# NeoCare Diagnostics Pvt. Ltd.

604-A, Ecstasy Business Park, Citi of Joy, JSD Road, Mulund (W), Mumbai 400080
Phone : 91 8850833991, 022 25644465 Email : neocarediagnostics@gmail.com

MC-2180


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## METABOLIC SCREENING, URINE

| DISORDERS | ANALYTES | RESULTS | BIOLOGICAL REFERENCE INTERVAL | UNITS |
|---|---|---|---|---|
| **METABOLIC PROFILE** | | | | |
| **Amino Acid Metabolism** | | | All related analytes within acceptable limits | |
| **Fatty Acid Metabolism** | | | All related analytes within acceptable limits | |
| **Organic Acidurias** | | | All related analytes within acceptable limits | |
| **Carbohydrate / TCA Cycle / Mitochondrial Metabolism** | | | All related analytes within acceptable limits | |
| **Purine / Pyrimidine Metabolism** | | | All related analytes within acceptable limits | |
| **Peroxisomal Metabolism** | | | All related analytes within acceptable limits | |
| **Neurotransmitter Metabolism** | | | All related analytes within acceptable limits | |

Method : Gas Chromatography Mass Spectrometry

### Clinical History

C/o Vomiting on and off the take any medicine also with Nausea, Liver enzymes Deranged with all oral Medicine.

### Analytical Interpretation

**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.

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## 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)**

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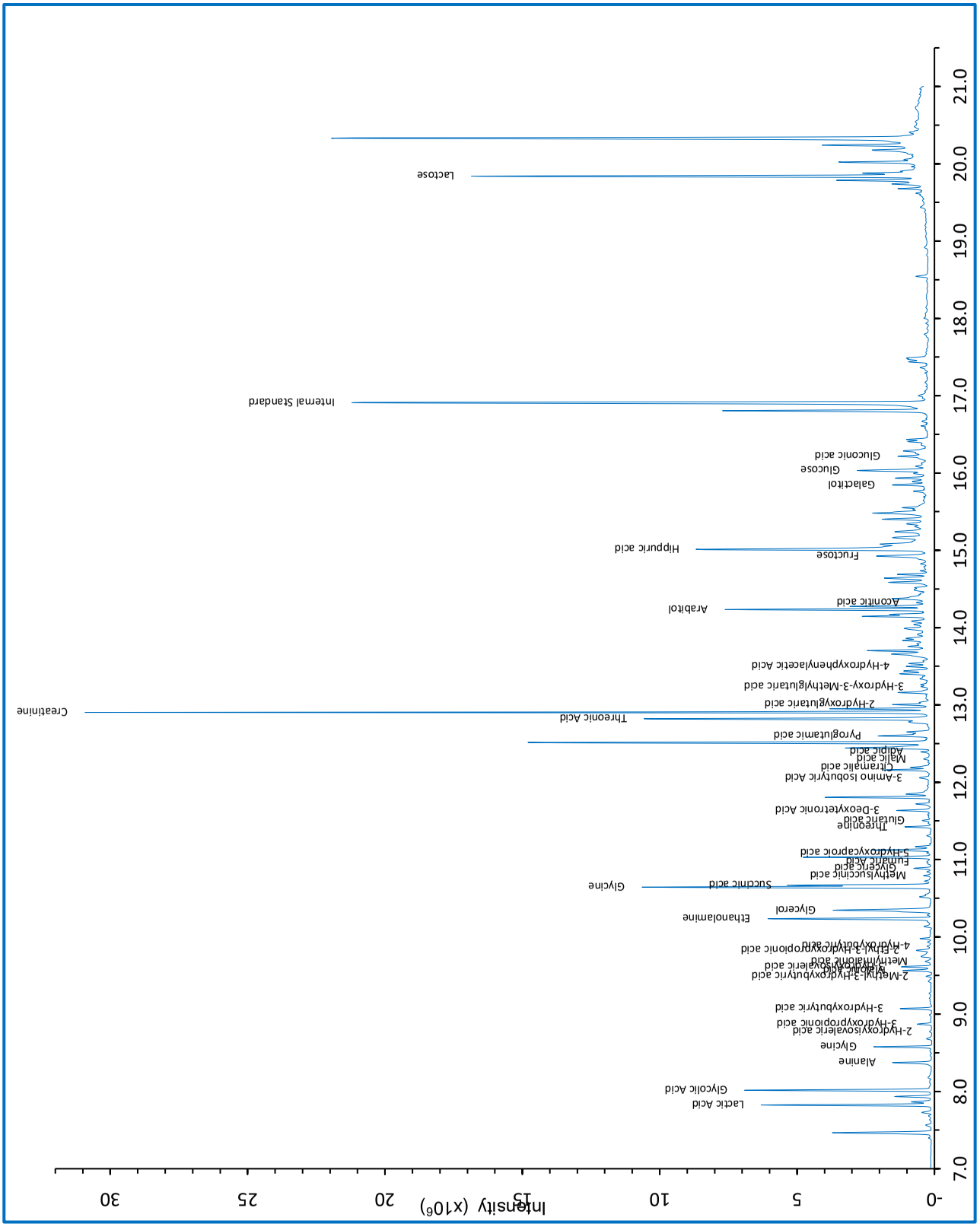

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## DETAILED REPORT OF ANALYTES TESTED FOR METABOLIC DISORDERS

### 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 | 0.27 | 0 - 2.9 |
| 2- Hydroxy Butyric Acid | 0 | 0 - 0.7 |
| 2- Hydroxy Isobutyric Acid | 0.23 | 0 - 4.6 |
| 2- Hydroxy Isovaleric Acid | 0.39 | 0 - 0.97 |
| 2- Hydroxy Sebasic Acid | 0 | 0 - 0.67 |
| 2- Keto Isocaproic Acid | 0 | 0 - 0.89 |
| 2- Methyl Acetoacetic Acid | 0 | 0 - 2 |
| 2- Oxoadipic Acid | 0 | 0 - 1.7 |
| 3- Amino Isobutyric Acid | 1.16 | 1 - 19 |
| 3- Hydroxy Adipic Acid | 0 | 0 - 6 |
| 3- Hydroxy Dodecanedioic Acid | 1.35 | 0 - 10 |
| 3- Hydroxy Hexanedioic Acid | 0 | 0 - 11 |
| 3- Hydroxy Isovaleric Acid | 1.42 | 0 - 29 |
| 3-(3- Hydroxy Phenyl) 3- Hydroxy Propionic Acid | 0 | 0 - 0.5 |
| 3- Hydroxy Phenylhydracrylic Acid | 1.44 | 0 - 15.3 |
| 3- Hydroxy Sebasic Acid | 0.25 | 0 - 3.8 |
| 3- Hydroxy Valeric Acid | 0 | 0 - 1.4 |
| 3- Methyl 2-Hydroxy Valeric Acid | 0 | 0 - 2.1 |
| 3- Methyl Glutaconic Acid | 0 | 0 - 8.3 |
| 4- Deoxy Tetronic Acid | 1.16 | 0 - 8 |
| 4- Hydroxy Benzoic Acid | 0.97 | 0 - 3.6 |
| 4- Hydroxy Cyclohexylacetic acid | 0 | 0 - 3 |
| 4- Hydroxy Phenyl Lactic Acid | 0 | 0 - 2.6 |

| Analyte | Result | BRI µmol/mmol Creatinine |
|---|---|---|
| 2- Deoxy Tetronic Acid | 5.26 | 5 - 57.2 |
| 2- Hydroxy Adipic Acid | 0 | 0 - 2.8 |
| 2- Hydroxy Glutaric Acid | 1.43 | 0 - 8 |
| 2- Hydroxy Isocaproic Acid | 0 | 0 - 5 |
| 2- Hydroxy Phenyl Acetic Acid | 0 | 0 - 0.46 |
| 2- Keto Glutaric Acid | 5.7 | 4 - 52 |
| 2- Methyl 3-Hydroxy Butyric Acid | 0.69 | 0 - 6.2 |
| 2- Methyl Glutaric Acid | 0 | 0 - 2.6 |
| 2,5 Furandicarboxylic Acid | 1.05 | 0 - 12 |
| 3- Deoxy Tetronic Acid | 1.19 | 0 - 1.8 |
| 3- Hydroxy Butyric Acid | 2.2 | 0 - 2.8 |
| 3- Hydroxy Glutaric Acid | 0 | 0 - 6.2 |
| 3- Hydroxy Isobutyric Acid | 0 | 0 - 97 |
| 3- Hydroxy Methyl Glutaric Acid | 1.04 | 0 - 15 |
| 3- Hydroxy Phenyl Acetic Acid | 0 | 0 - 8.1 |
| 3- Hydroxy Propionic Acid | 1.04 | 0 - 11.8 |
| 3- Hydroxy Suberic Acid | 0.37 | 0 - 5.6 |
| 3- Methoxy Benzene Propionic Acid | 0 | 0 - 11.9 |
| 3- Methyl Crotonyl Glycine | 0 | 0 - 2 |
| 3- Methyl Glutaric Acid | 0 | 0 - 1.2 |
| 4- Hydroxy 3- Methyl Benzoic Acid | 0 | 0 - 9.7 |
| 4- Hydroxy Butyric Acid | 0 | 0 - 2.8 |
| 4- Hydroxy Phenyl Acetic Acid | 4.1 | 0 - 29 |
| 4- Hydroxy Phenyl Pyruvic Acid | 0 | 0 - 0.4 |

Low Normal Borderline High

BRI - Biological Reference Interval

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### DISORDERS OF AMINO ACID & FATTY ACID METABOLISM AND ORGANIC ACIDURIAS

| Analyte | Result | BRI µmol/mmol Creatinine |
|---|---|---|
| 4- Hydroxy Proline | 0.84 | 0 - 2 |
| 5-Hydroxy Indole Acetic Acid | 0 | 0 - 7.2 |
| 7- Hydroxy Octanoic Acid | 0 | 0 - 2 |
| 8- Hydroxy Octanoyl Glycine | 0 | 0 - 0.2 |
| Acetyl Glycine | 0 | 0 - 1 |
| Alanine | 2.79 | 0.8 - 15.1 |
| Asparagine | 0.43 | 0 - 5.1 |
| Azelaic Acid | 0 | 0 - 15 |
| Butyryl Glycine | 0 | 0 - 2 |
| Citramalic Acid | 1.1 | 0 - 5.8 |
| 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 | 14.5 | 0 - 69 |
| Ethyl Malonic Acid | 0 | 0 - 4.2 |
| Furoic Acid | 1.57 | 0 - 28 |
| Glutamic Acid | 1.17 | 0 - 2.3 |
| Glutaric Acid | 0.41 | 0 - 2.6 |
| Glycerol | 5.9 | 0 - 19 |
| Glycolic Acid | 14.3 | 0 - 67 |
| Hawkinsin | 0 | 0 - 0.1 |
| Hexanoic Acid | 0 | 0 - 2.3 |
| Hippuric Acid | 36.4 | 0 - 603 |

| Analyte | Result | BRI µmol/mmol Creatinine |
|---|---|---|
| 5- Hydroxy Caproic Acid | 0.54 | 0 - 1.8 |
| 5- Hydroxy Methyl Furanoic Acid | 6.6 | 0 - 44 |
| 7- Hydroxy Octanoyl Glycine | 0 | 0 - 0.2 |
| Acetoacetic Acid | 0 | 0 - 0.82 |
| Adipic Acid | 1.15 | 0 - 2.8 |
| Argininosuccinic Acid | 0 | 0 - 0.6 |
| Aspartic Acid | 0.12 | 0 - 0.82 |
| Beta Alanine | 0.31 | 0 - 1.4 |
| Cis-Aconitic Acid | 2.21 | 0 - 36 |
| Citric Acid | 49.1 | 40 - 520 |
| Cysteine | 4.2 | 0 - 27 |
| 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 | 0 - 2.9 |
| Formiminoglutamic Acid | 0 | 0 - 1.5 |
| Glutaconic Acid | 0 | 0 - 3.1 |
| Glutamine | 8.5 | 1 - 37 |
| Glyceric Acid | 1.24 | 1 - 16.4 |
| Glycine | 39.6 | 3.2 - 95 |
| Glyoxylic Acid | 0 | 0 - 3.26 |
| Hexadecanoic (Palmitic ) Acid | 4.1 | 2.6 - 24.3 |
| Hexanoyl Glycine | 0 | 0 - 2 |
| Histidine | 22.5 | 0 - 106 |

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| Analyte | Result | BRI µmol/mmol Creatinine |
|---|---|---|
| Homocysteine | 0 | 0 - 1.2 |
| Homoserine | 0 | 0 - 0.66 |
| Indole Acetic acid | 0.86 | 0 - 4.2 |
| Isocitric Acid | 7.8 | 0 - 65 |
| Isovaleryl Glycine | 0 | 0 - 3.7 |
| Lactic Acid | 13.1 | 1.9 - 19.8 |
| Linoleinic Acid | 0 | 0 - 1.27 |
| Malonic Acid | 0 | 0 - 3.1 |
| Methionine | 0.33 | 0 - 1.4 |
| Methyl Citric Acid | 0 | 0 - 2 |
| Methyl Malic Acid | 0 | 0 - 0.2 |
| Methyl Succinic Acid | 1.1 | 0 - 6.2 |
| Mevalonolactone | 0 | 0 - 0 |
| N- Acetyl Aspartic Acid | 2.81 | 2.6 - 5.4 |
| N-Acetyl Proline | 0 | 0 - 2 |
| Nonadecanoic acid | 0 | 0 - 1 |
| Octanoic Acid | 0 | 0 - 7.7 |
| Oleic Acid | 0 | 0 - 13 |
| Orotic Acid | 0 | 0 - 1 |
| p-Cresol | 0 | 0 - 71 |
| Phenol | 3.81 | 0 - 12.8 |
| Phenylalanine | 4.5 | 0 - 11.3 |
| Phenyl Pyruvic Acid | 0 | 0 - 0.76 |
| Pipecolic Acid | 0 | 0 - 0.24 |
| Propionic Acid | 0.83 | 0 - 11.2 |

| Analyte | Result | BRI µmol/mmol Creatinine |
|---|---|---|
| Homogentisic Acid | 0 | 0 - 0 |
| Hydroxylysine | 0 | 0 - 1.2 |
| Isobutyryl Glycine | 0 | 0 - 9 |
| Isoleucine | 0.57 | 0 - 1.4 |
| Kynurenic Acid | 0 | 0 - 7.1 |
| Leucine | 0.45 | 0.1 - 2.3 |
| Lysine | 4.15 | 0.2 - 16 |
| Mandelic Acid | 0 | 0 - 0.48 |
| Methyl Adipic Acid | 0 | 0 - 5.6 |
| Methyl Fumaric Acid | 0 | 0 - 0.2 |
| Methyl Malonic Acid | 0.75 | 0 - 1.9 |
| Mevalonic Acid | 0 | 0 - 0.2 |
| N-Acetyl Alanine | 0 | 0 - 1 |
| N-Acetyl Glycine | 0 | 0 - 3.4 |
| N- Acetyl Tyrosine | 0 | 0 - 2 |
| Nonanoic acid | 0 | 0 - 1 |
| Octenedioic Acid | 0 | 0 - 2.8 |
| Ornithine | 2.95 | 2 - 8.8 |
| Oxalic Acid | 5.13 | 0 - 78 |
| Pentadecanoic acid | 0 | 0 - 1 |
| Phenyl Acetyl Glycine | 11.4 | 0 - 110 |
| Phenyl Lactic Acid | 0 | 0 - 0.49 |
| Pimelic Acid | 0.26 | 0 - 3.5 |
| Proline | 0.32 | 0 - 1.9 |
| Propionyl Glycine | 0 | 0 - 0.1 |

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## 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 |
|---|---|---|---|---|---|
| Pyroglutamic Acid | 5.9 | 4.5 - 34 | Pyruvic Acid | 6.4 | 2.6 - 32 |
| Sarcosine | 0.17 | 0 - 0.34 | Sebasic Acid | 0 | 0 - 0.24 |
| Serine | 9.3 | 0 - 35.8 | Stearic Acid | 0 | 0 - 6.6 |
| Suberic Acid | 0.81 | 0 - 2.1 | Suberyl Glycine | 0 | 0 - 1 |
| Succinic Acid | 9.38 | 0 - 16 | Succinyl Acetone | 0 | 0 - 0.001 |
| Tetracosanoic acid | 0 | 0 - 1 | Tetradecenoic Acid | 0 | 0 - 1 |
| Threonine | 2.41 | 0 - 8.1 | Tiglyl Glycine | 0 | 0 - 1.8 |
| Tridecanoic Acid | 0 | 0 - 1 | Tryptophan | 2.18 | 0 - 6.9 |
| Tyrosine | 8.6 | 0 - 23.2 | Uracil | 0.52 | 0 - 6.9 |
| Valine | 3.13 | 0 - 9.8 | Xanthurenic Acid | 0 | 0 - 0.96 |

### DISORDERS OF CARBOHYDRATE METABOLISM

| Analyte | Result | BRI µmol/mmol Creatinine | Analyte | Result | BRI µmol/mmol Creatinine |
|---|---|---|---|---|---|
| Arabitol | 11.5 | 0 - 36 | Erythronic Acid | 4.22 | 3.9 - 22.2 |
| Fructose | 7.78 | 0 - 35 | Galactitol | 2.9 | 0 - 13 |
| Galactonic Acid | 6.7 | 0 - 77 | Galactose | 4.9 | 0 - 8.5 |
| Gluconic Acid | 9.2 | 8.2 - 26.4 | Glucose | 17.5 | 10.3 - 56.7 |
| Glucuronic Acid | 4.17 | 3.7 - 20.6 | Glyceraldehyde 3- Phosphate | 0 | 0 - 1 |
| Lactic Acid | 13.1 | 1.9 - 19.8 | Lactose | 13.6 | 0 - 24.2 |
| Mannose | 0 | 0 - 2.5 | Ribose | 0 | 0 - 5 |
| Sucrose | 4.31 | 0 - 9.2 | Threonic Acid | 9.9 | 8.6 - 36.1 |
| Xylose | 0 | 0 - 67.1 | | | |

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## DETAILED REPORT OF ANALYTES TESTED FOR METABOLIC DISORDERS

### DISORDERS OF TCA CYCLE/MITOCHONDRIAL DYSFUNCTION

| Analyte | Result | BRI µmol/mmol Creatinine | Analyte | Result | BRI µmol/mmol Creatinine |
|---|---|---|---|---|---|
| 2- Keto Glutaric Acid | 5.7 | 4 - 52 | Alanine | 2.79 | 0.8 - 15.1 |
| Cis-Aconitic Acid | 2.21 | 0 - 36 | Citric Acid | 49.1 | 40 - 520 |
| Fumaric Acid | 0.39 | 0 - 1.8 | Isocitric Acid | 7.8 | 0 - 65 |
| Lactic Acid | 13.1 | 1.9 - 19.8 | Malic Acid | 1.04 | 0 - 5.3 |
| Methyl Fumaric Acid | 0 | 0 - 0.2 | Methyl Malic Acid | 0 | 0 - 0.2 |
| Pyruvic Acid | 6.4 | 2.6 - 32 | Succinic Acid | 9.38 | 0 - 16 |

### DISORDERS OF PURINE/PYRIMIDINE METABOLISM

| Analyte | Result | BRI µmol/mmol Creatinine | Analyte | Result | BRI µmol/mmol Creatinine |
|---|---|---|---|---|---|
| 2,8 Dihydroxyadenine | 0 | 0 - 0.001 | 5- Hydroxy Methyl Uracil | 0 | 0 - 0.0121 |
| Adenosine | 0 | 0 - 1 | Beta-Ureidopropionic Acid | 0.58 | 0 - 4.8 |
| Deoxyadenosine | 0 | 0 - 0.5 | Dihydro Thymine | 0 | 0 - 10 |
| Dihydro Uracil | 0 | 0 - 16.5 | Guanosine | 0 | 0 - 0.23 |
| Hypoxanthine | 3.06 | 0 - 24.1 | Inosine | 0 | 0 - 3.1 |
| Orotic Acid | 0 | 0 - 1 | Pseudouridine | 4.82 | 0 - 47.3 |
| Thymine | 0 | 0 - 1.7 | Uracil | 0.52 | 0 - 6.9 |
| Uric Acid | 48.9 | 24 - 398 | Uridine | 0.1 | 0 - 1.351 |
| Xanthine | 1.5 | 0 - 3.75 | | | |

### PEROXISOMAL DISORDERS

| Analyte | Result | BRI µmol/mmol Creatinine | Analyte | Result | BRI µmol/mmol Creatinine |
|---|---|---|---|---|---|
| 2,6 Dimethyloctanedioic Acid | 0 | 0 - 0.001 | 3-Methyl Adipic Acid | 0 | 0 - 5.6 |
| Adipic Acid | 1.15 | 0 - 2.8 | Malic Acid | 0.94 | 0 - 5.3 |
| Oxalic Acid | 5.13 | 0 - 78 | Sebasic Acid | 0 | 0 - 0.24 |
| Suberic Acid | 0.81 | 0 - 2.1 | | | |

Low Normal Borderline High

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## 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 | 1.15 | 0 - 2.8 |
| Quinolinic Acid | 2.11 | 0-9.1 |
| Vanillyl Mandelic Acid | 0.95 | 0 - 3.6 |

| Analyte | Result | BRI µmol/mmol Creatinine |
|---|---|---|
| 4- Hydroxy Mandelic Acid | 1.03 | 0.91 - 1.7 |
| Homovanillic Acid | 1.33 | 0 - 5.3 |
| Sebasic Acid | 0 | 0 - 0.24 |

Low Normal Borderline High

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## 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 ---**