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AMPATH
ACCURATE DIAGNOSTICS. AMERICAN STANDARDS.
In collaboration with
UPMC LIFE CHANGING MEDICINE

LABORATORY REPORT

PATIENT INFORMATION
Baby SIDRA FATIMA .

AGE	: 1Y 4m
GENDER	: Female
PRIORITY	: Routine
OP / IP / DG #	:

REFERRED BY
DR.SHALINE PHE
LIFETIME CARE LABS
Lab MR #: 5702705

SPECIMEN INFORMATION

SAMPLE TYPE	: **Urine**
ORDER REQ. NO	: OREQ-LAB-22-3006861
LAB ORDER. NO	: 2215177170
COLLECTED ON	: 11-May-2022 12:00
RECEIVED ON	: 11-May-2022 18:47
REPORT STATUS	: **Completed**

BIOCHEMISTRY

Test Name (Methodology)	Result	Flag	Units	Biological Reference Interval
Organic Acid Screen - Quantitative - Urine				
Organic Acid Screen - Qualitative - Urine	.			

Dr. Mohammad Ibrahim Shaik
Consultant
16-May-2022 20:20

Checked by Mr. Vinod Kumar Mahendrekar
Senior Lab Technician

-----Analytical Report Attached-----

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 pyramidines. 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	1.29	6.70
2	2-Hydroxyisobutyric acid	0.11	0.50
3	Caproic acid	0.27	0.50
4	Glycolic acid	1.26	3.99
5	Oxalic acid	**1.53**	1.00
6	2-Hydroxybutyric acid	0.00	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	**27.52**	0.50
11	3-Hydroxybutyric acid	**262.37**	5.28
12	3-Hydroxyisobutyric acid	0.00	13.02
13	2-Hydroxyisovaleric acid	0.00	0.50
14	2-Methyl-3-hydroxybutyric acid	1.38	7.65
15	Malonic acid	0.00	0.50
16	3-Hydroxyisovaleric acid	**7.15**	6.10
17	2-Keto-isovaleric acid-oxime	0.00	0.50

S.No.	Compound Name	Results	Cut off(%)
18	Methylmalonic acid	0.00	5.34
19	Ethylhydracrylic acid	4.03	6.22
20	Urea	0.00	968.40
21	4-Hydroxybutyric acid	0.00	0.50
22	2-Hydroxyisocaproic acid	0.00	0.50
23	3-Hydroxyvaleric acid	0.00	0.50
24	Acetoacetic acid	0.00	0.10
25	2-Hydroxy-3-Methylvaleric acid	**0.77**	0.50
26	Benzoic acid	0.00	29.13
27	Acetoacetic Acid	**1.67**	0.10
28	Octanoic acid	0.00	0.59
29	2-Propylhydroxyglutaric acid	0.00	0.50
30	2-Methyl-3-hydroxyvaleric acid	0.00	0.50
31	Glycerol	0.00	2.72
32	Phosphoric acid	**120.63**	72.70
33	2-Methyl-3-hydroxyvaleric acid	0.42	0.50
34	Ethylmalonic acid	0.00	7.45

S.No.	Compound Name	Results	Cut off(%)
35	2-Ketoisocaproic acid	0.00	0.73
36	Acetylglycine	0.21	0.50
37	Phenylacetic acid	0.00	0.62
38	Maleic acid	**1.16**	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	0.00	10.36
44	Propionylglycine	0.00	0.50
45	Acetylglycine	0.00	0.50
46	Mevalonolactone	0.00	0.50
47	Mevalonic lactone	0.00	0.50
48	Isobutyrylglycine	0.00	0.50
49	2-propyl-3-hydroxy-pentanoic acid	**0.54**	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	0.00	0.50
53	3-Methylglutaric acid	0.54	10.48
54	2-Propyl-3-ketopentanoic acid	0.00	0.50
55	Propionylglycine1	0.00	0.50
56	Isobutyrylglycine	0.00	0.50
57	2-Deoxytetronic acid	0.00	12.88
58	Butyrylglycine	0.00	0.50
59	3-Methylglutaconic acid(E)	4.22	5.12
60	Glutaconic acid	**0.95**	0.50
61	Succinylacetone	0.00	0.50
62	Trimethylsilyl(TMS) decanoate	0.00	0.50
63	2-Propyl-5-OH-pentanoic	0.00	0.50
64	2-Pentenedioic acid 3-methyl bis(TMS) ester	4.22	14.15
65	Isovaleroglycine TMS ester	0.00	0.69
66	Butyrylglycine	0.00	0.50
67	Malic acid	0.00	1.20
68	Adipic acid	**20.15**	13.00

S.No.	Compound Name	Results	Cut off(%)
69	Isovalerylglycine	0.00	0.50
70	2-Hexenedioic acid	0.00	19.55
71	5-Oxoproline	1.12	10.48
72	3-Methyladipic acid	1.07	31.21
73	Thiodiglycolic acid	**1.43**	0.10
74	2-Propyl-glutaric acid	**1.35**	0.10
75	7-Hydroxoctanoic acid	0.00	0.06
76	5-Hydroxy-2-furoic acid	3.55	9.02
77	Tiglylglycine- 2TMS	0.00	0.50
78	3-Methylcrotonoylglycine	0.00	1.05
79	Tiglylglycine-1TMS	0.00	0.53
80	3-Methylcrotonoylglycine	0.00	0.52
81	2-Hydroxyglutaric acid	3.04	9.49
82	3-Hydroxyglutaric acid	0.32	0.50
83	Phenyllactic acid	0.00	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	0.00	169.10
88	4-Hydroxybenzoic acid	2.91	22.38
89	4-Hydroxyphenylacetic acid	0.56	139.99
90	2-Ketoglutaric acid-oxime	0.00	28.54
91	Hexanoylglycine	0.00	0.50
92	Phenylpyruvic acid-oxime	0.00	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	0.00	0.50
99	2-Keto-adipic	0.22	11.43
100	Aconitic acid	9.30	304.07
101	Orotic acid	0.00	2.50
102	3-Methoxy-4-hydroxybenzoic acid	0.00	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	0.00	21.54
106	Isocitric acid	5.42	62.91
107	Citric acid	4.36	978.29
108	Homogentisic Acid	0.00	0.99
109	Hippuric acid-1TMS	85.35	336.99
110	Methylcitric acid(1)	0.00	1.81
111	3-(3-Hydroxyphenyl)-3-hydroxypropionic acid	0.00	4.99
112	Methylcitric acid(2)	0.00	1.49
113	3-Hydroxyoctenedioic acid	0.00	11.68
114	3-Hydroxysuberic acid	0.00	8.95
115	Vanillylmandelic acid	28.86	156.72
116	Sebacic acid	0.00	11.52
117	Decadienedioic acid	0.34	4.64
118	4-Hydroxyphenyllactic acid	**46.93**	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	0.00	0.50
123	Palmitic acid	**31.07**	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	0.00	0.50
128	N-Acetyltyrosine	0.00	0.10
129	Uric acid	0.00	21.49
130	3-6-Epoxydodecanedioic acid	0.52	8.58
131	3-hydroxydodecanedioic acid	**6.40**	2.25
132	3-6-Epoxytetradecanedioic acid	0.00	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-Methylgutaconic 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-hydroxybutyrl CoA	
13	Beta Ketothiolase deficiency	Beta Ketothiolase	
14	Isobutyrylglycinuria	Isobutyryl CoA dehydrogenase	
15	3-Hydroxyisobutyric acidurias	3-Hydroxyisobutyryl CoA dehydrogenase or methylmalonic semialdehyde dehydrgenase	
Other Amino acid metabolism disorders			
16	Alkaptonuria	Homogentisic dioxygenase	
17	Classical Phenylketonuria	Phenylalanine Hydroxylase	
18	Tyrosinemia Type 1	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 trancarbamylase	
24	Citrullinemia Type I	Argininosuccinate synthtase	
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 academia type II	Electron transport flavoprotein (ETF) ubiquinone reductase	
29	MCAD	Medium chain acyl CoA dehdrogenase	
30	MCKAT	Medium chain 3-ketoacyl CoA thiolase	
31	SCAD	Short chain acyl CoA dehydrogenase	
32	SCHAD	Short chain3-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	Glycerloluria (X-linked)	Glycerol kinase	
40	Hyperoxaluria Type 1	Alanine:glyoxalate aminotransferase	
41	Hyperoxaluria Type 2	D-glyceric dehdrogenase	
42	Malonic acidemia	Malonyl CoA decarboxylase	
43	Mevalonic aciduria	Mevalonate kinase	

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

----------End of report ----------