Disorders Alphabetical by Abbreviation

updated 1-2020

Abbreviation	Disorders	Classification	Recommended Uniform Screening Panel (RUSP) Classification
2M3HBA	2-Methyl 3 Hydroxy Butyric Aciduria	Organic Acid Disorder	Secondary Condition
2MBG	2-Methyl Butyryl-CoA Dehydrogenase Deficiency	Organic Acid Disorder	Secondary Condition (called 2-Methylbutyrylglycinuria on RUSP)
3MCC	3-Methylcrotonyl CoA Carboxylase Deficiency	Organic Acid Disorder	Core Condition
3MGA	3-Methylglutaconic Aciduria	Organic Acid Disorder	Secondary Condition
ALD	X-Linked Adrenoleukodystrophy	Peroxisomal Disorder	Core Condition
ARG	Argininemia, Arginase Deficiency	Amino Acid Disorder	Secondary Condition
ASA	Arginosuccinic Aciduria	Amino Acid Disorder	Core Condition
BIO	Biotinidase Deficiency	Metabolic Disorder of Biotin Recycling	Core Condition
BIOPT (BS)	Biopterin Defect in Cofactor Biosynthesis	Amino Acid Disorder	Secondary Condition
BIOPT (Reg)	Biopterin Defect in Cofactor Regeneration	Amino Acid Disorder	Secondary Condition
BKT	Beta-Ketothiolase Deficiency	Organic Acid Disorder	Core Condition
CACT	Carnitine/Acylcarnitine Translocase Deficiency	Fatty Acid Oxidation Disorder	Secondary Condition
CAH	Congenital Adrenal Hyperplasia	Endocrine Disorder	Core Condition
Cbl A, B	Methylmalonic Acidemia caused by cobalamin A or cobalamin B deficiencies	Organic Acid Disorder	Core Condition
Cbl C, D	Methylmalonic Acidemia with Homocystinuria (Cbl C, D)	Organic Acid Disorder	Core Condition
CCHD	Critical Congenital Heart Disease	Other	Core Condition
CF	Cystic Fibrosis	Other	Core Condition
CH	Congenital Hypothyroidism	Endocrine Disorder	Core Condition
CIT II	Citrullinemia Type II	Amino Acid Disorder	Secondary Condition
CIT I	Citrullinemia Type I	Amino Acid Disorder	Core Condition
CPS	Carbamoyltransferase Deficiency, Carbamoyl Phosphate Synthetase I Deficiency	Amino Acid Disorder	Not on RUSP
CPT I	Carnitine Palmitoyl Transferase Deficiency Type 1	Fatty Acid Oxidation Disorder	Secondary Condition
CPT II	Carnitine Palmitoyl Transferase Deficiency Type 2	Fatty Acid Oxidation Disorder	Secondary Condition
CUD	Carnitine Uptake Defect	Fatty Acid Oxidation Disorder	Secondary Condition
DE RED	2,4 Dienoyl CoA Reductase Deficiency	Fatty Acid Oxidation Disorder	Secondary Condition
EME	Ethylmalonic Encephalopathy	Organic Acid Disorder	Not on RUSP
FIGLU	Formiminoglutamic Acidemia, Glutamate Formiminotransferase Deficiency (FIGLU)	Fatty Acid Oxidation Disorder	Not on RUSP
GA I	Glutaric Acidemia Type 1	Organic Acid Disorder	Core Condition
GA II	Glutaric Acidemia Type 2	Fatty Acid Oxidation Disorder	Secondary Condition
GALE	Galactoepimerase Deficiency	Disorder of Galactose Metabolism	Secondary Condition
GALK	Galactokinase deficiency	Disorder of Galactose Metabolism	Secondary Condition
GALT	Galactosemia (Classical)	Disorder of Galactose Metabolism	Core Condition
HCY	Homocystinuria	Amino Acid Disorder	Core Condition
Hearing	Hearing Loss	Other	Core Condition
Hemoglobin Bart's	Alpha-Thalassemia (Bart's Hb)	Hemoglobin Disorder	Secondary Conditoin
Hemoglobin S	S,S Disease (Sickle Cell Disease)	Hemoglobin Disorder	Core Condition
Hemoglobin SC	S,C Disease	Hemoglobin Disorder	Core Condition
Hemoglobin Sβ° Thal	S, Beta-Thalassemia	Hemoglobin Disorder	Core Conditoin
Hgb FAC	Hemoglobin C Trait (Carrier)	Hemoglobin Trait	Not on RUSP
Hgb FAD	Hemoglobin D Trait (Carrier)	Hemoglobin Trait	Not on RUSP
Hgb FAE	Hemoglobin E Trait (Carrier)	Hemoglobin Trait	Not on RUSP
Hgb FAS	Hemoglobin S Trait (Carrier)	Hemoglobin Trait	Not on RUSP
Hgb Var	Hemoglobinopathies (Various other)	Hemoglobin Disorder	Secondary Condition
HMG	3-Hydroxy-3-Methylglutaryl CoA Lyase Deficiency	Organic Acid Disorder	Core Condition
IBG	Isobutyryl-CoA dehydrogenase deficiency	Fatty Acid Oxidation Disorder	Secondary Condition
IVA	Isovaleric Acidemia	Organic Acid Disorder	Core Condition
LCHAD	Long Chain Hydroxyacyl-CoA Dehydrogenase Deficiency	Fatty Acid Oxidation Disorder	Core Condition
M/SCHAD	Medium/Short Chain 3-Hydroxyacyl-CoA Dehydrogenase Deficiency	Fatty Acid Oxidation Disorder	Secondary Condition
MAL	Malonic Aciduria	Organic Acid Disorder	Secondary Conditions
MCAD	Medium Chain Acyl-CoA Dehydrogenase Deficiency	Fatty Acid Oxidation Disorder	Core Condition
MCD	Multiple CoA Carboxylase Deficiency	Organic Acid Disorder	Core Condition (Called Holocarboxylase Synthetase deficiency on RUSP)

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MET	Hypermethionemia	Amino Acid Disorder	Secondary Condition
MMA	Methylmalonic Acidemia	Organic Acid Disorder	Core Condition
MSUD	Maple Syrup Urine Disease	Amino Acid Disorder	Core Condition
OTC	Ornithine Transcarbamylase Deficiency	Amino Acid Disorder	Not on RUSP
PC	Pyruvate Carboxylase Deficiency	Amino Acid Disorder	Not on RUSP
PHE	Benign Hyperphenylalaninemia	Amino Acid Disorder	Secondary Condition
PKU	Phenylketonuria (Classic)	Amino Acid Disorder	Core Condition
PPA	Propionic Acidemia	Organic Acid Disorder	Core Condition
RMD	Homocystinuria due to MTHFR (5,10-methylenetetrahydrofolate reductasedeficiency, Remethylation Defect)	Amino Acid Disorder	Not on RUSP
SCAD	Short Chain Acyl-CoA Dehydrogenase Deficiency	Fatty Acid Oxidation Disorder	Secondary Condition
SCID	Severe Combined Immunodeficiency Syndrome	Immune Disorder	Core Condition
Sβ-Thal	S, βeta-thalassemia (Sickle Beta Thalassemia)	Hemoglobin Disorder	Core Condition
T-Cell Lymphopenias	T-cell related lymphocyte deficiencies	Immune Disorder	Secondary Condition
TFP	Trifunctional Protein Deficiency	Fatty Acid Oxidation Disorder	Core Condition
TYR I	Tyrosinemia Type I	Amino Acid Disorder	Core Condition
TYR II	Tyrosinemia Type II	Amino Acid Disorder	Secondary Condition
TYR III	Tyrosinemia Type III	Amino Acid Disorder	Secondary Condition
Variant Hemoglobins	Other Hemoglobinopathies	Hemoglobin Disorder	Secondary Condition
VLCAD	Very Long Chain Acyl-CoA Dehydrogenase Deficiency	Fatty Acid Oxidation Disorder	Core Condition
	Spinal Muscular Atrophy	Neuromuscular Disorder	Core Condition