MEDGENOME | ACTIA Inherited Disease Genetics

CAP ACCREDITED COLLEGE of AMERICAN PATHOLOGISTS

BabySecure

Advanced Newborn Screening portfolio to secure the baby from the impact of Inherited Disorders

What is BabySecure Newborn Screening and why it is important?

BabySecure Newborn screening test is a simple test done 24 hours after (not before) a baby is born to identify serious disorders the baby might have been born with. These disorders, if undetected and untreated, can have adverse consequences for the baby. If a baby is born with a disorder, BabySecure helps identify it early so that treatment can start before the disorder causes any harm to the baby.

When a BabySecure Newborn Screening test done?

The test is done by performing a painless heel prick on the baby and collecting a few drops of blood, 24 to 72 hours, after birth.

Is there a time limit for the BabySecure Newborn Screening test?

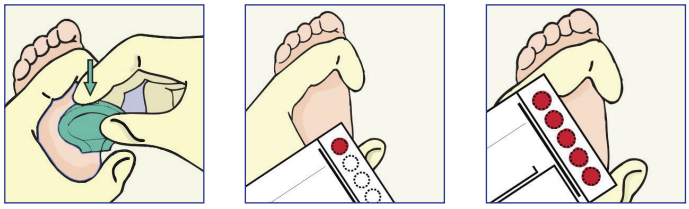
No, even if you miss the 72-hour window, you can still do the test. There is no age limit but sooner the better because if a disorder is identified, treatment can start immediately.

Can disorders be prevented?

These disorders are inherited and cannot be prevented but they are treatable. Even healthy looking babies and those with no family history of such disorders may have them. If a baby is born with a disorder, early detection and treatment is the only solution. Failure to start the treatment in time may result in serious consequences.

How the BabySecure Newborn Screening test is performed?

The first step is to make a painless heel prick. A few drops of blood from the baby's heel are then placed on a special type of filter paper. The paper is allowed to dry and is then sent to the lab where tests are performed using **Tandem Mass Spectrometry (TMS)** and other technologies.



Test details

Test Code	Test Name	Disorders Covered	Sample Type	Turnaround Time (TAT)
MGM2704	Newborn Screening (NBS+) - Comprehensive Panel	65	Dried Blood spots(FTA Cards)	24 - 48 Hours
MGM2705	Newborn Screening 1 (NBS1)	61	Dried Blood spots(FTA Cards)	24 - 48 Hours

BabySecure Newborn Screening Panels

	Disorders	NBS+	NBS1
	Disorders Screened by Tandem Mass Spectrometry (TMS)		
A	**Fatty Acid Oxidation Disorders**		
1	Carnitine / Acylcarnitine Translocase Deficiency	✓	✓
2	3-Hydroxy Long Chain Acyl-CoA Dehydrogenase Deficiency	✓	✓
3	Medium Chain Acyl-CoA Dehydrogenase Deficiency	✓	✓
4	Neonatal Carnitine Palmitoyl Transferase Deficiency Type II	✓	✓
5	Very Long Chain Acyl-CoA Dehydrogenase Deficiency	✓	✓
6	Carnitine Palmitoyl Transferase Deficiency Type I#	✓	✓
7	2,4-Dienoyl-CoA Reductase Deficiency	✓	✓
8	Multiple Acyl-CoA Dehydrogenase Deficiency	✓	✓
9	Short-chain Acyl-CoA Dehydrogenase Deficiency	✓	✓
10	Trifunctional Protein Deficiency	✓	✓
11	Short chain Hydroxy Acyl-CoA Dehydrogenase Deficiency	✓	✓
12	Medium Chain Ketoacyl-CoA Thiolase Deficiency	✓	✓
B	**Organic Acid Disorders**		
13	3-Hydroxy-3-Methylglutaryl-CoA Lyase Deficiency	✓	✓
14	Glutaric Acidemia Type I	✓	✓
15	Isobutyryl-CoA Dehydrogenase Deficiency	✓	✓
16	Isovaleric Acidemia	✓	✓
17	2-Methylbutyryl-CoA Dehydrogenase Deficiency	✓	✓
18	3-Methylcrotonyl-CoA Carboxylase Deficiency	✓	✓
19	3-Methylglutaconyl-CoA Hydratase Deficiency	✓	✓
20	2-Methyl-3-Hydroxybutyric Aciduria	✓	✓
	Methylmalonic Acidemias		
21	Methymalonyl-CoA Mutase Deficiency	✓	✓
22	Methylmalonic Acidemia (Cobalamin Disorders)	✓	✓
23	Methylmalonic Acidemia with Homocystinuria	✓	✓
24	Maternal Vitamin B12 Deficiency	✓	✓
25	Mitochondrial Acetoacetyl-CoA Thiolase Deficiency	✓	✓
26	Propionic Acidemia	✓	✓
27	Multiple CoA Carboxylase Deficiency	✓	✓
28	Malonic Aciduria	✓	✓
C	**Amino Acid Disorders**		
29	Argininemia	✓	✓
30	Argininosuccinic Aciduria	✓	✓
31	5-Oxoprolinuria	✓	✓

	Disorders	NBS+	NBS1
32	Carbamoylphosphate Synthetase Deficiency#	✓	✓
33	Ornithine Transcarbamylase Deficiency#	✓	✓
34	Citrullinemia	✓	✓
35	Citrullinemia Type II#	✓	✓
36	Homocystinuria	✓	✓
37	Hypermethioninemia	✓	✓
38	Hyperammonemia, Hyperornithinemia, Homocitrullinuria Syndrome#	✓	✓
39	Hyperornithinemia with Gyral Atrophy#	✓	✓
40	Maple Syrup Urine Disease	✓	✓
	Phenylketonuria		
41	*Classic Phenylketonuria*	✓	✓
42	*Benign Hyperphenylalaninemia*	✓	✓
43	*Defects of Biopterin Cofactor Biosynthesis*	✓	✓
44	*Defects of Biopterin Cofactor Regeneration*	✓	✓
	Tyrosinemia		
45	*Transient Neonatal Tyrosinemia*	✓	✓
46	*Tyrosinemia Type I#*	✓	✓
47	*Tyrosinemia Type II*	✓	✓
48	*Tyrosinemia Type III*	✓	✓
49	Nonketonic Hyperglycemia#	✓	✓
D	**Other Disorders**		
50	Liver Disease	✓	✓
51	Carnitine Uptake Deficiency	✓	✓
E	**Other Observations**		
52	Hyperalimentation	✓	✓
53	Medium Chain Triglyceride Oil Administration	✓	✓
54	Treatment with Benzoate, Pyvalic Acid, or Valproic Acid	✓	✓
55	Presence of EDTA Coagulants in Blood Specimen	✓	✓
	Disorders Screened by Other Technologies		
56	Congenital Hypothyroidism	✓	✓
57	Galactosemia	✓	✓
58	Congenital Adrenal Hyperplasia	✓	✓
59	Glucose-6-Phosphate Dehydrogenase Deficiency	✓	✓
60	Biotinidase Deficiency	✓	✓
	Phenylketonuria		
61	Cystic Fibrosis	✓	✓
62	Sickle Cell Anemia	✓	✗
63	Sickle-C Disease	✓	✗
64	S-ßeta Thalassemia	✓	✗
65	Hb Variants	✓	✗