

## Annexure 1

Patient Name: DNANB001A001L381

### 1. Amino acids:

(Amino acid & Urea Cycle Disorders)

S.No	Metabolite	Conc. (uM)	Reference Ranges	S.No	Metabolite	Conc. (uM)	Reference Ranges
1	Alanine	363.26	103-742	8	Methionine	20.88	5-41
2	Arginine	6.37	1-41	9	Ornithine	42.88	10-263
3	Aspartic acid	45.78	10-345	10	Phenylalanine	71.98	10-102
4	Citrulline	8.95	5-43	11	Proline	207.95	87-441
5	Glutamic acid	463.93	152-708	12	Tyrosine	120.74	15-259
6	Glycine	360.28	0-1142	13	Valine	106.55	52-322
7	Leucine	158.14	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)	44.98	5-125	8	Methylmalonylcarnitine (C4DC)	0.20	0.1-1.25
2	Total Carnitines	—	-	9	Isovalerylcarnitine (C5)	1.03	0.01-1
3	Acetylcarnitine (C2)	24.14	1.4-80	10	3-methylcrotonylcarnitine (C5:1)	0.07	0.01-0.9
4	Propionylcarnitine (C3)	1.96	0.18-6.3	11	Glutarylcarnitine (C5DC)	0.15	0.01-2.99
5	Malonylcarnitine (C3DC)	0.21	0.1-0.45	12	3-OH- Isovalerylcarnitine (C5OH)	0.83	0.01-0.9
6	Butyrylcarnitine (C4)	0.51	0.08-1.7	13	Hexanoylcarnitine (C6)	0.05	0.01-0.95
7	3-OH- Butyrylcarnitine (C4OH)	0.47	0.01-1.29	14	Methylglutarylcarnitine (C6DC)	0.08	0.01-0.23

S.No	Metabolite	Conc. (uM)	Reference Ranges	S.No	Metabolite	Conc. (uM)	Reference Ranges
15	Octanoylcarnitine (C8)	0.06	0.01-0.6	26	Palmitoylcarnitine (C16)	2.25	0.34-10.35
16	Octenoylcarnitine (C8:1)	0.08	0.01-0.7	27	Hexadecenoylcarnitine (C16:1)	0.06	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)	0.12	0.01-0.45	29	Hexadecenoylcarnitine (C16OH)	0.01	0.01-0.1
19	Decadienoyl carnitine (C10:2)	0.04	0.01-0.22	30	Stearoylcarnitine (C18)	0.90	0.21-2.03
20	Dodecanoyl carnitine (C12)	0.35	0.02-0.6	31	Octadecadienoyl carnitine (C18:2)	0.10	0.1-0.73
21	Dodecenoyl carnitine (C12:1)	0.02	0.01-0.5	32	Octadecenoylcarnitine (C18:1)	0.77	0.5-7
22	Myristoyl carnitine (C14)	0.33	0.01-1.22	33	3-Hydroxylinoleyl carnitine (C18:2OH)	0.02	0.01-0.03
23	Tetradecenoyl carnitine (C14:1)	0.20	0.01-0.8	34	3-OH-Octadecenoylcarnitine (C18:1OH)	0.02	0.01-0.1
24	Tetradecadienoyl carnitine (C14:2)	0.02	0-0.2	35	3-OH-Stearoylcarnitine (C18OH)	0.02	0.01-0.1
25	3-OH-Tetradecenoyl carnitine (C14OH)	0.02	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:** DNANB001A001L381

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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