

TEST INFORMATION

NM Newborn Screening

1. Congenital Adrenal Hyperplasia
2. Sickle Cell
3. Hb S/beta-thalassemia
4. Biotinidase Deficiency
5. Congenital hypothyroidism
6. Galactosemia
7. Hearing deficiency
8. Cystic Fibrosis

Expanded Screening

Fatty Acid Oxidation Disorders (FOA)

9. Carnitine uptake defect
10. Long-chain L-3-OH acyl-CoA dehydrogenase deficiency
11. Medium chain acyl-CoA dehydrogenase deficiency
12. Trifunctional protein deficiency
13. Very long-chain acyl-CoA dehydrogenase deficiency

Amino Acid Disorders

14. Homocystinuria
15. Tyrosinemia type I
16. Phenylketonuria
17. Maple syrup urine disease

Urea Cycle disorders

18. Argininosuccinic acidemia
19. Citrullinemia

Organic Acidemia Disorders

20. 3-Hydroxy 3-Methyl Glutaric Aciduria (HGM)
21. 3-methylcrotonyl-CoA deficiency
22. Mitochondrial acetoacetyl-CoA thiolase deficiency
23. Isovaleric acidemia
24. Methylmalonic acidemia
25. Proponic acidemia
26. Multiple carboxylase deficiency
27. Glutaric acidemia type I