Disorders Recommended by US Department of Health and Human Services

Secretary's Advisory on Heritable and Genetic Disorders

Amino Acid Metabolic Disorder	***Annual Incidence (National)***
PKU – Phenylketonuria	1/ 13,947
MSUD - Maple Syrup Urine Disease	1/250,000
HCY - Homocystinuria	1/300,000
CIT - Citrullinemia	1/60,000
ASA - Arginosuccinic	1/60,000
TYR - Tyrosinemia	1/63,000
Organic Acid Metabolism Disorders	
IVA - Isovaleric Acidemia	1/50,000
GA – Glutaric Acidemia	1/30,000
HMG 3 - Hydroxy 3 Methylglutaryl -CoA Lyase	1/300,000
MCD - Multiple Carboxylase Deficiency	1/50,000
MUT - Methylmalonic acidemia due to Mutase deficiency	1/50,000
Cbl A,B - Methylmalonic acidemia cbl a and cbl b forms	1/200,000
3MCC - 3 Methylcrotonyl - COA Carboxylase deficiency	1/75,000
PROP - Propionic acidemia	1/50,000
BKT - Beta - Ketothiolase deficiency	1/200,000
Fatty Acid Oxidation Disorders	
MCAD - Medium - Chain acyl - CoA dehydrogenase deficiency	1/10,000
VLCAD - Very Long Chain acyl - CoA dehydrogenase deficiency	1/200,000
LCHAD - Long - Chain 3-OH acyl - CoA dehydrogenase deficiency	1/50,000
TFP - Trifunctional protein deficiency	1/200,000
CUD - Carnitine uptake defect	1/200,000
Hemoglobinopathies	
Hb SS - Sickle Cell Anemia	1/3,721 1/400 in African Americans
Hb S/Th - Hb S/ Beta - Thalassemia	1/25,000
Hb S/C - Hb S/C disease	1/20,000
Others	
CH - Congenital hypothyroidism	1/3,044
BIOT - Biotinidase deficiency	1/72,000
CAH - Congenital adrenal hyperplasia	1/20,000
GALT - Classical Galactosemia	1/50,000
HEAR - Hearing loss	3/1,000
CF - Cystic Fibrosis	1/5,000