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Acylcarnitines in dried blood spots

Centre: Italy

Final Report 2024

prepared by
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Note: This annual report is intended for participants of the ERNDIM Acylcarnitines in dried blood spots (ACDB) 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 performance of your laboratory, unless ERNDIM is required to disclose performance data by a relevant government agency. For details please see 'ERNDIM Terms and conditions' and the ERNDIM Privacy Policy on www.erndim.org.

1. Introduction

The ERNDIM Acylcarnitine in dried blood spots scheme offers dried blood spots obtained from confirmed patients with confirmed diagnoses to enable laboratories to gain or maintain experience to identify organoacidopathies and fatty acid β-oxidation defects. The scheme is organised by Dr Cristiano Rizzo (Bambino Gesù Children's Hospital, Rome, Italy) 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.

2. Geographical distribution of participants

In 2024 45 laboratories from many different countries participated in the ACDB Rome scheme.
There was no educational participant in 2024 (one in 2019). 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.

[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.

Participants and new applicants will distributed between the Heidelberg, London and Rome acylcarnitine in dried blood spots schemes which are run separately. The three organising laboratories each participate in the other's scheme by rotation.

Country	Number of participants
Belgium	6
Bulgaria	1
Croatia	1
Czech Republic	2
Germany	1
Greece	1
Israel	3
Lebanon	1
Lithuania	2
Malaysia	3
Morocco	1
Portugal	2
Saudi Arabia	1
Singapore	1
Slovakia	1
Slovenia	1
Spain	8
Switzerland	2
Taiwan	1
Turkey	1
United Kingdom	1
United States	4

3. Design and logistics of the scheme including sample information

As usual, the samples used in 2024 were authentic human blood spot samples, 5 from affected patients and one from a healthy individual.

All samples selected by the Scientific Advisor are prepared from 30-50µl of lithium heparin anticoagulated whole blood on Whatman (Schleicher & Schuell) 903™ paper. All samples are obtained following local ethical and consent guidelines.

In 2024 CSCQ dispatched the ACDB 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 scan/chromatograms can be uploaded on the CSCQ website

4. Schedule of the scheme

Time schedule in the 2024 ERNDIM ACDB Rome scheme.

	1st Submission Round	2nd Submission Round
Sample ID's:	ACDB-IR-2024-A ACDB-IR-2024-B ACDB-IR-2024-C	ACDB-IR-2024-D ACDB-IR-2024-E ACDB-IR-2024-F
Shipment of samples	07 February, 2024	
Start of analysis (clinical data available)	12 March 2024	03 June 2024
Reminder for result submission	26 March 2024	17 June 2024
Results submission deadline:	02 April 2024	01 July 2024
Interim reports available on CSCQ website	28 May 2024	14 August 2024

To be able to continue this scheme we need a steady supply of new patient samples. Several laboratories have donated samples to the ACDB 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 ACDB scheme in the following year.

Samples included in the 2024 ERNDIM ACDB Rome scheme.

Survey	Sample no.	Diagnosis
24-05-IR	ACDB-IR-2024-A	CPT1
	ACDB-IR-2024-B	GA1; GCDH
	ACDB-IR-2024-C	cblC
24-07-IR	ACDB-IR-2024-D	Methylmalonic aciduria cblA type
	ACDB-IR-2024-E	SBCADD
	ACDB-IR-2024-F	VLCAD

The scheme format was kept identical to those of previous years. Samples were shipped by regular mail. Details regarding stability of samples are provided in the sample package.
Interim reports were generated by the evaluation program developed by CSCQ.
Origin of patients: all urine samples have been provided by the scheme organizers or specified participants.

5. Results

Returned results in the 2024 ERNDIM ACDB Rome scheme.

	Survey 1	Survey 2
Receipt of results	42	44
No answer	3	1

6. Web site reporting

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

- **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.**
- **Diagnosis**
  - **Don't enter the diagnosis 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.

7. Scoring and evaluation of results

A scoring system was developed in 2012 and approved by the ERNDIM Scientific Advisory Board. Similar to other qualitative (proficiency testing) ERNDIM schemes, the maximum score for a sample is 4 points.
Qualitative results and diagnostic proficiency of the 2024 samples were scored using the criteria given below. These criteria have been set by the Scientific Advisor, approved by the Scientific Advisory Board. The final decision about scoring of the scheme is made in the Scientific Advisory Board (SAB) during the Autumn meeting (November 28-29th, 2024).
General criteria used to score results

Item	Description of scoring criteria	Score
Quantitative results	Correct classification of quantitative results (i.e. normal or increased) according to reference values	1
	Incorrect classification of quantitative results	0
Qualitative results	Correct results according to criteria set for the sample	1
	Incorrect: minimally required results not reported	0
Diagnostic proficiency	Correct according to criteria set for the sample	2
	Partially correct	1
	Unsatisfactory or misleading	0
	Maximum total score	**4**

Starting with the 2014 schemes the concept of 'critical error' is introduced to the assessment of the qualitative schemes. Labs failing to make a correct diagnosis of a sample considered eligible for this category will be deemed not to have reached a satisfactory performance even if their total points for the year is sufficient according to the requirement set by the SAB. The classification of samples to be judged for critical error was undertaken at the SAB meeting held November 28-29th, 2024.

7.1. Score for satisfactory performance

A minimum of 17 points out of a maximum of 24 (≥70%) is necessary for a satisfactory performance. In instances where the SAB agrees that a sample will be classed as an Educational Sample, the scores associated with the sample will be not included in the performance evaluation of the participating laboratories' overall scheme.

We are required to define "Participation" for the purpose of the ERNDIM Annual Certificate which covers all ERNDIM schemes. For this acylcarnitine in dried blood spots scheme we have defined "Participation" as requiring two returns during the year. Failure to meet this requirement will result in the certificate of participation showing 'non-submitter' rather than 'satisfactory' or 'unsatisfactory'.

8. Results of samples and evaluation of reporting

8.1. Patient A

Carnitine Palmitoyl Transferase I Deficency

Patient details provided to participants

Patient with hypoglycemic crises at 6 months of age. He currently has language and learning difficulties at school

Patient details

Analytical performance

45 laboratories who were registered with the Rome section of the Scheme. Results were returned by 42 labs (92%).

Diagnosis / Interpretative proficiency

45 laboratories who were registered with the Rome section of the Scheme. Results were returned by 42 labs (92%).
41/42 (97%) respondents reported a Carnitine Palmitoyl Transferase I Deficency as the most likely diagnosis. One respondent reported LCHAD/MTP as the most likely diagnosis.
2 respondents considered supplementation with carnitine, 2 respondents CPT2, 2 respondents COASY Protein-Associated Neurodegeneration (CoPAN), 4 respondent Medium Chain Acyl-CoA dehydrogenase deficiency, 2 respondent HMG, 1 respondent liver disfunction /renal metabolic acidosis as alternative diagnosis.
41 respondents suggested these follow up test to confirm the diagnosis: 36 respondents suggested the CPT1A mutation analysis, 14 respondents suggested enzyme assay in cultured fibroblasts, 13 respondents suggested plasma carnitine/acylcanitines analysis, 3 respondents suggested urinary carnitine analysis, 6 respondent suggested organic acid analysis, 3 respondents suggested ammonia, 9 respondents liver enzymes and CPK assay. The implementation of appropriate dietary management to avoiding fasting (carbohydrate rich diet, low fat diet supplement with MCT and essential fatty acids) was mentioned by 8 respondents.

8.2. Patient B

Glutaryl-CoA dehydrogenase deficiency (glutaric acidemia type 1)

Patient details provided to participants

Patient presented dystonic spastic hemiparesis at the age of 2 years

Patient details

Analytical performance

45 laboratories who were registered with the Rome section of the Scheme. Results were returned by 42 labs (92%).

Diagnosis / Interpretative proficiency

41/42 (97%) respondents considered glutaryl-CoA dehydrogenase deficiency (glutaric acidemia type I) as the most likely diagnosis. One respondent considered the sample normal.
Suggested follow up test to confirm the diagnosis or guide further investigation were mutation analysis of glutaryl-CoA dehydrogenase (GCDH) gene (n=35), urinary organic acids (n=35), enzyme assay in cultured fibroblast (n=13), plasma carnitine/acylcanitines analysis (n=6),urinary glutarylcarnitine (n=4)
The implementation of appropriate dietary management (carnitine supplementation, lysine restricted diet) was mentioned by 12 respondents. Referral to a metabolic physician was mentioned by 10 respondents

Recommendations

Glutaric acidemia type 1 (GA1) is an autosomal recessive neurometabolic disorder caused by glutaryl-CoAdehydrogenase (GCDH) deficiency (GDD) due to mutations in the GCDH gene (19p13.2). GCDH is involved in Lysine,L-hydroxylysine and L-tryptophane catabolic pathways. Normal levels of glutarylcarnitine (C5DC) in DBS orplasma do not rule out GA1.
A positive MS/MS screening result should be confirmed by one or more alternative techniques, including:
1) Urinary organic acids (glutaric acid and 3-hydroxy-glutaric acid) by GC/MS. GA1 is divided into two arbitrarily defined biochemical subtypes: high excreters of urinary glutaric acid (GA) and low excreters. Urinary organic acid analysis suggests GA1 if it reveals 3-OH-glutaric aciduria with or without increased glutaric acid
2)plasma carnitine/acylcarnitine analysis
3) urinary quantification of glutarylcarnitine
4) The diagnosis is confirmed by identification of biallelic pathogenic variants in GCDH gene or, when molecular genetic test results are uncertain, by detection of significantly reduced activity of the enzyme glutaryl-CoA dehydrogenase (GCDH) in cultured fibroblasts or leukocytes.
Treatment includes: dietary management with carnitine supplementation and lysine restricted diet

8.3. Patient C

Combined methylmalonic aciduria and homocystinuria type cblC

Patient details provided to participants

Patient presented ipotonia, neutropenia and microcephaly at birth. Maculopathy was found at 2 months of age. He currently has intellectual disability and visual impairment.

Patient details

Analytical performance

45 laboratories who were registered with the Rome section of the Scheme. Results were returned by 42 labs (92%).

Diagnosis / Interpretative proficiency

33/42 (78%) respondents considered a disorders of intracellular cobalamin metabolism, 10 respondents suggested a normal profile, 1 respondent suggested mitochondrial acetoacetyl-CoA-thiolase deficiency, 1 respondent suggested long chain Acyl-CoA dehydrogenase deficiency, 1 respondent suggested Carnitine uptake disorder, as the most likely diagnosis.
21 respondents considered other disorders of cobalamin uptake, transport, and metabolism; dietary vitamin B12 deficiency, propionic acidemia or other forms of methylmalonic acidemias, 2 respondents considered Homocystinuria or MTHFR deficiency, 1 respondent considered Cohen syndrome as alternative diagnosis.
Suggested follow up test to confirm the diagnosis or guide further investigation were: genetic analyses of genes related to elevation of C3-carnitine (panel including MMACHC gene and other genes involved in the absorption, transport and intracellular metabolism of cobalamins and propionic pathway disorders) (n=22), urinary organic acids (n=28), plasma total Hcys (n=20), serum Vit. B12 (n=18), plasma aminoacids (n=11), plasma methylmalonate (n=21), cblC complementation studies in fibroblasts (n=2), plasma carnitine/acylcanitines analysis (n=3), plasma, SAM and SAH (n=2)

8.4. Patient D

Methylmalonic aciduria, vitamin B12-responsive, cblA type

Patient details provided to participants

Patient presented with recurrent vomiting, dysphagia nystagmus and hypotonus of the lower limbs at two months of life. He currently has psychomotor retardation and language delay. In treatment with carnitine, vitamin B12 and folate.

Patient details

Analytical performance

45 participants. 2 participants did not provide a response.

Diagnosis / Interpretative proficiency

40/43 (93%) respondents considered a disorders of intracellular cobalamin metabolism, methylmalonic acidemia, or propionic acidemia, as the most likely diagnosis.
1 respondent suggested carnitine uptake deficiency, 1 respondent suggested b-ketothiolase deficency, 1 respondent mt-ATPase6 deficency as the most likely diagnosis.
Suggested follow up test to confirm the diagnosis or guide further investigation were: genetic analyses of genes related to elevation of C3-carnitine, (transport and intracellular metabolism of cobalamins and propionic pathway disorders) (n=34), urinary organic acids (n=40), plasma total Hcys (n=28), serum Vit. B12 (n=21), plasma aminoacids (n=17), plasma methylmalonate(n=16), DBS methylmalonate (n=4)complementation studies in fibroblasts (n=3), plasma carnitine/acylcanitines analysis (n=6), plasmaSAM and SAH (n=3)

8.5. Patient E

Short/Branched-Chain Acyl-CoA Dehydrogenase Deficiency
(2-Methylbutyryl-CoA Dehydrogenase Deficiency)

Patient details provided to participants

Patient presented hyporeactivity, sleepiness and hypertonic crises at 20 days of life. He currently has no symptoms and is on a free diet

Patient details

Analytical performance

45 laboratories who were registered with the Rome section of the Scheme. Results were returned by 43 labs (95%). 6 laboratories reported normal C5-carnitine levels

Diagnosis / Interpretative proficiency

37/43 (86%) respondents considered 2-Methylbutyryl-CoA Dehydrogenase Deficiency (Short/Branched-Chain Acyl-CoA Dehydrogenase Deficiency; SBCADD) or Isovaleryl-CoA Dehydrogenase Deficiency (Isovaleric acidemia;IVA) as the most likely diagnosis.
2 respondents considered normal the acylcarnitine profile.
3 respondents considered Carnitine palmitoyltransferase II deficiency, 1 Very long Chain Acyl-CoA Dehydrogenase deficiency and 1 a 3-Methylcrotoyl-CoA Carboxylase deficency as the most likely diagnosis.
2 respondent suggested in addition, the possibility of false positives due to contamination by pivalate containing drugs or cosmetic products containing neopentanoate
Suggested follow up test to confirm the diagnosis or guide further investigation were mutation analysis of Short/Branched-Chain Acyl-CoA dehydrogenase (ACADSB) or/and Isovaleryl-CoA Dehydrogenase (IVD) genes (n=31), urinary organic acids (n=37), enzyme assay in cultured fibroblast (n=3), plasma carnitine/acylcanitinesanalysis (n=5).
5 respondents performed a II tier test which provided the chromatographyc separation of the C5 isomers(isovalerylcarnitine, 2-methylbutyrilcarnitine, pivaloylcarnitine, valerylcarnitine) on the blood spot.
The implementation of appropriate dietary management (carnitine supplementation, protein restricted diet) was mentioned by 6 respondents. Referral to a metabolic physician was mentioned by 4 respondents.

8.6. Patient F

Very Long Chain Acyl-CoA Dehydrogenase deficiency

Patient details provided to participants

Diagnosis at birth by NBS. Sporadic episodes of myalgia in the lower limbs at the age of 6 years.

Patient details

Analytical performance

45 laboratories who were registered with the Rome section of the Scheme. Results were returned by 44 labs(98%).

Diagnosis / Interpretative proficiency

44/44 (100%) respondents considered Very long Chain Acyl-CoA Dehydrogenase deficiency (VLCADD)as the most likely diagnosis

The alternative differential diagnosis suggested by respondents included:

a) Carnitine-acylcarnitine translocase (CACT) deficiency (n=3)

b) Carnitine Palmitoyl transferase type 2 (CPT2) deficiency (n=11)

c) Mitochondrial trifunctional protein (MTP) deficiency/long-chain 3-Hydroxyacyl-CoA dehydrogenase (LCHAD) deficiency/long-chain 3-Ketoacyl-CoA Thiolase (LCKAT) deficiency (n=2)

d) Multiple Dehydrogenase Deficency (glutaric aciduria type 2) (n=2)

e) TANGO2 (n=1)

Suggested follow up test to confirm the diagnosis or guide further investigation were: mutation analysis of ACADVLgene (n=41), urinary organic acids (n=24), VLCAD activity in fibroblasts and lymphocytes (n=22), plasma carnitine/acylcanitines analysis (n=19)

9. Scores of participants

All data transfer, the submission of data as well as the request and viewing of reports proceed via the ACDB-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 CPT1			Patient B GA1; GCDH			Patient C cblC			
	A	I	Total	A	I	Total	A	I	Total	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	--	--	--	--	--	--	--	--	--	0
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	0	0	0	8
11	2	2	4	2	2	4	2	2	4	12
12	2	2	4	2	2	4	0	0	0	8
13	2	2	4	2	2	4	1	2	3	11
14	2	2	4	2	2	4	0	2	2	10
15	1	2	3	2	2	4	2	0	2	9
16	2	2	4	2	2	4	2	2	4	12
17	--	--	--	--	--	--	--	--	--	0
18	2	2	4	2	2	4	2	2	4	12
19	2	2	4	2	2	4	2	2	4	12
20	1	1	2	2	2	4	0	0	0	6
21	2	2	4	2	2	4	2	2	4	12
22	2	2	4	2	2	4	0	0	0	8
23	2	1	3	2	2	4	0	2	2	9
24	2	2	4	2	2	4	2	2	4	12
25	2	2	4	2	2	4	2	2	4	12

Lab n°	Patient A CPT1			Patient B GA1; GCDH			Patient C cblC			
	A	**I**	**Total**	**A**	**I**	**Total**	**A**	**I**	**Total**	**Total**
26	2	2	4	0	0	0	0	1	1	5
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	0	0	0	8
32	2	2	4	2	2	4	0	0	0	8
33	2	2	4	2	2	4	2	2	4	12
34	1	0	1	2	2	4	2	2	4	9
35	2	2	4	2	2	4	2	2	4	12
36	2	2	4	2	2	4	0	0	0	8
37	2	2	4	2	2	4	2	2	4	12
38	--	--	--	--	--	--	--	--	--	0
39	2	2	4	2	2	4	0	0	0	8
40	2	2	4	2	2	4	2	2	4	12
41	1	2	3	2	2	4	0	0	0	7
42	2	2	4	2	2	4	2	2	4	12
43	2	1	3	2	2	4	0	0	0	7
44	2	2	4	0	1	1	2	2	4	9
45	2	2	4	2	2	4	0	2	2	10

Detailed scores – Round 2

Lab n°	Patient D Methylmalonic aciduria cblA type			Patient E SBCADD			Patient F VLCAD			
	A	I	Total	A	I	Total	A	I	Total	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	0	0	0	2	2	4	8
7	--	--	--	--	--	--	--	--	--	0
8	2	2	4	2	2	4	2	2	4	12
9	2	2	4	0	0	0	2	2	4	8
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	0	1	1	2	2	4	9
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	0	0	0	2	1	3	7
21	0	2	2	2	2	4	2	2	4	10
22	2	2	4	2	2	4	2	2	4	12
23	--	--	--	2	2	4	2	2	4	8
24	2	2	4	2	2	4	2	2	4	12
25	2	1	3	2	2	4	2	2	4	11
26	0	1	1	0	0	0	0	1	1	2
27	2	2	4	0	1	1	2	2	4	9
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

Lab n°	Patient D Methylmalonic aciduria cblA type			Patient E SBCADD			Patient F VLCAD			
	A	**I**	**Total**	**A**	**I**	**Total**	**A**	**I**	**Total**	**Total**
31	2	2	4	2	2	4	2	2	4	12
32	0	1	1	2	2	4	2	2	4	9
33	2	2	4	2	2	4	2	2	4	12
34	2	2	4	1	0	1	2	2	4	9
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	2	4	12
43	2	2	4	2	2	4	2	2	4	12
44	0	1	1	2	2	4	2	2	4	9
45	2	2	4	2	2	4	2	2	4	12

Total scores

Lab n°	A	B	C	D	E	F	Cumulative score	Cumulative score (%)	Critical error
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	4	4	4	4	0	4	20	83	
7	--	--	--	--	--	--	0	0	
8	4	4	4	4	4	4	24	100	
9	4	4	4	4	0	4	20	83	
10	4	4	0	4	4	4	20	83	
11	4	4	4	4	4	4	24	100	
12	4	4	0	4	4	4	20	83	
13	4	4	3	4	4	4	23	96	
14	4	4	2	4	4	4	22	92	
15	3	4	2	4	1	4	18	75	
16	4	4	4	4	4	4	24	100	
17	--	--	--	4	4	4	12	50	
18	4	4	4	4	4	4	24	100	
19	4	4	4	4	4	4	24	100	
20	2	4	0	4	0	3	13	54	
21	4	4	4	2	4	4	22	92	
22	4	4	0	4	4	4	20	83	
23	3	4	2	--	4	4	17	71	
24	4	4	4	4	4	4	24	100	
25	4	4	4	3	4	4	23	96	
26	4	0	1	1	0	1	7	29	CE
27	4	4	4	4	1	4	21	88	
28	4	4	0	4	4	4	20	83	
29	4	4	4	4	4	4	24	100	
30	4	4	4	4	4	4	24	100	
31	4	4	0	4	4	4	20	83	
32	4	4	0	1	4	4	17	71	
33	4	4	4	4	4	4	24	100	

Lab n°	A	B	C	D	E	F	Cumulative score	Cumulative score (%)	Critical error
34	1	4	4	4	1	4	18	75	CE
35	4	4	4	4	4	4	24	100	
36	4	4	0	4	4	4	20	83	
37	4	4	4	4	4	4	24	100	
38	--	--	--	4	4	4	12	50	
39	4	4	0	4	4	4	20	83	
40	4	4	4	4	4	4	24	100	
41	3	4	0	4	4	4	19	79	
42	4	4	4	4	4	4	24	100	
43	3	4	0	4	4	4	19	79	
44	4	1	4	1	4	4	18	75	
45	4	4	2	4	4	4	22	92	

Performance

	Number of labs	% total labs
Satisfactory performers (≥ 70 % of adequate responses)	39	87
Unsatisfactory performers (< 70 % adequate responses and/or critical error)	5	11
Partial and non-submitters	4	9

Overall Proficiency

Sample	Diagnosis	Analytical (%)	Interpretation (%)	Total (%)
ACDB-IR-2024-A	CPT1	95	94	95
ACDB-IR-2024-B	GA1; GCDH	95	96	96
ACDB-IR-2024-C	cblC	63	70	67
ACDB-IR-2024-D	Methylmalonic aciduria cblA type	91	95	93
ACDB-IR-2024-E	SBCADD	85	86	86
ACDB-IR-2024-F	VLCAD	98	98	98

10. Preview of the scheme in 2025

Sample distribution	5th February 2025
Start of analysis of Survey 2025/1 - Website open	17th March 2025
Survey 2025/1 - Results submission	7th April 2025
Survey 2025/1 - Reports	20th May 2025
Start of analysis of Survey 2025/2 - Website open	2nd June 2025
Survey 2025/2 – Results submission	23rd June 2025
Survey 2025/2 - Reports	4th August 2025
Annual Report 2025	Jan-Mar 2026

11. 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 ACDB scheme this certificate will indicate if results were submitted and whether satisfactory performance was achieved in the scheme.

12. Questions, Comments and Suggestions

If you have any questions, comments or suggestions please address to the Scientific Advisor of the scheme, Dr. Cristiano Rizzo (cristiano.rizzo@opbg.net) and/or to the ERNDIM Administration Office (admin@erndim.org)

Date of report, 2025-03-05
Name and signature of Scientific Advisor

Dr. Cristiano Rizzo
Scientific Advisor

Please note:
This annual report is intended for participants of the ERNDIM ACDB scheme. The contents should not be used for any publication without permission of the scheme advisor

APPENDIX 1 – Change log (changes since last version)

Version Number	Published	Amendments
1	16th May 2025	2024 annual report published

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