Baby' ①st test Comprehensive Screening test lists

	Amino acids disorders
01	classical phenylketonuria (PKU)
02	variant PKU
03	guanosine triphosphate cyclohydrolase 1 (GTPCH) deficiency (biopterin deficiency)
04	6-pyruvoyl-tetrahydropterin synthase (PTPS) deficiency (biopterin deficiency)
05	dihydropteridine reductase (DHPR) deficiency (biopterin deficiency)
06	pterin-4α-carbinolamine dehydratase (PCD) deficiency (biopterin deficiency)
07	argininemia/arginase deficiency
08	argininosuccinic acid lyase deficiency (ASAL deficiency)
09	citrullinemia, Type I/argininosuccinic acid synthetase deficiency (ASAS deficiency)
10	citrullinemia, Type II (citrin deficiency)
11	gyrate atrophy of the choroid and retina
12	homocitrullinuria, hyperornithinemia, hyperammonemia – HHH
13	homocystinuria/cystathionine beta-synthase deficiency (CBS deficiency)
14	methionine adenosyltransferase deficiency (MAT deficiency)
15	maple syrup urine disease – (MSUD)
16	prolinemia
17	tyrosinemia, Type I
18	tyrosinemia, Type II
19	tyrosinemia, Type III
20	tyrosinemia, Type transient
21	ornithine transcarbamylase deficiency (OTC deficiency)
22	remethylation defects
	Organic Acid Disorders
23	2-methyl-3-hydroxybutyryl-CoA dehydrogenase deficiency
24	2-methylbutyryl-CoA dehydrogenase deficiency
25	3-hydroxy-3-methylglutaryl-CoA lyase deficiency (HMGCoA lyase deficiency)
26	3-methylcrotonyl-CoA carboxylase deficiency (3MCC deficiency)
27	3-methylglutaconic aciduria (MGA), Type I (3-methylglutaconyl-CoA hydratase deficiency)
28	beta-ketothiolase deficiency (BKT)
29	ethylmalonic encephalopathy (EE)
30	glutaric acidemia type-1 (GA-1)
31	isobutyryl-CoA dehydrogenase deficiency
32	isovaleric acidemia (IVA)
33	malonic aciduria
34	methylmalonic acidemia, mut –
35	methylmalonic acidemia, mut 0
36	methylmalonic acidemia (Cbl A, B)
37	methylmalonic acidemia (Cbl C, D)
38	multiple carboxylase deficiency (MCD)
39	propionic acidemia (PA)
	Fatty Acid Oxidation Disorders
40	carnitine transporter deficiency
41	carnitine-acylcarnitine translocase deficiency (CAT deficiency)
42	carnitine palmitoyl transferase deficiency-type 1 (CPT-1 deficiency)
43	carnitine palmitoyl transferase deficiency-type 2 (CPT-2 deficiency)
44	long chain hydroxyacyl-CoA dehydrogenase deficiency (LCHAD deficiency)
45	medium chain acyl-CoA dehydrogenase deficiency (MCAD deficiency)
46	medium/short chain L-3-hydroxy acyl-CoA dehydrogenase deficiency (M/SCHAD deficiency)
47	multiple acyl-CoA dehydrogenase deficiency (MAD deficiency)/glutaric acidemia type-2 (GA-2)
48	short chain acyl-CoA dehydrogenase deficiency (SCAD deficiency)
49	trifunctional protein deficiency (TFP deficiency)
50	very long chain acyl-CoA dehydrogenase deficiency (VLCAD deficiency)
	Other Metabolic Disorders
51	classical galactosemia
52	biotinidase deficiency
53	Cystic Fibrosis
54	congenital adrenal hyperplasia
55	congenital hypothyroidism
56	Glucose -6- phospate Dehydroginase (G6PD)