List of conditions covered in New Born Screening

Disorder	Method	Frequency
I) Endocrine Disorders		
Hypothyroidism (TSH)	Photometry	1: 4000
Adrenogenital Syndrome (17-OH Progesterone)	Photometry	1: 11000
II) Hemoglobinopathies		
HbS, beta-Thal, HbH etc.	CA	Depending on the country up to >1:10
III) Others		
G6PDH Deficiency	Photometry	Depending on the country: >1:10
Galactosemia	Photometry	1: 60000
Biotinidase Deficiency	Photometry	1: 75000
Cystic Fibrosis (Immuno Reactive Trypsin)	Photometry	1: 4000
IIIa) Disorders of Amino Acid Metabolism		
PKU (Phenylketonuria) Hyperphenylalaninemia	MS/MS	1:5.500
Disorders of biopterin cofactors biosynthesis (Hyperphenylalaninemia)	MS/MS	1: 500000
Disorders of biopterin cofactors regeneration (Hyperphenylalaninemia)	MS/MS	1:250000
PBGS Deficiency (Porphobilinogen Synthase); (Tyrosinemia Type 1)	Photometry	< 1: 100000
Tyrosinemia Type 2	MS/MS	Tyrosine levels may not be sufficiently elevated for detection!
Tyrosinemia Type 3	MS/MS	Tyrosine levels may not be sufficiently elevated for detection!
Maple Syrup Disease (MSUD)	MS/MS	1:150000
Hypermethioninemia/ Homocystinuria	MS/MS	<1:100000
Arginase Deficiency	MS/MS	n.a
Argininosuccinate Synthase Deficiency	MS/MS	n.a.
Argininosuccinate Lyase Deficiency	MS/MS	n.a.
IIIb) Urea Cycle Disorders		
Ornithine Aminotransferase Deficiency	MS/MS	The diagnosis in the neonatal presentation of OAT deficiency is difficult as hyperornithinaemia is absent
Citrullinemia Type I	MS/MS	1<100000
Citrullinemia Type II (ASA)		1:150000
Argininemia	MS/MS	1:250000
IIIc) Fatty Acid Oxidation Disorders		
Carnitine uptake defect	MS/MS	1:50000

Long Chain 3-OH acyl CoA dehydrogenase deficiency (LCHAD)	MS/MS	1:50000 (see Trifunctional Protein deficiency!)
Medium Chain 3-OH acyl CoA dehydrogenase deficiency (MCAD)	MS/MS	1:11000
Trifunctional Protein Deficiency	MS/MS	See LCHAD!
Very long chain acyl CoA dehydrogenase deficiency	MS/MS	1:75000
Dienoyl reductase deficiency	MS/MS	1: 2000000
Carnitine Palmitoyl Transferase I deficiency	MS/MS	1:300000 May not be reliably detected in the first few days of life
Carnitine Palmitoyl Transferase Type II deficiency	MS/MS	1:250000 (detection as neonatal form is extremely rare)
Glutaric academia type II	MS/MS	1:250000
Medium/short chain 3-OH acyl CoA dehydrogenase deficiency	MS/MS	1:2000000
Medium chain ketoacyl CoA dehydrogenase deficiency	MS/MS	1:2000000
Short chain acyl-CoA dehydrogenase deficiency	MS/MS	1:30000
Carnitine/acylcarnitine translocase deficiency	MS/MS	1:300000
IIId) Organic Acid Disorders		
Glutaric Aciduria Type I	MS/MS	1:100000
Methylmalonic acidemia (A, B)	MS/MS	1:100000
Methylmalonic academia (Mut)	MS/MS	1:40000 (combined with A, B)
3-Methyl Crotonyl CoA carboxylase deficiency	MS/MS	1:50000
3-Hydroxy 3-Methylglutaric aciduria	MS/MS	1:250000
Beta-Ketothiolase deficiency	MS/MS	1:300000
Multiple carboxylase deficiency	MS/MS	1:250000
Propionic acidemia	MS/MS	1:150000
2-Methyl- 3- hydroxybutyric aciduria	MS/MS	1:1000000
2-Methylbutyryl CoA dehydrogenase deficiency	MS/MS	<1:100000
3-Methylglucaconic aciduria	MS/MS	1:100000
Isobutyryl CoA dehydrogenase deficiency	MS/MS	1:100000
Malonic aciduria	MS/MS	1:300000
Methylmalonic acidemia (Cbl, C, B)	MS/MS	1:100000