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Diagnostic Proficiency Testing

Centre: United Kingdom

Final Report 2025

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.

1. Geographical distribution of participants

The ERNDIM Diagnostic Proficiency Testing (DPT) Scheme is the ultimate external quality assessment scheme for biochemical genetics laboratories. In 2025, 21 labs participated to the Proficiency Testing Scheme UK

For the first survey, 20 and second survey 20 laboratories submitted results with 1 non-submitter for both surveys.

Country	Number of participants
Australia	1
Ireland	1
Netherlands	1
New Zealand	1
Spain	1
United Kingdom	16

[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 and Urine MPS 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 patients: all urine samples, excluding the common sample, have been provided by patients seen locally in Sheffield, UK. The common sample was sent by the Scientific Advisor for the France DPT scheme in 2025.

Patient A: MPS VI
Patient B: 3-Methylglutaconyl-CoA hydratase deficiency
Patient C: ASA
Patient D: Glutaric aciduria type 3 (GA3)
Patient E: Alpha mannosidosis
Patient F: Isovaleric aciduria (IVA)

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. The samples are stable for the duration of the scheme's submission calendar when stored under defined conditions.
Mailing: samples were sent by DHL; FedEx or the Swiss Post at room temperature.

3. Tests

The minimal required test panel for participation in any DPT scheme includes creatinine, dip stick, amino acids, organic acids, oligosaccharides, quantitative GAG screening and purines-pyrimidines. It is strongly recommended to have the following tests available for DPT-NL: GAG subtype analysis (by electrophoresis, TLC or LC-MS/MS), sialic acid, creatine-guanidinoacetate and polyols-sugars. Please note that in DPT schemes it is allowed to obtain results from partner laboratories when this is routine clinical practice. It is required to indicate in the report that results were obtained from a cluster lab.

4. Schedule of the scheme

- February 10, 2025: shipment of samples
- March 17, 2025: start analysis of samples of the first survey
- April 7, 2025: deadline for result submission (Survey 1)
- May 16, 2025: interim report with preliminary scores of Survey 1 published
- June 2, 2025: start analysis of samples of the second survey
- June 23, 2025: deadline for result submission (Survey 2)
- August 9, 2025: interim report with preliminary scores of Survey 2 published
- October 9, 2025: DPT meeting, ERNDIM symposium Madrid, Spain
- March, 2026: annual report with final scoring published

5. Results

20 of 21 labs returned results for both surveys. Only 1 laboratory asked for an extension to the deadline which was granted. 1 laboratory returned no results.

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

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. The scores were calculated only for laboratories submitting results.

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 2025 have been also scored by the scientific advisor for the Swiss DPT scheme. At the SAB meeting on 27th/28th November 2025, the definitive scores were 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 2025, no participants in the UK DPT scheme received a critical error.

ERNDIM provides a single certificate for all its schemes with details of participation and performance. In addition, performance support letters will be issued if the performance is evaluated as unsatisfactory. In 2025, no performance support letters will be sent by the Scheme Advisor for the UK DPT scheme.

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)

7.1. Score for satisfactory performance

At least 17 points from the maximum of 24. However, as one of the samples has this year been deemed to be educational, the scores for this sample will not be taken into consideration. Therefore the minimum score for 2025 for the UK DPT scheme is 14 out of 20.

8. Results of samples and evaluation of reporting

As 1 laboratory returned no results, I will discuss results in terms of 'out of 20'.

8.1. Patient A

Mucopolysaccharidosis Type VI (Maroteaux-Lamy disease)

Patient details provided to participants

15-year-old boy. Dysmorphic features, scoliosis, size -1.5 SD, normal intellectual development. Under treatment.

Patient details

This is the common sample distributed to all DPT centers. A separate presentation is available on ERNDIM website. The patient is a 15-year-old boy, born after a normal pregnancy and delivery. From 2 months to 6 years of age, he had frequent upper airways infections. His height at 1 year of age was +3 SD. Pectus carinatum was noted at 3 years of age. At 10 years of age, he presented with scoliosis with platyspondyly, dorsal kyphosis, a narrow cervical canal and decreased visual and hearing acuity. At 15 years of age, his intellectual development is normal, and his height is -1.5 SD. He underwent back surgery with arthrodesis of T10 to T12. At that time, urinary MPS analysis was requested because of spine abnormalities and dysmorphic features. Diagnosis of MPS VI was confirmed by measuring arylsulfatase B activity in leucocytes. Since then, he has received weekly enzyme replacement therapy.

Analytical performance

- 17/20 participants scored 2 marks
    - Identified presence of dermatan sulphate by GAG fractionation
- 3/20 participants scored 1 mark
    - Identified increased GAG concentration but did not do GAG fractionation

Diagnosis / Interpretative proficiency

- 17/20 participants scored 2 marks
- 3/20 participants scored 1 mark
- All the participants who scored 2 marks for analysis interpreted the result correctly.
- All the participants who scored 1 mark for analysis also scored 1 mark for interpretation

Recommendations

All participants gave helpful recommendations

- Enzyme assay to confirm the diagnosis – 18/20
- Genetic analysis – 16/20

MPS VI or Maroteaux-Lamy disease is due to N-acetylgalactosamine-4-sulfatase deficiency. Coded by the *ARSB* gene.

Scoring

(Marking scheme used by all the DPT scheme organisers)

- **Analytical**
- Increase of dermatan sulphate (score 2)
- Increase of glycosaminoglycans without GAGs fractionation (score 1)
- **Interpretation**
- Mucopolysaccharidosis type VI (score 2)
- Unspecified or wrong mucopolysaccharidosis, or diagnosis according to the clinical presentation (score 1)

Overall impression

Proficiency for this sample was very good with all participants scoring either 2 or 4 marks dependent on whether the laboratory performed GAG fractionation or not.

8.2. Patient B

3-Methylglutaconyl-CoA hydratase deficiency

Patient details provided to participants

Epilepsy since teenager. Adult onset leukoencephalopathy

Analytical performance

- 18/20 participants scored 2 marks for analysis
    - Noted the increased 3 methylglutaconic acid and the increased 3 hydroxy isovaleric acid
- 2/20 participants scored 1 mark for analysis
    - Did not report the increased excretion of 3 hydroxy isovaleric acid

Diagnosis / Interpretative proficiency

20/20 participants scored 2 marks

- All participants correctly identified this sample as having come from a patient with 3 methylglutaconyl-CoA hydratase deficiency (aka 3 methylglutaconic aciduria type 1)
- Many participants wrote other 3 methylglutaconic acidurias as their alternate diagnosis with some saying this was unlikely given the level of excretion of 3 hydroxy isovaleric acid.
- Due to the increased excretion of ethylmalonic acid in this sample, a couple of participants also mentioned multiple acyl-CoA dehydrogenase deficiency, ethylmalonic acidaemia or SCADD as other alternate diagnoses but with the caveat that these were much less likely on clinical grounds.

Recommendations

- Molecular analysis of the AUH gene - 16/20
- Referral to adult metabolic consultant/team - 13/20
- Repeat organic acid analysis - 7/20
- 3 methyl glutaconyl enzyme assay (in leucocytes or fibroblasts) - 4/20
- Acylcarnitines (some mentioned to help determine significance of the increased EMA) - 9/20

Scoring

- Analytical
- Increased 3 methyl glutaconic acid – 1 mark
- Increased 3 OH IVA – 1 mark
- (there is also raised EMA in this sample – cause unknown in this case)

- Interpretation
- 3 methyl glutaconyl hydratase deficiency – 2 marks
- 3 methylglutaconic aciduria – 1 mark

Overall impression

Overall proficiency was very good.

8.3. Patient C

Argininosuccinic aciduria, argininosuccinate lyase (ASL) deficiency

Patient details provided to participants

Presented at 1 year of age with developmental delay. Liver pathology a later feature.

Patient details

The sample was obtained from a 15 years old girl with Argininosuccinic aciduria due to arginine succinate lyase deficiency.

Analytical performance

- 20/20 participants scored 2 marks
- All detected increased argininosuccinic acid and its associated anhydrides
- Mean ASA concentration = 842.4 umol/mmol (7 labs provided a quantitative result)
- 9/20 commented on increased arginine – felt to be due to treatment
- Mean arginine concentration = 56.9 umol/mmol (SCH ref range 0 - 11)

Diagnosis / Interpretative proficiency

- 20/20 scored 2 marks
- All participants concluded that this sample was from a patient with Argininosuccinic aciduria

Recommendations

- Urgent blood ammonia - 20/20
- Urgent plasma amino acids - 18/20
- Mutation analysis of ASL gene to confirm diagnosis - 18/20
- Genetic testing (no gene named) – 2/20
- Testing of siblings/family members – 7/20
- Liver function tests – 7/20
- Urine orotic acid – 3/20 (10 participants commented on the fact that excretion of orotic acid was normal/not detected)

Scoring

- Analytical

Detection of ASA (argininosuccinic acid) – 2 marks

- Interpretation

ASA (argininosuccinic aciduria) – 2 marks

Overall impression

Proficiency was 100% for this sample.

Multiple distributions of similar samples

Compare this to the ASA sample that was sent as the common sample in 2023 when 6/20 participants scored 0 marks (though concentration of ASA in that sample was much less – mean value 46 umol/mmol.

8.4. Patient D

Glutaric aciduria type 3 (GA3) due to succinate-hydroxymethylglutarate-CoA transferase deficiency, encoded by C7orf10.

Patient details provided to participants

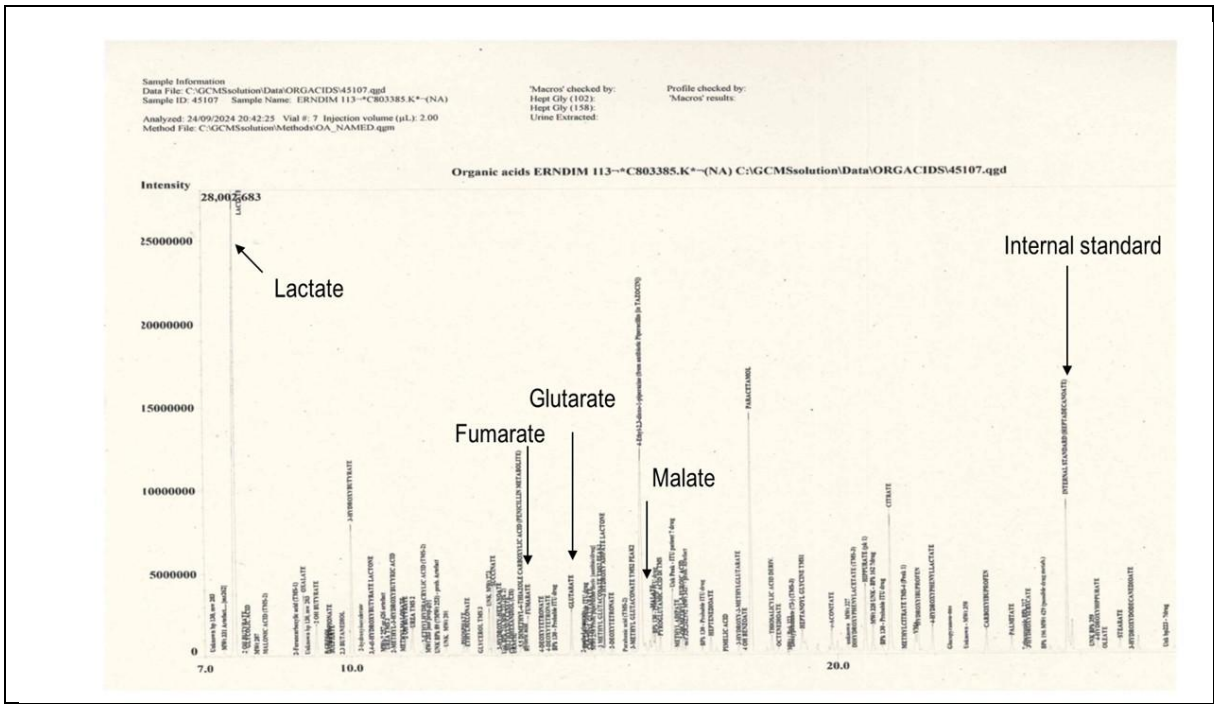
Failure to thrive in infancy. Presented acutely unwell later in life with hypoglycaemia, acidosis and requiring ventilatory support.

Patient details

- This sample was obtained from a patient with biochemically and genetically confirmed Glutaric aciduria type 3 (GA3)
- Succinate-hydroxymethylglutarate-CoA transferase deficiency, encoded by C7orf10 (converts glutarate to glutaryl-CoA)
- The patient presented acutely unwell at the age of 29 years, following a brief vomiting illness, and was admitted to the ICU requiring ventilation.
- Urine organic acids showed increased excretion of glutarate, lactate, fumarate and malate without increased acylglycines suggestive of a possible mitochondrial disorder. High dose riboflavin was administered which was followed by a rapid clinical recovery.
- It was subsequently found that the patient had been the first described case of GA3 (Bennett et al, JIMD (1991) 14:165-173) but had been lost to follow up.
- This case was presented as a poster at the SSIEM meeting in September 2014 and has also been used in the Sheffield QLOU scheme in 2014 (sample 221).

Analytical performance

- 14/20 participants commented on the increased glutarate in this sample.
- Most participants also commented on the increased excretion of lactate, malate, fumarate and 3 hydroxy butyrate.
- This was a dilute urine sample
- (mean creatinine conc. of participants results = 0.77mmol/L)



Diagnosis / Interpretative proficiency

– No participants gave glutaric aciduria type 3 as either the diagnosis or the alternate diagnosis.
– Most laboratories mentioned that the pattern of results on organic acid analysis (increased lactate,

malate and fumarate) could be suggestive of a mitochondrial disorder.
– Made scoring difficult.

Recommendations

- Plasma lactate – 19/20
- Acylcarnitines – 13/20
- Plasma amino acids – 15/20
- Mitochondrial genetics panel – 8/20
- Many other recommendations provided due to the non-specific findings

Scoring

Analytical

- Increased glutarate on OAs – 2 marks
- Increased lactate and TCA metabolites, with no mention of increased glutarate – 1 mark

Interpretive

- Increased glutaric acid with appropriate further tests (acylcarnitines, fatty acid oxidation flux) – 2 marks
- MADD – 1 mark
- Mitochondrial disorder – 1 mark

Overall impression

This was a difficult sample. The disorder is rare (and often thought of as benign) but one which metabolic laboratories need to be aware of. Biochemically it is difficult to distinguish between glutaric aciduria type 3, generalised mitochondrial dysfunction or riboflavin responsive glutaric aciduria type 2 on riboflavin. One of the key differences is the lack of acylglycines in GA3. Participants that failed to follow up on the increased glutarate appropriately scored lower.

At the Scientific Advisory Board meeting held November 2025 it was decided that this sample should be educational. Therefore the scores for this sample will not contribute to the final score in 2025 and the minimum score required has been lowered appropriately.

8.5. Patient E

Alpha mannosidosis due to alpha mannosidase deficiency

Patient details provided to participants

Skeletal dysplasia

Analytical performance

- 14/20 laboratories reported an oligosaccharide result
- 13/14 laboratories identified an abnormal pattern
- 1/14 reported a normal pattern
- 6/20 did not report an oligosaccharide result

1 laboratory reported that they had sent the sample away for oligosaccharide analysis but did not receive the result before the deadline. I am not aware that this laboratory asked for an extension

Diagnosis / Interpretative proficiency

- Of those 14 laboratories who performed oligosaccharide analysis, 13 gave the correct diagnosis and scored full marks for this sample
- 3 laboratories scored 1 mark for recommending oligosaccharide analysis
- 1 laboratory performed oligosaccharide analysis and reported a normal profile – scored 0 for this sample
- 3 laboratories did not perform nor suggest oligosaccharide analysis therefore scored 0 marks for this sample

Recommendations

- Genetic confirmation (MAN2B1 gene) - 9/20
- Genetic confirmation (no gene named) - 3/20
- Enzyme assay - 13/20 (including one lab who did not reach the correct diagnosis but felt, on clinical info provided, alpha mannosidosis needed investigating)
- Repeat urine for oligos – 3/20
- Oligos if not already done – 3/20 (different 3 to those above)
- Refer to metabolic consultant/team - 8/20

Not all labs who scored 4 marks for this sample suggested both genetic and enzyme analysis but did suggest at least one of these follow up tests.

Scoring

- Analytical

Abnormal oligosaccharide pattern - 2 marks

- Interpretation

Alpha mannosidosis – 2 marks

Suggesting oligosaccharide analysis if not done (or results not yet back) - 1 mark

Overall impression

Overall proficiency for this sample was just under 70%. Some laboratories did not perform oligosaccharide analysis.

8.6. Patient F

Isovaleric aciduria (IVA)due to isovaleryl-CoA dehydrogenase deficiency.

Patient details provided to participants

Poor feeding and drowsiness.

The patient was diagnosed at the age of 2 weeks. The sample was collected at the age of 32 years.

Analytical performance

- All participants identified the markedly increased excretion of isovalerylglycine, with many commenting on the absence of increased excretion of 3 hydroxy isovaleric acid, likely to be due to this being a non-crisis sample taken from a patient under treatment.
- All participants scored 2 marks for analysis

Diagnosis / Interpretative proficiency

- All participants correctly gave isovaleric aciduria/isovaleric acidaemia as the most likely diagnosis with no suggestions for alternative diagnoses provided given the clear nature of the analytical findings.
- All participants scored 2 marks for interpretation.

Recommendations

- Acylcarnitines (DBS or plasma) - 19/20
- Urgent ammonia (given clinical details of drowsiness) - 10/20
- Mutation analysis (IVD gene) - 18/20
- Mutation analysis (gene not named) - 2/20
- Referral to metabolic consultant/team - 15/20
- Testing of siblings/family members - 5/20
- Repeat sample for repeat organic acid analysis - 5/20
- Blood glucose (given clinical details of poor feeding) - 2/20

Scoring

- Analytical

Increased isovalerylglycine on OAs – 2 marks

- Interpretation

Isovaleric acidaemia – 2 marks

Overall impression

100% proficiency 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 MPS VI			Patient B 3-Methylglutaconyl-CoA hydratase deficiency			Patient C ASA			
	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	1	1	2	2	2	4	2	2	4	10
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	2	2	4	12
13	2	2	4	2	2	4	2	2	4	12
14	1	1	2	2	2	4	2	2	4	10
15	2	2	4	2	2	4	2	2	4	12
16	2	2	4	2	2	4	2	2	4	12
17	2	2	4	1	2	3	2	2	4	11
18	2	2	4	2	2	4	2	2	4	12
19	1	1	2	2	2	4	2	2	4	10
20	2	2	4	1	2	3	2	2	4	11
21	--	--	--	--	--	--	--	--	--	0

Detailed scores – Round 2

Lab n°	Patient D Glutaric aciduria type 3 (GA3)			Patient E Alpha mannosidosis			Patient F Isovaleric aciduria (IVA)			
	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	--	--	--	0	1	1	2	2	4	5
5	--	--	--	2	2	4	2	2	4	8
6	--	--	--	2	2	4	2	2	4	8
7	--	--	--	0	1	1	2	2	4	5
8	--	--	--	0	0	0	2	2	4	4
9	--	--	--	2	2	4	2	2	4	8
10	--	--	--	2	2	4	2	2	4	8
11	--	--	--	0	0	0	2	2	4	4
12	--	--	--	2	2	4	2	2	4	8
13	--	--	--	2	2	4	2	2	4	8
14	--	--	--	0	0	0	2	2	4	4
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	--	--	--	0	1	1	2	2	4	5
20	--	--	--	0	0	0	2	2	4	4
21	--	--	--	--	--	--	--	--	--	0

Total scores

Lab n°	A	B	C	D	E	F	Cumulative score	Cumulative score (%)	Critical error
1	4	4	4	--	4	4	20	100	
2	4	4	4	--	4	4	20	100	
3	4	4	4	--	4	4	20	100	
4	4	4	4	--	1	4	17	85	
5	4	4	4	--	4	4	20	100	
6	2	4	4	--	4	4	18	90	
7	4	4	4	--	1	4	17	85	
8	4	4	4	--	0	4	16	80	
9	4	4	4	--	4	4	20	100	
10	4	4	4	--	4	4	20	100	
11	4	4	4	--	0	4	16	80	
12	4	4	4	--	4	4	20	100	
13	4	4	4	--	4	4	20	100	
14	2	4	4	--	0	4	14	70	
15	4	4	4	--	4	4	20	100	
16	4	4	4	--	4	4	20	100	
17	4	3	4	--	4	4	19	95	
18	4	4	4	--	4	4	20	100	
19	2	4	4	--	1	4	15	75	
20	4	3	4	--	0	4	15	75	
21	--	--	--	--	--	--	0	0	

Performance

	Number of labs	% total labs
Satisfactory performers (≥ 70 % of adequate responses)	20	95
Unsatisfactory performers (< 70 % adequate responses and/or critical error)	0	0
Partial and non-submitters	1	5

Overall Proficiency

Sample	Diagnosis	Analytical (%)	Interpretation (%)	Total (%)
DPT-UK-2025-A	MPS VI	93	93	93
DPT-UK-2025-B	3-Methylglutaconyl-CoA hydratase deficiency	95	100	98
DPT-UK-2025-C	ASA	100	100	100
DPT-UK-2025-D	Glutaric aciduria type 3 (GA3)	--	--	--
DPT-UK-2025-E	Alpha mannosidosis	65	73	69
DPT-UK-2025-F	Isovaleric aciduria (IVA)	100	100	100

10. Annual meeting of participants

This took place in Madrid on 9th and 10th October 2025. This year was an ERNDIM workshop as opposed to a participant meeting before the SSIEM due to the ICIEM being held in 2025.

We remind you that attending the annual meeting 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

- Following 2 years as a pilot scheme, **'Lipids In Serum' (LIS)** will be organised as a full scheme starting in 2026 in collaboration with MCA laboratory. The scientific advisors of this scheme are dr Susan Goorden (Rotterdam, NL) and dr Marie van Dijk (Amsterdam, NL). LIS is a quantitative scheme in which several lipids relevant to IMD diagnostics are included. Some of the lipids included in LIS are new, while others have been in the Special Assays Serum scheme for some years already. Some lipids will be removed from SAS in 2026 (see details in the ERNDIM scheme catalogue.
- **Control materials** are provided by SKML/MCA laboratory since a few years. These are no longer related to EQA materials and have been produced separately. Two concentration levels for each group of analytes are available. The most suitable low and high concentration levels are defined by the scientific advisors of the schemes. Analytes and their concentrations will be similar in consecutive batches of control material. These reference materials can be ordered at MCA laboratory (https://www.erndimqa.nl/). Participants are encouraged to use them as internal control samples, but they cannot be used as calibrators. On the ERNDIMQA website a new section for data

management completes the ERNDIM internal Quality Control System. Laboratories have the option to submit results and request reports showing their result in the last run in comparison to defined acceptance limits, their own historical data and the mean of all laboratories using the same batch control material. Control materials for cystine in leukocytes are being tested, while aminoa cids in urine and CSF are under development. Control materials for neurotransmitters in CSF have been discontinued due to stability issues.

- **Training**:

  After successful webinars on amino acids, acylcarnitines, organic acids and purines-pyrimidines in 2024 and 2025 ERNDIM will organise two additional workshops on special assays in 2026. These workshops will focus on technical aspects of measuring metabolites. Dates of these workshops will be announced by email and on the ERNDIM website and registration will be required.

  An SSIEM Academy training course will be organised in 2026. Detail will be available on the SSIEM website

- **Urine samples**: To be able to continue this scheme we need a steady supply of new and interesting patient samples. Several laboratories have donated samples 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

  When a donated sample is used, the participating lab donating the sample will have a 20% discount on the DPT scheme fee in the next scheme year.

  For the DPT scheme we need at least 300 ml of urine from a patient affected with an established inborn error of metabolism, accompanied by a short clinical report. If possible, please collect 1500 ml of urine: this sample can be used as the common sample and be circulated to all labs participating to the DPT schemes. Each urine sample must be collected from a single patient. Please don't send a pool of urines, except if urine has been collected during a short period of time from the same patient. As soon as possible after collection, the urine sample must be heated at 50 °C for 20 minutes. Make sure that this temperature is achieved in the entire urine sample, not only in the water bath. Then aliquot the sample in 10 ml plastic tubes (minimum 48 tubes), add stoppers and freeze. Be careful to constantly homogenize the urine while aliquoting the sample. Send the aliquots on dry ice by rapid mail or express transport to:

  Mrs Joanne Croft
  Dept of Clinical Chemistry
  Sheffield Children's NHS Foundation
  Trust, Western Bank
  Sheffield, S10 2TH
  United Kingdom

  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

If you are not performing 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. 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.

14. Questions, Suggestions and Complaints

If you have any questions, comments or suggestions please address to the Scientific Advisor of the scheme, Joanne Croft 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, 2026-03-14
Name and signature of Scientific Advisor

Mrs Joanne Croft
Dept of Clinical Chemistry
Sheffield Children's NHS Foundation
Trust, Western Bank
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APPENDIX 1. Change log (changes since the last version)

Version Number	Published	Amendments
1	March 17 2026	2025 annual report published

END