ERNDIM Administration Office
c/o EMQN CIC
Third Floor, ICE Building,
3 Exchange Quay, Salford
M5 3ED, United Kingdom.
Tel: +44 161 757 4952
Fax: +44 161 850 1145
Email: admin@erndim.org

Scientific Coordination
Judit Garcia Villoria
Servicio de Bioquímica y Genética Molecular
Sección de Errores Cong del Metab
Hospital Clínic de Barcelona
c/ Mejía Lequerica s/n.Ed Helios III Pl bx
08028, Barcelona
Spain
Email: admin@erndim.org

Scheme Organisation
1) Alessandro Salemma; 2) Rose Defossez
CSCQ, Swiss Center for Quality Control
2 chemin du Petit-Bel-Air
CH-1225 Chêne-Bourg
Switzerland
Email: erndim.survey@cscq.ch

Published: 24th March 2026[1]

Qualitative Organic Acids
Centre: Spain

Final Report 2025

prepared by
Dr. Judit Garcia Villoria and Dr. Margarida Silva

Note: This annual report is intended for participants of the ERNDIM QLOU Barcelona 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 terms and conditions on page18 and the ERNDIM Privacy Policy on www.erndim.org.

1. Introduction

The ERNDIM Qualitative Organic Acids in urine scheme offers urine samples obtained from confirmed patients with confirmed diagnoses to enable laboratories to gain or maintain experience to identify organic acid disorders. The scheme is organised by Judit Garcia Barcelona Scheme in conjunction with CSCQ, the Swiss organisation for quality assurance in medical laboratories.

As in previous years, samples were sent out to cover the spectrum of what is typically observed in the metabolic laboratory. A mix of clearly diagnostic profiles and some more challenging profiles were provided. As in previous years normal profiles were also sent out. The requirement to interpret a normal profile, as such, is as important as correctly identifying abnormal profiles. Correctly identifying a profile as normal can avoid unnecessary further investigation and distress to the patient and family.

In 2025 seventy six laboratories from many different countries participated in the QLOU Barcelona scheme, without any educational participants (0 in 2024, 0 in 2023, 0 in 2022). Educational participants take part in all aspects of the scheme and receive interim reports with scores, but performance is not indicated on the ERNDIM certificate of performance.

Participants and new applicants will distributed between the QLOU-Barcelona, -Heidelberg and -Sheffield schemes which are run separately. The three organising laboratories each participate in the other's scheme by rotation.

[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.

2. Geographical distribution of participants

There are 76 participants with the following geographic distribution:

Country	Number of participants
Argentina	1
Brazil	2
Chile	1
China	1
Colombia	1
Cyprus	1
France	21
Germany	1
Greece	1
Hong Kong	5
India	5

Country	Number of participants
Italy	14
Lebanon	1
Philippines	1
Portugal	2
Qatar	1
Saudi Arabia	1
Singapore	1
Spain	11
Turkey	3
Uruguay	1

3. Design and logistics of the scheme including sample information

The scheme has been designed and planned by Judit García Villoria as Scientific Advisor and coordinated by CSCQ, both appointed by and according to procedures laid down the ERNDIM Board.

As usual, the samples used in 2025 were authentic human urine samples, 5 from affected patients and 1 from healthy individuals. In 2025 CSCQ dispatched the QLOU EQA samples to the scheme participants and provides a website for on-line submission of results and access to scheme reports. Existing QLOU, ACDB, DPT and Urine MPS scheme participants can log on to the CSCQ results submission website at: https://cscq.hcuge.ch/cscq/ERNDIM/Initial/Initial.php
Labelled copies of chromatograms can be uploaded on the CSCQ website.

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

Origin of patients: 3 samples were collected through the Scientific Advisor's institution (Hospital Clínic de Barcelona). Others were donated by the Scientific Advisor for QLOU-Heidelberg (Joachim Janda, Centre for Metabolic Diseases Heidelberg, Germany), the Scientific Advisor for DPT-Switzerland (Déborah Mathis, Inselspital University Hospital Bern, Switzerland) and an ERNDIM participant (Laboratorio de Pesquisa Neonatal, Instituto de la Seguridad Social – BPS, Uruguay)

Patient A: Normal sample
Patient B: 3-Methylcrotonyl-CoA carboxylase deficiency
Patient C: Glutaric aciduria type I
Patient D: Methylmalonic aciduria Mut0
Patient E: Alkaptonuria
Patient F: Propionic acidemia

The samples have been heat-treated. They were pre-analysed in the Scientific Advisors 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. Samples were shipped by DHL, FedEx or the Swiss Post at room temperature. Details regarding stability of samples are provided in the sample package. The samples are stable for the duration of the scheme's submission calendar when stored under defined conditions

To be able to continue this scheme we need a steady supply of new patient samples. Several laboratories have donated samples to the Urine QLOU scheme in the past, for which they are gratefully acknowledged. If you have one or more samples available and are willing to donate these to the scheme,

please contact us at admin@erndim.org. Laboratories which donate samples that are used in the scheme are eligible for a 20% discount on their participation in the QLOU scheme in the following year

4. Tests

Required method is the determination of organic acids

5. Schedule of the scheme

- Febrary 5, 2025: shipment of samples of Survey 1 and Survey 2
- May 6, 2025: analysis start, clinical data available and submission availability on the website (Survey 1)
- May 27, 2025: deadline for result submission (Survey 1)
- October 21, 2025: interim report of Survey 1 available on the website
- August 26, 2024: analysis start, clinical data available and submission availability on the website (Survey 2)
- September 16, 2024: deadline for result submission (Survey 2)
- October 28, 2025: interim report of Survey 2 available on the website
- March, 2025: annual report

6. Results

	Survey 1	Survey 2
Receipt of results	74	72
No answer	2	4

7. Web site reporting

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

- Selection of tests: **don't select a test if you will not perform it**, otherwise the evaluation program includes it in the report.
- Results
  - Give quantitative data as much as possible.
  - 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.**
- Recommendations = **advice for further investigation**.
  - Scored together with the interpretative score.
  - Advice for treatment are not scored.
  - **Don't give advice for further investigation in "Comments on diagnosis"**: it will not be included in the evaluation program.

8. Scoring and evaluation of results

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.

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. For further information, please refer to the Framework for Assessment and Education for Qualitative Schemes on our website (https://eqa.erndim.org/information/view/14)

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 Barcelona 2025 have been also scored by Dr Camilla Scott, from QLOU Shefield. At the SAB meeting in 27th and 28th November, the definitive scores have been finalized. 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 2025, the SAB decided that sample C has to be considered as a critical error for the labs who failed to identify an increase of 3-hydroxyglutaric acid and not give the diagnosis of Glutaric aciduria type I, and not provide any further recommendations to rule out it.

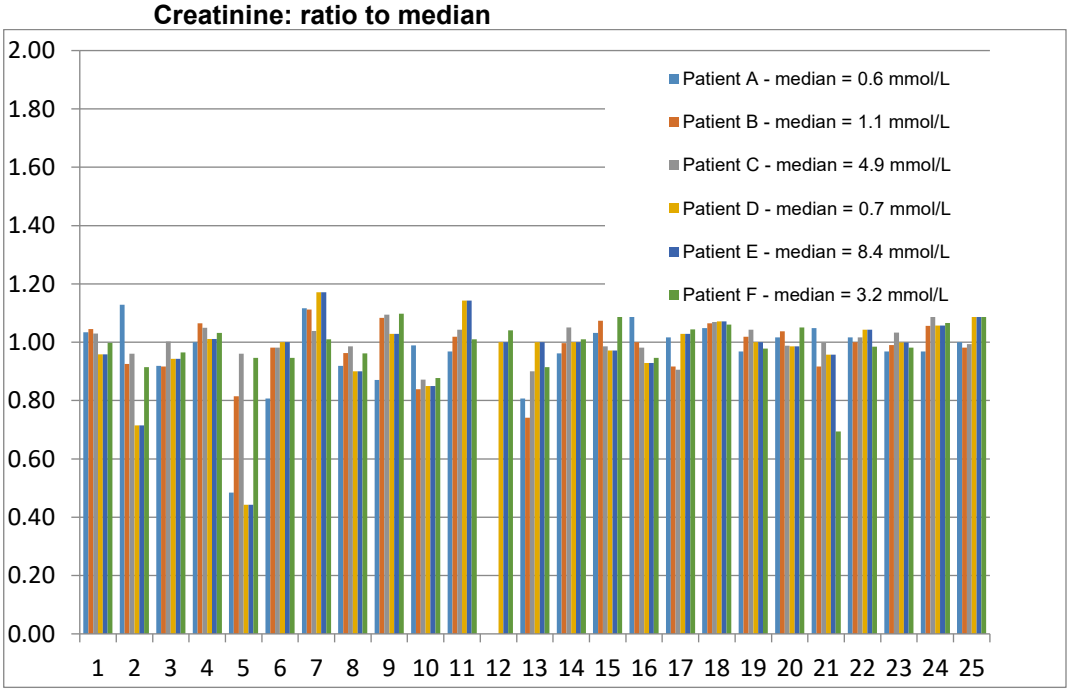
A certificate of participation will be issued for participation and it will be additionally notified whether the participant has received a performance support letter. This performance support letter is sent out if the performance is evaluated as unsatisfactory. One performance support letters will be sent by the Scheme Advisor for 2025. Any partial submitters will receive a letter from the ERNDIM Executive Administrator, Sara Gardner.

8.1. Score for satisfactory performance

At least 17 points from the maximum of 24 (71%). If your laboratory is assigned poor performance and you wish to appeal against this classification please email the ERNDIM Administration Office (erndim@mft.nhs.uk), with full details of the reason for your appeal, within one month receiving your Performance Support Letter.

9. Results of samples and evaluation of reporting

9.1. Creatinine measurement for all samples



9.2. Patient A

Normal sample

Patient details provided to participants

Patient with anorexia and autistic behavior.

Patient details

The urine sample was collected from a voluntary individual. No abnormalities were detected, except for a slight increase of glycerol probably due to external contamination and detection of paracetamol metabolite.

Analytical performance

-74 out of 76 active participants submitted results for Sample A.
-61 laboratories (82%) reported the result as a profile without significant alterations.
-8 participants (11%) detected a paracetamol metabolite, and 7 participants (10%) reported elevated glycerol levels. Some laboratories also noted slight increases or traces of acylglycines.
-One laboratory reported elevated levels of methylmalonic acid, sebacic acid, 3-hydroxybutyric acid, glutaric acid, and fumaric acid, suggesting a possible vitamin B12 deficiency.

Diagnosis / Interpretative proficiency

-67 laboratories (94%) correctly reported the sample as normal, either in the definitive diagnosis or as an alternative diagnosis.
-One participant diagnosed glycerol kinase deficiency as the most probable condition, but did not mention the possibility of contamination or recommend repeating the organic acid profile with a new sample.
-Another participant diagnosed vitamin B12 deficiency or hyperglycinemia based on elevated tiglylglycine. A different laboratory suggested vitamin B12 deficiency due to increased levels of methylmalonic acid, sebacic acid, 3-hydroxybutyric acid, glutaric acid, and fumaric acid.

Recommendations

Some participants recommended analyzing plasma and urine aminoacids, acylcarnitine profile, VLCFA, purines and pyrimidines, creatin metabolism, thiamine concentration, cholestanol, oxysterols, lactate, pyruvate and ammoniemia. In addition, NGS genetic screening.

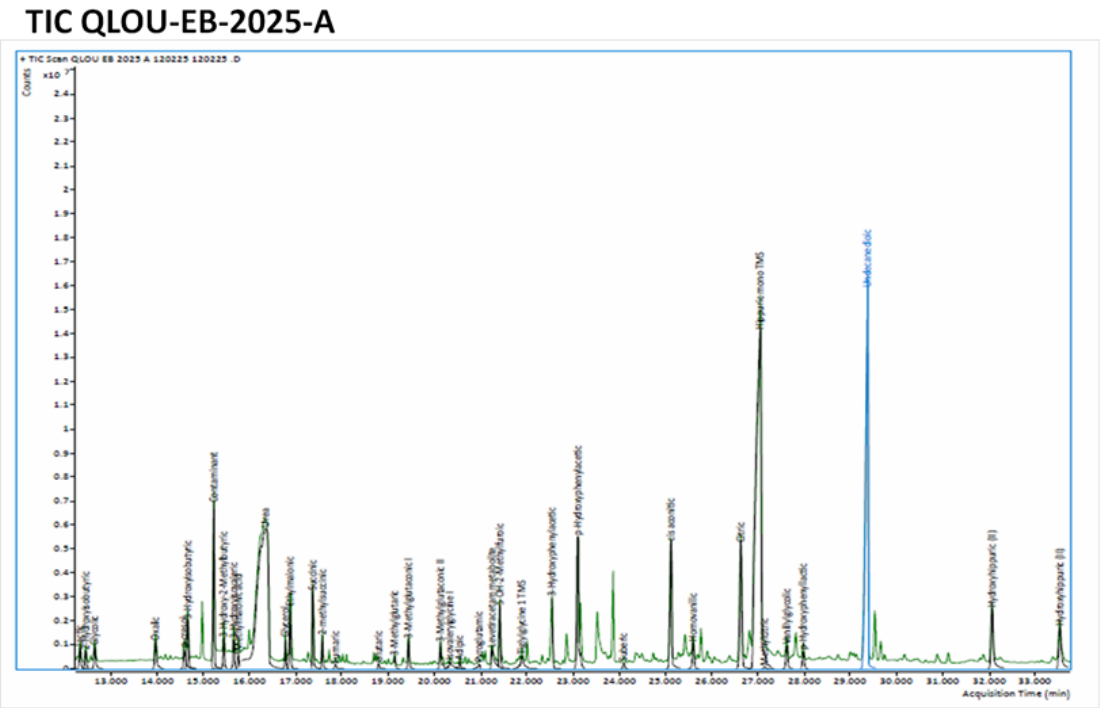
Scoring

- Analytical results: 2 points are awarded for reporting a normal profile or detecting glycerol and/or a paracetamol metabolite.
- Interpretation of results:
  -1 point is awarded if there is a suggestion to repeat the organic acid profile using a new urine sample
  -2 points are awarded for identifying the sample as normal or commenting that the increase in glycerol may be due to contamination.

Overall impression

The overall performance was 98%.

Chromatogram sample A:



Organic acids in urine were extracted with ethylacetate without oximation. After solvent evaporation TMS derivatization with bis-trimethylsilyl-trifluoroacetamide (BSTFA) was performed. The organic acids were analyzed by GC-MS using the 60 m x 0.25 mm ID HP-5MS capillary column.

9.3. Patient B

3-Methylcrotonyl-CoA carboxylase (3MCC) deficiency

Patient details provided to participants

Diagnosed in childhood. Adult – 37-year-old now well.

Patient details

A now 58-year-old man who was diagnosed at the age of 37 after diagnosis of 3-Methylcrotonylglycinuria (3MCC) in his newborn nephew in 2004.

Analytical performance

-74 out of 76 active participants submitted results for Sample B.
-60 participants (81%) detected increases in both key metabolites: 3-hydroxyisovaleric acid and 3 methylcrotonylglycine.
-73 laboratories reported elevated 3-hydroxyisovaleric acid, and 61 laboratories detected increased 3 methylcrotonylglycine.
-14 participants detected only the increase in 3-hydroxyisovaleric acid

Diagnosis / Interpretative proficiency

-59 laboratories (80%) reported 3-methylcrotonyl-CoA carboxylase (3MCC) deficiency as the correct diagnosis.
-9 participants (12%) suggested diagnoses such as biotinidase deficiency, holocarboxylase synthetase deficiency, or multiple carboxylase deficiency, without mentioning 3MCC or recommending further differential diagnosis.
-5 laboratories (6,7%) that only detected 3-hydroxyisovaleric acid diagnosed isovaleric aciduria, despite the absence of elevated isovalerylglycine

Recommendations

The majority of the participants recommended analysis of:
- Biotin in plasma

- Acylcarnitine profile in DBS or plasma
- Biotinidase activity in DBS and serum
- Molecular analysis of *MCC1, MMC2*, *HLCS* and *BTD* genes

Scoring

- Analytical results:

  -1 point is awarded for detecting increased 3-hydroxyisovaleric acid. 1 point is awarded for detecting increased 3-methylcrotonylglycine

  -2 points are given for the detection of 2-hydroxyglutarate.

- Interpretation of results:

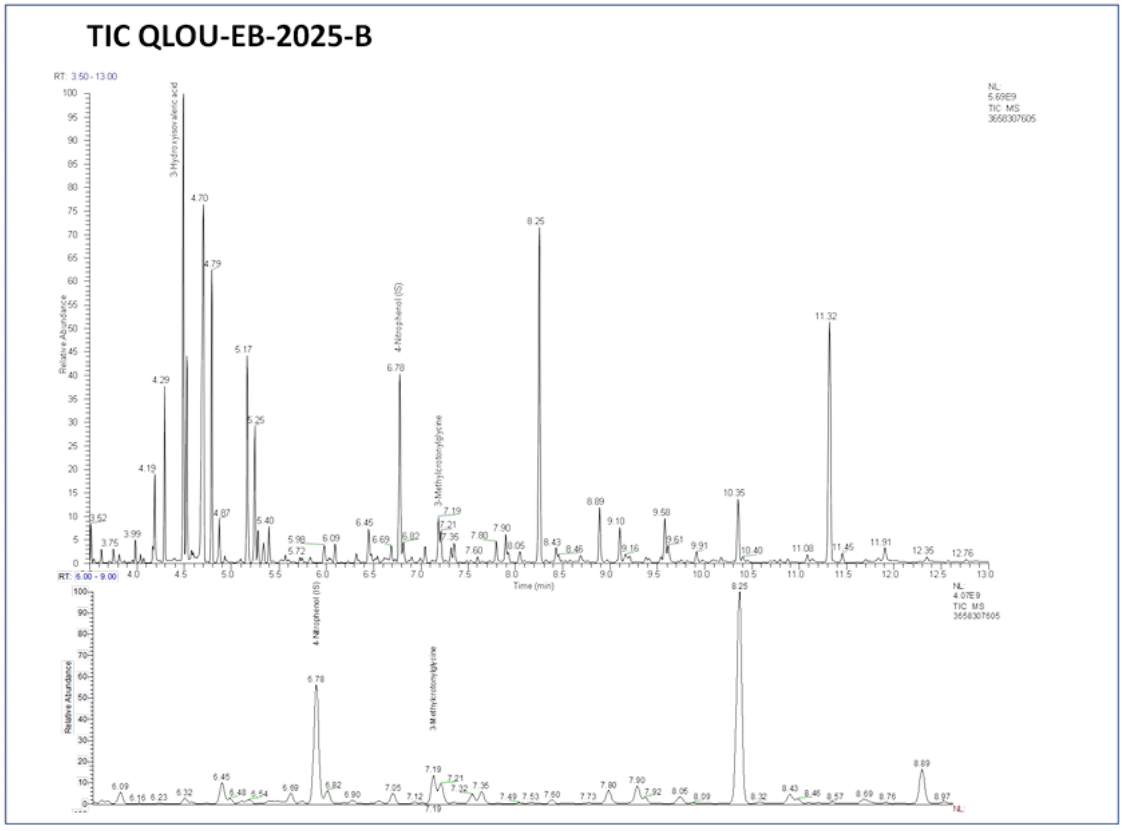
  -1 point is awarded for suggesting biotinidase, holocarboxylase synthetase, or multiple carboxylase deficiencies. An additional 1 point is awarded if the recommendation includes differential diagnosis of 3MCC through enzymatic or molecular studies.

  -2 points are awarded for correctly identifying 3MCC deficiency as the primary or an alternative diagnosis.

Overall impression

The overall performance was 89%

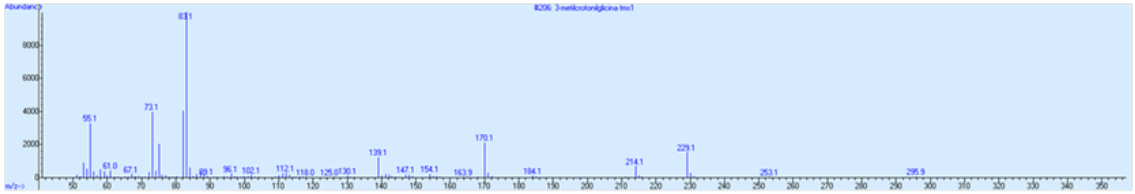
Chromatogram sample B:



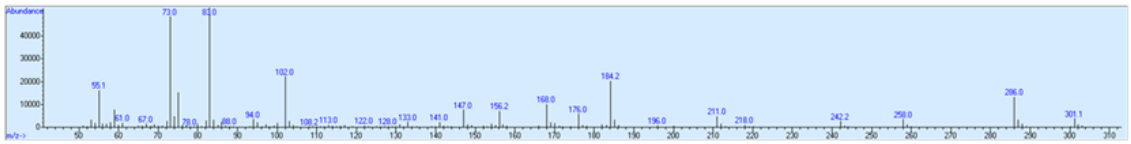
Organic acids in urine were extracted with ethylacetate without oximation. After solvent evaporation TMS derivatization with bis-trimethylsilyl-trifluoroacetamide (BSTFA) was performed. The organic acids were analyzed by GC-MS using the 60 m x 0.25 mm ID HP-5MS capillary column.

Ms spectrum of 3-methylcrotonylglycine

3-methylcrononylglycine mono-TMS



3-methylcrononylglycine di-TMS



9.4. Patient C

Glutaric aciduria type I

Patient details provided to participants

Patient who presented at age 2 with acute encephalopathy.

Patient details

The patient was diagnosed with glutaric aciduria type I during the first episode of acute encephalopathy. The diagnosis was confirmed by mutation analysis of the GCDH gene. The sample was collected at age 25 during treatment.

Analytical performance

-A total of 74 out of 76 active participants submitted results for Sample C.
-All 74 participants (100%) correctly detected an increase in glutarate.
-69 laboratories (93%) also detected an increase in 3-hydroxyglutarate

Diagnosis / Interpretative proficiency

-The majority of laboratories, 73 out of 74 (99%), correctly reported glutaric aciduria type I or glutaryl-CoA dehydrogenase deficiency as the diagnosis.
-One participant incorrectly reported a diagnosis of glutaric aciduria type II.

Recommendations

The majority of the participants recommended analysis of acylcarnitines in plasma, or urine, plasmatic aminoacid measurement and molecular studies of GCDH gene

Scoring

- Analytical results:

-1 point awarded for detection of increased glutarate.
-1 point awarded for detection of increased 3-hydroxyglutarate

- Interpretation of results: 2 points are awarded for the correct diagnosis of glutaric aciduria type I.

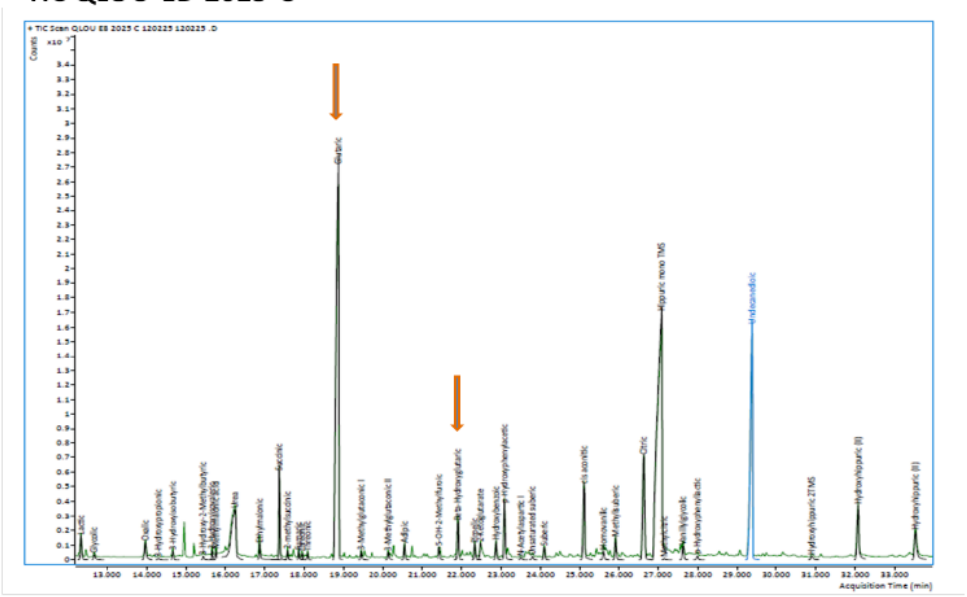
CRITICAL ERRORS: One laboratory reported the case as glutaric aciduria type II but failed to detect the elevated levels of 3-hydroxyglutaric acid and made errors in the clinical recommendations.

Overall impression

The overall performance was 98%.

Chromatogram sample C:

TIC QLOU-EB-2025-C



Organic acids in urine were extracted with ethylacetate without oximation. After solvent evaporation TMS derivatization with bis-trimethylsilyl-trifluoroacetamide (BSTFA) was performed. The organic acids were analyzed by GC-MS using the 60 m x 0.25 mm ID HP-5MS capillary column.

9.5. Patient D

Methylmalonyl-CoA mutase deficiency

Patient details provided to participants

Patient on treatment. Now listless, sleepy, had vomited 2x.

Patient details

The sample was collected from a male with normal pregnancy, birth and infancy. Metabolic crisis at age 2 and 11 with loss of abilities and residual dystonic hemiparesis prompted metabolic investigations resulting in suspicion of classic methylmalonic acidemia. Diagnosis was confirmed by identification of biallelic pathogenic variants in *MMUT.*

Analytical performance

- 72 laboratories of 76 active participants submitted results for sample D.
- In addition to the pathological excretion of key metabolites: Methylmalonic acid, 2-methylcitric acid and 3-hydroxypropionic, high excretion of lactate was present too.
-The 100 % of participants detected the increase of methylmalonate levels.
- 63 laboratories (87,5%) reported the increase of 2-methylcitrate levels.
- 44 participants (61%) detected high amounts of 3-hydroxypropionate levels.
- 37 (51%) laboratories observed an increase of lactate levels..

Diagnosis / Interpretative proficiency

- 71 participants (99%) reported methylmalonic aciduria as the correct diagnosis.
- One laboratory established the diagnosis of combined malonic and methylmalonic aciduria as most likely diagnostic, due to the identification of increase of both malonic and methylmalonic acids. As alternative diagnosis reported malonic aciduria

Recommendations

The majority of the laboratories recommended performing both blood aminoacids and acylcarnitines, and mutation analysis. Some of them directly diagnosed as methylmalonyl-CoA mutase deficiency and recommended measuring enzymatic activity in fibroblasts if the mutations are not found.

Scoring

- Analytical results:
  -1 point is given for the detection of high levels of methylmalonic acid.
  -1 point is given for reporting increase of 2-methylcitrate levels or 3-hydroxypropionic

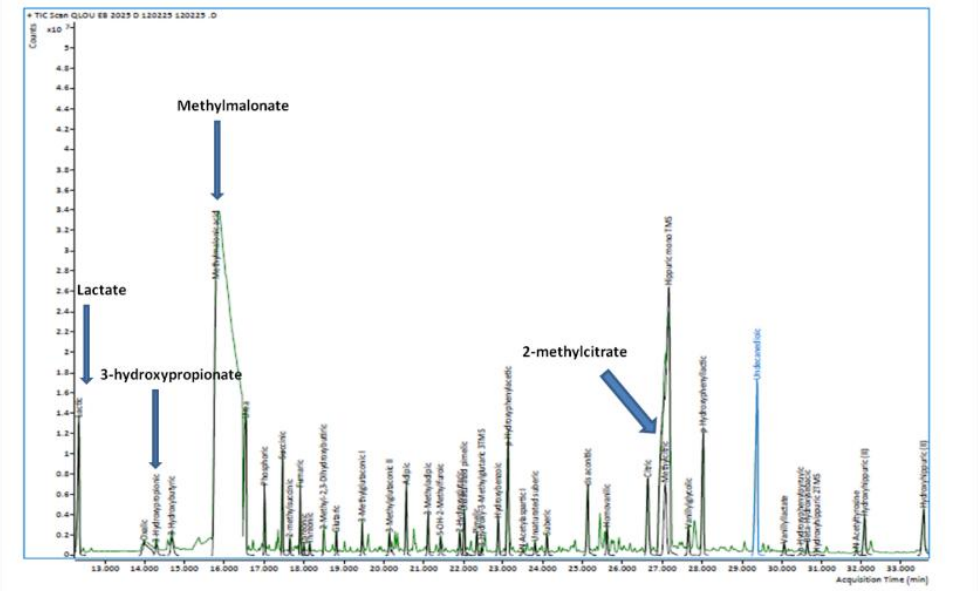
- Interpretative results: 2 points for the correct diagnosis of methylmalonic aciduria as the most likely diagnostic.

Overall impression

The overall performance was 98%.

Chromatogram sample D:

TIC QLOU EB 2025-D



Organic acids in urine were extracted with ethylacetate without oximation. After solvent evaporation TMS derivatization with bis-trimethylsilyl-trifluoroacetamide (BSTFA) was performed. The organic acids were analyzed by GC-MS using the 60 m x 0.25 mm ID HP-5MS capillary column.

9.6. Patient E

Alkaptonuria

Patient details provided to participants

Sine one month of age the mother noticed diapers stained black in several opportunities. Currently under treatment.

Patient details

The sample was from a patient with Alkaptonuria at the age of 10 years-old and under treatment. Clinically, the mother noticed diapers stained black in several opportunities since he was 1 month of age. Several studies were performed on blood and urine, but it was not possible to confirm a causal diagnosis, during infancy.
When he was 3 years of age, the analysis of homogentisic acid was performed and the diagnosis was established. He did not have development delay or cognitive impairment. He receives vitamin C and follow protein-limited diet.

Analytical performance

-72 laboratories of 76 active participants submitted results for sample E.
-100 % of the laboratories identified the increased homogentisic acid.

Diagnosis / Interpretative proficiency

-100% of participants reported Alkaptonuria as correct diagnosis.

Recommendations

The majority of the laboratories recommended performing molecular study of HGD gene and monitoring of plasma tyrosine levels if the patients is on Nitisinone treatment.

Scoring

- Analytical results: 2 points are given for the detection of homogentisic acid.
- Interpretation of results: 2 points are given for the diagnosis of Alkaptonuria.

Overall impression

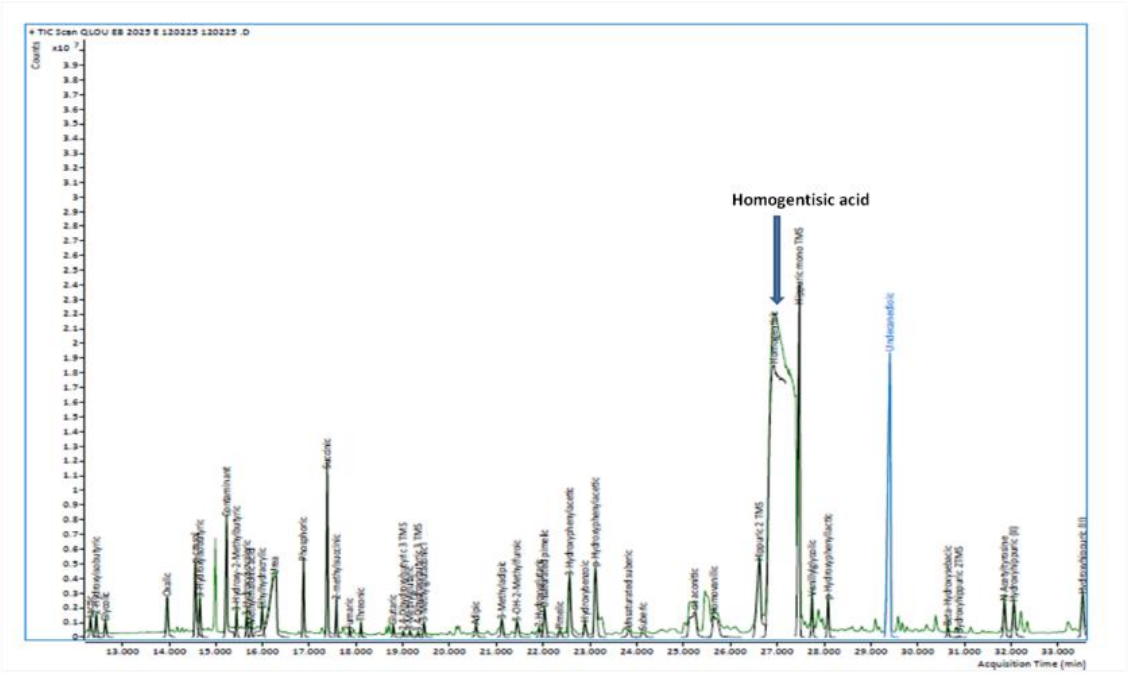
The overall performance was 100%.

Multiple distributions of similar samples
The same proficiency was obtained in other circulation in 2022.

Circulation	2022
Overall performance	97%

Chromatogram sample E:

TIC QLOU EB 2025-E



Organic acids in urine were extracted with ethylacetate without oximation. After solvent evaporation TMS derivatization with bis-trimethylsilyl-trifluoroacetamide (BSTFA) was performed. The organic acids were analyzed by GC-MS using the 60 m x 0.25 mm ID HP-5MS capillary column.

9.7. Patient F

Propionic aciduria

Patient details provided to participants
Patient with severe intellectual deficit and recurrent epilepsy. The diagnosis was performed in the first year of life. Currently under treatment.

Patient details
The sample is from a patient diagnosed of propionic aciduria at one year of age presenting with severe intellectual deficit and recurrent epilepsy. Currently is an adult patient under treatment.

Analytical performance
- 72 laboratories of 76 active participants submitted results for sample F.
- In addition to the pathological excretion of key metabolites: 3-hydroxypropionate, 2-methylcitrate and propionylglycine, the increase of other organic acids from isoleucine catabolic pathway such a tiglylglycine and 2-methyl-3-hydroxybutyrate, were detected. High excretion of lactate and fumarate were present too. The presence of drug metabolites such as ethosuximide and carbamazepine can also be detected.
- 70 participants (97%) detected the increase of 3-hydroxypropionate and 2-methylcitrate.

- 69 laboratories (96%) reported increased propionylglycine levels.
- 63 participants (88%) reported increased levels of tiglylglicine.
-In addition, 20% of the labs reported an increase of lactate and fumarate and of ethosuxmide or carbamazepine metabolites.

Diagnosis / Interpretative proficiency
- 70 laboratories (97%) reported propionic aciduria as the correct diagnosis.
- One laboratory established the diagnosis of multiple carboxylase deficiency or biotinidase deficiency. Other participant reported the diagnosis of multiple carboxylase deficiency or Carbonic anhydrase VA (CA-VA) deficiency. In any case they put the alternative diagnosis of propionic acidemia.

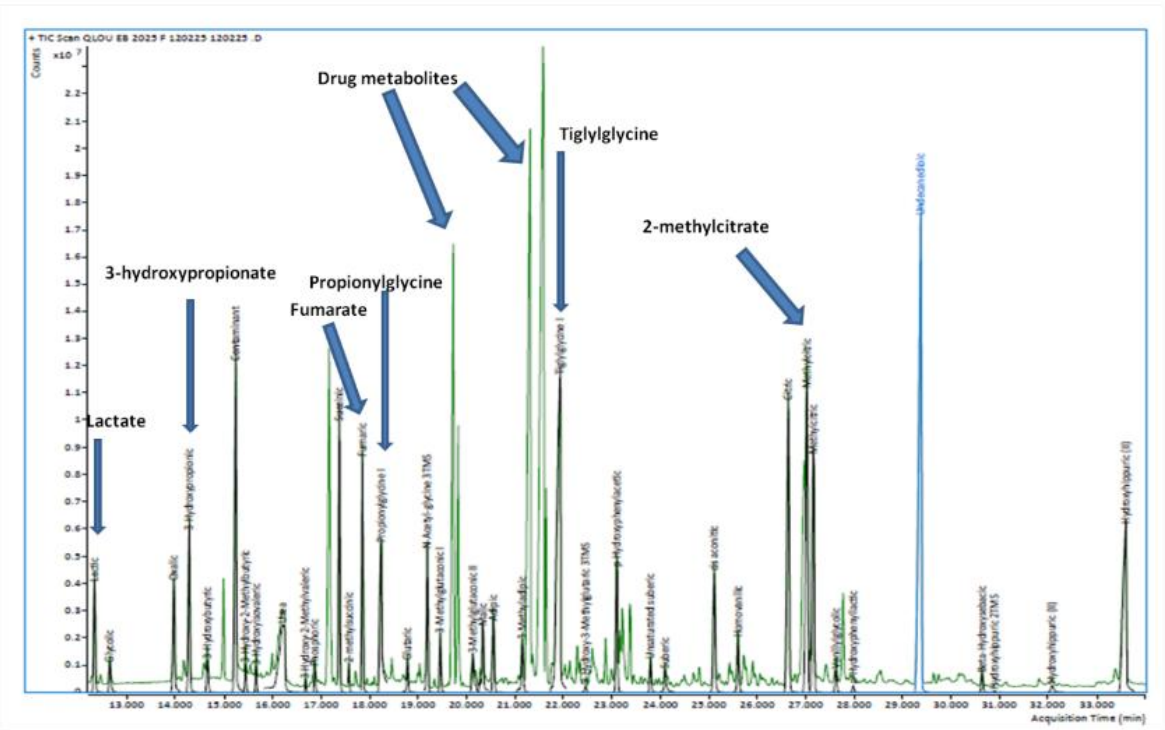
Recommendations
The majority of the laboratories recommend performing both blood aminoacids and acylcarnitines, and mutation analysis in *PCCA* and *PCCB* genes. If mutations are not found, the propionyl-CoA carboxylase activity can be measured in fibroblasts.

Scoring
- Analytical results:
  -1 point is given if a key metabolite is missed.
  -2 points are given for detecting the three key metabolites: 3-hydroxypropionate, 2-methylcitrate and propionylglycine.
- Interpretation of results:
  - 1 point is given when the correct diagnosis is not reported but with the recommendations it would be able to get
  - 2 points for the correct diagnosis of propionic acidemia as the most likely diagnostic or alternative.

Overall impression
The overall performance was 97%.

Chromatogram sample F:

TIC QLOU EB 2025-E



Organic acids in urine were extracted with ethylacetate without oximation. After solvent evaporation TMS derivatization with bis-trimethylsilyl-trifluoroacetamide (BSTFA) was performed. The organic acids were analyzed by GC-MS using the 60 m x 0.25 mm ID HP-5MS capillary column.

10. 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 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			Patient B			Patient C			Total
	Normal sample			3-Methylcrotonyl-CoA carboxylase deficiency			Glutaric aciduria type I			
	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	1	**3**	1	1	**2**	2	2	**4**	**9**
7	2	2	**4**	2	2	**4**	2	2	**4**	**12**
8										**No submission**
9	2	2	**4**	2	1	**3**	2	2	**4**	**11**
10	2	2	**4**	2	2	**4**	2	2	**4**	**12**
11	2	2	**4**	2	2	**4**	1	2	**3**	**11**
12	2	2	**4**	1	0	**1**	2	2	**4**	**9**
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**	1	1	**2**	2	2	**4**	**10**
16	2	2	**4**	2	1	**3**	2	2	**4**	**11**
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	0	0	**0**	2	2	**4**	2	1	**3**	**7**
21	2	2	**4**	2	1	**3**	2	2	**4**	**11**
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**	1	1	**2**	2	2	**4**	**10**
27	2	2	**4**	2	2	**4**	2	2	**4**	**12**
28	2	2	**4**	1	0	**1**	2	2	**4**	**9**

Lab n°	Patient A			Patient B			Patient C			Total
	Normal sample			3-Methylcrotonyl-CoA carboxylase deficiency			Glutaric aciduria type I			
	A	I	Total	A	I	Total	A	I	Total	
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	1	0	**1**	2	2	**4**	2	2	**4**	**9**
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**	2	2	**4**	**11**
42	2	2	**4**	1	0	**1**	1	2	**3**	**8**
43	2	2	**4**	1	1	**2**	2	2	**4**	**10**
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**	1	2	**3**	2	2	**4**	**11**
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**	1	0	**1**	2	2	**4**	**9**
52	2	2	**4**	2	2	**4**	2	2	**4**	**12**
53	2	2	**4**	1	1	**2**	1	2	**3**	**9**
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**	1	0	**1**	2	2	**4**	**9**
57	2	2	**4**	2	2	**4**	2	2	**4**	**12**
58	2	2	**4**	1	2	**3**	2	2	**4**	**11**
59	2	2	**4**	2	2	**4**	2	2	**4**	**12**
60	2	2	**4**	2	2	**4**	2	2	**4**	**12**
61	2	2	**4**	2	2	**4**	2	2	**4**	**12**
62	2	2	**4**	2	2	**4**	2	2	**4**	**12**
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**	1	0	**1**	**9**
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**

Lab n°	Patient A			Patient B			Patient C			Total
	Normal sample			3-Methylcrotonyl-CoA carboxylase deficiency			Glutaric aciduria type I			
	A	I	Total	A	I	Total	A	I	Total	
70	2	2	**4**	2	2	**4**	2	2	**4**	**12**
71	2	2	**4**	1	1	**2**	2	2	**4**	**10**
72	2	2	**4**	1	2	**3**	1	2	**3**	**10**
73	2	2	**4**	2	2	**4**	2	2	**4**	**12**
74										**No submission**
75	2	2	**4**	2	2	**4**	2	2	**4**	**12**
76	2	2	**4**	2	2	**4**	2	2	**4**	**12**

Detailed scores –Round 2

Lab n°	Patient D			Patient E			Patient F			Total
	Methylmalonic aciduria Mut0			Alkaptonuria			Propionic acidemia			
	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**	1	2	**3**	**11**
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**	1	2	**3**	**11**
12	2	2	**4**	2	2	**4**	2	2	**4**	**12**
13	2	2	**4**	2	2	**4**	1	2	**3**	**11**
14	2	2	**4**	2	2	**4**	2	2	**4**	**12**
15	2	2	**4**	2	2	**4**	1	2	**3**	**11**
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	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**	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**

Lab n°	Patient D			Patient E			Patient F			Total
	Methylmalonic aciduria Mut0			Alkaptonuria			Propionic acidemia			
	A	I	Total	A	I	Total	A	I	Total	
37	1	2	**3**	2	2	**4**	1	2	**3**	**10**
38	1	2	**3**	2	2	**4**	1	2	**3**	**10**
39	2	2	**4**	2	2	**4**	2	1	**3**	**11**
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	2	**4**	**12**
43	1	2	**3**	2	2	**4**	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**	2	2	**4**	2	2	**4**	**12**
48	2	2	**4**	2	2	**4**	2	2	**4**	**12**
49										**No submission**
50	2	2	**4**	2	2	**4**	2	2	**4**	**12**
51	2	2	**4**	2	2	**4**	2	2	**4**	**12**
52	2	2	**4**	2	2	**4**	2	2	**4**	**12**
53										**No submission**
54	1	0	**1**	2	2	**4**	2	2	**4**	**9**
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										**No submission**
59	2	2	**4**	2	2	**4**	2	2	**4**	**12**
60	2	2	**4**	2	2	**4**	2	2	**4**	**12**
61	2	2	**4**	2	2	**4**	2	2	**4**	**12**
62	2	2	**4**	2	2	**4**	2	2	**4**	**12**
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**	1	2	**3**	**11**
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	1	**3**	**11**
71	2	2	**4**	2	2	**4**	2	2	**4**	**12**
72	2	2	**4**	2	2	**4**	2	2	**4**	**12**
73	2	2	**4**	2	2	**4**	2	2	**4**	**12**

Lab n°	Patient D			Patient E			Patient F			Total
	Methylmalonic aciduria Mut0			Alkaptonuria			Propionic acidemia			
	A	I	Total	A	I	Total	A	I	Total	
74										**No submission**
75	2	2	**4**	2	2	**4**	2	2	**4**	**12**
76	1	2	**3**	2	2	**4**	1	2	**3**	**10**

Total scores

Lab nº	A	B	C	D	E	F	Cumulative score	Cumulative score (%)	Performance
1	4	4	4	4	4	4	24	100	
2	4	4	4	4	4	4	24	100	
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	3	2	4	4	4	4	21	88	
7	4	4	4	4	4	4	24	100	
8				4	4	3	11	46	Partial-submission
9	4	3	4	4	4	4	23	96	
10	4	4	4	4	4	4	24	100	
11	4	4	3	4	4	3	22	92	
12	4	1	4	4	4	4	21	88	
13	4	4	4	4	4	3	23	96	
14	4	4	4	4	4	4	24	100	
15	4	2	4	4	4	3	21	88	
16	4	3	4	4	4	4	23	96	
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	0	4	3	4	4	4	19	79	
21	4	3	4	4	4	4	23	96	
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	2	4	4	4	4	22	92	
27	4	4	4	4	4	4	24	100	
28	4	1	4	4	4	4	21	88	
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	1	4	4	4	4	4	21	88	
37	4	4	4	3	4	3	22	92	
38	4	4	4	3	4	3	22	92	
39	4	4	4	4	4	3	23	96	
40	4	4	4	4	4	4	24	100	
41	4	3	4	4	4	4	23	96	
42	4	1	3	4	4	4	20	83	

Lab nº	A	B	C	D	E	F	Cumulative score	Cumulative score (%)	Performance
43	4	2	4	3	4	4	21	88	
44	4	4	4	4	4	4	24	100	
45	4	4	4	4	4	4	24	100	
46	4	3	4	4	4	4	23	96	
47	4	4	4	4	4	4	24	100	
48	4	4	4	4	4	4	24	100	
49	4	4	4				12	50	Partial-submission
50	4	4	4	4	4	4	24	100	
51	4	1	4	4	4	4	21	88	
52	4	4	4	4	4	4	24	100	
53	4	2	3				9	38	Partial-submission
54	4	4	4	1	4	4	21	88	
55	4	4	4	4	4	4	24	100	
56	4	1	4	4	4	4	21	88	
57	4	4	4	4	4	4	24	100	
58	4	3	4				11	46	Partial-submission
59	4	4	4	4	4	4	24	100	
60	4	4	4	4	4	4	24	100	
61	4	4	4	4	4	4	24	100	
62	4	4	4	4	4	4	24	100	
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	1	4	4	3	20	83	CE (Sample C)
67	4	4	4	4	4	4	24	100	
68	4	4	4	4	4	4	24	100	
69	4	4	4	4	4	4	24	100	
70	4	4	4	4	4	3	23	96	
71	4	2	4	4	4	4	22	92	
72	4	3	3	4	4	4	22	92	
73	4	4	4	4	4	4	24	100	
74									Non-submission
75	4	4	4	4	4	4	24	100	
76	4	4	4	3	4	3	22	92	

Performance

	Number of labs	% total labs
Satisfactory performers (≥ 71 % of adequate responses)	71	93.4
Unsatisfactory performers (< 71 % adequate responses and/or critical error)	1	1.3
Partial and non-submitters	4	5.3

Overall Proficiency

Sample ID	Diagnosis	Proficiency (%)
QLOU-EB-2025-A	Normal sample	97
QLOU-EB-2025-B	3-Methylcrotonyl-CoA carboxylase deficiency	89
QLOU-EB-2025-C	Glutaric aciduria type I	97
QLOU-EB-2025-D	Methylmalonic aciduria Mut0	98
QLOU-EB-2025-E	Alkaptonuria	100
QLOU-EB-2025-F	Propionic acidemia	97

11. Annual meeting of participants

A QLOU Participant meeting was held during the ERNDIM symposium in Madrid, Spain on 10th October 2025.

Every two years an on-line meeting to discuss QLOU samples will be organised. The next online QLOU meeting is anticipated to be in 2027.

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

- New **reference materials** are now provided by MCA Laboratories: they are not related to EQA samples anymore. There are two concentration levels for each group of analytes. The most suitable low and high concentration levels are defined by the respective scientific advisors. Analytes and their concentrations will be approximately the same in consecutive batches of control material. These reference materials can be ordered through the MCA website. Participants are encouraged to use them as internal control, but they cannot be used as calibrants. On the website a new section for data management completes the ERNDIM internal Quality Control System. Laboratories have the option to submit results and request reports showing their result in the last run in comparison to defined acceptance limits, their own historical data and the mean of all laboratories using the same batch control material.
- A set of **organic acid mixtures** has been developed by Dr Herman ten Brink in Amsterdam, following request and advice from ERNDIM. The product is currently available at: at: https://www.vumc.com/departments/clinical-chemistry/metaboliclaboratory/organic-synthesis-laboratory/organic-acids-mixture.htm
- **Urine samples**: we remind you that every year, each participant must provide to the scheme organizer at least 200 ml of urine from a patient affected with an established inborn error of metabolism or “normal” urine, together with a short clinical report. If possible, please collect 700 ml of urine: this sample can be sent to all labs participating from the three QLOU schemes. 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. For “normal” urine, the sample must be collected from a symptomatic patient (don’t send urine from your kids!). Send the aliquots on dry ice by rapid mail or express transport to:

  Dr. Judit García Villoria
  Hospital Clínic de Barcelona
  Division of Inborn Errors of Metabolism
  c/Mejía Lequerica s/n Edificio Helios III, pb

08028 Barcelona, Spain
Telephone (+)34 93 2275672
Fax (+)34 93 2275668

13. Reminders

- We remind you that the diagnosis of the sample should be done with the organic acid profile.
- For the normal samples for scoring adequately is very important to enter “Normal profile” in “Key metabolites” as well as, enter “Normal profile” in diagnosis box.
- Reccomendation= advice for further investigation is scored together with the interpretative score. Advice for treatment is not scored. Don’t give advice for further investigation in “Comments on diagnosis”: it will not be included in the evaluation program.

14. Tentative schedule and fee in 2026

Sample distribution	4th February 2026
Start of analysis of Survey 2026/1 Website open	5th May 2026
Survey 2026/1 - Results submission	26th May 2026
Survey 2026/1 - Reports	June 2026
Start of analysis of Survey 2026/2 Website open	17th August 2026
Survey 2026/2 – Results submission	31st August 2026
Survey 2026/2 - Reports	October 2026
Annual Report 2026	January-March 2027

15. 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. For the QLOU scheme this certificate will indicate if results were submitted and whether satisfactory performance was achieved in the scheme.

16. Questions, Suggestions and Complaints

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

Most complaints received by ERNDIM consist of minor misunderstandings or problems with samples, which can usually be resolved via direct contact with the ERNDIM administrative staff. If you wish to file a formal complaint, please email your complaint with details of your issue to admin@erndim.org or contact us through our website at https://www.erndim.org/contact-us/

Date of report, 2026-03-03
Name and signature of Scientific Advisor

Dr. Judit García Villoria
Hospital Clínic de Barcelona
Division of Inborn Errors of Metabolism
c/Mejía Lequerica s/n Edificio Helios III, pb
08028 Barcelona, Spain

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

Version Number	Published	Amendments
1	24 March 2026	2025 annual report published

END