آزمایشگاه پاتوبیولوژی نور
NOOR Pathobiology laboratory

DIAGNOSABLE DISORDERS BY LC/MS/MS (TANDEM MASS)

Amino Acid Disorders:
Argininemia
Argininosuccinic aciduria
Citrullinemia
Homocystinuria
Maple syrup urine disease (MSUD)
Phenylketonuria (PKU)
Tyrosinemia type 1
Fatty Acid Oxidation Disorders:
CACT deficiency
CPT1 deficiency
CPT2 deficiency
Carnitine uptake deficiency
Glutaric acidemia type 2
LCHADD/TFP deficiency
MCADD
Short-chain acyl-CoA deficiency (SCADD)
VLCADD
Organic Acidemias :
Glutaric acidemia type 1
2M3HBA deficiency
3MCC deficiency
Beta-Ketothiolase deficiency
HMG-CoA lyase deficiency
Holocarboxylase/multiple carboxylase def.
Isobutyryl Co-A dehydrogenase def.
Malonic aciduria

Methylmalonicacidemias
Propionic acidemia
Isovaleric acidemia
Short/branched chain acyl-CoA dehydrogenase deficiency

DETECTABLE COMPONENTS BY LC/MS/MS (TANDEM MASS)

Compound Name
Alanine
Arginine
Aspartic acid
Citroline
Glutamic acid
Glycine
Leucine
Methionine
Ornithine
Phenylalanine
Proline
Tyrosine
Valin
C0(Free carnitine)
C2(Acetylcarnitine)
C3(Propionylcarnitine)
C4(Butyrylcarnitine)
C4OH(3-OH-Butyrlcarnitine)
C4DC
C5(Isovalerycarnitine)
C5OH(3-OH-Isovalerylcarnitine)
C5DC(Glutarylcarnitine)
C6(Hexanoylcarnitine)
C8(Octanoylcarnitine)
C8:(Octanoylcarnitine)
C10(Decanoylcarnitine)
C10:(Decanoylcarnitine)
C12(Dodecanoylcarnitine)

Compound Name
C14(Tetradecanoylcarnitine)
C14:1(Tetradecanoylcarnitine)
C14:2(Tetradecanoylcarnitine)
C14OH(3-OH-Tetradecanoylcarnitine)
C16(Hexadecanoylcarnitine)
C16:1(Hexadecanoylcarnitine)
C16:1OH(3-OH-Hexadecanoylcarnitine)
C16OH(3-OH-Hexadecanoylcarnitine)
C18(Stearoylcarnitine)
C18:1(Oleoylcarnitine)
C18:1OH(3-OH-Oleylcanitine)
C18:2OH(3-OH-Linoleylcarnitine)
C18OH
C0/(C16+C18:1)
C3/C2
C5/C3
C8/C10
C16OH/C16
Val/Phe
Leu/Ala
Phe/Tyr
Cit/Arg
Orn/Cit
Arg/Orn