

### Introduction

These tests are intended to detect disorders that could result in complications such as early mortality or lifelong disability. Generally, most states only test for selected disorders and not all states do the same screening tests. It is important to be informed about any and all tests that are available for a new born baby

Source: <http://americanpregnancy.org/labor-and-birth/newborn-testing/>

### Disorders

Disorder	Statistical Occurance
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1. 3-Methylcrotonyl-CoA carboxylase deficiency (3MCC)	
2. 3-OH 3-CH3 glutaric aciduria (HMG) or 3-hydroxy-3-methylglutaryl-CoA lyase deficiency	
3. Argininosuccinic acid lyase deficiency (ASAL)	1 in 70,000
4. Beta-ketothiolase deficiency (BKD)	1 in 75,000
5. Biotinidase deficiency (BIOT)	
6. Carnitine transporter deficiency (CTD) or carnitine uptake defect (CUD)	1 in 40,000
7. Citrullinemia (CIT)	
8. Congenital adrenal hyperplasia (CAH)	1 in 18,000
9. Congenital hypothyroidism (HYPOTH):	1 in 3,000
10. Cystic fibrosis (CF)	1 in 30,000-60,000
11. Galactosemia (GALT)	
12. Glucose-6-phosphate dehydrogenase deficiency (G6PD)	
13. Glutaric acidemia type I (GA I)	
14. Hb S/Beta-thalassemia (Hb S/Th)	
15. Hb S/C disease (Hb S/C)	
16. Hearing deficiency	3-4 in 1,000
17. Homocystinuria (HCY)	1 in 200,000
18. Isovaleric academia (IVA)	1 in 230,000
19. Long-chain L-3-OH acyl-CoA dehydrogenase deficiency (LCHAD)	
20. Maple syrup urine disease (MSUD)	< 1 in 100,000
21. Medium chain acyl-CoA dehydrogenase deficiency (MCAD)	
22. Methylmalonic acidemia (MMA, Cbl A,B)	
23. Methylmalonic acidemia with Homocystinuria (MMA + HCU)	
24. Medium chain acyl-CoA dehydrogenase deficiency (MCAD)	1 in 16,000
25. Phenylketonuria (PKU)	1 in 25,000
26. Propionic acidemia (PROP)	
27. Sickle cell anemia (SCA)	1 in 400 African Americans
28. Trifunctional protein deficiency (TFP)	
29. Tyrosinemia type I (TYR I)	1 in 100,000
30. Very long-chain acyl-CoA dehydrogenase deficiency (VLCAD)	



### The most common disorders tested for

- Phenylketonuria (PKU)
- Congenital Hypothyroidism
- Galactosemia
- Maple Syrup Urine Disease
- Homocystinuria, Biotinidase
- Sickle Cell Disease
- Congenital Adrenal Hyperplasia
- Cystic Fibrosis.

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Published 26th August, 2018.  
Last updated 26th August, 2018.  
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