Test Name	Result	Unit	Biological Reference Interval
THYROID STIMULATING HORMONE (TSH) CONGENITAL HYPOTHYROIDISM Method : Enzyme Linked Immunoassay	2.20	uIU/ml	Normal < 10 Borderline 10 - 20 Presumptive Positive > 20
17 – α HYDROXY PROGESTERONE CONGENITAL ADRENAL HYPERPLASIA Method : Enzyme Linked Immunoassay	10.46	nmol/L	Normal< 37.5 Bordline 37.5 - 90 Presumptive Positive> 90
GLUCOSE 6 PHOSPHATE DEHYDROGENASE G6PD DEFICIENCY Method : Enzymatic, Kinetic	5.48	U/gHb	Deficient ≤ 2 Borderline 2.1 - 4.9 Normal ≥ 5
TOTAL GALACTOSE / GALACTOSEMIA Method : Enzymatic, End Point	9.68	mg/dL	Normal < 10 Borderline 10 - 20 Presumptive Positive> 20
BIOTINIDASE / BIOTIDINASE DEFICIENCY Method : Enzymatic, End Point	4.00	nmol/min/ml	Normal > 2.8 Presumptive Positive ≤ 2.8
IMMUNOREACTIVE TRYPSINOGEN (IRT) CYSTIC FIBROSIS Method : Enzyme Immunoassay	64.02	ng/mL	Normal < 68 Presumptive Positive ≥ 68
Clinical History	Fever, Cough, Vomitting from 8 days fever still there		
Analytical Interpretation	The levels of all analytes are within normal limits. Hence, The levels of all analytes are within normal limits. Hence, Screen Negative for tested disorders.		

Scan to Validate

METABOLIC SCREENING, DRY BLOOD SPOT

DISORDERS	ANALYTES	RESULTS	BIOLOGICAL REFERENCE INTERVAL	UNITS
METABOLIC PROFILE				
	Amino Acids Profile	**All related analytes within acceptable limits**		
	Acylcarnitine Profile	**See Interpretation below**		

Method : Tandem Mass Spectrometry

Clinical History

Fever, Cough, Vomitting from 8 days and Fever

Analytical Interpretation

The level of Malonylcarnitine+3-hydroxy butyrylcarnitine(C3DC+C4OH) is marginally higher than normal range.

Though C3DC+C4OH levels are elevated in Malonic Acidemia caused by Malonyl-CoA decarboxylase deficiency, marginally elevated levels may not be clinically significant.

Kindly correlate the report with clinical and therapeutic history of the patient.

Test Information

In metabolic disorders, deficiency of specific enzymes causes disruption of metabolic pathways leading to accumulation of abnormal metabolites in blood. Altered levels of these metabolites are suggestive of specific congenital metabolic disorder. The TMS analysis of Dry Blood Spot Specimen allows simultaneous screening of Amino Acid Disorders, Fatty Acid Oxidation Disorders, Organic Acidurias.

Disclaimer

Metabolic Screening process assists in the detection of metabolic disorders. However, due to various factors such as age, health status and treatment at the time of specimen collection, genetic variability, prematurity, quality of specimen etc, the screen may not detect the presence or absence of potentially detectable disorder. While a positive screening result identifies newborns at an increased risk, a negative screening result does not rule out possibility of the disorder. The positive screening test result should be confirmed with confirmatory tests. It is recommended for repeat screening for infants when specimens are collected before 48 hours of age.

- HOSPITAL : -

Tandem Mass Spectrometry

AMINO ACIDS PROFILE

Analyte	Result µM/L	BRI µM/L	Analyte	Result µM/L	BRI µM/L
Alanine		0 - 480	Arginine	29.4	0 - 130
Citrulline	8	10 - 40	Glycine	38.8	0 - 623

Leucine+Isoleucine	159	0 - 200	Methionine	2.78	0 - 44
Ornithine	2.98	0 - 393	Phenylalanine	80.2	0 - 130
Tyrosine	48.2	0 - 181	Valine	118	0 - 182

ACYLCARINITINE PROFILE

Analyte	Result µM/L	BRI µM/L	Analyte	Result µM/L	BRI µM/L
Free Carnitine (C0)	15.3	9 - 57	Acetylcarnitine (C2)	12.8	3 - 45
Propionylcarnitine (C3)	0.84	0 - 5.81	Malonylcarnitine+3-hydroxy butyrylcarnitine(C3DC+C4OH)	0.57	0 - 0.55
Butyrylcarnitine (C4)	0.23	0 - 1.7	Methylmalonylcarnitine+3-hydroxy isovalerylcarnitine (C4DC+C5OH)	0.18	0 - 0.4
Isovalerylcarnitine (C5)	0.19	0 - 0.65	3-methylcrotonylcarnitine (C5:1)	0.01	0 - 0.2
Glutaryl Carnitine (C5DC)	0.13	0 - 0.41	Hexanoylcarnitine (C6)	0.08	0 - 0.23
Octanoyl Carnitine (C8)	0.1	0 - 0.39	Decanoylcarnitine (C10)	0.21	0 - 0.5
Decenoylcarnitine (C10:1)	0.18	0 - 0.5	Dodecanoylcarnitine (C12)	0.14	0 - 0.42
Tetradecanoylcarnitine (C14)	0.1	0 - 0.41	Tetradecenoylcarnitine (C14:1)	0.09	0 - 0.39
Tetradodecenoylcarnitine (C14:2)	0.03	0 - 0.06	Palmitoylcarnitine (C16)	0.71	0.76 - 6.46
3-Hydroxypalmitoylcarnitine (C16-OH)	0.01	0 - 0.21	Stearoylcarnitine (C18)	0.39	0.31 - 1.8
Oleylcarnitine (C18:1)	0.54	0.36 - 2.4	3-Hydroxylinoleoylcarnitine (C18:1-OH)	0.01	0 - 0.15
3Hydroxylinoleoylcarnitine (C18-OH)	0.01	0 - 0.17			

LIST OF DISORDERS SCREENED

Amino Acid Metabolism Disorders		25	Medium chain Ketoacyl CoA thiolase deficiency
1	Phenylketonuria (PKU)	26	Very long chain Acyl-CoA dehydrogenase deficiency
2	Benign hyperphenylalaninemia	27	Long chain L-3-hydroxy acyl-CoA dehydrogenase deficiency
3	Defects of Biopterin cofactor biosynthesis	28	Trifunctional protein deficiency
4	Defects of biopterin cofactor regeneration	29	Multiple acyl-CoA dehydrogenase deficiency/Glutaric acidemia type II
5	Maple syrup urine disease	30	Medium chain L-3 hydroxy acyl coA dehydrogenase deficiency
6	Classical Citrullinemia	**Organic Acid Metabolism Disorders**	
7	Citrullinemia type-2 (citrin deficiency)	31	Glutaric acidemia type I
8	Arginosuccinate synthase (ASS) deficiency	32	Isovaleric acidemia
9	Argininosuccinic aciduria	33	2-Methylbutyryl-CoA-dehydrogenase deficiency
10	Hyperornithinemia, Hyperammoninemia Hyperhomocitrullinemia (HHH) syndrome	34	Propionic acidemia
11	Arginase deficiency/Argininemia	35	Methylmalonyl-CoA mutase deficiency
12	Neonatal Tyrosinemia	36	Methylmalonic acidemia
13	Tyrosinemia I	37	Methylmalonic acidemia (mutase)
14	Tyrosinemia type II	38	3-Methylcrotonyl-CoA carboxylase deficiency
15	Tyrosinemia III	39	Malonic aciduria
16	Homocystinuria	40	3-Hydroxy 3-methyl glutaric acidemia
17	Hypermethioninemia	41	β-Ketothiolase deficiency
Fatty Acid Metabolism Disorders		42	Multiple CoA carboxylase deficiency
18	Carnitine transporter deficiency (Carnitine uptake deficiency)	43	Isobutaryl CoA dehydrogenase deficiency
19	Carnitine/acylcarnitine Translocase deficiency	44	Malonyl-CoA decarboxylase deficiency
20	Carnitine palmitoyl transferase deficiency type I	45	Holocarboxylase deficiency
21	Carnitine palmitoyl transferase deficiency type II	46	3-Methyl glutaconyl CoA hydratase deficiency
22	Short chain Acyl CoA dehydrogenase deficiency		
23	Short chain Hydroxy Acyl CoA dehydrogenase deficiency		
24	Medium chain Acyl CoA dehydrogenase deficiency		

--- End of Report ---