Additional file 3. Diseases included in the neonatal screening program of Catalonia.

Inherited metabolic diseases

Phenylketonuria and Hyperphenyalaninemias

Maple syrup urine disease

Tyrosinemia type I

Citrulinemia

Homocystinuria (CBS deficiency)

Glutaric aciduria type I

Isovaleric acidemia

Methylmalonic aciduria

Methylmalonic aciduria with Homocystinuria

3-Hydroxy-3-methylglutaryl-CoA lyase deficiency

ß-Ketothiolase deficiency

Propionic acidemia

Medium chain acyl-CoA dehydrogenase deficiency

Very long chain acyl-CoA dehydrogenase deficiency

Mitochondrial trifunctional protein deficiency

Multiple acyl-CoA dehydrogenase deficiency

Primary carnitine deficiency

Carnitine palmitoyltransferase 1 deficiency

Carnitine palmitoyltransferase 2 deficiency

Carnitine-acylcarnitine translocase deficiency

Other diseases

Cystic Fibrosis

Congenital Hypothyroidism

Sickle cell disease

Severe combined immunodeficiency