

**Metabolic Screening Laboratory  
Dr J.D. Shoemaker MD PhD  
Dept of Biochemistry  
1402 S Grand Ave  
St Louis, MO 63104  
(314)977-9230**

**Urine Organic Compounds**

Date: 4/16/09  
Time: 9:27 AM

To:  
SSM Cardinal Glennon Children's Medical Center  
1425 S Grand Ave  
St. Louis, MO 63104

Re:  
Name: Hawkinsinuria  
Clinic / Req #:

Date Collected:

Results: This sample contained 1.72 uM creatinine /ml  
and other peaks considered to be:

\_\_\_\_\_ Within Normal Limits

\_\_\_X\_\_\_ Abnormal

\_\_\_\_\_ Unidentified, Mass Spectra Available on Request

\_\_\_\_\_ Tentatively Identified

\_\_\_X\_\_\_ Positively Identified

Excretion of hawkinsin and 4-hydroxycyclohexylacetate indicates hawkinsinuria in this child with hemolytic anemia. Massive excretion of pyroglutamic acid is found. Pyroglutamic acidemia accounts for acidosis in this child.

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James D. Shoemaker MD Director

**Quantified Target Panel**

**Metabolic Screening Lab  
Dr J.D. Shoemaker MD PhD  
Urine Organic Compounds**

**St Louis University**

Patient Name                      Hawkinsinuria  
Data File                            OCM104 10/17/08

	mM/M		mM/M		mM/M
Organic Acids	Creatinine	Neurotransmitters	Creatinine	Acyl Glycines	Creatinine
Lactic	1365.98	GABA	1.36	Propionyl Glycine	0.00
Pyruvic	43.26	Homovanillic	15.56	Butyryl Glycine	0.00
Glycolic	333.54	Normetanephrine	0.32	Hexanoyl Glycine	0.80
Hexanoic	15.67	Vanillylmandelic	5.70	Phenylpropionyl Glycine	1.48
2-Hydroxybutyric	8.27	Metanephrine	2.02	Suberyl Glycine	62.36
3-Hydroxybutyric	58.86	5-HIAA	16.37	Isobutyryl Glycine	1.85
4-Hydroxybutyric	13.51	MHPG	3.36	Isovaleryl Glycine	0.00
Acetoacetic	1.23	Ethanolamine	7.60	Tiglyl Glycine	0.09
5-Hydroxycaproic	4.13	Phosphoethanolamine	0.79	Beta-Met Crotonyl Glycine	0.08
b-Lactate	327.45				
Octanoic	12.34	Nutritionals		Carbohydrates	
Succinic	1190.50				
Methylsuccinic	3.73	Kynurenine	6.08	Arabinitol	223.50
Glutaric	66.68	Formiminoglutamic	13.33	Dulcitol	17.94
a-Ketoglutarate	1059.48	4-Pyridoxic	2.95	Erythritol	140.37
Fumaric	756.11	Pantothenic	59.48	Inositol	76.68
Malic	1764.96	Xanthurenic	0.09	Mannitol	99.33
Adipic	57.83	Kynurenine	21.53	Ribitol	67.10
Suberic	27.45	Quinolinic	3.55	Sorbitol	107.69
Sebacic	14.00	Orotic	37.28	Threitol	46.14
Glyceric	1400.72	d-Aminolevulinic	144.53	Xylitol	37.25
Methylmalonic	10.71	3-Methylhistidine	45.95	Galactonic Acid	59.60
Ethylmalonic	16.76	1-Methylhistidine	14.73	Glucaric Acid	16.50
Homogentisic	6.27	Niacinamide	2.11	Gluconic Acid	136.73
Phenylacetic	20.83	Pseudouridine	138.67	Glucuronic Acid	41.36
Phenyllactic	0.87	2-Deoxytetrone	111.42	Arabinose	157.35
Phenylpyruvic	17.77	Xanthine	17.32	Fructose	142.82
p-Hydroxyphenylacetic	573.34	Hypoxanthine	28.45	Fucose	72.22
p-Hydroxyphenyllactic	78.47	Urocanic	2.84	Galactose	153.09
p-Hydroxyphenylpyruvic	89.09	Ascorbic	120.04	Glucose	1544.71
Succinylacetone	10.15	Glycerol	448.17	Isomaltose	4.83
3-Hydroxyisovaleric	93.09	Glycerol-3-Phosphate	12.74	Lactose	373.17
Phosphate	1477.01			Ribose	39.39
Citric	479.57			Sucrose	171.47
Isocitric	323.62			Xylose	77.78
Hippuric	465.14			Levoglucofan	57.64
Uric	542.76				
<b>Pyroglutamic</b>	<b>8094.46</b>			Amino Acids	
Uracil	8.03				
Thymine	0.07	Glycine	687.14	Lysine	49.77
Tartaric	120.52	Alanine	88.33	Histidine	144.74
Oxalic	138.70	Sarcosine	79.00	Threonine	109.57
7-Methylguanine	83.73	B-Alanine	5.58	Homoserine	0.68
2-Oxoadipic	24.04	B-Aminoisobutyric	22.88	Phosphoserine	4.52
		Serine	18.83	Methionine	3.95
Miscellaneous		Proline	115.44	Cysteine	562.41
		Hydroxyproline	3.80	Homocysteine	0.00
2-Amino adipic	13.89	Aspartic	14.26	Cystathionine	2.67
2-Hydroxy adipic	1.70	Asparagine	102.31	Cystine	0.00
2-Hydroxyglutaric	5.33	N-Acetyl Aspartic	5.63	Homocystine	0.30
2-Hydroxysebacic	10.68	N-Acetyl Cysteine	92.27	Phenylalanine	84.94
2-Methylcitric	36.12	N-Acetyl Tyrosine	26.43	Tyrosine	99.44
3-Hydroxyglutaric	92.65	Ornithine	1.83	Tryptophan	82.15
3-Hydroxy-3-methylglutaric	42.02	Glutamic	114.70		
3-Hydroxyisobutyric	56.60	Glutamine	578.83		
3-Hydroxysebacic	79.13	Pipecolic	4.60		
3-Methylglutaconic	36.93	Leucine	36.19		
3-Methylglutaric	5.01	Ketoleucine	2.34		
4-Hydroxycyclohexylacetic	3512.36	Valine	50.99		
Benzoic	20.16	Ketovaline	1.75		
cis-Aconitic	2.61	Isoleucine	22.47		
Sialic	110.54	Ketoisoleucine	1.26		

This Sample Contained 0.86 uMoles Creatinine/Sample

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Patient Name : Hawkinsinuria  
 Clinic/Ref# :  
 Data File : :\OCMD\Completed\OCM104.D  
 Acq On : 17 Oct 2008 16:46  
 Instrument ID : US61633459

Pk#	RT	Area%	Urine Constituents - Best fit from library	Ref#	Qual
1	3.61	0.09	1,4-Cyclohexadiene, 6-isopropenyl-2,4-dimethyl-1,3-	119715	12
2	3.70	0.40	Silanamine, N,N'-methanetetraylbis[1,1,1-trimethyl-	47703	64
3	4.01	0.07	3,8-Dioxa-2,9-disiladec-5-ene, 2,2,9,9-tetramethyl-	79474	10
4	4.11	0.29	1-Propene, 3-propoxy-	3770	16
5	4.33	0.78	LACTIC ACID DI-TMS	1499	53
6	5.11	0.03	4-Hexenoic acid, 4-methyl-6-(fluorodimethylsilyl)-6	109089	12
7	5.26	0.05	Decanoic acid, tert-butyl dimethylsilyl ester	115969	43
8	5.43	0.06	N,N-Diethyl-1,1,1-trimethylsilylamine	20621	10
9	5.60	2.32	LACTIC ACID DI-TMS	1499	87
10	5.89	0.53	LACTIC ACID DI-TMS	1499	50
11	6.16	0.13	13C3 Lactic Acid TMS - 5.47	187	42
12	6.53	0.49	SARCOSINE DI-TMS	1472	78
13	6.91	0.59	GLYCINE DI-TMS	52	80
14	7.15	0.14	ALPHA HYDROXYBUTYRIC ACID DI-TMS	97	72
15	7.34	0.10	ALANINE DI-TMS	79	91
16	7.41	0.05	Quinoline, 2-(1-methylethyl)-	37147	16
17	7.52	0.21	BETA-LACTATE DI-TMS	1640	64
18	7.99	0.37	4-HYDROXY BUTYRIC ACID DI-TMS	98	64
19	8.13	0.05	2-HYDROXY PENTANOIC ACID DI-TMS	141	83
20	8.31	0.04	BETA AMINO BUTYRIC ACID DI-TMS	90	58
21	8.52	0.03	1,1,1,3,5,5,7,7,7-Nonamethyl-3-(trimethylsiloxy)tet	166197	14
22	8.71	0.06	METHYLSUCCINIC ACID DI-TMS	172	53
23	8.87	0.04	3 METHYL 2-OXO BUTYRATE DI-TMS (ketovaline),10.591	1909	40
24	9.31	0.16	3-HYDROXY-ISOVALERIC, DI-TMS	137	53
25	9.46	0.06	METHYLMALONIC ACID DI-TMS	134	50
26	9.54	0.07	VALINE DI-TMS	127	91
27	9.89	0.07	Succinic Acid DI-TMS 12.01	82	50
28	10.41	0.03	2-METHYL ALANINE DI-TMS	89	45
29	10.71	0.03	Dipropylacetic acid, trimethylsilyl ester	68261	25
30	11.10	1.06	Urea, N,N'-bis(trimethylsilyl)-	60204	60
31	11.40	2.98	Glycerol-3-Phosphate TMS 22.90	71	72
32	11.93	0.11	PROLINE DI-TMS	121	59
33	12.41	6.03	4-HYDROXY BUTYRIC ACID DI-TMS	98	56
34	12.59	0.03	3,8-Dioxa-2,9-disiladec-5-ene, 2,2,9,9-tetramethyl-	79474	30
35	12.77	0.05	Citric Acid TMS 23.90	12	40
36	12.92	0.07	4-HYDROXY BUTYRIC ACID DI-TMS	98	59
37	13.15	1.99	GLYCERIC ACID TRI-TMS	323	87
38	13.37	0.22	2 METHYL 3-OXOBUTYRATE DI-TMS	124	72
39	13.60	0.17	4 DE-O TETRONIC TMS3 THREO	1408	50
40	14.02	0.86	SERINE TRI-TMS	321	46
41	14.82	0.17	THREONINE N,O,O-TRI-TMS	348	86
42	15.41	0.24	SILANE, 2-ETHOXYETHOXY TRIMETHYL-	1110	47
43	16.04	0.83	3,4 DIHYDROXY BUTYRIC ACID TRI-TMS	359	81
44	16.50	0.07	3-hydroxyadipic acid tri-TMS, 21.53	352	37
45	17.35	0.20	BUTANEDIOIC ACID, TMS-OXY-, BIS-TMS ESTER	415	49
46	17.56	0.54	BUTANEDIOIC ACID, TMS-OXY-, BIS-TMS ESTER	415	49
47	18.06	8.18	PYROGLUTAMIC ACID DI-TMS	2441	91
48	18.81	1.26	CREATININE ENOL TRI-TMS	1457	96
49	18.94	2.08	(R*,S*)-2,3-Dihydroxybutanoic acid, tris(trimethyls	145448	16
50	19.31	3.91	2-Keto-l-gluconic acid, penta(O-trimethylsilyl)- ,	300	36
51	19.45	0.63	2-HYDROXYGLUTARIC TRI-TMS, 857	461	87
52	19.76	0.12	301,344 @ 19.64	337	95
53	19.95	0.36	2-HEXENEDIOIC ACID DI-TMS, E-	222	49
54	20.06	1.05	4-Trimethylsilyloxycyclohexylacetate, trimethylsily	261	99
55	20.23	0.30	233,178,163 @ 19.98	293	49
56	20.33	1.14	GLUTAMIC ACID TRI-TMS	1524	50
57	20.52	1.04	PARA-HYDROXYPHENYLACETIC ACID DI-TMS	1475	95
58	20.64	0.22	Xylonic acid, 2,3,5-tris-O-(trimethylsilyl)-, .gamm	158612	97
59	20.82	0.08	3-hydroxyadipic acid tri-TMS, 21.53	352	28
60	20.92	0.66	TARTARIC ACID TETRA-TMS, 20.676	666	93
61	21.09	0.28	ARABINOSE TETRA-TMS	670	95
62	21.26	0.22	3-HYDROXYDODECANEDIOIC ACID-tri-TMS, 29.14	1754	47

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63	21.42	0.39	Bis(trimethylsilyl)bromosuccinate	147376	12
64	21.58	0.30	3-hydroxyglutarate, 19.19	292	56
65	21.68	0.09	Suberic Acid di-TMS, 21.593	217	58
66	21.96	0.23	3-hydroxyglutarate, 19.19	292	41
67	22.07	0.58	Levoglucozan, tris(trimethylsilyl)-, 21.775	204	86
68	22.26	0.32	GALACTOSE PENTA-TMS	875	94
69	22.60	1.87	XYLITOL PENTA-TMS	838	95
70	22.73	0.93	myo-inositol hexa-TMS, 27.886	274	42
71	23.02	1.31	n-Octanoic acid, pentamethyl-disilyl ester	107897	7
72	23.11	0.28	(2,2-Dimethyl-propionyl)-tris(trimethylsilyl)silane	143280	9
73	23.24	2.89	1,3-Dihydroxyacetone dimer	43364	1
74	23.44	0.50	GALACTONIC ACID HEXA-TMS	985	45
75	23.52	0.25	NONANEDIOIC ACID DI-TMS AZELAIC	1410	78
76	23.62	0.56	D-XYLOPYRANOSE TETRA-TMS	676	47
77	23.73	0.64	Galactonic acid, 2,3,4,5,6-pentakis-O-(trimethylsil	170	38
78	23.88	0.36	.ALPHA.-D-GALACTOSIDE, METHYL TETRAKIS-O-TMS	794	50
79	24.18	2.40	Ethylglucuronide per-TMS, 26.738	312	23
80	24.62	0.26	.ALPHA.-D-GLUCOPYRANURONIC ACID, 1,2,3,4-TETRAKIS-O	910	47
81	24.85	2.91	.BETA.-D-GALACTOFURANOSE, 1,2,3,5,6-PENTAKIS-O- TMS	877	91
82	24.99	0.49	7-Chloro-4-[p-[1-dimethylaminocyclohexylmethyl]anil	169341	38
83	25.16	0.81	d6 Sebacic Acid TMS 25.07	195	98
84	25.38	0.28	.BETA.-D-GALACTOFURANOSIDE, ETHYL 2,3,5,6-TETRAKIS-	810	78
85	25.49	1.10	P-HYDROXYPHENYL LACTIC ACID TRI-TMS	576	98
86	25.64	3.96	D-MANNOPYRANOSE PENTA-TMS	889	94
87	25.74	0.59	GALACTOSE PENTA-TMS	875	81
88	25.84	0.87	Histidine per-TMS, 25.539	283	97
89	25.91	0.31	D-RIBOFURANOSE TETRA-TMS	682	45
90	26.06	0.62	N,O-Bis(trimethylsilyl)-L-phenylalanine	130125	37
91	26.24	0.56	MANNITOL HEXA-TMS	977	46
92	26.35	1.97	Glucose oxime hexakis(trimethylsilyl)	188995	38
93	26.73	1.07	Galactonic acid, 2,3,4,5,6-pentakis-O-(trimethylsil	170	60
94	26.89	4.23	.ALPHA.-D-GALACTOPYRANOSE, 1,2,3,4,6-PENTAKIS-O- TM	882	87
95	27.01	0.21	GALACTOSE PENTA-TMS	875	47
96	27.09	0.09	GALACTOSE PENTA-TMS	875	94
97	27.17	1.25	Hexanedioic acid, 3-oxo-, tris(trimethylsilyl) deri	163597	10
98	27.27	0.50	PALMITIC ACID TMS	334	93
99	27.34	0.64	SACCHARIC TMS 6	1552	56
100	27.39	0.81	.ALPHA.-L-GALACTOFURANOSE, 6-DEOXY-1,2,3,5-TETRAKIS	721	72
101	27.45	0.59	ALLO-INOSITOL HEXA-TMS	968	40
102	27.58	0.17	3-hydroxysebacic acid tri-TMS, 27.33	211	46
103	27.66	0.08	Galactonic acid, 2,3,5,6-tetrakis-O-(trimethylsilyl	181667	25
104	27.73	0.24	GLYCERIC ACID TRI-TMS	323	52
105	27.92	0.24	d-Glucitol, 2-(acetylamino)-2-deoxy-1,3,4,5,6-penta	188024	38
106	27.99	0.72	2-Keto-l-gluconic acid, penta(O-trimethylsilyl)- ,	300	40
107	28.06	1.10	d6 Inositol TMS 28.02	257	91
108	28.30	0.29	300, 257 @ 28.114 YB0706	309	62
109	28.37	0.13	PENTANEDIOIC ACID, 2,4-BIS TMS-OXY -, BIS-TMS ESTER	714	72
110	28.43	0.09	D-XYLOPYRANOSE TETRA-TMS	676	52
111	28.55	0.14	Estra-1,3,5(10)-trien-17-one, 2,3-bis[(trimethylsil	177189	43
112	28.70	0.16	7-methylguanidine di-TMS, 28.483	234	38
113	28.89	0.29	LINOLEIC ACID TMS	422	92
114	29.06	0.19	5-Hydroxy Indole Acetic Acid TRI-TMS 28.75	126	97
115	29.13	1.10	I.S.:5-deutero-tryptophan per-TMS, 28.890	235	42
116	29.27	0.40	ALTRONIC ACID, 2,3,5,6-TETRAKIS-O-TMS-, LACTONE	736	47
117	29.38	0.34	Pregnan-20-one, 3,11,21-tris[(trimethylsilyl)oxy]-,	188336	8
118	29.53	0.13	Gulonic acid, 2,3,5,6-tetrakis-O-(trimethylsilyl)-,	181655	20
119	29.59	0.12	Gulonic acid, 2,3,5,6-tetrakis-O-(trimethylsilyl)-,	181659	45
120	29.66	0.34	D-Fructose, 1,3,4,5,6-pentakis-O-(trimethylsilyl)-	186541	35
121	29.74	0.43	.ALPHA.-D-GALACTOPYRANOSE, 1,2,3,4,6-PENTAKIS-O- TM	882	53
122	29.82	0.34	2-(N-Methyl-N-nitroamino)-4-[2,2,3,3,3-pentafluoro-	189252	1
123	29.90	0.12	ARABINO-HEXARIC ACID, 2-DEOXY-3,4,5-TRIS-O- TMS-, B	905	50
124	29.96	0.17	GALACTOSE PENTA-TMS	875	46
125	30.21	1.21	.BETA.-D-GALACTOFURANOSE, 1,2,3,5,6-PENTAKIS-O- TMS	877	42
126	30.52	0.31	Sebacic acid, bis(trimethylsilyl) ester	150522	7
127	30.64	0.27	D-RIBOSE TETRA-TMS	671	53

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128	30.73	0.23	Piperazine, 2,5-diketo-, bis(trimethylsilyl)-	96993	42
129	30.93	0.34	1-Propene-1,2,3-tricarboxylic acid, tris(trimethyls	168231	12
130	31.05	0.08	Chloral hydrate glucuronide per-TMS, 29.71	294	25
131	31.17	1.43	HAWKINSIN PENTA-TMS, 1653	2635	90
132	31.22	0.40	HAWKINSIN PENTA-TMS, 1653	2635	90
133	31.33	0.56	FUMARIC ACID DI-TMS, 12.938	1641	40
134	31.56	0.09	2,4,6 1H,3H,5H-PYRIMIDINETRIONE, 5- 3-CHLORO-2- TMS	567	41
135	31.81	0.34	Palmitate monoglyceride di-TMS, 31.663	263	76
136	32.13	0.19	D-XYLOPYRANOSE TETRA-TMS	676	46
137	32.19	0.28	D-RIBOSE TETRA-TMS	671	43
138	32.36	0.18	.BETA.-D-GALACTOFURANOSIDE, ETHYL 2,3,5,6-TETRAKIS-	810	53
139	32.51	1.59	GALACTOSE PENTA-TMS	875	58
140	32.64	0.12	D-XYLOPYRANOSE TETRA-TMS	676	62
141	32.93	0.38	N2-(4-Chlorophenyl)-2,2-diphenyl-N1,N1-dipropylacet	172370	10
142	33.14	0.28	Octadecanoic acid, 2,3-bis[(trimethylsilyl)oxy]prop	184625	7
143	33.38	1.40	.ALPHA.-D-GALACTOPYRANOSE, 1,2,3,4,6-PENTAKIS-O- TM	882	80
144	33.77	0.11	Xanthumin	128369	1
145	33.86	0.05	7-Fluoro-3,4-dihydro-3-[4-[trifluoromethyl]phenyl]-	163401	22
146	33.93	0.06	211,375 @ 32.477	272	55
147	34.05	0.08	1H-Indole-2,3-dione, 1-(tert-butyldimethylsilyl)-7-	177515	25
148	34.54	0.08	Zirconium, bis(.eta.5-2,4-cyclopentadien-1-yl)bis(.	187956	9
149	37.39	0.06	3.beta.,5-Bis(trimethylsiloxy)-5.alpha.-cholestane	186898	87
150	37.59	0.06	d3 Serine TMS 13.902	191	20

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File : D:\OCMD\Completed\OCM104  
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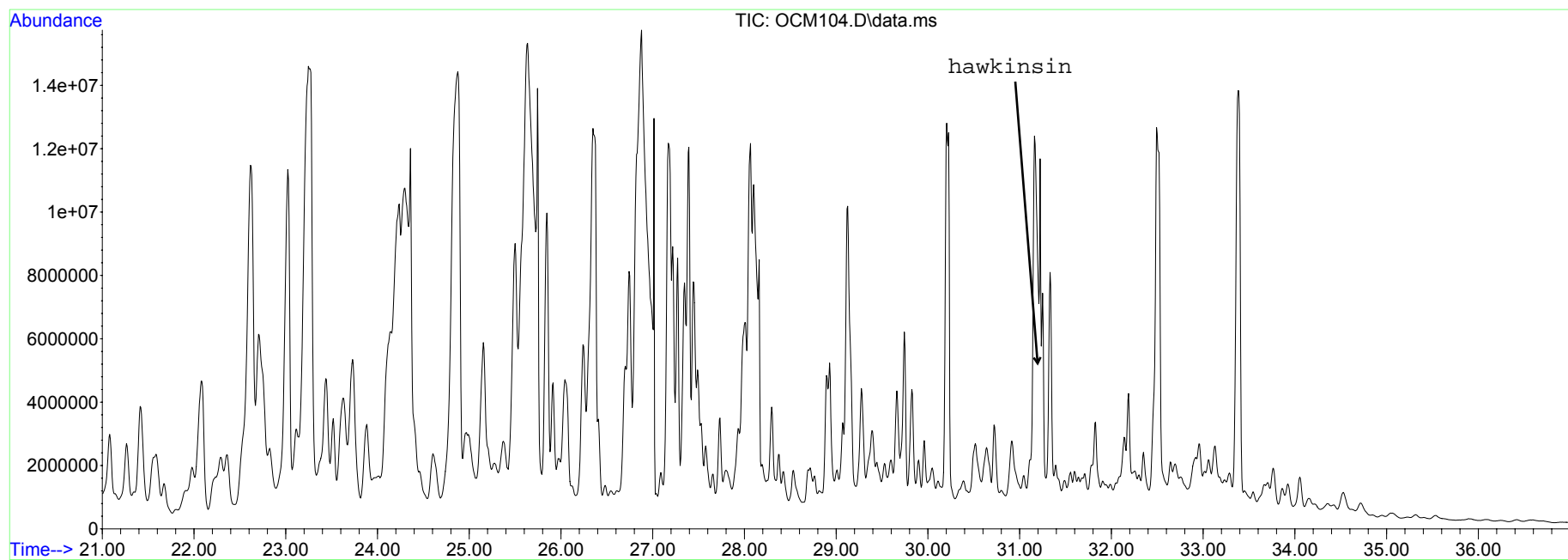
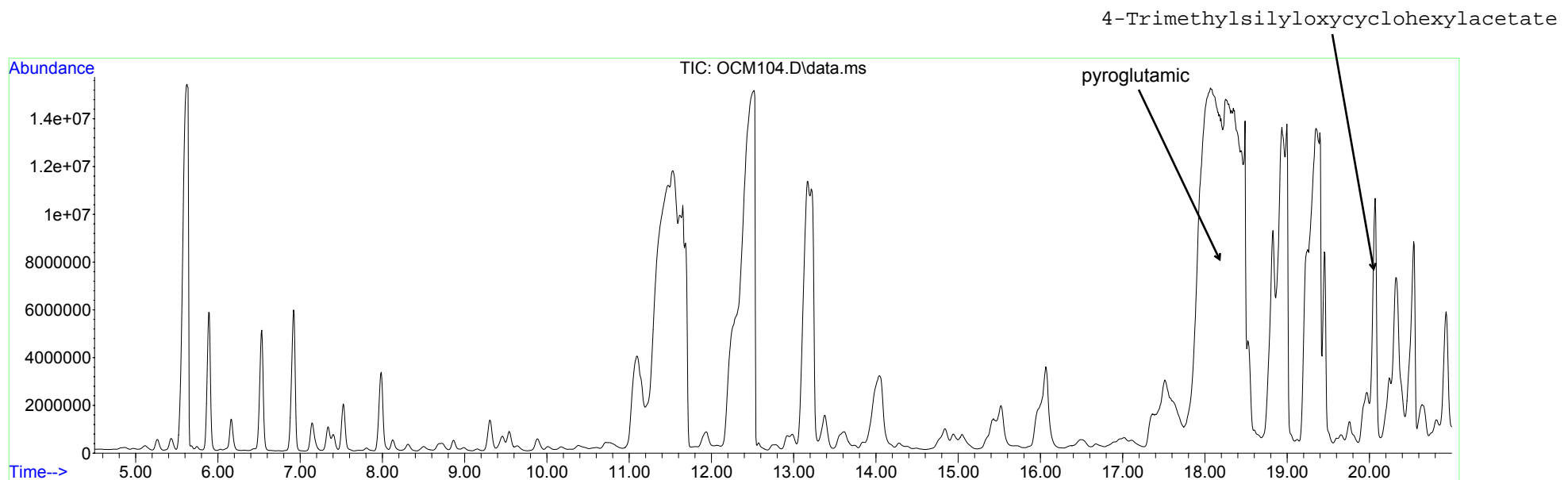


Table I.

Inborn errors diagnosed in 13,242 patients using the Urease method Prior to State Newborn Screening by Tandem Mass Spectrometry **Diagnoses in Red would probably not have been made by liquid or solid phase extraction organic acid analysis, or tandem mass spectrometry.**

Of 14,919 Samples, from 13,242 patients  
1691 Abnormal 11%  
164 confirmed Inborn Errors  
+ 20 MADD or cystinuria  
184 Total 1.38%  
48 involved non-acidic metabolites  
28 involved orotate or hexanoylglycine  
35-70% increase due to urease method

Of 184 Inborn Errors:

14	<b>Urea cycle</b>	2	HMG
18	MCAD	3	SCAD
14	LCHAD	2	<b>Hyperoxaluria</b>
10	MMA	4	<b>Renal Fanconi</b>
7	MSUD	3	<b>Molybdenum Cofactor Def</b>
5	Propionic		
9	<b>Galactosemia</b>	2	<b>Iminoaciduria</b>
12	Glutaric I	4	<b>Cystinuria</b>
5	Biotinidase	3	Adrenoleukodystrophy
4	Glutaric II		
2	Pyruvate Dehydrogenase,		
2	<b>Glyceroluria</b>		
2	<b>Hereditary Fructose Intolerance</b>		
2	<b>FIGLU uria,</b>		
2	<b>Homocystinuria</b>		
2	Glycerate kinase def		
2	<b>Hartnup's</b>		
2	<b>Hypophosphatasia</b>		
3	3-methylglutaconic		
3	3-methylglutaconic		
3	Adrenoleukodystrophy		
3	<b>Cystathioninase def</b>		
4	beta-methylcrotonyl CoA carboxylase		
4	<b>Sialuria</b>		
6	<b>Pyroglutamic</b>		

Single cases:

PKU  
Alkaptonuria  
Fumaric  
**2-Hydroxyglutaric**  
**Proline Oxidase,**  
Succinic Semi-aldehyde dehydrogenase Deficiency  
**Canavan's Disease**  
**Renal Glycosuria,**  
Pyruvate dehydrogenase def  
**Hawkinsinuria**  
Hepato-Renal Tyrosinemia