ERNDIM Administration Office
c/o EMQN CIC, Unit 4, Enterprise House,
Manchester Science Park, Pencroft Way,
Manchester M15 6SE, United Kingdom
Tel: +44 161 7574952
Fax: +44 161 850 1145
Email: admin@erndim.org

Scientific Coordination
Mrs Joanne Croft
Dept of Clinical Chemistry
Sheffield Children's NHS Foundation
Trust, Western Bank
Sheffield, S10 2TH
United Kingdom
Tel: +44(0)114 271 7000 Ext 17267
Fax: +44(0)114 276 6205
Email: joanne.croft4@nhs.net

Scheme Organisation
CSCQ (Quality Control Centre, Switzerland)
1)Alessandro Salemma 2)Nicola Braik
2 chemin du Petit-Bel-Air
1225 Chêne-Bourg
Switzerland,
Tel: +41 22 305 52 36
Email: 1)alessandro.salemma@hcuge.ch
2)nicola.braik@hcuge.ch

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Diagnostic Proficiency Testing

Centre: United Kingdom

Final Report 2023

prepared by
Mrs Joanne Croft

Note: This annual report is intended for participants of the ERNDIM DPT UK scheme. The contents should not be used for any publication without permission of the Scientific Advisor.

The fact that your laboratory participates in ERNDIM schemes is not confidential, however, the raw data and performance scores are confidential and will only be shared within ERNDIM for the purpose of evaluating your laboratories performance, unless ERNDIM is required to disclose performance data by a relevant government agency. For details please see the terms and conditions on page18 and the ERNDIM Privacy Policy on www.erndim.org.

In 2023, 20 labs participated in the UK Diagnostic Proficiency Testing Scheme.

1. Geographical distribution of participants

All 20 participants submitted results for both submission rounds.

Country	Number of participants
Australia	1
Czech Republic	1
Ireland	1
New Zealand	1
Spain	1
United Kingdom	15

[1] If this report is not Version 1 for this scheme year, go to APPENDIX 1 for details of the changes made since the last version of this document.

2. Design and logistics of the scheme including sample information

The scheme has been designed and planned by Joanne Croft as Scientific Advisor and coordinated by Alessandro Salemma as scheme organiser (sub-contractor on behalf of CSCQ), both appointed by and according to procedures laid down the ERNDIM Board.

CSCQ dispatches DPT EQA samples to the scheme participants and provides a website for on-line submission of results and access to scheme reports. Existing DPT scheme participants can log on to the CSCQ results submission website at:

https://cscq.hcuge.ch/cscq/ERNDIM/Initial/Initial.php

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

Origin of samples: all urine samples sent this year (apart from the common sample) were historical samples held in the freezers at the scheme organizers laboratory.

The samples have been heat-treated. They were pre-analysed in our institute after 3 days incubation at ambient temperature (to mimic possible changes that might arise during transport). In all six samples the typical metabolic profiles were preserved after this process.
Mailing: bulk samples were sent to CSCQ in Switzerland at room temperature using TNT. Samples were then aliquoted and an aliquot sent back to the organising laboratory for confirmatory testing. Aliquots were then couriered to all participating laboratories in February 2023.

3. Tests

Analyses of amino acids, organic acids, mucopolysaccharides, oligosaccharides and purines/pyrimidines were required in 2023.

4. Schedule of the scheme

- 8th February, 2023: shipment of samples of Survey 1 and Survey 2
- 13th March, 2023: analysis start and website submission available for Survey 1
- 3rd April, 2023: deadline for result submission (Survey 1)
- 5th June, 2023: analysis start and website submission available for Survey 2
- 26th June, 2023: deadline for result submission (Survey 2)
- June, 2023: interim report of Survey 1 by e-mail
- August, 2023: interim report of Survey 2 by e-mail
- 21st Sept, 2023: on line DPT UK participants meeting
- 30th Nov, 2023: SAB meeting in Prague
- Feb 2024: annual report with final scoring issued by e-mail.

5. Results

20 of 20 labs returned results for both surveys.

	Survey 1	**Survey 2**
Receipt of results	20	20
No answer	0	0

6. Web site reporting

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

- Selection of tests: **don't select a test if you will not perform it**, otherwise the evaluation program includes it in the report.
- Results
  - Give quantitative data as much as possible.

- Enter the key metabolites with the evaluation **in the tables** even if you don't give quantitative data.
- If the profile is normal: enter "Normal profile" in "Key metabolites".
- **Don't enter results in the "comments" window**, **otherwise your results will not be included in the evaluation program.**

- Recommendations = **advice for further investigation**.
  - Scored together with the interpretative score.
  - Advice for treatment are not scored.
  - **Don't give advice for further investigation in "Comments on diagnosis"**: it will not be included in the evaluation program.

7. Scoring and evaluation of results

Information regarding procedures for establishment of assigned values, statistical analysis, interpretation of statistical analysis etc. can be found in generic documents on the ERNDIM website.
The scoring system has been established by the International Scientific Advisory Board of ERNDIM. Two criteria are evaluated: 1) analytical performance, 2) interpretative proficiency also considering recommendations for further investigations.

A	Analytical performance	Correct results of the appropriate tests	2
		Partially correct or non-standard methods	1
		Unsatisfactory or misleading	0
I	Interpretative proficiency & Recommendations	Good (diagnosis was established)	2
		Helpful but incomplete	1
		Misleading or wrong diagnosis	0

The total score is calculated as a sum of these two criteria. The maximum to be achieved is 4 points per sample.

Scoring and certificate of participation: scoring is carried by a second assessor who changes every year as well as by the scientific advisor. The results of DPT UK 2023 have also been scored by Dr Christine Saban, the scientific advisor for the DPT France scheme. At the SAB meeting in November 2023, the definitive scores have been finalized. The concept of critical error was introduced in 2014. A critical error is defined as an error resulting from seriously misleading analytical findings and /or interpretations with serious clinical consequences for the patient. Thus labs failing to make a correct diagnosis of a sample considered as eligible for this category will be deemed not to have reached a satisfactory performance even if their total points for the year exceed the limit set at the SAB. For 2023 no participants in the UK DPT scheme will receive a critical error.

A certificate of participation will be issued and it will be additionally include whether the participant has received a performance support letter. This performance support letter is sent out if the performance is evaluated as unsatisfactory. One performance support letter will be sent by the Scheme Advisor for 2023.

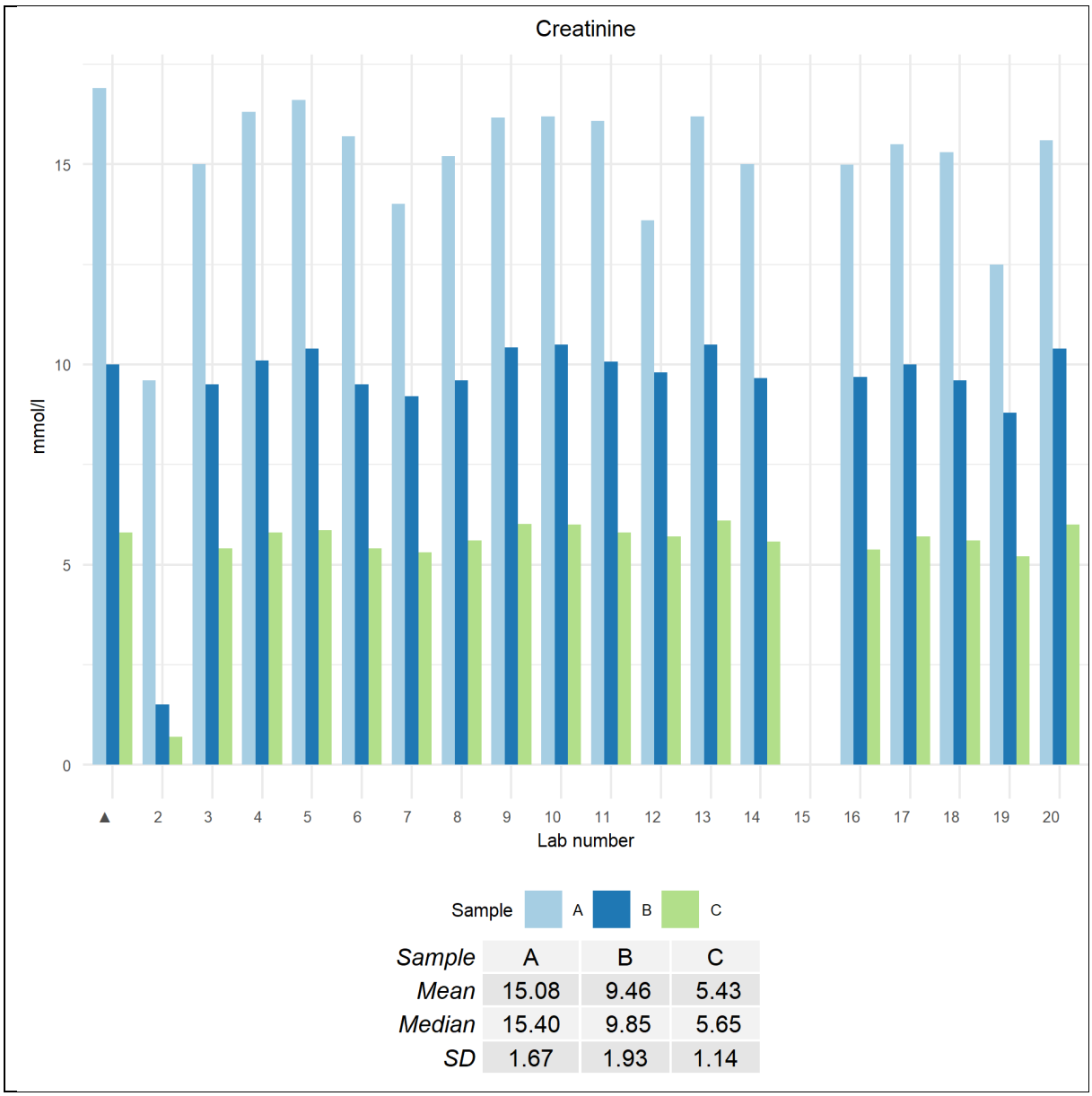
7.1. Score for satisfactory performance

The minimum satisfactory score is 17 out of 24. Whether Sample E (urine from a patient with LCHADD, collected while the patient was well) should be classed as educational or not was discussed at the SAB meeting in November. It was concluded that Sample E should not be classed as educational as other Scientific Advisors felt that the clinical details provided and the level of metabolites seen by organic acid analysis were enough to correctly diagnose (as indeed 9 participants did).

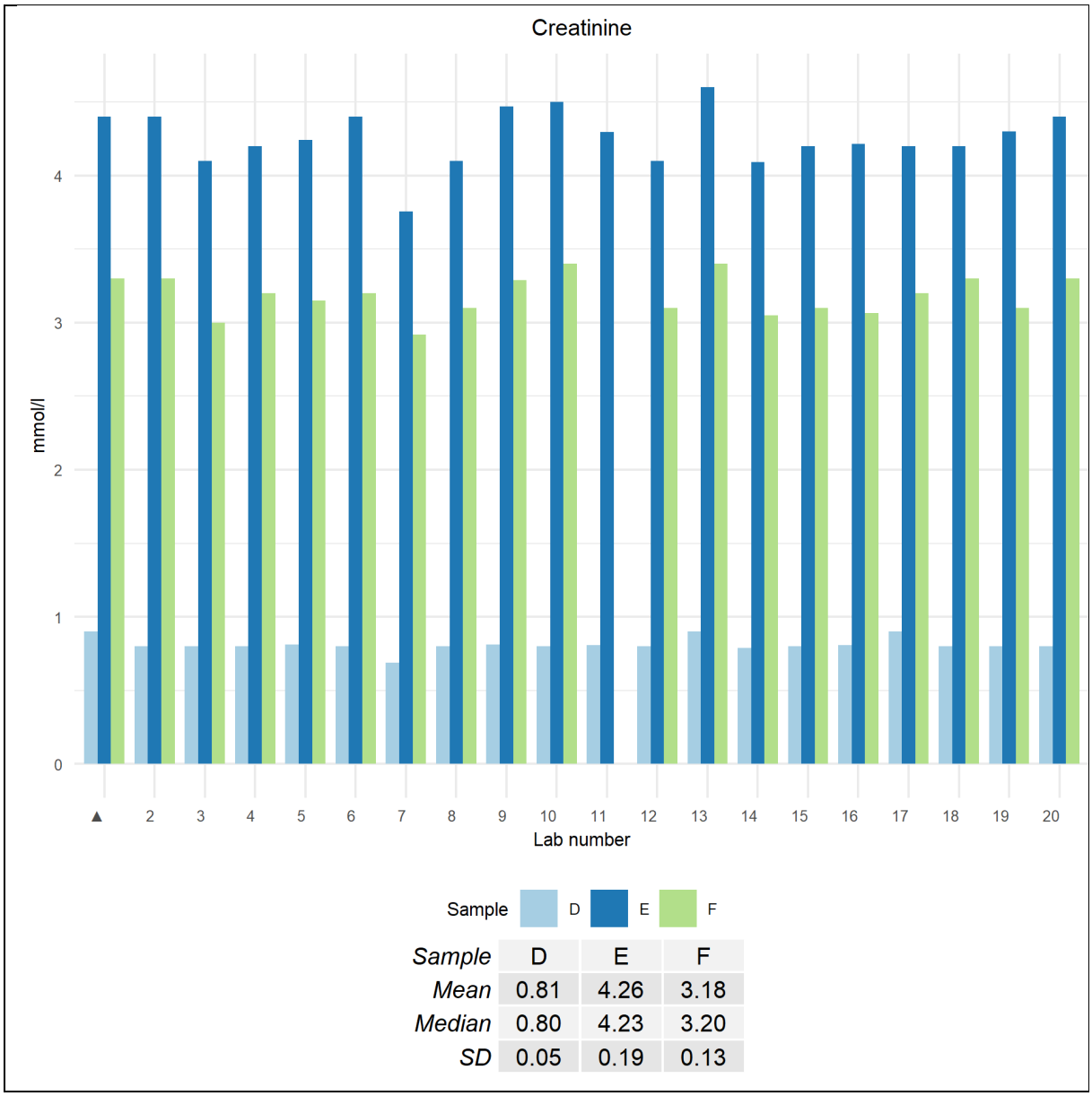
8. Results of samples and evaluation of reporting

8.1. Creatinine measurement for all samples

Values provided by participating laboratories for creatinine concentration in the main were similar (see graphs below). For samples A, B and C it appears that one laboratory (lab 15) did not provide creatinine values. However, due to this laboratory changing the method used to measure creatinine the software couldn't 'pick up' the creatinine results for the graph (their results are A = 15.2, B = 9.9, C = 5.6). This wasn't an issue for the second survey.
For sample F, 1 participant (lab 11) did not provide a creatinine result.
For samples A, B and C, laboratory 2 gave lower creatinine concentrations, all of which are lower than -2 S.Ds from the mean. This has contributed to this laboratory reporting an increased glycosaminoglycan concentration and glycine concentration for sample B. For samples D, E and F this was not an issue.



Sample	A	B	C
Mean	15.08	9.46	5.43
Median	15.40	9.85	5.65
SD	1.67	1.93	1.14



Sample	D	E	F
Mean	0.81	4.26	3.18
Median	0.80	4.23	3.20
SD	0.05	0.19	0.13

8.2. Patient A

Argininosuccinate lyase deficiency

Patient details provided to participants

This boy was referred at the age of 7 years with attention deficit disorder. The sample was collected at the age of 25 years on the specific treatment.

Patient details

This was the common sample sent to all Diagnostic Proficiency Scheme participants.

Marking scheme

(used by all the DPT scheme organisers)

- Analytical
  - Detecting increased concentration of argininosuccinic acid and/or it's anhydrides: 2 marks
- Interpretation

- Argininosuccinic aciduria due to argininosuccinate lyase deficiency: 2 marks
- Other urea cycle disorder(s): 1 mark

Analytical performance

- 14/20 participants scored 2 marks
- 6/20 participants scored 0 marks
  - did not detect the argininosuccinic acid or it's anhydrides
- Very low concentration of argininosuccinic acid in this sample
  - 7 participants provided a quantitative result (umol/mmol creatinine)
  - median = 34, mean = 46, min, max 0.08 - 150
- Citrulline was also not increased
- No participants reported an increase in orotic acid

We perform amino acid analysis by ion exchange chromatography (Biochrom analyser) and when we analysed this sample the argininosuccinic acid (ASA) eluted under the leucine peak. The Biochrom programme has now been altered to improve separation between ASA and leucine and the laboratory runs an ASA standard (spiked calibrant) regularly to check that we still have separation between these 2 amino acids. We also performed thin layer chromatography on this sample but due to the low concentration of the ASA the typical rocket shaped spot was not observed.

Diagnosis / Interpretative proficiency

- 14/20 participants scored 2 marks
- 6/20 participants scored 0 marks
- All the participants who scored 2 marks for analysis interpreted the analytical findings correctly.
- All the participants who scored 0 marks for analysis did not conclude to the correct diagnosis
  - (5 - no diagnosis reached, 1 - MPS III)

Recommendations

Recommendations are those provided by the 14 laboratories who gave the correct diagnosis:

- Plasma amino acids - 13/14
- Blood ammonia - 11/14
- Mutation analysis of ASL gene - 13/14
- Quantitative orotic acid - 4/14
- Refer to metabolic team (or check already under care of) - 10/14
- Test siblings – 4/14
- Enzyme studies - 3/14

Overall impression

This was a difficult sample with a low concentration of argininosuccinic acid. From all the DPT schemes, 20 out of 98 participants did not detect the argininosuccinic acid. Therefore it has been decided by the Scientific Advisory Board that no critical errors for this sample will be given.

8.3. Patient B

Sample from a healthy adult male with no known inborn error of metabolism.

Patient details provided to participants

Muscle pains after prolonged exercise.

Patient details

This sample was donated by an adult male with no known inborn error of metabolism.

Marking Scheme

- Analytical
    - Performing at least 3 analyses (not including the 'pre-investigations') and finding no significant abnormality – 2 marks
    - Performing less than 3 analyses and finding no significant abnormality – 1 mark
    - Reporting a significant abnormality – 0 marks

- Interpretation
    - Concluding no significant abnormality (or similar) – 2 marks
    - Concluding the wrong diagnosis – 0 marks
    - Leaving diagnosis section blank or putting n/a – 0 marks

Analytical performance

Analytical performance was good for this sample with 16/20 participants scoring 2 marks and 4/20 scoring 1 mark. Those participants who did not score 2 marks for analysis commented on an abnormal finding that no other participants had found.

Diagnosis / Interpretative proficiency

- 18/20 scored 2 marks
- 2/20 scored 1 mark
- 15/16 participants who scored 2 marks for analysis correctly interpreted the analytical findings and reported as 'no significant abnormality' or similar. The remaining participant who scored 2 marks for analysis reported, as the most likely diagnosis, the sample having come from a patient with carnitine palmitoyl transferase type 2 (CPT2) deficiency.
- 3 participants who had found an abnormality on analysis concluded that this was not clinically significant (so scored 3 overall for this sample). 1 participant concluded to glycogen storage disorder type 5.

There were many different alternative diagnoses provided by participants for this sample:

- Muscle glycogen storage disorder
- Fatty acid oxidation defect
- Mitochondrial myopathies
- Mucopolysaccharidosis disorders
- Lipin 1 deficiency
- Myoadenylate deaminase deficiency

Recommendations

Given the clinical details of muscle pain after prolonged exercise, 15/20 participants suggested that creatine kinase should be measured. Another very popular and sensible recommendation was to measure acylcarnitines (19/20) as some of the fatty acid oxidation disorders can present in adulthood with muscle pain e.g. CPT2 deficiency. Quite a few participants (8/20) mentioned that glycogen storage disorders should be considered.

Overall impression

As has been seen previously when samples from individuals with no inborn error of metabolism have been used in the DPT scheme there were a lot of diagnoses and recommendations provided.
Overall, proficiency for this sample was good.

8.1. Patient C

Medium chain Acyl-CoA dehydrogenase deficiency (MCADD)

Patient details provided to participants

Presented at A and E – drowsy and not feeding. Sample collected during time of intercurrent illness.

Patient details

The patient was diagnosed at the time of presentation and the sample collected a few months later during a period of intercurrent illness.

Marking Scheme

- Analytical
  - Detecting hexanoylglycine, suberylglycine and phenylpropanoylglycine – 2 marks
- Interpretation
  - Medium chain acyl CoA dehydrogenase (MCAD) deficiency – 2 marks

Analytical performance

19/20 participants reported the increased concentrations of hexanoylglycine, phenylpropionylglycine and suberylglycine in this sample and therefore scored 2 marks for analysis. 1 participant did not report the increased phenylpropionylglycine and therefore scored 1 for analysis.

Diagnosis / Interpretative proficiency

All participants correctly gave MCADD as the diagnosis. 7/20 participants also stated that other disorders of fatty acid oxidation should be considered with 6/7 specifically mentioning glutaric aciduria type 2/multiple acyl CoA dehydrogenase deficiency/riboflavin responsive disorders.

Recommendations

- Urgent glucose - 5/20
  - (one additional lab recommended 'routine biochemical parameters')
- Acylcarnitines (either DBS or plasma) - 18/20
- Carnitine monitoring – 2/20
  - (1 participant did not mention acylcarnitines/carnitine)
- Mutation analysis of the ACADM gene - 19/20
- Referral to metabolic team - 18/20
- Sibling testing - 9/20
- Enzyme assay – 3/20
- Other recommendations included the need for an emergency regimen and avoidance of fasting. 4 participants mentioned doing urgent ammonia due to the clinical information provided.

Overall impression

This was a straightforward sample with very good proficiency.

8.1. Patient D

Hypophosphatasia diagnosed within the first 6 months of life due to the clinical finding of rickets. Likely to be the infantile form.

Patient details provided to participants

Failure to thrive. ? rickets. Found to have low alkaline phosphatase.

Patient details

This sample came from a patient who was found to have rickets during the first year of life. Biochemical investigation showed persistently low alkaline phosphatase levels and increased phosphoethanolamine.

Marking Scheme

- Analytical
  - Detecting increased phosphoethanolamine – 2 marks
  - Stating that phosphoethanolamine is at the high end of the reference range – 1 mark
- Interpretive
  - Hypophosphatasia – 2 marks
  - Stating that Hypophosphatasia cannot be excluded – 1 mark

Analytical performance

- 16 participants scored 2 marks
- 2 participants scored 1 mark (i.e. stated PEA was at high end of ref range)
- 2 participants scored 0 marks (stated PEA was normal)
- 18 participants provided a quantitative result for phosphoethanolamine
  - Median = 51.6, mean = 52.2, min, max = 11.0, 75.0
  - Our result for this sample = 43 µmol/mmol creat. (ref. < 10)
- We don't have an age related reference range – intend to correct this! The range is higher in infants (this sample was collected at 5 months of age)

Diagnosis / Interpretative proficiency

Most laboratories (16/20) scored 2 marks. The other 4 laboratories scored 1 mark as they stated that hypophosphatasia could not be excluded (these included the participants who reported phosphoethanolamine as not increased).

Recommendations

All participants gave useful recommendations for further follow up/definitive diagnosis

- These included:
  - measurement of serum/plasma alkaline phosphatase (or reviewing previous results) - (5/20)
  - pyridoxal 5 phosphate or pyridoxal phosphate:pyridoxic acid ratio - (17/20)
  - bone profile including zinc and magnesium – (4/20)
  - bone radiology - (4/20)
  - mutation analysis of the ALPL gene – (17/20)
  - consider ERT – (3/20) (asfotase alfa)
  - multidisciplinary team input – (12/20)

Overall impression

Proficiency for this sample was good with no participants scoring 0 marks (although it may be possible that some laboratories concluded to the correct diagnosis based on the clinical details provided alone).

8.1. Patient E

Long chain hydroxy acyl CoA dehydrogenase deficiency

Patient details provided to participants

Cardiomyopathy, hypotonia and hypoketotic hypoglycaemia. Sample collected at 13 years of age when on treatment and well.

Patient details

This sample was donated by a patient with long chain hydroxy acyl CoA dehydrogenase (LCHAD) deficiency. The sample was collected when the patient was on treatment and well which accounts for the low concentration of the relevant metabolites in this sample.

Marking Scheme

- Analytical
    - Identifying increased hydroxydicarboxylic acids on organic acids – 2 marks
- Interpretation
    - LCHADD – 2 marks
    - Mitochondrial trifunctional protein deficiency – 2 marks
    - Fatty acid oxidation defect – 2 marks
    - Suggestion to do acylcarnitine analysis (plasma or DBS) if diagnosis not correct – 1 mark

Analytical performance

- 9/20 participants scored 2 marks
- 1/20 participant scored 1 mark
- 10/20 participants scored 0 marks

Diagnosis / Interpretative proficiency

- 12/20 participants scored 2 marks
- 8/20 participants scored 1 mark

Many participants who did not detect the key metabolites still included a fatty acid oxidation defect in their differential diagnosis based on the clinical history provided. No participants scored zero for this sample.

Recommendations

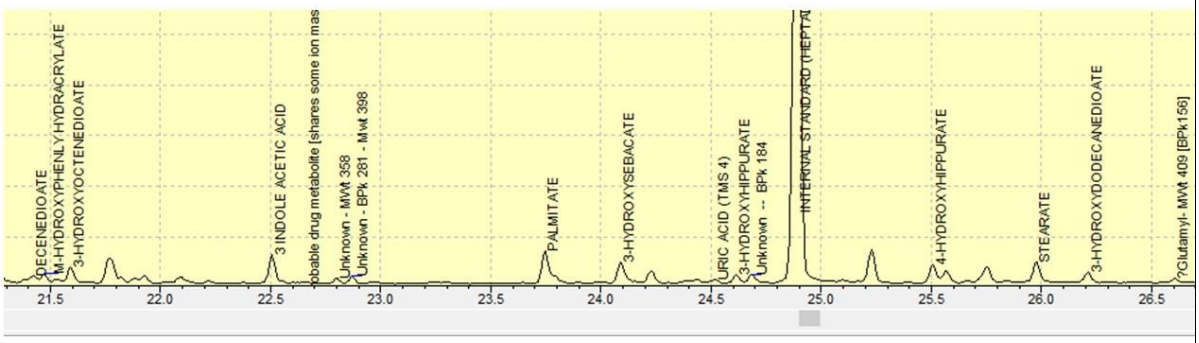
- Acylcarnitine analysis (DBS or plasma) – 20/20
- Many suggested genetic analysis but only 4 specifically gave the HADHA/HADHB genes, these being some of the participants who had given LCHADD as their primary diagnosis.

Overall impression

The concentration of the relevant metabolites in this sample, collected from a patient with LCHAD deficiency but who was on treatment and well at the time, were small. Many participants who did not detect the key metabolites by urine organic acid analysis included a fatty acid oxidation defect in their differential diagnosis based on the clinical history provided.

No critical errors are to be awarded for this sample as all participants gave a correct/helpful diagnosis and recommendations.

Though proficiency for this sample is low (64% overall) it was not deemed to be suitable as an educational sample by the Scientific Advisory Board due to the detectable metabolites and the clinical details provided. Therefore marks for this sample are to be taken into account in the total score.



Sample E Organic Acid Chromatogram - RT 21.5 to 26.5 minutes

8.1. Patient F

Cystinuria.

Patient details provided to participants

Epilepsy

Patient details

This sample was donated by a patient with cystinuria. We made up the clinical details for this case as we have diagnosed cases of cystinuria in the past as incidental findings. Therefore any participants who gave cystinuria-hypotonia syndrome as the diagnosis were scored with full marks. Given the clinical details this would need to be followed up as a potential diagnosis if this was a real case.

Marking Scheme

- Analytical
  - Identifying increased cystine, arginine, ornithine and lysine – 2 marks
  - Identifying increased concentration of less than all 4 amino acids – 1 mark

- Interpretation
  - Cystinuria – 2 marks
  - Hypotonia/cystinuria syndrome (2p21 deletion syndrome) – 2 marks

Analytical performance

All 20 participants detected the increased concentration of the basic amino acids therefore scoring 2 marks.
17 provided quantitative results (µmol/mmol creatinine)

Amino Acid	Mean	Median	Min	Max
Cystine	279	273	173	408
Arginine	752	745	491	972
Ornithine	346	356	221	429
Lysine	1296	1317	890	1668

Diagnosis / Interpretative proficiency

All participants gave either cystinuria or hypotonia-cystinuria syndrome as the primary diagnosis, therefore scoring 2 marks.

Even though this sample came from a patient with cystinuria, participants quite rightly pointed out that with the clinical details of epilepsy that the contiguous gene syndrome giving rise to hypotonia - cystinuria syndrome was also a possibility.

Recommendations

- All participants gave helpful recommendations
    - Repeat urine amino acids – (7/20)
    - Referral to paediatric renal team – (11/20)
    - Refer to metabolic team – (6/20)
    - Referral to metabolic team to investigate for other causes of epilepsy – (1/20)
    - Mutation analysis of the SLC3A1 gene – (16/20)
    - Mutation analysis of the SLC7A9 gene – (17/20)
    - Mutation analysis for the 2p21 deletion syndrome – (8/20)

Overall impression

Proficiency for this sample was excellent (100%).

9. Scores of participants

All data transfer, the submission of data as well as the request and viewing of reports proceed via the DPT-CSCQ results website. The results of your laboratory are confidential and only accessible to you (with your username and password). The anonymous scores of all laboratories are accessible to all participants and only in your version is your laboratory highlighted in the leftmost column.

If your laboratory is assigned poor performance and you wish to appeal against this classification please email the ERNDIM Administration Office (admin@erndim.org), with full details of the reason for your appeal, within one month receiving your Performance Support Letter. Details of how to appeal poor performance are included in the Performance Support Letter sent to poor performing laboratories

Detailed scores – **Round 1**

Lab n°	**Patient A** **Argininosuccinate lyase deficiency.**			**Patient B** **No known inherited metabolic disorder.**			**Patient C** **Medium chain Acyl CoA dehydrogenase deficiency (MCADD)**			
	A	**I**	**Total**	**A**	**I**	**Total**	**A**	**I**	**Total**	**Total**
1	2	2	4	2	2	4	2	2	4	12
2	0	0	0	1	2	3	2	2	4	7
3	2	2	4	2	2	4	2	2	4	12
4	0	0	0	2	2	4	2	2	4	8
5	2	2	4	2	2	4	2	2	4	12
6	2	2	4	2	2	4	2	2	4	12
7	0	0	0	2	2	4	1	2	3	7
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	0	0	0	1	2	3	2	2	4	7
12	0	0	0	1	2	3	2	2	4	7
13	2	2	4	2	2	4	2	2	4	12
14	2	2	4	1	1	2	2	2	4	10
15	2	2	4	2	1	3	2	2	4	11
16	2	2	4	2	2	4	2	2	4	12
17	2	2	4	2	2	4	2	2	4	12
18	0	0	0	2	2	4	2	2	4	8
19	2	2	4	2	2	4	2	2	4	12
20	2	2	4	2	2	4	2	2	4	12

Detailed scores – Round 2

Lab n°	Patient D Hypophosphatasia			Patient E LCHADD			Patient F Cystinuria			
	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	0	2	2	2	2	4	10
3	2	2	4	2	2	4	2	2	4	12
4	2	1	3	0	2	2	2	2	4	9
5	2	2	4	2	2	4	2	2	4	12
6	2	2	4	0	1	1	2	2	4	9
7	2	2	4	2	2	4	2	2	4	12
8	2	2	4	2	2	4	2	2	4	12
9	2	1	3	0	1	1	2	2	4	8
10	0	1	1	0	1	1	2	2	4	6
11	2	2	4	0	1	1	2	2	4	9
12	2	2	4	2	2	4	2	2	4	12
13	2	2	4	0	1	1	2	2	4	9
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	0	1	1	2	2	4	9
17	0	1	1	0	1	1	2	2	4	6
18	2	2	4	0	1	1	2	2	4	9
19	1	2	3	1	2	3	2	2	4	10
20	1	2	3	2	2	4	2	2	4	11

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	0	3	4	4	2	4	17	71	
3	4	4	4	4	4	4	24	100	
4	0	4	4	3	2	4	17	71	
5	4	4	4	4	4	4	24	100	
6	4	4	4	4	1	4	21	88	
7	0	4	3	4	4	4	19	79	
8	4	4	4	4	4	4	24	100	
9	4	4	4	3	1	4	20	83	
10	4	4	4	1	1	4	18	75	
11	0	3	4	4	1	4	16	67	
12	0	3	4	4	4	4	19	79	
13	4	4	4	4	1	4	21	88	
14	4	2	4	4	4	4	22	92	
15	4	3	4	4	4	4	23	96	
16	4	4	4	4	1	4	21	88	
17	4	4	4	1	1	4	18	75	
18	0	4	4	4	1	4	17	71	
19	4	4	4	3	3	4	22	92	
20	4	4	4	3	4	4	23	96	

Performance

	Number of labs	% total labs
Satisfactory performers (≥ 70 % of adequate responses)	19	95
Unsatisfactory performers (< 70 % adequate responses and/or critical error)	1	5
Partial and non-submitters	0	0

Overall Proficiency

Sample	Diagnosis	Analytical (%)	Interpretation (%)	Total (%)
DPT-UK-2023-A	Argininosuccinate lyase deficiency.	70	70	70
DPT-UK-2023-B	No known inherited metabolic disorder.	90	95	93
DPT-UK-2023-C	Medium chain Acyl CoA dehydrogenase deficiency (MCADD)	98	100	99
DPT-UK-2023-D	Hypophosphatasia	85	90	88
DPT-UK-2023-E	LCHADD	48	80	64
DPT-UK-2023-F	Cystinuria	100	100	100

10. Annual meeting of participants

This took place in Jerusalem on Tuesday 29th August 2023, before the SSIEM Meeting. Workshops for the different DPT schemes are held prior to the ERNDIM meeting of participants. However, as the UK DPT Scientific Advisor was unable to attend in Jerusalem, an on-line meeting was held on Thursday 21st September for the UK DPT participants which was led by Joanne Croft and Claire Hart, Deputy Scientific Advisor for the UK DPT scheme. We have found that these on-line meetings are better attended than the workshops, and that multiple staff members from one participating laboratory can join if they so wish.

We remind you that attending these meetings is an important part of the proficiency testing. The goal of the program is to **improve** the competence of the participating laboratories, which includes the critical review of all results with a discussion about improvements.

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

- **Training**: SSIEM Academy training courses.
  - A 2 day course has been organized for Monday and Tuesday 22nd/23rd April 2024 in Amsterdam, The Netherlands. The program includes:
    - Peroxisomal Disorders
    - Purine and Pyrimidine Disorders
    - Trace elements and metals disorders

- Lysosomal Disorders
- Metabolic neurodegenerative disorders

- **Urine samples**: we remind you that every year, each participant must provide to the scheme organizer at least 200 ml of urine from a patient affected with an established inborn error of metabolism or “normal” urine, together with a short clinical report. If possible, please collect 1500 ml of urine: this sample can be sent to all labs participating to one of the DPT schemes (the ‘common’ sample). Each urine sample must be collected from a single patient (don’t send urine spiked with pathological compounds). Please don’t send a pool of urines, except if urine has been collected on a short period of time from the same patient. Please contact either Joanne Croft (Joanne.Croft4@nhs.net) or Claire Hart (Claire.Hart10@nhs.net) for further information and for a consent form.

Mrs Joanne Croft
Dept of Clinical Chemistry
Sheffield Children’s NHS Foundation
Trust, Western Bank
Sheffield, S10 2TH
United Kingdom
Tel: +44(0)114 271 7000 Ext 17267
Fax: +44(0)114 276 6205
Email: joanne.croft4@nhs.net

Please send us an e-mail on the day you send the samples.

12. Reminders

We remind you that to participate to the DPT-scheme, you must perform at least:
- Amino acids
- Organic acids
- Oligosaccharides
- Mucopolysaccharides
- Purines and pyrimidines

If you do not perform one of these assays, you can send the samples to another lab (cluster lab) but you are responsible for the results.
Please send quantitative data for amino acids and, as much as possible, for organic acids.

13. Tentative schedule in 2024

Sample distribution	7th February 2024
Start of analysis of Survey 2024/1 Website open	12th March 2024
Survey 2024/1 - Results submission	2nd April 2024
Survey 2024/1 - Reports	June 2024
Start of analysis of Survey 2024/2	3rd June 2024
Survey 2024/2 – Results submission	24th June 2024
Survey 2024/2 - Reports	August 2024
Annual meeting of participants	3rd September 2024
Annual Report 2024	December 2024

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

15. Questions, Comments and Suggestions

If you have any questions, comments or suggestions please address to the Scientific Advisor of the scheme, Mrs Joanne Croft (joanne.croft4@nhs.net) and/or to the ERNDIM Administration Office (admin@erndim.org)

Date of report, 2024-02-02

Mrs Joanne Croft
Dept of Clinical Chemistry
Sheffield Children's NHS Foundation
Trust, Western Bank
Sheffield, S10 2TH
United Kingdom
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Fax: +44(0)114 276 6205
Email: joanne.croft4@nhs.net

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

Version Number	Published	Amendments
1	02 February 2024	2023 annual report published

END