# Annexure 1

**Patient Name:** DNANB001A001L387

## 1. Amino acids:

*(Amino acid & Urea Cycle Disorders)*

| S.No | Metabolite | Conc. (uM) | Reference Ranges | S.No | Metabolite | Conc. (uM) | Reference Ranges |
|---|---|---|---|---|---|---|---|
| 1 | Alanine | 333.20 | 103-742 | 8 | Methionine | 28.05 | 5-41 |
| 2 | Arginine | 4.35 | 1-41 | 9 | Ornithine | 61.34 | 10-263 |
| 3 | Aspartic acid | 51.18 | 10-345 | 10 | Phenylalanine | 75.84 | 10-102 |
| 4 | Citrulline | 6.37 | 5-43 | 11 | Proline | 189.73 | 87-441 |
| 5 | Glutamic acid | 503.67 | 152-708 | 12 | Tyrosine | 80.92 | 15-259 |
| 6 | Glycine | 511.00 | 0-1142 | 13 | Valine | 140.41 | 52-322 |
| 7 | Leucine | 154.91 | 27-324 | | | | |

## 2. Amino acids molar ratios:

| S.No | Ratios | Values | Ranges | S.No | Ratios | Values | Ranges |
|---|---|---|---|---|---|---|---|
| 1 | Met / Leu | — | <0.42 | 4 | Leu / Ala | — | 0.12-1.00 |
| 2 | Met / Phe | — | <0.70 | 5 | Leu / Tyr | — | 0.50-3.50 |
| 3 | Phe / Tyr (PKU) | — | <2.00 | | | — | |

## 3. Acylcarnitines:

*(fatty Acid Oxidation defects & Organic Acid Disorders)*

| S.No | Metabolite | Conc. (uM) | Reference Ranges | S.No | Metabolite | Con. (uM) | Reference Ranges |
|---|---|---|---|---|---|---|---|
| 1 | Free CN (C0) | 33.00 | 5-125 | 8 | Methylmalonylcarnitine (C4DC) | 0.34 | 0.1-1.25 |
| 2 | Total Carnitines | — | - | 9 | Isovalerylcarnitine (C5) | 0.88 | 0.01-1 |
| 3 | Acetylcarnitine (C2) | 33.49 | 1.4-80 | 10 | 3-methylcrotonylcarnitine (C5:1) | 0.05 | 0.01-0.9 |
| 4 | Propionylcarnitine (C3) | 3.62 | 0.18-6.3 | 11 | Glutarylcarnitine (C5DC) | 3.60 | 0.01-2.99 |
| 5 | Malonylcarnitine (C3DC) | 0.46 | 0.1-0.45 | 12 | 3-OH- Isovalerylcarnitine (C5OH) | 0.54 | 0.01-0.9 |
| 6 | Butyrylcarnitine (C4) | 1.05 | 0.08-1.7 | 13 | Hexanoylcarnitine (C6) | 0.05 | 0.01-0.95 |
| 7 | 3-OH-Butyrylcarnitine (C4OH) | 0.66 | 0.01-1.29 | 14 | Methylglutarylcarnitine (C6DC) | 0.17 | 0.01-0.23 |

| S.No | Metabolite | Conc. (uM) | Reference Ranges | S.No | Metabolite | Con. (uM) | Reference Ranges |
|---|---|---|---|---|---|---|---|
| 15 | Octanoylcarnitine (C8) | 0.17 | 0.01-0.6 | 26 | Palmitoylcarnitine (C16) | 1.89 | 0.34-10.35 |
| 16 | Octenoylcarnitine (C8:1) | 0.13 | 0.01-0.7 | 27 | Hexadecenoylcanitine (C16:1) | 0.10 | 0.01-1.4 |
| 17 | Decanoylcarnitine (C10) | 0.06 | 0.02-0.65 | 28 | 3-Hydroxypalmitoleyl carnitine (C16:1OH) | 0.02 | 0.01-0.1 |
| 18 | Decenoylcarnitine (C10:1) | 0.06 | 0.01-0.45 | 29 | Hexadecenoylcanitine (C16OH) | 0.00 | 0.01-0.1 |
| 19 | Decadienoyl carnitine (C10:2) | 0.07 | 0.01-0.22 | 30 | Stearoylcarnitine (C18) | 0.50 | 0.21-2.03 |
| 20 | Dodecanoyl carnitine (C12) | 0.17 | 0.02-0.6 | 31 | Octadecadienoyl carnitine (C18:2) | 0.10 | 0.1-0.73 |
| 21 | Dodecenoyl carnitine (C12:1) | 0.04 | 0.01-0.5 | 32 | Octadecenoylcarnitine (C18:1) | 0.78 | 0.5-7 |
| 22 | Myristoyla carnitine (C14) | 0.21 | 0.01-1.22 | 33 | 3-Hydroxylinoleoyl carnitine (C18:2OH) | 0.02 | 0.01-0.03 |
| 23 | Tetradecenoyl carnitine (C14:1) | 0.09 | 0.01-0.8 | 34 | 3-OH-Octadecenoylcarnitine (C18:1OH) | 0.02 | 0.01-0.1 |
| 24 | Tetradecadienoyl carnitine(C14:2) | 0.05 | 0-0.2 | 35 | 3-OH-Stearoylcarnitine (C18OH) | 0.01 | 0.01-0.1 |
| 25 | 3-OH-Tetradecenoyl carnitine (C14OH) | 0.01 | 0-0.2 | | | | |

## 4. Acylcarnitine molar ratios:

| S.No | Ratios | Values | Ranges | S.No | Ratios | Values | Ranges |
|---|---|---|---|---|---|---|---|
| 1 | C4 / C3 | — | <1.18 | 6 | C0 / ( C16 + C18) | — | <70 |
| 2 | C3 / C0 | — | <0.27 | 7 | C5 / C2 | — | <0.16 |
| 3 | C3 / C2 | — | <0.45 | 8 | C5 / C3 | — | <0.29 |
| 4 | C8 / C10 | — | < 1.50 | 9 | C5DC / C3 | — | <0.27 |
| 5 | C8 / C2 | — | <0.03 | 10 | C5DC / C16 | — | <0.68 |

# Annexure 2

**Patient Name:** DNANB001A001L387

Results Biochemical Parameters:

| Biochemical Parameters | | |
|---|---|---|
| **Assay** | **Result** | **Reference Ranges** |
| Thyroid Stimulating Hormone (TSH)<br>(Congenital Hypothyroidism (CH) | — | < 15 ulU/mL |
| 17-hydroxyprogrestrone (17-OHP)<br>(Congenital Adrenal Hyperplasia (CAH) | — | <30 ng/mL (BW >2250g)<br><50 ng/mL (BW<2250g) |
| G6PD enzyme activity<br>(G6PD Deficiency) | — | > 1.5 U/gHb |
| Total Galactose (TGAL)<br>(Galactosemia (GAL) | — | < 15 mg/dL |
| Immunoreactive trypsinogen (IRT)<br>(Cystic Fibrosis -CF) | — | < 90 µg/L |
| Biotinidase (BIOT)<br>(Biotinidase) | — | 31.6 - 388 U |

*********End Of Report********

Disclaimer: The laboratory values in this report represent "screening" results and are intended to identify NEWBORNS at risk for selected disorders and may need for more definitive testing. "NORMAL" refers to the analyte(s) measured. NOT ALL BABIES AT RISK for screened disorders will be detected and the above results should be clinically correlated with the following factors at the time of collection: age, birth weight or current weight, prematurity, nutrition, health status, and treatments (IV glucose, transfusions, antibiotics, TPN/hyperalimentation, etc.

## Disorders Included in the test panel

| Disorders list would go here |
| --- |