

## Annexure 1

Patient Name: DNANB001A001L307

### 1. Amino acids:

(Amino acid & Urea Cycle Disorders)

S.No	Metabolite	Conc. (uM)	Reference Ranges	S.No	Metabolite	Conc. (uM)	Reference Ranges
1	Alanine	302.25	103-742	8	Methionine	23.62	5-41
2	Arginine	7.01	1-41	9	Ornithine	64.40	10-263
3	Aspartic acid	70.28	10-345	10	Phenylalanine	62.38	10-102
4	Citrulline	6.21	5-43	11	Proline	180.62	87-441
5	Glutamic acid	503.02	152-708	12	Tyrosine	56.42	15-259
6	Glycine	587.57	0-1142	13	Valine	100.83	52-322
7	Leucine	154.75	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	Conc. (uM)	Reference Ranges
1	Free CN (C0)	52.04	5-125	8	Methylmalonylcarnitine (C4DC)	0.16	0.1-1.25
2	Total Carnitines	—	-	9	Isovalerylcarnitine (C5)	1.06	0.01-1
3	Acetylcarnitine (C2)	30.16	1.4-80	10	3-methylcrotonylcarnitine (C5:1)	0.09	0.01-0.9
4	Propionylcarnitine (C3)	2.26	0.18-6.3	11	Glutaryl carnitine (C5DC)	2.06	0.01-2.99
5	Malonylcarnitine (C3DC)	0.02	0.1-0.45	12	3-OH- Isovalerylcarnitine (C5OH)	0.60	0.01-0.9
6	Butyrylcarnitine (C4)	0.35	0.08-1.7	13	Hexanoylcarnitine (C6)	0.09	0.01-0.95
7	3-OH- Butyrylcarnitine (C4OH)	0.42	0.01-1.29	14	Methylglutaryl carnitine (C6DC)	0.24	0.01-0.23

S.No	Metabolite	Conc. (uM)	Reference Ranges	S.No	Metabolite	Con. (uM)	Reference Ranges
15	Octanoylcarnitine (C8)	0.12	0.01-0.6	26	Palmitoylcarnitine (C16)	3.44	0.34-10.35
16	Octenoylcarnitine (C8:1)	0.11	0.01-0.7	27	Hexadecenoylcarnitine (C16:1)	0.14	0.01-1.4
17	Decanoylcarnitine (C10)	0.07	0.02-0.65	28	3-Hydroxypalmitoleyl carnitine (C16:1OH)	0.00	0.01-0.1
18	Decenoylcarnitine (C10:1)	0.05	0.01-0.45	29	Hexadecenoylcarnitine (C16OH)	0.02	0.01-0.1
19	Decadienoyl carnitine (C10:2)	0.06	0.01-0.22	30	Stearoylcarnitine (C18)	0.85	0.21-2.03
20	Dodecanoyl carnitine (C12)	0.32	0.02-0.6	31	Octadecadienoyl carnitine (C18:2)	0.07	0.1-0.73
21	Dodecenoyl carnitine (C12:1)	0.11	0.01-0.5	32	Octadecenoylcarnitine (C18:1)	0.87	0.5-7
22	Myristoyla carnitine (C14)	0.31	0.01-1.22	33	3-Hydroxylinoleoyl carnitine (C18:2OH)	0.00	0.01-0.03
23	Tetradecenoyl carnitine (C14:1)	0.14	0.01-0.8	34	3-OH-Octadecenoylcarnitine (C18:1OH)	0.01	0.01-0.1
24	Tetradecadienoyl carnitine (C14:2)	0.08	0-0.2	35	3-OH-Stearoylcarnitine (C18OH)	0.02	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:** DNANB001A001L307

**Results Biochemical Parameters:**

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

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## Disorders Included in the test panel

Disorders list would go here

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