

## 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	Conc. (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	Conc. (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	Hexadecenoylcarnitine (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	Hexadecenoylcarnitine (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	Myristoyl carnitine (C14)	0.21	0.01-1.22	33	3-Hydroxylinoleyl 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) (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.

NBS Report for DNANB001A001L387

## Disorders Included in the test panel

S.No	Amino Acid Disorders	S.No	Acylcarnitine and Organic acid Disorders
1	(ARG) Argininemia	1	(CACT) Carnitine Acylcarnitine Translocase Deficiency
2	(ASA) Argininosuccinic Aciduria	2	(CPT-IA) Carnitine Palmitoyltransferase Type I Deficiency
3	(CIT-I) Citrullinemia, Type I	3	(CPT-II) Carnitine Palmitoyltransferase Type II Deficiency
4	(CIT-II) Citrullinemia Type II	4	(LCHAD) Long-Chain L-3 Hydroxyacyl-CoA Dehydrogenase Deficiency
5	(CPS-1) Carbomyl phosphate synthetase 1 deficiency	5	(DE-RED) 2,4-Dienoyl-CoA Reductase Deficiency
6	(BIOPT-BS) defects of biopterin cofactor biosynthesis	6	(CUD) Carnitine Uptake Defect
7	(BIOPT-RG) defects of biopterin cofactor regeneration	7	(MCAD) Medium-Chain Acyl-CoA Dehydrogenase Def.
8	(HCY) Homocystinuria	8	(MADD) Multiple Acy-CoA Dehydrogenase Deficiency
9	(H-PHE) Hyperphenylalaninemia	9	(SCAD) Short-chain Acyl-CoA Dehydrogenase Deficiency
10	(HHH syndrome 1) Hyperammonemia, Hyperornithinemia and Homocitrullinemia	10	(M/SCHAD) Medium/Short-Chain L-3-Hydroxyacyl-CoA Dehydrogenase Deficiency
11	(HOGA) Hyperornithinemia with Gyral Atrophy	11	(TFP) Trifunctional Protein Deficiency
12	(H-PHE) Benign Hyperphenylalaninemia	12	(CBL C, D) Methylmalonic acidemia with homocystinuria
13	Hyperprolinemia	13	(IVA) Isovaleric Acidemia
14	Hyperalimentation	14	(GA1) Glutaric Acidemia, Type I
15	(MET) Hypermethioninemia	15	(2MBG) 2-Methylbutyryl-CoA Dehydrogenase Deficiency
16	(MSUD) Maple Syrup Urine Disease	16	(MCD) Multiple CoA Carboxylase Deficiency
17	(5-OXO) 5-Oxoprolinuria	17	(3MCC) 3-Methylcrotonyl-CoA Carboxylase Deficiency
18	Ornithine transcarbamylase deficiency	18	(3MGA) 3-Methylglutaconyl-CoA Hydratase Deficiency
19	(PKU) Classic Phenylketonuria	19	(MMA) Methylmalonic Acidemias
20	Pyruvate decarboxylase deficiency	20	(MUT) Methylmalonyl-CoA Mutase Deficiency
21	(TYR-1) Tyrosinemia	21	(VLCAD) Very Long-Chain Acyl-CoA Dehydrogenase Deficiency
22	(TYR-II) Tyrosinemia	22	(BKT) Mitochondrial Acetoacetyl-CoA Thiolase Deficiency
23	(TYR-III) Tyrosinemia	23	(MCAT) Medium-chain Ketoacyl-CoA Thiolase Deficiency
24	Liver Disease	24	Short chain Hydroxy Acyl-CoA Dehydrogenase Deficiency
		25	Maternal Vitamin B12 Deficiency
		26	(MAL) Malonic Aciduria
		27	(PROP) Propionic Acidemia
		28	GA-II Multiple Acyl-CoA Dehydrogenase Deficiency GAI
		29	(HMG) 3-Hydroxy-3-methylglutaryl-CoA Lyase Deficiency
		30	(IBG) Isobutyryl-CoA Dehydrogenase Deficiency
		31	(MCT )Medium Chain Triglyceride Oil Administration
		32	Methylmalonic Acidemia (Cobalamin disorders) Cbl A, B