

UCHSC Expanded Newborn Screening Program

Disorders Reliably Detected in Newborns by MS/MS Technology:

Amino Acid Disorders:

Argininosuccinic Aciduria (ASA Lyase Deficiency)
Citrullinemia (ASA Synthetase Deficiency)
Homocystinuria (CBS Deficiency)
Hypermethioninemias
Maple Syrup Urine Disease (MSUD)
Phenylketonuria (PKU) and other hyperphenylalaninemias
some Tyrosinemias

Fatty Acid Oxidation Disorders*:

Carnitine/Acylcarnitine Translocase Deficiency
3-Hydroxy Long Chain Acyl-CoA Dehydrogenase (LCHAD) Deficiency
Medium Chain Acyl-CoA Dehydrogenase (MCAD) Deficiency
Multiple Acyl-CoA Dehydrogenase Deficiency (Glutaric Acidemia-Type II, GA II)
Neonatal Carnitine Palmitoyl Transferase Deficiency-Type II (CPT II)
Short Chain Acyl-CoA Dehydrogenase (SCAD) Deficiency
Trifunctional Protein (TFP) Deficiency
Very Long Chain Acyl-CoA Dehydrogenase (VLCAD) Deficiency

Organic Acid Disorders*:

3-Hydroxy-3-Methylglutaryl-CoA Lyase (HMG CoA) Deficiency
Glutaric Acidemia-Type I (GA I)
Isovaleric Acidemia (IVA)
3-Methylcrotonyl-CoA Carboxylase (3MCC) Deficiency
3-Methylglutaconyl-CoA Hydratase Deficiency
Methylmalonic Acidemias (MMA)
Mitochondrial Acetoacetyl-CoA Thiolase (3-Ketothiolase) Deficiency
Propionic Acidemia (PA, PCC Deficiency)
Multiple-CoA Carboxylase Deficiencies

Abnormal Metabolites:

Amino Acids:

Citrulline
Citrulline
Methionine
Methionine
Leucine/Isoleucine, Valine
Phenylalanine, Tyrosine
Tyrosine

Carnitine Esters:

C16, C18, C18:1
C16OH, C18:1OH
C8, C10:1
C5, C5DC, C8
C16, C18, C18:1
C4
C16OH, C18:1OH
C14, C14:1, C16, C18

C5OH
C5DC
C5
C5OH
C5OH
C3
C5OH, C5:1
C3
C3, C5OH

* The screen for some fatty acid oxidation & organic acid disorders is less sensitive after the age of seven days.

For questions, information or screening kits, contact:

Robert Binard or Mike Wootner
UCHSC Biochemical Genetics Laboratory
Phone: (303) 724-3826
Email: robert.binard@uchsc.edu
mike.wootner@uchsc.edu
Information Online: www.uchsc.edu/newbornscreening

Send specimen cards to:

Domestic (mail via U.S. Postal Service):

UCHSC Biochemical Genetics Laboratory
Mail Stop 8313
P.O. Box 6511
Aurora, CO 80045

International (ship via DHL, UPS, FedEx, etc.):

UCHSC Biochemical Genetics Laboratory
Bldg. RC-1 North, Rm. P18-4403C
12800 E. 19th Av.
Aurora, CO 80045 U.S.A.