รายชื่อ 40 โรคพันธุกรรมเมแทบอลิกที่คัดกรองในทารกแรกเกิด ศูนย์บริการตรวจคัดกรองทารกแรกเกิด โรงพยาบาลรามาธิบดี

List of inborn metabolic diseases in expanded newborn screening: RAMA-eNBS

Disorders of amino acid metabolism

1.	Phenylketonuria
2.	Tetrahydrobiopterin defects
3.	Tyrosinemia type 1
4.	Tyrosinemia type 2
5.	Tyrosinemia type 3
6.	Maple syrup urine disease
7.	Homocystinuria
8.	Hypermethioninemia (methionine adenosyltransferase deficiency)

Urea cycle disorders

9.	Citrullinemia type 1
10.	Citrullinemia type 2 (citrin deficiency)
11.	Argininosuccinic aciduria
12.	Argininemia
13.	Hyperammonemia-hyperornithinemia-homocitrullinuria
14.	Ornithine transcarbamylase (OTC) deficiency
15.	Proximal urea cycle disorders (Carbamoylphosphate synthetase 1 deficiency and N-acetylglutamate synthase deficiency)

Disorders of organic acid metabolism

16.	Methylmalonic acidemia
17.	Propionic acidemia
18.	Adenosylcobalamin synthesis defects
19.	Combined methylmalonic acidemia and homocystinuria
20.	Malonic aciduria
21.	Isobutyryl-CoA dehydrogenase deficiency
22.	Isovaleric acidemia
23.	2-Methylbutyryl-CoA dehydrogenase deficiency
24.	Glutaric acidemia type 1
25.	Beta-Ketothiolase deficiency
26.	3-Hydroxy-3-methylglutaryl-CoA lyase deficiency
27.	3-Methylcrotonyl-CoA carboxylase deficiency
28.	3-Methylglutaconyl-CoA hydratase deficiency
29.	Multiple carboxylase deficiency

Disorders of fatty acid oxidation

30.	Primary systemic carnitine deficiency (Carnitine uptake defect)
31.	Carnitine palmitoyltransferase type 1 (CPT1) deficiency
32.	Carnitine palmitoyltransferase type 2 (CPT2) deficiency
33.	Carnitine-acylcarnitine translocase (CACT) deficiency
34.	Short-chain acyl-CoA dehydrogenase (SCAD) deficiency
35.	Short-chain hydroxyacyl-CoA dehydrogenase (SCHAD) deficiency
36.	Medium-chain acyl-CoA dehydrogenase (MCAD) deficiency
37.	Very long-chain acyl-CoA dehydrogenase (VLCAD) deficiency
38.	Long-chain hydroxyacyl-CoA dehydrogenase (LCHAD) deficiency
39.	Trifunctional protein deficiency
40.	Multiple acyl-CoA dehydrogenase (MAD) deficiency