Annexure 1

Patient Name: **DNANB001A001J696**

1. Amino acids:

(Amino acid & Urea Cycle Disorders)

S.No	Metabolite	Conc. (uM)	Reference Ranges	S.No	Metabolite	Conc. (uM)	Reference Ranges
1	Alanine	366.25	103-742	8	Methionine	32.47	5-41
2	Arginine	3.95	1-41	9	Ornithine	45.54	10-263
3	Aspartic acid	27.73	10-345	10	Phenylalanine	103.57	10-102
4	Citrulline	10.08	5-43	11	Proline	224.31	87-441
5	Glutamic acid	597.97	150-708	12	Tyrosine	57.71	15-259
6	Glycine	464.26	0-1142	13	Valine	98.33	52-322
7	Leucine	168.13	27-324				

2. Amino acids molar ratios:

S.No	Ratios	Values	Ranges	S.No	Ratios	Values	Ranges
1	Met / Leu	0.19	<0.42	4	Leu / Ala	0.46	0.12-1.00
2	Met / Phe	0.31	<0.70	5	Leu / Tyr	2.91	0.50-3.50
3	Phe / Tyr (PKU)	1.79	<2.00	6			

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)	23.94	5-125	8	Methylmalonylcarnitine (C4DC)	0.40	0.10-1.25
2	Total Carnitines	77.94	10-184	9	Isovalerylcarnitine (C5)	0.39	0.01-1.00
3	Acetylcarnitine (C2)	38.25	1.4-80	10	3-methylcrotonylcarnitine (C5:1)	0.09	0.01-0.90
4	Propionylcarnitine (C3)	3.94	0.18-6.3	11	Glutarylcarnitine (C5DC)	0.02	0.01-2.99
5	Malonylcarnitine (C3DC)	0.07	0.01-0.45	12	3-OH- Isovalerylcarnitine (C5OH)	0.10	0.01-0.90
6	Butyrylcarnitine (C4)	0.93	0.08-1.70	13	Hexanoylcarnitine (C6)	0.32	0.01-0.95
7	3-OH-Butyrylcarnitine (C4OH)	0.32	0.01-1.29	14	Methylglutarylcarnitine (C6DC)	0.07	0.01-0.23

S.No	Metabolite	Conc. (uM)	Reference Ranges	S.No	Metabolite	Con. (uM)	Reference Ranges
15	Octanoylcarnitine (C8)	0.06	0.01-0.60	26	Palmitoylcarnitine (C16)	4.55	0.34-10.35
16	Octenoylcarnitine (C8:1)	0.11	0.01-0.70	27	Hexadecenoylcanitine (C16:1)	0.21	0.01-1.4
17	Decanoylcarnitine (C10)	0.14	0.02-0.65	28	3-Hydroxypalmitoleyl carnitine (C16:1OH)	0.02	0.01-0.10
18	Decenoylcarnitine (C10:1)	0.07	0.01-0.45	29	Hexadecenoylcanitine (C16OH)	0.01	0.01-0.10
19	Decadienoyl carnitine (C10:2)	0.01	0.01-0.22	30	Stearoylcarnitine (C18)	1.04	0.21-2.03
20	Dodecanoyl carnitine (C12)	0.07	0.02-0.6	31	Octadecadienoyl carnitine (C18:2)	0.05	0.10-0.73
21	Dodecenoyl carnitine (C12:1)	0.14	0.01-0.5	32	Octadecenoylcarnitine (C18:1)	2.26	0.5-7.0
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)	0.11	0.01-0.80	34	3-OH-Octadecenoylcarnitine (C18:1OH)	0.00	0.01-0.10
24	Tetradecadienoyl carnitine(C14:2)	0.01	0.00-0.20	35	3-OH-Stearoylcarnitine (C18OH)	0.010	0.01-0.10
25	3-OH-Tetradecenoyl carnitine (C14OH)	0.010	0.00-0.20				

4. Acylcarnitine molar ratios:

S.No	Ratios	Values	Ranges	S.No	Ratios	Values	Ranges
1	C4 / C3	0.24	<1.18	6	C0 / (C16 + C18)	4.29	<70
2	C3 / C0	0.16	<0.27	7	C5 / C2	0.010	<0.16
3	C3 / C2	0.10	<0.45	8	C5 / C3	0.10	<0.29
4	C8 / C10	0.42	< 1.50	9	C5DC / C3	0.00	<0.27
5	C8 / C2	0.002	<0.03	10	C5DC / C16	0.00	<0.68

Annexure 2

Patient Name: DNANB001A001J696

Results Biochemical Parameters:

Biochemical Parameters		
Assay	**Result**	**Reference Ranges**
Thyroid Stimulating Hormone (TSH) (Congenital Hypothyroidism (CH)	2.40	< 15 ulU/mL
17-hydroxyprogestrone (17-OHP) (Congenital Adrenal Hyperplasia (CAH)	19.30	<30 nmol/L (BW >2250g) <50 nmol/L (BW<2250g)
G6PD enzyme activity (G6PD Deficiency)	3.30	> 1.5 U/gHb
Total Galactose (TGAL) (Galactosemia (GAL)	5.70	< 15 mg/dL
Immunoreactive trypsinogen (IRT) (Cystic Fibrosis -CF)	55.20	< 90 µg/L
Biotinidase (BIOT) (Biotinidase)	64.10	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			
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) Mutiple CoA Carboxylase Deficiency
17	(5-OXO) 5-Oxoprolinurial	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 GAII
		29	(HMG) 3-Hydroxy-3-methylglutaryl-CoA Lyase Deficiency
		30	(IBG) Isobutyrl-CoA Dehydrogenase Deficiency
		31	(MCT)Medium Chain Triglyceride Oil Administration
		32	Methylmalonic Acidemia (Cobalamin disorders) Cbl A, B