

Administration Office

c/o EMQN CIC, Unit 4, Enterprise House,
Manchester Science Park, Pencroft Way,
Manchester M15 6SE, United Kingdom.

Tel: +44 161 757 4952

Fax: +44 161 850 1145

Email: admin@erndim.org

Scientific Coordination

Mrs C Scott and Miss S Colyer
NHS
Department of Clinical
Chemistry and Newborn Screening
The Children's Hospital
Sheffield
S10 2TH
United Kingdom

Scheme Organisation

CSCQ (Quality Control Centre, Switzerland)
2 chemin du Petit-Bel-Air
1225 Chêne-Bourg
Switzerland,
Tel: +41 22 305 52 36
Email: cscq@hcuge.ch

Published: 02 February 2024¹

Qualitative Organic Acids

Centre: United Kingdom

Final Report 2023

prepared by
Mrs C Scott and Miss S Colyer

Note: This annual report is intended for participants of the ERNDIM QLOU Sheffield scheme. The contents should not be used for any publication without permission of the Scientific Advisor.

The fact that your laboratory participates in ERNDIM schemes is not confidential, however, the raw data and performance scores are confidential and will only be shared within ERNDIM for the purpose of evaluating your laboratories performance, unless ERNDIM is required to disclose performance data by a relevant government agency. For details please see the ERNDIM Privacy Policy on www.erndim.org

In 2023, 74 labs participated to the Qualitative Organic Acid Scheme, Sheffield.

1. Geographical distribution of participants

For the first survey, 73 and second survey 72 laboratories submitted results.

Country	Number of participants
Australia	6
Belgium	7
Finland	2
Hungary	1
Ireland	1
Israel	3
Japan	6
Malaysia	3
New Zealand	1
Norway	1
Pakistan	1
Poland	3
Slovakia	2

¹ If this report is not Version 1 for this scheme year, go to APPENDIX 1 for details of the changes made since the last version of this document.

South Africa	2
Spain	1
Sweden	2
United Kingdom	16
United States of America	16

2. Design and logistics of the scheme including sample information.

The scheme has been designed and planned by Camilla Scott as Scientific Advisor, Sharon Colyer as Deputy Scientific Advisor and coordinated by Alessandro Salemma scheme organiser (sub-contractor on behalf of CSCQ), both appointed by and according to procedures laid down the ERNDIM Board. CSCQ dispatches QLOU EQA samples to the scheme participants and provides a website for on-line submission of results and access to scheme reports. Existing QLOU scheme participants can log on to the CSCQ results submission website at:

<https://cscq.hcuge.ch/cscq/ERNDIM/Initial/Initial.php>

2 surveys	Round 1: patients A, B and C
	Round 2: patients D, E and F

Origin of patients: all urine samples have been provided by the scheme organizers.

The samples used in 2023 were human urine samples. Three patient sample donations, and one control sample donation were collected through Sheffield Children's NHS Foundation Trust, Sheffield UK. A further two samples were selected and donated by the Scientific Advisor for QLOU-Heidelberg, Joachim Janda through the Center for Metabolic Diseases Heidelberg, Heidelberg, Germany.

The samples have been heat-treated. They were pre-analysed in our institute after 3 days incubation at ambient temperature (to mimic possible changes that might arise during transport). In all six samples the typical metabolic profiles were preserved after this process. Details regarding stability of (reconstituted) samples are provided in the sample package.

Mailing: samples were sent by DHL; FedEx or the Swiss Post at room temperature.

3. Tests

Analyses of qualitative organic acids.

4. Schedule of the scheme

- Feb 8, 2023: shipment of samples by CSCQ
- May 9, 2023: analysis start and website submission 1st round (A-C)
- May 30, 2023: results submission deadline
- August 29, 2023: analysis start and website submission 2nd round (D-F)
- Sep 27, 2023: deadline for result submission (Survey 2)
- August 01, 2023: report of Survey 1 by e-mail
- November 11, 2023: report of Survey 2 by e-mail
- January 20, 2024: annual report with scoring.

5. Results

	Survey 1	Survey 2
Receipt of results	73	72
No answer	1	2

6. Web site reporting

The website reporting system is compulsory for all centres. Please carefully read the following advice:

- Results
 - Enter the key metabolites with the evaluation **in the tables**.
 - If the profile is normal: enter "Normal profile" in "Key metabolites".
 - **Don't enter results in the "comments" window, otherwise your results will not be included in the evaluation program.**
- Recommendations = **advice for further investigation**.
 - Scored together with the interpretative score.
 - Advice for treatment are not scored but may be used in the overall assessment.
 - **Don't give advice for further investigation in "Comments on diagnosis":** it will not be included in the evaluation program.
 - Enter the key metabolites with the evaluation **in the tables** even if you don't give quantitative data.
 - If the profile is normal: enter "Normal profile" in "Key metabolites".
 - **Don't enter results in the "comments" window, otherwise your results will not be included in the evaluation program.**

7. Scoring and evaluation of results

Information regarding procedures for establishment of assigned values, statistical analysis, interpretation of statistical analysis etc. can be found in generic documents on the ERNDIM website.

The scoring system has been established by the International Scientific Advisory Board of ERNDIM. Two criteria are evaluated: 1) analytical performance, 2) interpretative proficiency also considering recommendations for further investigations.

A	Analytical performance	Correct results of the appropriate tests	2
		Partially correct or non-standard methods	1
		Unsatisfactory or misleading	0
I	Interpretative proficiency & Recommendations	Good (diagnosis was established)	2
		Helpful but incomplete	1
		Misleading or wrong diagnosis	0

The total score is calculated as a sum of these two criteria. The maximum to be achieved is 4 points per sample. The scores were calculated only for laboratories submitting results.

Scoring and certificate of participation: scoring is carried by a second assessor who changes every year as well as by the scientific advisor. The results of QLOU US have also been second scored by Joachim Janda, QLOU SA, Heidelberg. At the SAB meeting in November 2023 the definitive scores have been finalised. The concept of critical error was introduced in 2014. A critical error is defined as an error resulting from seriously misleading analytical findings and /or interpretations with serious clinical consequences for the patient. Thus, labs failing to make a correct diagnosis of a sample considered as eligible for this category will be deemed not to have reached a satisfactory performance even if their total points for the year exceed the limit set at the SAB. For 2023, the SAB decided that critical error would be awarded for sample B (MMA) and sample F (Alkaptonuria) if the diagnosis was missed and/or there were no suggestions for appropriate tests that would ultimately reach the correct diagnosis.

A certificate of participation will be issued for participation, and it will be additionally notified whether the participant has received a performance support letter. This performance support letter is sent out if the performance is evaluated as unsatisfactory. Two performance support letters will be sent by the Scheme Advisor for 2023. Two critical error letters will be sent out for this scheme for 2023. Any partial submitters will receive a letter from the ERNDIM Executive Administrator, Sara Gardner.

7.1. Score for satisfactory performance

At least 17 points from the maximum of 36 (72%).

8. Results of samples and evaluation of reporting

8.1. Patient A

Normal

Patient details provided to participants.

Hot flushes at night, 48-year-old, female.

Patient details

Sample A was donated from healthy volunteer undergoing the menopause with no underlying medical conditions.

Analytical performance

Number of labs:

No abnormal organic acids	71
Methylmalonic acid	1
3-hydroxy isobutyric acid	1

Diagnosis / Interpretative proficiency

Number of labs:

No significant abnormality	72
Methylmalonic aciduria	1

Recommendations

None

Scoring

Analytical:

No abnormal findings 2 points

Clinical:

Normal profile 2 points

Overall impression

A normal profile that was identified as normal by nearly all participants in this scheme. As in previous distributions, overinterpretation of normal controls can sometimes result in low scoring for individual participants.

8.2. Patient B

Methylmalonic Aciduria

Patient details provided to participants.

Intellectual disability, 3-year-old male.

Patient details

Sample B was donated from a known patient diagnosed with Methylmalonyl-CoA Mutase deficiency in early childhood.

Analytical performance

Number of labs:

Methylmalonic acid	71
Propionic Acid	1
Orotic acid	1

Diagnosis / Interpretative proficiency

Number of labs:

Methylmalonic aciduria	71
Ornithine transcarbamylase deficiency	1
Propionic acidaemia	1

Recommendations

Total homocysteine, plasma amino acids, plasma acylcarnitine's, quantitative methylmalonic acid, vitamin B12 measurement, folic acid measurement. Mutational analysis of the *MMUT* gene.

Scoring

Analytical:

Methylmalonic acid 2 points

Clinical:

Methylmalonic aciduria 2 points

Overall impression

Good scoring survey with an easy recognisable patten.

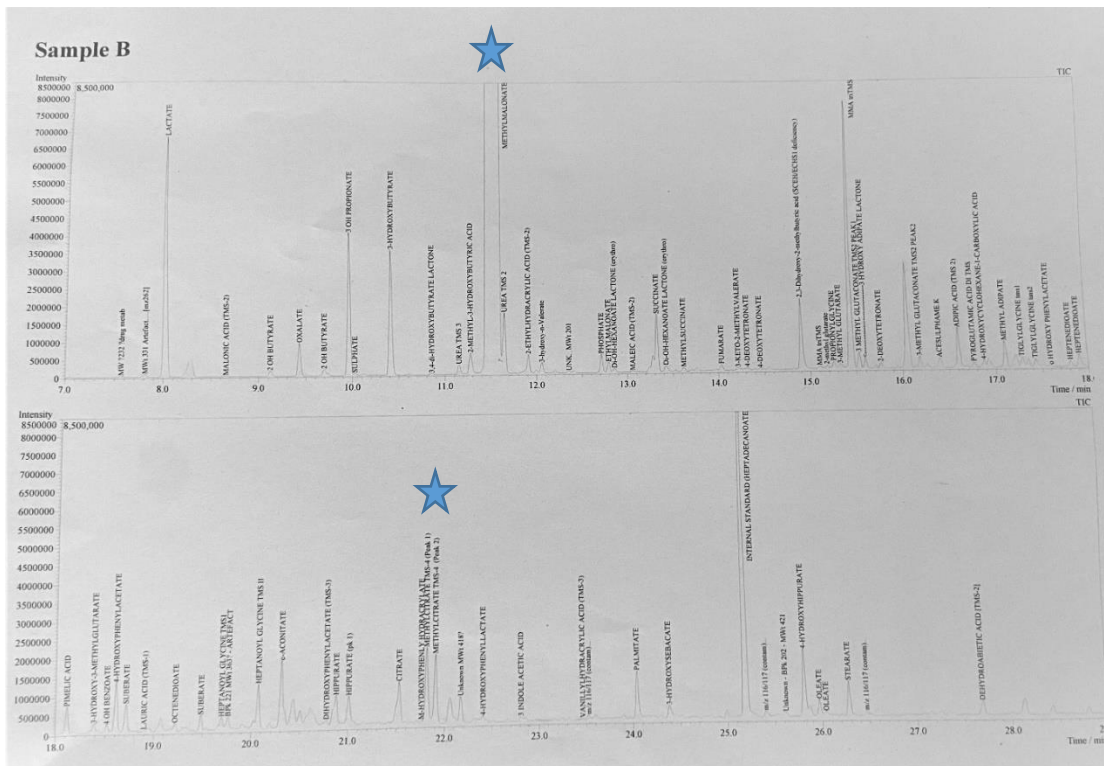


Figure two. The chromatogram for sample B demonstrating significantly increased excretion of methylmalonic acid and methyl citrate.

8.3 Patient C

Ornithine Transcarbamylase Deficiency

Patient details provided to participants.

Muscle Weakness, memory problems, 25-year-old female.

Patient details

Patient C was donated from a sister laboratory and is from an adult patient with known Ornithine Transcarbamylase (OTC) deficiency. The patient is also being investigated for possibly secondary mitochondrial dysfunction.

Analytical performance

Number of labs:

Orotic Acid	67
-------------	----

Diagnosis / Interpretative proficiency

Number of labs:

Urea Cycle Defect/OTC	67
MADD	2
5 Oxoprolinase	1
AADC	1

Recommendations

Plasma ammonia, plasma, and urine amino acids, quantitative orotic acid, mutational analysis of the genes associated with defects of the urea cycle including *OTCD*.

Scoring

Analytical:

Orotic acid	2 points
-------------	----------

Clinical:

UCD/OTC	2 points
---------	----------

Overall impression

Most participants identified this sample as attributable to a urea cycle defect. There was also evidence of secondary mitochondrial dysfunction.

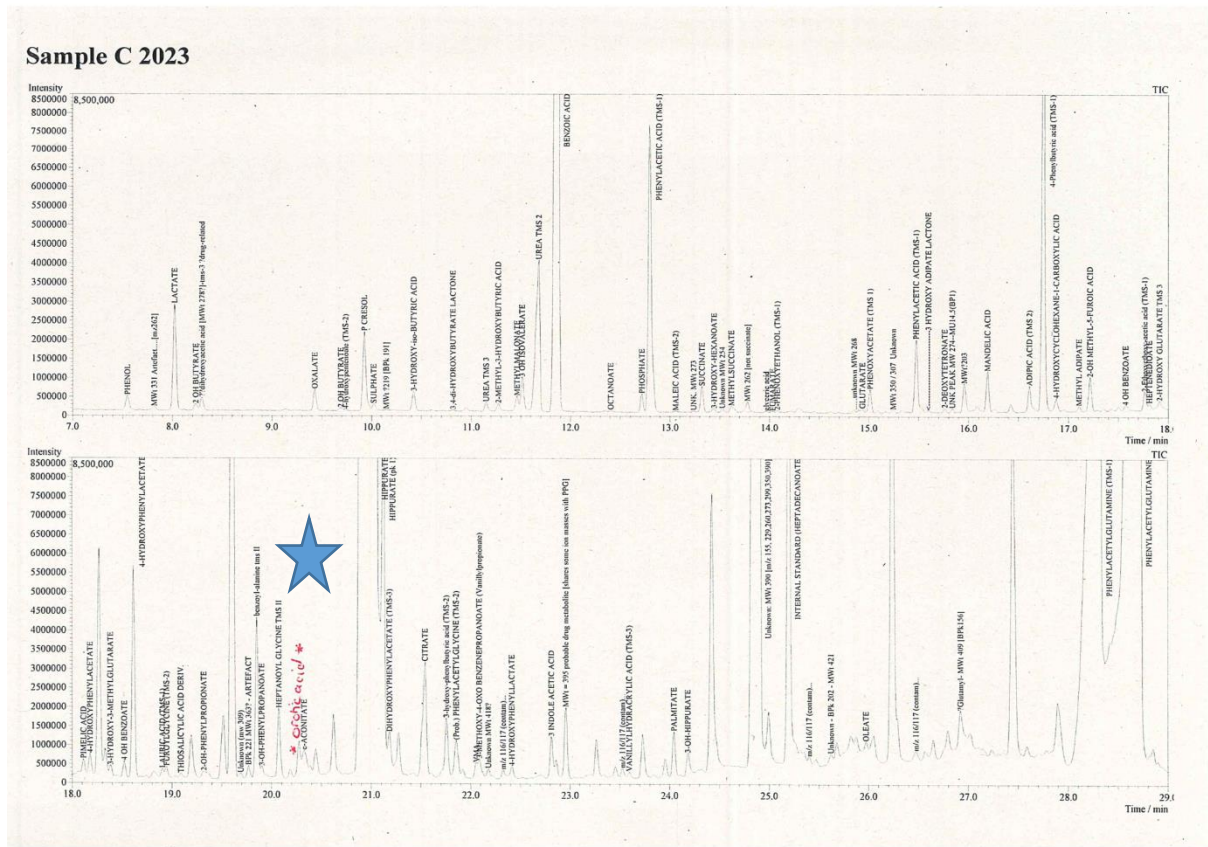


Figure three. The chromatogram for sample C demonstrating increased excretion of orotic acid. On our chromatogram the orotic acid co-elutes with c-aconitate.

8.4 Patient D

Barth Syndrome

Patient details provided to participants.

Cardiomyopathy Screen, 6-year-old male.

Patient details

Sample D was donated from an older child with biochemical and genetically confirmed Barth Syndrome.

Analytical performance

Number of labs:

3 Methylglutaconic Acid	72
3 Methyl Glutaric Acid	71

Diagnosis / Interpretative proficiency

Number of labs:

Barth Syndrome	67
Other secondary 3 Methylglutaconic aciduria	4
Other	1

Recommendations

Molecular analysis of the TAZ gene.

Scoring

Analytical:

3 Methylglutaconic acid	1 point
3 Methyl Glutaric Acid	1 point

Interpretative:

Barth Syndrome	2 points
3 Methylglutaconic aciduria	1 point

Overall impression

Most participants identified the correct metabolites and suggested Barth Syndrome as the most likely diagnosis scoring the maximum of 4 points. A small number did not specify Barth Syndrome in their report and scored 3 points. The clinical details provided were highly suggestive of Barth Syndrome.

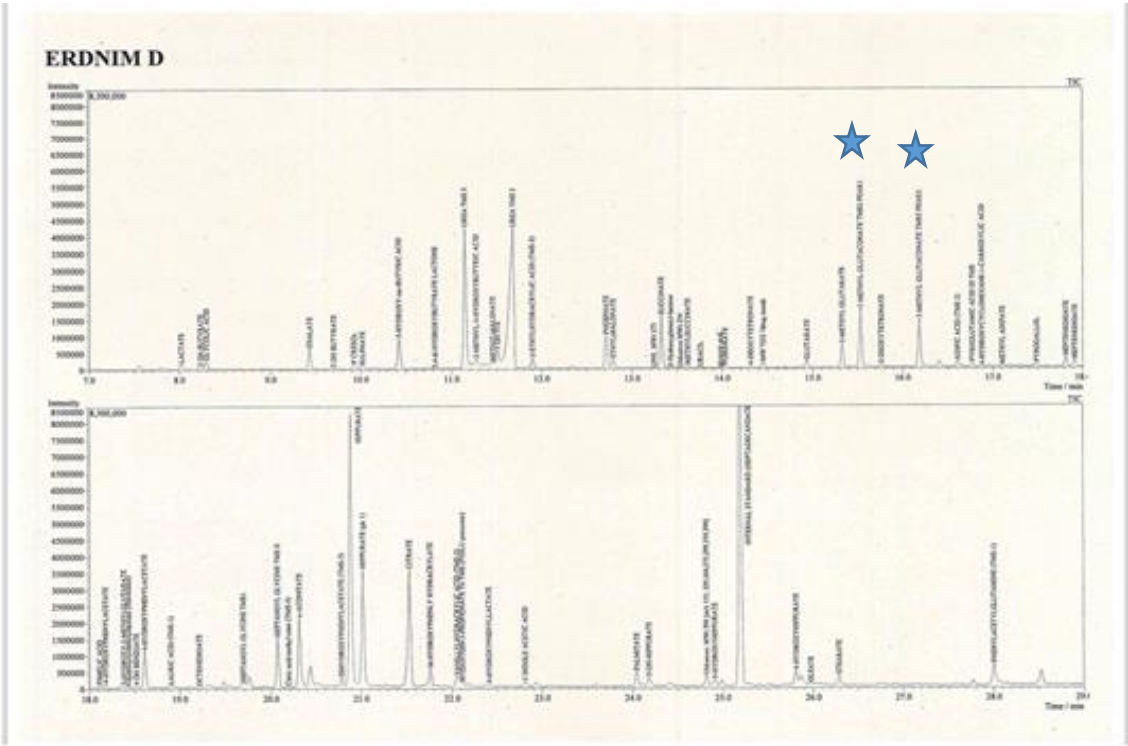


Figure four. The chromatogram for sample C demonstrating increased excretion of 3-Methylglutaconic acid.

8.5 Patient E

3-Methyl Glutaconyl Hydratase Deficiency

Patient details provided to participants

Adult onset leukoencephalopathy, 25-year-old male.

Patient details

Sample E was donated from a patient who had recently been diagnosed with 3-Methyl Glutaconyl Hydratase Deficiency. This was subsequently confirmed by genetic analysis.

Analytical performance

Number of labs:

3 Methylglutaconic Acid	69
3-hydroxy isovaleric acid	53

Diagnosis / Interpretative proficiency

Number of labs:

Primary 3 Methylglutaconic aciduria	61
Secondary 3 Methylglutaconic aciduria	8

Recommendations

Molecular analysis of the AUH gene.

Scoring

Analytical:

3 Methylglutaconic acid	1 point
3 Hydroxy Isovaleric Acid	1 point

Interpretative:

3 Methylglutaconyl Hydratase Deficiency/Primary 3 Methylglutaconic aciduria	2 points
3 Methylglutaconic aciduria	1 point

Overall impression

The key to the correct diagnosis was the identification of the increased 3 hydroxyisovaleric acid along with the clinical details. Most participants identified the key metabolites and reached the correct diagnosis. There was an increased excretion of ethylmalonic acid in this patient, the origin of which remains unknown.

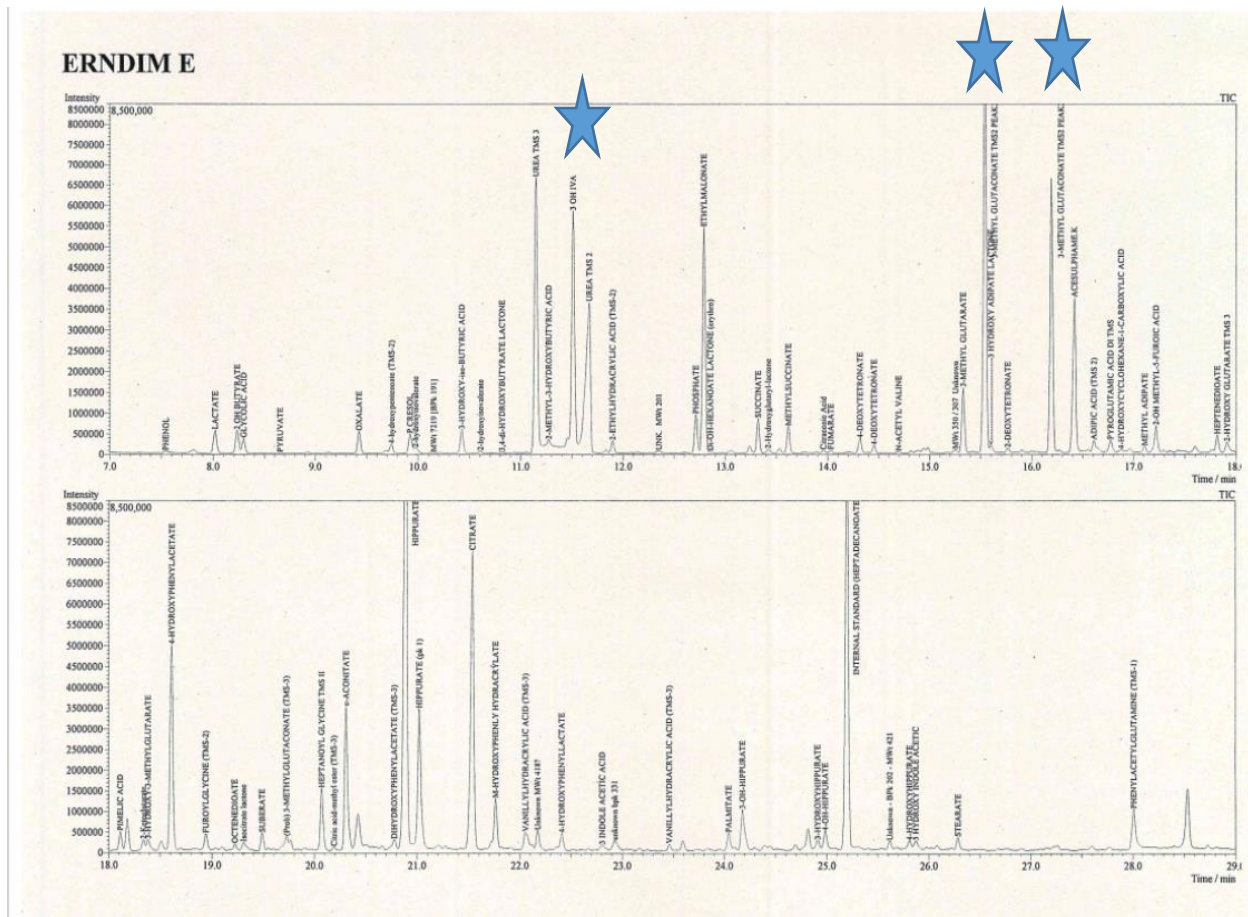


Figure five. The chromatogram for sample E demonstrating increased excretion of 3-hydroxyisovaleric acid and 3-methyl glutaconic acid.

8.6 Patient F

Alkaptonuria

Patient details provided to participants

Joint pain & Swelling, 60 year old male.

Patient details

Patient F was donated by a sister laboratory and is from a patient with confirmed Alkaptonuria.

Analytical performance

Number of labs:

Homogentisic acid 71
3 Methylglutaric acid 1

Diagnosis / Interpretative proficiency

Alkaptonuria 71
Other 1

Recommendations

Molecular testing of the HGD gene

Scoring

Analytical:

Homogentisic acid

2 points

Clinical:

Alkaptonuria

2 points

Overall impression

High scoring sample with no analytical or interpretive difficulties.

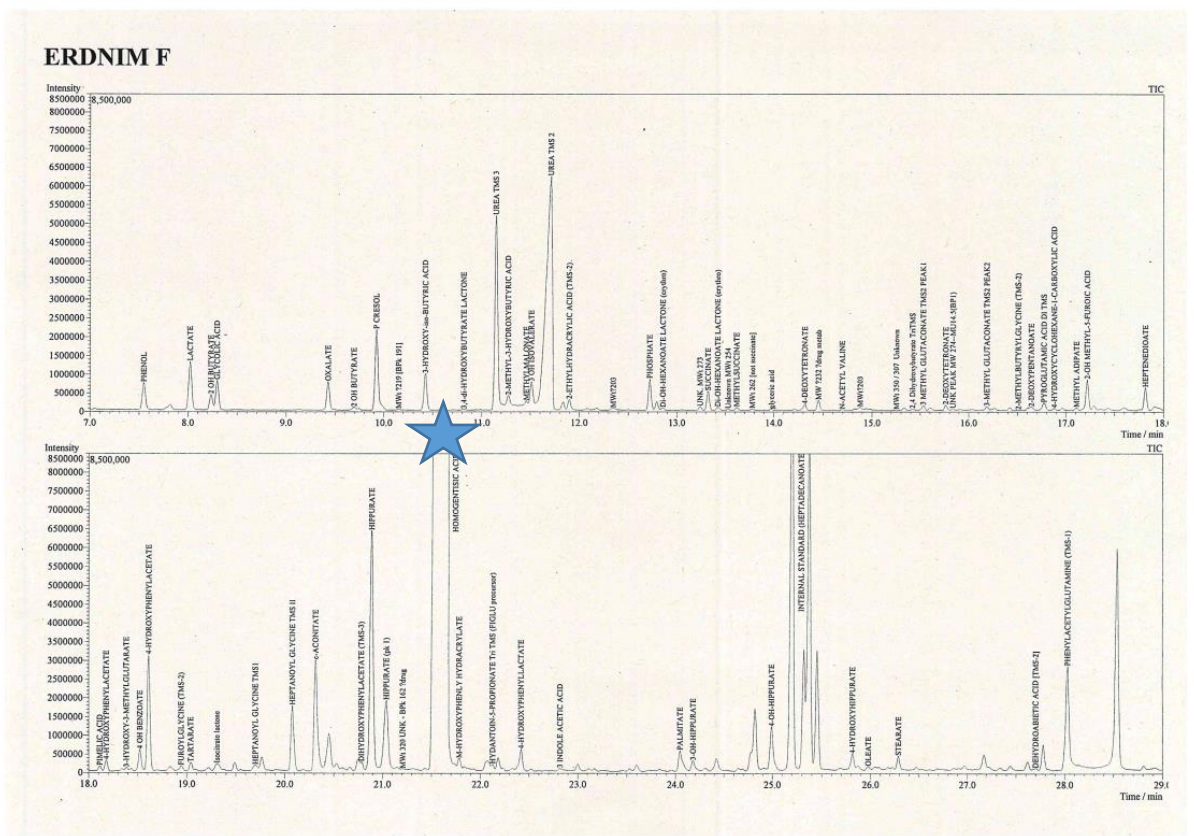


Figure Six. The chromatogram for sample F demonstrating increased excretion homogentisic acid.

9. Scores of participants

All data transfer, the submission of data as well as the request and viewing of reports proceed via the DPT-CSCQ results website. The results of your laboratory are confidential and only accessible to you (with your username and password). The anonymous scores of all laboratories are accessible to all participants and only in your version is your laboratory highlighted in the leftmost column.

If your laboratory is assigned poor performance and you wish to appeal against this classification please email the ERNDIM Administration Office (admin@erndim.org), with full details of the reason for your appeal, within one month receiving your Performance Support Letter. Details of how to appeal poor performance are included in the Performance Support Letter sent to poor performing laboratories.

Detailed scores – Round 1

Lab n°	Patient A Normal.			Patient B Methylmalonic aciduria.			Patient C Ornithine Transcarbamylase deficiency.			Total
	A	I	Total	A	I	Total	A	I	Total	
1	2	2	4	2	2	4	2	2	4	12
2	2	2	4	2	2	4	2	2	4	12
3	2	2	4	2	2	4	2	2	4	12
4	2	2	4	2	2	4	2	2	4	12
5	2	2	4	2	2	4	2	2	4	12
6	2	2	4	2	2	4	2	2	4	12
7	2	2	4	2	2	4	2	2	4	12
8	2	2	4	2	2	4	2	2	4	12
9	2	2	4	2	2	4	2	2	4	12
10	2	2	4	2	2	4	2	2	4	12
11	2	2	4	2	2	4	2	2	4	12
12	2	2	4	2	2	4	2	2	4	12
13	2	2	4	2	2	4	2	2	4	12
14	2	2	4	2	2	4	2	2	4	12
15	2	2	4	2	2	4	2	2	4	12
16	2	2	4	2	2	4	2	2	4	12
17	2	2	4	2	2	4	2	2	4	12
18	2	2	4	2	2	4	2	2	4	12
19	2	2	4	2	2	4	2	2	4	12
20	2	2	4	2	2	4	2	0	2	10
21	2	2	4	2	2	4	0	0	0	8
22	2	2	4	2	2	4	2	2	4	12
23	2	2	4	2	2	4	2	2	4	12

24	2	2	4	2	2	4	2	2	4	12
25	2	2	4	2	2	4	2	2	4	12
26	2	2	4	2	2	4	2	2	4	12
27	2	2	4	2	2	4	2	2	4	12
28	2	2	4	2	2	4	2	2	4	12
29	2	2	4	2	2	4	2	2	4	12
30	2	2	4	2	2	4	2	2	4	12
31	2	2	4	2	2	4	2	2	4	12
32	2	2	4	2	2	4	2	2	4	12
33	2	2	4	2	2	4	2	2	4	12
34	2	2	4	2	2	4	2	2	4	12
35	2	2	4	2	2	4	2	2	4	12
36	2	2	4	2	2	4	2	2	4	12
37	2	2	4	2	2	4	2	2	4	12
38	2	2	4	2	2	4	0	1	1	9
39	2	2	4	2	2	4	2	2	4	12
40	2	2	4	2	2	4	2	2	4	12
41	2	2	4	2	2	4	2	2	4	12
42	2	2	4	1	1	2	2	2	4	10
43	2	2	4	2	2	4	2	2	4	12
44	2	2	4	2	2	4	2	2	4	12
45	2	2	4	2	2	4	0	2	2	10
46	2	2	4	2	2	4	2	2	4	12
47	2	2	4	2	2	4	2	2	4	12
48	2	2	4	2	2	4	2	2	4	12
49	0	0	0	0	0	0	0	0	0	0
50	2	2	4	2	2	4	2	2	4	12
51	2	2	4	2	2	4	2	2	4	12
52	0	0	0	2	2	4	2	2	4	8
53	2	2	4	2	2	4	2	2	4	12
54	2	2	4	2	2	4	2	2	4	12
55	2	2	4	2	2	4	2	2	4	12
56	2	2	4	2	2	4	2	2	4	12
57	2	2	4	2	2	4	2	2	4	12
58	2	2	4	2	2	4	2	2	4	12
59	2	2	4	2	2	4	2	2	4	12

60	2	2	4	2	2	4	2	1	3	11
61	2	2	4	2	2	4	0	0	0	8
62	0	0	0	0	0	0	0	0	0	0
63	2	2	4	2	2	4	2	2	4	12
64	2	2	4	2	2	4	2	2	4	12
65	2	2	4	2	2	4	2	2	4	12
66	2	2	4	2	2	4	2	2	4	12
67	2	2	4	2	2	4	2	2	4	12
68	2	2	4	2	2	4	2	2	4	12
69	2	2	4	2	2	4	2	2	4	12
70	2	2	4	2	2	4	2	2	4	12
71	2	2	4	2	2	4	2	2	4	12
72	2	2	4	2	2	4	0	0	0	8
73	2	2	4	2	2	4	2	2	4	12
74	2	2	4	2	2	4	2	2	4	12

Detailed scores – Round 2

Lab n°	Patient D Barth Syndrome			Patient E 3 Methyl Glutaconyl-CoA hydratase deficiency			Patient F Alkaptonuria			Total
	A	I	Total	A	I	Total	A	I	Total	
1	2	2	4	2	2	4	2	2	4	12
2	2	2	4	1	0	1	2	2	4	9
3	2	2	4	2	2	4	2	2	4	12
4	2	2	4	2	2	4	2	2	4	12
5	2	2	4	2	2	4	2	2	4	12
6	2	2	4	2	2	4	2	2	4	12
7	2	2	4	2	2	4	2	2	4	12
8	2	2	4	2	2	4	2	2	4	12
9	2	2	4	2	2	4	2	2	4	12
10	2	2	4	2	1	3	2	2	4	11
11	2	2	4	2	2	4	2	2	4	12
12	2	2	4	2	2	4	2	2	4	12
13	2	2	4	2	2	4	2	2	4	12
14	2	2	4	2	2	4	2	2	4	12
15	2	2	4	2	2	4	2	2	4	12
16	2	2	4	2	2	4	2	2	4	12
17	2	2	4	2	2	4	2	2	4	12
18	2	2	4	2	2	4	2	2	4	12
19	2	2	4	2	2	4	2	2	4	12
20	2	2	4	2	2	4	2	2	4	12
21	0	0	0	0	0	0	0	0	0	0
22	2	2	4	2	2	4	2	2	4	12
23	2	2	4	2	2	4	2	2	4	12
24	2	2	4	2	2	4	2	2	4	12
25	2	2	4	2	2	4	2	2	4	12
26	2	2	4	2	2	4	2	2	4	12
27	2	2	4	1	1	2	2	2	4	10
28	2	2	4	2	2	4	2	2	4	12
29	2	2	4	1	0	1	2	2	4	9
30	2	2	4	2	2	4	2	2	4	12

31	2	2	4	2	2	4	2	2	4	12
32	2	2	4	1	2	3	2	2	4	11
33	2	2	4	2	2	4	2	2	4	12
34	2	2	4	2	2	4	2	2	4	12
35	2	2	4	2	2	4	2	2	4	12
36	2	2	4	2	2	4	2	2	4	12
37	2	2	4	2	1	3	2	2	4	11
38	2	2	4	1	2	3	2	2	4	11
39	2	2	4	2	2	4	2	2	4	12
40	2	2	4	2	2	4	2	2	4	12
41	2	2	4	2	2	4	2	2	4	12
42	2	2	4	0	0	0	2	2	4	8
43	2	2	4	1	2	3	2	2	4	11
44	2	2	4	2	2	4	2	2	4	12
45	2	2	4	2	2	4	2	2	4	12
46	2	2	4	2	2	4	2	2	4	12
47	2	2	4	1	2	3	2	2	4	11
48	2	2	4	2	2	4	2	2	4	12
49	2	2	4	2	2	4	2	2	4	12
50	2	2	4	2	1	3	2	2	4	11
51	2	2	4	2	2	4	2	2	4	12
52	2	2	4	2	2	4	2	2	4	12
53	2	2	4	2	2	4	2	2	4	12
54	2	2	4	2	2	4	2	2	4	12
55	2	1	3	1	1	2	2	2	4	9
56	2	2	4	2	2	4	2	2	4	12
57	2	2	4	2	2	4	2	2	4	12
58	0	0	0	0	0	0	0	0	0	0
59	2	2	4	2	2	4	2	2	4	12
60	4	0	4	1	1	2	2	2	4	10
61	2	0	2	1	1	2	2	2	4	8
62	2	1	3	2	2	4	2	2	4	11
63	2	2	4	2	2	4	2	2	4	12
64	2	2	4	2	2	4	2	2	4	12
65	2	2	4	2	2	4	2	2	4	12
66	2	2	4	2	2	4	2	2	4	12

67	2	1	3	2	2	4	2	2	4	11
68	2	2	4	0	1	1	2	2	4	9
69	2	2	4	1	2	3	2	2	4	11
70	2	2	4	1	2	3	2	2	4	11
71	2	2	4	1	1	2	2	2	4	10
72	2	1	3	2	1	3	0	0	0	6
73	2	2	4	2	2	4	2	2	4	12
74	2	2	4	2	2	4	2	2	4	12

Total scores

Lab n°	A	B	C	D	E	F	Cumulative score	Cumulative score (%)
1	4	4	4	4	4	4	24	100
2	4	4	4	4	1	4	21	88
3	4	4	4	4	4	4	24	100
4	4	4	4	4	4	4	24	100
5	4	4	4	4	4	4	24	100
6	4	4	4	4	4	4	24	100
7	4	4	4	4	4	4	24	100
8	4	4	4	4	4	4	24	100
9	4	4	4	4	4	4	24	100
10	4	4	4	4	3	4	23	96
11	4	4	4	4	4	4	24	100
12	4	4	4	4	4	4	24	100
13	4	4	4	4	4	4	24	100
14	4	4	4	4	4	4	24	100
15	4	4	4	4	4	4	24	100
16	4	4	4	4	4	4	24	100
17	4	4	4	4	4	4	24	100
18	4	4	4	4	4	4	24	100
19	4	4	4	4	4	4	24	100
20	4	4	2	4	4	4	22	92
21	4	4	0	0	0	0	8	33
22	4	4	4	4	4	4	24	100
23	4	4	4	4	4	4	24	100
24	4	4	4	4	4	4	24	100
25	4	4	4	4	4	4	24	100
26	4	4	4	4	4	4	24	100
27	4	4	4	4	2	4	22	92
28	4	4	4	4	4	4	24	100
29	4	4	4	4	1	4	21	88
30	4	4	4	4	4	4	24	100
31	4	4	4	4	4	4	24	100
32	4	4	4	4	3	4	23	96
33	4	4	4	4	4	4	24	100

34	4	4	4	4	4	4	24	100
35	4	4	4	4	4	4	24	100
36	4	4	4	4	4	4	24	100
37	4	4	4	4	3	4	23	96
38	4	4	1	4	3	4	20	83
39	4	4	4	4	4	4	24	100
40	4	4	4	4	4	4	24	100
41	4	4	4	4	4	4	24	100
42	4	2	4	4	0	4	18	75
43	4	4	4	4	3	4	23	96
44	4	4	4	4	4	4	24	100
45	4	4	2	4	4	4	22	92
46	4	4	4	4	4	4	24	100
47	4	4	4	4	3	4	23	96
48	4	4	4	4	4	4	24	100
49	0	0	0	4	4	4	12	50
50	4	4	4	4	3	4	23	96
51	4	4	4	4	4	4	24	100
52	0	4	4	4	4	4	20	83
53	4	4	4	4	4	4	24	100
54	4	4	4	4	4	4	24	100
55	4	4	4	3	2	4	21	88
56	4	4	4	4	4	4	24	100
57	4	4	4	4	4	4	24	100
58	4	4	4	0	0	0	12	50
59	4	4	4	4	4	4	24	100
60	4	4	3	4	2	4	21	88
61	4	4	0	2	2	4	16	67
62	0	0	0	3	4	4	11	46
63	4	4	4	4	4	4	24	100
64	4	4	4	4	4	4	24	100
65	4	4	4	4	4	4	24	100
66	4	4	4	4	4	4	24	100
67	4	4	4	3	4	4	23	96
68	4	4	4	4	1	4	21	88
69	4	4	4	4	3	4	23	96

70	4	4	4	4	3	4	23	96
71	4	4	4	4	2	4	22	92
72	4	4	0	3	3	0	14	58
73	4	4	4	4	4	4	24	100
74	4	4	4	4	4	4	24	100

Performance

	Number of labs	% total labs
Satisfactory performers (≥ 70 % of adequate responses)	69	93
Unsatisfactory performers (< 70 % adequate responses and/or critical error)	3	4
Partial and non-submitters	3	4

Overall Proficiency

Sample	Diagnosis	Total (%)
QLOU-US-2023-A	Normal	97
QLOU-US-2023-B	Methylmalonic Aciduria	99
QLOU-US-2023-C	Ornithine Transcarbamylase Deficiency	93
QLOU-US-2023-D	Barth Syndrome	98
QLOU-US-2023-E	3 Methylglutaconyl-CoA hydratase Deficiency	85
QLOU-US-2023-F	Alkaptonuria	99

10. Information from the Executive Board and the Scientific Advisory Board

- **Urine samples:** we remind you that every year, each participant must provide to the scheme organizer at least 300 ml of urine from a patient affected with an established inborn error of metabolism or “normal” urine, together with a short clinical report. Each urine sample must be collected from a single patient (don't send urine spiked with pathological compounds). Please don't send a pool of urines, except if urine has been collected on a short period of time from the same patient.

As soon as possible after collection, the urine sample must be heated at 50 °C for 20 minutes. Make sure that this temperature is achieved in the entire urine sample, not only in the water bath. Then aliquot the sample in 10 ml plastic tubes (minimum 48 tubes), add stoppers and freeze. Be careful to constantly homogenize the urine while aliquoting the sample. Send the aliquots on dry ice by rapid mail or express transport to:

Mrs C Scott and Miss S Colyer
NHS
Department of Clinical
Chemistry and Newborn Screening
The Children's Hospital
Sheffield
S10 2TH
United Kingdom

Please send us an e-mail on the day you send the samples.

11. Tentative schedule and 2024

Sample distribution	7 th February 2024
Start of analysis of Survey 2024/1 Website open	7 th May 2024
Survey 2024/1 - Results submission	28 th May 2024
Survey 2024/1 - Reports	June 2024
Start of analysis of Survey 2023/2 Website open	19 th August 2024
Survey 2024/2 – Results submission	09 th September 2024
Survey 2024/2 - Reports	October 2024
Annual Report 2024	January 2025

12. ERNDIM certificate of participation

A combined certificate of participation covering all EQA schemes will be provided to all participants who take part in any ERNDIM scheme.

13. Questions, Comments and Suggestions

If you have any questions, comments or suggestions please address to the Scientific Advisor of the scheme, Mrs C Scott and/or to the ERNDIM Administration Office (admin@erndim.org)

Date of report, 2024-01-20

Name and signature of Scientific Advisor



Mrs C Scott and Miss S Colyer
NHS
Department of Clinical Chemistry and Newborn Screening
The Children's Hospital Sheffield
S10 2TH
United Kingdom

APPENDIX 1. Change log (changes since the last version)

Version Number	Published	Amendments
1	02 February 2024	2023 annual report published

END