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Urine Mucopolysaccharides

Centre: The Netherlands

Final Report 2024

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

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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.

1. Introduction

The ERNDIM Urine Mucopolysaccharide scheme offers (1) urine samples obtained from confirmed MPS patients to enable laboratories to gain or maintain experience to identify MPS patients and (2) proficiency testing for laboratories providing urine screening of mucopolysaccharidoses. The scheme is organized by University Medical Centre Utrecht, the Netherlands in conjunction with MCA, the Dutch organization for quality assurance in medical laboratories (MCA laboratory, Winterswijk, the Netherlands) and CSCQ, the Swiss organization for quality assurance in medical laboratories.

[1] If this report is not Version 1 for this scheme year, go to Page 22 for details of the changes made since the last version of this document.

2. Geographical distribution of participants

In 2024, 91 laboratories from many different countries registered for the Urine MPS scheme. The number of participants is relatively stable over the years (2021: 87, 2022: 88, 2023: 88 participants). In 2024 there were 3 educational participants. Educational participants take part in all aspects of the scheme and receive interim reports with scores, but performance is not indicated on the ERNDIM certificate of performance.

Country	Number of participants
Argentina	2
Australia	4
Austria	1
Belgium	5
Brazil	2
Canada	4
Chile	1
China	1
Colombia	1
Croatia	1
Cyprus	1
Czech Republic	1
Estonia	1
France	5
Germany	10
Greece	1
Hong Kong	1
Italy	4

Country	Number of participants
Latvia	1
Malaysia	2
Netherlands	3
New Zealand	2
Norway	1
Poland	1
Portugal	2
Qatar	1
Singapore	1
South Africa	2
Spain	4
Sweden	1
Switzerland	2
Turkey	1
Ukraine	1
United Kingdom	13
United States	6
Uruguay	1

3. Design and logistics of the scheme including sample information

The scheme has been designed and planned by Dr. Berthil Prinsen as Scientific Advisor and coordinated by Dr. Alessandro Salemma and Dr. Nicola Braik (sub-contractors on behalf of CSCQ) and Dr. Cas Weykamp (sub-contractors on behalf of MCA Laboratories) as scheme organizers, all appointed by and according to procedures laid down the ERNDIM Board.
As a subcontractor of ERNDIM, MCA prepares lyophilized sample aliquots and dispatches UMPS EQA samples to the scheme participants by courier. CSCQ provides a website for online submission of results and access to scheme reports. Existing 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: samples UMPS-NL-2024-A, B and C
	Round 2: samples UMPS-NL-2024-D, E and F

As usual, the samples used in 2024 were authentic human urine samples, five from MPS patients and one from a non-MPS individual. Three samples were obtained from the sample repository at Erasmus MC, Rotterdam, The Netherlands. One sample was obtained from the UMC Utrecht, Utrecht, the Netherlands, one sample was obtained from the General University Hospital, Prague, Czech Republic and one sample was obtained from the Hospices Civils de Lyon – CHU de Lyon, Lyon, France. Samples were selected by the Scientific Advisor and tested for suitability in the Scientific Advisor's laboratory (UMC Utrecht, Utrecht, the Netherlands). Integrity of the samples was checked after preparation of the lyophilized aliquots in the Scientific Advisor's laboratory before shipment to participants. Details regarding stability of (reconstituted) samples are provided in the sample package.

UMPS-NL-2024-A	Female, 27 years	MPS-I
UMPS-NL-2024-B	Male, 16 years	MPS-VII
UMPS-NL-2024-C	Male, 40 years	Control subject
UMPS-NL-2024-D	Male, 7 years	MPS-III
UMPS-NL-2024-E	Male, 55 years	MPS-I
UMPS-NL-2024-F	Male, 20 years	MPS-IV

4. Tests

Tests required for participation in the Urine MPS scheme are creatinine analysis and GAG analysis (quantitative (total) GAG and GAG-subtyping, either qualitative by electrophoresis/TLC or mass-spectrometry). Participants are asked to interpret the GAG concentration according to age-matched reference values (i.e normal or increased), interpret GAG subfractions (i.e. normal or increased CS (chondroitin-sulphate), HS (heparan-sulphate), DS (dermatan-sulphate) and KS (keratan-sulphate) and to give the most likely diagnosis.

5. Schedule of the scheme

- 6 February 2024: sample dispatch
- 11 March 2024: analysis start (survey 1)
- 15 April 2024: website available for result submission (survey 1)
- 13 May 2024: deadline for result submission (survey 1)
- 27 June 2024: interim report of survey 1 available for download
- 8 July 2024: analysis start (survey 2)
- 12 August 2024: website available for result submission (survey 2)
- 09 September 2024: deadline for result submission (survey 2)
- 21 October 2024: interim report of survey 2 available for download
- January, 2025: annual report with final scoring, confirmed by the SAB, available for download

6. Results submitted

83 out of the 91 labs that were registered returned results for both surveys.

	Survey 1	**Survey 2**
Receipt of results	88	86
No report	3	5

7. Website reporting

Website reporting system is compulsory for all participants. Please note, the website includes a section to specify methods. Method specification is required for correct evaluation of the quantitative results (method specific statistics for DMB, harmine, Alcian Blue, CPC and mass-spectrometry). Unfortunately, not all participants have specified their methods.

Since 2017, an evaluation program made by Dr. Albe from CSCQ is used to evaluate and score results submitted by participants. The use of this software enabled production of customised interim reports and the annual report, i.e. including scores, for each individual participant.

8. Scoring and evaluation of results

Information regarding procedures for establishment of assigned values, statistical analysis, interpretation of statistical analysis can be found in generic documents on the ERNDIM website.
The scoring system has been established by the Scientific Advisory Board of ERNDIM. Scores are allocated to different elements of the results reported. Two aspects are evaluated: 1) analytical performance, 2) interpretative proficiency. The total score is calculated as a sum of these two aspects. Similar to other qualitative (proficiency testing) ERNDIM schemes, the maximum score for a sample is 4 points. The scores were calculated only for laboratories submitting results.

A	Analytical performance	Correct results of the appropriate tests	2
		Partially correct or missing results	1

		Unsatisfactory or misleading	0
I	Interpretative proficiency	Correct (differential) diagnosis was established	2
		Helpful, but (partially) incorrect	1
		Misleading or wrong diagnosis	0

The specific criteria applied to score the results of the samples included in the 2024 scheme are given under item 9. These criteria have been set by the Scientific Advisor, approved by the Scientific Advisory Board, and have been devised on the basis of (1) for each sample: the type of MPS, (2) current possibilities of routine MPS testing, and (3) actual achievable results for a particular sample.
The final decision about scoring was made in the Scientific Advisory Board (SAB) during the autumn meeting (28th November – 29th November, 2024 in Leiden for the 2024 scheme).

A note on scoring of diagnostic proficiency and the use of check boxes and the comment box:
To indicate the most likely diagnosis check boxes must be used to facilitate evaluation of results. The use of the 'comments' box in the website form is recommended to explain your interpretation of results and recommendations. Comments will be taken into account to score interpretation.
For example, we have noted in previous surveys that it may be hard to distinguish MPS I and VI. In the case of increased DS with normal or undetectable HS, checking just the MPS VI box may result in lower than maximum marks if this actually was a MPS I sample. In this case we advise to check the MPS VI box and explain in the comments box that MPS I (and perhaps II) cannot be excluded on the basis of the results. Or alternatively the boxes for MPS I, II and VI could be checked with a comment entered explaining that MPS VI is more likely.
It is important to realize, when no diagnosis is selected a comment or recommendation is mandatory that needs to explain why the diagnosis 'no diagnosis' is selected. This information is essential for correct scoring of your samples.

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, the SAB decided that samples UMPS-NL-2024-A, UMPS-NL-2024-D and UMPS-NL-2024-E were eligible for critical error. For UMPS-NL-2024-B and UMPS-NL-2024-F, it was decided to score the sample. UMPS-NL-2024-C (control subject) was not eligible for critical error.

Score required for satisfactory performance: at least 17 points from the maximum of 24 (70%).
From the 88 regular (non-educational) participants 83 participants (94%) submitted results for two rounds of which 71 achieved satisfactory performance (2 reports submitted, score ≥17, no critical error). In 2024, there was 1 non-submitter (no-results submitted) and 2 partial-submitters (1 survey submitted instead of 2 reports submitted). Twelve participants did not accomplish satisfactory performance. Two participants withdrew from the scheme.

A certificate of participation, including a statement on performance (satisfactory yes/no) will be issued for participation. In addition, performance support letters will be sent out if the performance is evaluated as unsatisfactory. Twelve performance support letters were sent by the Scheme Advisor for 2024. Any partial submitters or non-submitters will receive a letter from the ERNDIM office.

9. Results of the samples and evaluation of reporting

9.1. Creatinine and total GAG results of all samples

Quantitative results of creatinine and total GAG were summarised in the two interim reports.

Parameter/Method	UMPS-NL-2024-A	UMPS-NL-2024-B	UMPS-NL-2024-C	UMPS-NL-2024-D	UMPS-NL-2024-E	UMPS-NL-2024-F
Creatinine (mmol/L)						
Average	3,34	2,54	6,07	2,52	5,81	4,23
SD	0,35	1,82	4,03	0,17	0,42	0,27

Parameter/Method	UMPS-NL-2024-A	UMPS-NL-2024-B	UMPS-NL-2024-C	UMPS-NL-2024-D	UMPS-NL-2024-E	UMPS-NL-2024-F
Median	3,31	2,34	5,70	2,50	5,76	4,20
N	87	88	88	86	86	86
GAG quantitative (mg/mmol creat) DMB-test						
Average			1,9	22,5	26,8	11,6
SD			1,2	6,5	7,1	3,5
Median			1,7	22,7	26,6	11,3
N			58	60	60	60
GAG quantitative (mg/mmol creat) Alcian blue colorimetric tests						
Average	26,0	20,0	3,0	23,2	37,8	14,1
SD	0,0	0,0	0,0	0,3	2,5	2,7
Median	26,0	20,0	3,0	23,2	37,8	14,1
N	1	1	2	2	2	2
GAG quantitative (mg/mmol creat) Uronic acids - carbazole/harmine method						
Average	6,0	52,9	1,0	6,8	6,9	2,5
SD	0,0	0,0	0,4	6,1	2,8	0,6
Median	6,0	52,9	1,0	7,5	7,8	2,6
N	1	1	2	3	3	3
GAG quantitative (mg/mmol creat) LC-MS/MS GAG fragments (Saville method)						
Average	24,2	18,7	1,5	70,2	36,4	9,6
SD	0,0	0,0	0,1	68,4	5,1	4,0
Median	24,2	18,7	1,5	70,2	36,4	9,6
N	1	1	2	2	2	2

Quantitative GAG results were evaluated separately for most methods (DMB, Alcian Blue, Harmine/carbazole, CPC/turbidity). Most participants use DMB for quantitative total GAG analysis (approximately 65% in 2023).
Figure 1 gives an overview of the different mass-spectrometry related techniques that are used for GAG-analysis in the UMPS-scheme in 2017 and 2023. The number of participants that use these techniques for GAG-analysis is slightly increasing over the years (approximately 20% in 2023).

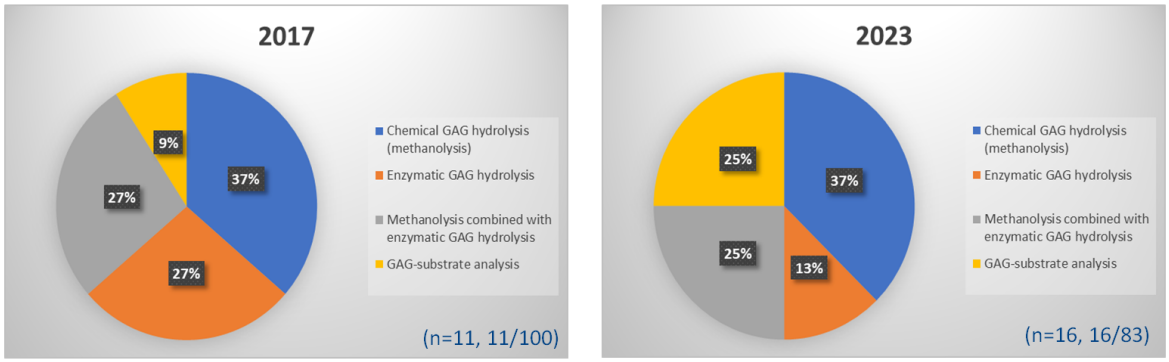


Figure 1: Overview of the different mass-spectrometry related techniques that are used for GAG-analysis in the UMPS-scheme in 2017 and 2023 (data are presented at the ERNDIM Workshop in Porto, SSIEM 2-6 September 2024).

9.2. Sample UMPS-NL-2024-A; MPS-I

Patient details
This urine sample was obtained from an adult female of 27 years old with MPS-I. Diagnosis was confirmed by enzyme testing.

Analytical performance
Nearly all participants (79/81, 98%) that performed GAG-screening did report an abnormal GAG-screening test result (e.g. DMB-test). Elevated DS was reported by 78/81 (96%) and 46/78 (59%) participants reported elevated HS. The analytical performance of this sample was 94%.

Diagnosis / Interpretative proficiency
Eighteen participants (21%) concluded that this sample was obtained from a patient with MPS-I. In total 74 participants (74/88, 84%) reported a differential diagnosis including MPS-I in various combinations with MPS-II, VI and VII. One participant reported no abnormalities for quantitative GAGs and GAG-subtyping and did not give an advice for follow-up. The diagnostic performance of this sample was 88% and the total performance was 91%.

Diagnosis	N	%
MPS I/MPS II/MPS VI/MPS VII	20	23,8
MPS I	18	21,4
MPS I/MPS VI/MPS VII	8	9,5
MPS I/MPS II/MPS VI	8	9,5
MPS I/MPS II/MPS VII	7	8,3
MPS I/MPS II	7	8,3
MPS VI	5	6,0
MPS I/MPS VI	2	2,4
MPS I/MPS VII	2	2,4
MPS II	1	1,2
MPS III	1	1,2
MPS I/MPS III	1	1,2
MPS IV	1	1,2

Diagnosis	N	%
MPS I/MPS III/MPS VII	1	1,2
Normal	1	1,2
No Diagnosis	1	1,2
N results	84	100
N non-submitters	7	
N registered	91	

Scoring

- Analytical results: Elevated (total) GAG: 1 mark, elevated DS: 1 mark.
- Interpretation: MPS-I mentioned in the differential diagnosis (based on elevated DS): 2 marks. Combinations of MPS-II, VI or VII based on elevated DS: 1 mark.
- Critical error: Reporting a normal profile as the most likely diagnosis was considered as a critical error for this sample (n=1).

9.3. Sample UMPS-NL-2024-B; MPS-VII

Patient details

This urine sample was obtained from a patient of 16 years old with MPS-VII. Diagnosis was confirmed by enzyme testing.

Analytical performance

A considerable number of participants observed a mild elevated (total) GAGs concentration (78/81, 96%). Since CS, DS and HS all contain glucuronic acid residues, elevation of these GAG-species could theoretically be expected in a MPS-VII urine sample. Elevated CS was reported by 33/72 (46%) participants, elevated DS by 41/79 (52%) participants and elevated HS by 24/78 (31%) participants. In some electrophoresis methods CS and KS are not well separated and it could be possible that some participants have misinterpreted the CS elevation in this sample as being KS. Elevated KS was reported by 10/68 (15%) participants. The analytical performance of this sample was 80%.

Diagnosis / Interpretative proficiency

Many different differential diagnoses were reported (see table below). MPS-VII was reported by 8 participants (10%) as a single possible diagnosis. A number of participants (25/88) included MPS-VII in their differential diagnosis. In total 33 participants (33/88, 38%) reported MPS-VII in their (differential) diagnosis. Fifteen participants reported ‘normal profile/no diagnosis’ as diagnosis (15/88, 17%), 9 participants (9/88, 10%) concluded that this sample was obtained from a patient with MPS-VI, while 11 participants reported MPS-IV (11/88, 12,5%). The diagnostic proficiency was only 43%, which was much lower compared to the analytical performance.

It is possible that a number of participants do not have experience with MPS-VII samples and perhaps do not include MPS-VII in their differential diagnosis when CS or DS are elevated. Several labs missed marks in the diagnostic follow-up of the results, because MPS-VII was not mentioned in the differential diagnosis. The total proficiency of this sample was 61%.

Diagnosis	N	%
MPS IV	11	13,6
MPS VI	9	11,1
No Diagnosis	9	11,1
MPS VII	8	9,9
MPS I/MPS II/MPS VI/MPS VII	8	9,9
MPS VI/MPS VII	7	8,6
Normal	6	7,4
MPS I/MPS II/MPS VI	5	6,2
MPS III	4	4,9
MPS I/MPS II/MPS VII	3	3,7
MPS IV/MPS VII	3	3,7

Diagnosis	N	%
MPS I/MPS II	1	1,2
MPS VII/No Diagnosis	1	1,2
MPS III/No Diagnosis	1	1,2
MPS I/MPS VI/MPS VII	1	1,2
MPS II/MPS III	1	1,2
MPS VII/Normal	1	1,2
MPS I/MPS II/MPS III	1	1,2
MPS I/MPS II/MPS III/MPS VI/MPS VII	1	1,2
N results	81	100
N non-submitters	10	
N registered	91	

Scoring
- Analytical results: Elevated (total) GAG: 1 mark, elevated DS and/or CS: 1 mark.
- Interpretation: MPS-VII mentioned in the differential diagnosis (based on elevated DS/CS): 2 marks. Combinations of MPS-I, II, IV and VI based on elevated DS/CS: 1 mark.
- Critical error: This sample was not considered eligible for critical error.

9.4. Sample UMPS-NL-2024-C; Normal profile

Patient details
This sample was obtained from a healthy male subject of 40 years old.

Analytical performance
All participants (80/80, 100%) reported a normal quantitative GAG-screening test. GAG-subtyping was reported as normal by 80/81 participants (99%), although two participants (2/75, 3%) noticed an increased HS-excretion. The analytical performance of this sample was 98%.

Diagnosis / Interpretative proficiency
As is usual for normal samples, most participants (80/81, 99%) correctly concluded that this urine samples was not a sample of a patient with a MPS. One participant concluded that this sample was obtained from a patient with MPS-III. The diagnostic performance was 96% and the overall proficiency of this sample was 97%.

Diagnosis	N	%
Normal	74	91,4
No Diagnosis	6	7,4
MPS III	1	1,2
No Diagnosis/Normal	0	0,0
N results	81	100
N non-submitters	10	
N registered	91	

Scoring
- Analytical results: Normal (total) GAGs and normal GAG-subtyping were each scored 1 mark.
- Interpretation: A normal profile and other combinations with normal profile/no diagnosis were scored 2 marks.
- Critical error: The sample was not considered eligible for critical error.

9.5. Sample UMPS-NL-2024-D; MPS-III

Patient details
This sample was obtained from a male subject of 7 years old with MPS-IIIC. Diagnosis was confirmed by enzyme testing.

Analytical performance
Most of the participants reported elevated quantitative GAGs (76/77, 99%) and the majority of the participants (68/80, 85%) reported an elevated HS as well. Remarkedly, 11% of the participants (9/80) reported a normal HS-excretion, while 7% of the participant (5/76) reported an elevated DS. The analytical performance of this sample was 91%.

Diagnosis / Interpretative proficiency
In total 71/85 participants concluded that this sample was obtained from a patient with MPS-III. Three participants reported a normal profile as the most likely diagnosis. Of these, one participant did not notice any abnormalities (normal quantitative GAGs and GAG-subtyping), while the other two participants reported elevated quantitative GAGs with normal GAG-subtyping. For this sample reporting a normal profile was considered to be a critical error. The diagnostic performance of this sample was 82% and the total performance was 87%.

Diagnosis	**N**	**%**
MPS III	62	74,7
Normal	3	3,6
MPS I/MPS II/MPS VI	2	2,4
MPS I/MPS II/MPS III/MPS VII	2	2,4
No Diagnosis	2	2,4
MPS III/MPS VII	2	2,4
MPS I/MPS II/MPS VI/MPS VII	2	2,4
MPS II/MPS III/MPS IV/MPS VI	1	1,2
MPS IV	1	1,2
MPS III/No Diagnosis	1	1,2
MPS I/MPS II/MPS III/MPS IV/MPS VII	1	1,2
MPS I/MPS II/MPS VII	1	1,2
MPS III/Normal	1	1,2
MPS IV/Normal	1	1,2
MPS I/MPS II/MPS III/MPS VI/MPS VII	1	1,2
N results	83	100
N non-submitters	8	
N registered	91	

Scoring
- Analytical results: Elevated (total) GAG and elevated HS were each scored 1 mark.
- Interpretation: MPS-III was scored 2 marks.
- Critical error: Reporting a normal profile as the most likely diagnosis was considered as a critical error for this sample (n=3).

9.6. Sample UMPS-NL-2024-E; MPS-I

Patient details
This urine sample was obtained from an adult male of 55 years old with MPS-I. Diagnosis was confirmed by enzyme testing.

Analytical performance
98% of the participants (76/77) reported elevated quantitative (total) GAGs. Most of the participants found that GAG-subtyping was abnormal and elevated DS was reported by 99% of the participants (79/80). One lab detected elevated quantitative GAGs, but did not perform a GAG-subtyping. The analytical performance of this sample 97%.

Diagnosis / Interpretative proficiency
Six participants reported MPS-I as diagnosis. In total 76 participants (76/85, 89%) reported a differential diagnosis including MPS-I in various combinations with MPS-II, VI and VII. A normal profile was reported by one participant. The diagnostic performance of this sample was 91% and the total performance was 94%.

Diagnosis	N	%
MPS I/MPS II/MPS VI/MPS VII	27	32,5
MPS I/MPS II	16	19,3
MPS I/MPS II/MPS VII	14	16,9
MPS I/MPS II/MPS VI	12	14,5
MPS I	6	7,2
MPS VI	3	3,6
MPS I/MPS II/MPS III/MPS VII	1	1,2
MPS II	1	1,2
Normal	1	1,2
MPS II/MPS IV/MPS VI	1	1,2
No Diagnosis	1	1,2
N results	83	100
N non-submitters	8	
N registered	91	

Scoring
- Analytical results: Elevated (total) GAG: 1 mark, elevated DS: 1 mark.
- Interpretation: MPS-I mentioned in the differential diagnosis (based on elevated DS): 2 marks. Combinations of MPS-II, VI or VII based on elevated DS: 1 mark.
- Critical error: Reporting a normal profile as the most likely diagnosis was considered as a critical error for this sample (n=1).

9.7. Sample UMPS-NL-2024-F; MPS-IV

Patient details
This urine sample was obtained from a male subject of 20 years old with MPS-IVA. Diagnosis was confirmed by enzyme testing.

Analytical performance
Abnormal (total) quantitative GAGs was reported by 92% of the participants (71/77). Six participants reported normal (total) quantitative GAGs (8%). From the 72 participants that submitted a result for KS, 49 (68%) reported that KS was elevated. N-acetyl-galactosamine-6-sulphatase deficiency in MPS-IVA may lead to storage of chondroitin-6-suphate and indeed 26 participants (35%) reported elevated CS. Markedly, 7 participants (9%) noticed that DS was present and 5 participants (7%) observed the presence of HS in this sample. The analytical performance of this sample was 82%.

Diagnosis / Interpretative proficiency
MPS-IVA was reported as the most likely diagnosis by 61 participants (61/85, 72%). Three participants reported MPS IV in combination with normal/no diagnosis, while nine participants (11%) reported a normal GAG-subtyping result. Similar MPS-IVA samples were also circulated in 2022 and 2023. In 2022, the total performance was 57% and 69% in 2023. The diagnostic performance in this sample was 72% and the total performance 77%.

Diagnosis	N	%
MPS IV	55	67,1
Normal	9	11,0
No Diagnosis	6	7,3
MPS IV/MPS VII	3	3,7
MPS IV/No Diagnosis	2	2,4
MPS I/MPS II/MPS VI/MPS VII	2	2,4
MPS IV/Normal	1	1,2
MPS VI/MPS VII	1	1,2
MPS I/MPS II/MPS III/MPS VI/MPS VII	1	1,2
MPS III/MPS VII	1	1,2
MPS III	1	1,2
N results	82	100
N non-submitters	9	
N registered	91	

Scoring

- Analytical results: Elevated (total) GAG: 1 mark, elevated KS or CS: 1 mark.
- Interpretation: MPS-IV mentioned in the differential diagnosis (based on elevated KS or CS): 2 marks.
- Critical error: The sample was not considered eligible for critical error.

10. Scores of participants

All data transfer, i.e. the submission of data as well as viewing and downloading of reports proceed via the CSCQ results website. The results of your laboratory are confidential and only accessible to you (with your username and password).The anonymous scores of all laboratories are accessible to all participants and only in your version is your laboratory highlighted in the leftmost column (available from https://cscq.hcuge.ch/cscq/ERNDIM/Initial/Initial.php).

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 you 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°	Sample 1 This urine sample was obtained from a patient with MPS-I.			Sample 2 This urine sample was obtained from a patient with MPS-VII.			Sample 3 This urine sample was obtained from a subject with no indication for an inborn error of metabolism.			
	A	**I**	**Total**	**A**	**I**	**Total**	**A**	**I**	**Total**	**Total**
1	2	2	4	2	2	4	2	2	4	12
2	--	--	--	--	--	--	--	--	--	0
3	2	2	4	2	2	4	2	2	4	12
4	2	2	4	2	1	3	2	2	4	11

Lab n°	Sample 1 This urine sample was obtained from a patient with MPS-I.			Sample 2 This urine sample was obtained from a patient with MPS-VII.			Sample 3 This urine sample was obtained from a subject with no indication for an inborn error of metabolism.			
	A	**I**	**Total**	**A**	**I**	**Total**	**A**	**I**	**Total**	**Total**
5	2	2	4	0	0	0	2	2	4	8
6	1	0	1	2	2	4	1	2	3	8
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	0	2	2	2	4	10
10	1	2	3	2	0	2	2	2	4	9
11	2	2	4	2	2	4	2	2	4	12
12	2	2	4	1	0	1	2	2	4	9
13	2	2	4	2	2	4	2	2	4	12
14	2	2	4	1	0	1	1	0	1	6
15	2	2	4	2	2	4	2	2	4	12
16	2	2	4	2	2	4	2	0	2	10
17	2	2	4	2	0	2	2	2	4	10
18	2	2	4	2	1	3	2	2	4	11
19	2	2	4	2	2	4	2	2	4	12
20	2	2	4	2	2	4	2	2	4	12
21	2	2	4	2	0	2	2	2	4	10
22	2	2	4	2	2	4	2	2	4	12
23	2	2	4	2	2	4	2	2	4	12
24	2	2	4	2	2	4	2	2	4	12
25	2	2	4	1	0	1	2	2	4	9
26	2	2	4	2	2	4	2	2	4	12
27	2	2	4	1	0	1	2	2	4	9
28	2	2	4	2	2	4	2	2	4	12
29	1	0	1	1	0	1	2	2	4	6
30	2	2	4	1	0	1	2	2	4	9
31	2	2	4	1	0	1	2	2	4	9
32	2	2	4	1	0	1	2	2	4	9
33	1	0	1	1	0	1	2	2	4	6
34	1	0	1	1	0	1	2	2	4	6

Lab n°	Sample 1 This urine sample was obtained from a patient with MPS-I.			Sample 2 This urine sample was obtained from a patient with MPS-VII.			Sample 3 This urine sample was obtained from a subject with no indication for an inborn error of metabolism.			
	A	**I**	**Total**	**A**	**I**	**Total**	**A**	**I**	**Total**	**Total**
35	2	2	4	2	2	4	2	2	4	12
36	0	0	0	0	0	0	1	0	1	1
37	2	2	4	2	2	4	2	2	4	12
38	2	2	4	0	0	0	2	2	4	8
39	1	0	1	1	0	1	2	2	4	6
40	2	2	4	2	0	2	2	2	4	10
41	2	2	4	2	0	2	2	2	4	10
42	2	2	4	2	0	2	2	2	4	10
43	--	--	--	--	--	--	--	--	--	0
44	2	2	4	2	2	4	2	2	4	12
45	2	2	4	2	2	4	2	2	4	12
46	2	2	4	1	0	1	2	2	4	9
47	2	2	4	1	2	3	2	2	4	11
48	2	2	4	1	0	1	2	2	4	9
49	2	2	4	2	1	3	2	2	4	11
50	2	2	4	2	2	4	2	2	4	12
51	2	2	4	2	2	4	2	2	4	12
52	2	2	4	1	0	1	2	2	4	9
53	2	1	3	1	0	1	2	2	4	8
54	2	2	4	1	0	1	2	2	4	9
55	2	2	4	1	0	1	2	2	4	9
56	2	2	4	2	0	2	2	2	4	10
57	2	2	4	2	2	4	2	2	4	12
58	2	2	4	2	0	2	2	2	4	10
59	2	1	3	1	0	1	2	2	4	8
60	2	2	4	1	0	1	2	2	4	9
61	2	2	4	2	2	4	2	2	4	12
62	2	2	4	1	0	1	2	2	4	9
63	2	2	4	2	0	2	2	2	4	10
64	2	1	3	2	1	3	2	2	4	10

Lab n°	Sample 1 This urine sample was obtained from a patient with MPS-I.			Sample 2 This urine sample was obtained from a patient with MPS-VII.			Sample 3 This urine sample was obtained from a subject with no indication for an inborn error of metabolism.			
	A	**I**	**Total**	**A**	**I**	**Total**	**A**	**I**	**Total**	**Total**
65	2	2	4	1	0	1	2	2	4	9
66	2	2	4	1	2	3	2	2	4	11
67	2	2	4	1	0	1	2	2	4	9
68	2	2	4	2	0	2	2	2	4	10
69	2	2	4	2	1	3	2	2	4	11
70	2	2	4	2	2	4	2	2	4	12
71	2	2	4	2	2	4	2	2	4	12
72	2	2	4	2	0	2	2	2	4	10
73	2	2	4	1	0	1	2	2	4	9
74	1	0	1	1	0	1	2	2	4	6
75	2	1	3	2	0	2	2	2	4	9
76	2	2	4	2	2	4	2	2	4	12
77	2	2	4	2	1	3	1	0	1	8
78	2	2	4	2	2	4	2	2	4	12
79	2	2	4	2	1	3	2	2	4	11
80	2	2	4	1	0	1	2	2	4	9
81	2	2	4	2	0	2	2	2	4	10
82	2	2	4	2	2	4	2	2	4	12
83	--	--	--	--	--	--	--	--	--	0
84	2	2	4	1	0	1	2	2	4	9
85	2	2	4	2	1	3	2	2	4	11
86	2	2	4	2	2	4	2	2	4	12
87	1	0	1	1	0	1	2	2	4	6
88	2	1	3	2	0	2	2	2	4	9
89	2	1	3	2	0	2	2	2	4	9
90	2	2	4	2	2	4	2	2	4	12
91	2	2	4	2	2	4	2	2	4	12

Detailed scores – Round 2

Lab n°	Sample 4 This urine sample was obtained from a patient with MPS-III.			Sample 5 This urine sample was obtained from a patient with MPS-I.			Sample 6 This urine sample was obtained from a patient with MPS-IV.			
	A	**I**	**Total**	**A**	**I**	**Total**	**A**	**I**	**Total**	**Total**
1	2	2	4	2	2	4	2	2	4	12
2	--	--	--	--	--	--	--	--	--	0
3	1	0	1	2	2	4	1	0	1	6
4	1	0	1	2	2	4	2	2	4	9
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	1	2	3	11
9	2	2	4	2	2	4	2	2	4	12
10	2	2	4	1	2	3	0	0	0	7
11	2	2	4	2	2	4	2	2	4	12
12	2	2	4	2	2	4	2	2	4	12
13	1	0	1	2	0	2	2	2	4	7
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	1	2	3	11
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	1	0	1	9
21	2	2	4	2	2	4	0	0	0	8
22	2	2	4	2	2	4	2	2	4	12
23	2	2	4	2	2	4	1	0	1	9
24	2	2	4	2	2	4	1	0	1	9
25	2	2	4	2	2	4	2	2	4	12
26	2	2	4	2	2	4	2	2	4	12
27	2	2	4	2	2	4	2	2	4	12
28	2	2	4	2	2	4	1	0	1	9
29	2	2	4	2	2	4	2	2	4	12

Lab n°	Sample 4 This urine sample was obtained from a patient with MPS-III.			Sample 5 This urine sample was obtained from a patient with MPS-I.			Sample 6 This urine sample was obtained from a patient with MPS-IV.			
	A	**I**	**Total**	**A**	**I**	**Total**	**A**	**I**	**Total**	**Total**
30	2	2	4	2	2	4	2	2	4	12
31	2	2	4	2	2	4	0	0	0	8
32	2	2	4	2	2	4	1	0	1	9
33	--	--	--	--	--	--	--	--	--	0
34	1	0	1	1	0	1	1	0	1	3
35	2	2	4	2	2	4	2	2	4	12
36	2	2	4	2	2	4	2	2	4	12
37	2	2	4	2	2	4	2	2	4	12
38	2	2	4	2	2	4	1	0	1	9
39	1	0	1	1	0	1	1	0	1	3
40	2	2	4	2	2	4	2	2	4	12
41	2	2	4	2	2	4	0	0	0	8
42	2	2	4	2	2	4	2	2	4	12
43	--	--	--	--	--	--	--	--	--	0
44	2	2	4	2	2	4	2	2	4	12
45	2	2	4	2	2	4	2	2	4	12
46	2	2	4	2	2	4	2	2	4	12
47	2	2	4	2	2	4	1	0	1	9
48	--	--	--	--	--	--	--	--	--	0
49	2	0	2	2	2	4	2	2	4	10
50	2	2	4	2	2	4	2	2	4	12
51	2	2	4	2	2	4	2	2	4	12
52	2	2	4	2	2	4	2	2	4	12
53	2	2	4	2	2	4	2	2	4	12
54	1	2	3	2	2	4	2	2	4	11
55	2	2	4	2	0	2	2	2	4	10
56	2	2	4	2	2	4	2	0	2	10
57	2	2	4	2	2	4	1	0	1	9
58	1	0	1	2	2	4	1	0	1	6
59	1	0	1	2	2	4	1	0	1	6
60	2	2	4	2	2	4	1	2	3	11
61	2	2	4	2	2	4	2	2	4	12

Lab n°	Sample 4 This urine sample was obtained from a patient with MPS-III.			Sample 5 This urine sample was obtained from a patient with MPS-I.			Sample 6 This urine sample was obtained from a patient with MPS-IV.			
	A	**I**	**Total**	**A**	**I**	**Total**	**A**	**I**	**Total**	**Total**
62	2	2	4	2	2	4	2	2	4	12
63	2	2	4	2	2	4	2	2	4	12
64	2	2	4	2	2	4	2	2	4	12
65	2	2	4	2	2	4	2	2	4	12
66	2	0	2	2	2	4	1	2	3	9
67	0	0	0	0	0	0	0	0	0	0
68	0	0	0	2	2	4	2	2	4	8
69	2	2	4	2	2	4	2	2	4	12
70	2	0	2	2	2	4	2	0	2	8
71	2	2	4	2	2	4	2	2	4	12
72	2	2	4	2	2	4	2	2	4	12
73	2	2	4	1	0	1	2	0	2	7
74	1	2	3	2	1	3	1	2	3	9
75	2	2	4	2	2	4	2	2	4	12
76	2	2	4	2	2	4	2	2	4	12
77	2	2	4	2	1	3	2	2	4	11
78	2	2	4	2	2	4	1	0	1	9
79	2	0	2	2	2	4	2	2	4	10
80	1	0	1	2	0	2	2	2	4	7
81	2	2	4	2	2	4	2	2	4	12
82	2	2	4	2	2	4	1	2	3	11
83	--	--	--	--	--	--	--	--	--	0
84	1	2	3	2	2	4	2	0	2	9
85	1	0	1	2	2	4	1	0	1	6
86	2	2	4	2	2	4	2	2	4	12
87	1	0	1	1	0	1	0	0	0	2
88	2	2	4	2	2	4	2	2	4	12
89	2	2	4	2	2	4	2	2	4	12
90	2	2	4	2	2	4	2	2	4	12
91	2	2	4	2	2	4	2	2	4	12

Total scores

Lab n°	1	2	3	4	5	6	Cumulative score	Cumulative score (%)	Critical error
1	4	4	4	4	4	4	24	100	
2	--	--	--	--	--	--	0	0	
3	4	4	4	1	4	1	18	75	
4	4	3	4	1	4	4	20	83	
5	4	0	4	4	4	4	20	83	
6	1	4	3	4	4	4	20	83	
7	4	4	4	4	4	4	24	100	
8	4	4	4	4	4	3	23	96	
9	4	2	4	4	4	4	22	92	
10	3	2	4	4	3	0	16	67	
11	4	4	4	4	4	4	24	100	
12	4	1	4	4	4	4	21	88	
13	4	4	4	1	2	4	19	79	
14	4	1	1	4	4	4	18	75	
15	4	4	4	4	4	4	24	100	
16	4	4	2	4	4	3	21	88	
17	4	2	4	4	4	4	22	92	
18	4	3	4	4	4	4	23	96	
19	4	4	4	4	4	4	24	100	
20	4	4	4	4	4	1	21	88	
21	4	2	4	4	4	0	18	75	
22	4	4	4	4	4	4	24	100	
23	4	4	4	4	4	1	21	88	
24	4	4	4	4	4	1	21	88	
25	4	1	4	4	4	4	21	88	
26	4	4	4	4	4	4	24	100	
27	4	1	4	4	4	4	21	88	
28	4	4	4	4	4	1	21	88	
29	1	1	4	4	4	4	18	75	
30	4	1	4	4	4	4	21	88	
31	4	1	4	4	4	0	17	71	
32	4	1	4	4	4	1	18	75	
33	1	1	4	--	--	--	6	25	

Lab n°	1	2	3	4	5	6	Cumulative score	Cumulative score (%)	Critical error
34	1	1	4	1	1	1	9	38	
35	4	4	4	4	4	4	24	100	
36	0	0	1	4	4	4	13	54	CE
37	4	4	4	4	4	4	24	100	
38	4	0	4	4	4	1	17	71	
39	1	1	4	1	1	1	9	38	
40	4	2	4	4	4	4	22	92	
41	4	2	4	4	4	0	18	75	
42	4	2	4	4	4	4	22	92	
43	--	--	--	--	--	--	0	0	
44	4	4	4	4	4	4	24	100	
45	4	4	4	4	4	4	24	100	
46	4	1	4	4	4	4	21	88	
47	4	3	4	4	4	1	20	83	
48	4	1	4	--	--	--	9	38	
49	4	3	4	2	4	4	21	88	
50	4	4	4	4	4	4	24	100	
51	4	4	4	4	4	4	24	100	
52	4	1	4	4	4	4	21	88	
53	3	1	4	4	4	4	20	83	
54	4	1	4	3	4	4	20	83	
55	4	1	4	4	2	4	19	79	
56	4	2	4	4	4	2	20	83	
57	4	4	4	4	4	1	21	88	
58	4	2	4	1	4	1	16	67	CE
59	3	1	4	1	4	1	14	58	
60	4	1	4	4	4	3	20	83	
61	4	4	4	4	4	4	24	100	
62	4	1	4	4	4	4	21	88	
63	4	2	4	4	4	4	22	92	
64	3	3	4	4	4	4	22	92	
65	4	1	4	4	4	4	21	88	
66	4	3	4	2	4	3	20	83	
67	4	1	4	0	0	0	9	38	

Lab n°	1	2	3	4	5	6	Cumulative score	Cumulative score (%)	Critical error
68	4	2	4	0	4	4	18	75	CE
69	4	3	4	4	4	4	23	96	
70	4	4	4	2	4	2	20	83	
71	4	4	4	4	4	4	24	100	
72	4	2	4	4	4	4	22	92	
73	4	1	4	4	1	2	16	67	CE
74	1	1	4	3	3	3	15	62	
75	3	2	4	4	4	4	21	88	
76	4	4	4	4	4	4	24	100	
77	4	3	1	4	3	4	19	79	
78	4	4	4	4	4	1	21	88	
79	4	3	4	2	4	4	21	88	
80	4	1	4	1	2	4	16	67	CE
81	4	2	4	4	4	4	22	92	
82	4	4	4	4	4	3	23	96	
83	--	--	--	--	--	--	0	0	
84	4	1	4	3	4	2	18	75	
85	4	3	4	1	4	1	17	71	
86	4	4	4	4	4	4	24	100	
87	1	1	4	1	1	0	8	33	
88	3	2	4	4	4	4	21	88	
89	3	2	4	4	4	4	21	88	
90	4	4	4	4	4	4	24	100	
91	4	4	4	4	4	4	24	100	

Performance

	Number of labs	% total labs
Satisfactory performers (≥ 70 % of adequate responses)	71	86%
Unsatisfactory performers (< 70 % adequate responses and/or critical error)	12	14%
Partial and non-submitters	2	1

Overall Proficiency

Sample	Diagnosis	Analytical (%)	Interpretation (%)	Total (%)
UMPS-NL-2024-A	MPS-I	**94**	**88**	**91**
UMPS-NL-2024-B	MPS-VII	**80**	**43**	**61**
UMPS-NL-2024-C	Control subject	**98**	**95**	**97**
UMPS-NL-2024-D	MPS-III	**91**	**82**	**87**
UMPS-NL-2024-E	MPS-I	**97**	**91**	**94**
UMPS-NL-2024-F	MPS-IV	**82**	**72**	**77**

11. Tentative schedule for 2025

Sample distribution	4 February
Start of analysis of Survey 2025-1 (website open)	14 April
Survey 2025-1 - Results submission deadline	12 May
Survey 2025-1 – Interim Reports	23 June
Start of analysis of Survey 2025-2 (website open)	11 August
Survey 2025-2 – Results submission deadline	8 September
Survey 2025-2 – Interim Reports	20 October
Annual Report 2025	January- March 2026

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

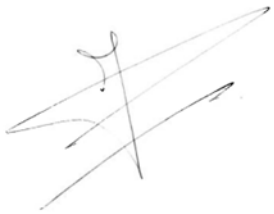
13. Questions, Comments and Suggestions

If you have any questions, comments or suggestions please address to the Scientific Advisor of the scheme, Dr. H.C.M..T. Prinsen, b.prinsen@umcutrecht.nl and/or to the ERNDIM Administration Office (admin@erndim.org).

Please read:
In order to achieve a satisfactory performance for the UMPS scheme, it is strongly recommended to perform both quantitative (total) GAG-analysis and GAG-subtyping for each sample. No points are rewarded when experiments are not performed.

The urine samples in this scheme are obtained from MPS-patients that are confirmed by enzyme testing or DNA-analysis. We notice that it is very difficult to obtain sufficient urine of MPS-patients (off treatment). If you have an urine sample of a MPS patient available, please do contact the scientific advisors (Dr. H.C.M.T. Prinsen or Dr. G.J.G. Ruijter). When the sample is suitable and selected for this scheme, your laboratory gets a discount for the next year.

Date of report, 28-03-2025



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

Version Number	Published	Amendments
1	15-04-2025	2024 annual report published

END