



Laboratory Investigation Report

Patient Name Centre Age/Gender OP/IP No/UHID MaxID/Lab ID Collection Date/Time Ref Doctor Reporting Date/Time

> **Clinical Biochemistry**

Result Unit **Bio Ref Interval**

Uric Acid Random Urine

Uricase, Colorimetric

Test Name

Uric Acid 18.06 mg/dL

Ref Range Male <40 Yrs 9 - 63 >=40 Yrs 6 - 114Female < 40 Yrs 6 - 71 >= 40 Yrs 4 - 93

Kindly correlate with clinical findings

*** End Of Report ***

Dr. Poonam. S. Das, M.D.

Principal Director-Max Lab & Blood Bank Services

Dr. Dilip Kumar M.D. Associate Director & Manager Quality

Dr. Nitin Dayal, M.D. Principal Consultant & Head, Haematopathology

Test Performed at :910 - Max Hospital - Saket M S S H, Press Enclave Road, Mandir Marg, Saket, New Delhi, Delhi 110017 Booking Centre: 1104 - Max Smart- MSSSH,

The authenticity of the report can be verified by scanning the Q R Code on top of the page

Page 1 of 1

Max Lab Limited (A Wholly Owned Subsidiary of Max Healthcare Institute Ltd.)

Max Super Speciality Hospital, Saket (West Block), 1, Press Enclave Road, Saket, New Delhi - 110 017, Phone: +91-11-6611 5050 (CIN No.: U85100DL2021PLC381826)

Liability of Max Healthcare for deficiency of services, or other errors and omissions shall be limited to fee paid by the patient for the relevant laboratory services.



NAME: Age:
PATIENT ID: ACCESSION NO:
COLLECTED ON: REPORTED ON:
REFERRED BY:

METABOLIC SCREENING, URINE

METABOLIC PROFILE

Amino Acidopathies All related analytes within acceptable limits

Fatty Acid Metabolism Disorders

All related analytes within acceptable limits

Organic Acidurias All related analytes within acceptable limits

Carbohydrate Metabolism Disorders

All related analytes within acceptable limits

TCA Cycle / Mitochondrial Dysfunction

All 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 and Recommendation

The levels of all analytes tested are within acceptable limits. Hence, Screen Negative for tested disorders.

No congenital metabolic abnormality was found in the excretion of amino Acids, organic Acids, fatty Acids, sugars, sugar Acids, sugar alcohols, nucleic Acids and nucleic Acid bases when screened for the target metabolic marker compounds.

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



NAME: Age:
PATIENT ID: ACCESSION NO:
COLLECTED ON: REPORTED ON:
REFERRED BY:

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

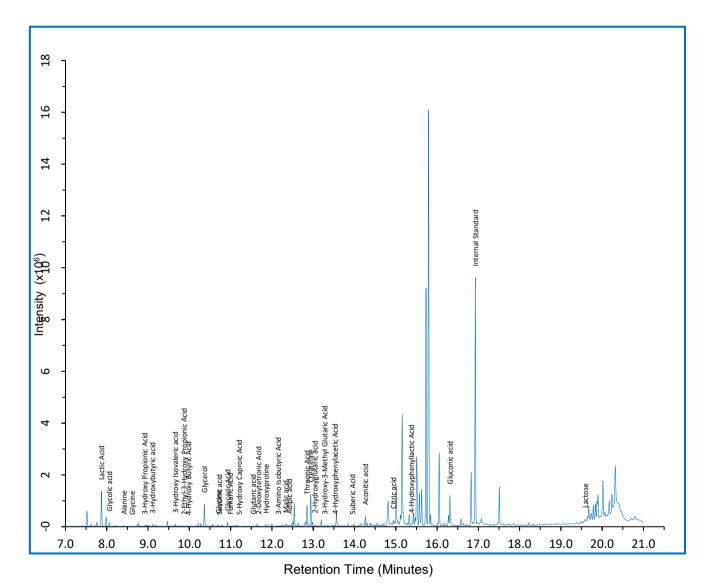


NAME: Age:

PATIENT ID: ACCESSION NO:

COLLECTED ON: REPORTED ON:

REFERRED BY:



Metabolic Profile by GC/MS - Total Ion Chromatogram of Urinary Metabolomes



NAME:
PATIENT ID:
COLLECTED ON:
REFERRED BY:

Age: 03 Years
ACCESSION NO
REPORTED ON

DETAILED REPORT OF ANALYTES TESTED FOR METABOLIC DISORDERS

Analyte Result profitmed Creatinine Profit P	DISORDERS OF AMINO ACID & FATTY ACID METABOLISM AND ORGANIC ACIDURIAS				
2- Ethyl 3-Hydroxy Propionic Acid		BRI µmol/mmol Creatinine			
2- Hydroxy Butyric Acid		4.8 - 22.4			
2- Hydroxy Isobutyric Acid		0 - 2.8			
2- Hydroxy Isovaleric Acid 0 0 - 11.9 2- Hydroxy Phenyl Acetic Acid 0 0 - 0.67 2- Keto Isocapric Acid 0 0 - 0.67 2- Keto Isocapric Acid 0 0 - 0.67 2- Methyl Acetoacetic Acid 0 0 - 7 2- Methyl Acetoacetic Acid 0 0 - 7 2- Methyl Acetoacetic Acid 0 0 0 - 2 2- Methyl Acetoacetic Acid 0 0 0 - 5 2- Methyl Glutaric Acid 0 0 0 - 5 2- Furandicarboxylic Acid 0 0 0 - 5 3- Hydroxy Adipic Acid 0 0 0 - 5 3- Hydroxy Adipic Acid 0 0 0 - 5 3- Hydroxy Dodecanedioic Acid 0 0 0 - 10 3- Hydroxy Isovaleric Acid 0 0 0 - 11 3- Hydroxy Isovaleric Acid 0 0 0 - 11 3- Hydroxy Phenyl 3- Propionic Acid 0 0 0 - 0.5 3- Hydroxy Phenyl 3- Propionic Acid 0 0 0 - 0.5 3- Hydroxy Phenyl 3- Propionic Acid 0 0 0 - 0.5 3- Hydroxy Phenyl 3- Propionic Acid 0 0 0 - 0.5 3- Hydroxy Phenyl Acetic Acid 0 0 0 - 0.5 3- Hydroxy Sebasic Acid 0 0 0 - 0.1 3- Hydroxy Sebasic Acid 0 0 0 - 0.1 3- Hydroxy Sebasic Acid 0 0 0 - 0.1 3- Hydroxy Sebasic Acid 0 0 0 - 0.1 3- Hydroxy Valeric Acid 0 0 0 - 0.1 3- Hydroxy Valeric Acid 0 0 0 - 0.1 3- Hydroxy Valeric Acid 0 0 0 - 1.4 3- Methyl Clutaric Acid 0 0 0 - 1.4 3- Methyl Clutaric Acid 0 0 0 - 1.4 3- Methyl Glutaric Acid 0 0 0 - 1.9 4- Deoxy Tetronic Acid 0 0 0 - 19 4- Hydroxy 3- Methyl Benzoic Acid 2.5 1		4 - 35			
2- Hydroxy Sebasic Acid		0 - 5			
2- Keto Isocapric Acid		0 - 20			
2- Methyl Acetoacetic Acid		0 - 117			
2- Oxoadipic Acid 0 0 - 5 3- Amino Isobutyric Acid 0 1.4 - 6.2 3- Hydroxy Adipic Acid 0 0 0 - 5 3- Hydroxy Adipic Acid 0 0 0 - 5 3- Hydroxy Dodecanedioic Acid 0 0 0 - 10 3- Hydroxy Hexanedioic Acid 0 0 0 - 11 3- Hydroxy Isovaleric Acid 0 0 0 - 11 3- Hydroxy Isovaleric Acid 0 0 0 - 11 3- Hydroxy Phenyl 3- Propionic Acid 0 0 0 - 31 3- Hydroxy Phenyl 3- Propionic Acid 0 0 0 - 31 3- Hydroxy Phenylhydracrylic Acid 0 0 0 - 31 3- Hydroxy Sebasic Acid 0 0 0 - 9.1 3- Hydroxy Valeric Acid 0 0 0 - 1.4 3- Methyl 2-Hydroxy Valeric Acid 0 0 0 - 19 4- Deoxy Tetronic Acid 0 0 0 - 8 2,5 Furandicarboxylic Acid 0 0 0 - 9 3- Deoxy Tetronic Acid 0 0 0 - 9 3- Hydroxy Butyric Acid 0 0 0 0 - 10 3- Hydroxy Butyric Acid 0 0 0 0 - 11 3- Hydroxy Isobutyric Acid 0 0 0 0 - 9.1 3- Hydroxy Phenyl Acetic Acid 0 0 0 0 - 9.1 3- Hydroxy Suberic Acid 0 0 0 - 1.4 3- Methyl 2-Hydroxy Valeric Acid 0 0 0 0 - 19 4- Deoxy Tetronic Acid 0 0 0 - 8		0 - 7			
3- Amino Isobutyric Acid 3- Hydroxy Adipic Acid 3- Hydroxy Adipic Acid 3- Hydroxy Dodecanedioic Acid 3- Hydroxy Butyric Acid 3- Hydroxy Butyric Acid 3- Hydroxy Butyric Acid 3- Hydroxy Butyric Acid 3- Hydroxy Glutaric Acid 3- Hydroxy Isobutyric Acid 3- Hydroxy Isobutyric Acid 3- Hydroxy Isobutyric Acid 3- Hydroxy Phenyl 3- Propionic Acid 3- Hydroxy Phenyl 3- Propionic Acid 3- Hydroxy Phenyl Hydracylic Acid 3- Hydroxy Phenyl Hydracylic Acid 3- Hydroxy Phenyl Acetic Acid 3- Hydroxy Phenyl Acetic Acid 3- Hydroxy Phenyl Acetic Acid 3- Hydroxy Sebasic Acid 3- Hydroxy Sebasic Acid 3- Hydroxy Valeric Acid 3- Hydroxy Valeric Acid 3- Hydroxy Valeric Acid 3- Methyl 2-Hydroxy Valeric Acid 4- Deoxy Tetronic Acid 4- Hydroxy 3- Methyl Benzoic Acid		0 - 2.6			
3- Hydroxy Adipic Acid 0 0 - 5 3- Hydroxy Butyric Acid 0 94 3- Hydroxy Dodecanedioic Acid 1.5 0 - 10 3- Hydroxy Hexanedioic Acid 0 0 - 11 3- Hydroxy Isovaleric Acid 0 3.77 3.1 - 23.1 3- Hydroxy Isovaleric Acid 0.14 0 - 0.5 3- Hydroxy Phenyl 3- Propionic Acid 0.14 0 - 0.5 3- Hydroxy Phenyl Hydracrylic Acid 0.19 0 - 31 3- Hydroxy Phenylhydracrylic Acid 0.79 0 - 9.1 3- Hydroxy Sebasic Acid 0.79 0 - 9.1 3- Hydroxy Valeric Acid 0 0 0 - 1.4 3- Methyl 2-Hydroxy Valeric Acid 0 0 0 - 1.9 3- Methyl Glutaconic Acid 0 0 0 - 19 3- Methyl Glutaconic Acid 0 0 0 - 19 4- Deoxy Tetronic Acid 0 0 - 8 4- Hydroxy 3- Methyl Benzoic Acid 0 4- Hydroxy 3- Methyl Benzoic Acid 0 0 0 - 8		0 - 11			
3- Hydroxy Dodecanedioic Acid 3- Hydroxy Hexanedioic Acid 3- Hydroxy Hexanedioic Acid 3- Hydroxy Isovaleric Acid 3- Hydroxy Isovaleric Acid 3- Hydroxy Isovaleric Acid 3- Hydroxy Sebasic Acid 3- Hydroxy Phenyl Acetic Acid 3- Hydroxy Phenyl Acetic Acid 3- Hydroxy Phenylhydracrylic Acid 3- Hydroxy Phenylhydracrylic Acid 3- Hydroxy Sebasic Acid 3- Hydroxy Sebasic Acid 3- Hydroxy Valeric Acid 3- Hydroxy Valeric Acid 3- Hydroxy Valeric Acid 3- Methyl 2-Hydroxy Valeric Acid 3- Methyl Glutaconic Acid 3- Methyl Glutaconic Acid 4- Deoxy Tetronic Acid 2-12 3- Hydroxy Glutaric Acid 3- Hydroxy Isobutyric Acid 3- Hydroxy Methyl Glutaric Acid 3- Hydroxy Phenyl Acetic Acid 3- Hydroxy Phenyl Acetic Acid 3- Hydroxy Phenyl Acetic Acid 3- Hydroxy Propionic Acid 3- Hydroxy Suberic Acid 3- Methyl Crotonyl Glycine 3- Methyl Glutaric Acid 4- Hydroxy 3- Methyl Benzoic Acid 2-16		1.9 - 15.2			
3- Hydroxy Hexanedioic Acid 0 0 - 11 3- Hydroxy Isobutyric Acid 0 3.77 3.1 - 23.1 3- Hydroxy Phenyl 3- Propionic Acid 0.14 0 - 0.5 3- Hydroxy Phenyl Acetic Acid 0 3- Hydroxy Phenylhydracrylic Acid 0.19 0 - 31 3- Hydroxy Propionic Acid 0 3- Hydroxy Sebasic Acid 0.79 0 - 9.1 3- Hydroxy Suberic Acid 0 3- Hydroxy Valeric Acid 0 0 3- Hydroxy Valeric Acid 0 0 3- Methyl 2-Hydroxy Valeric Acid 0 0 0 - 1.4 3- Methyl Glutaconic Acid 0 0 0 - 19 4- Hydroxy 3- Methyl Benzoic Acid 0 4- Hydroxy 3- Methyl Benzoic Acid 0 4- Hydroxy 3- Methyl Benzoic Acid 0 0 0 - 8		0 - 11.1			
3- Hydroxy Isovaleric Acid 3.77 3.1 - 23.1 3- Hydroxy Methyl Glutaric Acid 2.12 3- Hydroxy Phenyl 3- Propionic Acid 0.14 0 - 0.5 3- Hydroxy Phenyl Acetic Acid 0 3- Hydroxy Phenylhydracrylic Acid 0 3- Hydroxy Phenylhydracrylic Acid 0 3- Hydroxy Sebasic Acid 0.79 0 - 9.1 3- Hydroxy Suberic Acid 0 3- Hydroxy Suberic Acid 0 3- Hydroxy Suberic Acid 0 3- Methyl 2-Hydroxy Valeric Acid 0 3- Methyl Glutaric Acid 0 4- Deoxy Tetronic Acid 0 4- Hydroxy 3- Methyl Benzoic Acid 0 4- Hydroxy 3- Methyl Benzoic Acid		0 - 4.6			
3- Hydroxy Phenyl 3- Propionic Acid 0.14 0 - 0.5 3- Hydroxy Phenyl Acetic Acid 0 3- Hydroxy Phenylhydracrylic Acid 0.79 0 - 9.1 3- Hydroxy Sebasic Acid 0.79 0 - 9.1 3- Hydroxy Suberic Acid 0.95 3- Hydroxy Valeric Acid 0 0 - 1.4 3- Methoxy Benzene Propionic Acid 0 0 - 1.4 3- Methoxy Benzene Propionic Acid 0 0 - 5 3- Methyl Crotonyl Glycine 0 3- Methyl Glutaconic Acid 0 0 - 19 3- Methyl Glutaric Acid 0 0 - 8 4- Hydroxy 3- Methyl Benzoic Acid 0 4- Hydroxy 3- Methyl Benzoic Acid 0 0 - 8		0 - 137			
3- Hydroxy Phenylhydracrylic Acid 1.9 0 - 31 3- Hydroxy Propionic Acid 0 3- Hydroxy Sebasic Acid 0.79 0 - 9.1 3- Hydroxy Suberic Acid 0.95 3- Hydroxy Valeric Acid 0 0 - 1.4 3- Methoxy Benzene Propionic Acid 0 3- Methyl 2-Hydroxy Valeric Acid 0 0 - 5 3- Methyl Crotonyl Glycine 0 3- Methyl Glutaconic Acid 0 0 - 19 3- Methyl Glutaric Acid 0 4- Hydroxy 3- Methyl Benzoic Acid 0 4- Hydroxy 3- Methyl Benzoic Acid 0 4- Hydroxy 3- Methyl Benzoic Acid 0 0 - 8		0 - 12			
3- Hydroxy Sebasic Acid 0.79 0 - 9.1 3- Hydroxy Suberic Acid 0.95 3- Hydroxy Valeric Acid 0.95 3- Methyl 2-Hydroxy Valeric Acid 0.5 3- Methyl Glutaconic Acid 0.5 3- Methyl Glutaconic Acid 0.5 3- Methyl Glutaconic Acid 0.5 3- Methyl Glutaric Acid 0.5 4- Hydroxy 3- Methyl Benzoic Acid 0.5 4- Hydroxy 3- Hydr		0 - 11			
3- Hydroxy Valeric Acid 3- Methyl 2-Hydroxy Valeric Acid 3- Methyl Glutaconic Acid 3- Methyl Glutaconic Acid 4- Deoxy Tetronic Acid 0 - 1.4 3- Methoxy Benzene Propionic Acid 3- Methyl Crotonyl Glycine 3- Methyl Glutaric Acid 0 - 19 4- Hydroxy 3- Methyl Benzoic Acid 4- Hydroxy 3- Methyl Benzoic Acid 2.61		0 - 36			
3- Methyl 2-Hydroxy Valeric Acid 0 0 - 5 3- Methyl Crotonyl Glycine 0 3- Methyl Glutaconic Acid 0 0 - 19 3- Methyl Glutaric Acid 0 4- Deoxy Tetronic Acid 2.1 0 - 8 4- Hydroxy 3- Methyl Benzoic Acid 2.61		0 - 5.6			
3- Methyl Glutaconic Acid 0 0 - 19 4- Deoxy Tetronic Acid 2.1 0 - 8 4- Hydroxy 3- Methyl Benzoic Acid 2.61		0 - 11.9			
4- Deoxy Tetronic Acid 2.1 0 - 8 4- Hydroxy 3- Methyl Benzoic Acid 2.61		0 - 10			
		0 - 3			
4- Hydroxy Benzoic Acid (2.12) 0 - 16 4- Hydroxy Butyric Acid (0.65)		0 - 28.6			
		0 - 10			
4- Hydroxy Cyclohexylacetic acid 0 0 - 3 4- Hydroxy Phenyl Acetic Acid (23.8)		3.2 - 180.2			

Low Normal Borderline

Highlew Delhi - 110 017, Phone: +BRL Biological Reference Interval

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NAME: BARY SANGHAVI SINGH Age: 03 Yes

PATIENT ID : COLLECTED ON : 2
REFERRED BY : §

ACCESSI REPORT

DETAILED REPORT OF ANALYTES TESTED FOR METABOLIC DISORDERS

DISORDERS OF AMINO ACID & FATTY ACID METABOLISM AND ORGANIC ACIDURIAS						
Analyte	Result		BRI µmol/mmol Creatinine	Analyte	Result	BRI µmol/mmol Creatinine
4- Hydroxy Phenyl Lactic Acid	1.1		0 - 8.7	4- Hydroxy Phenyl Pyruvic Acid	0	0 - 0.4
4- Hydroxy Proline	11.54		0 - 135	5- Hydroxy Caproic Acid	0.49	0 - 1.9
5-Hydroxy Indole Acetic Acid	0		0 - 11.5	5- Hydroxy Methyl Furanoic Acid	0	0 - 44
7- Hydroxy Octanoic Acid	0		0 - 2	7- Hydroxy Octanoyl Glycine	0	0 - 0.2
8- Hydroxy Octanoyl Glycine	0		0 - 0.2	Acetoacetic Acid	0	0 - 5.8
Acetyl Glycine	0		0 - 1	Adipic Acid	1.7	0 - 35
Alanine	6.82		6.7 - 17.9	Argininosuccinic Acid	0	0 - 0.1
Aspargine	0.22		0 - 1.6	Aspartic Acid	0.37	0 - 2.46
Azelic Acid	0		0 - 46.7	Benzene dicarboxylic Acid	9.2	0 - 109
Beta Alanine	0		0 - 0.96	Butyryl Glycine	0	0 - 2
Cis-Aconitic Acid	27.4		14.7 - 93	Citramalic Acid	1.4	0 - 11.2
Citric Acid	65.2		33.2 - 652	Cystathionine	0	0 - 1
Cysteine	1.85		0 - 45.4	Decanoic Acid	0	0 - 1
Decanoyl Glycine	0		0 - 1	Dimethylglycine	0	0 - 1
Docosanoic acid	0		0 - 1	Dodecanoic Acid	0	0 - 0.05
Dodecendioic Acid	0		0 - 0.05	Eicosadecanoic Acid	0	0 - 1
Eicosadecenoic Acid	0		0 - 1	Ethanolamine	9.52	0 - 165
Ethyl Hydracrylic Acid	0.29		0 - 13.3	Ethyl Malonic Acid	0	0 - 14.6
Formiminoglutamic Acid	0.2		0 - 1.61	Furoic Acid	3.4	0 - 28
Glutaconic Acid	0		0 - 0	Glutamic Acid	17.1	0 - 21
Glutamine	73.8		69 - 127	Glutaric Acid	0.48	0 - 5.3
Glyceric Acid	4.9		0 - 37.3	Glycine	198	184 - 508
Glycolic Acid	14.7		0 - 198	Glycoxylic Acid	0	0 - 15.9

Low Normal Borderline

Highlew Delhi - 110 017, Phone: + BRL Biological Reference Interval

Page 5 | 10



 NAME :
 Age : 03 Years

 PATIENT ID :
 ACCESSION NO

 COLLECTED ON :
 3:25
 REPORTED ON

REFERRED BY :

DETAILED REPORT OF ANALYTES TESTED FOR METABOLIC DISORDERS

Analyte	Result	BRI µmol/mmol Creatinine	Analyte	Result	BRI µmol/mmol Creatinine
Hawkinsin	0	0 - 0.1	Hexadecanoic (Palmitic) Acid	0.3	0.1 - 8.2
Hexanoic Acid	0	0 - 2.3	Hexanoyl Glycine	0	0 - 2.9
Hippuric Acid	14.8	0 - 145	Histidine	46.4	0 - 339
Homocysteine	0	0 - 4.19	Homogentisic Acid	0	0 - 0
Homoserine	0	0 - 0.66	Hydroxylysine	0	0 - 0.97
Indole Acetic acid	0	0 - 3.4	Isobutyrylglycine	0	0 - 2.9
Isocitric Acid	10.4	0 - 141	Isoleucine	2.11)	0 - 16.6
Isovalerylglycine	0	0 - 10	Kynurenic Acid	0	0 - 16.508
Lactic Acid	83.1	5 - 118	Leucine	3.22	2 - 24
Linoleinic Acid	0	0 - 1.27	Lysine	5.4	0 - 81.2
Malonic Acid	0	0 - 28.8	Mandelic Acid	0	0 - 64
Methionine	0.84	0 - 2.4	Methyl Adipic Acid	0	0 - 5
Methyl Citric Acid	0	0 - 8.5	Methyl Fumaric Acid	0	0 - 0.2
Methyl Malic Acid	0	0 - 0.2	Methyl Malonic Acid	0	0 - 6
Methyl Succinic Acid	0	0 - 6	Mevalonic Acid	0	0 - 0.3
Mevalonolactone	0	0 - 0	N-Acetyl Alanine	0	0 - 1
N- Acetyl Aspartic Acid	7.6	6 - 40.8	N-Acetyl Glycine	0	0 - 3.4
N-Acetyl Proline	0	0 - 2	N- Acetyl Tyrosine	0	0 - 10
Nonadecanoic acid	0	0 - 1	Nonanoic acid	0	0 - 1
Octanoic Acid	0	0 - 7.7	Octenedioic Acid	0	0 - 2.8
Oleic Acid	0	0 - 0.4	Ornithine	0	0 - 11.8
Orotic Acid	0	0 - 4.3	Oxalic Acid	0	0 - 19
p-Cresol	0	0 - 11	Pentadecanoic acid	0	0 - 1

Low Normal Borderline

Highlew Delhi - 110 017, Phone: +BRL Biological Reference Interval

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REFERRED BY:

NAME: Age: 03 Years PATIENT ID: **ACCESSION NO COLLECTED ON:** 8:25 **REPORTED ON**

DETAILED REPORT OF ANALYTES TESTED FOR METABOLIC DISORDERS

DISORDERS OF AMINO ACID & FATTY ACID METABOLISM AND ORGANIC ACIDURIAS					
Analyte	Result		BRI µmol/mmol Creatinine	Analyte	F
Phenol	4.2		0 - 53	Phenyl Acetyl Glycine	6.39
Phenyl Alanine	20.4		0 - 71.6	Phenyl Lactic Acid	0
Phenyl Pyruvic Acid	0		0 - 2	Pimelic Acid	0.3
Pipecolic Acid	0		0 - 0.24	Proline	
Propionic Acid	1.7		0 - 11.2	Propionyl Glycine	0
Pyroglutamic Acid	21.6		0 - 79	Pyruvic Acid	2.5
Sarcosine	0.46		0 - 2	Sebasic Acid	0
Serine	20.7		0 - 177.3	Stearic Acid	0
Suberic Acid	0.75		0 - 10.1	Suberyl Glycine	0
Succinic Acid	7.5		4.9 - 81.3	Succinyl Acetone	0
Tetracosanoic acid	0		0 - 1	Tetradecenoic Acid	0
Threonine	21.8		0 - 58.2	Tiglyl Glycine	0
Tridecanoic Acid	0		0 - 1	Tryptophan	13.8
Tyrosine	10.7		6.2 - 59	Uracil	1.9
Valine	7.4		3.3 - 19.8	Xanthurenic Acid	0

Analyte	Result	BRI µmol/mmol Creatinine
Phenyl Acetyl Glycine	6.39	0 - 110
Phenyl Lactic Acid	0	0 - 4
Pimelic Acid	0.3	0 - 14
Proline	6.33	0 - 12.7
Propionyl Glycine	0	0 - 0.1
Pyruvic Acid	2.5	0 - 40.5
Sebasic Acid	0	0 - 12.3
Stearic Acid	0	0 - 8
Suberyl Glycine	0	0 - 1
Succinyl Acetone	0	0 - 0
Tetradecenoic Acid	0	0 - 1
Tiglyl Glycine	0	0 - 6.7
Tryptophan	13.8	2 - 46
Uracil	1.9	0 - 20.9
Xanthurenic Acid	0	0 - 4

DISORDERS OF CARBOHYDRATE METABOLISM

Analyte	Result	BRI µmol/mmol Creatinine
Arabinose	4.1	0 - 19.4
Fructose	2.1	0 - 35
Galactonic Acid	5.8	0 - 127.7
Gluconic Acid	40.9	0 - 122

Analyte	Result	BRI µmol/mmol Creatinine
Erythronic Acid	10.9	3.9 - 22.2
Galactitol	0.6	0 - 12
Galactose	7	0 - 49
Glucose	18.9	0 - 80

Low Normal Borderline (CIN No.: U85100DL2021PLC381826)

Highlew Delhi - 110 017, Phone: + BRL5 Biological Reference Interval

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NAME: PATIENT ID: **COLLECTED ON:** REFERRED BY:

Age: 03 Years **ACCESSION NO REPORTED ON**

DETAILED REPORT OF ANALYTES TESTED FOR METABOLIC DISORDERS

DISORDERS OF CARBOHYDRATE METABOLISM					
Analyte	Result	BRI µmol/mmol Creatinine			
Glucuronic Acid	0	0 - 75.7			
Lactic Acid	83.1	5 - 118			
Mannose	0.6	0 - 3.3			
Sucrose	4.3	0 - 91			
Xylose	0	0 - 67.1			

Analyte	Result	BRI μmol/mmol Creatinine
Glyceraldehyde 3- Phosphate	0	0 - 1
Lactose	11.7	0 - 52.4
Ribose	0.35	0 - 5
Threonic Acid	3.27	1.7 - 13.7

DISORDERS OF TCA CYCLE/MITOCHONDRIAL DYSFUNCTION

Analyte	Result	BRI µmol/mmol Creatinine
2- Keto Glutaric Acid	21.8	0 - 117
Cis-Aconitic Acid	27.4	14.7 - 93
Fumaric Acid	0.25	0 - 9.9
Lactic Acid	83.1	5 - 118
Methyl Fumaric Acid	0	0 - 0.2
Pyruvic Acid	2.5	0 - 40.5

Analyte	Result	BRI µmol/mmol Creatinine
Alanine	6.82	6.7 - 17.9
Citric Acid	65.2	33.2 - 652
Isocitric Acid	10.4	0 - 141
Malic Acid	0.3	0 - 16.2
Methyl Malic Acid	0	0 - 0.2
Succinic Acid	7.5	4.9 - 81.3

DISORDERS OF PURINE/PYRIMIDINE METABOLISM

Analyte	Result	BRI µmol/mmol Creatinine
2,8 Dihydroxyadenine	0	0 - 0
Adenosine	1.59	0 - 11
Deoxyadenosine	0	0 - 27.3
Dihydro Uracil	0	0 - 20
Hypoxanthine	(10.95)	0 - 60

Analyte	Result	BRI µmol/mmol Creatinine
5- Hydroxy Methyl Uracil	0	0 - 0.0121
Beta-Ureidopropionic Acid	0.75	0 - 4.8
Dihydro Thymine	0	0 - 3
Guanosine	0	0 - 11
Inosine	0	0 - 3.1

Low Normal Borderline (CIN No.: U85100DL2021PLC381826)

Highlew Delhi - 110 017, Phone: + BRL5 Biological Reference Interval

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NAME: Age: 03 Years PATIENT ID: **ACCESSION NO COLLECTED ON: REPORTED ON** REFERRED BY:

DETAILED REPORT OF ANALYTES TESTED FOR METABOLIC DISORDERS

DISORDERS OF I	DISORDERS OF PURINE/PYRIMIDINE METABOLISM					
Analyte	Result	BRI μmol/mmol Creatinine	Analyte	Result	BRI µmol/mmol Creatinine	
Orotic Acid	0	0 - 4.3	Pseudouridine	20.26	0 - 162	
Thymine	0	0 - 1.7	Uracil	1.9	0 - 20.9	
Uric Acid	46.7)	0 - 462	Uridine	0.86	0 - 9	
Xanthine	10.2	0 - 62				

PEROXISOMAL DISC	ORDERS				
Analyte	Result	BRI µmol/mmol Creatinine	Analyte	Result	BRI µmol/mmol Creatinine
2,6 Dimethyloctanedioic Acid	0	0 - 0	3-Methyl Adipic Acid	1.46	0 - 3.554
Adipic Acid	1.7	0 - 35	Malic Acid	0.3	0 - 16.2
Oxalic Acid	0	0 - 19	Sebasic Acid	0	0 - 12.3
Suberic Acid	0.75	0 - 10.1			

DISORDERS OF NEUROTRANSMITTER METABOLISM					
Analyte	Result	BRI μmol/mmol Creatinine	Analyte	Result	BRI µmol/mmol Creatinine
3- Hydroxy Mandelic Acid	0	0 - 0.1	4- Hydroxy Mandelic Acid	1.76	1.3 - 10
Adipic Acid	1.7	0 - 35	Homovanillic Acid	2.7	0 - 13.8
Quinolinic Acid	1.2	0-26.2	Sebasic Acid	0	0 - 12.3
Vanillyl Mandelic Acid	1.88	0 - 16.1			



LIST OF DISORDERS SCREENED

٩mi	ino 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			96	Methylmalc Cbl A, B)
3	3- Hydroxyisobutyryl-CoA-Deacy			97	Methylmalc Deficiency
4	5-Oxoprolinuria			98	Methylmak
5	Alkaptonuria	53	Transient Tyrosinemia in Infancy	99	Mevalonic ,
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)	Fatt	ty Acid Oxidation Disorders	106	Pyruvate Dehydrogenase Phosphatase Deficienc
14	Cystathioninuria	61	2, 4 - Dienoy CoA Reductase Deficiency	Carl	bohydrate 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) Very Long-Chain Acyl- CoA Dehydrogenase	114	Heredetary Fructose Intolerance
23	Glycerol Kinase Deficiency	70	Deficiency (VLCAD)	115	Lactose Intolerance
24	Glycine Encephalopathy	Org	anic 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)	Puri	ine / Pyrimidine Metabolism Disorde
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	Per	oxisomal 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
	Iminoglycinuria	88	Glutaric Aciduria Type III		Zellweger Like Syndrome (ZLS)
42	Lysinuric Protein Intolerance	89	Glutathionuria		Zellweger Syndrome
			Histidinemia		rotransmitter Metabolism Disorders
43	Maple Syrup Urine Disease (MSUD)	90	Historia	IACE	
43 44					
42 43 44 45 46	Maple Syrup Urine Disease (MSUD) N-Acetyl Glutamate Synthetase Deficiency Ornithine Transcarbamylase (OTC) Deficiency	90 91 92	Hyperpipecolatemia Isobutyryl-CoA Dehydrogenase Deficiency	134	Neuroblastoma

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