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Acylcarnitines in dried blood spots

Centre: London United Kingdom

Final Report 2023

prepared by
Mr Charles Turner

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 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 Mr Charles Turner (Evelina London Children's Hospital) 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 2023 45 laboratories from many different countries participated in the ACDB London scheme.
There was no educational participant in 2023. 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 were 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	Country	Number of participants
Australia	3	Oman	1
Brazil	2	Poland	2
Canada	4	Qatar	1
Chili	1	Taiwan	1
Germany	1	Turkey	2
Ireland	1	United Kingdom	13
Italia	11	United States	1
New Zealand	1		

3. Design and logistics of the scheme including sample information

The scheme has been designed and planned by Mr Charles Tuner as Scientific Advisor and coordinated by CSCQ, the Swiss organisation for quality assurance in medical laboratories, both appointed by and according to procedures laid down by the ERNDIM Board. As usual, the samples used in 2023 were authentic human blood spot samples, all 6 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.
In 2023 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 2023 ERNDIM ACDB London scheme.

	1st Submission Round	2nd Submission Round
Sample ID's:	ACDB-UL-2023-A ACDB-UL-2023-B ACDB-UL-2023-C	ACDB-UL-2023-D ACDB-UL-2023-E ACDB-UL-2023-F
Shipment of samples	February 8th, 2023	
Start of analysis (clinical data available)	March 13th, 2023	June 5th, 2023
Reminder for result submission	March 27th, 2023	June 19th, 2023
Results submission deadline:	April 3rd, 2023	June 26th, 2023
Interim reports available on CSCQ website	February 2nd, 2024	February 2nd, 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 2023 ERNDIM ACDB London scheme.

Survey	Sample no.	Diagnosis
23-03-ACL	ACDB-UL-2023-A	Glutaric Acidaemia Type 1 OMIM 231670
	ACDB-UL-2023-B	VLCADD OMIM 201475
	ACDB-UL-2023-C	Propionyl CoA Carboxylase deficiency (OMIM 606054)
23-06-ACL	ACDB-UL-2023-D	Carnitine Palmitoyl Transferase Deficiency (CPT2) OMIM 600649
	ACDB-UL-2023-E	Methylmalonyl CoA mutase deficiency (MMA) OMIM 251000
	ACDB-UL-2023-F	Methylmalonyl CoA mutase deficiency (MMA) OMIM 251000

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 blood samples have been provided by the scheme organizers

Patient A: Glutaric Acidaemia Type 1 OMIM 231670
Patient B: VLCADD OMIM 201475
Patient C: Propionyl CoA Carboxylase deficiency (OMIM 606054)
Patient D: Carnitine Palmitoyl Transferase Deficiency (CPT2) OMIM 600649
Patient E: Methylmalonyl CoA mutase deficiency (MMA) OMIM 251000
Patient F: Methylmalonyl CoA mutase deficiency (MMA) OMIM 251000

5. Results

Returned results in the 2023 ERNDIM ACDB London scheme.

	Survey 1	**Survey 2**
Receipt of results	45	42
No answer	0	3

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 2023 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 30th, 2023).

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 on November 30th, 2023.

7.1. Score for satisfactory performance

At least 17 points from the maximum of 24 (70%). 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

Glutaric Acidaemia Type 1 due to mutations in the GCDH gene (19p13.2)OMIM 231670

Patient details provided to participants

30-year-old man presented in infancy with macrocephaly and dystonia. Today under treatment.

Patient details

Information given was: 30 year old man presented in infancy with macrocephaly and dystonia. Today under treatment. The sample was from a patient with Glutaryl-CoA dehydrogenase deficiency (glutaric acidaemia type 1, GA1). This, the common sample for 2023, was circulated to all three sections of the ERNDIM ACDB scheme

Analytical performance

Results were returned by all 45 participants in the ACDB London scheme. 44/45 detected the elevated glutaryl carnitine.

Diagnosis / Interpretative proficiency

All 44 participants who detected C5DC (glutaryl) carnitine included GA1 in their differential diagnosis.

Recommendations

Follow-up tests suggested to clarify/confirm the diagnosis were: urine organic acid analysis, including quantitative urinary 3-hydroxyglutarate: n=37, mutation analysis of the GCDH gene: n=37, enzyme activity in fibroblasts or leucocytes: n=11 (2 of these would only proceed to enzyme activity measurement if mutation analysis was not definitive), repeat acylcarnitine analysis: n=10, and urinary glutaryl carnitine measurement: n=5. One participant measured glutarate and 3-hydroxyglutarate on the original dried blood spot.

Clinical recommendations included an emergency regimen, a low lysine diet and carnitine supplementation, particularly since the free carnitine in this patient was low.

Scoring

The scoring criteria were 2 points for increased C5DC and/or appropriate ratio such as C5DC/C8 or C5DC/C16 (whether numerical or qualitative comment) and 2 points for GA1 as the primary or secondary likely diagnosis, with appropriate recommendations (UORG, genetics and/or enzyme analysis)

Overall impression

Performance overall was 96% for this sample. Most participants found the key metabolite and made the correct interpretation.

8.2. Patient B

The patient had very long chain acyl CoA dehydrogenase deficiency (VLCADD), OMIM 201475, and was well at the time of sampling.

Patient details provided to participants

Hypoglycaemia, hypotonia

Patient details

3y Old, presented with Hypoglycaemia, hypotonia. The sample was from a patient with very long chain acyl CoA dehydrogenase deficiency.

Analytical performance

The majority of participants 34/45 recognised that the tetradecenoyl (C14:1) carnitine or a C14:1 based ratio was disproportionately raised, giving an analytical proficiency of 75.6%.

Diagnosis / Interpretative proficiency

Those laboratories who found the C14:1 raised interpreted the pattern as potentially indicating VLCADD and made appropriate recommendations for confirmatory tests. Interpretative proficiency was 70%. The 10 labs who did not find the C14:1 raised considered the profile most likely normal. One included a possible unspecified fatty acid oxidation defect in their differential diagnosis and one CPT1 deficiency.

Recommendations

Of those considering VLCADD the most common follow-up tests were mutation analysis of the ACADVL gene: n=30, plasma acylcarnitine assay: n=26, and VLCADD enzyme activity in cultured cells: n=9 (with a further 2 if no definitive mutation found). Of those who did not include VLCADD 6 felt no further testing was indicated. 4 suggested some further testing, including plasma acylcarnitine analysis and tests to exclude hyperinsulinism

Scoring

The scoring criteria were 2 points for increased or clearly elevated C14:1 and/or C14:1 ratios and 2 points for VLCADD as primary or secondary diagnosis and comments for diagnostic confirmation: enzyme activity, ACADVL gene

Overall impression

This sample caused a problem for a number of labs. There was a clear split between the majority of labs who considered the C14:1 and/or C14:1 based ratios to be clearly indicative of VLCADD deficiency, and a minority of labs who considered the profile normal. Overall proficiency was only 72.8%.

8.3. Patient C

Propionyl CoA Carboxylase deficiency (OMIM 606054)

Patient details provided to participants

Acidosis, hyperammonaemia in infancy, on treatment

Patient details

This patient presented in the neonatal period with hyperammonaemic encephalopathy and acidosis. This sample was taken when the patient was well, on treatment, at a routine clinic visit.

Analytical performance

The profile from this patient showed an extremely high propionyl carnitine and high free carnitine: detected by all participants

Diagnosis / Interpretative proficiency

42/45 participants included propionic acidaemia in their differential diagnosis. One participant only mentioned methylmalonic acidaemia. Two gave no interpretation at all.

Recommendations

41/45 participants suggested suitable follow-up tests to distinguish propionic acidaemia from one of the methylmalonic acidaemias and confirm the diagnosis. The majority suggested urinary organic acid analysis. 4 suggested no follow-up tests, including the two who gave no interpretation at all.

Scoring

The scoring criteria were 2 points for increased or clearly elevated C3 and/or C3 ratios and elevated C0 and 2 points for propionic acidaemia on carnitine as primary or secondary diagnosis and comments for diagnostic confirmation: e.g UOA, methylcitrate & MMA, genotyping, enzyme activity

Overall impression

This was a very clear profile from a patient with propionic acidaemia on carnitine supplementation. All participants detected the grossly raised propionyl (C3) carnitine and free carnitine (C0). 42/43 of those who gave an interpretation included the correct diagnosis in their differential, the other only suggested methylmalonic acidaemia but suggested further tests which would have led to the correct diagnosis.

8.4. Patient D

Carnitine Palmitoyl Transferase Deficiency (CPT2) OMIM 600649

Patient details provided to participants

Hypoketotic hypoglycaemia

The sample was from a patient who initially presented with hypoketotic hypoglycaemia. The sample was taken at a routine clinic visit. The diagnosis was carnitine palmitoyl transferase type 2 deficiency OMIM 600649

Analytical performance

38/42 respondents commented on the low free carnitine. The concentrations of long chain acylcarnitines (C16, C18 and C18:1) were reported as normal by 13, 10, and 7 laboratories respectively. 1 laboratory commented on a high C16 acylcarnitine. Only 5 laboratories reported the (C16+C18:1)/C2 ratio, all of whom found it elevated.

Diagnosis / Interpretative proficiency

Only 14/42 respondents included CPT2 in their differential diagnosis. Overall interpretative proficiency was 59.5%

Recommendations

Those laboratories who included CPT2 in their differential diagnosis all suggested appropriate genotyping for CPT2.

Scoring

The sample was designated educational. Had it been included the scoring criteria would have been 2 points for low free carnitine C0 with disproportionately elevated long chain acylcarnitines C14-C18 or ratios e.g. (C16+C18:1)/C2 (whether numerical or qualitative comment) and 2 points for CPT2 as the primary or secondary likely diagnosis or unspecified long chain fatty acid disorder with suitable follow-up tests which would result in diagnosis

Overall impression

Overall proficiency for this sample was poor: 58.9%. The changes in the acylcarnitine profile were subtle and complicated by the relatively low free carnitine which resulted in no absolute elevation of long chain species. However, the (C16+C18:1)/C2 ratio was clearly elevated and should have allowed the diagnosis to be made. The difficulty of the sample means that it has been designated educational and removed from the overall scoring.

8.5. Patient E

Methylmalonyl CoA mutase deficiency (MMA) OMIM 251000 on carnitine supplements

Patient details provided to participants

Acidosis, hyperammonaemia in infancy, on treatment

Patient details

The sample was from a patient with Methylmalonyl CoA mutase deficiency (MMA) OMIM 251000, on carnitine supplements

Analytical performance

All participants reported on the grossly raised propionyl (C3) carnitine, giving an analytical proficiency of 100%. 23/42 participants also commented on a raised methylmalonyl (C4DC) carnitine.

Diagnosis / Interpretative proficiency

41/42 participants included MMA in their differential diagnosis, the other lab only mentioned raised propionyl carnitine and did not provide a diagnosis. All suggested appropriate follow-up tests to confirm/clarify the diagnosis. Overall proficiency of interpretation was 97.6%

Recommendations

41/42 respondents would proceed to urine organic acid analysis to clarify the diagnosis and guide further follow-up. 23 would measure plasma methylmalonic acid and 23 plasma total homocysteine. 33 would confirm the diagnosis by genotyping of the appropriate genes, with a further 4 suggesting enzyme activity measurements.

Scoring

The scoring criteria were 2 points for increased or clearly elevated C3 and/or C3 ratios such as C3/C2 or C3/C16 (whether numerical or qualitative comment) and 2 points for MMA as primary or secondary diagnosis and comments for diagnostic confirmation: e.g UOA, methylcitrate & MMA, genotyping, enzyme activity

Overall impression

This sample was straightforward. Overall proficiency was 98.8%

8.6. Patient F

Methylmalonyl CoA mutase deficiency (MMA) OMIM 251000

Patient details provided to participants

Acidosis, mild hyperammonaemia

Patient details

The sample was from a patient with Methylmalonyl CoA mutase deficiency (MMA) OMIM 251000, soon after presentation with acidosis, and mild hyperammonaemia. Carnitine supplementation had only just been started.

Analytical performance

Overall analytical performance was very good: 97.6%. 41/42 recognised the elevated C3 in this sample. The one laboratory who did not seemed to have a pre- or post analytical sample swap.

Diagnosis / Interpretative proficiency

All but one of the 41 labs who recognised the elevated C3 carnitine included MMA in their differential and suggested appropriate tests to clarify the diagnosis. Interpretive proficiency was 92.9%

Recommendations

40/42 respondents would proceed to urine organic acid analysis to clarify the diagnosis and guide further follow-up. 12 would measure plasma methylmalonic acid and 21 plasma total homocysteine. 31 would confirm the diagnosis by genotyping of the appropriate genes, with a further 4 suggesting enzyme activity measurements.

Scoring

The scoring criteria were 2 points for increased or clearly elevated C3 and/or C3 ratios and 2 points for MMA as primary or secondary diagnosis and comments for diagnostic confirmation: e.g. UOA, plasma MMA, genotyping, enzyme assay

Overall impression

The majority of laboratories found no difficulty in identifying the abnormalities in this sample, Overall proficiency was 95.2%

9. Scores of participants

All data transfer, the submission of data as well as the request and viewing of reports proceed via the 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

		Patient A			Patient B			Patient C			
	Lab No	Glutaric Acidaemia Type 1 OMIM 231670			Very long chain acyl CoA dehydrogenase deficiency VLCADD OMIM 201475			Propionyl CoA carboxylase deficiency (OMIM 606054)			
		A	I	Total	A	I	Total	A	I	Total	Total
	1	2	2	4	0	0	0	2	2	4	8
	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	0	0	0	2	2	4	8
	9	2	2	4	2	2	4	2	2	4	12
	10	2	2	4	2	2	4	2	2	4	12
	11	2	2	4	2	2	4	2	2	4	12
	12	2	2	4	2	2	4	2	2	4	12
	13	2	2	4	2	2	4	2	2	4	12
	14	2	2	4	2	2	4	2	2	4	12
	15	2	2	4	0	0	0	2	2	4	8
	16	2	2	4	2	2	4	2	2	4	12
	17	2	2	4	2	2	4	2	2	4	12
	18	2	2	4	2	2	4	2	2	4	12
	19	2	2	4	2	2	4	2	2	4	12
	20	2	2	4	2	1	3	2	1	3	10
	21	2	2	4	0	0	0	2	2	4	8
	22	2	2	4	2	2	4	2	2	4	12
	23	2	2	4	0	0	0	2	2	4	8
	24	2	2	4	2	2	4	2	2	4	12
	25	2	2	4	0	0	0	2	2	4	8
	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	1	3	2	1	3	10
	30	2	2	4	2	2	4	2	2	4	12
	31	2	2	4	0	0	0	2	2	4	8
	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	0	0	0	2	2	4	8
	37	2	2	4	2	0	2	2	2	4	10
	38	2	2	4	2	2	4	2	2	4	12
	39	2	0	2	0	0	0	2	0	2	4
	40	2	2	4	2	2	4	2	2	4	12
	41	2	2	4	2	2	4	2	2	4	12
	42	0	0	0	0	0	0	2	0	2	2
	43	2	1	3	2	2	4	2	1	3	10
	44	2	2	4	0	1	1	2	2	4	9
	45	2	2	4	2	0	2	2	2	4	10

Detailed scores – Round 2

	Lab No	Carnitine Palmitoyl Transferase Deficiency (CPT2) OMIM 600649 Educational Sample			Methylmalonyl CoA mutase deficiency (MMA) OMIM 251000			Methylmalonyl CoA mutase deficiency (MMA) OMIM 251000			
		A	I	Total	A	I	Total	A	I	Total	Total
	1	–	–	–	2	2	4	2	2	4	8
	2	–	–	–	2	2	4	2	2	4	8
	3	–	–	–	2	2	4	2	2	4	8
	4	–	–	–	2	2	4	2	2	4	8
	5	–	–	–	2	2	4	2	2	4	8
	6	–	–	–	2	2	4	2	2	4	8
	7	–	–	–	2	2	4	2	2	4	8
	8	–	–	–	2	2	4	2	2	4	8
	9	–	–	–	2	2	4	2	2	4	8
	10	–	–	–	2	2	4	2	2	4	8
	11	–	–	–	2	2	4	2	2	4	8
	12	–	–	–	2	2	4	2	2	4	8
	13										
	14	–	–	–	2	2	4	2	2	4	8
	15	–	–	–	2	2	4	2	2	4	8
	16	–	–	–	2	2	4	2	2	4	8
	17	–	–	–	2	2	4	2	2	4	8
	18	–	–	–	2	2	4	2	2	4	8
	19	–	–	–	2	2	4	2	2	4	8
	20	–	–	–	2	2	4	2	2	4	8
	21	–	–	–	2	2	4	2	2	4	8
	22	–	–	–	2	2	4	2	2	4	8
	23	–	–	–	2	2	4	2	2	4	8
	24	–	–	–	2	2	4	2	2	4	8
	25	–	–	–	2	2	4	2	2	4	8
	26	–	–	–	2	2	4	2	2	4	8
	27	–	–	–	2	2	4	2	2	4	8
	28	–	–	–	2	2	4	2	2	4	8
	29	–	–	–	2	2	4	2	1	3	7
	30	–	–	–	2	2	4	2	2	4	8
	31	–	–	–	2	2	4	2	1	3	7
	32	–	–	–	2	2	4	2	2	4	8
	33	–	–	–	2	2	4	2	2	4	8
	34	–	–	–	2	2	4	2	2	4	8
	35	–	–	–	2	2	4	2	2	4	8

	36	–	–	–	2	2	4	0	0	0	4
	37	–	–	–	2	2	4	2	2	4	8
	38	–	–	–	2	2	4	2	2	4	8
	39										
	40										
	41	–	–	–	2	2	4	2	2	4	8
	42	–	–	–	2	0	2	2	0	2	4
	43	–	–	–	2	2	4	2	2	4	8
	44	–	–	–	2	2	4	2	2	4	8
	45	–	–	–	2	2	4	2	2	4	8

Total scores

	Lab n°	A	B	C	D	E	F	Cumulative score	Cumulative score (%)	Critical error
	1	4	0	4	–	4	4	16	80	
	2	4	4	4	–	4	4	20	100	
	3	4	4	4	–	4	4	20	100	
	4	4	4	4	–	4	4	20	100	
	5	4	4	4	–	4	4	20	100	
	6	4	4	4	–	4	4	20	100	
	7	4	4	4	–	4	4	20	100	
	8	4	0	4	–	4	4	16	80	
	9	4	4	4	–	4	4	20	100	
	10	4	4	4	–	4	4	20	100	
	11	4	4	4	–	4	4	20	100	
	12	4	4	4	–	4	4	20	100	
	13	4	4	4				12	60	
	14	4	4	4	–	4	4	20	100	
	15	4	0	4	–	4	4	16	80	
	16	4	4	4	–	4	4	20	100	
	17	4	4	4	–	4	4	20	100	
	18	4	4	4	–	4	4	20	100	
	19	4	4	4	–	4	4	20	100	
	20	4	3	3	–	4	4	18	90	
	21	4	0	4	–	4	4	16	80	
	22	4	4	4	–	4	4	20	100	
	23	4	0	4	–	4	4	16	80	
	24	4	4	4	–	4	4	20	100	
	25	4	0	4	–	4	4	16	80	
	26	4	4	4	–	4	4	20	100	
	27	4	4	4	–	4	4	20	100	
	28	4	4	4	–	4	4	20	100	
	29	4	3	3	–	4	3	17	85	
	30	4	4	4	–	4	4	20	100	
	31	4	0	4	–	4	3	15	75	
	32	4	4	4	–	4	4	20	100	
	33	4	4	4	–	4	4	20	100	
	34	4	4	4	–	4	4	20	100	
	35	4	4	4	–	4	4	20	100	
	36	4	0	4	–	4	0	12	60	
	37	4	2	4	–	4	4	18	90	
	38	4	4	4	–	4	4	20	100	
	39	2	0	2				4	20	

	40	4	4	4				12	60	
	41	4	4	4	–	4	4	20	100	
	42	0	0	2	–	2	2	6	30	
	43	3	4	3	–	4	4	18	90	
	44	4	1	4	–	4	4	17	85	
	45	4	2	4	–	4	4	18	90	

Performance

	Number of labs	% total labs
Satisfactory performers (≥ 70 % of adequate responses)	40	89
Unsatisfactory performers (< 70 % adequate responses and/or critical error)	2	4
Partial and non-submitters	3	7

Overall Proficiency

Sample	Diagnosis	Analytical (%)	Interpretation (%)	Total (%)
ACDB-UL-2023-A	Glutaric Acidaemia Type 1 OMIM 231670	98	94	96
ACDB-UL-2023-B	VLCADD OMIM 201475	76	70	73
ACDB-UL-2023-C	Propionyl CoA Carboxylase deficiency (OMIM 606054)	100	92	96
ACDB-UL-2023-D	Carnitine Palmitoyl Transferase Deficiency (CPT2) OMIM 600649	--	--	--
ACDB-UL-2023-E	Methylmalonyl CoA mutase deficiency (MMA) OMIM 251000	100	98	99
ACDB-UL-2023-F	Methylmalonyl CoA mutase deficiency (MMA) OMIM 251000	98	93	95

10. Preview of the scheme in 2024

Erin Emmett, (Erin.Emmett@gstt.nhs.uk) will take over as Scientific Advisor for the 2024 scheme.

Sample distribution	8th February 2024
Start of analysis of Survey 2024/1 Website open	13th March 2024
Survey 2024/1 - Results submission	2rd April 2024
Survey 2024/1 - Reports	April/May 2024
Start of analysis of Survey 2023/2 Website open	5th June 2024
Survey 2024/2 – Results submission	19th June 2024
Survey 2024/2 - Reports	July/August 2024
Annual Report 2024	January 2025

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, Mr Charles Turner (chas.turner@kcl.ac.uk) and/or to the ERNDIM Administration Office (admin@erndim.org)

Date of report, 2024-04-26
Name and signature of Scientific Advisor

Mr Charles Turner
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	2nd May 2024	2023 annual report published

END