ERNDIM Administration Office
c/o EMQN CIC
Third Floor, ICE Building,
3 Exchange Quay, Salford
M5 3ED, United Kingdom.
Tel: +44 161 757 4952
Fax: +44 161 850 1145
Email: admin@erndim.org

Scientific Coordination
Dr. H.C.M.T. Prinsen
UMC Utrecht
Dept of Genetics, section Metabolic Diagnostics
KC.02.069.1
3584 CX Utrecht
The Netherlands
Email: admin@erndim.org

Scheme Organization
CSCQ (Quality Control Centre, Switzerland)
Alessandro Salemma, Rose Defossez
2 chemin du Petit-Bel-Air
1225 Chêne-Bourg
Switzerland,
Tel: +41 22 305 52 36
Email: erndim.survey@cscq.ch

Scheme Organization
Dr. R.M. Schoeman
Streekziekenhuis Koningin Beatrix
MCA Laboratory
Beatrixpark 1
7101 BN Winterswijk
The Netherlands
Email: ma.office@skbwinterswijk.nl

Published: 12th February 2026[1]

Urine Mucopolysaccharides
Centre: The Netherlands

Final Report 2025

prepared by
Dr. H.C.M.T. (Berthil) Prinsen and Dr. G.J.G. (George) Ruijter

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.

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 performance of your laboratory, unless ERNDIM is required to disclose performance data by a relevant government agency. For details please see 'ERNDIM Terms and conditions' and the ERNDIM Privacy Policy on www.erndim.org.

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 APPENDIX 1 for details of the changes made since the last version of this document.

2. Geographical distribution of participants

In 2025, 86 laboratories from many different countries have registered for the Urine MPS scheme. The number of participants is relatively stable over the years (2022: 88, 2023: 88, 2024: 91 participants). In 2025, there were 2 educational participants. One participant was withdrawn for the scheme. 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
Undefined country	2
Argentina	1
Australia	4
Austria	1
Belgium	5
Brazil	2
Canada	3
Chile	1
China	1
Colombia	1
Croatia	1
Cyprus	1
Czech Republic	1
Estonia	1
France	5
Germany	8
Hong Kong	1
Italy	4
Japan	1
Latvia	1
Malaysia	2
Netherlands	4
New Zealand	2
Norway	1
Poland	1
Portugal	2
Singapore	1
South Africa	2
Spain	4
Sweden	1
Switzerland	2
Turkey	1
Ukraine	1
United Kingdom	13
United States	6

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. Rose Defossez (sub-contractors on behalf of CSCQ) and Dr. R.M. Schoeman (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: UMPS-NL-2025-A, B and C
	Round 2: UMPS-NL-2025-D, E and F

As usual, the samples used in 2025 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 UMC Utrecht, Utrecht, the Netherlands. One sample was obtained from the Erasmus MC, Rotterdam, The Netherlands, one sample was obtained from the General University Hospital, Prague, Czech Republic and one sample was obtained from the Heidelberg University Hospital, Heidelberg, Germany. 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. The samples are stable for the duration of the scheme's submission calendar when stored under defined conditions.

UMPS-NL-2025-A	Male, 25 years old	MPS-III
UMPS-NL-2025-B	Male, 7 years old	Control subject
UMPS-NL-2025-C	Female, 3 years old	MPS-I
UMPS-NL-2025-D	Female, 31 years old	MPS-I
UMPS-NL-2025-E	Male, 10 years old	MPS-IV
UMPS-NL-2025-F	Male, 42 years old	MPS-II

4. Tests

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

5. Schedule of the scheme

- 4 February 2025: sample dispatch
- 10 March 2025: analysis start (survey 1)
- 14 April 2025: website available for result submission (survey 1)
- 12 May 2025: deadline for result submission (survey 1)
- 23 June 2025: interim report of survey 1 available for download
- 7 July 2025: analysis start (survey 2)
- 11 August 2025: website available for result submission (survey 2)
- 08 September 2025: deadline for result submission (survey 2)
- 20 October 2025: interim report of survey 2 available for download
- January, 2026: annual report with final scoring, confirmed by the SAB, available for download

6. Results submitted

All participants (n = 86) that were registered returned results for both surveys.

	Survey 1	**Survey 2**
Receipt of results	86	86
No report	0	0

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. 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)

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 2025 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 (27th November – 28th November, 2025 in Leiden for the 2025 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 2025, the SAB decided that samples UMPS-NL-2025-A, UMPS-NL-2025-C, UMPS-NL-2025-E and UMPS-NL-2025-F were eligible for critical error. For UMPS-NL-2025-D, it was decided to score the sample. UMPS-NL-2025-B (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 84 regular (non-educational) participants 84 participants (100%) submitted results for two rounds of which 77 achieved satisfactory performance (2 reports submitted, score ≥17, no critical error). Six participants did not accomplish satisfactory performance. One participant was withdrawn 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. Six performance support letters were sent by the Scheme Advisor for 2025. 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-2025-A	UMPS-NL-2025-B	UMPS-NL-2025-C	UMPS-NL-2025-D	UMPS-NL-2025-E	UMPS-NL-2025-F
Creatinine (mmol/L)						
Average	7,71	6,32	1,50	4,64	1,42	2,78
SD	0,42	0,41	0,18	0,31	0,17	0,22
Median	7,69	6,28	1,46	4,57	1,40	2,76
N	84	84	84	85	84	84
GAG quantitative (mg/mmol creat) Dimethyl Methylene Blue tests						
Average	5,6	6,5	64,0	7,1	19,4	26,5
SD	1,6	1,9	18,7	2,1	6,1	6,9
Median	5,8	6,0	65,1	7,0	18,9	26,4
N	56	56	58	59	58	58
GAG quantitative (mg/mmol creat) Alcian blue colorimetric tests						
Average	6,2	8,8	71,2	8,9	23,3	29,7
SD	1,7	0,9	12,4	1,9	4,1	4,3
Median	6,2	8,8	71,2	8,0	23,9	30,5
N	2	2	2	3	3	3
GAG quantitative (mg/mmol creat) CPC turbidity method						
Average	16,7	5,9	78,9	24,2	13,0	40,7
SD	6,7	0,2	18,9	11,6	2,3	6,7
Median	16,7	5,9	78,9	24,2	13,0	40,7
N	2	2	2	2	2	2

9.2. Creatinine and total GAG results of all samples

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. The number of participants that use mass-spectrometry for GAG-analysis is increasing over the years (>20% in 2025).

We noticed that GAG-values obtained by DMB-testing that are in the range of the upper limit of normal may be interpretated differentially. For sample UMPS-NL-2025-A, the mean GAG-value in the scientific advisor laboratory was approximately 7.8 mg/mmol creatinine (n<6.7 mg/mmol creatinine) and slightly elevated. Several participants reported their value as normal, while other participants that use DMB-testing as well, reported their results as clearly abnormal. In the table below, reference values for DMB-testing are included, where the upper limit of normal (ULN) was shown as average + 1.2 SD and not average + 2 SD or higher. This strategy was used to reduce the number of false negatives, since it is known that GAG-elevations can be very subtle and patients with MPS-III and MPS-IV can be easily missed using DMB-testing alone. Several participants perform GAG-analyse by DMB-testing and subtle differences can be present between the different laboratories. Therefore, it is strongly advisable to create you our own in house reference values for the DMB-test.

Age	Max value (avg+1.2SD)	AVG	SD	n	+1SD	+2SD
0-1 months	71,1	49,9	17,7	31	67,6	85,3
1 -6 months	54,4	37,6	14	31	51,6	65,6
6 -12 months	36,9	26,6	8,6	21	35,2	43,8
1-2 year	32,5	23	7,9	40	30,9	38,8
2-4 year	26,1	19,3	5,7	68	25	30,7
4-6 year	18,3	13,4	4,1	45	17,5	21,6
6-10 year	15,2	10,4	4	42	14,4	18,4
10-15 year	11,2	7,8	2,8	46	10,6	13,4
15-20 year	8,1	5,3	2,3	29	7,6	9,9
> 20 year	6,7	4,5	1,8	37	6,3	8,1

(Courtesy of Dr. G.J.G. (George) Ruijter, method is described in PMID 1597005)

9.3. Sample UMPS-NL-2025-A; MPS-III

Patient details
This urine sample was obtained from a male patient with MPS-IIIA on treatment (SCT). The actual age at urine collection was 17 years. Since this was the first time that a patient on treatment was included in the scheme, the age of the patient was adapted to 25 years to make the diagnosis slightly easier. Diagnosis was confirmed by enzyme and genetic testing.

Analytical performance
A majority of the participants reported elevated quantitative GAGs (58/86, 67%) and most of the participants reported an elevated HS (76/86, 88%). Remarkedly, 12% of the participants (10/81) reported elevated DS and 3 participants (3/86, 3%) reported a normal HS-excretion. The analytical performance of this sample was 78%.

Diagnosis / Interpretative proficiency
In total 70/86 participants (81%) concluded that this sample was obtained from a patient with MPS-III. Three participants reported a no abnormalities (normal GAGs and normal GAG-subtyping) in this sample and reported a normal profile as the most likely diagnosis. Three laboratories reported no diagnosis and made relevant comments in the comment box why no diagnosis was reported. These additional comments prevented the 3 participants from a critical error. For this sample reporting a normal profile was considered to be a critical error. The diagnostic performance of this sample was 81% and the total performance was 80%.

Diagnosis	**N**	**%**
MPS III	59	68,6
MPS I/MPS II/MPS III/MPS VII	6	7,0
MPS I/MPS II/MPS VI/MPS VII	3	3,5
Normal	3	3,5
No Diagnosis	3	3,5
MPS I/MPS II/MPS III	2	2,3
MPS I/MPS II	2	2,3
MPS IV/Normal	2	2,3
MPS VII	1	1,2
MPS III/MPS IV	1	1,2
MPS III/MPS VII	1	1,2
MPS IV	1	1,2
MPS I/MPS II/MPS VII	1	1,2
MPS I/MPS II/MPS III/MPS VI/MPS VII	1	1,2

Diagnosis	N	%
N results	86	100
N non-submitters	2	
N registered	88	

Scoring
- Analytical results: Elevated (total) GAGs and elevated HS were each scored 1 mark. The detection of MPS-IIIA substrate was scored 2 marks.
- 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).

9.4. Sample UMPS-NL-2025-B; Normal

Patient details
This urine sample was obtained from a healthy male subject of 7 years old.

Analytical performance
Nearly all particpants (85/86, 99%) reported a normal quantitative GAG-screening test. GAG-subtyping was reported as normal by 82/86 participants (95%), although 2 participants (2/80, 3%) noticed increased DS-excretion and 1 participant (1/80, 1%) noticed increased HS-excretion. The analytical performance of this sample was 96%.

Diagnosis / Interpretative proficiency
As is usual for normal samples, most participants (82/86, 95%) correctly concluded that this urine sample was not a sample of a patient with a MPS. Four participants concluded that this sample was obtained from a patient with a MPS (different combinations). The diagnostic performance was 94% and the overall proficiency of the sample was 96%.

Diagnosis	N	%
Normal	70	81,4
No Diagnosis	5	5,8
No Diagnosis/Normal	3	3,5
MPS IV/Normal	2	2,3
MPS IV	2	2,3
MPS I/MPS II/MPS III/MPS VII/Normal	1	1,2
MPS I/MPS II	1	1,2
MPS VI/Normal	1	1,2
MPS VII	1	1,2
N results	86	100
N non-submitters	2	
N registered	88	

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-2025-C; MPS-I

Patient details
The urine sample was obtained from a patient of 3 years old with MPS-I. Diagnosis was confirmed by

enzyme and genetic testing.

Analytical performance
All participants (86/86, 100%) reported elevated quantitative (total) GAGs. Nearly all participants found that GAG-subtyping was abnormal (85/86, 99%) and elevated DS was reported by 99% of the participants (82/83). Forty-six participants (46/81, 59%) reported elevated HS as well, while 16 particpants (16/81, 20%) reported normal HS. The analytical performance of this sample was 99%.

Diagnosis / Interpretative proficiency
Thirteen participants (15% 13/86) reported MPS-I as diagnosis. In total 80 participants (80/86, 93%) reported a differential diagnosis including MPS-I in various combinations with MPS-II, VI and VII. None of the participants reported a normal profile. The diagnostic performance of this sample was 98% and the total performance was 99%.

Diagnosis	N	%
MPS I/MPS II/MPS VI/MPS VII	25	29,1
MPS I	13	15,1
MPS I/MPS II/MPS VII	10	11,6
MPS I/MPS VI/MPS VII	8	9,3
MPS I/MPS II/MPS VI	7	8,1
MPS I/MPS II	6	7,0
MPS I/MPS VII	4	4,7
MPS I/MPS VI	3	3,5
MPS VI	3	3,5
MPS II	1	1,2
MPS I/MPS III/MPS VI/MPS VII	1	1,2
MPS VII	1	1,2
MPS I/MPS II/MPS III/MPS VI/MPS VII	1	1,2
MPS I/MPS II/MPS IV/MPS VII	1	1,2
MPS I/MPS III	1	1,2
MPS I/MPS III/MPS VII	1	1,2
N results	86	100
N non-submitters	2	
N registered	88	

Scoring
- Analytical results: Elevated (total) GAGs and elevated DS were each scored 1 mark. The detection of MPS-I substrate was scored 2 marks.
- 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=0).

9.6. Sample UMPS-NL-2025-D; MPS-I

Patient details
This urine sample was obtained from an adult female of 31 years old. Diagnosis was confirmed by enzyme and genetic testing.

Analytical performance
The analytical performance in the sample was lower in contrast to sample UMPS-NL-2025-C. Quantitative (total) GAGs is sample UMPS-NL-2025-D (corrected for age) was lower (8.1 mg/mmol creatinine, n< 6.7 mg/mmol creatinine, 1,2 fold increased) compared to UMPS-NL-2025-D (57.9

mg/mmol creatinine, n< 26 mg/mmol creatinine, 2.2 fold increased). Although (total) GAG excretion was not so high in this urine sample, 78% of the participants (67/86) reported their quantitative (total) GAGs value as elevated. A majority of the participants found that GAG-subtyping was abnormal (66/86, 77%). Elevated DS was reported by 79% (64/81) and elevated HS was reported by 54% (43/80). Normal DS was reported by 15% (12/81) and normal HS was reported by 33% (26/80) of the participants. Eight participants reported no abnormalities (normal GAGs and normal GAG-subtyping) in this sample and reported a normal profile/no diagnosis as the most likely diagnosis, suggesting that this urine sample was challenging sample for a number of participants. The analytical performance of this sample was 77%.

Diagnosis / Interpretative proficiency
Thirteen participants (13/86, 15%) reported MPS-I as diagnosis. In total 60 participants (60/86, 70%) reported a differential diagnosis including MPS-I in various combinations with MPS-II, VI and VII. A normal profile was reported by 8 participants (8/86, 9%). The diagnostic performance of this sample was 72%. The total performance was 75% and was lower than expected.

Diagnosis	N	%
MPS I/MPS II/MPS VI/MPS VII	21	24,4
MPS I	13	15,1
MPS VI	8	9,3
MPS I/MPS II/MPS VII	7	8,1
MPS III	6	7,0
MPS I/MPS VI/MPS VII	6	7,0
Normal	5	5,8
MPS I/MPS II/MPS VI	4	4,7
MPS I/MPS II	3	3,5
No Diagnosis	3	3,5
MPS VII	2	2,3
MPS I/MPS III/MPS VII	1	1,2
MPS I/MPS IV/MPS VI/MPS VII	1	1,2
MPS VI/MPS VII	1	1,2
MPS I/MPS VI	1	1,2
MPS I/MPS III/MPS VI/MPS VII	1	1,2
MPS III/MPS VII	1	1,2
MPS I/MPS VII	1	1,2
MPS I/MPS II/MPS III/Normal	1	1,2
N results	86	100
N non-submitters	0	
N registered	86	

Scoring
- Analytical results: Elevated (total) GAGs and elevated DS were each scored 1 mark. The detection of MPS-I substrate was scored 2 marks.
- Interpretation: MPS-I, or 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: The sample was not considered eligible for critical error.

9.7. Sample UMPS-NL-2025-E; MPS-IV

Patient details
This urine sample was obtained from a male patient with MPS-IVA. Diagnosis was confirmed by enzyme testing.

Analytical performance
Abnormal (total) quantitative GAGs was reported by 91% of the participants (78/86) and 8 participants reported normal (total) quantitative GAGs (8/86, 9%). Fifty-nine participants reported elevated KS (59/71, 83%). N-acetyl-galactosamine-6-sulphatase deficiency in MPS-IVA may lead tot storage of chondroitin-6-sulphate and indeed 32 participants (32/73, 44%) reported elevated CS. Markedly, 9 participants (9/79, 11%) noticed that DS was present and 7 participants (7/79, 9%) observed the presence of HS in this sample. The analytical performance of this sample was 88%.

Diagnosis / Interpretative proficiency
MPS-IV was reported as the most likely diagnosis by 73 participants (73/86, 85%). Seven participants (7/86, 8%) reported a normal profile/no diagnosis as the most likely diagnosis. Five out of seven participants made relevant comments in the comment box. These additional comments prevented the 5 participants from a critical error. Two participants reported no abnormalities (normal GAGs and normal GAG-subtyping). For this sample reporting a normal profile was considered to be a critical error. Similar MPS-IVA samples were also circulated in 2023 and 2024. In 2023, the total performance was 69% and 77% in 2024. The diagnostic performance for this sample was 85% and the total performance was 87%. This observation suggests that the techniques that participants use for diagnosis of patients with MPS including MPS-IVA has improved over the years (e.g. (NRE-GAG) mass-spectrometry).

Diagnosis	N	%
MPS IV	66	77,6
Normal	5	5,9
MPS IV/Normal	2	2,4
MPS IV/MPS VI	2	2,4
No Diagnosis	2	2,4
MPS I/MPS II/MPS VI/MPS VII	2	2,4
MPS IV/MPS VII/No Diagnosis	1	1,2
MPS VII	1	1,2
MPS IV/MPS VII	1	1,2
MPS I/MPS II/MPS III/MPS IV/MPS VI/MPS VII	1	1,2
MPS I/MPS II/MPS III	1	1,2
MPS III	1	1,2
N results	85	100
N non-submitters	1	
N registered	86	

Scoring
- Analytical results: Elevated (total) GAGs and elevated CS or KS were each scored 1 mark. The detection of MPS-IVA substrate was scored 2 marks.
- Interpretation: MPS-IV mentioned (based on elevated CS or KS) 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=2).

9.8. Sample UMPS-NL-2025-F; MPS-II

Patient details
This urine sample was obtained from an adult male of 42 years old with MPS-II. Diagnosis was confirmed by enzyme and genetic testing.

Analytical performance
Nearly all participants reported elevated quantitative (total) GAGs (83/86, 97%). Most of the participants found that GAG-subtyping was abnormal (80/86, 93%) and elevated DS was reported by 94% of the participants (76/81). Elevated HS was reported by 75% of the participants (59/79). Markedly, 4 participants reported normal DS (4/81, 5%) and 11 participants noticed no elevation of HS (11/79, 14%). One participant reported no abnormalities (normal GAGs and normal GAG-subtyping). The analytical

performance of this sample was 95%.

Diagnosis / Interpretative proficiency
Eight participants reported MPS-II as diagnosis. In total 80 participants (80/86, 93%) reported a differential diagnosis including MPS-II in various combinations with MPS-I, VI and VII. A normal profile was reported by 1 participant. The diagnostic performance of this sample was 93% and total performance was 94%.

Diagnosis	N	%
MPS I/MPS II/MPS VI/MPS VII	25	29,4
MPS I/MPS II	20	23,5
MPS I/MPS II/MPS VII	15	17,6
MPS II	8	9,4
MPS I/MPS II/MPS VI	7	8,2
MPS III	2	2,4
MPS I/MPS II/MPS III	1	1,2
MPS I/MPS II/MPS III/MPS IV/MPS VI/MPS VII	1	1,2
MPS IV	1	1,2
MPS I/MPS II/MPS III/MPS VI/MPS VII	1	1,2
Normal	1	1,2
MPS I	1	1,2
MPS I/MPS II/MPS IV/MPS VII	1	1,2
MPS I/MPS II/MPS III/MPS VII	1	1,2
N results	85	100
N non-submitters	1	
N registered	86	

Scoring
- Analytical results: Elevated (total) GAGs and elevated DS were each scored 1 mark. The detection of MPS-II substrate was scored 2 marks.
- Interpretation: MPS-II, or MPS-II mentioned in the differential diagnosis (based on elevated DS): 2 marks. Combinations of MPS-I, 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).

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 1 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°	Sample 1 This urine sample was obtained from a patient with MPS-III.			Sample 2 This urine sample was obtained from a control subject.			Sample 3 This urine sample was obtained gfrom a patient with MPS-I.			
	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	0	0	0	2	2	4	2	2	4	8
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	1	2	3	2	2	4	1	2	3	10
8	1	2	3	2	2	4	2	2	4	11
9	2	2	4	2	2	4	2	2	4	12
10	1	2	3	2	2	4	2	2	4	11
11	1	2	3	2	2	4	2	2	4	11
12	1	0	1	2	2	4	2	2	4	9
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	2	2	4	12
17	1	2	3	2	2	4	2	2	4	11
18	2	0	2	2	2	4	2	0	2	8
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	2	4	2	2	4	12
22	2	2	4	2	2	4	2	2	4	12
23	2	2	4	2	2	4	2	2	4	12
24	1	2	3	2	2	4	2	2	4	11
25	1	2	3	2	2	4	2	2	4	11
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	2	2	4	12
29	2	2	4	2	2	4	2	2	4	12
30	2	2	4	2	2	4	2	2	4	12

Lab n°	Sample 1 This urine sample was obtained from a patient with MPS-III.			Sample 2 This urine sample was obtained from a control subject.			Sample 3 This urine sample was obtained gfrom a patient with MPS-I.			
	A	**I**	**Total**	**A**	**I**	**Total**	**A**	**I**	**Total**	**Total**
31	2	0	2	2	2	4	2	2	4	10
32	1	0	1	2	2	4	2	2	4	9
33	2	2	4	2	2	4	2	2	4	12
34	2	2	4	2	2	4	2	2	4	12
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	2	2	4	12
39	2	2	4	2	2	4	2	2	4	12
40	2	2	4	2	2	4	2	2	4	12
41	2	2	4	2	2	4	2	2	4	12
42	2	2	4	2	2	4	2	2	4	12
43	1	2	3	2	2	4	2	2	4	11
44	1	2	3	2	2	4	2	2	4	11
45	1	2	3	2	2	4	2	2	4	11
46	2	2	4	2	2	4	2	2	4	12
47	1	0	1	2	2	4	2	2	4	9
48	2	2	4	1	0	1	2	2	4	9
49	1	2	3	2	2	4	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	2	2	4	2	2	4	12
53	1	2	3	2	2	4	2	2	4	11
54	1	2	3	2	2	4	2	2	4	11
55	1	0	1	2	2	4	2	2	4	9
56	1	2	3	2	2	4	2	2	4	11
57	1	2	3	2	2	4	2	2	4	11
58	1	2	3	2	2	4	2	2	4	11
59	2	2	4	2	2	4	2	2	4	12
60	2	2	4	2	2	4	2	0	2	10
61	1	0	1	2	2	4	2	2	4	9

Lab n°	Sample 1 This urine sample was obtained from a patient with MPS-III.			Sample 2 This urine sample was obtained from a control subject.			Sample 3 This urine sample was obtained gfrom a patient with MPS-I.			
	A	**I**	**Total**	**A**	**I**	**Total**	**A**	**I**	**Total**	**Total**
62	1	2	3	2	2	4	2	2	4	11
63	2	2	4	2	2	4	2	2	4	12
64	1	2	3	2	0	2	2	2	4	9
65	1	2	3	2	2	4	2	2	4	11
66	1	0	1	2	2	4	2	2	4	9
67	2	0	2	1	2	3	2	2	4	9
68	2	2	4	2	2	4	2	2	4	12
69	1	0	1	2	2	4	2	2	4	9
70	2	2	4	2	2	4	2	2	4	12
71	2	0	2	2	2	4	2	2	4	10
72	0	0	0	2	2	4	2	2	4	8
73	2	2	4	2	2	4	2	2	4	12
74	1	2	3	2	2	4	2	2	4	11
75	1	2	3	2	2	4	2	2	4	11
76	0	0	0	1	0	1	2	2	4	5
77	2	2	4	2	2	4	2	2	4	12
78	0	0	0	1	0	1	2	2	4	5
79	1	0	1	2	2	4	2	2	4	9
80	2	2	4	1	0	1	2	2	4	9
81	2	2	4	2	2	4	2	2	4	12
82	2	2	4	2	2	4	2	2	4	12
83	1	2	3	2	2	4	2	2	4	11
84	2	2	4	2	2	4	2	2	4	12
85	2	2	4	2	2	4	2	2	4	12
86	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-I.			Sample 5 This urine sample was obtained from a patient with MPS-IV.			Sample 6 This urine sample was obtained from an adult patient with MPS-II.			
	A	I	Total	A	I	Total	A	I	Total	Total
1	2	2	4	2	2	4	2	2	4	12
2	2	0	2	2	2	4	2	2	4	10
3	0	0	0	2	2	4	2	2	4	8
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	1	2	3	2	2	4	1	2	3	10
8	2	0	2	1	2	3	2	2	4	9
9	1	0	1	1	2	3	2	2	4	8
10	2	2	4	2	2	4	1	0	1	9
11	2	2	4	2	2	4	2	2	4	12
12	1	0	1	2	2	4	2	2	4	9
13	0	0	0	2	2	4	2	2	4	8
14	2	2	4	2	2	4	2	2	4	12
15	1	0	1	2	2	4	2	2	4	9
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	0	1	2	2	4	2	2	4	9
20	2	2	4	2	2	4	2	2	4	12
21	2	2	4	2	2	4	2	2	4	12
22	2	2	4	2	2	4	2	2	4	12
23	2	2	4	2	2	4	2	2	4	12
24	1	2	3	2	2	4	2	2	4	11
25	1	2	3	1	0	1	2	2	4	8
26	2	2	4	--	--	--	2	2	4	8
27	1	0	1	2	2	4	2	2	4	9
28	2	2	4	2	2	4	2	2	4	12
29	1	0	1	2	2	4	2	2	4	9
30	1	0	1	2	2	4	2	2	4	9

Lab n°	Sample 4 This urine sample was obtained from a patient with MPS-I.			Sample 5 This urine sample was obtained from a patient with MPS-IV.			Sample 6 This urine sample was obtained from an adult patient with MPS-II.			
	A	**I**	**Total**	**A**	**I**	**Total**	**A**	**I**	**Total**	**Total**
31	2	2	4	2	2	4	2	2	4	12
32	1	2	3	2	2	4	2	2	4	11
33	1	0	1	2	2	4	2	2	4	9
34	2	2	4	2	2	4	--	--	--	8
35	2	2	4	2	2	4	2	2	4	12
36	2	2	4	2	2	4	1	2	3	11
37	1	0	1	2	2	4	2	2	4	9
38	2	0	2	1	2	3	2	2	4	9
39	0	0	0	2	2	4	2	2	4	8
40	2	2	4	2	2	4	2	2	4	12
41	2	2	4	2	2	4	2	2	4	12
42	2	2	4	2	2	4	2	2	4	12
43	2	2	4	2	2	4	2	2	4	12
44	1	2	3	2	2	4	2	2	4	11
45	1	2	3	2	2	4	2	2	4	11
46	1	2	3	2	2	4	2	2	4	11
47	2	2	4	2	2	4	2	2	4	12
48	2	2	4	1	0	1	2	2	4	9
49	1	2	3	2	2	4	2	2	4	11
50	2	0	2	2	0	2	1	0	1	5
51	2	0	2	2	2	4	2	2	4	10
52	2	2	4	2	2	4	2	2	4	12
53	2	2	4	2	2	4	2	2	4	12
54	2	2	4	2	0	2	2	2	4	10
55	2	2	4	1	2	3	2	2	4	11
56	0	0	0	2	2	4	2	2	4	8
57	1	2	3	2	2	4	2	2	4	11
58	2	2	4	2	2	4	2	2	4	12
59	1	2	3	2	2	4	2	2	4	11
60	2	2	4	1	0	1	2	2	4	9
61	2	2	4	1	2	3	2	2	4	11
62	2	2	4	2	2	4	2	0	2	10

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

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	4	4	4	2	4	4	22	92	
3	0	4	4	0	4	4	16	67	CE
4	4	4	4	4	4	4	24	100	
5	4	4	4	4	4	4	24	100	
6	4	4	4	4	4	4	24	100	
7	3	4	3	3	4	3	20	83	
8	3	4	4	2	3	4	20	83	
9	4	4	4	1	3	4	20	83	
10	3	4	4	4	4	1	20	83	
11	3	4	4	4	4	4	23	96	
12	1	4	4	1	4	4	18	75	
13	4	4	4	0	4	4	20	83	
14	4	4	4	4	4	4	24	100	
15	4	4	4	1	4	4	21	88	
16	4	4	4	4	4	4	24	100	
17	3	4	4	4	3	4	22	92	
18	2	4	2	4	4	4	20	83	
19	4	4	4	1	4	4	21	88	
20	4	4	4	4	4	4	24	100	
21	4	4	4	4	4	4	24	100	
22	4	4	4	4	4	4	24	100	
23	4	4	4	4	4	4	24	100	
24	3	4	4	3	4	4	22	92	
25	3	4	4	3	1	4	19	79	
26	4	4	4	4	--	4	20	83	
27	4	4	4	1	4	4	21	88	
28	4	4	4	4	4	4	24	100	
29	4	4	4	1	4	4	21	88	
30	4	4	4	1	4	4	21	88	
31	2	4	4	4	4	4	22	92	
32	1	4	4	3	4	4	20	83	
33	4	4	4	1	4	4	21	88	

Lab n°	1	2	3	4	5	6	Cumulative score	Cumulative score (%)	Critical error
34	4	4	4	4	4	--	20	83	
35	4	4	4	4	4	4	24	100	
36	4	4	4	4	4	3	23	96	
37	4	4	4	1	4	4	21	88	
38	4	4	4	2	3	4	21	88	
39	4	4	4	0	4	4	20	83	
40	4	4	4	4	4	4	24	100	
41	4	4	4	4	4	4	24	100	
42	4	4	4	4	4	4	24	100	
43	3	4	4	4	4	4	23	96	
44	3	4	4	3	4	4	22	92	
45	3	4	4	3	4	4	22	92	
46	4	4	4	3	4	4	23	96	
47	1	4	4	4	4	4	21	88	
48	4	1	4	4	1	4	18	75	
49	3	4	4	3	4	4	22	92	
50	4	4	4	2	2	1	17	71	
51	4	4	4	2	4	4	22	92	
52	4	4	4	4	4	4	24	100	
53	3	4	4	4	4	4	23	96	
54	3	4	4	4	2	4	21	88	
55	1	4	4	4	3	4	20	83	
56	3	4	4	0	4	4	19	79	
57	3	4	4	3	4	4	22	92	
58	3	4	4	4	4	4	23	96	
59	4	4	4	3	4	4	23	96	
60	4	4	2	4	1	4	19	79	
61	1	4	4	4	3	4	20	83	
62	3	4	4	4	4	2	21	88	
63	4	4	4	0	0	0	12	50	CE
64	3	2	4	3	4	4	20	83	
65	3	4	4	3	4	4	22	92	
66	1	4	4	4	4	4	21	88	
67	2	3	4	4	4	4	21	88	

Lab n°	1	2	3	4	5	6	Cumulative score	Cumulative score (%)	Critical error
68	4	4	4	4	4	4	24	100	
69	1	4	4	4	3	4	20	83	
70	4	4	4	1	4	1	18	75	
71	2	4	4	2	4	4	20	83	
72	0	4	4	3	3	4	18	75	
73	4	4	4	2	4	4	22	92	
74	3	4	4	3	4	4	22	92	
75	3	4	4	4	1	4	20	83	
76	0	1	4	3	1	4	13	54	CE
77	4	4	4	3	2	4	21	88	
78	0	1	4	4	4	4	17	71	CE
79	1	4	4	1	1	4	15	62	
80	4	1	4	4	1	4	18	75	
81	4	4	4	4	4	4	24	100	
82	4	4	4	4	4	4	24	100	
83	3	4	4	4	0	4	19	79	CE
84	4	4	4	4	4	4	24	100	
85	4	4	4	4	4	4	24	100	
86	4	4	4	0	4	4	20	83	

Performance

	Number of labs	% total labs
Satisfactory performers (≥ 70 % of adequate responses)	77	90%
Unsatisfactory performers (< 70 % adequate responses and/or critical error)	6	7%
Partial and non-submitters	0	0%
Educational participants	2	2%
Scheme withdrawal	1	1%

Overall Proficiency

Sample	Diagnosis	Analytical (%)	Interpretation (%)	Total (%)
UMPS-NL-2025-A	MPS-IIIA	78	81	80
UMPS-NL-2025-B	Control subject	97	94	96
UMPS-NL-2025-C	MPS-I	99	98	99
UMPS-NL-2025-D	MPS-I	77	72	75
UMPS-NL-2025-E	MPS-IVA	88	85	87
UMPS-NL-2025-F	MPS-II	95	93	94

11. Tentative schedule for 2026

Sample distribution	10 February 2026
Start of analysis of Survey 2026-1. Website open	9 March 2026
Survey 2026-1 - Results submission	11 May 2026
Survey 2026-1 - Reports	22 June 2026
Start of analysis of Survey 2026-2	6 July 2026
Survey 2026-2 – Results submission	7 September 2026
Survey 2026-2 - Reports	19 October 2026
Annual Report 2026	January – March 2027

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, Suggestions and Complaints

If you have any questions, comments or suggestions please address to the Scientific Advisor of the scheme, Dr. H.C.M..T. (Berthil) Prinsen, 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/

Please read:

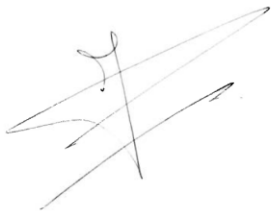
In order to achieve a satisfactory performance for the UMPS scheme, it is strongly recommended to perform both quantitative (total) GAG-analysis and GAG-subtyping for each sample. No points are rewarded when experiments are not performed.

The interpretative proficiency will be scored based on the submitted results of the analytical section. If a laboratory selects all possible diagnoses 1 mark will be subtracted.

In patients with MPS-I, II and VII DS and HS are elevated. In these patients DS is generally higher compared to HS. Therefore, the scientific advisors decided that the scoring criteria for MPS-I, II and VII are based on elevated DS (1 mark), while elevated HS is not scored. Ideally, 1 mark should be rewarded for both elevated DS and HS.

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 (preferable 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, 12-02-2026



Dr. H.C.M.T. Prinsen
UMC Utrecht
Dept of Genetics, section Metabolic Diagnostics
KC02.069.1
3584 CX Utrecht
The Netherlands

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

Version Number	Published	Amendments
1	12th February 2026	2025 annual report published

END