BABY'S NAME :	**DHYANS SINGH**	**SAMPLE ID :**	B3066087
BABY'S DATE OF BIRTH :	-	**AGE :**	1.4 Years **GENDER:** Male
SAMPLE COLLECTED :		**SAMPLE RECEIVED :**	
SAMPLE REPORTED :		**HOSPITAL :**	-
		REFERRED BY :	-

METABOLIC SCREENING, URINE

DISORDERS	ANALYTES	RESULTS	BIOLOGICAL REFERENCE INTERVAL	UNITS
METABOLIC PROFILE				
Amino Acidopathies			All related analytes within acceptable limits	
Fatty Acid Metabolism Disorders			All related analytes within acceptable limits	
Organic Acidurias				
	4- Hydroxy Phenyl Lactic Acid	**3.2**	**0 - 2**	**µmol/mmol of Creatinine**
	Lactic Acid	**147.5**	**5 - 118**	**µmol/mmol of Creatinine**
			All other related analytes within acceptable limits	
Carbohydrate / TCA Cycle / Mitochondrial Dysfunction				
	Succinic Acid	**166.4**	**4.9 - 81.3**	**µmol/mmol of Creatinine**
			All other related analytes within acceptable limits	
Purine / Pyrimidine Metabolism Disorders			All related analytes within acceptable limits	
Peroxisomal Disorders			All related analytes within acceptable limits	
Neurotransmitter Metabolism Disorders			All related analytes within acceptable limits	

Method : Gas Chromatography Mass Spectrometry

Clinical History

Nil

Analytical Interpretation

Urine metabolic profile revealed elevated excretions of 4- Hydroxy Phenyl Lactic Acid (4HPL). Lactic Acid and Succinic Acid.

Urine Lactic acid levels are elevated by a number of nonspecific factors such as bacterial overgrowth of the GI tract, shock, poor perfusion, B-vitamin deficiency, mitochondrial dysfunction or damage, and anemia. Mild to moderate lactic acidosis may result in various conditions such as apnea, septicemia, seizures, respiratory or cardiac insufficiency, systemic inflammatory response syndrome, severe physical trauma or severe depletion of body fluids and TPN administration.

4 HPL is a tyrosine metabolite and elevated levels of 4HPL are commonly associated with disorders of tyrosine metabolism, increased tyrosine intake, bacterial gut metabolism, short bowel syndrome, sepsis, lactic acidosis or liver disease or damage. A slightly elevated level of 4HPL in a newborn may be transient and not necessarily indicate a serious condition, especially if the baby is feeding well and developing normally.

BABY'S NAME :	**DHYANS SINGH**	**SAMPLE ID :**	B3066087	
BABY'S DATE OF BIRTH :	-	**AGE :**	1.4 Years	**GENDER:** Male
SAMPLE COLLECTED :		**SAMPLE RECEIVED :**		
SAMPLE REPORTED :		**HOSPITAL :**	-	
		REFERRED BY :	-	

Elevated level of Succinic acid may indicate a relative deficiency of riboflavin and/or coenzyme Q10, bacterial degradation of unabsorbed glutamine supplement or bacterial infections. Elevated levels may also indicate mitochondrial dysfunction. Succinic acid is a metabolite of gamma aminobutyric acid (GABA), hence supplementation with GABA, disrupted GABA metabolism or increased GABA levels may also increase succinic acid. Very high levels of succinic acid occur in rare genetic disorder - Succinate Dehydrogenase deficiency. Elevated succinic acid in urine is also linked to autism.

Please correlate the report with other clinical and therapeutic history as well as other laboratory diagnostic findings.

Test Information

This GC/MS analysis of urine allows simultaneous detection and quantitation of 135 metabolic disorders.

In metabolic disorders, deficiency of specific enzymes causes disruption of metabolic pathways leading to accumulation of abnormal metabolites in body. In order to maintain physiological homeostasis, body rapidly excretes these excess abnormal metabolites in urine. Hence these metabolites are detected in urine much earlier than the actual rise in their blood level. GC/MS method detects these abnormal metabolites in urine thus making presymptomatic detection of a metabolic disorder possible.

These urinary metabolites act as specific and precise biomarkers for identification of congenital metabolic disorder as well as mild nutritional deficiencies.

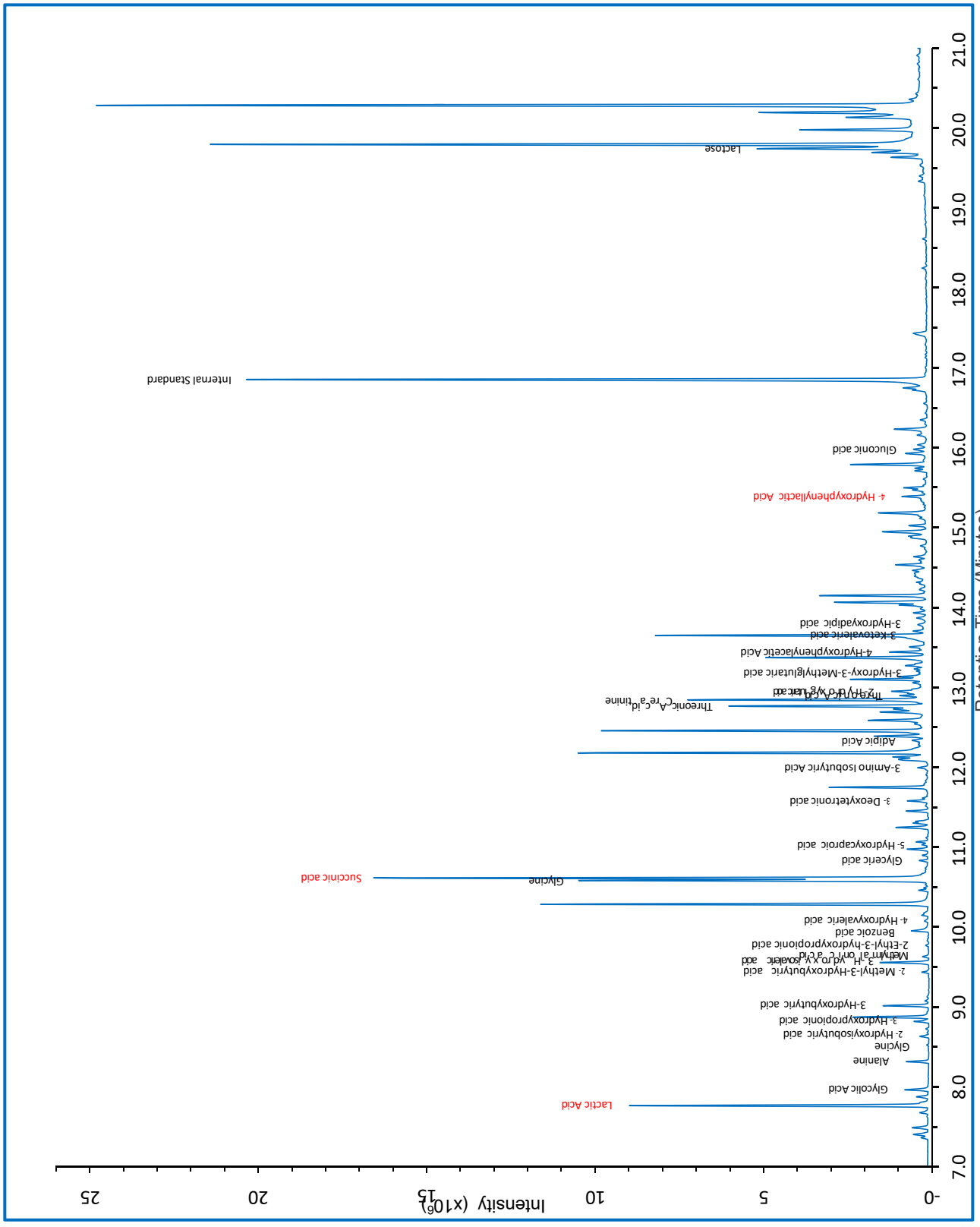
Disclaimer

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

Dr. Milind Chincholkar
M.D. (Pathology)

Dr. Jyoti Sawant
Ph.D. (Med. Biochemistry)

BABY'S NAME :	**DHYANS SINGH**	**SAMPLE ID :**	**B3066087**
BABY'S DATE OF BIRTH :	-	**AGE :** 1.4 Years	**GENDER:** Male
SAMPLE COLLECTED :		**SAMPLE RECEIVED :**	
SAMPLE REPORTED :		**HOSPITAL :** -	
		REFERRED BY : -	



BABY'S NAME :	**DHYANS SINGH**		**SAMPLE ID :**	**B3066087**	
BABY'S DATE OF BIRTH :	-		**AGE :**	1.4 Years	**GENDER:** Male
SAMPLE COLLECTED :			**SAMPLE RECEIVED :**		
SAMPLE REPORTED :			**HOSPITAL :**	-	
			REFERRED BY :	-	

DETAILED REPORT OF ANALYTES TESTED FOR METABOLIC DISORDERS

DISORDERS OF AMINO ACID & FATTY ACID METABOLISM AND ORGANIC ACIDURIAS

Analyte	Result	BRI µmol/mmol Creatinine
2- Amino Adipic Acid	0	0 - 4.3
2- Ethyl 3-Hydroxy Propionic Acid	0.74	0 - 19.9
2- Hydroxy Butyric Acid	0	0 - 2
2- Hydroxy Isobutyric Acid	3.13	2.9 - 19.5
2- Hydroxy Isovaleric Acid	0	0 - 11.9
2- Hydroxy Sebasic Acid	0	0 - 0.67
2- Keto Isocaproic Acid	0	0 - 7
2- Methyl Acetoacetic Acid	0	0 - 2
2- Oxoadipic Acid	0	0 - 5
3- Amino Isobutyric Acid	1.81	1.4 - 6.2
3- Hydroxy Adipic Acid	1.1	0 - 5
3- Hydroxy Dodecanedioic Acid	1.32	0 - 10
3- Hydroxy Hexanedioic Acid	0	0 - 11
3- Hydroxy Isovaleric Acid	6.2	3.1 - 23.1
3-(3- Hydroxy Phenyl) 3- Hydroxy Propionic Acid	0	0 - 0.5
3- Hydroxy Phenylhydracrylic Acid	1.83	0 - 31
3- Hydroxy Sebasic Acid	0.87	0 - 9.1
3- Hydroxy Valeric Acid	0	0 - 1.4
3- Methyl 2-Hydroxy Valeric Acid	0	0 - 5
3- Methyl Glutaconic Acid	0	0 - 19
4- Deoxy Tetronic Acid	1.76	0 - 8
4- Hydroxy Benzoic Acid	0	0 - 16
4- Hydroxy Cyclohexylacetic acid	0	0 - 3
4- Hydroxy Phenyl Lactic Acid	3.2	0 - 2

Analyte	Result	BRI µmol/mmol Creatinine
2- Deoxy Tetronic Acid	53	4.8 - 22.4
2- Hydroxy Adipic Acid	0	0 - 2.8
2- Hydroxy Glutaric Acid	2.8	0 - 8
2- Hydroxy Isocaproic Acid	0	0 - 5
2- Hydroxy Phenyl Acetic Acid	0	0 - 20
2- Keto Glutaric Acid	5.15	0 - 52
2- Methyl 3-Hydroxy Butyric Acid	0	0 - 7
2- Methyl Glutaric Acid	0	0 - 2.6
2,5 Furandicarboxylic Acid	1.49	0 - 11
3- Deoxy Tetronic Acid	2.53	1.9 - 15.2
3- Hydroxy Butyric Acid	5.1	0 - 11.1
3- Hydroxy Glutaric Acid	0	0 - 4.6
3- Hydroxy Isobutyric Acid	0	0 - 137
3- Hydroxy Methyl Glutaric Acid	2.1	0 - 12
3- Hydroxy Phenyl Acetic Acid	0	0 - 11
3- Hydroxy Propionic Acid	1.59	0 - 22
3- Hydroxy Suberic Acid	0.46	0 - 5.6
3- Methoxy Benzene Propionic Acid	0	0 - 11.9
3- Methyl Crotonyl Glycine	0	0 - 10
3- Methyl Glutaric Acid	0	0 - 3
4- Hydroxy 3- Methyl Benzoic Acid	0	0 - 28.6
4- Hydroxy Butyric Acid	1.46	0 - 10
4- Hydroxy Phenyl Acetic Acid	4.11	0 - 19
4- Hydroxy Phenyl Pyruvic Acid	0	0 - 0.4

Low Normal Borderline High

BRI - Biological Reference Interval

BABY'S NAME :	DHYANS SINGH	**SAMPLE ID :**	B3066087	
BABY'S DATE OF BIRTH :	-	**AGE :**	1.4 Years	**GENDER:** Male
SAMPLE COLLECTED :		**SAMPLE RECEIVED :**		
SAMPLE REPORTED :		**HOSPITAL :**	-	
		REFERRED BY :	-	

DETAILED REPORT OF ANALYTES TESTED FOR METABOLIC DISORDERS

DISORDERS OF AMINO ACID & FATTY ACID METABOLISM AND ORGANIC ACIDURIAS

Analyte	Result	BRI µmol/mmol Creatinine
4- Hydroxy Proline	10.7	0 - 135
5-Hydroxy Indole Acetic Acid	0	0 - 11.5
7- Hydroxy Octanoic Acid	0	0 - 2
8- Hydroxy Octanoyl Glycine	0	0 - 0.2
Acetyl Glycine	0	0 - 1
Alanine	3.34	3 - 17.9
Asparagine	0.52	0 - 1.6
Azelaic Acid	0	0 - 9
Butyryl Glycine	0	0 - 2
Citramalic Acid	1.43	0 - 11.2
Cystathionine	0	0 - 1
Decanoic Acid	0	0 - 1
Dimethylglycine	0	0 - 1
Dodecanoic Acid	0	0 - 0.05
Eicosadecanoic Acid	0	0 - 1
Ethanolamine	10.3	0 - 165
Ethyl Malonic Acid	0	0 - 14.6
Furoic Acid	2.28	0 - 28
Glutamic Acid	8.9	0 - 21
Glutaric Acid	0	0 - 5.3
Glycerol	5.7	0 - 25
Glycolic Acid	4.13	0 - 198
Hawkinsin	0	0 - 0.1
Hexanoic Acid	0	0 - 2.3
Hippuric Acid	0	0 - 145

Analyte	Result	BRI µmol/mmol Creatinine
5- Hydroxy Caproic Acid	0.68	0 - 1.9
5- Hydroxy Methyl Furanoic Acid	1.96	0 - 44
7- Hydroxy Octanoyl Glycine	0	0 - 0.2
Acetoacetic Acid	0	0 - 1.5
Adipic Acid	1.75	0 - 35
Argininosuccinic Acid	0	0 - 0.1
Aspartic Acid	0.79	0 - 2.46
Beta Alanine	0.19	0 - 0.96
Cis-Aconitic Acid	1.8	0 - 17
Citric Acid	79.5	40 - 520
Cysteine	5.14	0 - 45.4
Decanoyl Glycine	0	0 - 1
Docosanoic acid	0	0 - 1
Dodecendioic Acid	0	0 - 0.05
Eicosadecenoic Acid	0	0 - 1
Ethyl Hydracrylic Acid	0.65	0 - 13.3
Formiminoglutamic Acid	0	0 - 1.61
Glutaconic Acid	0	0 - 0
Glutamine	78	69 - 127
Glyceric Acid	1.87	0 - 37.3
Glycine	401.2	184 - 508
Glyoxylic Acid	0	0 - 15.9
Hexadecanoic (Palmitic) Acid	1.12	0.1 - 8.2
Hexanoyl Glycine	0	0 - 2.9
Histidine	50.5	0 - 339

Low Normal Borderline High

BRI - Biological Reference Interval

BABY'S NAME :	DHYANS SINGH		**SAMPLE ID :**	**B3066087**	
BABY'S DATE OF BIRTH :	-		**AGE :**	1.4 Years	**GENDER:** Male
SAMPLE COLLECTED :			**SAMPLE RECEIVED :**		
SAMPLE REPORTED :			**HOSPITAL :**	-	
			REFERRED BY :	-	

DETAILED REPORT OF ANALYTES TESTED FOR METABOLIC DISORDERS

DISORDERS OF AMINO ACID & FATTY ACID METABOLISM AND ORGANIC ACIDURIAS

Analyte	Result	BRI µmol/mmol Creatinine
Homocysteine	0	0 - 4.19
Homoserine	0	0 - 0.66
Indole Acetic acid	0.81	0 - 3.4
Isocitric Acid	12.6	0 - 141
Isovaleryl Glycine	0	0 - 4.3
Lactic Acid	147.5	5 - 118
Linoleinic Acid	0	0 - 1.27
Malonic Acid	0	0 - 9.7
Methionine	0.65	0 - 2.4
Methyl Citric Acid	0	0 - 2.7
Methyl Malic Acid	0	0 - 0.2
Methyl Succinic Acid	0.85	0 - 6
Mevalonolactone	0	0 - 0.001
N- Acetyl Aspartic Acid	8.23	6 - 40.8
N-Acetyl Proline	0	0 - 2
Nonadecanoic acid	0	0 - 1
Octanoic Acid	0	0 - 4
Oleic Acid	0	0 - 0.4
Orotic Acid	0	0 - 4.3
p-Cresol	0	0 - 11
Phenol	4.93	0 - 53
Phenylalanine	16.2	0 - 71.6
Phenyl Pyruvic Acid	0	0 - 2
Pipecolic Acid	0	0 - 0.24
Propionic Acid	0.65	0 - 11.2

Analyte	Result	BRI µmol/mmol Creatinine
Homogentisic Acid	0	0 - 0
Hydroxylysine	0	0 - 0.97
Isobutyryl Glycine	0	0 - 2.9
Isoleucine	5.27	0 - 16.6
Kynurenic Acid	0	0 - 16.508
Leucine	8.91	2 - 24
Lysine	18.5	0 - 81.2
Mandelic Acid	0	0 - 64
Methyl Adipic Acid	0	0 - 3.554
Methyl Fumaric Acid	0	0 - 0.2
Methyl Malonic Acid	1.6	0 - 6
Mevalonic Acid	0	0 - 0.3
N-Acetyl Alanine	0	0 - 1
N-Acetyl Glycine	0	0 - 3.4
N- Acetyl Tyrosine	0	0 - 10
Nonanoic acid	0	0 - 1
Octenedioic Acid	0	0 - 2.8
Ornithine	2.19	0 - 11.8
Oxalic Acid	0	0 - 19
Pentadecanoic acid	0	0 - 1
Phenyl Acetyl Glycine	13.7	0 - 110
Phenyl Lactic Acid	0	0 - 4
Pimelic Acid	2.43	0 - 14
Proline	4.57	0 - 12.7
Propionyl Glycine	0	0 - 0.1

Low Normal Borderline High

BRI - Biological Reference Interval

BABY'S NAME :	DHYANS SINGH		**SAMPLE ID :**	**B3066087**	
BABY'S DATE OF BIRTH :	-		**AGE :**	1.4 Years	**GENDER:** Male
SAMPLE COLLECTED :			**SAMPLE RECEIVED :**		
SAMPLE REPORTED :			**HOSPITAL :**	-	
			REFERRED BY :	-	

DETAILED REPORT OF ANALYTES TESTED FOR METABOLIC DISORDERS

DISORDERS OF AMINO ACID & FATTY ACID METABOLISM AND ORGANIC ACIDURIAS

Analyte	Result	BRI µmol/mmol Creatinine
Pyroglutamic Acid	5.8	0 - 67
Sarcosine	0.39	0 - 2
Serine	29.4	0 - 177.3
Suberic Acid	0	0 - 10.1
Succinic Acid	166.4	4.9 - 81.3
Tetracosanoic acid	0	0 - 1
Threonine	10.9	0 - 58.2
Tridecanoic Acid	0	0 - 1
Tyrosine	13.9	6.2 - 59
Valine	7.2	3.3 - 19.8

Analyte	Result	BRI µmol/mmol Creatinine
Pyruvic Acid	9.52	0 - 40.5
Sebasic Acid	0	0 - 1.4
Stearic Acid	0	0 - 8
Suberyl Glycine	0	0 - 1
Succinyl Acetone	0	0 - 0.001
Tetradecenoic Acid	0	0 - 1
Tiglyl Glycine	0	0 - 6.7
Tryptophan	12.5	2 - 46
Uracil	0.69	0 - 9.7
Xanthurenic Acid	0	0 - 4

DISORDERS OF CARBOHYDRATE METABOLISM

Analyte	Result	BRI µmol/mmol Creatinine
Arabinose	0	0 - 36
Fructose	10.4	0 - 35
Galactonic Acid	10.8	0 - 127.7
Gluconic Acid	7.5	0 - 122
Glucuronic Acid	0	0 - 75.7
Lactic Acid	147.5	5 - 118
Mannose	0	0 - 3.3
Sucrose	14.4	0 - 91
Xylose	0	0 - 67.1

Analyte	Result	BRI µmol/mmol Creatinine
Erythronic Acid	4.2	3.9 - 22.2
Galactitol	0	0 - 52
Galactose	9.7	0 - 49
Glucose	25.2	0 - 80
Glyceraldehyde 3- Phosphate	0	0 - 1
Lactose	23.1	0 - 52.4
Ribose	0	0 - 5
Threonic Acid	11.4	1.7 - 43.7

Low Normal Borderline High

BRI - Biological Reference Interval

BABY'S NAME :	DHYANS SINGH		**SAMPLE ID :**	**B3066087**	
BABY'S DATE OF BIRTH :	-		**AGE :**	1.4 Years	**GENDER:** Male
SAMPLE COLLECTED :			**SAMPLE RECEIVED :**		
SAMPLE REPORTED :			**HOSPITAL :**	-	
			REFERRED BY :	-	

DETAILED REPORT OF ANALYTES TESTED FOR METABOLIC DISORDERS

DISORDERS OF TCA CYCLE/MITOCHONDRIAL DYSFUNCTION

Analyte	Result	BRI µmol/mmol Creatinine
2- Keto Glutaric Acid	5.15	0 - 52
Cis-Aconitic Acid	1.8	0 - 17
Fumaric Acid	0.92	0 - 9.9
Lactic Acid	147.5	5 - 118
Methyl Fumaric Acid	0	0 - 0.2
Pyruvic Acid	9.52	0 - 40.5

Analyte	Result	BRI µmol/mmol Creatinine
Alanine	3.34	3 - 17.9
Citric Acid	79.5	40 - 520
Isocitric Acid	12.6	0 - 141
Malic Acid	1.2	0 - 16.2
Methyl Malic Acid	0	0 - 0.2
Succinic Acid	166.4	4.9 - 81.3

DISORDERS OF PURINE/PYRIMIDINE METABOLISM

Analyte	Result	BRI µmol/mmol Creatinine
2,8 Dihydroxyadenine	0	0 - 0.001
Adenosine	1.19	0 - 11
Deoxyadenosine	0	0 - 27.3
Dihydro Uracil	0	0 - 20
Hypoxanthine	6.6	0 - 60
Orotic Acid	0	0 - 4.3
Thymine	0	0 - 1.7
Uric Acid	26.2	0 - 462
Xanthine	5.8	0 - 62

Analyte	Result	BRI µmol/mmol Creatinine
5- Hydroxy Methyl Uracil	0	0 - 0.0121
Beta-Ureidopropionic Acid	1.11	0 - 4.8
Dihydro Thymine	0	0 - 3
Guanosine	0	0 - 11
Inosine	0	0 - 3.1
Pseudouridine	4.6	0 - 47.3
Uracil	0.69	0 - 9.7
Uridine	2.31	0 - 9

PEROXISOMAL DISORDERS

Analyte	Result	BRI µmol/mmol Creatinine
2,6 Dimethyloctanedioic Acid	0	0 - 0.001
Adipic Acid	1.75	0 - 35
Oxalic Acid	0	0 - 19
Suberic Acid	0	0 - 10.1

Analyte	Result	BRI µmol/mmol Creatinine
3-Methyl Adipic Acid	0	0 - 3.554
Malic Acid	1.2	0 - 16.2
Sebasic Acid	0	0 - 1.4

Low Normal Borderline High

BRI - Biological Reference Interval

BABY'S NAME :	**DHYANS SINGH**		**SAMPLE ID :**	**B3066087**	
BABY'S DATE OF BIRTH :	-		**AGE :**	1.4 Years	**GENDER:** Male
SAMPLE COLLECTED :			**SAMPLE RECEIVED :**		
SAMPLE REPORTED :			**HOSPITAL :**	-	
			REFERRED BY :	-	

DETAILED REPORT OF ANALYTES TESTED FOR METABOLIC DISORDERS

DISORDERS OF NEUROTRANSMITTER METABOLISM

Analyte	Result	BRI µmol/mmol Creatinine
3- Hydroxy Mandelic Acid	0	0 - 0.1
Adipic Acid	1.75	0 - 35
Quinolinic Acid	5.6	0-26.2
Vanillyl Mandelic Acid	3.94	0 - 16.1

Analyte	Result	BRI µmol/mmol Creatinine
4- Hydroxy Mandelic Acid	1.59	1.3 - 10
Homovanillic Acid	4.7	0 - 13.8
Sebasic Acid	0	0 - 1.4

Low Normal Borderline High

BRI - Biological Reference Interval

LIST OF DISORDERS SCREENED

Amino Acid Metabolism Disorders		48	Pyruvate Carboxylase Deficiency	94	Malonic Acidemia (MAL)
1	2- Ketoadipic Aciduria	49	Pyruvate Dehydrogenase (E1) Deficiency	95	Methylmalonic Acidemia (MMA) - Cbl C, D
2	2-Oxoglutaric Aciduria	50	Renal Fancony Syndrome	96	Methylmalonic Aciduria, cbl A and cbl B forms (MMA, Cbl A, B)
3	3- Hydroxyisobutyryl-CoA-Deacylase Deficiency	51	Saccharopinuria	97	Methylmalonic Semialdehyde Dehydrogenase Deficiency
4	5-Oxoprolinuria	52	Serum Carnosinase Deficiency	98	Methylmalonyl- CoA Mutase Deficiency (MUT)
5	Alkaptonuria	53	Transient Tyrosinemia in Infancy	99	Mevalonic Acidemia
6	Aminoacylase 1 Deficiency	54	Tryptophanuria with Dwarfism	100	Multiple Carboxylase Deficiency
7	Argininemia	55	Tyrosinemia caused by a Liver Dysfunction	101	Propionic Acidemia (PPA)
8	Argininosuccinic Aciduria	56	Tyrosinemia Type I	**TCA Cycle & Mitochondrial Dysfunction**	
9	Benign Hyperphenylalaninemia	57	Tyrosinemia Type II	102	Cytochrome C Oxidase deficiency
10	Biotinidase Deficiency	58	Tyrosinemia Type III	103	Cytochrome aa3-b deficiency
11	Carbamoylphosphate Synthetase 1- Deficiency	59	Valinemia	104	Leigh's Syndrome
12	Citrullinemia	60	Xanthurenic Aciduria	105	Mitochondrial Encephalopathy
13	Citrullinemia type II (CIT II)	**Fatty Acid Oxidation Disorders**		106	Pyruvate Dehydrogenase Phosphatase Deficiency
14	Cystathioninuria	61	2, 4 - Dienoy CoA Reductase Deficiency	**Carbohydrate Metabolism Disorders**	
15	Cystinuria	62	Carnitine Transport Defect	107	D-Glyceric Aciduria
16	Defects of Biopterin Cofactor Biosynthesis (BIOPT BS)	63	Glutaric Aciduria Type II	108	Endogenous Sucrosuria
17	Defects Of Biopterin Cofactor Regeneration (BIOPT REG)	64	Long-Chain 3- Hydroxyacyl-CoA Dehydrogenase Deficiency (LCHAD)	109	Fructose-1,6-Diphosphatase Deficiency
18	Dicarboxylic Aminoaciduria	65	Medium/Short-Chain 3-Hydroxyacyl-CoA Dehydrogenase Deficiency (SCHAD)	110	Fructosuria
19	Dihydrolipoyl Dehydrogenase (E3) Deficiency	66	Medium-Chain Acyl- CoA Dehydrogenase Deficiency (MCAD)	111	Galactokinase Deficiency (GALK)
20	Dimethylglycinuria	67	Medium-Chain Ketoacyl- CoA Thiolase Deficiency (MCKAT)	112	Galactose Epimerase Deficiency (GALE)
21	Ethanolaminosis	68	Mitochondrial Trifunctional Protein Deficiency (MTPD)	113	Galactosemia
22	Familial Renal Iminoglycinuria	69	Short-Chain Acyl- CoA Dehydrogenase Deficiency (SCAD)	114	Heredetary Fructose Intolerance
23	Glycerol Kinase Deficiency	70	Very Long-Chain Acyl- CoA Dehydrogenase Deficiency (VLCAD)	115	Lactose Intolerance
24	Glycine Encephalopathy	**Organic Acidurias**		116	Pentosuria
25	GTP Cyclohydrolase (GTPCH) Deficiency	71	2-Aminoadipic Aciduria	117	Transaldolase Deficiency
26	Hartnup Disease	72	2-Hydroxyglutaric Aciduria	118	Transient Galactosemia
27	Hawkinsinuria	73	2-Methyl 3-Hydroxy Butyric Aciduria (2M3HBA)	**Purine / Pyrimidine Metabolism Disorders**	
28	Histidinuria - Renal Tubular Defect	74	2-Methylbutyryl-CoA Dehydrogenase Deficiency (2MBG)	119	Adenine Phosphoribosyl Transferase Deficiency
29	Homocystinuria	75	3-Aminoisobutyric Aciduria	120	Adenosine Deaminase Deficiency
30	Hydroxylysinuria	76	3-Hydroxy-3-Methylglutaric Aciduria (HMG CoA Lyase Deficiency)	121	Beta Ureidopropionase Deficiency
31	Hyperhydroxyprolinemia	77	3-Methylcrotonyl CoA Carboxylase Deficiency	122	Dihydropyrimidinase Deficiency
32	Hyperbasic Aminoaciduria	78	3-Methylglutaconic Aciduria	123	Hyperuric Acidemia
33	Hyperbeta-Alaninemia	79	4-Hydroxybutyric Aciduria	124	Lesch - Nyhan Syndrome
34	Hyperglycinuria	80	Barth Syndrome	125	Orotic Aciduria
35	Hyperleucine - Isoleucinemia	81	Beta- Ketothiolase Deficiency (BKT)	126	Partial Deficiency of Hypoxanthine- Adenine Phosphoribosyl Transferase
36	Hypermethioninemia	82	Canavan Disease	127	Thymine- Uraciluria
37	Hyperornithinemia- Hyper ammoninemia- Hyper homocitrullinemia (HHH) Syndrome	83	Ethylmalonic Aciduria	128	Xanthinuria
38	Hyperprolinemia type I	84	Formiminoglutamic Aciduria	**Peroxisomal Disorders**	
39	Hyperprolinemia type-II	85	Fumarate Hydratase Deficiency	129	Infantile Refsum Disease (IRD)
40	Hypersarcosinemia	86	GABA Transaminase Deficiency	130	Neonatal Adrenoleukodystrophy
41	Imidazole Aminoaciduria	87	Glutaric Aciduria Type I	131	Primary Hyperoxaluria
42	Iminoglycinuria	88	Glutaric Aciduria Type III	132	Zellweger Like Syndrome (ZLS)
43	Lysinuric Protein Intolerance	89	Glutathionuria	133	Zellweger Syndrome
44	Maple Syrup Urine Disease (MSUD)	90	Histidinemia	**Neurotransmitter Metabolism Disorders**	
45	N-Acetyl Glutamate Synthetase Deficiency	91	Hyperpipecolatemia	134	Neuroblastoma
46	Ornithine Transcarbamylase (OTC) Deficiency	92	Isobutyryl-CoA Dehydrogenase Deficiency	135	Pheochromocytoma
47	Phenylketonuria (PKU)	93	Isovaleric Acidemia		

--- End of Report ---

BABY'S NAME :	**DHYANS SINGH**		**AGE :**	1.4 Years	**GENDER:** Male
SAMPLE ID :	**B3066087**	**HOSPITAL :** -	**REFERRED BY :**	-	
COLLECTED ON :		**RECEIVED ON :**	**REPORTED ON :**		

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

Nil

Analytical Interpretation

The Newborn Screening test on Dried Blood Spot sample using Tandem MS for Amino Acids & Acylcarnitine profile for 46 metabolic conditions (list enclosed) showed that the levels of Free Carnitine (C0), Palmitoylcarnitine (C16), Stearoylcarnitine (C18), Oleylcarnitine (C18:1) are lower than normal range.

Lower levels of Palmitoylcarnitine (C16), Stearoylcarnitine (C18) and Oleylcarnitine (C18:1) are clinically not significant.
Low C0 levels may indicate Carnitine Uptake Defect (CUD). However, the secondary marker of this disorder - (C0+C2+C3+C16+C18+C18:1/citrulline) ratio is not elevated. Low C0 levels also occur in carnitine deficiency.

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.

This test has been performed at our referral Lab

Dr. Jyoti Sawant
Ph.D. (Med. Biochemistry)

Dr. Milind Chincholkar
M.D. (Pathology)

BABY'S NAME :	DHYANS SINGH		**AGE :**	1.4 Years	**GENDER:** Male
SAMPLE ID :	B3066087		**COLLECTED ON :**		
RECEIVED ON :			**REPORTED ON :**		
REFERRED BY :	-		**HOSPITAL :**	-	

Tandem Mass Spectrometry

AMINO ACIDS PROFILE

Analyte	Result µM/L	BRI µM/L
Alanine	292	0 - 480
Citrulline	8	10 - 40
Leucine+Isoleucine	129	0 - 200
Ornithine	86.4	0 - 393
Tyrosine	67.8	0 - 181

Analyte	Result µM/L	BRI µM/L
Arginine	40.6	0 - 130
Glycine	205	0 - 623
Methionine	9.92	0 - 44
Phenylalanine	64.2	0 - 130
Valine	142	0 - 182

ACYLCARINITINE PROFILE

Analyte	Result µM/L	BRI µM/L
Free Carnitine (C0)	4.2	9 - 57
Propionylcarnitine (C3)	0.64	0 - 5.81
Butyrylcarnitine (C4)	0.15	0 - 1.7
Isovalerylcarnitine (C5)	0.1	0 - 0.65
Glutaryl Carnitine (C5DC)	0.03	0 - 0.41
Octanoyl Carnitine (C8)	0.03	0 - 0.39
Decenoylcarnitine (C10:1)	0.02	0 - 0.5
Tetradecanoylcarnitine (C14)	0.06	0 - 0.41
Tetradodecenoylcarnitine (C14:2)	0.01	0 - 0.06
3-Hydroxypalmitoylcarnitine (C16-OH)	0.01	0 - 0.21
Oleylcarnitine (C18:1)	0.2	0.36 - 2.4
3Hydroxylinoleoylcarnitine (C18-OH)	0.01	0 - 0.17

Analyte	Result µM/L	BRI µM/L
Acetylcarnitine (C2)	3.79	3 - 45
Malonylcarnitine+3-hydroxy butyrylcarnitine(C3DC+C4OH)	0.06	0 - 0.35
Methylmalonylcarnitine+3-hydroxy isovalerylcarnitine (C4DC+C5OH)	0.23	0 - 0.4
3-methylcrotonylcarnitine (C5:1)	0.03	0 - 0.2
Hexanoylcarnitine (C6)	0.05	0 - 0.23
Decanoylcarnitine (C10)	0.03	0 - 0.5
Dodecanoylcarnitine (C12)	0.03	0 - 0.42
Tetradecenoylcarnitine (C14:1)	0.04	0 - 0.39
Palmitoylcarnitine (C16)	0.49	0.76 - 6.46
Stearoylcarnitine (C18)	0.18	0.31 - 1.8
3-Hydroxylinoleoylcarnitine (C18:1-OH)	0.04	0 - 0.15

Low Normal Borderline High

BRI - Biological Reference Interval

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 ---