Abbreviations and Alternate Names for The Additional NBS Disorders

ORGANIC ACIDS

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 DIOSORDERS

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