Questions?
Call +91 99006 55114/17

NeoGen Labs

PATIENT DATA	FILTER PAPER DATA	SUBMITTER DATA
Name: Kishan	**Filter Paper No:** 99093285	**Name:** Mr. Nithin K. J.
Birth Date: 01 Aug 2024	**Collected:** 04 Dec 2025 02:21	**T:** +91 96322 88711
Sex: Male	**Received:** 04 Dec 2025 14:58	**Hospital:** Sparsh Hospital, Hennur
Weight (gms): 3300	**Transfused:**	**Hospital No:**
Gestation (weeks): 40	**Panel:** SMA	**T:**
Mother: Manasa	**Reported:** 09 Dec 2025 15:44	**Physician:** Dr. Ann Agnes Mathew
T: +91 72598 75115	**Specimen:** Dried Blood Spot (DBS)	**T:**

LABORATORY REPORT FOR First Step™ NEWBORN SCREENING TEST

SMA (qPCR) Survival Motor Neuron 1 (SMN 1) gene (Ch 5 Exon 7)	**ABNORMAL** **Probable Spinal Muscular Atrophy. Deletion of SMN1 Exon 7** Recommend a confirmatory test for SMN 1 gene mutation and SMN 2 copy number quantification. Referral to a pediatric neurologist / neuro-geneticist at the earliest for further work up.

QNS Quantity Not Sufficient **NA** Not Applicable Residual specimen is stored for retesting purposes.

NOTES:
1 year 4 month old, sick baby. C/o hypotonia.
This test is meant to screen for SMA but cannot differentiate between SMA type 1, SMA type 2, SMA type 3 or SMA type 4. SMA screening is carried out for SMN 1 gene mutation (deletion) of Exon 7 region, that accounts for approximately 95% of SMA disorders detected. For this reason, a negative screening result does not completely rule out the possibility of the disorder.

For NeoGen Labs Pvt. Ltd.
Authorised Signatory
Dr. ROHIT CARIAPPA
Lab Director

For NeoGen Labs Pvt Ltd
Dr. Siva Adarsh
Consultant Biochemist PhD

This test is CAP accredited vide certification number 8962127

End of Report

NeoGen Labs Private Limited
UCF Center, 2nd Floor, Hennur Main Road
Lingarajapuram, Bangalore 560 084, Karnataka INDIA
T: +91 80 2580 5600 **E:** report@neogenlabs.com **W:** www.neogenlabs.com
CIN: U33125GJ2006PTC048588

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1st Step Screening Panel

Disorders Screened by Tandem Mass Spectrometry

Acylcarnitine Profile - Fatty Acid Oxidation Disorders

1. Carnitine / Acylcarnitine Translocase Deficiency (CACT)
2. 3-Hydroxy Long Chain Acyl-CoA Dehydrogenase Deficiency (LCHAD)
3. Medium Chain Acyl-CoA Dehydrogenase Deficiency (MCAD)
4. Neonatal Carnitine Palmitoyl Transferase Deficiency Type II (CPT-II)
5. Very Long Chain Acyl-CoA Dehydrogenase Deficiency (VLCAD)
6. Carnitine Palmitoyl Transferase Deficiency Type IA (CPT-IA) [1]
7. 2,4-Dienoyl-CoA Reductase Deficiency (DE-RED)
8. Multiple Acyl-CoA Dehydrogenase Deficiency (MADD or GA-II)
9. Short-chain Acyl-CoA Dehydrogenase Deficiency (SCAD)
10. Trifunctional Protein Deficiency (TFP)
11. Short chain Hydroxy Acyl-CoA Dehydrogenase Deficiency (SCHAD)
12. Medium Chain Ketoacyl-CoA Thiolase Deficiency (MCKAT)

Acylcarnitine Profile - Organic Acid Disorders

13. 3-Hydroxy-3-Methylglutaryl-CoA Lyase Deficiency (HMG)
14. Glutaric Acidemia Type I (GA-1)
15. Isobutyryl-CoA Dehydrogenase Deficiency (IBG)
16. Isovaleric Acidemia (IVA)
17. 2-Methylbutyryl-CoA Dehydrogenase Deficiency (2MBG)
18. 3-Methylcrotonyl-CoA Carboxylase Deficiency (3MCC)
19. 3-Methylglutaconyl-CoA Hydratase Deficiency (3MGA)
20. 2-Methyl-3-Hydroxybutyric Aciduria (2M3HBA)
21. Methymalonyl-CoA Mutase Deficiency (MUT)
22. Methylmalonic Acidemia (Cobalamin Disorders): Cbl A, B
23. Methylmalonic Acidemia with Homocystinuria: Cbl C, D
24. Maternal Vitamin B12 Deficiency
25. Mitochondrial Acetoacetyl-CoA Thiolase Deficiency (BKT)
26. Propionic Acidemia (PROP)
27. Multiple CoA Carboxylase Deficiency (MCD)
28. Malonic Aciduria (MAL)

Amino Acid Profile - Amino Acid Disorders

29. Argininemia (ARG)
30. Argininosuccinic Aciduria (ASA Lyase)
31. 5-Oxoprolinuria (5-OXO or Pyroglutamic Aciduria)
32. Carbamoylphosphate Synthetase Deficiency (CPS) [1]
33. Ornithine Transcarbamylase Deficiency (OTC) [1]
34. Citrullinemia (CIT-I or ASA Synthetase)
35. Citrullinemia Type II (CIT-II) / Citrin Deficiency [1]
36. Homocystinuria (HCY)
37. Hypermethioninemia (MET)
38. Hyperammonemia, Hyperornithinemia, Homocitrullinuria Syndrome (HHH Syndrome) [1]
39. Hyperornithinemia with Gyral Atrophy (HOGA) [1]
40. Maple Syrup Urine Disease (MSUD)
41. Phenylketonuria (PKU)
42. Benign Hyperphenylalaninemia (H-PHE)
43. Defects of Biopterin Cofactor Biosynthesis (BIOPT BS)
44. Defects of Biopterin Cofactor Regeneration (BIOPT REG)
45. Transient Tyrosinemia of the Newborn (TTN)
46. Tyrosinemia Type I (TYR - I) [1]
47. Tyrosinemia Type II (TYR - II)
48. Tyrosinemia Type III (TYR-III)
49. Nonketotic Hyperglycinemia (NKHG) [1]

Other

50. Hyperalimentation
51. Medium Chain Triglyceride Oil Administration
52. Treatment with Benzoate, Pyvalic Acid, or Valproic Acid
53. Liver Disease
54. Presence of EDTA Coagulants in Blood Specimen
55. Carnitine Uptake Deficiency (CUD)

Disorders Screened by Other Technologies

56. Congenital Hypothyroidism (CH)
57. Galactosemia (TGAL)
58. Congenital Adrenal Hyperplasia (CAH)
59. Glucose-6-Phosphate Dehydrogenase Deficiency (G6PD)
60. Biotinidase Deficiency (BIOT)
61. Cystic Fibrosis (CF)
62. Sickle Cell Anemia (Hb S/S)
63. Sickle-C Disease (Hb S/C)
64. S-βeta Thalassemia (Hb S/βTh)
65. Hb Variants (Var Hb)
66. βeta Thalassemia Major [1]
67. Spinal Muscular Atrophy (SMA)

Newborn Screening Limitations

Due to biologic variability in newborns and differences in time of onset for the various disorders, newborn Screening will not identify all newborns with these conditions. While a positive screening result identifies newborns at an increased risk to justify a diagnostic work-up, a negative screening result does not rule out the possibility of a disorder. Healthcare providers should remain vigilant for any signs or symptoms of these disorders in their patients. The screening results are to be clinically correlated. The screening services and materials are not a substitute for medical advice, diagnosis, or treatment.

Normal Ranges

Acylcarnitine (µM)

Analyte	1 to 7 days		8+ days	
	Lower Limit	Upper Limit	Lower Limit	Upper Limit
Free CN	5.00	150	5.00	125
C2	2.00	80.0	2.00	50.0
C3	0.20	6.00	0.20	4.00
C3DC	< 0.36		< 0.15	
C4	< 1.00		< 1.00	
C5	< 1.00		< 0.80	
C5:1	0.01 - 0.35		0.01 - 0.04	
C4-OH	< 0.80		< 0.80	
C5-OH	< 1.00		< 1.00	
C8	< 0.40		< 0.30	
C10	< 0.50		< 0.50	
C5DC	< 0.17		< 0.15	
C12	< 0.50		< 0.50	
C14:1	0.01	0.7	0.01	0.52
C14	< 0.85		< 0.60	
C16	0.06	10.0	0.06	6.00
C16-OH	< 0.16		< 0.10	

Amino Acids (µM)

Analyte	Lower Limit	Upper Limit
Val	< 325	
Leu-Ile	< 375	
Met	6.00	62.0
Cit	3.00	75.0
Phe	< 150	
Tyr	< 300	
Orn	< 450	
Arg	< 110	
Ala	< 960	
GLY	< 800	
SUAC	< 0.75	

References for the Biological Reference Intervals **https://www.neogenlabs.com/references.pdf**

[1] There is a lower probability of detection of this disorder during the immediate newborn period