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Acylcarnitines in Dried Blood Spots

Centre: Germany

Final Report 2025

prepared by
Dr Joachim Janda

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.

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1. Introduction

The ERNDIM ACDB scheme offers dried blood spots (DBS) obtained from 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 Joachim Janda (Metabolic Center Heidelberg) in conjunction with CSCQ, the Swiss organisation for quality assurance in medical laboratories.

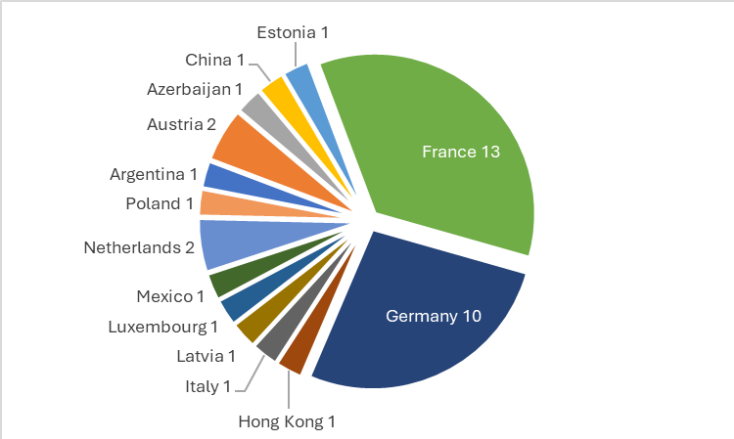
As in previous years, samples were sent out to cover the spectrum of what is typically observed in the metabolic laboratory. A mix of clearly diagnostic profiles and some more challenging profiles were provided. As in previous years normal profiles were also sent out. The requirement to interpret a normal 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, 47 laboratories from many different countries registered in the ACDB Heidelberg scheme with no educational participants. Educational participants can 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 be distributed between the Heidelberg, London and Rome acylcarnitine in DBS schemes which are run separately. The three organising laboratories each participate in the other's scheme by rotation.



Country	Number of participants
Argentina	1
Austria	2
Azerbaijan	1
China	1
Estonia	1
France	13
Germany	10
Hong Kong	1

Country	Number of participants
Italy	1
Latvia	1
Luxembourg	1
Mexico	1
Netherlands	2
Poland	1
Turkey	9
Ukraine	1

3. Design and logistics of the scheme including sample information

As in earlier ACDB schemes, the samples used in 2025 were authentic human DBS samples, five from affected patients and one from a healthy individual.
All samples selected by the Scientific Advisor are typically prepared from 30-50 µL of lithium heparin (or EDTA) anticoagulated whole blood on Whatman (Schleicher & Schuell) 903™ paper. All samples are obtained following local ethical and consent guidelines.
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

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 Heidelberg scheme.

	1st Submission Round	2nd Submission Round
Sample IDs	ACDB-DH-2025-**A** ACDB-DH-2025-**B** ACDB-DH-2025-**C**	ACDB-DH-2025-**D** ACDB-DH-2025-**E** ACDB-DH-2025-**F**
Shipment of samples	February 7, 2025	
Start of analysis (clinical data available)	17 March 2025	02 June 2025
Reminder for result submission	31 March 2025	16 June 2025
Results submission deadline	07 April 2025	23 June 2025
Interim reports available on CSCQ website	after 10 July 2025	after 31 October 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 Heidelberg scheme.

Survey	Sample no.	Diagnosis
25-03-ACH	ACDB-DH-2025-A	Very-Long-Chain Acyl-CoA Dehydrogenase deficiency
	ACDB-DH-2025-B	Mild Very-Long-Chain Acyl-CoA Dehydrogenase deficiency
	ACDB-DH-2025-C	Multiple Acyl-CoA dehydrogenase deficiency
25-06-ACH	ACDB-DH-2025-D	Normal control sample
	ACDB-DH-2025-E	Methylmalonic aciduria due to methylmalonyl-CoA mutase deficiency
	ACDB-DH-2025-F	3-hydroxy-3-methylglutaryl-CoA lyase deficiency

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 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: Unless indicated otherwise, the DBS samples were provided by the scheme organizers.

Patient A:	VLCADD	Common sample distributed to the participants of all ACDB centres provided by the ACDB centre London
Patient B:	VLCADD	
Patient C:	MAD	
Patient D:	Normal	
Patient E:	MMA mut(0)	
Patient F:	HMG CLD	

Prior to the distribution of the first round, a validation set of samples was returned from the CSCQ to the organising laboratory and re-analysed.

5. Results

Returned results in the 2025 ERNDIM ACDB Heidelberg scheme.

	Survey 1	Survey 2
Receipt of results	47	47
No answer	5	7

6. Web site reporting

The website reporting system is compulsory for all centres. The participants are reminded to carefully read and consider the following advice:

- **Results**
  - Give as much quantitative data as possible.
  - Enter the key metabolites with the evaluation **in the tables** even if you do not give quantitative data.
  - If the profile is normal: enter “Normal profile” in “Key metabolites”.
  - **Do not enter results in the “comments” field**, **otherwise your results will not be included in the evaluation program.**
- **Diagnosis**
  - **Do not 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 on treatment will not be scored.
  - **Do not give recommendations in “Comments on diagnosis” field**: 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 our website (https://eqa.erndim.org/information/view/14)

Qualitative results and diagnostic proficiency of the 2025 samples were scored using the criteria given below. These criteria have been set by the Scientific Advisor and approved by the SAB of ERNDIM. A second evaluation of this year's results was carried out by Erin Emmett, scientific advisor of the ACDB London scheme. The final decision on scoring of the scheme has been made by the SAB during its autumn meeting (27 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 17 November 2025

Score for satisfactory performance

A minimum of 17 points out of a maximum of 24 (71%) is necessary for a satisfactory performance.

The ERNDIM Annual Certificate covers all ERNDIM schemes in which a laboratory has participated during the scheme year. For the ACDB scheme, “participation” is defined 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.

Further patient details

This was a common sample provided by the ACDB centre London.

This sample was collected from a 57-year-old male with history of episodes of rhabdomyolysis associated with intense exercise. He received genetic confirmation of Very-Long-Chain Acyl-CoA Dehydrogenase (VLCAD) deficiency at the age of 55 following another episode of rhabdomyolysis requiring admission to A&E with a CK of >10,000 IU/L. Patient now well controlled on a lower fat diet with avoidance of large intakes of alcohol, and use of SOS carbohydrate supplement prior to increased exercise. At the time of sample collection, the patient was not formally using MCT supplementation.

The acylcarnitine profile shows a clear VLCADD profile with elevated C14:1 carnitine and grossly elevated associated ratios, e. g. C14:1/C4, C14:1/C6, or C14:1/C8. C14:0 carnitine is slightly elevated.

Analytical performance

The metabolite levels reported most frequently as elevated (E) or even grossly elevated (GE) in this sample were those of C14:1 carnitine (GE: n=32, E: n=8), C14:2 carnitine (GE: n=8, E: n=23), C14:0 carnitine (GE: n=5, E: n=26), and C16:1 carnitine (GE: n=2, E: n=19). Most participants also reported on diagnostic ratios, such as the C14:1/C12:1 ratio (GE: n=8, E: n=4) or other ratios based on C14:1 carnitine.
One participant reported a normal acylcarnitine profile.

Evaluation criteria: Two points are awarded for reporting C14:1 carnitine and/or associated ratios at least as elevated.

Diagnosis / Interpretative proficiency

VLCADD was reported as principal diagnosis by 36 participants (86%) and as an alternative by one participant. Five participants chose CPT2 deficiency as their principal diagnosis. It was also the most frequently mentioned alternative diagnosis (n=7). In some cases, the participants interpreted their analytical results as MADD (n=6 as alternative diagnosis), LCHADD (n=1), TFPD (n=1) or as normal (n=1).

Evaluation criteria: Two points are given if VLCADD is reported as diagnosis (principal or alternative) and an appropriate test for confirmation is recommended. One point is awarded if only one of both criteria is met.

One participant, who did not detect an elevated C14:1 carnitine concentration reported a mild CPT II deficiency as diagnosis and actively excluded a VLCADD in the report. Another participant, who failed to detect relevant metabolites reported a normal profile. Both of these were considered critical errors, that have been approved at the autumn SAB meeting.

Recommendations

The vast majority of participating labs (n=40) recommended to perform genetic testing to confirm their suggested diagnoses. Enzymatic analyses (n=25) or the determination of organic acids in urine (n=24) were also frequently recommended. Ten participants recommended to analyse acylcarnitines in plasma, and nine participants gave advice on therapeutic measures.

Overall impression

This sample was not challenging for most participants. The analytical proficiency achieved was 98% and the interpretative proficiency was 88%.

8.2. Patient B

Mild Very-Long-Chain Acyl-CoA Dehydrogenase deficiency

Patient details provided to participants

Girl aged 11, diagnosed by NBS. Now recurrent nausea without vomiting.

Further patient details

The sample was taken from an 11-year-old girl with elevated C14:1 level in newborn screening, which normalised in the second card. However, VLCAD enzyme activity in leukocytes was 18% of controls and molecular analysis of *ACADVL* showed compound heterozygosity for two deletions confirming VLCAD deficiency. She has never had any metabolic decompensations, but in the last outpatient visit recurrent episodes of nausea in the last 6 months were reported. CK and aminotransferases were normal and apart from obesity, the physical examination was normal. The nausea was interpreted as most likely triggered by psychosocial factors.

Compared to sample A, the pattern of the indicative acylcarnitines in this VLCAD sample is less pronounced, i.e., the C14:1 carnitine concentration is only slightly elevated. However, several of the associated ratios are elevated, e.g., C14:1/C4, C14:1/C6, and C14:1/C8.

Analytical performance

Most participants referred to C14:1 and C14:2 carnitines and categorised their concentrations as elevated (n=26 and n=11, respectively), while four participants reported one or both metabolites as normal. Many of them also reported C14:1-based ratios, and parameters such as C14:1/C2, C14:1/C6, C14:1/C8 and C14:1/C16 were mostly rated as elevated (n=26 in total, compared to two ratings as normal), while C14:1/C12:1 was exclusively categorised as normal in this sample.

The analytical results of the participants (n=15) who reported other metabolites did not indicate a clear trend towards specific acylcarnitines. A normal profile was reported by seven labs.

Evaluation criteria: Two points are awarded for reporting C14:1 carnitine and/or associated ratios at least as elevated.

Diagnosis / Interpretative proficiency

VLCADD was reported as principal diagnosis by 21 (50%) and as alternative by four (9.5%) participants. Seven participants suggested a normal acylcarnitine profile, while seven other participants considered MADD deficiency as their interpretation. Three labs interpreted their analytical results as a typical profile indicating a long period of fasting. Other interpretations included carnitine deficiency (n=3), MCADD (n=2), LCHADD, CPT2 and CPT1 deficiency (n=1 each).

Evaluation criteria: The criteria were set as in sample A, i.e., two points are given if VLCADD is considered as diagnosis. Recommending an appropriate further test to diagnose VLCADD is awarded with 1 pt.

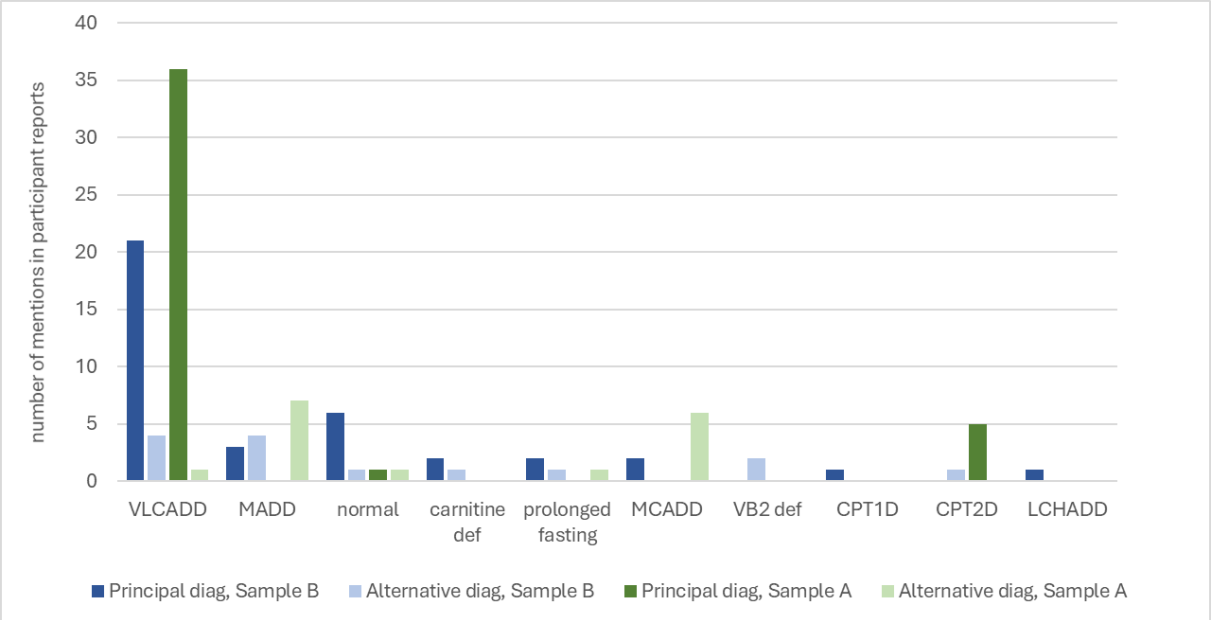
Due to the only subtle increase of the C14:1 concentration, a comparably large part of the participants did not consider VLCADD. Instead, the interpretations were much broader as depicted below. Thus, it was suggested that it should be excluded from critical error evaluation, which was approved at the autumn SAB meeting.

Recommendations

The most frequently recommended additional tests to perform were genetic analyses (n=27), urinary organic acids (n=25), enzymatic activity (n=12) or repeating the acylcarnitines in dried blood spots (n=12) or plasma (n=11).

Overall impression

Compared to the rather clear profile in sample A, this sample was much more challenging. The analytical and interpretational proficiency was 64% and 71%, respectively.



Comparison of feedback received for samples A and B

This sample was one of the more difficult ones in the 2025 scheme of ACDB Heidelberg. The evaluation showed that those laboratories that included specific diagnostic ratios for VLCADD in their analysis results were also more likely to be the participants who were able to make the correct diagnosis.

It may therefore advisable to consider an implementation of C14:1-based diagnostic ratios such as C14:1/C2 or ratios to certain medium-chain acylcarnitines[2].

[2] Chace et al. (2001): Electrospray Tandem Mass Spectrometry for Analysis of Acylcarnitines in Dried Postmortem Blood Specimens Collected at Autopsy from Infants with Unexplained Cause of Death, Clin. Chem. 47, 1166–1182. https://doi.org/10.1093/clinchem/47.7.1166

McHugh, et al. (2011): Clinical validation of cutoff target ranges in newborn screening of metabolic disorders by tandem mass spectrometry: A worldwide collaborative project, Genet. Med. 13, 230–254. https://doi.org/10.1097/gim.0b013e31820d5e67

Tajima et al (2024): Using the C14:1/Medium-Chain Acylcarnitine Ratio Instead of C14:1 to Reduce False-Positive Results for Very-Long-Chain Acyl-CoA Dehydrogenase Deficiency in Newborn Screening in Japan, Int. J. Neonatal Screen. 10, 15. https://doi.org/10.3390/ijns10010015

8.3. Patient C

Multiple Acyl-CoA dehydrogenase deficiency

Patient details provided to participants

Toddler with normal development with recurrent episodes of trembling.

Further patient details

The sample is from a now 3-year-old patient identified in newborn screening with elevations of medium chain acylcarnitines (C8 < C10) and elevated C12, C14 and C14:1. Clinically, the patient had been asymptomatic. Confirmatory biochemical analysis showed elevated medium chain acylcarnitines in DBS and normal urinary organic acids. Molecular analysis revealed two variants in *ETFDH* that had not been reported before (one interpreted as VUS and one as likely pathogenic) indicating multiple acyl-CoA dehydrogenase (MAD) deficiency. VLCAD and MCAD enzyme activity was normal. On riboflavine treatment the patient is still asymptomatic with normal development and no decompensations.

The acylcarnitines profile shows elevated C8 carnitine with several increased associated ratios (e.g., C8/C6, C8/C2 or C8/C12). Concentrations of C6, C10 and C10:1 carnitine are also elevated, and C8 > C10. Therefore, the profile may be mistaken for that of MCAD deficiency.

Analytical performance

The feedback from the participants on the results of their analyses reflects the profile described very well: Elevated or grossly elevated concentrations of C8, C10, C6 and C10:1 carnitines were reported by 98%, 88%, 69% and 48% of labs, respectively. 19 participants reported an elevated C8/C2 ratio and seven participants an elevated C8/C10 ratio, which was also rated as normal by 10 other participants. Other analytes reported as elevated by few participants were C8:1, C12, C14, and C16 carnitines.

Evaluation criteria: One point is awarded for reporting one of the analytical parameters C6, C8, C10:0, C10:1 carnitine, or C8-based ratios at least as elevated. Two points are given for reporting two or more of them.

Diagnosis / Interpretative proficiency

In their interpretations most participants stated MCADD as principal (n=31) or alternative diagnosis (n=8). The correct diagnosis, MADD, however, was only considered by 17 participants. Other interpretations considered by some labs included riboflavin deficiency (n=7) or mitochondrial disorders (n=3).

In this sample, it is particularly striking that a large number of participants have settled on one diagnosis without considering an alternative.

Evaluation criteria: Two points are given if MADD is considered as diagnosis and a suitable test for differentiation is recommended. One point is given if only one of both criteria is met.

Recommendations

Most participants (n=37) recommended analysing organic acids in urine, which in this particular case is useful for differentiating the ambiguous profile. For confirmation, the majority of laboratories recommended genetic testing (n=35) or enzymatic activity analysis (n=11). Repeating acylcarnitine analysis in plasma was suggested by ten laboratories and in dried blood spots by six laboratories.

Overall impression

The participants demonstrated an excellent analytical performance and, in most cases, reported multiple of the relevant analytes (99% proficiency). However, a majority of the participants have decided on MCADD in their interpretation without considering MADD as a possible alternative, resulting in only 68% interpretative proficiency.

8.4. Patient D

Normal control sample

Patient details provided to participants

Middle-aged man who recurrently suffered from joint pain and apathy.

Further patient details

The sample was taken from a colleague in our lab not known to have a metabolic deficiency. In the initial and validation experiments, acyl carnitine concentrations were within normal ranges except for a slightly increased C5OH concentration to the upper normal range.

Analytical performance

A normal AC profile was reported by 21 participants (53%) in their analytical results. Seventeen participants (43%) reported C5OH (or in combination with the isobaric C4DC), and elevated C3 concentrations were stated by 12 participants (30%). Two labs described the C3 concentration as normal. Other ACs were only mentioned sporadically as conspicuous, e.g., C3DC, C5:1, C18 or C18:1.

Evaluation criteria: Full score is awarded for considering a normal profile or absence of a metabolic disorder.

Diagnosis / Interpretative proficiency

Twenty-five participants considered a normal sample, i. e. the absence of a metabolic disorder in this sample, even though four of them reported elevated analyte concentrations. However, the majority of participants who detected analyte concentrations above their reference ranges suggested diagnoses: Eleven labs reported Vit. B_{12} deficiency and/or a disorder of cobalamin metabolism, four participants mentioned C5OH-related disorders, e. g. 3MCC or 3MGA, and three labs reported CPT2 deficiency.
In this sample, in addition to the primary and alternative diagnoses, many participants indicated further disorders in the comments field of the “interpretation” section that are either broader in scope or cannot be diagnosed through the analysis of acylcarnitines, e.g., mitochondrial or peroxisomal disorders, mevalonic aciduria, Gaucher, renal dysfunction, McArdle disease, or Duchenne muscular dystrophy.

Evaluation criteria: Full score is awarded for considering a normal profile or absence of a metabolic disorder.

Recommendations

Frequently recommended follow-up tests given by the participants were analysis of urinary organic acids (n=22), determination of amino acids in plasma or urine (n=9), cobalamin in plasma (n=10) or genetic testing (n=12).

Overall impression

Compared to earlier circulations of normal samples, this one seemed to be difficult for many participants. Only a small proportion of participants who reported elevated analyte levels considered that the increases might be non-specific. The combined proficiency for this sample was 63%.

8.5. Patient E

Methylmalonic aciduria due to methylmalonyl-CoA mutase deficiency

Patient details provided to participants

Metabolic decompensation after gastrointestinal infection. Now on treatment.

Further patient details

The sample was taken from a girl now 5 years old with mut° methylmalonic aciduria (MMA), identified by newborn screening. She already had metabolic acidosis when results of NBS became available and she had frequent metabolic decompensations thereafter. Diagnosis was confirmed by molecular analysis showing two pathogenic variants in *MMUT*.
The acylcarnitines profile shows grossly elevated propionyl (C3) carnitine with increased associated ratios (e. g. C3/C0 or C3/C2). In addition, slightly increased tiglyl (C5:1) carnitine and decreased palmitoyl (C16) carnitine concentrations can be observed.

Analytical performance

All participants who submitted results reported elevated (n=2) or grossly elevated (n=38) C3 concentrations and, in most cases, also mentioned elevated associated analyte ratios. Some participants also referred to concentration levels of other acylcarnitines in their reports, e. g. C4DC (9 normal, 5 elevated), C5:1 (9 elevated), C2 (5 elevated, 1 normal, 2 decreased) or C16 (3 decreased).

Evaluation criteria: Two points are given for reporting C3 carnitine and/or associated ratios at least as elevated.

Diagnosis / Interpretative proficiency

The most frequently mentioned principal diagnosis for this sample was propionic aciduria (n=30), followed by MMA (n=13). Three participants suggested a cobalamin-related MMA. These diagnoses were also most frequently mentioned as alternative, however, eight participants stated PA as diagnosis without considering MMA. Less frequently mentioned alternative diagnoses were Vit. B_{12} deficiency or multiple carboxylase deficiency.

Evaluation criteria: One point was given for reporting MMA as principal or alternative diagnosis and one point for a helpful recommendation enabling to find the correct diagnosis, e. g. urinary organic acids.

Recommendations

The most frequently recommended additional tests to perform were organic acids in urine (n=39), genetic analyses (n=35), plasma amino acids (n=14) and measuring enzymatic activity (n=8). Some participants specified the latter to measure biotinidase acitivity to narrow down their reported alternative diagnoses.

Overall impression

The participants showed a great analytical performance (100%), however, not all participants reported MMA, thus the interpretive proficiency was 91% only.

8.6. Patient F

3-hydroxy-3-methylglutaryl-CoA lyase deficiency

Patient details provided to participants

Seven-year-old girl with a hypoglycemic seizure as a toddler. Today well.

Further patient details

The sample was taken from a girl in whom 3-hydroxy-3-methylglutaryl-CoA lyase (HMG CL) deficiency was suspected in NBS and further confirmed by organic acid analysis. Initially, she had metabolic acidosis and mild hyperammonemia which normalized after iv glucose and carnitine. Treatment was started with a low protein diet and oral carnitine. At age 5, she had a metabolic decompensation with hypoglycemia caused by a febrile infection. Her cognitive development is in the lower normal range and she has a deficit of white matter in cranial MRI. The family refused genetic testing.

The key acylcarnitines for HMG CLD, C5OH and C6DC, are observed in only slightly elevated concentrations in this sample.

Analytical performance

In their feedback on this sample, the participating laboratories most frequently reported that C5OH carnitine levels were elevated (n=30), significantly elevated (n=2) or normal (n=2). Ten laboratories found elevated C6DC levels, while four classified them as normal. One participant reported an elevated concentration of free carnitine, while 15 others described it as normal. Other acylcarnitines (C2, C3, C5:1) were mentioned less frequently and were classified as normal in all cases.

Evaluation criteria: Two points are given for reporting the concentration of at least one of the key metabolites, C5OH or C6DC, as elevated or grossly elevated.

Diagnosis / Interpretative proficiency

Due to the frequently reported isolated increase in C5OH, most participants opted for corresponding diagnoses, with 3MCC being the most frequently mentioned (n=26), followed by HMG CLD (n=23) and 3MGA (n=17). Diagnoses such as multiple carboxylase deficiency or beta-ketothiolase deficiency were only mentioned as alternatives and less frequently (n=12 and n=6, respectively). Nine participants reported a normal AC profile for this sample.

Evaluation criteria: Reporting HMG CLD as principal or alternative diagnosis yields two points. Recommending organic acid analyses as an additional test yields one point.

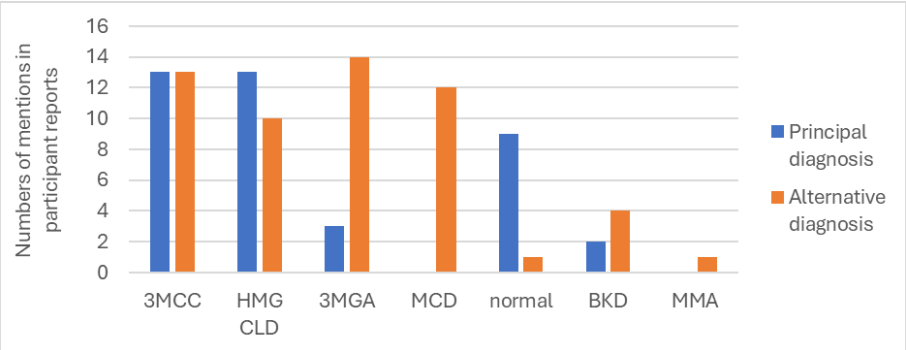
Due to the only subtle increase of the key metabolites in this sample, it was suggested to exclude the it from critical error assessment, which was approved at the autumn SAB meeting.

Recommendations

Most laboratories recommended to analyse urinary organic acids (n=34) or perform genetic testing (n=28). Measuring enzymatic activity was recommended by 10 labs.

Overall impression

Despite only marginally increased concentrations of the key metabolites, most participants performed well, resulting in an analytical proficiency of 83%, while interpretative proficiency was 74%.



Distribution of interpretations received for sample F

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 receiving your Performance Support Letter. Details of how to appeal poor performance are included in the Performance Support Letter sent to poor performing laboratories.

9.1. Detailed scores – Round 1

Lab n°	Patient A VLCADD			Patient B VLCADD			Patient C MAD			
	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	0	2	0	0	0	2	1	3	5
6	2	2	4	2	2	4	2	1	3	11
7	2	2	4	2	2	4	2	2	4	12
8	2	2	4	2	2	4	2	1	3	11
9	2	2	4	2	2	4	2	1	3	11
10	2	2	4	2	2	4	2	2	4	12
11	2	2	4	2	2	4	2	1	3	11
12	2	2	4	2	2	4	2	2	4	12
13	2	2	4	0	0	0	2	1	3	7
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	0	0	0	2	1	3	7
17	2	2	4	2	2	4	2	2	4	12
18	2	2	4	2	1	3	2	2	4	11
19	2	2	4	2	2	4	2	1	3	11
20	2	2	4	2	2	4	2	1	3	11
21	2	2	4	2	2	4	2	0	2	10
22	2	2	4	2	2	4	2	2	4	12
23	2	0	2	0	0	0	2	1	3	5
24	2	2	4	2	1	3	2	1	3	10
25	2	2	4	2	2	4	2	2	4	12
26	2	2	4	0	1	1	2	1	3	8

Lab n°	Patient A VLCADD			Patient B VLCADD			Patient C MAD			
	A	I	Total	A	I	Total	A	I	Total	Total
27	2	2	4	2	2	4	2	1	3	11
28	2	2	4	0	0	0	2	1	3	7
29	2	2	4	2	2	4	2	2	4	12
30	2	0	2	2	2	4	2	2	4	10
31	2	2	4	2	2	4	2	1	3	11
32	2	2	4	0	0	0	2	1	3	7
33	0	0	0	0	0	0	0	0	0	0
34	2	2	4	0	0	0	2	1	3	7
35	2	2	4	2	2	4	2	1	3	11
36	2	2	4	0	1	1	2	2	4	9
37	2	0	2	2	2	4	2	0	2	8
38	2	2	4	2	2	4	2	2	4	12
39	2	2	4	0	1	1	2	1	3	8
40	0	0	0	0	0	0	0	0	0	0
41	2	2	4	0	1	1	2	1	3	8
42	2	2	4	0	1	1	2	1	3	8
43	0	0	0	0	0	0	0	0	0	0
44	2	2	4	0	1	1	2	1	3	8
45	0	0	0	0	1	1	1	1	2	3
46	0	0	0	0	0	0	0	0	0	0
47	0	0	0	0	0	0	0	0	0	0

9.2. Detailed scores – Round 2

Lab n°	Patient D Normal			Patient E MMA mut(0)			Patient F HMG CLD			
	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	0	0	0	2	2	4	2	1	3	7
5	2	2	4	2	2	4	0	0	0	8
6	0	0	0	2	1	3	2	2	4	7
7	0	0	0	2	2	4	2	1	3	7
8	2	2	4	2	2	4	2	2	4	12
9	2	2	4	2	1	3	2	1	3	10
10	0	0	0	2	2	4	2	2	4	8
11	0	0	0	2	1	3	2	1	3	6
12	2	2	4	2	1	3	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	0	0	0	0	0	0	0	0	0	0
16	0	0	0	2	2	4	2	2	4	8
17	2	2	4	2	2	4	0	1	1	9
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	2	2	4	2	2	4	8
22	2	2	4	2	2	4	2	2	4	12
23	0	0	0	2	1	3	0	0	0	3
24	0	0	0	2	2	4	2	2	4	8
25	0	0	0	2	1	3	2	2	4	7
26	2	2	4	2	2	4	2	1	3	11
27	2	2	4	2	2	4	2	2	4	12
28	0	0	0	2	2	4	2	1	3	7
29	0	0	0	2	2	4	2	1	3	7
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	1	3	11
33	0	0	0	0	0	0	0	0	0	0

Lab n°	Patient D Normal			Patient E MMA mut(0)			Patient F HMG CLD			
	A	I	Total	A	I	Total	A	I	Total	Total
34	2	2	4	2	2	4	2	2	4	12
35	2	2	4	2	2	4	0	0	0	8
36	2	2	4	2	2	4	0	0	0	8
37	0	0	0	0	0	0	0	0	0	0
38	0	0	0	2	2	4	2	1	3	7
39	0	0	0	2	2	4	2	1	3	7
40	0	0	0	0	0	0	0	0	0	0
41	2	2	4	2	2	4	0	1	1	9
42	0	0	0	2	1	3	0	1	1	4
43	0	0	0	0	0	0	0	0	0	0
44	2	2	4	2	2	4	2	2	4	12
45	2	2	4	2	2	4	2	2	4	12
46	0	0	0	0	0	0	0	0	0	0
47	0	0	0	0	0	0	0	0	0	0

9.3. Total scores

Lab n°	A	B	C	D	E	F	Cumulative score	Cumulative score in %	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	0	4	3	19	79	
5	2	0	3	4	4	0	13	54	
6	4	4	3	0	3	4	18	75	
7	4	4	4	0	4	3	19	79	
8	4	4	3	4	4	4	23	96	
9	4	4	3	4	3	3	21	88	
10	4	4	4	0	4	4	20	83	
11	4	4	3	0	3	3	17	71	
12	4	4	4	4	3	4	23	96	
13	4	0	3	4	4	4	19	79	
14	4	4	4	4	4	4	24	100	
15	4	1	4	0	0	0	9	38	
16	4	0	3	0	4	4	15	62	
17	4	4	4	4	4	1	21	88	
18	4	3	4	4	4	4	23	96	
19	4	4	3	4	4	4	23	96	
20	4	4	3	4	4	4	23	96	
21	4	4	2	0	4	4	18	75	
22	4	4	4	4	4	4	24	100	
23	2	0	3	0	3	0	8	33	CE
24	4	3	3	0	4	4	18	75	
25	4	4	4	0	3	4	19	79	
26	4	1	3	4	4	3	19	79	
27	4	4	3	4	4	4	23	96	
28	4	0	3	0	4	3	14	58	
29	4	4	4	0	4	3	19	79	
30	2	4	4	4	4	4	22	92	
31	4	4	3	4	4	4	23	96	
32	4	0	3	4	4	3	18	75	
33	0	0	0	0	0	0	0	0	

Lab n°	A	B	C	D	E	F	Cumulative score	Cumulative score in %	Critical error
34	4	0	3	4	4	4	19	79	
35	4	4	3	4	4	0	19	79	
36	4	1	4	4	4	0	17	71	
37	2	4	2	0	0	0	8	33	
38	4	4	4	0	4	3	19	79	
39	4	1	3	0	4	3	15	62	
40	0	0	0	0	0	0	0	0	
41	4	1	3	4	4	1	17	71	
42	4	1	3	0	3	1	12	50	
43	0	0	0	0	0	0	0	0	
44	4	1	3	4	4	4	20	83	
45	0	1	2	4	4	4	15	62	CE
46	0	0	0	0	0	0	0	0	
47	0	0	0	0	0	0	0	0	

9.4. Performance

	Number of labs	% total labs
Satisfactory performers (≥ 71 % of adequate responses)	33	70
Unsatisfactory performers (< 71 % adequate responses and/or critical error)	7	15
Partial and non-submitters	7	15

9.5. Overall Proficiency

Sample	Diagnosis	Analytical (%)	Interpretation (%)	Total (%)
ACDB-DH-2025-A	VLCADD	98	88	93
ACDB-DH-2025-B	VLCADD	64	71	68
ACDB-DH-2025-C	MAD	99	68	83
ACDB-DH-2025-D	Normal	63	63	63
ACDB-DH-2025-E	MMA mut(0)	100	91	96
ACDB-DH-2025-F	HMG CLD	83	74	78

10. Tentative 2026 schedule

Sample distribution	4th February 2026
Start of analysis of Survey 2026/1 Website open	17th March 2026
Survey 2026/1 - Results submission	07th April 2026
Survey 2026/1 - Reports	May 2026
Start of analysis of Survey 2026/2 Website open	1st June 2026
Survey 2026/2 – Results submission	22nd June 2026
Survey 2026/2 - Reports	July/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.

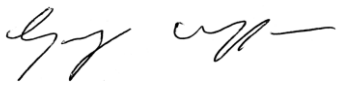
12. Questions, Suggestions and Complaints

If you have any questions, comments or suggestions please address to the Scientific Advisor of the scheme, Dr. Joachim Janda 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, 2026-01-21
Name and signature of Scientific Advisor

Dr. J. Janda
Scientific Advisor
Laboratory of Metabolic Diseases



Prof. Dr. G. F. Hoffmann
Director
Department of General Paediatrics

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	21 January 2026	2025 annual report published

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