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Qualitative Organic Acids

Centre: United Kingdom

Final Report 2024

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 2024, 75 labs participated to the Qualitative Organic Acid Scheme, Sheffield.

1. Geographical distribution of participants

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

Country	Number of participants
Australia	6
Belgium	7
Finland	1
Germany	1

[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.

Hungary	2
Ireland	1
Israel	3
Japan	6
Malaysia	3
New Zealand	1
Norway	1
Pakistan	1
Poland	3
Slovakia	1
South Africa	2
Sweden	2
United Kingdom	16
United States	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 2024 were human urine samples. Four patient sample donations, and one control sample donation were collected through Sheffield Children's NHS Foundation Trust, Sheffield UK. A sample was also 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 7, 2024: shipment of samples by CSCQ
- May 7, 2024: analysis start and website submission 1st round (A-C)
- May 28, 2024: results submission deadline
- August 26, 2024: analysis start and website submission 2nd round (D-F)

- Sep 9, 2024: deadline for result submission (Survey 2)
- July 23, 2024: report of Survey 1 by e-mail
- October 17, 2024: report of Survey 2 by e-mail
- January 31, 2025: annual report with scoring.

5. Results

	Survey 1	Survey 2
Receipt of results	73	73
No answer	2	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 may 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 may 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 may 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 Judith Garcia, QLOU SA, Barcelona. At the SAB meeting in November 2024 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 2024, the SAB decided that critical error would be awarded for sample A (MCADD) if the diagnosis was missed.

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. No performance support letters will be sent by the Scheme Advisor for 2024. Four critical error letters will be sent out for this scheme for 2024. 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%). However, for 2024, due to an error with the clinical details on the CSCQ website at the time of results submission, for samples B and C poor performance criteria have been excluded in the overall score. Subsequently, for 2024 there were no poor performers by score.

8. Results of samples and evaluation of reporting

8.1. Patient A

24-05-OUS Medium-chain acyl-CoA dehydrogenase deficiency. (MCADD).

Patient details provided to participants.
Presumptive positive picked up on NBS, unwell on day 5 of life. Sample taken when well. Female aged 5 years.

Patient details

This sample was donated from a patient diagnosed with Medium-Chain Acyl-CoA Dehydrogenase Deficiency (MCADD).

Analytical performance

	Number of labs:
Hexanoyl Glycine	70
Suberyl or Phenyl propionyl Glycine	62
No abnormal metabolites	3

Diagnosis / Interpretative proficiency

	Number of labs:
MCADD	69
Normal	4

Recommendations
Molecular analysis *ACADM* gene, urine organic acids, metabolic referral, avoid fasting.

Scoring
Analytical:

Hexanoyl Glycine & Suberyl Glycine or Phenyl Propinoyl Glycine	2 points

Clinical:

MCADD	2 points

Overall impression
This was a good scoring sample with 95% of the participants identifying MCADD. Four participants missed MCADD as a diagnosis. The organic acid profile was typical for MCADD with increased Hexanoyl Glycine, Suberyl Glycine and Phenyl Propinoyl Glycine. Missing the diagnosis of MCADD in this sample was discussed at the Scientific Advisory Board held in November 2024 in Leiden and it was deemed that this would be classed as a Critical Error. Four participants were awarded Critical Error.

8.2. Patient B

24-05-OUS Maple Syrup Urine Disease.

Patient details provided to participants.

The clinical details circulated with the sample were incorrect on the participants website 'recurrent abdominal pain, vegan diet, 8 year old female:

The clinical details that should have been circulated with this sample were 'developmental delay' 4 year old male.

Patient details.

This sample was donated from a patient with diagnosed Maple Syrup Urine Disease who was well at the time of sampling.

Analytical performance

	Number of labs:
Branched-chain oxo and hydroxyacids.	72

Diagnosis / Interpretative proficiency

	Number of labs:
MSUD	66
Other disorder	6
No disorder	1

Recommendations

Follow up with plasma amino acids and molecular analysis of E1 alpha BCKDH1A, E1 beta BCKDH1B, E2 DBT, E3 DLD.

Scoring

Analytical:

Branched-chain oxo and hydroxyacids	2 points

Clinical:

MSUD	2 points

Overall impression

This was a good scoring survey with nearly all participants identifying the correct metabolites for the diagnosis of MSUD. The vast majority concluding this sample was from a patient with MSUD as the most likely diagnosis. Three participants were very specific in their interpretation of the enzyme/gene defect opting for the E3/DLD or E2 subunit only.

Because the incorrect clinical details had been posted on the CSCQ website the score for this sample was excluded for poor performance. Overall scoring of the sample also considered the incorrect clinical details provided at the time of results submission.

8.3 Patient C

24-05-OUS No known metabolic defect, dietary vitamin B12 deficiency.

Patient details provided to participants.

The clinical details circulated with the sample were incorrect on the participants website: Extra pyramidal signs and chronic kidney disease, male, 2 years old.

The clinical details that should have been circulated with this sample were 'Recurrent abdominal pain, 8-year-old, female on a diary free diet.'

Patient details

This sample was donated by a healthy child noted to have dairy milk intolerance. However, post donation this child was diagnosed with dietary vitamin B12 deficiency. This is a child with no known underlying metabolic defect.

Analytical performance

	Number of labs:
No significant abnormality	66
Increased MMA	4
Other	3

Diagnosis / Interpretative proficiency

Clinical:

	Number of labs:
Normal	69
Possible B12 deficiency	4

Recommendations

Recommendations were directed towards investigations for the clinical symptoms provided.

Scoring

Analytical:

Normal/slightly increased MMA	2 points

Clinical:

Normal/B12 deficiency	2 points

Overall impression

This sample was donated from a patient with no known metabolic disease at the time of sampling, who subsequently was diagnosed with dietary vitamin B12 deficiency. Scoring reflected the clinical presentation of this child. The clinical details provided with this sample were incorrect at the time of results submission. Due to the incorrect details being given on the website the results of this sample were excluded from unsatisfactory performance.

8.4 Patient D

QLOU-US-2024-D Ethylmalonic aciduria

Patient details provided to participants.
Developmental delay, 4 year old, male

Patient details
This is a historical sample donated from a patient with persistent ethylmalonic acid excretion. Monitored and treated by the metabolic team.

Analytical performance

	Number of labs:
Ethylmalonic Acid increased	71
Normal EMA excretion	2

Diagnosis / Interpretative proficiency

	Number of labs:
SCADD Deficiency /Ethylmalonic encephalopathy	71
Mitochondrial respiratory chain	
Other	2

Recommendations
Participants suggested plasma acylcarnitines and molecular analysis of the various genes associated with increased excretion of ethylmalonic acid (ACADS, ETHE1, ETFDH/A/B etc.).

Scoring
Analytical:

Ethylmalonic acid	2 points

Clinical:

Any condition associated with increased EMA	2 points

Overall impression
Most of the participants identified the increased ethylmalonic acid excretion. A definitive diagnosis could not be given in this patient, but appropriate investigations and recommendations were scored.

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8.5 Patient E

QLOU-US-2024-E Methylmalonic Acidaemia

Patient details provided to participants.
Extrapyramidal signs, chronic kidney disease, 2 year old male.

Patient details
This sample was donated from a patient with methylmalonic acidaemia (methylmalonyl-CoA mutase (MUT) deficiency).

Analytical performance

	Number of labs:
Methylmalonic acid	73
Methyl citrate	73

Diagnosis / Interpretative proficiency

	Number of labs:
Methylmalonic Acidaemia	73

Recommendations
Acylcarnitine analysis, assessment of vitamin B12 status and mutational analysis of the MMUT gene to confirm the diagnosis. Plasma amino acids and total homocysteine to exclude a cobalamin defect. Trial of vitamin B12 supplementation to assess responsiveness.

Scoring
Analytical:

Methylmalonic acid	1 point
Methyl citrate	1 point

Interpretative:

Methylmalonic Acidaemia	2 points

Overall impression
This sample scored 100 percent diagnostic and analytical proficiency and was the highest scoring sample for 2024.

8.6 Patient F

QLOU-US-2024-F Aromatic L-amino acid decarboxylase deficiency.

Patient details provided to participants.

Predominantly truncal hypotonia and intermittent dystonic posturing. On treatment. 8-year-old male.

Analytical performance

	Number of labs:
Vanillactate	60
N-acetyl Tyrosine	71

Diagnosis / Interpretative proficiency

Aromatic L-amino acid decarboxylase deficiency	61
Tyrosinaemia	6
Normal	5
Other	2

Recommendations

Biogenic amines (CSF), enzyme studies, mutational analysis of the DDC gene.

Scoring

Analytical:

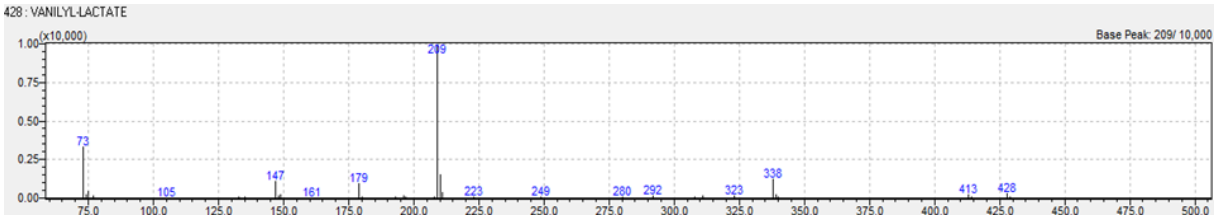
Vanillactate	2 points

Clinical:

AADC	2 points

Overall impression

This was a difficult sample taken from a patient with previously diagnosed Aromatic L-amino acid decarboxylase deficiency having undergone intracerebral gene therapy with the additional complication of parenteral nutrition. The overall proficiency for this difficult sample was good at eighty-four percent, and a significant improvement compared to the last circulation of an AADC sample in which diagnostic proficiency was only 56 percent. The ion spectrum for Vanillactate has been included below.



9. Scores of participants

All data transfer, the submission of data as well as the request and viewing of reports proceed via the QLOU-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 in the table below.

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 MCADD			Patient B MSUD.			Patient C No evidence for a metabolic defect.			
	A	I	Total	A	I	Total	A	I	Total	Total
1	2	2	4	2	2	4	2	2	4	12
2	0	0	0	2	2	4	2	2	4	8
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	1	3	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	0	0	0	8
21	2	2	4	2	2	4	2	2	4	12
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	1	1	2	10
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	2	2	4	12
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	1	3	0	0	0	7
42	1	0	1	2	2	4	2	2	4	9
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	2	2	4	12
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	2	2	4	2	2	4	2	2	4	12
50	2	2	4	2	2	4	2	2	4	12
51	2	2	4	2	0	2	2	1	3	9
→ **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	2	4	2	0	2	2	2	4	10
56	2	2	4	2	2	4	2	2	4	12
57	2	2	4	2	2	4	2	0	2	10
58	2	2	4	2	2	4	2	2	4	12
59	2	2	4	2	2	4	2	1	3	11

60	2	2	4	2	0	2	2	2	4	10
61	0	0	0	0	0	0	0	0	0	0
62	2	2	4	2	2	4	2	2	4	12
63	2	2	4	2	0	2	2	2	4	10
64	2	2	4	2	2	4	2	2	4	12
65	0	0	0	2	2	4	2	2	4	8
66	2	2	4	2	2	4	2	2	4	12
67	2	2	4	2	2	4	2	2	4	12
68	0	0	0	2	0	2	2	2	4	6
69	2	2	4	2	2	4	2	2	4	12
70	2	2	4	2	2	4	2	2	4	12
71	0	0	0	0	0	0	0	0	0	0
72	1	0	1	2	2	4	2	2	4	9
73	2	2	4	0	0	0	2	2	4	8
74	2	2	4	2	2	4	2	2	4	12
75	2	2	4	2	2	4	2	2	4	12

Detailed scores – Round 2

Lab n°	Patient D			Patient E			Patient F			
	A	I	Total	A	I	Total	A	I	Total	Total
1	2	2	4	2	2	4	0	0	0	8
2	2	2	4	2	2	4	0	2	2	10
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	0	0	0	8
15	0	0	0	2	2	4	2	2	4	8
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	0	0	0	8
21	2	2	4	2	2	4	2	2	4	12
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	0	0	0	8
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	2	2	4	12
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	2	2	4	2	0	2	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	2	2	4	12
46	2	2	4	2	2	4	2	2	4	12
47	2	2	4	2	2	4	0	0	0	8
48	2	2	4	2	2	4	2	2	4	12
49	2	2	4	2	2	4	2	2	4	12
50	0	0	0	2	2	4	2	2	4	8
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	0	0	0	8
55	2	2	4	2	2	4	0	0	0	8
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	2	4	12
61	0	0	0	0	0	0	0	0	0	0
62	2	2	4	2	2	4	0	0	0	8
63	2	1	3	2	2	4	2	2	4	11
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	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	0	0	0	8
71	0	0	0	0	0	0	0	0	0	0
72	2	2	4	2	2	4	2	2	4	12
73	2	2	4	2	2	4	0	2	2	10
74	2	2	4	2	2	4	0	0	0	8
75	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	0	20	83
2	0	4	4	4	4	2	18	75
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	3	4	4	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	0	20	83
15	4	4	4	0	4	4	20	83
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	0	4	4	0	16	67
21	4	4	4	4	4	4	24	100
22	4	4	4	4	4	4	24	100
23	4	4	4	4	4	4	24	100
24	4	4	2	4	4	4	22	92
25	4	4	4	4	4	4	24	100
26	4	4	4	4	4	4	24	100
27	4	4	4	4	4	4	24	100
28	4	4	4	4	4	0	20	83
29	4	4	4	4	4	4	24	100
30	4	4	4	4	4	4	24	100
31	4	4	4	4	4	4	24	100
32	4	4	4	4	4	4	24	100

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	4	4	24	100
38	4	4	4	4	4	4	24	100
39	4	4	4	4	4	4	24	100
40	4	4	4	4	4	4	24	100
41	4	3	0	4	4	4	19	79
42	1	4	4	4	4	2	19	79
43	4	4	4	4	4	4	24	100
44	4	4	4	4	4	4	24	100
45	4	4	4	4	4	4	24	100
46	4	4	4	4	4	4	24	100
47	4	4	4	4	4	0	20	83
48	4	4	4	4	4	4	24	100
49	4	4	4	4	4	4	24	100
50	4	4	4	0	4	4	20	83
51	4	2	3	4	4	4	21	88
→ **52**	4	4	4	4	4	4	24	100
53	4	4	4	4	4	4	24	100
54	4	4	4	4	4	0	20	83
55	4	2	4	4	4	0	18	75
56	4	4	4	4	4	4	24	100
57	4	4	2	4	4	4	22	92
58	4	4	4	4	4	4	24	100
59	4	4	3	4	4	4	23	96
60	4	2	4	4	4	4	22	92
61	0	0	0	0	0	0	0	0
62	4	4	4	4	4	0	20	83
63	4	2	4	3	4	4	21	88
64	4	4	4	4	4	4	24	100
65	0	4	4	4	4	4	20	83
66	4	4	4	4	4	4	24	100
67	4	4	4	3	4	4	23	96
68	0	2	4	4	4	4	18	75

69	4	4	4	4	4	4	24	100
70	4	4	4	4	4	0	20	83
71	0	0	0	0	0	0	0	0
72	1	4	4	4	4	4	21	88
73	4	0	4	4	4	2	18	75
74	4	4	4	4	4	0	20	83
75	4	4	4	4	4	4	24	100

Performance

	Number of labs	% total labs
Satisfactory performers (≥ 70 % of adequate responses)	69	92
Unsatisfactory performers (< 70 % adequate responses and/or critical error)	4	5
Partial and non-submitters	2	3

Overall Proficiency

Sample	Diagnosis	Total (%)
QLOU-US-2024-**A**	Medium-chain acyl-CoA dehydrogenase deficiency	**94**
QLOU-US-2024-**B**	Maple Syrup Urine Disease	96
QLOU-US-2024-**C**	No metabolic disease	95
QLOU-US-2024-**D**	Ethylmalonic Aciduria	97
QLOU-US-2024-**E**	Methylmalonic Aciduria	100
QLOU-US-2024-**F**	Aromatic L-amino decarboxylase deficiency	84

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 2025

Sample distribution	5th February 2025
Start of analysis of Survey 2025/1 Website open	6th May 2025
Survey 2025/1 - Results submission	27th May 2025
Survey 2025/1 - Reports	June 2025
Start of analysis of Survey 2025/2 Website open	18th August 2025
Survey 2025/2 – Results submission	08th September 2025
Survey 2025/2 - Reports	October 2025
Annual Report 2025	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.

1. 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, 2025-01-31
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 2025	2024 annual report published

END