Disorder	Core Condition
Homocystinuria due to MTHFR (5,10-methylenetetrahydrofolate reductasedeficiency, Remethylation Defect)	RMD	Amino Acid Disorder	Not on RUSP

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Hypermethionemia	MET	Amino Acid Disorder	Secondary Condition
Isobutyryl-CoA dehydrogenase deficiency	IBG	Fatty Acid Oxidation Disorder	Secondary Condition
Isovaleric Acidemia	IVA	Organic Acid Disorder	Core Condition
Long Chain Hydroxyacyl-CoA Dehydrogenase Deficiency	LCHAD	Fatty Acid Oxidation Disorder	Core Condition
Malonic Aciduria	MAL	Organic Acid Disorder	Secondary Conditions
Maple Syrup Urine Disease	MSUD	Amino Acid Disorder	Core Condition
Medium Chain Acyl-CoA Dehydrogenase Deficiency	MCAD	Fatty Acid Oxidation Disorder	Core Condition
Medium/Short Chain 3-Hydroxyacyl-CoA Dehydrogenase Deficiency	M/SCHAD	Fatty Acid Oxidation Disorder	Secondary Condition
Methylmalonic Acidemia	MMA	Organic Acid Disorder	Core Condition
Methylmalonic Acidemia caused by cobalamin A or cobalamin B deficiencies	Cbl A, B	Organic Acid Disorder	Core Condition
Methylmalonic Acidemia with Homocystinuria (Cbl C, D)	Cbl C, D	Organic Acid Disorder	Core Condition
Multiple CoA Carboxylase Deficiency	MCD	Organic Acid Disorder	Core Condition (Called Holocarboxylase Synthetase deficiency on RUSP)
Ornithine Transcarbamylase Deficiency	OTC	Amino Acid Disorder	Not on RUSP
Other Hemoglobinopathies	Variant Hemoglobins	Hemoglobin Disorder	Secondary Condition
Phenylketonuria (Classic)	PKU	Amino Acid Disorder	Core Condition
Propionic Acidemia	PPA	Organic Acid Disorder	Core Condition
Pyruvate Carboxylase Deficiency	PC	Amino Acid Disorder	Not on RUSP
S, Beta-Thalassemia	Hemoglobin Sβ° Thal	Hemoglobin Disorder	Core Conditoin
S, βeta-thalassemia (Sickle Beta Thalassemia)	Sβ-Thal	Hemoglobin Disorder	Core Condition
S,C Disease	Hemoglobin SC	Hemoglobin Disorder	Core Condition
S,S Disease (Sickle Cell Disease)	Hemoglobin S	Hemoglobin Disorder	Core Condition
Severe Combined Immunodeficiency Syndrome	SCID	Immune Disorder	Core Condition
Short Chain Acyl-CoA Dehydrogenase Deficiency	SCAD	Fatty Acid Oxidation Disorder	Secondary Condition
Spinal Muscular Atrophy	SMA	Neuromuscular Disorder	Core Condition
T-cell related lymphocyte deficiencies	T-Cell Lymphopenias	Immune Disorder	Secondary Condition
Trifunctional Protein Deficiency	TFP	Fatty Acid Oxidation Disorder	Core Condition
Tyrosinemia Type I	TYR I	Amino Acid Disorder	Core Condition
Tyrosinemia Type II	TYR II	Amino Acid Disorder	Secondary Condition
Tyrosinemia Type III	TYR III	Amino Acid Disorder	Secondary Condition
Very Long Chain Acyl-CoA Dehydrogenase Deficiency	VLCAD	Fatty Acid Oxidation Disorder	Core Condition
X-Linked Adrenoleukodystrophy	ALD	Peroxisomal Disorder	Core Condition