METASCREEN TEST REPORT

Name:

Address:

Child's Name:
Birth Type:
Child Order:

Attending Doctor:

Your child's urinary specimen has been tested. The details of your newborn's urinary testing are shown below.

Date of Birth
Newborn Screening Panel
Specimen Collection Date
Specimen Laboratory Receipt Date
Report Date

Metabolic Screening Test Results:

Amino acid and organic acid metabolism disorders	No abnormality detected
Sugar metabolism disorders	No abnormality detected
Fatty acid metabolism disorders	No abnormality detected
Peroxisomal disorders	No abnormality detected
Purine & pyrimidine metabolism disorders	No abnormality detected
Lactic acid, hyperpyruvic acid metabolic disorders	No abnormality detected
Other inborn errors of metabolism in the subscribed panel	No abnormality detected

Comments:

Revision Remarks:

Dr.
This is a computer generated report, no signature is required.

Enclosed:
1. List of diseases covered in the tested panel

Remarks:

1. This test is intended for 6 month-old baby or younger. Accuracy of results for babies more than 6-months of age is not guaranteed. The clinician is requested to correlate this report with other clinical findings and medical history of patient. Since some metabolites may be found to be increased in several conditions other than the disorders screened for, such as in case of administered IV fluids, or if the patient is on certain medicine, it is important to take into considerations such conditions while interpreting the result.

2. All dates displayed on this page are in the format DD/MM/YYYY.

Meta100+ Panel

Amino acid and organic acid metabolism disorders

1. Propionic aciduria
2. Holocarboxylase synthetase deficiency
3. Methylmalonic aciduria (Cbl C & Cbl D)
4. Methylmalonic aciduria
5. Methylmalonic aciduria (Cbl A & Cbl B)
6. Malonic aciduria
7. Isobutyryl-CoA dehydrogenase deficiency
8. 2-methylbutyryl-CoA dehydrogenase deficiency
9. Methylmalonic semialdehyde dehydrogenase deficiency
10. Beta-ketothiolase deficiency
11. Isovaleric aciduria
12. 3-methylcrotonylglycinuria
13. 3-methylglutaconic aciduria (Type I – hydratase deficiency)
14. Barth Syndrome
15. 3-hydroxy-3-methylglutaric aciduria
16. Glutaric aciduria type II
17. Glutaric aciduria type I
18. Mevalonate kinase deficiency
19. Glyceroluria
20. Phenylketonuria (phenylalanine hydroxylase deficiency)
21. Hyperphenylalaninuria (variant, benign)
22. 2-Methyl 3-hydroxy butyric aciduria
23. Tyrosinuria type I (hepatorenal tyrosinuria)
24. Tyrosinuria type II (oculocutaneous tyrosinuria)
25. Tyrosinuria type III (4-hydroxyphenylpyruvate dioxygenase deficiency)
26. Transient tyrosinuria in the newborn
27. Tyrosinuria caused by a liver disease
28. Maple syrup urine disease
29. N-acetylglutamate synthase deficiency
30. Carbamylphosphate synthetase deficiency
31. Ornithine transcarbamylase deficiency
32. Citrullinuria (argninosuccinate synthase deficiency)
33. Citrullinuria type II (citrin deficiency)
34. Argininosuccinic aciduria
35. Argininuria
36. Hypermethioninuria (MAT I/III deficiency)
37. Homocystinuria cystathionine beta-synthase deficiency
38. Alkaptonuria
39. Tada Syndrome
40. Encephalopathy due to hydroxykynureninuria
41. Valinuria
42. Hyperleucine- isoleucinuria
43. Dihydrolipoyl dehydrogenase (E3) deficiency
44. Beta-hydroxyisobutyryl CoA deacylase deficiency
45. Histidinuria
46. Hartnup Syndrome
47. Lysinuric protein intolerance
48. Alpha-ketoadipic aciduria
49. Saccharopinuria
50. Seizures-intellectual deficit due to hydroxylysinuria
51. Cystathinionuria
52. Hyperprolinuria type I
53. Hyperprolinuria type II
54. Hyper hydroxyprolinuria
55. 2-hyroxy glutaric aciduria
56. Hawkinsinuria
57. Biotinidase deficiency
58. Fumarate hydratase deficiency
59. Hyperornithinuria-hyperammoninuria-hyperhomocitrullinuria syndrome

Sugar metabolism disorders

60. Classic galactosuria
61. Galactokinase deficiency
62. Galactose epimerase deficiency
63. Transient galactosuria
64. D-glyceric aciduria
65. Fructose-1,6-diphosphatase deficiency
66. Endogeneous sucrosuria
67. Lactose intolerance

Fatty acid metabolism disorders

68. Short-chain acyl-CoA dehydrogenase deficiency
69. Medium-chain acyl-CoA dehydrogenase deficiency
70. Long-chain 3-OH acyl-CoA dehydrogenase deficiency
71. Medium/short-chain L-3-OH acyl-CoA DH deficiency
72. Ethylmalonic encephalopathy
73. Dicarboxylic aciduria

Peroxisomal disorders

74. Zellweger syndrome
75. Neonatal adrenoleukodystrophy
76. Infantile refsum disease
77. Zellweger-like syndrome
78. Primary hyperoxaluria

Purine & pyrimidine metabolism disorders

79. Adenosine deaminase deficiency
80. Lesch-Nyhan syndrome
81. Kelley-Seegmiller syndrome
82. Adenine phosphoribosyl transferase deficiency
83. Hereditary xanthinuria
84. Orotic aciduria
85. Dihydropyrimidine dehydrogenase deficiency
86. Dihydropyrimidinase deficiency
87. Beta-ureido propionase deficeincy

Lactic acid, hyperpyruvic acid metabolism disorders

88. Pyruvate dehydrogenase (E1) deficiency
89. Pyruvate dehydrogenase phosphatase deficiency
90. Pyruvate carboxylase deficiency
91. Pyruvate decarboxylase deficiency
92. Leigh syndrome
93. Cytochrome C oxidase deficiency
94. De Toni-Fanconi-Debre Syndrome

Other inborn errors of metabolism

95. Hyperglycinuria
96. Sarcosinuria
97. Imidazole aminoaciduria
98. Formiminoglutamic aciduria
99. Carnosinuria
100. Canavan disease
101. Glutathione synthetase deficiency
102. Gamma-glutamyl transpeptidase deficiency
103. Succinic semialdehyde dehydrogenase deficiency
104. Hyperpipecolaturia
105. Neonatal intrahepatic cholestasis caused by citrin deficiency
106. Beta-aminoisobutyric aciduria