# Organic acid disorders (GC/MS)

Methylmalonic Acidemia

MMA-semialdehyde dehydrogenase deficiency

Propionic Acidemia

Beta Ketothiolase Deficiency

Isovaleric Acidemia

3-Methylcrotonyl-CoA Carboxylase Deficiency

Multiple Carboxylase Deficiency

3-OH-3-Methylglutaric Aciduria

3-Methylglutaconic Aciduria

Glutaric Aciduria Type I

Glutaric Aciduria Type II

3-OH-Isobutyric Aciduria

5-Oxoprolinuria

2-Hydroxyglutaric Acidemia

4-Hydroxybutyric Aciduria

Mevalonic Aciduria

Glyceroluria

Primary Hyperoxaluria Type I

L-Glyceric Acidemia (Primary Hyperoxaluria Type II )

2-Ketoadipic Aciduria

Alkaptunuria

Urea Cycle Disorder / OTC Deficiency

Phenylketonuria

Maple Syrup Urine Disease

Tyrosinemia Type I

MCAD Deficiency

Canavan Disease

Lactic Aciduria (Condition)

Respiratory chain disease

Ketosis (Condition)

Dicarboxylic Aciduria

Ketotic dicarboxylic aciduria

Non-ketotic dicarboxylic aciduria

Cobalamin deficiency

3-Hydroxydicarboxylic Aciduria (Condition)

Zellweger Syndrome

Metabloic disorders of purine and pyrimidine

Valproic acid treatment

Ethosuccimide treatment

Glycerol treatment

Carbocysteine administration

Aspirin administration

Sodium benzoate administration

MCT milk feeding

Food containing vanilla taking

Special fomula containing vanilla feeding

Ascorbic acid infusion

Neonatal transient hypertyrosinemia

Severe liver dysfunction

Hypoxia

Acetonemic vomiting

Neuroblastoma

Mitochondriopathy

# Metabolic Screening by LC/MS/MS

## Amino Acid Disorders

Phenylketonuria (PKU)

Biopterin Cofactor Deficiencies (GTPCH/ PTPS/ DHPR/ PCD Deficiency)

Argininemia/Arginase Deficiency

Argininosuccinic Acid Lyase Deficiency (ASAL Deficiency)

Citrullinemia, Type I /Argininosuccinic Acid Synthetase Deficiency (ASAS Deficiency)

Citrullinemia, Type I I (Citrin Deficiency)

Homocitrullinuria, Hyperornithinemia, Hyperammonemia – HHH

Homocystinuria Type I (Cystathionine beta-synthase Deficiency (CBS Deficiency))

Homocystinuria Type I I (Defect in MethylcobalamineSynthesis )

Homocystinuria Type I II (MTHFR, MTR, MTRR, Cbl D v1, Cbl G Deficiencies)

Methionine Adenosyltransferase Deficiency (MAT Deficiency)

Maple Syrup Urine Disease (MSUD)

Ornithine Transcarbamylase Deficiency (OTC Deficiency)

CarbamoylphosphateSynthetase Deficiency (CPS Deficiency)

Prolinemia

Tyrosinemia Type I, II, III

Gyrate atrophy of the choroid and retina

Ketotic hyperglycinemia

Non-ketotic hyperglycinemia

## Organic acid disorders

3-Hydroxy-3-Methylglutaryl-CoA Lyase Deficiency (HMG-CoA Lyase Deficiency)

Isobutyryl-CoA Dehydrogenase Deficiency (IBD Deficiency)

2-Methylbutyryl-CoA Dehydrogenase Deficiency (Short/Branched-Chain Acyl-CoA Dehydrogenase Deficiency)

3-Methylcrotonyl-CoA Carboxylase Deficiency (3MCC Deficiency)

3-methylglutaconic aciduria (MGA Type I) (3-methylglutaconyl-CoA Hydratase Deficiency)

Cytosolic ß-Ketothiolase Deficiency

Mitochondrial Acetoacetyl-CoA Thiolase Deficiency (3-Oxothiolase Deficiency)

Glutaric Acidemia-Type I (GA I)

Isovaleric Acidemia (IVA)

MethylmalonicAcidemias

Succinyl-CoA synthetase Deficiency (SUCLA2 Deficiency)

Propionic Acidemia (PA)

Multiple-CoA Carboxylase Deficiency / HolocarboxylaseSynthetase Deficiency (MCD)

Multiple-CoA Carboxylase Deficiency / Biotinidase Deficiency

2-methyl-3-hydroxybutyryl-CoA Dehydrogenase Deficiency (2M3HBA Deficiency)

Ethylmalonic Encephalopathy (EE)

## Fatty Acid disorders

Primary Carnitine Deficiency (PCD Deficiency)

Secondary Carnitine Deficiencies

Carnitine/acylcarnitine Translocase Deficiency (CACT Deficiency)

Carnitine PalmitoylTransferase Deficiency-type 1 (CPT 1 Deficiency)

Carnitine PalmitoylTransferase Deficiency-type 2 (CPT 2 Deficiency)

Multiple acyl-CoA Dehydrogenase Deficiency (MAD Deficiency)/Glutaric acidemia type-2 (GA-2)

Short-Chain Acyl-CoA Dehydrogenase Deficiency (SCAD Deficiency)

Short Chain Hydroxy Acyl-CoA Dehydrogenase Deficiency (SCHAD Deficiency)

Medium-Chain Acyl-Coenzyme A Dehydrogenase Deficiency (MCAD Deficiency)

Very Long Chain Acyl-CoA Dehydrogenase Deficiency (VLCAD)

LCHAD / TFP Deficiency