Diagnoses Included in the Inborn Errors of Metabolism Information System (IBEM-IS) Data Registry

Amino Acidemias

- Maple syrup urine disease
- Homocystinuria (CBS, MTHFR, Cbl D variant 1, Cbl E, Cbl G)
- Tyrosinemia
- Arginemia
- Argininosuccinate acidemia (ASA)
- Citrullinemia Type I (argininosuccinate synthetase)
- Citrullinemia Type II (citrin deficiency)
- Hypermethioninemia
- Defects of biopterin cofactor biosynthesis/regeneration
- Hyperphenylalaninemia/phenylketonuria

Fatty Acid Oxidation

- Carnitine uptake deficiency (CUD)
- CACT deficiency
- CPT-1 deficiency
- CPT-2 deficiency
- SCAD deficiency
- SBCAD deficiency
- MCAD deficiency
- LCHAD deficiency
- Trifunctional protein deficiency
- VLCAD deficiency
- Dienoyl-CoA reductase deficiency
- Medium-chain ketoacyl-CoA thiolase (MCKAT) deficiency
- Glutaric acidemia type II
- Medium/Short-chain L-3-hydroxy acyl-CoA dehydrogenase (M/SCHAD) deficiency

Organic Acidemias

- 2-methylbutyryl-CoA dehydrogenase deficiency
- Isovaleric acidemia
- Glutaric acidemia type I
- Isobutyryl-CoA dehydrogenase deficiency
- 3-MCC deficiency
- 2-methyl 3-hydroxybutyryl CoA dehydrogenase (2M3HBA) deficiency
- Holocarboxylase synthetase deficiency
- 3-methylglutaconic aciduria type I
- Beta-ketothiolase deficiency
- Succinyl CoA-3-keto transferase (SCOT) deficiency
- 3-hydroxy 3-methylglutaryl (HMG) CoA lyase deficiency
- Propionic Acidemia
- MMA (Mut-, Mut0, cbl A, cbl B, cbl D variant 2)
- MMA + Hcy (Cbl C, Cbl D, Cbl F, Transcobalamin II)
- Malonic acidemia

Non-MS/MS Conditions

- Biotinidase deficiency
- GALT deficiency
- Galactokinase (GALK) deficiency galactosemia
- UDP-galactose-4-epimerase (GALE) deficiency galactosemia