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Acylcarnitines in dried blood spots
Centre: Italy

Final Report 2025

prepared by
Dr. Cristiano Rizzo

Note: This annual report is intended for participants of the ERNDIM Acylcarnitine in dried blood spots scheme. The contents should not be used for any publication without permission of the Scientific Advisor.

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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 (metabolic center Roma) 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 2025 46 laboratories from many different countries participated in the ACDB Rome scheme.
There were no educational participants in 2025. 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 are 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.

[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.

Country	Number of participants
Belgium	6
Bulgaria	1
Croatia	1
Czech Republic	2
Greece	1
Israel	3
Lebanon	1
Lithuania	2
Malaysia	2
Oman	1
Portugal	2
Saudi Arabia	1
Singapore	1
Slovakia	1
Slovenia	1
Spain	8
Switzerland	3
Taiwan	1
Turkey	1
United Kingdom	2
United States	5

3. Design and logistics of the scheme including sample information

As usual, the samples used in 2025 were authentic human blood spot samples from affected patients. 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.

CSCQ dispatches the ACDB EQA samples to the scheme participants and provides a website for online 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

Participants are also encouraged to make use of the option to upload labelled copies of scans and/or chromatograms on the CSCQ website together with their analytical and interpretative results.

4. Schedule of the scheme

Time schedule in the 2025 ERNDIM ACDB Rome scheme.

	1st Submission Round	2nd Submission Round
Sample ID's:	ACDB-IR-2025-A ACDB-IR-2025-B ACDB-IR-2025-C	ACDB-IR-2025-D ACDB-IR-2025-E ACDB-IR-2025-F
Shipment of samples	February 5th 2025	
Start of analysis (clinical data available)	March 17th 2025	June 2nd 2025
Reminder for result submission	March 31st 2025	June 16th 2025
Results submission deadline:	April 7th 2025	June 23rd 2025
Interim reports available on CSCQ website	July 17th 2025	October 15th 2025

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 2025 ERNDIM ACDB Rome scheme.

Survey	Sample no.	Diagnosis
25-03-ACDB	ACDB-IR-2025-A	VLCADD
	ACDB-IR-2025-B	MCADD
	ACDB-IR-2025-C	SCADD
25-06-ACDB	ACDB-IR-2025-D	VLCADD
	ACDB-IR-2025-E	GAII
	ACDB-IR-2025-F	GAI

The scheme format was kept identical to those of previous years. All samples selected by the Scientific Advisor have been tested for suitability in the Scientific Advisor's laboratory before and after the shipping process. Samples were sent by DHL, FedEx or the Swiss Post at room temperature. The samples are stable for the duration of the scheme's submission calendar when stored under defined conditions. Interim reports were generated by the evaluation program developed by CSCQ.

Origin of patient samples: all urine samples have been provided by the scheme organizers or specified participants.

5. Results

Returned results in the 2025 ERNDIM ACDB Rome scheme.

	Survey 1	**Survey 2**
Receipt of results	43	41
No answer	3	5

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

Qualitative results and diagnostic proficiency of the samples were scored using the criteria given below. These criteria have been set by the Scientific Advisor and approved by the ERNDIM Scientific Advisory Board (SAB). A second evaluation of this year's results was carried out by Dr Joachim Janda, Scientific Advisor for the ACDB Heidelberg scheme. The final decision on the scoring in the scheme was made by the SAB at its autumn meeting (27th November 2025).

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**

ERNDIM applies the concept of 'critical error' in the scoring of results. In principle this is a category of error that would be unacceptable to the majority of labs and would have a serious adverse effect on patient management. 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 are 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 on 27 November 2025

7.1. Score for satisfactory performance

At least 17 points from the maximum of 24 (71%) are required for satisfactory performance.

We are required to define "Participation" for the purpose of the ERNDIM Annual Certificate which covers all ERNDIM schemes. For the 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

Very long Chain Acyl-CoA Dehydrogenase deficiency

Patient details provided to participants

Occasional cramps and dark urine as child. Episodes of rhabdomyolysis as young adult, two episodes requiring dialysis.

Analytical performance

46 laboratories were registered with the Rome section of the Scheme. Results were returned by 42 labs (91%). 41/42 (97%) respondents considered C14:1 concentration increased.

Interpretative proficiency

42/42 (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=2) b) Carnitine Palmitoyl transferase type 2 (CPT2) deficiency (n=8) 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 Deficiency (glutaric aciduria type 2) (n=5) e) TANGO2 (n=1) Suggested follow up test to confirm the diagnosis or guide further investigation were: mutation analysis of ACADVL gene (n=39), urinary organic acids (n=18), VLCAD activity in fibroblasts and lymphocytes (n=13), plasma carnitine/acylcarnitines analysis (n=18) The implementation of appropriate dietary management (carnitine supplementation, high carbohydrates low fat diet, avoidance of fasting) was mentioned by 8 respondents. Referral to a metabolic physician was mentioned by 6 respondents.

Acylcarnitines	n (quant)	n (qual)	Median value	SD
c14:1	41	42	2,89	0,92
c14	32	32	0,62	0,15
c14:2	31	32	0,37	0,16
c16:1	18	18	0,56	0,10
c14:1/c2	12	12	0,52	0,25
c14:1/c16	15	15	2,40	0,59

8.2. Patient B

Medium Chain Acyl-CoA Dehydrogenase deficiency

Patient details provided to participants

Neonatal onset, with hypoglycemic and hyperammonemic decompensation on the second day of life. To date, stable clinical picture and follows a normolipidic diet.

Analytical performance

46 laboratories were registered in Rome section of the Scheme. Results were turned by 41 labs (89%). Significant increases were found in C6, C8, C10:1, acylcarnitine species. The most used ratios were C8/C2 and C8/C10. All respondents considered C6, C8, C10:1 concentration increased.

Interpretative proficiency

40/41 (97%) respondents considered Medium Chain Acyl-CoA Dehydrogenase deficiency (MCAD) as the most likely diagnosis and 1 respondent considered Short Chain Acyl-CoA Dehydrogenase Deficiency/ Isobutyryl-CoA Dehydrogenase Deficiency as the most likely diagnosis. The alternative differential diagnosis suggested by respondents included: a) Multiple Dehydrogenase Deficiency (glutaric aciduria type 2) (n=5) b) Short Chain Acyl-CoA Dehydrogenase Deficiency/ Isobutyryl-CoA Dehydrogenase Deficiency (n=1) Suggested follow up test to confirm the diagnosis or guide further investigation were mutation analysis of ACADM gene (n=39), urinary organic acids (n=34), urinary acylglycines (n=14) enzyme assay in cultured fibroblast (n=7), plasma carnitine/acylcarnitines analysis (n=20). The implementation of appropriate dietary management (carnitine supplementation, high carbohydrates low fat diet, avoidance of fasting) was mentioned by 13 respondents. Referral to a metabolic physician was mentioned by 7 respondents.

Acylcarnitines	n (quant)	n (qual)	Median value	SD
c6	40	41	1,20	0,25
c8	40	41	2,90	0,68
c10:1	34	35	0,68	0,33
c10	17	18	0,19	0,04
c8/c10	24	24	15,71	2,46
c8/c2	17	17	1,22	2,17

8.3. Patient C

Short Chain Acyl-CoA Dehydrogenase Deficiency

Patient details provided to participants

At the age of 4, a diagnosis of epilepsy was made. Therapy started first with Valproate then with ethosuximide with disappearance of the crises

Analytical performance

46 laboratories were registered in Rome section of the Scheme. Results were turned by 41 labs (89%). 39 respondents found increased C4-carnitine. 1 laboratory reported no alterations in the acylcarnitine profile. The acylcarnitine pattern with increased C4-carnitine is highly suggestive for SCADD or IBDD.

Interpretative proficiency

39/41 (95%) respondents considered Short Chain Acyl-CoA Dehydrogenase Deficiency (SCADD; OMIM 201470), 1 respondent considered Ethylmalonic encephalopathy (ETHE1), 1 respondent considered normal acylcarnitines profile as the most likely diagnosis. The alternative differential diagnosis suggested by respondents included: a) Isobutyryl-CoA Dehydrogenase Deficiency (IBDD; OMIM 611283) (n=23) b) Ethylmalonic encephalopathy (ETHE1) (n=10) c) Glutaric aciduria type II (n=1) d) Secondary effect of valproate therapy (n=2) Suggested follow up test to confirm the diagnosis or guide further investigation were mutation analysis of ACADS, ACAD8, ETHE1 genes (n=36), urinary organic acids (n=36), enzyme assay in cultured fibroblast (n=6), plasma acylcarnitines analysis (n=18), urine acylcarnitines analysis (n=6). Urinary organic acid analysis and resolution between butyrylcarnitine and isobutyrylcarnitine using the second-tier LC-MS/MS method in DBS are required to further establish the diagnosis. The final confirmation is by molecular genetic testing.

Acylcarnitines	n (quant)	n (qual)	Median value	SD
c4	38	40	1,35	0,23
c4/c3	10	10	1,04	0,13
c4/c2	11	11	0,17	0,04
c4/c8	7	7	20,56	1,80
c5	13	13	0,20	0,05

8.4. Patient D

Very Long Chain Acyl-CoA dehydrogenase deficiency (OMIM 609575)

Patient details provided to participants

She underwent multiple hospitalizations for rhabdomyolysis and metabolic decompensation. To date, she follows the low-lipid home diet integrated with MCT and evening cornstarch with good compliance.

Analytical performance

Very long-chain acyl-CoA dehydrogenase deficiency (VLCADD) is caused by mutations in ACADVL gene and leads to the accumulation of C14-C18 acylcarnitines and low free carnitine. The phenotypic presentation is heterogenous, including cardiomyopathy, hypoketotic hypoglycemia, liver disease, excercise intolerance and rhabdomyolisis. It is associated with three major phenotypes: severe infantile VLCADD, moderately severe infantile/childhood VLCADD and late onset myopathic VLCADD. The diagnosis is confirmed by molecular genetic testing showing two pathogenic mutations in the ACADVL gene. In VLCAD patients it's necessary to avoid fasting, limit exercise and cold/heat exposure. Dietary treatment in VLCADD involves a low long-chain fat diet in combination with medium chain triglycerides.
46 laboratories were registered with the Rome section of the Scheme. Analytical results were returned by 41 labs (89%). 100% respondents founded C14:1-carnitine elevated.

Interpretative proficiency

Interpretative proficiency results were returned by 41 labs (89%). 41/41 (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=10) c) Mitochondrial trifunctional protein (MTP) deficiency/long-chain 3-Hydroxyacyl-CoA dehydrogenase (LCHAD) deficiency/longchain 3-Ketoacyl-CoA Thiolase (LCKAT) deficiency (n=2) d) Multiple Dehydrogenase Deficency (glutaric aciduria type 2) (n=1) e) MCAD (n=1) Suggested follow up test to confirm the diagnosis or guide further investigation were: mutation analysis of ACADVL gene (n=37), urinary organic acids (n=19), VLCAD activity in fibroblasts and lymphocytes (n=16), plasma carnitine/acylcanitines analysis (n=23).

Acylcarnitines	n (quant)	n (qual)	Median value	SD
c14:1	41	41	1,49	0,49
c14:2	35	35	0,24	0,06
c14	34	34	0,63	0,11
c16:1	22	22	0,66	0,14
c14:1/c2	14	14	0,22	0,04
c14:1/c16	15	16	0,90	0,20

8.5. Patient E

Multiple Acyl-CoA Dehydrogenase Deficiency (Glutaric Acidemia II)

Patient details provided to participants

At one year of age she was hospitalized for hyperthermia with asthenia and hypotonia, To date, she follows therapy with riboflavin and carnitine; psychomotor development and language are appropriate to age.

Analytical performance

46 laboratories were registered in Rome section of the Scheme. Results were turned by 41 labs (89%). 100% respondents founded C4, C5, C6, C8, C10, C12, C14, C14: 1, C14:2, C18:1 and C18:2 carnitines elevated.

Interpretative proficiency

Interpretative proficiency results were returned by 41 labs (89%). 40/41 (97%) respondents considered Multiple Acyl-CoA dehydrogenase deficiency (MADD) and one respondent considered Very long-chain acyl-CoA dehydrogenase deficiency (VLCAD) as the most likely diagnosis. The suggested alternative diagnoses were: defects in FAD synthase, riboflavin transporters or the mitochondrial flavin adenine dinucleotide transporter (MTF) (14 respondents); Medium chain Acyl-CoA deficiency (3 respondents); Carnitine palmitoyltransferase II (5 respondents); Very Long Chain Acyl-CoA dehydrogenase deficiency (3 respondent); Carnitine-acylcarnitine translocase deficiency (3 respondent); riboflavin deficiency (4 respondents); Short Chain Acyl-CoA dehydrogrnase deficiency (1 respondent); TANGO 2 (1 respondent); unspecified mitochondrial disorder (1 respondent); autosomal recessive polycystic kidney disease (1 respondent), Zellweger syndrome (1 respondent), alterations in sterol biosynthesis (1 respondent), MADD like disease associated with Sertraline use (1 respondent) , nitric oxide abuse (1 respondent). Suggested follow up test to confirm the diagnosis or guide further investigation were mutation analysis of ETFA, ETFB, ETFDH, FLAD1, SLC52A1, SLC52A2 SLC52A3, SLC25A32 genes (n=39), urinary organic acids (n=35), enzyme assay in cultured fibroblast (n=4), plasma acylcarnitines analysis (n=17), plasma riboflavin (n=5). It is important for patients with MADD to strictly avoid fasting to prevent hypoglycemia and crises of metabolic acidosis. The implementation of appropriate dietary management and therapy (riboflavin, carnitine, coenzyme Q10, glycine supplementation, triheptanoine, protein and fat restricted diet and high carbohydrate, ketogenic diet) was mentioned by 8 respondents. Referral to a metabolic physician was mentioned by 4 respondents. The clinical presentation and treatment with riboflavin and carnitine fit with the MADD diagnosis. Patients with MADD can present symptoms from infancy to adulthood. The most common symptoms are muscle weakness, exercise intolerance, and/or muscle pain, although metabolic decompensation with episodes of rhabdomyolysis can also be seen. Fever can trigger metabolic decompensation.

Acylcarnitines	n (quant)	n (qual)	Median value	SD
c8	41	41	0,79	0,14
c10	40	40	1,35	0,42
c5	41	41	0,98	0,34
c14:1	38	38	0,78	0,27
c6	37	37	0,44	0,10
c12	29	29	0,71	0,17

8.6. Patient F

Glutaryl-CoA Dehydrogenase deficiency (glutaric acidemia type I) OMIM 231670

Patient details provided to participants

Diagnosed at 17 days of age. At birth he presented with large pseudocysts in the bilateral frontal paraventricular area. Follows pharmacological therapy (carnitine) and dietary therapy (reduced intake of lysine). To date, the child's clinical picture is very good: never metabolic decompensations, absence of neurological signs and macrocrania.

Analytical performance

Glutaric acidemia type 1 (GA1) is an autosomal recessive neurometabolic disorder caused by glutaryl-CoA dehydrogenase (GCDH) deficiency (GDD) due to mutations in the GCDH gene (19p13.2). GCDH is involved in Llysine, L-hydroxylysine and L-tryptophane catabolic pathways. Based on the acylcarnitine profile, investigation of organic acids in urine should be performed (GDD leads to the accumulation of glutaric, 3-hydroxyglutaric and glutaconic acids and glutarylcarnitine in body tissues). The diagnosis is confirmed by enzyme assay and GCDH mutation analysis. Presymptomatic detection can be offered through newborn screening programs implemented in many countries. Prenatal diagnosis is available. Therapy includes low-lysine diet, carnitine supplementation, and emergency treatment during episodes of decompensation. 46 laboratories were registered with the Rome section of the Scheme. Analytical results were returned by 40 labs (86%). 100% respondents founded glutarylcarnitine elevated.

Interpretative proficiency

40/40 (100%) respondents considered glutaryl-CoA dehydrogenase deficiency (glutaric acidemia type I) as the most likely diagnosis. Suggested follow up test to confirm the diagnosis or guide further investigation were mutation analysis of glutaryl-CoA dehydrogenase (GCDH) gene (n=37), urinary organic acids (n=38), enzyme assay in cultured fibroblast (n=14), plasma carnitine/acylcanitines analysis (n=14), urinary glutarylcarnitine (n=3) The implementation of appropriate dietary management (carnitine supplementation, lysine restricted diet) was mentioned by 10 respondents. Referral to a metabolic physician was mentioned by 1 respondent.

Acylcarnitines	n (quant)	n (qual)	Median value	SD
c5dc	40	40	2,92	1,45
c5dc/c16	12	12	3,71	1,40
c5dc/c8	12	12	86,02	757,65
c0	13	13	33,00	8,33
c5dc/c5oh	7	7	6,39	2,24

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.

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 of 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 VLCADD			Patient B MCADD			Patient C SCADD			
	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	0	0	0	8
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	1	3	11
8	2	2	4	2	2	4	2	1	3	11
9	2	2	4	2	2	4	2	2	4	12
10	2	2	4	2	0	2	2	2	4	10
11	2	2	4	2	2	4	2	2	4	12
12	2	1	3	2	1	3	2	2	4	10
13	2	2	4	2	2	4	2	2	4	12
14	2	2	4	2	2	4	2	2	4	12
15	2	2	4	2	2	4	2	2	4	12
16	2	2	4	2	2	4	2	2	4	12
17	2	2	4	2	2	4	2	2	4	12
18	2	2	4	2	2	4	2	2	4	12
19	2	2	4	2	2	4	2	2	4	12
20	2	2	4	2	2	4	2	2	4	12
21	2	1	3	2	1	3	2	2	4	10
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

Lab n°	Patient A VLCADD			Patient B MCADD			Patient C SCADD			
	A	I	Total	A	I	Total	A	I	Total	Total
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	0	0	0	0	0	0	4
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	0	0	0	0	0	0	0	0	0	0
37	2	2	4	2	2	4	2	2	4	12
38	2	2	4	2	2	4	2	2	4	12
39	0	0	0	0	0	0	0	0	0	0
40	2	2	4	2	2	4	2	2	4	12
41	0	0	0	0	0	0	0	0	0	0
42	0	0	0	0	0	0	0	0	0	0
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	0	0	0	8

Detailed scores – Round 2

Lab n°	Patient D VLCADD			Patient E GAII			Patient F GAI			
	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	0	0	0	0	0	0	0	0	0	0
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	1	3	2	2	4	2	2	4	11
13	2	2	4	2	2	4	2	2	4	12
14	2	2	4	2	2	4	2	2	4	12
15	2	2	4	2	2	4	2	2	4	12
16	2	2	4	2	2	4	2	2	4	12
17	2	2	4	2	2	4	2	2	4	12
18	2	2	4	2	2	4	2	2	4	12
19	2	2	4	2	2	4	2	2	4	12
20	2	2	4	2	2	4	2	2	4	12
21	0	0	0	0	0	0	0	0	0	0
22	2	2	4	2	2	4	2	2	4	12
23	2	2	4	2	2	4	2	2	4	12
24	2	2	4	2	2	4	2	2	4	12
25	2	2	4	2	2	4	2	2	4	12
26	2	2	4	2	2	4	2	2	4	12
27	2	2	4	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	0	0	0	8
30	2	2	4	2	2	4	2	2	4	12
31	2	2	4	2	2	4	2	2	4	12

Lab n°	Patient D VLCADD			Patient E GAII			Patient F GAI			
	A	I	Total	A	I	Total	A	I	Total	Total
32	2	2	4	2	2	4	2	2	4	12
33	2	2	4	2	2	4	2	2	4	12
34	0	0	0	0	0	0	0	0	0	0
35	2	2	4	2	2	4	2	2	4	12
36	2	2	4	2	2	4	2	2	4	12
37	2	2	4	2	1	3	2	2	4	11
38	2	2	4	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	0	0	0	0	0	0	0	0	0	0
42	0	0	0	0	0	0	0	0	0	0
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

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	0	0	0	12	50	
4	4	4	0	4	4	4	20	83	
5	4	4	4	4	4	4	24	100	
6	4	4	4	4	4	4	24	100	
7	4	4	3	4	4	4	23	96	
8	4	4	3	4	4	4	23	96	
9	4	4	4	4	4	4	24	100	
10	4	2	4	4	4	4	22	92	
11	4	4	4	4	4	4	24	100	
12	3	3	4	3	4	4	21	88	
13	4	4	4	4	4	4	24	100	
14	4	4	4	4	4	4	24	100	
15	4	4	4	4	4	4	24	100	
16	4	4	4	4	4	4	24	100	
17	4	4	4	4	4	4	24	100	
18	4	4	4	4	4	4	24	100	
19	4	4	4	4	4	4	24	100	
20	4	4	4	4	4	4	24	100	
21	3	3	4	0	0	0	10	42	
22	4	4	4	4	4	4	24	100	
23	4	4	4	4	4	4	24	100	
24	4	4	4	4	4	4	24	100	
25	4	4	4	4	4	4	24	100	
26	4	4	4	4	4	4	24	100	
27	4	4	4	4	4	4	24	100	
28	4	4	4	4	4	4	24	100	
29	4	0	0	4	4	0	12	50	
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	

Lab n°	A	B	C	D	E	F	Cumulative score	Cumulative score (%)	Critical error
34	4	4	4	0	0	0	12	50	
35	4	4	4	4	4	4	24	100	
36	0	0	0	4	4	4	12	50	
37	4	4	4	4	3	4	23	96	
38	4	4	4	4	4	4	24	100	
39	0	0	0	4	4	4	12	50	
40	4	4	4	4	4	4	24	100	
41	0	0	0	0	0	0	0	0	
42	0	0	0	0	0	0	0	0	
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	0	4	4	4	20	83	

Performance

	Number of labs	% total labs
Satisfactory performers (≥ 71 % of adequate responses)	38	83
Unsatisfactory performers (< 71 % adequate responses and/or critical error)	1	2
Partial and non-submitters	7	15

Overall Proficiency

Sample	Diagnosis	Analytical (%)	Interpretation (%)	Total (%)
ACDB-IR-2025-A	VLCADD	91	89	90
ACDB-IR-2025-B	MCADD	89	85	87
ACDB-IR-2025-C	SCADD	85	83	84
ACDB-IR-2025-D	VLCADD	89	88	89
ACDB-IR-2025-E	GAII	89	88	89
ACDB-IR-2025-F	GAI	87	87	87

10. Tentative schedule in 2026

Sample distribution	04 February 2026
Start of analysis of Survey 2026/1	17 March 2026
Survey 2026/1 – results submission	07 April 2026
Survey 2026/1 – report	19 May 2026
Start of analysis of Survey 2026/2	01 June 2026
Survey 2026/2 – results submission	22 June 2026
Survey 2026/2 – report	03 August 2026
Annual meeting of participants	August 2026
Annual report 2026	January-March 2027

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, Suggestions and Complaints

If you have any questions, comments or suggestions please address to the Scientific Advisor of the scheme, Dr Cristiano Rizzo 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, 2026-01-28
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 the last version)

Version Number	Published	Amendments
1	03 February 2026	2025 annual report published

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