

|                    |                      |              |                 |
|--------------------|----------------------|--------------|-----------------|
| Name               | : Master. KETAN BALU | Barcode No   | : 23586704      |
| Age /Gender        | : 13M/Male           | Reg. No      | : 0352301190060 |
| Referring by       | :                    | SPP Code     | : SPL-PU-011    |
| Referring Customer | :                    | Collected On | : 19-01-2023    |
| Primary Sample     | : Urine              | Received On  | : 20-01-2023    |
| Sample Tested In   | : Urine              | Reported On  | : 24-01-2023    |

**ORGANIC ACIDS, URINE @**  
(GCMS)

**Impression:-** Elevated levels of 3-Hydroxybutyric acid, Acetoacetic acid suggestive of mild Ketosis.

**Comment**

Organic acids are intermediate metabolites of all major groups of organic cellular components: aminoacids, lipids carbohydrates, purines and pyrimidines. Organic acidurias are biochemically heterogeneous group of inborn errors of metabolism biochemically characterized by accumulation of either pathological amounts of normal metabolites or metabolites which are normally not present in physiological conditions but are formed as result of activation of alternate pathways. The incidence of individual organic acid metabolism varies from 1:10,000 to 1:1,000,000 live births. All possible disease entities included, the incidence of conditions in which informative organic acid profiles could be detected in urine is likely to approach to 1:1000 live birth. Important organic acidurias include Propionic aciduria, Methylmalonic aciduria, Isovaleric aciduria, Maple syrup urine disease (MSUD), Glutaric aciduria type 1 and Multiple carboxylase deficiency.

| S.No. | Compound Name                  | Results | Cut off(%) |
|-------|--------------------------------|---------|------------|
| 1     | Lactic acid                    | 2.52    | 6.70       |
| 2     | 2-Hydroxyisobutyric acid       | 0.02    | 0.50       |
| 3     | Caproic acid                   | 0.27    | 0.50       |
| 4     | Glycolic acid                  | 1.26    | 3.99       |
| 5     | Oxalic acid                    | 0.21    | 1.00       |
| 6     | 2-Hydroxybutyric acid          | 0.05    | 0.50       |
| 7     | Glyoxylic acid-oxime           | 0.38    | 15.17      |
| 8     | 3-Hydroxypropionic acid        | 0.90    | 1.95       |
| 9     | Pyruvic acid-oxime             | 7.37    | 32.61      |
| 10    | Valproic acid                  | 0.10    | 0.50       |
| 11    | 3-Hydroxybutyric acid          | 1.20    | 5.28       |
| 12    | 3-Hydroxyisobutyric acid       | 0.05    | 13.02      |
| 13    | 2-Hydroxyisovaleric acid       | 0.05    | 0.50       |
| 14    | 2-Methyl-3-hydroxybutyric acid | 1.38    | 7.65       |
| 15    | Malonic acid                   | 0.05    | 0.50       |
| 16    | 3-Hydroxyisovaleric acid       | 1.88    | 6.10       |
| 17    | 2-Keto-isovaleric acid-oxime   | 0.05    | 0.50       |

| S.No. | Compound Name                  | Results | Cut off(%) |
|-------|--------------------------------|---------|------------|
| 18    | Methylmalonic acid             | 0.05    | 5.34       |
| 19    | Ethylhydracrylic acid          | 4.03    | 6.22       |
| 20    | Urea                           | 0.05    | 968.40     |
| 21    | 4-Hydroxybutyric acid          | 0.05    | 0.50       |
| 22    | 2-Hydroxyisocaproic acid       | 0.05    | 0.50       |
| 23    | 3-Hydroxyvaleric acid          | 0.05    | 0.50       |
| 24    | Acetoacetic acid               | 0.05    | 0.10       |
| 25    | 2-Hydroxy-3-Methylvaleric acid | 0.10    | 0.50       |
| 26    | Benzoic acid                   | 0.05    | 29.13      |
| 27    | Acetoacetic Acid               | 0.05    | 0.10       |
| 28    | Octanoic acid                  | 0.05    | 0.59       |
| 29    | 2-Propylhydroxyglutaric acid   | 0.05    | 0.50       |
| 30    | 2-Methyl-3-hydroxyvaleric acid | 0.05    | 0.50       |
| 31    | Glycerol                       | 0.05    | 2.72       |
| 32    | Phosphoric acid                | 12.85   | 72.70      |
| 33    | 2-Methyl-3-hydroxyvaleric acid | 0.42    | 0.50       |
| 34    | Ethylmalonic acid              | 0.05    | 7.45       |

| S.No. | Compound Name                     | Results | Cut off(%) |
|-------|-----------------------------------|---------|------------|
| 35    | 2-Ketoisocaproic acid             | 0.05    | 0.73       |
| 36    | Acetylglycine                     | 0.21    | 0.50       |
| 37    | Phenylacetic acid                 | 0.05    | 0.62       |
| 38    | Maleic acid                       | 0.45    | 0.55       |
| 39    | Succinic acid                     | 2.07    | 129.90     |
| 40    | Methylsuccinic acid               | 1.37    | 8.62       |
| 41    | Glyceric acid                     | 2.31    | 2.93       |
| 42    | Uracil                            | 0.84    | 19.65      |
| 43    | Fumaric acid                      | 0.05    | 10.36      |
| 44    | Propionylglycine                  | 0.05    | 0.50       |
| 45    | Acetylglycine                     | 0.05    | 0.50       |
| 46    | Mevalonolactone                   | 0.05    | 0.50       |
| 47    | Mevalonic lactone                 | 0.05    | 0.50       |
| 48    | Isobutyrylglycine                 | 0.05    | 0.50       |
| 49    | 2-propyl-3-hydroxy-pentanoic acid | 0.14    | 0.50       |
| 50    | Mesaconic acid                    | 0.95    | 11.36      |
| 51    | Glutaric acid                     | 0.90    | 8.44       |

| S.No. | Compound Name                               | Results | Cut off(%) |
|-------|---------------------------------------------|---------|------------|
| 52    | 3-Methylglutaconic acid                     | 0.05    | 0.50       |
| 53    | 3-Methylglutaric acid                       | 0.54    | 10.48      |
| 54    | 2-Propyl-3-ketopentanoic acid               | 0.05    | 0.50       |
| 55    | Propionylglycine1                           | 0.05    | 0.50       |
| 56    | Isobutyrylglycine                           | 0.05    | 0.50       |
| 57    | 2-Deoxytetronic acid                        | 0.05    | 12.88      |
| 58    | Butyrylglycine                              | 0.05    | 0.50       |
| 59    | 3-Methylglutaconic acid(E)                  | 4.22    | 5.12       |
| 60    | Glutaconic acid                             | 0.02    | 0.50       |
| 61    | Succinylacetone                             | 0.05    | 0.50       |
| 62    | Trimethylsilyl(TMS) decanoate               | 0.05    | 0.50       |
| 63    | 2-Propyl-5-OH-pentanoic                     | 0.05    | 0.50       |
| 64    | 2-Pentenedioic acid 3-methyl bis(TMS) ester | 4.22    | 14.15      |
| 65    | Isovaleroglycine TMS ester                  | 0.05    | 0.69       |
| 66    | Butyrylglycine                              | 0.05    | 0.50       |
| 67    | Malic acid                                  | 0.05    | 1.20       |
| 68    | Adipic acid                                 | 5.01    | 13.00      |

| S.No. | Compound Name                   | Results | Cut off(%) |
|-------|---------------------------------|---------|------------|
| 69    | Isovalerylglycine               | 0.05    | 0.50       |
| 70    | 2-Hexenedioic acid              | 0.05    | 19.55      |
| 71    | 5-Oxoproline                    | 1.12    | 10.48      |
| 72    | 3-Methyladipic acid             | 1.07    | 31.21      |
| 73    | Thiodiglycolic acid             | 0.05    | 0.10       |
| 74    | 2-Propyl-glutaric acid          | 0.07    | 0.10       |
| 75    | 7-Hydroxooctanoic acid          | 0.05    | 0.06       |
| 76    | 5-Hydroxy-2-furoic acid         | 3.55    | 9.02       |
| 77    | Tiglylglycine- 2TMS             | 0.05    | 0.50       |
| 78    | 3-Methylcrotonoylglycine        | 0.05    | 1.05       |
| 79    | Tiglylglycine-1TMS              | 0.05    | 0.53       |
| 80    | 3-Methylcrotonoylglycine        | 0.05    | 0.52       |
| 81    | 2-Hydroxyglutaric acid          | 3.04    | 9.49       |
| 82    | 3-Hydroxyglutaric acid          | 0.32    | 0.50       |
| 83    | Phenylactic acid                | 0.05    | 5.79       |
| 84    | Pimelic acid                    | 0.90    | 15.16      |
| 85    | 3-Hydroxy-3-methylglutaric acid | 10.58   | 30.52      |

| S.No. | Compound Name                   | Results | Cut off(%) |
|-------|---------------------------------|---------|------------|
| 86    | 3-Hydroxyphenylacetic acid      | 0.56    | 1.94       |
| 87    | 2-Ketoglutaric acid-oxime       | 0.05    | 169.10     |
| 88    | 4-Hydroxybenzoic acid           | 2.91    | 22.38      |
| 89    | 4-Hydroxyphenylacetic acid      | 0.56    | 139.99     |
| 90    | 2-Ketoglutaric acid-oxime       | 0.05    | 28.54      |
| 91    | Hexanoylglycine                 | 0.05    | 0.50       |
| 92    | Phenylpyruvic acid-oxime        | 0.05    | 0.50       |
| 93    | N-Acetylaspartic acid           | 0.26    | 7.84       |
| 94    | 2-Hydroxyadipic acid            | 0.58    | 3.34       |
| 95    | Octenedioic acid                | 1.51    | 8.37       |
| 96    | 3-Hydroxyadipic acid            | 1.63    | 16.35      |
| 97    | Suberic acid                    | 6.60    | 11.51      |
| 98    | 3-Methylglutaconic acid         | 0.05    | 0.50       |
| 99    | 2-Keto-adipic                   | 0.22    | 11.43      |
| 100   | Aconitic acid                   | 9.30    | 304.07     |
| 101   | Orotic acid                     | 0.05    | 2.50       |
| 102   | 3-Methoxy-4-hydroxybenzoic acid | 0.05    | 5.79       |

| S.No. | Compound Name                               | Results | Cut off(%) |
|-------|---------------------------------------------|---------|------------|
| 103   | Homovanillic acid                           | 10.24   | 42.35      |
| 104   | Azelaic acid                                | 0.56    | 21.63      |
| 105   | Hippuric acid- 2TMS                         | 0.05    | 21.54      |
| 106   | Isocitric acid                              | 5.42    | 62.91      |
| 107   | Citric acid                                 | 4.36    | 978.29     |
| 108   | Homogentisic Acid                           | 0.05    | 0.99       |
| 109   | Hippuric acid-1TMS                          | 85.35   | 336.99     |
| 110   | Methylcitric acid(1)                        | 0.05    | 1.81       |
| 111   | 3-(3-Hydroxyphenyl)-3-hydroxypropionic acid | 0.05    | 4.99       |
| 112   | Methylcitric acid(2)                        | 0.05    | 1.49       |
| 113   | 3-Hydroxyoctenedioic acid                   | 0.05    | 11.68      |
| 114   | 3-Hydroxysuberic acid                       | 0.05    | 8.95       |
| 115   | Vanillylmandelic acid                       | 28.86   | 156.72     |
| 116   | Sebacic acid                                | 0.05    | 11.52      |
| 117   | Decadienedioic acid                         | 0.34    | 4.64       |
| 118   | 4-Hydroxyphenyllactic acid                  | 10.20   | 12.51      |
| 119   | 4-Hydroxyphenylpyruvic acid-oxime           | 1.78    | 1.90       |

| S.No. | Compound Name                  | Results | Cut off(%) |
|-------|--------------------------------|---------|------------|
| 120   | 2-Hydroxyhippuric acid         | 1.78    | 20.66      |
| 121   | 3- Indoleacetic acid           | 1.33    | 149.51     |
| 122   | Suberylglycine                 | 0.05    | 0.50       |
| 123   | Palmitic acid                  | 10.52   | 23.34      |
| 124   | 2-Hydroxysebacic acid          | 0.76    | 10.34      |
| 125   | 3-Hydroxysebacic acid          | 5.79    | 15.60      |
| 126   | 2-Hydroxyhippuric acid         | 1.34    | 20.66      |
| 127   | Dodecanedioic acid             | 0.05    | 0.50       |
| 128   | N-Acetyltyrosine               | 0.05    | 0.10       |
| 129   | Uric acid                      | 0.05    | 21.49      |
| 130   | 3-6-Epoxydodecanedioic acid    | 0.52    | 8.58       |
| 131   | 3-hydroxydodecanedioic acid    | 1.20    | 2.25       |
| 132   | 3-6-Epoxytetradecanedioic acid | 0.05    | 6.17       |

#### Disorders detected by estimating organic acid in Urine

| S.No.                                                    | Disorder Name                   | Enzyme/Transport defect   |
|----------------------------------------------------------|---------------------------------|---------------------------|
| Disorders Of Propionate metabolism and Related Cofactors |                                 |                           |
| 1                                                        | Propionic Acidemia              | Propionic CoA Carboxylase |
| 2                                                        | Multiple Carboxylase Deficiency | Holocarboxylase synthase  |

| S.No.                                             | Compound Name                        | Results                                                                              | Cut off(%) |
|---------------------------------------------------|--------------------------------------|--------------------------------------------------------------------------------------|------------|
| 3                                                 | Methylmalonic Acidemia (MMA)         | MethylmalonylCoA mutase, Mitochondrial Cobalamin reductase, Cob(i)alamin transferase |            |
| 4                                                 | Methylmalonic Acidemia (MMA)         | MethylmalonylCoA isomerase                                                           |            |
| 5                                                 | Methylmalonic Acidemia (MMA)         | Lysosomal release                                                                    |            |
| Disorders of Branched Chain Amino Acid Metabolism |                                      |                                                                                      |            |
| 6                                                 | MSUD                                 | Branched chain ketoacid dehydrogenase complex                                        |            |
| 7                                                 | Isovaleric Acidemia                  | Isovaleryl CoA dehydrogenase                                                         |            |
| 8                                                 | 3-Methylcrotonyl glycinuria          | 3-Methylcrotonyl CoA carboxylase                                                     |            |
| 9                                                 | 3-Methylglutaconic aciduria          | 3-Methylglutaconyl CoA hydratase                                                     |            |
| 10                                                | 3-Hydroxy -3-methylglutaric aciduria | 3-Hydroxy -3-methylglutaryl CoA Lyase                                                |            |
| 11                                                | 2-Methylbutyrylglycinuria            | 2-Methylbutyryl-CoA dehydrogenase                                                    |            |
| 12                                                | 2-Methyl-3-hydroxybutyric aciduria   | 2-Methyl-3-hydroxybutyryl CoA                                                        |            |
| 13                                                | Beta ketothiolase deficiency         | Beta ketothiolase                                                                    |            |
| 14                                                | Isobutyrylglycinuria                 | Isobutyryl CoA dehydrogenase                                                         |            |
| 15                                                | 3-Hydroxyisobutyric acidurias        | 3-Hydroxyisobutyryl CoA dehydrogenase or methylmalonic semialdehyde dehydrogenase    |            |
| Other Amino acid metabolism disorders             |                                      |                                                                                      |            |
| 16                                                | Alkaptonuria                         | Homogentisic dioxygenase                                                             |            |
| 17                                                | Classical Phenylketonuria            | Phenylalanine Hydroxylase                                                            |            |
| 18                                                | Tyrosinemia Type I                   | Fumarylacetoacetase                                                                  |            |
| 19                                                | Tyrosinemia Type II                  | Tyrosine aminotransferase                                                            |            |
| 20                                                | Tyrosinemia Type III                 | 4 Hydroxyphenylpyruvate dioxygenase                                                  |            |
| 21                                                | Glutaric Acidemia Type 1             | Glutaryl CoA dehydrogenase                                                           |            |

| S.No.                          | Compound Name                  | Results                                                    | Cut off(%) |
|--------------------------------|--------------------------------|------------------------------------------------------------|------------|
| 22                             | 2-Ketoadipic aciduria          | 2-ketoadipic dehydrogenase                                 |            |
| urea Cycle Disorders           |                                |                                                            |            |
| 23                             | OTC Deficiency (X-linked)      | Ornithine transcarbamylase                                 |            |
| 24                             | Citrullinemia Type I           | Argininosuccinate synthetase                               |            |
| 25                             | Citrullinemia Type II          | Citrin deficiency                                          |            |
| Fatty Acid oxidation disorders |                                |                                                            |            |
| 26                             | LCHAD                          | Long chain acyl CoA dehydrogenase deficiency               |            |
| 27                             | TFP deficiency                 | Trifunctional Protein                                      |            |
| 28                             | Glutaric acidemia type II      | Electron transport flavoprotein (ETF) ubiquinone reductase |            |
| 29                             | MCAD                           | Medium chain acyl CoA dehydrogenase                        |            |
| 30                             | MCKAT                          | Medium chain 3-ketoacyl CoA thiolase                       |            |
| 31                             | SCAD                           | Short chain acyl CoA dehydrogenase                         |            |
| 32                             | SCHAD                          | Short chain 3-hydroxy acyl CoA dehydrogenase               |            |
| 33                             | Dicarboxylic aciduria          | Enzyme defect in beta oxidation of 6-10 carbon fatty acid  |            |
| 34                             | 3-hydroxydicarboxylic aciduria | Long chain 3 hydroxy acyl CoA dehydrogenase deficiency     |            |
| Other Miscellaneous Disorders  |                                |                                                            |            |
| 35                             | Biotinidase Deficiency         | Biotinidase                                                |            |
| 36                             | 4-OH butyric aciduria          | Succinic semialdehyde dehydrogenase                        |            |
| 37                             | Canavan disease                | N-aspartoacylase                                           |            |
| 38                             | Ethylmalonic encephalopathy    | Unknown (ETHE1 gene)                                       |            |
| 39                             | Glyceroluria (X-linked)        | Glycerol kinase                                            |            |
| 40                             | Hyperoxaluria Type 1           | Alanine:glyoxalate aminotransferase                        |            |
| 41                             | Hyperoxaluria Type 2           | D-glyceric dehydrogenase                                   |            |
| 42                             | Malonic acidemia               | Malonyl CoA decarboxylase                                  |            |
| 43                             | Mevalonic aciduria             | Mevalonate kinase                                          |            |

| S.No. | Compound Name                  | Results                          | Cut off(%) |
|-------|--------------------------------|----------------------------------|------------|
| 44    | 5-Oxoprolinuria                | Glutathione synthase             |            |
| 45    | Zellweger Syndrome             | Peroxisomal disorder             |            |
| 46    | 2-Hydroxy glutaric<br>acidemia | 2-hydroxyglutarate dehydrogenase |            |
| 47    | Glyceric acidemia              | Glycerate kinase                 |            |

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