# Questions?

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| PATIENT DATA | FILTER PAPER DATA | SUBMITTER DATA |
|---|---|---|

## LABORATORY REPORT FOR First Step™ NEWBORN SCREENING TEST

| | |
|---|---|
| **Acylcarnitine Profile** (TMS)<br>Fatty Acid Oxidation & Organic Acid Disorders | **Within Normal Limits** |
| **Amino Acid Profile** (TMS)<br>Amino Acid Disorders, Urea Cycle Disorders | **Within Normal Limits** |
| **G6PD Deficiency** (FEA)<br>Glucose-6-Phosphate Dehydrogenase (G6PD)<br>**Normal**: > 2 U/gHb **Deficient:** < 2 U/gHb | **Within Normal Limits**<br>G6PD Enzyme Activity > 7.8 U/gHb |
| **Congenital Adrenal Hyperplasia** (FEIA)<br>17-hydroxyprogesterone (17-OHP)<br>**Normal**: < 20 ng/mL **High:** > 20 ng/mL | **Within Normal Limits**<br>17-OHP < 0.05 ng/mL |
| **Cystic Fibrosis** (FEIA)<br>Immunoreactive Trypsinogen (IRT)<br>Not valid after 2 months of age<br>**Normal**: < 110 µg/L **High:** > 110 µg/L | **NA** |
| **Galactosemia** (Fluorimetry)<br>Total Galactose (TGAL)<br>**Normal**: < 20 mg/dL **High:** > 20 mg/dL | **Within Normal Limits**<br>TGAL < 4 mg/dL |
| **Congenital Hypothyroidism** (FEIA)<br>Thyroid Stimulating Hormone (TSH)<br>**Normal**: < 10 µIU/mL **High:** > 10 µIU/mL | **Within Normal Limits**<br>TSH = 1.71 µIU/mL |
| **Biotinidase Deficiency** (FEA)<br>Biotinidase (BIOT)<br>**Normal**: > 40 U **Deficient:** < 40 U | **Within Normal Limits**<br>Biotinidase Enzyme Activity = 286.45 U |
| **Hemoglobinopathies** (HPLC)<br>Sickle Cell & Other Hemoglobinopathies<br>Not valid after 3 months of age | **NA** |

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

**NOTES**:
3 year 9 month old baby.

For NeoGen Labs Pvt. Ltd.
Authorised Signatory
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For NeoGen Labs Pvt. Ltd
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End of Report

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NeoGen Labs

## Disorders Detected By Tandem Mass Spectrometry

### Acylcarnitine Profile - Fatty Acid Oxidation Disorders

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

### Acylcarnitine Profile - Organic Acid Disorders

13 1 3-Hydroxy-3-Methylglutaryl-CoA Lyase Deficiency (HMG)
14 2 Glutaric Acidemia Type I (GA-I)
15 3 Isobutyryl-CoA Dehydrogenase Deficiency (IBG)
16 4 Isovaleric Acidemia (IVA)
17 5 2-Methylbutyryl-CoA Dehydrogenase Deficiency (2MBG)
18 6 3-Methylcrotonyl-CoA Carboxylase Deficiency (3MCC)
19 7 3-Methylglutaconyl-CoA Hydratase Deficiency (3MGA)
20 8 2-Methyl-3-Hydroxybutyric Aciduria (2M3HBA)
21 9 Methymalonyl-CoA Mutase Deficiency (MUT)
22 10 Methylmalonic Acidemia (Cobalamin disorders): Cbl A, B
23 11 Methylmalonic Acidemia with Homocystinuria: Cbl C, D
24 12 Maternal Vitamin B12 Deficiency
25 13 Mitochondrial Acetoacetyl-CoA Thiolase Deficiency (BKT)
26 14 Propionic Acidemia (PROP)
27 15 Multiple CoA Carboxylase Deficiency (MCD)
28 16 Malonic Aciduria (MAL)

### Amino Acid Profile - Amino Acid Disorders

29 1 Argininemia (ARG)
30 2 Argininosuccinic Aciduria (ASA Lyase)
31 3 5-Oxoprolinuria (5-OXO or Pyroglutamic Aciduria)
32 4 Carbamoylphosphate Synthetase Deficiency (CPS)[1]
33 5 Ornithine Transcarbamylase Deficiency (OTC)[1]
34 6 Citrullinemia (CIT-I or ASA Synthetase)
35 7 Citrullinemia Type II (CIT-II) / Citrin Deficiency[1]
36 8 Homocystinuria (HCY)
37 9 Hypermethioninemia (MET)
38 10 Hyperammonemia, Hyperornithinemia, Homocitrullinuria Syndrome (HHH Syndrome)[1]
39 11 Hyperornithinemia with Gyral Atrophy (HOGA)[1]
40 12 Maple Syrup Urine Disease (MSUD)
41 13 Phenylketonuria (PKU)
42 14 Benign Hyperphenylalaninemia (H-PHE)
43 15 Defects of Biopterin Cofactor Biosynthesis (BIOPT BS)
44 16 Defects of Biopterin Cofactor Regeneration (BIOPT REG)
45 17 Transient Tyrosinemia of the Newborn (TTN)
46 18 Tyrosinemia Type I (TYR-I)[1]
47 19 Tyrosinemia Type II (TYR-II)
48 20 Tyrosinemia Type III (TYR-III)
49 21 Nonketotic Hyperglycinemia (NKHG)[1]

### Other

50 1 Hyperalimentation
51 2 Medium Chain Triglyceride Oil Administration
52 3 Treatment with Benzoate, Pyvalic Acid, or Valproic Acid
53 4 Liver Disease
54 5 Presence of EDTA Anticoagulants in blood specimen
55 6 Carnitine Uptake Deficiency (CUD)

## Disorders Detected By Other Technologies

56 1 Congenital Hypothyroidism (CH)
57 2 Galactosemia (TGAL)
58 3 Congenital Adrenal Hyperplasia (CAH)
59 4 Glucose-6-Phosphate Dehydrogenase Deficiency (G6PD)
60 5 Biotinidase Deficiency (BIOT)
61 6 Cystic Fibrosis (CF)
62 7 Sickle Cell Anemia (Hb S/S)
63 8 Sickle-C Disease (Hb S/C)
64 9 S-βeta Thalassemia (Hb S/βTh)
65 10 Hb Variants (Var Hb)

## Newborn Screening Limitations

**Due to biologic variability of newborns and differences in detection rates for the various disorders in the newborn period, 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 process is best coordinated with a physician. 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 |
| C4 | < 1.00 | | < 1.00 | |
| C5 | < 1.00 | | < 0.80 | |
| 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 | |

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

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

First Step Screening Panel

1st STEP Screening Test

Rohit Cariappa, PhD, DABCC

Where babies take their first step

# Questions?

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## ACYLCARNITINES

| Analyte | Result (μM) | Normal Range (μM) |
|---|---|---|
| FreeCN | 28.292 | 5 to 125 |
| C2 | 8.585 | 2 to 50 |
| C3 | 1.986 | 0.2 to 4 |
| C4 | 0.411 | <1 |
| C5:1 | 0.041 | 0.01 to 0.4 |
| C5 | 0.29 | <0.8 |
| C4-OH | 0.041 | <0.8 |
| C6 | 0.035 | 0.01 to 0.16 |
| C5-OH | 0.127 | <1 |
| C8:1 | 0.059 | 0 to 0.33 |
| C8 | 0.023 | <0.3 |
| C3DC(C8OH) | 0.035 | 0 to 0.15 |
| C10:2 | 0.001 | 0 to 0.1 |
| C10:1 | 0.012 | 0.01 to 0.25 |
| C10 | 0.001 | <0.5 |

| Analyte | Result (μM) | Normal Range (μM) |
|---|---|---|
| C4DC | 0.093 | 0.03 to 0.61 |
| C5DC(C10-OH) | 0.001 | <0.15 |
| C12:1 | 0.015 | 0.01 to 0.33 |
| C12 | 0.044 | <0.5 |
| C14:2 | 0.001 | 0 to 0.14 |
| C14:1 | 0.044 | 0.01 to 0.52 |
| C14 | 0.161 | <0.6 |
| C16:1 | 0.037 | 0 to 1 |
| C16 | 1.068 | 0.06 to 6 |
| C16-OH | 0.001 | <0.1 |
| C18:2 | 0.061 | 0 to 1 |
| C18:1 | 0.473 | 0.2 to 4 |
| C18 | 0.449 | 0.13 to 3 |
| C18:1-OH | 0.037 | 0.01 to 0.15 |
| C18-OH | 0.024 | 0 to 0.18 |

## AMINO ACIDS

| Analyte | Result (μM) | Normal Range (μM) |
|---|---|---|
| Gly MRM | 163.439 | 0 to 800 |
| Ala | 301.747 | <960 |
| Val | 173.874 | <325 |
| Gln (Glu) | 35.54 | 19 to 265.83 |
| Leu-Ile | 200.62 | <375 |
| Met | 28.424 | 6 to 62 |
| His | 16.657 | |
| SUAC | 0.14 | 0 to 0.75 |

| Analyte | Result (μM) | Normal Range (μM) |
|---|---|---|
| Phe | 85.481 | <150 |
| Tyr | 95.884 | <300 |
| Asp | 67.319 | 9 to 108.1 |
| Glu | 200.554 | 0 to 2000 |
| Ser | 60.745 | |
| Orn | 112.631 | <450 |
| Cit(119) | 59.738 | 3 to 75 |
| Arg | 69.75 | <110 |

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