

Supplementary file S1. List of 35 prospective disorders to be included in the panel for universal NBS in Bulgaria (based on Loeber JG, Platis D, Zetterström RH, Almashanu S, Boemer F, Bonham JR, et al. Neonatal Screening in Europe Revisited: An ISNS Perspective on the Current State and Developments Since 2010. Int J Neonatal Screen. 2021 Mar 5;7(1):15.)

1. 3-Hydroxy-3-methylglutaric aciduria
2. 3-Methylcrotonyl-CoA carboxylase deficiency/3-Methylglutacon aciduria/2-methyl-3-OH-butyric aciduria
3. Argininemia
4. Argininosuccinic aciduria
5. Beta-ketothiolase deficiency
6. Biotinidase deficiency
7. Carnitine acylcarnitine translocase deficiency
8. Carnitine palmitoyltransferase deficiency type I
9. Carnitine palmitoyltransferase type II / Carnitine acylcarnitine transporter deficiency
10. Carnitine uptake defect
11. Citrullinemia type I and II
12. Classical galactosemia
13. Cystic fibrosis
14. Glucose-6-phosphate dehydrogenase deficiency
15. Glutaric acidemia type I
16. Glutaric acidemia type II / multiple acyl coA dehydrogenase deficiency
17. Holocarboxylase synthetase deficiency
18. Homocystinuria

19. Isovaleric acidemia
20. Long-chain L-3-hydroxyacyl-CoA dehydrogenase deficiency / Trifunctional protein deficiency
21. Maple syrup urine disease
22. Medium-chain acyl-CoA dehydrogenase deficiency
23. Methionine adenosyl transferase I/III deficiency
24. Methylmalonic acidemia
25. Multiple carboxylase deficiency
26. Propionic acidemia
27. Remethylation disorders (methylenetetrahydrofolate reductase, methylcobalamine deficiencies)
28. Severe combined immunodeficiencies
29. Short-chain acyl-CoA dehydrogenase deficiency
30. Spinal muscular atrophy
31. Thalassemia
32. Tyrosinemia type I
33. Tyrosinemia type II
34. Very long-chain acyl-CoA dehydrogenase deficiency
35. x-Adrenoleukodystrophy