



Disorders List

The Newborn Screening Laboratory screens all Michigan Infants for more than 50 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-I)
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. Trifunctional protein deficiency (TFP)
26. Very long-chain acyl-CoA dehydrogenase deficiency (VLCAD)

Organic Acid Disorders

27. 2-Methyl-3-hydroxy butyric aciduria (2M3HBA)
28. 2-Methylbutyryl-CoA dehydrogenase deficiency (2MBG)
29. 3-hydroxy 3-methylglutaric aciduria (HMG)
30. 3-Methylcrotonyl-CoA carboxylase deficiency (3-MCC)

31. 3-Methylglutaconic aciduria (3MGA)
32. Beta-ketothiolase deficiency (BKT)
33. Glutaric acidemia type I (GA1)
34. Isovaleric acidemia (IVA)

Malonic Acidemia (MAL)

35. Methylmalonic acidemia cobalamin disorders (Cbl A,B)
36. Methylmalonic aciduria with homocystinuria (Cbl C,D)
37. Methylmalonic acidemia methylmalonyl-CoA mutase (MUT)
38. Multiple carboxylase deficiency (MCD)
39. Propionic acidemia (PROP)

Hemoglobinopathies

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

Endocrine Disorders

45. Congenital adrenal hyperplasia (CAH)
46. Congenital hypothyroidism (CH)

Lysosomal Storage Disorders

47. Glycogen Storage Disease Type II (Pompe)
48. Mucopolysaccharidosis Type I (MPS I)

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. X-linked Adrenoleukodystrophy (X-ALD)
55. Spinal muscular atrophy (SMA)
56. Guanidinoacetate methyltransferase (GAMT) deficiency
57. Hearing
58. Critical Congenital Heart Disease (CCHD)

Disorders Coming Soon

These conditions have been approved for addition to Michigan's panel, but implementation is in progress and screening has not yet begun.

- Mucopolysaccharidosis Type II (MPS II)