Disorders by Rusp Classification

updated 1/2020

Recommended Uniform Screening Panel (RUSP) Classification	Disorders	Abbreviation
Core Conditions	Arginosuccinic Aciduria	ASA
	Citrullinemia Type I	CIT I
	Homocystinuria	HCY
	Maple Syrup Urine Disease	MSUD
	Phenylketonuria (Classic)	PKU
	Tyrosinemia Type I	TYR I
	Galactosemia (Classical)	GALT
	Congenital Adrenal Hyperplasia	CAH
	Congenital Hypothyroidism	CH
	Long Chain Hydroxyacyl-CoA Dehydrogenase Deficiency	LCHAD
	Medium Chain Acyl-CoA Dehydrogenase Deficiency	MCAD
	Trifunctional Protein Deficiency	TFP
	Very Long Chain Acyl-CoA Dehydrogenase Deficiency	VLCAD
	S, βeta-thalassemia (Sickle Beta Thalassemia)	Sβ-Thal
	S,C Disease	Hemoglobin SC
	S,S Disease (Sickle Cell Disease)	Hemoglobin S
	Severe Combined Immunodeficiency Syndrome	SCID
	Biotinidase Deficiency	BIO
	3-Hydroxy-3-Methylglutaryl CoA Lyase Deficiency	HMG
	3-Methylcrotonyl CoA Carboxylase Deficiency	3MCC
	Beta-Ketothiolase Deficiency	BKT
	Glutaric Acidemia Type 1	GA I
	Isovaleric Acidemia	IVA
	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
	Propionic Acidemia	PPA
	Critical Congenital Heart Disease	CCHD
	Cystic Fibrosis	CF
	Hearing Loss	Hearing
	X-Linked Adrenoleukodystrophy	ALD
	Multiple CoA Carboxylase Deficiency (Called Holocarboxylase Synthetase deficiency on RUSP)	MCD
	S, Beta-Thalassemia	Hemoglobin Sβ° Thal
	Spinal Muscular Atrophy	SMA
Secondary Conditions	Argininemia, Arginase Deficiency	ARG
	Benign Hyperphenylalaninemia	PHE
	Biopterin Defect in Cofactor Biosynthesis	BIOPT (BS)
	Biopterin Defect in Cofactor Regeneration	BIOPT (Reg)
	Citrullinemia Type II	CIT II
	Hypermethionemia	MET
	Tyrosinemia Type II	TYR II
	Tyrosinemia Type III	TYR III

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	Galactoepimerase Deficiency	GALE
	Galactokinase deficiency	GALK
	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
	Glutaric Acidemia Type 2	GA 2
	Isobutyryl-CoA dehydrogenase deficiency	IBG
	Medium/Short Chain 3-Hydroxyacyl-CoA Dehydrogenase Deficiency	M/SCHAD
	Short Chain Acyl-CoA Dehydrogenase Deficiency	SCAD
	Hemoglobinopathies (Various other)	Hgb Var
	Other Hemoglobinopathies	Variant Hemoglobins
	T-cell related lymphocyte deficiencies	T-Cell Lymphopenias
	2-Methyl 3 Hydroxy Butyric Aciduria	2M3HBA
	3-Methylglutaconic Aciduria	3MGA
	2-Methyl Butyryl-CoA Dehydrogenase Deficiency (called 2-Methylbutyrylglycinuria on RUSP)	2MBG
	Malonic Aciduria	MAL
	Alpha-Thalassemia (Bart's Hb)	Hemoglobin Bart's
Non-RUSP	Carbamoyltransferase Deficiency, Carbamoyl Phosphate Synthetase I Deficiency	CPS
	Homocystinuria due to MTHFR (5,10-methylenetetrahydrofolate reductasedeficiency, Remethylation Defect)	RMD
	Ornithine Transcarbamylase Deficiency	OTC
	Pyruvate Carboxylase Deficiency	PC
	Formiminoglutamic Acidemia, Glutamate Formiminotransferase Deficiency (FIGLU)	FIGLU
	Hemoglobin C Trait (Carrier)	Hgb FAC
	Hemoglobin D Trait (Carrier)	Hgb FAD
	Hemoglobin E Trait (Carrier)	Hgb FAE
	Hemoglobin S Trait (Carrier)	Hgb FAS
	Ethylmalonic Encephalopathy	EME