



Name	: Baby DUMMY-1	Collected	: 8/9/2016 6:46:00PM
Lab No.	: 106120073	Received	: 8/9/2016 7:06:45PM
Age:	5 Years	Reported	: 27/10/2016 12:40:00PM
Gender:	Female	Report Status	: Final
A/c Status	: P	Ref By	: SELF

- | Test Name  | Results | Units | Bio. Ref. Interval |
|--|---------|-------|--------------------|
| 2. False positive results are seen in patients administered amino acid & carnitine supplements.  |         |       |                    |
| 3. Results should be clinically correlated. Any infant with clinical features suggestive of Inborn Errors of Metabolism must be investigated regardless of the result. |         |       |                    |
| 4. Test conducted on heel / finger prick blood.  |         |       |                    |
| 5. Genetic counseling available with prior appointment at National Reference Laboratory, New Delhi.  |         |       |                    |

#### Disorders Tested by Tandem Mass Spectrometry

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| <ul style="list-style-type: none"> <li>• Urea Cycle Disorders</li> <li>• Homocystinuria</li> <li>• Hypermethioninemia</li> <li>• Non-ketotic hyperglycinemia</li> <li>• Hyperornithinemia -Hyperammonemia- Homocitrullinuria (HHH)</li> <li>• Maple Syrup Urine Disease (MSUD)</li> <li>• Hypervalinemia</li> <li>• PKU/ Hyperphenylalaninemia</li> <li>• Tyrosinemia</li> <li>• Cobalmin defect</li> <li>• Lactic acidosis</li> <li>• Glutaric acidemia type 1</li> <li>• Isovaleric Acidemia</li> <li>• 3-Methyl crotonyl CoA carboxylase deficiency (3MCC)</li> <li>• 3-Methylglutaconyl CoA hydratase deficiency (3MGA)</li> <li>• 2-Methyl 3 Hydroxybutyryl CoA Dehydrogenase Deficiency (2MBDH)</li> <li>• 2-Methylbutyryl CoA Dehydrogenase Deficiency (2MBCD)</li> <li>• 2,4 Dienyl CoA Reductase Deficiency</li> <li>• 3 Hydroxy 3 Methylglutaryl CoA Lyase Deficiency (HMG)</li> </ul> | <ul style="list-style-type: none"> <li>• Multiple CoA Carboxylase deficiency</li> <li>• Mitochondrial Acetoacetyl CoA Thiolase (Ketothiolase) deficiency</li> <li>• Methylmalonic Acidemia</li> <li>• Methylmalonic acidemia with homocystinuria</li> <li>• Propionic Acidemia</li> <li>• Short chain acyl CoA Dehydrogenase deficiency (SCAD)</li> <li>• Medium chain acyl CoA Dehydrogenase deficiency (MCAD)</li> <li>• Mitochondrial Trifunctional protein deficiency</li> <li>• Longchain 3-Hydroxy acyl CoA Dehydrogenase deficiency (LCHAD)</li> <li>• Very long Chain Acyl CoA dehydrogenase deficiency (VLCAD)</li> <li>• Carnitine Transport Defect</li> <li>• Carnitine Palmitoyl Transferase Deficiency type 1 (CPT 1)</li> <li>• Carnitine Palmitoyl Transferase Deficiency type 2 (CPT 2)</li> <li>• Glutaric Acidemia Type 2</li> <li>• Carnitine Acylcarnitine Translocase Deficiency</li> </ul> |
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Dr. Manjeet Kaur  
PhD  
HOD Genetics

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Name	: Master DUMMY-1	Collected	: 8/9/2016 6:17:00PM
Lab No.	: 106120058	Received	: 8/9/2016 6:36:42PM
Age:	5 Years	Reported	: 26/9/2016 6:27:56PM
Gender:	Male	Report Status	: Final
A/c Status	: P	Ref By	: SELF

- | Test Name  | Results | Units | Bio. Ref. Interval |
|--|---------|-------|--------------------|
| 3. Results should be clinically correlated. Any infant with clinical features suggestive of Inborn Errors of Metabolism must be investigated regardless of the result. |         |       |                    |
| 4. Test conducted on heel / finger prick blood.  |         |       |                    |
| 5. Genetic counseling available with prior appointment at National Reference Laboratory, New Delhi.  |         |       |                    |

#### Disorders Tested by Tandem Mass Spectrometry

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