Metabolic Screening List of 140+ Metabolic Abnormalities by GC/MS Analysis

DISORDERS OF AROMATIC AMINO ACID METABOLISM	
1	Hyperphenylalaninemia
2	Phenylketonuria
3	BH4 deficiency
4	Tyrosinemia I II III
5	Transient neonatal, oculocutaneous & hepatorenal forms
6	Tyrosinemia Hepatorenal form only
7	Hawkinsinuria
8	Alcaptonuria
DISORDERS OF BRANCHED CHAIN AMINO ACID METABOLISM	
9	Maple syrup urine disease
10	Dihydrolipoyl dehydrogenase (E3) deficiency
11	Isovaleric academia
12	3-methylcrotonyl-CoA carboxylase deficiency
13	Holocarboxylase synthetase deficiency
14	Biotinidase deficiency
15	3-Methylglutaconic aciduria
16	3-Hydroxy-3-methylglutaric aciduria
17	3-oxothiolase deficiency
18	3-Oxoacid CoA transterase deficiency
19	Propionic academia
20	Methylmaonic academia
21	Malonyl-CoA decarboxylase deficiency
22	Hyperleucinemia
23	Hypervalinemia
24	3-hydroxyisobutyryl CoA deacylase deficiency
25	Ethylhydracrylic aciduria
26	2-Oxoadpic aciduria
DISORDERS OF DIBASIC AMINO ACID METABOLISM	
27	Glutaric aciduria TypeI
28	Hyperornithinemia-hyperammonemia- homocitrullinuria (HHH) syndrome
29	Lysinuric protein intolerance
30	N-acetylglutamate synthetase deficiency
DISORDERS OF THE UREA CYCLE	
31	Vitamin B 12 deficiency
32	Carbamyl phosphate synthase deficiency
33	Arigininosuccinase deficiency
34	Argininosuccinate synthase deficiency
35	Ornithine carbamoyltransferase deficiency
36	Argininemia
DISORDERS OF PYRIMIDINE METABOLISM	
37	Orotic aciduria

Metabolic Screening List of 140+ Metabolic Abnormalities by GC/MS Analysis

38 Dihydropyrimidine dehydrogenase deficiency
39 Adenosine deaminase deficiency
40 Lesch-Nyhan syndrome
41 Xanthinuria
42 Dihydropyridinase deficiency
43 Adenine phosphoribosyltransferase deficiency
44 Partial deficiency of hypoxanthine adenine phosphoribosyltransferase deficiency

DISORDERS OF FATTY ACID OXIDATION

45 Long chain acyl-CoA dehydrogenase deficiency
46 Medium chain acyl-CoA dehydrogenase deficiency
47 Short chain acyl-CoA dehydrogenase deficiency
48 Multiple acyl-CoA dehydrogenase deficiency (glutaric aciduria II)
49 Normals fed medium chain triglyceride formulas
50 MCT Milk fed
51 Very Long-chain acyl-CoA dehydrogenase deficiency (VLCAD)
52 Short-chain 3-hydroxyacyl-CoA dehydrogenase deficiency (SCHAD)
53 Long-chain 3-hydroxyacyl-CoA dehydrogenase deficiency (LCHAD)
54 Mitochondrial trifunctional protein Deficiency (MTPD)
55 Carnitine Palmitoyl Synthase I Deficiency(CPSID)
56 Carnitine Palmitoyl Synthase II Deficiency(CPSIID)
57 Carnitine Palmitoyl Synthase Deficiency(CPSD)
58 Carnitine transport defect

AMINO ACID DISORDERS

59 Hyperlysinemia
60 Arginosuccinase deficiency
61 Citrullinemia
62 Hartnup disorder
63 Renal Fanconi's
64 Histidinemia
65 Pyruvate carboxylase deficiency
66 Hyper- ß-alaninemia
67 Aminoadipic aciduria
68 Ketoadipic acidura
69 ß-Aminoisobutyric aciduria
70 Hyperornithinemia
71 Cystinuria
72 Hyperdibasicaminoaciduria
73 Lysinuric protein intolerance
74 Dicarboxylic aminoaciduria
75 Saccharopinuria
76 Cystathioninuria
77 Homocystinuria

Metabolic Screening List of 140+ Metabolic Abnormalities by GC/MS Analysis

78 Ethanolaminosis
79 Hyperglycinuria
80 Hyperprolinemia type I, Prolidase deficiency
81 Hydroxyprolinemia
82 Tryptophanuria
83 Lysine malabsorption
84 Mercaptolactate-cysteine disulfiduria
85 Formiminoglutamic aciduria
86 Hypermethioninemia
87 Hypophosphatasia
88 Molybdenum cofactor deficiency/Sulfite oxidase deficiency
89 Glutathione synthase deficiency
90 Hyperleucinuria
91 Hypervalinemia
92 Hypersarcosinemia
93 Hyperphenylalaniemia
94 Homoserinuria

MISCELLANEOUS DISORDERS

95 4-hydroxybutyric aciduria
96 Fumaric acidemia
97 Mevalonic acidemia
98 5-Oxoprolinuria
99 Canavan disease
100 Dehydration
101 Malabsorption syndromes
102 Lactose intolerance
103 Glyceric aciduria
104 Hyperoxaluria type I
105 Hyperoxaluria type II
106 Glyceroluria
107 Lactic acidosis
108 Pyruvate dehydrogenase
109 Intestinal bacterial overgrowth
110 Hepatic failure
111 Renal dysfunction
112 Chronic hypoxia
113 Acidosis, gluconeogenesis

CARBOHYDRATES

114 Aspartylglucosaminura
115 Galactosemia I
116 Galactosemia II
117 Galactosemia III

Metabolic Screening List of 140+ Metabolic Abnormalities by GC/MS Analysis

118 Transient galactosemia
119 Renal glycosuria
120 Hereditary fructose intolerance
121 Sialic Acid Storage Disease
122 Pentosuria
123 Diabetes mellitus type I
124 Diabetes mellitus type II
125 Gestational diabetes mellitus
126 Fructosuria
127 Fructose-1, 6-diphosphatase deficiency
128 Endogeneous sucrosuria

PEROXISOMAL DISEASE

129 Zellweger syndrome
130 Neonatal adrenoleukodystrophy
131 Infantile refsum disease (IRD)
132 Zellweger like syndrome (ZLS)
133 Primary hyperoxoluria

NEUROTRANSMITTERS

134 Pheochromocytoma
135 Neuroblastoma

TOXICOLOGIC

136 Aspirin Poisoning
137 Ethylene glycol poisoning
138 Tegretol Poisoning
139 Valproate toxicity

CITRIN DEFICIENCY

140 Neonatal intrahepatic cholestasis (NICCD)
141 Adult-onset type II citrullinemia (CTLN2)