

UNIFORM CONDITION PANEL
RECOMMENDED BY AMERICAN COLLEGE OF MEDICAL GENETICS

30 conditions, organized into 5 categories as follows:

Organic Acid Metabolism Disorders

IVA = Isovaleric acidemia

GA I = Glutaric acidemia type I

HMG = 3-OH 3-CH₃ glutaric aciduria

MCD = Multiple carboxylase deficiency

MUT = Methylmalonic acidemia (mutase deficiency)

3MCC = 3-Methylcrotonyl-CoA carboxylase deficiency

Cbl A,B = Methylmalonic acidemia

PROP = Propionic acidemia

BKT = Beta-ketothiolase deficiency

Fatty Acid Oxidation Disorders

MCAD = Medium-chain acyl-CoA dehydrogenase deficiency

VLCAD = Very long-chain acyl-CoA dehydrogenase deficiency

LCHAD = Long-chain L-3-OH acyl-CoA dehydrogenase deficiency

TFP = Trifunctional protein deficiency

CUD = Carnitine uptake defect

Amino Acid Metabolism Disorders

PKU = Phenylketonuria

MSUD = Maple syrup urine disease

HCY = Homocystinuria

CIT = Citrullinemia

ASA = Argininosuccinic acidemia

TYR I = Tyrosinemia type I

Hemoglobinopathies

SCA = Sickle cell anemia

Hb S/Th = Hb S/Beta-thalassemia

Hb S/C = Hb S/C disease

UNIFORM CONDITION PANEL RECOMMENDED BY ACMG

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Others

HYPOTH = Congenital hypothyroidism

BIOT = Biotinidase deficiency

CAH = Congenital adrenal hyperplasia

GALT = Galactosemia

HEAR = Hearing deficiency

G6PD = Glucose-6-phosphate dehydrogenase deficiency

CF = Cystic fibrosis

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