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Diagnostic Proficiency Testing

Centre: United Kingdom

Final Report 2024

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 ERNDIM Privacy Policy on www.erndim.org.

1. Geographical distribution of participants

In 2024, 20 labs participated in the UK Diagnostic Proficiency Testing Scheme. All 20 participants submitted results for both submission rounds.

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

2. Design and logistics of the scheme including sample information

[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.

The scheme has been designed and planned by Joanne Croft as Scientific Advisor and Claire Hart as Deputy Scientific Advisor and coordinated by Alessandro Salemma as scheme organiser (sub-contractor on behalf of CSCQ), all 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: Excluding the common sample (sent this year by the Swiss DPT scheme organiser), 1 sample sent this year was a historical sample held in the freezer at Sheffield Children's NHS Foundation Trust, 3 samples had been kindly donated by participating laboratories in the scheme and one was donated by a colleague in the 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 5 samples sent by the UK DPT scheme organiser the typical metabolic profiles were preserved after this process. Mailing: bulk samples were sent to CSCQ in Switzerland at room temperature using TNT/Fedex. 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 2024.

3. Tests

Analyses of amino acids, organic acids, mucopolysaccharides, oligosaccharides and purines/pyrimidines were required in 2024.

4. Schedule of the scheme

- 7th February, 2024: shipment of samples of Survey 1 and Survey 2
- 12th March, 2024: analysis start and website submission available for Survey 1
- 2nd April, 2024: deadline for result submission (Survey 1)
- 3rd June, 2024: analysis start and website submission available for Survey 2
- 24th June, 2024: deadline for result submission (Survey 2)
- May, 2024: interim report of Survey 1 by e-mail
- August, 2024: interim report of Survey 2 by e-mail
- 3rd Sept, 2024: DPT UK participants meeting, Porto
- 28th and 29th Nov, 2024: SAB meeting, Leiden
- Jan 2025: 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

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 2024 have also been scored by Dr George Ruijter, the scientific advisor for the DPT Netherlands scheme. At the SAB meeting in November 2024, 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 2024 2 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. Three performance support letters will be sent by the Scheme Advisor for 2024.

7.1. Score for satisfactory performance

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

8. Results of samples and evaluation of reporting

8.1. Creatinine measurement for all samples

A summary of values provided by participating laboratories for creatinine concentration are shown in the table below.

Creatinine values for each sample in the UK DPT scheme (mmol/L)

Sample	Mean	Median	SD	Min.	Max.
A	8.07	8.30	0.85	5.2	9.1
B	2.19	2.28	0.24	1.22	2.4
C	7.55	7.60	0.41	6.2	8.0
D	1.59	1.60	0.09	1.40	1.73
E	2.46	2.60	0.4	1.20	2.8
F	5.87	5.90	0.31	5.20	6.34

8.2. Patient A

Cytosolic malonyl-CoA decarboxylase deficiency (Malonic aciduria).

Patient details provided to participants
Diagnosed by family screening after sudden infant death of brother at 5 months of age in the context of an intercurrent viral infection. Dilated cardiomyopathy, normal development.

Patient details
Diagnosed aged 3 years. Sample collected aged 15 years. This was the common sample sent to all Diagnostic Proficiency Scheme participants.

Marking scheme
(used by all the DPT scheme organisers)

- Analytical
  - Detecting elevated malonic acid on organic acid analysis or C3DC-carnitine on acylcarnitine analysis: 2 marks (acylcarnitine analysis is not standard but was scored for this sample as it is very specific in this case)
- Interpretation
  - Malonyl-CoA decarboxylase deficiency/malonic aciduria as main diagnosis: 2 marks
  - Malonyl-CoA decarboxylase deficiency/malonic aciduria as alternative diagnosis: 1 mark
  - Combined malonic acidaemia methylmalonic acidaemia (CMAMMA) as main diagnosis: 1 mark
  - Therefore in the case of CMAMMA as main diagnosis and malonic acidaemia as alternative diagnosis, 2 marks would be awarded

Analytical performance

- 15/20 participants scored 2 marks
- 5/20 participants scored 0 marks
  - did not detect the malonic acid or C3DC carnitine

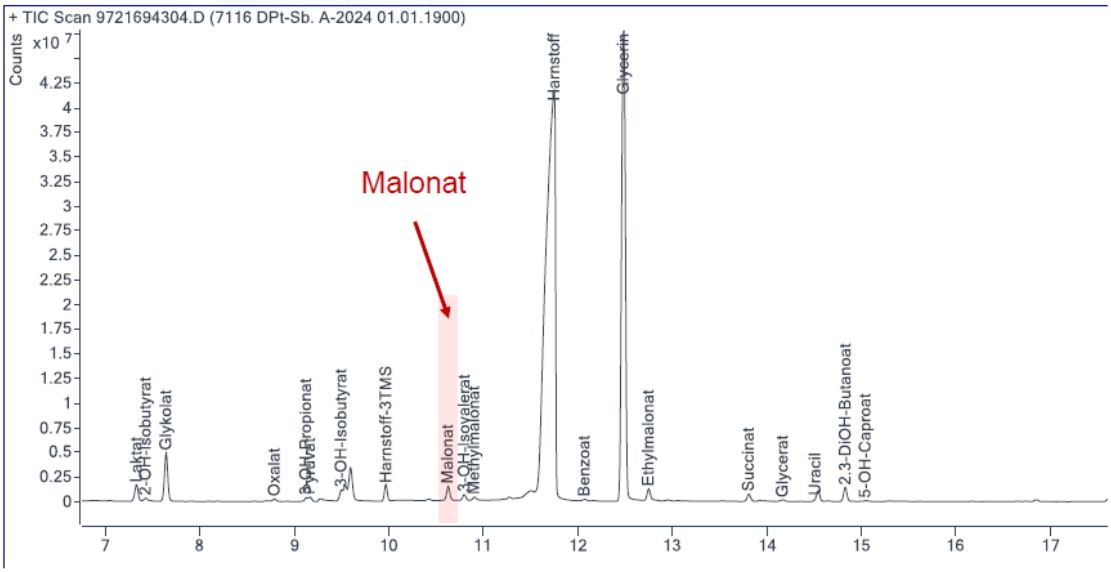


Image of organic acid chromatogram for Sample A. Kindly provided by Deborah Mathis.

Diagnosis / Interpretative proficiency

- 15/20 participants scored 2 marks
- 5/20 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

Recommendations

Recommendations are those provided by the 15 laboratories who gave the correct diagnosis:

- Repeat organic acids – 7/15
- Acylcarnitines - 14/15
- Mutation analysis of MLYCD gene - 12/15
- Refer to clinical metabolic team – 13/15

Overall impression

Performance for this sample was disappointing with 5 laboratories not identifying the malonic acid on organic acid analysis. As this was the common sample it was possible to compare performance with all the other DPT scheme participants. Performance for the UK scheme was lower than for the other schemes. Overall proficiency across all 5 schemes was 90% (compare to 75% for the UK scheme). Due to the relatively large number of participants who did not detect the malonic acid in this sample in the UK scheme it was decided at the SAB meeting that this sample was not eligible for critical error.

8.3. Patient B

MPS Type IV (Morquio)

Patient details provided to participants
Short stature

Patient details
This sample was donated by a patient with mucopolysaccharidosis type 4 who is an immigrant to the UK and who was already diagnosed. Sample collected at 12 years of age.

Marking Scheme

- Analytical
  - Identifying increased keratan sulphate: 2 marks
  - Increased glycosaminoglycan (GAG) concentration with recommendation to do electrophoresis/fractionation: 1 mark

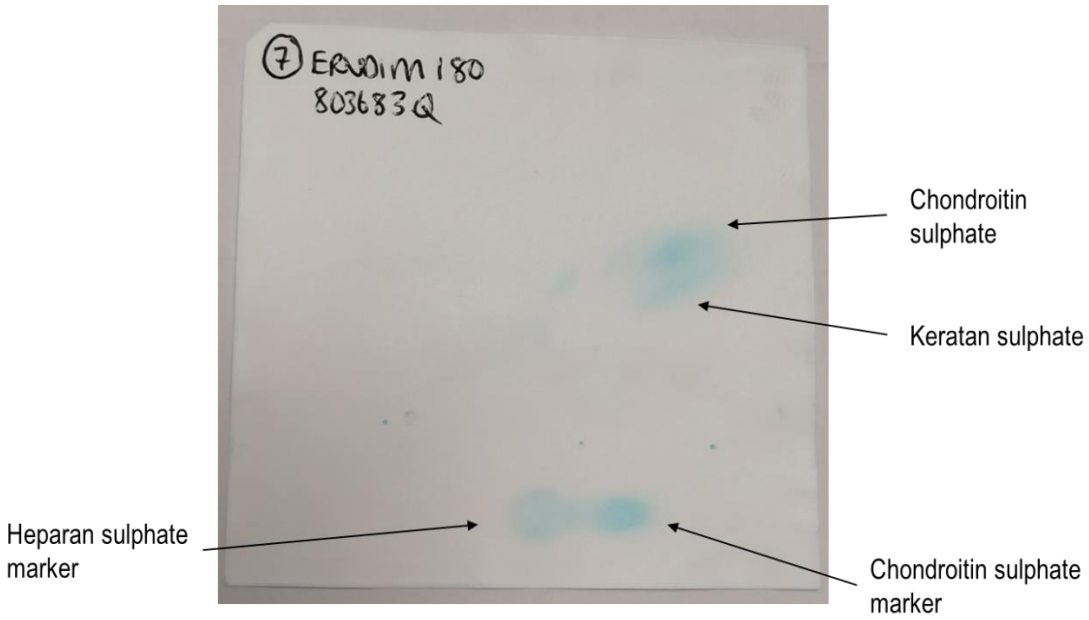
- Interpretation
  - MPS 4: 2 marks
  - Any MPS disorder or MPS4 as alternate: 1 mark

Analytical performance

Analytical performance was good for this sample with 18/20 participants scoring 2 marks and only 1 laboratory scoring 0 marks. The laboratory who scored 1 mark for analysis only provided a GAG quantitative result and stated that they would dispatch the sample for electrophoresis but did not give a result for this analysis. The participant who scored 0 marks for analysis performed GAG quantitation and interpreted the result as normal. Interestingly they reported a higher concentration than some other laboratories but interpreted the result differently. They also performed GAG fractionation (by LC-MS/MS) and reported abnormal presence of dermatan and heparan sulphate.

19/20 participants provided a quantitative GAG result (the remaining laboratory did electrophoresis only)
Mean = 20.6 mg/mmol creatinine
Median = 19.8
SD = 7.25
Min, Max = 10.1, 36.0

16 participants interpreted the result as elevated and 3 as normal (who all went on to do fractionation).



2D GAG Electrophoresis of Sample B

Diagnosis / Interpretative proficiency

- 19/20 scored 2 marks (including the lab who did not perform GAG fractionation)
- 1/20 scored 1 mark (the laboratory who reported a normal GAG concentration and abnormal presence of dermatan and heparan sulphate. Gave MPS1 or MPSV1 as diagnosis)

Recommendations

- Enzymatic confirmation (galactose 6 sulphatase) – 13/20
- Enzymatic confirmation (B galactosidase) – 10/20
- Enzymatic confirmation (enzyme not specified) – 4/20
    - Only 2 participants did not recommend enzymatic confirmation but both did recommend genetic analysis
- Genetic analysis (GALNS) – 13/20
- Genetic analysis (GLB1) – 9/20
- Discuss with/refer to metabolic clinician – 13/20
- Sibling/family investigation – 6/20

Overall impression

Proficiency for this sample was good (95% overall).

8.4. Patient C

Adenine phosphoribosyltransferase (APRT) deficiency

Patient details provided to participants
History of renal stones and urinary tract infections

Patient details

This sample was donated to us by another laboratory. Sample collected at 7 years of age.

Marking Scheme

- Analytical
    - Detection of 2,8-dihydroxyadenine: 2 marks
- Interpretation
    - Adenine phosphoribosyltransferase (APRT) deficiency: 2 marks
    - Other purine disorder: 1 mark
    - Recommendation to do purine analysis (if not done): 1 mark

Analytical performance

15/20 participants reported the 2,8-dihydroxyadenine in this sample and scored 2 marks for analysis. The remaining 5 participants scored 0 for analysis as the sample was not sent for purine analysis. It is noted that in the UK there are limited options for purine analysis and that the majority of participants will have had to either send the sample away for analysis or contacted the Purine Laboratory for their results (as previously agreed).

Diagnosis / Interpretative proficiency

All participants who reported the 2,8-dihydroxyadenine gave the correct diagnosis and scored 2 marks for interpretation. The remaining 5 laboratories scored 1 mark as they recommended purine analysis.

Recommendations

- Purine analysis (if not already done) – 5/20
- APRT activity in red blood cells – 9/20
- Mutation analysis of the APRT gene – 12/20
- Refer to appropriate clinical teams (nephrology/metabolic) – 10/20
- Stone analysis - 3/20
- Check renal function – 2/20

Overall impression

All participants who either sent the sample for purine analysis or who requested the result from the Purine laboratory scored 4 marks for this sample. Although purine analysis is not available in many laboratories it is important to remember these conditions as a cause of renal stones.

8.5. Patient D

No inborn error of metabolism.

Patient details provided to participants
Developmental delay. On dairy free diet.

Patient details
This sample came from a child on a dairy free diet whose diet is high in coconut oil which is high in medium chain triglycerides. They were also subsequently found to be Vitamin B12 deficient.

Marking Scheme

- Analytical
  - Performing at least 3 analyses (not including the 'pre-investigations') and finding no significant abnormality: 2 marks
  - There were some dicarboxylic acids and a slightly increased excretion of methylmalonic acid in this sample. Scoring of the organic acid findings have therefore been lenient.

- Interpretive
  - 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

- All participants scored 2 marks for analysis

Diagnosis / Interpretative proficiency

All participants scored 2 marks for diagnosis.
Alternative diagnoses provided included primary or secondary 3 methylglutaconic aciduria and to consider galactosaemia.

Recommendations

As seen in previous years, samples for which no significant abnormality is the diagnosis are often reported in the DPT scheme with a few recommendations for follow up tests.

- Galactosaemia testing (including Beutler test, galactose-1-phophate and galactitol) – 8/20
- Acylcarnitine analysis (in view of the slight DCAs on organic acids) – 5/20
- Plasma amino acids – 6/20
- Plasma methylmalonate – 1/20
- Plasma total homocysteine – 2/20
- No recommendations provided – 3/20
- Ask for further clinical information to guide investigations – 2/20

Overall impression

Proficiency for this sample was excellent.

8.6. Patient E

Isolated 3-methylcrotonyl-CoA carboxylase deficiency (3-Methylcrotonylglycinuria)

Patient details provided to participants
Diagnosed following newborn screening results of baby

Patient details

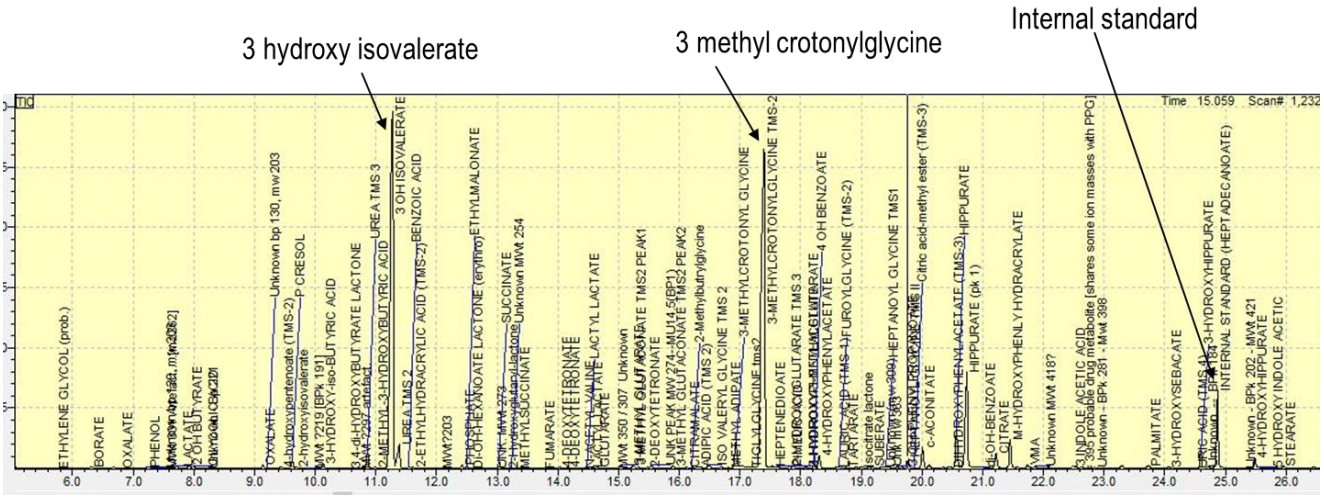
This sample was donated by the mother of a baby who had been found to have a very low free carnitine concentration during newborn screening (during the medium chain acyl CoA dehydrogenase newborn screening trial in the UK). The mother was tested and found to also have a very low free carnitine of 4 umol/L. Urine was analysed for organic acids and the diagnosis made. Mother is asymptomatic.

Marking Scheme

- Analytical
  - Identifying increased 3 hydroxy isovaleric acid and 3-methylcrotonylglycine on organic acids: 2 marks
- Interpretation
  - 3 methylcrotonylglycinuria (3MCC) – 2 marks

Analytical performance

- 20/20 participants scored 2 marks



Diagnosis / Interpretative proficiency

- 20/20 participants scored 2 marks

Alternative diagnoses suggested – 'multiple carboxylase deficiency is on the differential but is considered less likely in view of the lack of elevated lactate, methyl citrate and tiglyglycine'.
'Biotinidase deficiency – highly unlikely from the clinical history and would be atypical organic acid pattern for this'.

Recommendations

- Genetic confirmation (MCCC1 and MCCC2 genes) – 18/20
- Genetic testing (genes not named) – 2/20
- Enzyme analysis - 3/20
- Acylcarnitine analysis (DBS or plasma) – 20/20

- Biotinidase – 6/20
- Recommend discussion with/referral to metabolic team – 16/20
- Testing of family members/genetic counselling – 5/20

Overall impression

Proficiency was excellent for this sample which gave a clear pattern on organic acid analysis.

8.7. Patient F

Gyrate atrophy of retina and choroidea due to ornithine aminotransferase deficiency.

Patient details provided to participants
Retinal changes

Patient details

This sample was donated by a patient with ornithine aminotransferase deficiency.

Marking Scheme

- Analytical
  - Identifying increased ornithine concentration – 2 marks

- Interpretation
  - Ornithine aminotransferase deficiency/Gyrate atrophy of retina and choroidea – 2 marks

Analytical performance

18 participants identified the increased ornithine concentration in this sample. 2 participants scored 0 marks for analysis. 1 did not perform/report amino acid analysis and 1 failed to identify the increased ornithine concentration.

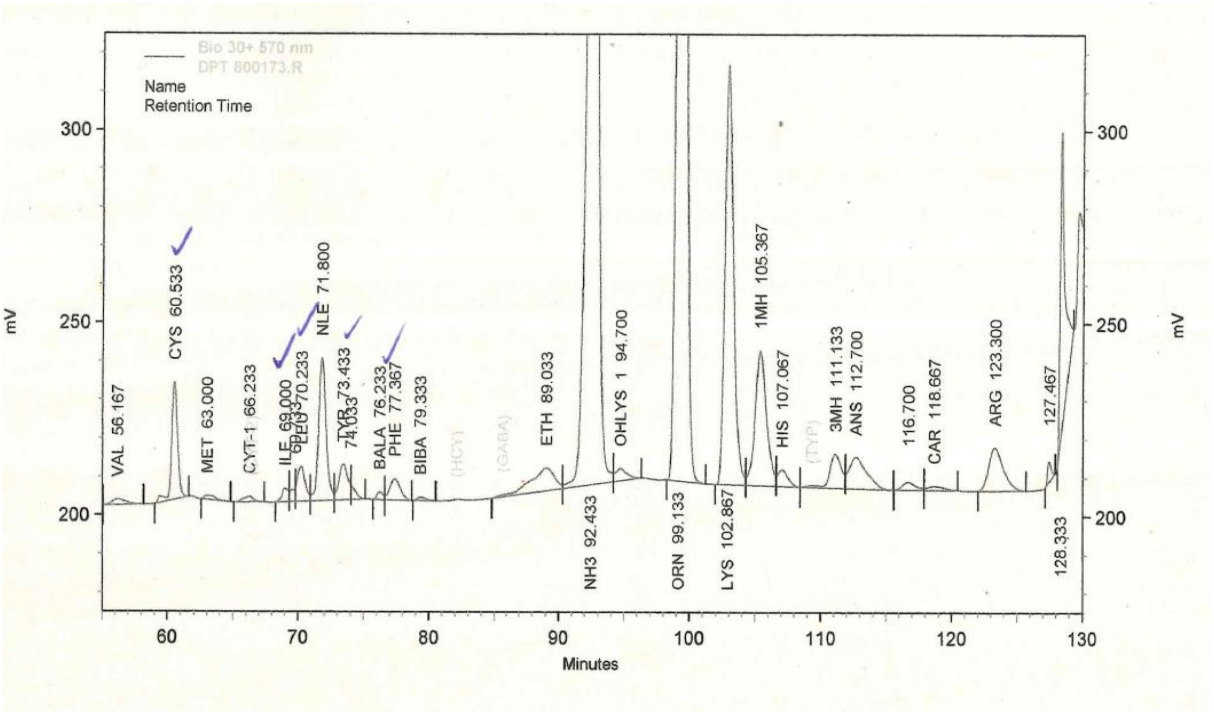
17 participants provided a quantitative value for ornithine (the 3 who did not include the 2 who scored 0 for analysis and 1 further lab who gave a qualitative result only).

Median = 879 umol/mmol creatinine
Mean = 886.9
SD = 354
Min, max = 348 – 2000

Sheffield Children's Hospital result = 1012 umol/mmol creatinine (ref. 0 – 5)

There was also increased concentration of lysine, cystine and arginine in this sample, likely due to competition for the renal tubular dibasic amino acid transporter.

It was confirmed at the SAB meeting in November 2024 that failure to identify the increased ornithine concentration in this sample constitutes a critical error and therefore 2 participants will receive a performance support letter for this sample. The laboratory who did perform amino acid analysis reported a grossly increased concentration of lysine. The method they report using is ion exchange chromatography with ninhydrin detection. As can be seen from the amino acid trace taken from our analysis of this sample using our Biochrom analyser, the peaks of ornithine and lysine are close together and I suspect there has been an issue with the separation of the peaks for this particular laboratory.


Bio 30+ 570 nm
DPT 800173.R
Name
Retention Time
mV
300
250
200
VAL 56.167
CYS 60.533
MET 63.000
CYT-1 66.233
ILE 69.000
LEU 70.233
NLE 71.800
TYR 73.433
BALA 76.233
PHE 77.367
BIBA 79.333
(HCY)
(GABA)
ETH 89.033
NH3 92.433
OHLYS 1 94.700
ORN 99.133
LYS 102.867
1MH 105.367
HIS 107.067
(TYP)
3MH 111.133
ANS 112.700
116.700
CAR 118.667
ARG 123.300
127.467
128.333
60
70
80
90
100
110
120
130
Minutes

Diagnosis / Interpretative proficiency

18/20 participants gave the correct diagnosis and scored 2 marks for interpretation.

Alternative diagnoses provided included lysinuric protein intolerance (2 labs), cystinuria (2 labs), hyperlysinaemia and HHH.

2/20 participants scored 0 marks for interpretation (one concluded to a peroxisomal disorder, the other to MPSIV). These were the 2 labs who also scored 0 for analysis.

Recommendations

- All participants who gave the correct diagnosis gave helpful recommendations
-
    - Plasma amino acids – 15/18
    - Genetic analysis (OAT gene) – 16/18
    - Referral to metabolic consultant/ team – 15/18
    - Plasma ammonia – 5/18
    - Investigate siblings and children – 6/18

Overall impression

Proficiency for this sample was good (90%) though it is disappointing that 1 laboratory did not report amino acids for this sample.

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 Malonic aciduria			Patient B MPS IV			Patient C APRT deficiency			
	A	I	Total	A	I	Total	A	I	Total	Total
1	0	0	0	2	2	4	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	0	1	1	9
5	2	2	4	0	1	1	2	2	4	9
6	2	2	4	2	2	4	2	2	4	12
7	2	2	4	2	2	4	2	2	4	12
8	2	2	4	2	2	4	0	1	1	9
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	0	0	0	2	2	4	2	2	4	8
13	2	2	4	2	2	4	2	2	4	12
14	0	0	0	1	2	3	0	1	1	4
15	0	0	0	2	2	4	0	1	1	5
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	0	1	1	9
20	2	2	4	2	2	4	2	2	4	12

Detailed scores – Round 2

Lab n°	Patient D No inborn error of metabolism.			Patient E 3-Methylcrotonyl-CoA carboxylase deficiency			Patient F Ornithine aminotransferase deficiency			
	A	I	Total	A	I	Total	A	I	Total	Total
1	2	2	4	2	2	4	2	2	4	12
2	2	2	4	2	2	4	2	2	4	12
3	2	2	4	2	2	4	2	2	4	12
4	2	2	4	2	2	4	2	2	4	12
5	2	2	4	2	2	4	2	2	4	12
6	2	2	4	2	2	4	2	2	4	12
7	2	2	4	2	2	4	2	2	4	12
8	2	2	4	2	2	4	2	2	4	12
9	2	2	4	2	2	4	2	2	4	12
10	2	2	4	2	2	4	2	2	4	12
11	2	2	4	2	2	4	2	2	4	12
12	2	2	4	2	2	4	0	0	0	8
13	2	2	4	2	2	4	2	2	4	12
14	2	2	4	2	2	4	2	2	4	12
15	2	2	4	2	2	4	2	2	4	12
16	2	2	4	2	2	4	0	0	0	8
17	2	2	4	2	2	4	2	2	4	12
18	2	2	4	2	2	4	2	2	4	12
19	2	2	4	2	2	4	2	2	4	12
20	2	2	4	2	2	4	2	2	4	12

Total scores

Lab n°	A	B	C	D	E	F	Cumulative score	Cumulative score (%)	Critical error
1	0	4	4	4	4	4	20	83	
2	4	4	4	4	4	4	24	100	
3	4	4	4	4	4	4	24	100	
4	4	4	1	4	4	4	21	88	
5	4	1	4	4	4	4	21	88	
6	4	4	4	4	4	4	24	100	
7	4	4	4	4	4	4	24	100	
8	4	4	1	4	4	4	21	88	
9	4	4	4	4	4	4	24	100	
10	4	4	4	4	4	4	24	100	
11	4	4	4	4	4	4	24	100	
12	0	4	4	4	4	0	16	67	CE
13	4	4	4	4	4	4	24	100	
14	0	3	1	4	4	4	16	67	
15	0	4	1	4	4	4	17	71	
16	4	4	4	4	4	0	20	83	CE
17	4	4	4	4	4	4	24	100	
18	0	4	4	4	4	4	20	83	
19	4	4	1	4	4	4	21	88	
20	4	4	4	4	4	4	24	100	

Performance

	Number of labs	% total labs
Satisfactory performers (≥ 70 % of adequate responses)	17	85
Unsatisfactory performers (< 70 % adequate responses and/or critical error)	3	15

Overall Proficiency

Sample	Diagnosis	Analytical (%)	Interpretation (%)	Total (%)
DPT-UK-2024-A	Malonic aciduria	75	75	75
DPT-UK-2024-B	MPS IV	93	98	95
DPT-UK-2024-C	APRT deficiency	75	88	81
DPT-UK-2024-D	No inborn error of metabolism.	100	100	100
DPT-UK-2024-E	3-Methylcrotonyl-CoA carboxylase deficiency	100	100	100
DPT-UK-2024-F	Ornithine aminotransferase deficiency	90	90	90

10. Annual meeting of participants

This took place in Porto on Tuesday 3rd September 2024, before the SSIEM Meeting. Workshops for the different DPT schemes are held prior to the ERNDIM meeting of participants.

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.

If you would rather that the UK DPT participant meeting is held on-line in the future please contact the ERNDIM administration office. It may be that an on-line meeting instead of or as well as the in-person meeting may be more productive as more participating laboratories and multiple staff members from participating laboratories will be able to contribute.

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

- **Training**: SSIEM Academy training courses.
  - A 2 day course has been organized for 28th/29th April 2025 in Prague, Czech Republic.
  - The program includes:
    - Mitochondrial disorders
    - Neurotransmitter disorders
    - Glycogen storage disorders
    - Congenital Disorders of glycosylation
- **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.

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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. Schedule in 2025

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

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, 2025-01-10

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

Version Number	Published	Amendments
1	21 January 2025	2024 annual report published

END