

Disorders List

The Newborn Screening Laboratory screens all Michigan Infants for more than fifty disorders.

Amino Acid Disorders

1. Argininemia (ARG)
2. Argininosuccinic acidemia (ASA)
3. Citrullinemia Type I (CIT-I)
4. Citrullinemia Type II (CIT-II)
5. Homocystinuria (HCY)
6. Hypermethioninemia (MET)
7. Maple syrup urine disease (MSUD)
8. Phenylketonuria (PKU)
 9. Benign hyperphenylalaninemia defect (H-PHE)
 10. Biopterin cofactor biosynthesis defect (BIOPT-BS)
 11. Biopterin cofactor regeneration defect (BIOPT-REG)
12. Tyrosinemia Type I (TYR-1)
 13. Tyrosinemia Type II (TYR-II)
 14. Tyrosinemia Type III (TYR-III)

Fatty Acid Oxidation Disorders

15. Carnitine acylcarnitine translocase deficiency (CACT)
16. Carnitine palmitoyltransferase I deficiency (CPT-1A)
17. Carnitine palmitoyltransferase II deficiency (CPT-II)
18. Carnitine uptake defect (CUD)
19. Dienoyl-CoA reductase deficiency (DERED)
20. Glutaric acidemia type II (GA-2)
21. Long-chain L-3-hydroxy acyl-CoA dehydrogenase deficiency (LCHAD)
22. Medium/short-chain L-3-hydroxy acyl-CoA dehydrogenase deficiency (M/SCHAD)
23. Medium-chain acyl-CoA dehydrogenase deficiency (MCAD)
24. Medium-chain ketoacyl-CoA thiolase deficiency (MCKAT)
25. Short-chain acyl-CoA dehydrogenase deficiency (SCAD)
26. Trifunctional protein deficiency (TFP)
27. Very long-chain acyl-CoA dehydrogenase deficiency (VLCAD)

Organic Acid Disorders

28. 2-Methyl-3-hydroxy butyric aciduria (2M3HBA)
29. 2-Methylbutyryl-CoA dehydrogenase deficiency (2MBG)
30. 3-hydroxy 3-methylglutaric glutaric aciduria (HMG)
31. 3-Methylcrotonyl-CoA carboxylase deficiency (3-MCC)
32. 3-Methylglutaconic aciduria (3MGA)
33. Beta-ketothiolase deficiency (BKT)
34. Glutaric acidemia type I (GA1)
35. Isobutyryl-CoA dehydrogenase deficiency (IBG)
36. Isovaleric acidemia (IVA)
37. Methylmalonic acidemia cobalamin disorders (Cbl A,B)
38. Methylmalonic aciduria with homocystinuria (Cbl C,D)
39. Methylmalonic acidemia methylmalonyl-CoA mutase (MUT)
40. Multiple carboxylase deficiency (MCD)
41. Propionic acidemia (PROP)

Hemoglobinopathies

42. S/Beta thalassemia
43. S/C disease
44. Sickle cell anemia
45. Variant hemoglobinopathies
46. Hemoglobin H disease

Endocrine Disorders

47. Congenital adrenal hyperplasia (CAH)
48. Congenital hypothyroidism (CH)

Other Disorders

49. Biotinidase deficiency (BIOT)
50. Galactosemia (GALT)
51. Cystic fibrosis (CF)
52. Severe combined immunodeficiency (SCID)
 53. T-cell related lymphocyte deficiencies

54. **Hearing**