MEDGENOME | ACTIA Inherited Disease Genetics

BabySecure+

by Gas Chromatography Mass Spectrometry (GCMS)

Advanced newborn screening test using Urine Organic Acid Analysis by GCMS to secure the baby from the impact of inherited disorders

Test done on urine sample using **Gas Chromatography Mass Spectrometry (GCMS)** platform that covers the screening for **100+ inherited metabolic disorders** in the baby.

Why BabySecure+ by GCMS is important?

The test is an useful adjunct to newborn screening follow-up, and it has the additional benefit of being able to identify many classes of biochemical compounds, such as amino acids, acylglycines, organic acids, purine and pyrimidines, neurotransmitters, and carbohydrates.

These disorders, if remain undetected and untreated, can have irreversible adverse outcomes. BabySecure+ tests helps identify babies with Inborn Errors of Metabolism (IEMs) so that treatment can start before any adverse consequences to the baby's health.

When is BabySecure+ by GCMS analysis done?

It is usually performed along with Amino acid and Acylcarnitine profile on Dried Blood Spot by Tandem Mass Spectrometry (TMS) as a second-tier test or as a confirmatory test. The test can also be performed as a routine screening test in a newborn.

It can also be used as a high-risk screening in case of appearance of neurologic signs, jaundice, metabolic acidosis, ketotic hypoglycemia, and hyperammonemia. It can be done as a confirmatory test of dieseases such as Orotic aciduria, Canavan disease or Alkaptonuria etc., too.

This test helps in disorder screening, confirmation and follow up monitoring of the inherited metabolic disorders such as Organic acidemia, amino acid disorders, Mitochondrial disorders, Fatty acid Oxidation disorders, Peroxisomal disorders, Purine-Pyrimidine metabolism defects and Carbohydrate metabolism disorders.

How the sample is collected for the BabySecure+ by GCMS?

The GCMS is a simple test done any time after 24 hours of birth of a newborn on an urine sample collected on a dried urine filter paper or Sterile Urine container.

- Urine samples from a newborn can be collected by placing the special filter paper card in the baby's diaper and checking every 30 minutes, ensuring that the urine is passed and absorbed over the filter. **(Figure 1)**
- Alternatively, urine sample can be collected in a sterile urine container (10-20 ml) and dipping the special filter paper card. **(Figure 2)**

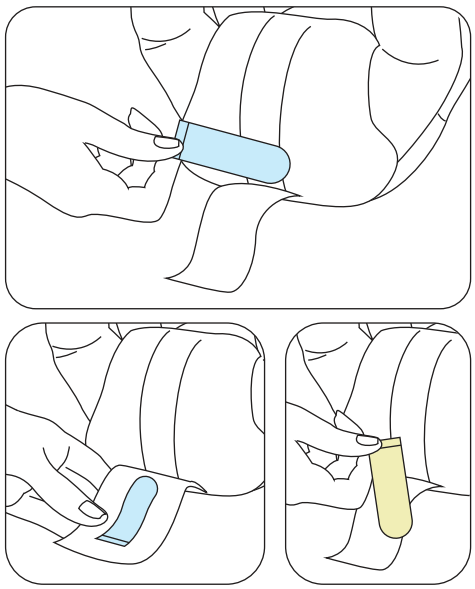

Figure 1

Figure 2

Allow the card to dry thoroughly by hanging them with clip for one hour. The dried urine strips can be shipped to MedGenome Labs in a zip lock pouch after appropriate labeling.

Test Details

Test Code	Test Name	Disorders covered	Sample type	TAT
MGM3297	Urine Organic Acid Analysis by GCMS	100+	Dried Urine Sample (DUS)	72 hours
MGM3298	NBS Duo - DBS Amino acid / Acylcarnitine profile by TMS and Urine Organic Acid Analysis by GCMS	100+	Dried Blood Spot (DBS) and Dried Urine Sample (DUS)	72 hours

Disorders covered in BabySecure+ by GCMS

A	Amino Acidopathies and Organic Acidemias
1	2-Hydroxyglutaric aciduria
2	2-ketoadipic aciduria
3	3-hydroxy-3-methylglutaryl-CoA-lyase deficiency
4	3-Hydroxyisobutyryl CoA Deacylase Deficiency
5	3-methylcrotonyl CoA carboxylase deficiency
6	3-methylglutaconic aciduria Type I
7	5-oxoprolinuria (Pyroglutamic aciduria)
8	Alkaptonuria
9	Aminoacylase I Deficiency
10	Argininemia
11	Argininosuccinic aciduria
12	Benign hyperphenylalaninemia
13	Beta-Aminoisobutyric Aciduria
14	Profound Biotinidase deficiency
15	Beta- ketothiolase deficiency (BKT)
16	Canavan disease
17	Carbamoyl Phosphate Synthetase-1 Deficiency
18	Carnosinuria
19	Citrullinemia Type 1
20	Citrullinemia type II
21	Cystinuria
22	Cystathioninuria
23	Defects of biopterin cofactor regeneration (BIOPT REG)
24	Dihydrolipoyl Dehydrogenase (E3) Deficiency (also known as MSUD Type 3)
25	Familial Renal iminoglycinuria
26	Formiminoglutamic aciduria
27	Glutaric aciduria type II
28	Glutaric aciduria type I
29	Glutathionuria
30	Hartnup Disease
31	Hawkinsinuria
32	Hyperornithinemia-hyperammoninemia-hyperhomocitrullinemia (HHH) syndrome
33	Histidinuria
34	Histidinemia
35	Homocystinuria
36	Hyperprolinemia type I
37	Hyperprolinemia type II
38	Hydroxylysinuria
39	Hyperhydroxyprolinemia
40	Hyperleucine-isoleucinemia
41	Hyperglycinuria (Ketotic)
42	Hyperglycinuria (non-ketotic)
43	Hypermethioninemia
44	Hypersarcosinemia
45	Hyperlysinemia Type 1
46	Isobutyryl-CoA dehydrogenase deficiency (IBD)
47	Iminoglycinuria
48	Imidazole Amino Aciduria
49	Isovaleric acidemia
50	Lysinuric protein intolerance
51	Malonic acidemia
52	Maple syrup urine disease (MSUD)
53	Methylmalonic aciduria, cblA and cblB forms
54	Methylmalonic acidemia and Homocystinuria -Cbl C, D
55	Methylmalonic semialdehydede hydrogenase deficiency
56	Methylmalonyl- CoA mutase deficiency (MUT)
57	Mevalonic aciduria
58	Multiple carboxylase deficiency
59	N-Acetylglutamate Synthetase Deficiency
60	NICCD
61	Ornithine Trans Carbamylase Deficiency
62	Phenylketonuria (PKU)
63	Propionic acidemia
64	Saccharopinuria (also known as Hyperlysinemia Type II)
65	Succinic semialdehyde dehydrogenase deficiency
66	Tyrosinemia caused by liver dysfunction
67	Transient neonatal tyrosinemia
68	Tryptophanuria with dwarfism
69	Tyrosinemia Type II
70	Tyrosinemia Type III (also covers transient neonatal tyrosinemia)
71	Tyrosinemia Type I
72	Valinemia
73	Xanthurenic aciduria

B	TCA Cycle/Mitochondrial Abnormality
74	Fumarate hydratase deficiency
75	Leigh syndrome
76	Pyruvate carboxylase deficiency
77	Pyruvate decarboxylase deficiency
78	Pyruvate dehydrogenase (E1) deficiency
79	Pyruvate dehydrogenase phosphatase deficiency
C	**Disorders of Fatty Acid Metabolism**
80	Dicarboxylic aminoaciduria
81	Ethyl Malonic Aciduria
82	Glycerol Kinase Deficiency
83	Medium chain acyl CoA dehydrogenase deficiency
84	Mitochondrial trifunctional protein deficiency
85	Very Long-chain acyl-CoA dehydrogenase deficiency
86	Short chain acyl CoA dehydrogenase dehydrogenase deficiency
D	**Peroxisomal Disorders**
87	Infantile refsum disease
88	Neonatal Adrenoleukodystrophy
89	Primary hyperoxaluria Type 1
90	Primary hyperoxaluria Type 2
91	Zellweger syndrome
92	Zellweger like syndrome

E	Disorders of Purine and Pyrimidine Metabolism
93	Adenine phosphoribosyl transferase deficiency
94	Dihydropyrimidinase Deficiency
95	Hyperuric acidemia
96	Lesch-Nyhan syndrome
97	Molybdenum cofactor deficiency
98	Orotic aciduria
99	Partial deficiency of hypoxanthine-guanine phosphoribosyltransferase
100	Thymine Uraciluria
101	Xanthinuria (Type I & II)
F	**Disorders of Sugars**
102	D-glyceric aciduria
103	Endogenous sucrosuria
104	Fructose-1 and 6-diphosphatase deficiency
105	Fructosuria
106	Galactokinase deficiency (GALK)
107	Galactose epimerase deficiency (GALE)
108	Lactic Acidosis
109	Transient Galactosemia
G	**Non-IEM Disorder**
110	Neuroblastoma

Talk to the Experts:

1800 103 3691

diagnostics@medgenome.com

www.medgenome.com

Follow us on LinkedIn