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 biosysnthesis 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. 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-Methylbutyryrl-CoA dehydrogenase deficiency (2MBG)
29. 3-hydroxy 3-methylglutaric glutaric 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)
35. Malonic Acidemia (MAL)
36. Methylmalonic acidemia cobalamin disorders (Cbl A,B)
37. Methylmalonic aciduria with homocystinuria (Cbl C,D)
38. Methylmalonic acidemia methylmalonyl-CoA mutase (MUT)
39. Multiple carboxylase deficiency (MCD)
40. Propionic acidemia (PROP)

Hemoglobinopathies

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

Endocrine Disorders

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

Lysosomal Storage Disorders

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

Other Disorders

50. Biotinidase deficiency (BIOT)
51. Galactosemia (GALT)
52. Cystic fibrosis (CF)
53. Severe combined immunodeficiency (SCID)
54. T-cell related lymphocyte deficiencies
55. X-linked Adrenoleukodystrophy (X-ALD)
56. Spinal muscular atrophy (SMA)
57. Guanidinoacetate methyltransferase (GAMT) deficiency
58. Hearing
59. Critical Congenital Heart Disease (CCHD)