โรคพันธุกรรมเมแทบอลิก 40 โรคที่คัดกรองในทารกแรกเกิด (Expanded newborn Screening)

Disorders of organic acid metabolism

1. Glutaric acidemia type 1 (GA1)
2. Isovaleric acidemia (IVA)
3. Methylmalonic acidemia (MMA) due to mutase deficiency
4. Propionic acidemia (PA)
5. Multiple carboxylase deficiency (MCD) including holocarboxylase deficiency and biotinidase deficiency
6. Adenosylcobalamin synthesis defects
7. Beta-Ketothiolase deficiency
8. 3-Hydroxy-3-Methylglutaryl-CoA (HMG-CoA) lyase deficiency
9. Isobutyryl-CoA dehydrogenase deficiency
10. 2-Methylbutyryl-CoA dehydrogenase deficiency or 2-methyl-3-hydroxybutyric acidemia or short/branched chain acyl-CoA dehydrogenase (SBCAD) deficiency
11. 3-Methylcrotonyl-CoA carboxylase deficiency (3-MCC)
12. 3-Methylglutaconyl-CoA hydratase (3-MGA) deficiency or 3-methylglutaconic aciduria
13. Malonic aciduria
14. Combined methylmalonic acidemia and homocystinuria (CblC, CblD, CblF deficiency) and transcobalamin II defects

Disorders of amino acid metabolism

15. Phenylketonuria (PKU)
16. Tetrahydrobiopterin (BH4) defects
17. Maple syrup urine disease (MSUD)
18. Tyrosinemia type 1 (TYR1)
19. Tyrosinemia type 2 (TYR2)
20. Tyrosinemia type 3 (TYR3)
21. Homocystinuria

22. Hypermethioninemia including methyltransadenosyltransferase I/II deficiency; glycine-N-methyltransferase (GNMT) deficiency; adenosylhomocysteine hydrolase deficiency
23. Hyperornithinemia with gyrate atrophy

Disorders of fatty acid oxidation

24. Medium-chain acyl-CoA dehydrogenase (MCAD) deficiency
25. Long-chain hydroxyacyl-CoA dehydrogenase (LCHAD) deficiency
26. Very long-chain acyl-CoA dehydrogenase (VLCAD) deficiency
27. Short-chain acyl-CoA dehydrogenase (SCAD) deficiency
28. Short-chain hydroxyacyl-CoA dehydrogenase (SCHAD) deficiency
29. Trifunctional protein deficiency
30. Multiple acyl-CoA dehydrogenase (MADD) deficiency (Glutaric aciedemia type 2)
31. Carnitine-acylcarnitine translocase (CACT) deficiency
32. Carnitine palmitoyltransferase type 1 (CPT1) deficiency
33. Neonatal carnitine palmitoyltransferase type 2 (CPT2) deficiency
34. Primary systemic carnitine deficiency (Carnitine uptake defect, CUD)

Urea cycle disorders

35. Citrullinemia type 1 (CIT1)
36. Citrullinemia type 2 or Citrin deficiency
37. Argininosuccinic aciduria
38. Argininemia (Arginase deficiency)
39. Hyperammonemia-Hyperornithinemia-Homocitrullinuria (HHH) syndrome
40. Ornithine transcarbamylase deficiency