Abbreviations and Alternate Names for the Additional Newborn Screening Disorders

Organic Acid Disorders

IVA – Isovaleric acidemia/aciduria or
IVD – Isovaleric Acid CoA dehydrogenase deficiency

GAI – Glutaric acidemia/aciduria Type 1, or
Glutaryl – CoA dehydrogenase deficiency

HMG – 3-hydroxy (OH), 3-methyl (CH_3) CoA lyase deficiency;
hydroxymethyl glutaric aciduria; or
HLD – HMG-CoA lyase deficiency

MCD – Multiple carboxylase deficiency; or
HCD – Holocarboxylase deficiency; holocarboxylase synthetase deficiency

MUT – Methylmalonic acidemia (mutase deficiency); methylmalonic aciduria, or MCM deficiency; – vitamin B_{12} non responsive methylmalonic aciduria due to methylmalonic CoA mutase deficiency.

3MCC – 3 methylcrotonyl CoA carboxylase deficiency; 3 methylcrotonylglycinuria

Cbl A,B – Methylmalonic aciduria/acidemia, vitamin B_{12} responsive; or
MMAA due to defect in synthesis of adenosylcobalamin cbl A or cb1B types

PROP – Propionic acidemia; propionyl-CoA carboxylase deficiency; ketotic hyperglycinemia

BKT – Beta-ketothiolase deficiency; alpha-methylacetoacetic aciduria; 2-methyl-3-hydroxybutyric acidemia; mitochondrial acetoacetyl-CoA thiolase deficiency; or
MAAT deficiency; or
T2 deficiency; 3-oxothiolase deficiency; 3-ketothiolase deficiency; or
3-KTD deficiency

Fatty Acid Oxidation Disorders

MCAD – Medium-chain acyl-CoA dehydrogenase deficiency; or
MCADD; or
ACADM deficiency

VLCAD – Very long-chain acyl-CoA dehydrogenase deficiency;
or VLCADD

LCHAD – Long-chain hydroxyacyl-CoA dehydrogenase deficiency;
or LCADD

TFP – Trifunctional protein deficiency; mitochondrial trifunctional protein deficiency

CUD – Carnitine uptake defect; primary carnitine deficiency; systemic carnitine deficiency or
CTD – Carnitine transporter deficiency

Amino Acid Disorders

PKU – Phenylketonuria

MSUD – Maple syrup urine disease; branched-chain ketoaciduria; ketoacid decarboxylase deficiency; branched-chain alpha-keto acid hydrogenase deficiency

HCY – Homocystinuria

TYR1 – Tyrosinemia type 1; hepatorenal tyrosinemia; fumaryl acetoacetase deficiency or
FAH deficiency – fumaryl acetoacetate hydrolase deficiency

Urea Cycle Disorders

CIT – Citrullinemia;
CTLNI;
ASS deficiency – Argininosuccinate synthetase deficiency

ASA – Arginine succinic aciduria; argininosuccinase deficiency; or
ASL deficiency – Argininosuccinate lyase deficiency; or
ASAL deficiency

Other

BIOT - Biotinidase deficiency