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Diagnostic Proficiency Testing Centre: Czech Republic

Final Report 2025

prepared by
Petr Chrastina

Note: This annual report is intended for participants of the ERNDIM DPT Czech Republic 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 in the ERNDIM Privacy Policy on www.erndim.org.

1. Geographical distribution of participants

Fifteen laboratories from 12 countries have participated in the Diagnostic Proficiency Testing scheme Czech Republic in 2025, for details see the below table:

Country	Number of participants
Austria	1
Croatia	1
Cyprus	1
Czechia	1
Finland	1
Germany	4
Latvia	1
Lithuania	1
Malaysia	1
Portugal	1
Slovakia	1
United Kingdom	1

[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 Petr Chrastina as Scientific Advisor and coordinated by Alessandro Salemma as scheme organizer (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://cscqwstest.hcuge.ch/qls/Initial/Initial.php

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

Origin of patients: All six urines were obtained from patients with known diagnoses. Four urine samples have been provided by the scheme organizers and by Department of Clinical Biochemistry of University Children's Hospital in Bratislava. The common sample was from DPT center France (distributed in all five DPT schemes).

In 2025 the samples have been heat-treated and apart from the common sample A, were re-analyzed in our department after receiving the samples from CSCQ (samples were shipped via courier at ambient temperature to mimic possible changes that might arise during transport). We prepared and checked all five samples, and the typical metabolic profiles were preserved after heat treatment in each one. 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

Analyses of amino acids, organic acids, mucopolysaccharides, oligosaccharides and purines/pyrimidines were required in 2025.

4. Schedule of the scheme

Sample distribution	05 February 2025
Start of analysis of Survey 2025/1	17 March 2025
Survey 2025/1 – results submission	07 April 2025
Survey 2025/1 – report	20 May 2025
Start of analysis of Survey 2025/2	02 June 2025
Survey 2025/2 – results submission	23 June 2025
Survey 2025/2 – report	04 August 2025
Annual meeting of participants	09 October 2025
Annual report 2025	January-March 2026

5. Results

14 of 15 labs returned results for both surveys by the deadline.

	Survey 1	**Survey 2**
Receipt of results	15	14
No answer	0	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 for scoring.**

- 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 for scoring.

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 Czech Republic 2025 have been also scored by George Ruijter, from DPT NL. At the SAB meeting on 27th November 2025, the definitive scores have been finalized.

ERNDIM applies the concept of 'critical error' in the scoring of results. In principle this is a category of error that would be unacceptable to the majority of labs and would have a serious adverse effect on patient management. Examples of such errors could be:

a) Failure to recognize a pre-defined set of diagnoses.
b) Missing a diagnosis when proficiency for that EQA sample is >95% (where proficiency for an EQA sample is the percentage of EQA participants that correctly identified the diagnosis of the sample).
c) Failure to perform a relevant test.
d) Identifying a 'normal' sample as having an IEM when it is clear that the sample was obtained from a patient not suspected of having an IEM and the findings reported were not identified by the rest of the participants and this diagnosis could potentially result in treatment that is harmful for the patient.

When a critical error is established for one or more samples, performance is not acceptable in that year, regardless of the number of points assigned. A critical error needs to be ratified by the ERNDIM Scientific Advisory Board. In 2025, critical errors were established for sample B and C.

A certificate of participation will be issued for participation, and it will be additionally notified whether the participant has received a performance support letter. This performance support letter is sent out if the performance is evaluated as unsatisfactory. Any partial submitters will receive a letter from the ERNDIM Executive Administrator, Sara Gardner.

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)

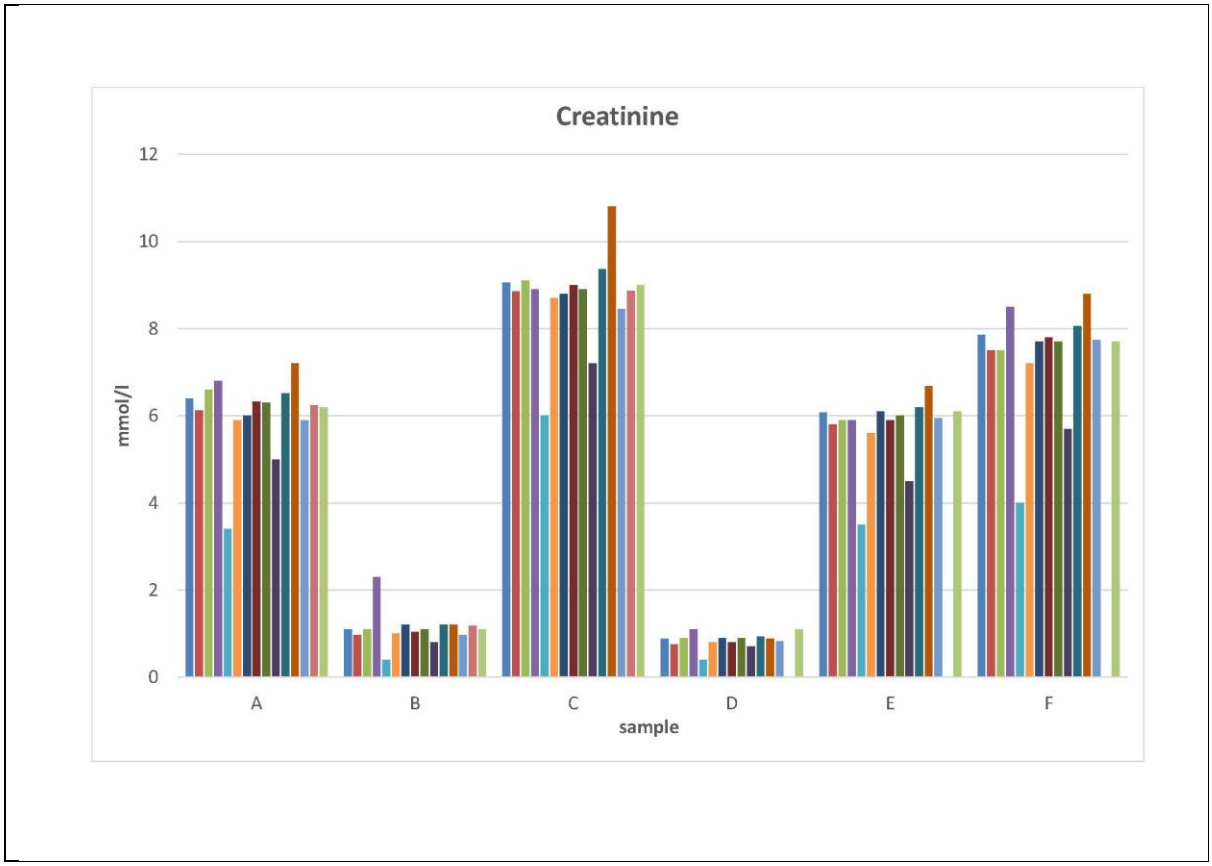
7.1. Score for satisfactory performance

Performance of the participant that obtained at least 17 points from the maximum of 24 (71%) and more within the calendar year and that did not receive "critical error" mark is considered satisfactory.

8. Results of samples and evaluation of reporting

8.1. Creatinine measurement for all samples

Creatinine determination was mostly satisfying with some outliers. There was a systematic error in two labs in both surveys.



8.2. Patient A

mucopolysaccharidosis type VI due to arylsulfatase B deficiency

Patient details provided to participants

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

Patient details

The sample was obtained from a 16-years old boy with mucopolysaccharidosis type VI due to arylsulfatase B deficiency. The diagnosis was confirmed by molecular genetic analysis.

Analytical performance

Eleven participants analyzed glycosaminoglycans (GAG) in urine and 13 participants performed GAG fractionation. Elevated excretion of glycosaminoglycans without report on dermatan sulfate elevation was considered as partially correct and scored with 1 point. Increased proportion of dermatan sulphate was scored as correct analytical result with 2 points. The analytical performance for this sample was good (83%).

Interpretative proficiency and recommendation

The diagnosis of mucopolysaccharidosis type VI was considered correct (2 points), while suspicion for MPS (other types of MPS or non-specified MPS) was considered helpful but incomplete (1 point). Confirmation of the diagnosis by measurement of arylsulfatase B in leukocytes/fibroblasts and/or mutation analysis of the ARSB gene was considered helpful. Recommendation to perform GAG fractionation for those participants who did not perform this analysis was also considered helpful (1 point). The proficiency score for this sample was good (80%).

Critical errors

No critical error for this sample.

Overall impression
Typical DPT sample with good proficiency score (82%).

8.3. Patient B

3-methylglutaconic aciduria type IV due to mutation in the TMEM70 gene

Patient details provided to participants
A two-day-old newborn with cyanosis, respiratory distress and severe metabolic acidosis was presented to the emergency department. Urine was collected at the age of 1 year.

Patient details
The sample was obtained from a 1-year-old boy with 3-methylglutaconic aciduria type IV due to mutation in the TMEM70 gene. The diagnosis was confirmed by molecular genetic analysis.

Analytical performance
All participants analyzed organic acids and 14 of them reported elevated excretion of 3-methylglutaconic acid. Such analytical finding was considered correct result and scored with 2 points. The analytical performance for this sample was very good (93%).

Interpretative proficiency and recommendation
The diagnosis of 3-methylglutaconic aciduria type IV or secondary 3-methylglutaconic aciduria due to mitochondrial dysfunction were considered correct (2 points), while suspicion for other types of 3-methylglutaconic aciduria or non-specified 3-methylglutaconic aciduria were considered helpful but incomplete (1 point). Confirmation of diagnosis by mutation analysis of TMEM70 gene were considered helpful. The proficiency score for this sample was good (83%).

Critical errors
The failure to recognize abnormal excretion of 3-methylglutaconic acid is considered by the ERNDIM SAB as a critical error, which would prevent establishing the correct diagnosis; critical error was assigned to one participant in our scheme.

Overall impression

Typical DPT sample with good proficiency score (88%).

8.4. Patient C

cystinuria

Patient details provided to participants
A 17-year-old woman presented to the emergency department with severe abdominal pain. A CT scan revealed a renal calculus. The sample was taken at the age of 17 years during the specific treatment.

Patient details
The sample was obtained from 17-years old woman with cystinuria. The diagnosis was confirmed by molecular genetic analysis.

Analytical performance
All participants analyzed urinary amino acids. Fourteen participants reported presence of increased excretion of cystine, such analytical finding was considered a correct analytical result and scored by 1 point. All participants reported dibasic hyperaminoaciduria, such analytical finding was considered a correct analytical result and scored by 1 point. The analytical performance was very good (97%).

Interpretative proficiency and recommendation
The diagnosis of cystinuria was considered the correct and scored with 2 points. Confirmation of diagnosis by mutation analysis of SLC3A1 and SLC7A9 genes was considered helpful. The proficiency score for this sample was very good (93%).

Critical errors
The failure to recognize abnormal excretion of cystine is considered by the ERNDIM SAB as a critical error, which would prevent establishing the correct diagnosis; critical error was assigned to one

participant in our scheme.

Overall impression

Easy DPT sample with very good proficiency score (95%).

8.5. Patient D

GM1-gangliosidosis due to beta-galactosidase deficiency

Patient details provided to participants

A seven-month-old boy was examined for transaminase elevation, facial dysmorphia, psychomotor retardation, and ptosis. Urine was collected at the age of 1 year.

Patient details

This sample was obtained from a 1-year-old boy with GM1-gangliosidosis due to beta-galactosidase deficiency. The diagnosis was confirmed by molecular genetic analysis.

Analytical performance

Eleven laboratories performed OLS analysis and all of them reported a correct analytical result "OLS profile characteristic for GM1-gangliosidosis", which was scored 2 points. The analytical performance was slightly suboptimal (79%).

Interpretative proficiency and recommendation

The diagnosis of GM1-gangliosidosis due to beta-galactosidase deficiency was considered correct and scored 2 points. Confirmation of the diagnosis by enzyme assay of beta-galactosidase activity in fibroblasts/leukocytes and/or mutation analysis of the GLB1 gene was considered helpful. The interpretation proficiency score for this sample was slightly suboptimal (79%).

Critical errors

No critical error for this sample.

Overall impression

Typical DPT sample with slightly suboptimal proficiency score (72%).

8.6. Patient E

mucopolysaccharidosis type I due to alpha-L-iduronidase deficiency

Patient details provided to participants

A 12-year-old boy was referred for metabolic screening because of an unknown disease of the finger joints, rheumatic origin was excluded. Urine was collected at the age of 13 years.

Patient details

The sample was obtained from a 13-year-old boy with mucopolysaccharidosis type I due to alpha-L-iduronidase deficiency, diagnosis was confirmed by molecular genetic analysis.

Analytical performance

Ten participants analyzed glycosaminoglycans (GAG) in urine and 12 participants performed GAG fractionation. Increased proportion of dermatan sulphate and/or heparan sulphate was scored as correct analytical result with 2 points. The analytical performance for this sample was good (86%).

Interpretative proficiency and recommendation

The diagnosis of mucopolysaccharidosis type I was considered correct (2 points), while suspicion for MPS (other types of MPS or non-specified MPS) was considered helpful but incomplete (1 point). Confirmation of the diagnosis by measurement of alpha-L-iduronidase in leukocytes/fibroblasts and/or mutation analysis of the IDUA gene was considered helpful. Recommendation to perform GAG fractionation for those participants who did not perform this analysis was also considered helpful (1 point). The proficiency score for this sample was good (89%).

Critical errors

No critical error for this sample.

Overall impression

Typical DPT sample with good proficiency score (88%).

8.7. Patient F

no IEM

Patient details provided to participants

This woman was referred for hearing impairment at the age of 51. The sample was taken at the age of 51.

Patient details

This sample was obtained from a 51-year-old woman with no evidence of an inherited metabolic disorder, despite extensive metabolic screening.

Analytical performance

All labs performed analysis of organic acids. Nine labs reported normal profile of organic acids, four labs reported mildly elevated excretion of lactic acids or pyruvic acids only. This analytical finding was considered correct and scored one point. Ten participants analyzed at least three of the five required methods, reporting a normal profile except for organic acids. Such an analytical findings were considered correct and scored one point. Overall, the analytical performance was good (82%).

Interpretative proficiency and recommendation

We considered the report of “no IEM” or “non-specific finding” a good diagnosis, which was scored with 2 points. The interpretative proficiency score for this sample was suboptimal (64%).

Critical errors

No critical error for this sample.

Overall impression

Typical DPT sample with slightly suboptimal proficiency score (73%).

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 mucopolysaccharidosis type VI			Patient B 3-methylglutaconic aciduria type IV			Patient C cystinuria			
	A	I	Total	A	I	Total	A	I	Total	Total
1	2	2	4	2	2	4	2	2	4	12
2	2	1	3	2	2	4	2	2	4	11
3	2	1	3	0	0	0	2	2	4	7
4	2	2	4	2	2	4	2	2	4	12
5	1	1	2	2	2	4	2	2	4	10
6	2	2	4	2	2	4	2	2	4	12
7	2	2	4	2	2	4	2	2	4	12
8	0	1	1	2	1	3	2	2	4	8
9	0	1	1	2	1	3	2	2	4	8
10	2	2	4	2	2	4	2	2	4	12
11	2	2	4	2	1	3	2	2	4	11
12	2	2	4	2	2	4	2	2	4	12
13	2	2	4	2	2	4	1	0	1	9
14	2	2	4	2	2	4	2	2	4	12
15	2	1	3	2	2	4	2	2	4	11

Detailed scores – Round 2

Lab n°	Patient D GM1-gangliosidosis			Patient E mucopolysaccharidosis type I			Patient F no IEM			
	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	2	2	4	2	2	4	8
3	2	2	4	2	2	4	2	2	4	12
4	2	2	4	2	2	4	2	0	2	10
5	2	2	4	2	2	4	2	2	4	12
6	2	2	4	2	2	4	2	0	2	10
7	2	2	4	2	2	4	2	2	4	12
8	2	2	4	0	1	1	2	2	4	9
9	0	0	0	0	1	1	1	2	3	4
10	0	0	0	2	2	4	1	2	3	7
11	2	2	4	2	2	4	2	2	4	12
12	2	2	4	2	2	4	2	0	2	10
13	2	2	4	2	1	3	0	0	0	7
14	--	--	--	--	--	--	--	--	--	0
15	2	2	4	2	2	4	1	0	1	9

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	3	4	4	0	4	4	19	79	
3	3	0	4	4	4	4	19	79	CE
4	4	4	4	4	4	2	22	92	
5	2	4	4	4	4	4	22	92	
6	4	4	4	4	4	2	22	92	
7	4	4	4	4	4	4	24	100	
8	1	3	4	4	1	4	17	71	
9	1	3	4	0	1	3	12	50	
10	4	4	4	0	4	3	19	79	
11	4	3	4	4	4	4	23	96	
12	4	4	4	4	4	2	22	92	
13	4	4	1	4	3	0	16	67	CE
14	4	4	4	--	--	--	12	50	
15	3	4	4	4	4	1	20	83	

Performance

	Number of labs	% total labs
Satisfactory performers (≥ 71 % of adequate responses)	11	73
Unsatisfactory performers (< 70 % adequate responses and/or critical error)	3	20
Partial and non-submitters	1	7

Overall Proficiency

Sample	Diagnosis	Analytical (%)	Interpretation (%)	Total (%)
DPT-CP-2025-A	mucopolysaccharidosis type VI	83	80	82
DPT-CP-2025-B	3-methylglutaconic aciduria type IV	93	83	88
DPT-CP-2025-C	cystinuria	97	93	95
DPT-CP-2025-D	GM1-gangliosidosis	79	79	79
DPT-CP-2025-E	mucopolysaccharidosis type I	86	89	88
DPT-CP-2025-F	no IEM	82	64	73

10. Annual meeting of participants

The annual meeting of participants of the Proficiency Testing Centre Czech Republic was held during ERNDIM 2025 Symposium 9th October 2025 in Madrid, Spain.

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

- In 2026, ERNDIM will launch a new scheme developed in response to participant feedback: Lipids in Serum (LIS). This quantitative scheme builds on two years of pilot testing and features a panel of lipids, some of which have been moved from the SAS scheme, and others newly introduced. Our aim is to expand the LIS panel over time to include additional diagnostically valuable markers.

- **Urine samples**: we remind you that every year, each participant must provide to the scheme organizer at least 300 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. 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. For “normal” urine, the sample must be collected from a symptomatic patient (don’t send urine from your kids!). As soon as possible after collection, the urine sample must be heated at 56 °C for 30 minutes. Make sure that this temperature is achieved in the entire

urine sample, not only in the water bath. Please send us an e-mail about possible samples and we will organize transport with your cooperation.

- If a sample donated by your laboratory is used as an EQA material in one of the Qualitative EQA schemes, you will qualify for a 20% discount on the cost of that specific scheme when you register for the following scheme year.

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. Tentative schedule in 2026

Sample distribution	04 February 2026
Start of analysis of Survey 2026/1	17 March 2026
Survey 2026/1 – results submission	07 April 2026
Survey 2026/1 – report	19 May 2026
Start of analysis of Survey 2026/2	01 June 2026
Survey 2026/2 – results submission	22 June 2026
Survey 2026/2 – report	03 August 2026
Annual meeting of participants	August 2026
Annual report 2026	January 2027

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, Suggestions and Complaints

If you have any questions, comments or suggestions please address to the Scientific Advisor of the scheme, Petr Chrastina and/or to the ERNDIM Administration Office (admin@erndim.org).

Most complaints received by ERNDIM consist of minor misunderstandings or problems with samples, which can usually be resolved via direct contact with the ERNDIM administrative staff. If you wish to file a formal complaint, please email your complaint with details of your issue to admin@erndim.org or contact us through our website at https://www.erndim.org/contact-us/

Date of report, 2026-01-22
Name and signature of Scientific Advisor

Petr Chrastina

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

Version Number	Published	Amendments
1	22 January 2026	2025 annual report published

END