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Urine Mucopolysaccharides
Centre: The Netherlands

Final Report 2020

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

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1. Introduction

The ERNDIM Urine Mucopolysaccharides 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 organised by Erasmus Medical Centre (Rotterdam, NL) in conjunction with SKML, the Dutch organisation for quality assurance in medical laboratories (MCA laboratory, Winterswijk, NL) and CSCQ, the Swiss organisation for quality assurance in medical laboratories.

2. Geographical distribution of participants

In 2020 97 laboratories from many different countries have registered for the Urine MPS scheme. The number of participants is relatively stable over the years (2016: 99, 2017: 102, 2018:100, 2019: 96 participants). One laboratory was an educational participant in 2020 (2 in 2019). 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	4

[1] If these scheme instructions are not Version 1 for this scheme year, go to **Error! Reference source not found.** for details of the changes made since the last version of this document.

Country	Number of participants
Brazil	1
Canada	5
Chile	1
Colombia	1
Croatia	1
Cyprus	1
Czechia	1
Denmark	1
Estonia	1
France	7
Germany	8
Greece	1
Hong Kong	1
Italia	6
Kingdom of Saudi Arabia	1
Latvia	1
Malaysia	2
Mexico	1
Netherlands	3
New Zealand	2
Norway	1
Poland	1
Portugal	2
Republic of Korea	1
Serbia	1
Singapore	1
Slovakia	1
South Africa	2
Spain	4
Sweden	1
Switzerland	1
Taiwan	1
Turkey	2
United Kingdom	14
United States of America	6
Uruguay	1

3. Design and logistics of the scheme including sample information

The scheme has been designed and planned by dr George Ruijter as Scientific Advisor and coordinated by dr Xavier Albe (sub-contractor on behalf of CSCQ) and dr Cas Weykamp (sub-contractor on behalf of SKML) as scheme organisers, all appointed by and according to procedures laid down the ERNDIM Board.

SKML prepares lyophilised sample aliquots and dispatches UMPS EQA samples to the scheme participants by courier. CSCQ provides a website for on-line 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 2020-1, 2 and 3
	Round 2: samples 2020-4, 5 and 6

As usual, the samples used in 2020 were authentic human urine samples, 5 from MPS patients and 1 from a non-MPS individual. One sample was donated by Dr Kairit Joost, Tartu University hospital, Tartu, Estonia and one sample by Dr Eresha Jasinge, Lady Ridgeway Hospital for Children, Colombo, Sri Lanka. The other 4 samples were from the sample repository at Erasmus MC, Rotterdam, The Netherlands. Samples were selected by the Scientific Advisor and tested for suitability in the Scientific Advisor's laboratory (Erasmus Medical Centre, Rotterdam, 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.

Sample 2020.01	male, 39 y	Normal sample
Sample 2020.02	female, 7 y	MPS VII
Sample 2020.03	male, 44 y	MPS II, attenuated phenotype
Sample 2020.04	female, 27 y	MPS I, Scheie phenotype
Sample 2020.05	female, 5 y	MPS IV A
Sample 2020.06	male, 35 y	MPS II, attenuated phenotype

4. Tests

Test required for participation in the Urine MPS scheme are creatinine concentration and GAG analysis (quantitative total GAG and/or GAG sub fractions, either qualitative by electrophoresis/TLC or quantitative by LC-MS/MS). 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

- February 11, 2020: shipment of samples
- March 1, 2020: analysis start (survey 1)
- April 1, 2020: website available for result submission (survey 1)
- June 1, 2020: extended deadline (due to COVID-19) for result submission of survey 1
- July 24, 2020: interim report of survey 1 available for download
- August 1, 2020: analysis start (survey 2)
- September 1, 2020: website available for result submission (survey 2)
- September 28, 2020: deadline for result submission (survey 2)
- November 12, 2020: interim report of survey 2 available for download
- January 12, 2021: annual report with final scoring, confirmed by the SAB, available for download

6. Results submitted

88 out of the 97 labs that were registered returned results for both surveys. Due to COVID-19, results were submitted late by 5 participants for survey 1. Four participants submitted results 1-3 weeks after the original deadline. Another participant could not submit results online due to closure of the website and submitted results of survey 1 by email four weeks after the deadline of June 1st.

	Survey 1	**Survey 2**
Receipt of results	91	89
No report	6	8

7. Web site 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, LC-MS/MS test results). Unfortunately, not all participants have specified their methods.

- **When you are submitting results for the Urine MPS scheme, please specify the methods used by your laboratory to investigate MPS.**

In 2017 an evaluation program made by dr Albe from CSCQ was used for the first time 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 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. 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 (diiferential) 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 2020 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 is made in the Scientific Advisory Board (SAB) during the autumn meeting (November 19-20, 2020 for the 2020 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. 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.

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 2020, the SAB decided that samples 2020.03, 2020.04, 2020.05 and 2020.06 were eligible for critical error (details provided under item 9).

Score required for satisfactory performance: at least 15 points from the maximum of 24 (62%).
Please see note on score required for adequate performance in 2021 under item 11.

From the 96 regular (non-educational) participants 79 (82%) achieved satisfactory performance (2 reports submitted, score ≥15, no critical error). Seventeen participants did not accomplish satisfactory performance, including 7 due to incomplete submission of results (i.e. no report or 1 survey report submitted instead of 2 reports). The number of incomplete submissions is relatively high in 2020, which may be related to the COVID-19 pandemic.

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. Ten performance support letters will be sent by the Scheme Advisor for the 2020 scheme year. Any partial submitters or non-submitters will receive a letter from the ERNDIM Executive Administrator, Sara Gardner.

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.

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. Quantitative GAG results were evaluated separately for most methods (DMB, Alcian Blue, Harmine/carbazole, CPC/turbidity). Most participants use DMB (approx. 70 %) for quantitative total GAG analysis. The number of participants using other GAG screening methods is smaller.

Parameter/Method	**MPS 2020.01**	**MPS 2020.02**	**MPS 2020.03**	**MPS 2020.04**	**MPS 2020.05**	**MPS 2020.06**
Creatinine (mmol/L)						
Average	6,43	1,69	7,45	3,27	4,13	5,92
SD	0,39	0,22	0,39	0,24	0,27	0,30
Median	6,42	1,68	7,45	3,29	4,16	5,91
N	89	89	89	86	87	85
GAG quantitative (mg/mmol creat) DMB colorimetric method						
Average	2,3	19,1	16,8	19,4	30,2	24,8
SD	1,5	6,7	4,8	4,7	10,4	6,7
Median	1,9	18,0	16,2	19,4	30,0	24,8
N	60	61	60	58	59	58
GAG quantitative (mg/mmol creat) Alcian blue colorimetric tests						
Average	3,4	20,8	17,1	18,5	97,8	27,2
SD	1,3	7,7	3,3	4,0	182,4	5,8
Median	3,7	19,1	16,2	17,4	25,4	26,3
N	6	6	6	6	6	6
GAG quantitative (mg/mmol creat) CPC turbidity method						
Average	2,2	18,2	27,3	28,3	55,8	39,6
SD	1,6	1,8	3,7	16,0	29,4	14,9
Median	2,2	18,2	27,3	28,3	55,8	39,6
N	2	2	2	2	2	2
GAG quantitative (mg/mmol creat) Uronic acids - carbazole/harmine method						
Average	0,3	4,5	3,2	2,6	6,5	3,2
SD	0,1	0,1	0,2	2,5	1,2	2,8
Median	0,3	4,5	3,2	1,5	6,9	2,6
N	2	2	2	3	3	3

9.2. Your results

9.3. Sample 2020.01 – Normal sample

Patient details
Sample from a 39-year-old male not suffering from an MPS disorder.

Analytical performance
93% (80/86) of the participants reported a normal result of their quantitative GAG screening test (e.g. DMB test). From the 6 participants that reported abnormal screening results, 3 concluded that this was a normal sample after all, based on GAG subtype analysis. Most participants indeed reported normal test results of GAG electrophoresis, TLC or LC-MS/MS. Two labs reported elevated DS and three increased HS. None of the participants reported increased KS.

Diagnosis / Interpretative proficiency
As usual for normal samples, most participants (80/89; 90%) correctly concluded that this was not an MPS sample. Three participants concluded a mucopolysaccharidosis in this sample (see table below).
Overall proficiency (based on points): 91%

Diagnosis	N	%
Normal	76	85,4
No Diagnosis	6	6,7
MPS III	2	2,2
MPS VI/Normal	1	1,1
No Diagnosis/Not performed	1	1,1
MPS VI	1	1,1
No Diagnosis/Normal	1	1,1
MPS III/Normal	1	1,1
N results	89	100
N non-submitters	8	
N registered	97	

Scoring
- Analytical results: normal results: score 2.
- Interpretation: normal: two points. ‘No diagnosis/normal’ or 'normal, but MPS not excluded': 1 point
- Critical error. This sample was not considered eligible for critical error.

9.4. Sample 2020.02 – MPS VII

Patient details
7-year old female MPS VII patient. Diagnosis was confirmed by enzyme testing.

Analytical performance
A considerable number of participants observed elevated total GAG (76/86; 88%) in this sample. Many labs commented that total GAG was only mildly elevated. Theoretically, since DS, CS and HS all contain glucuronic acid residues; elevation of these 3 GAG species could be expected in an MPS VII urine sample. In fact, only DS and CS are usually reported elevated in MPS VII urine (see also Fig. 1) and the following observations were reported in sample 2020.02: elevated DS, 24%, and elevated CS, 29%. In addition, 17% reported increased HS, and 18% increased KS. The results reported in this sample seem to confirm that in particular DS and CS are increased in MPS VII urine, but the results are not very clear.
In some electrophoresis methods CS and KS are not well separated and it might be that some labs have misinterpreted the CS elevation in this sample as being increased KS.

Analytical proficiency was 63%.

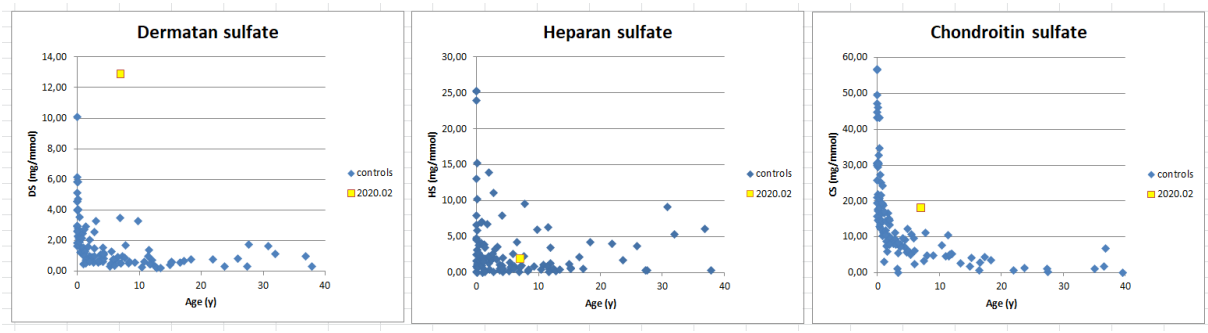

Figure 1. GAG analysed in sample 2020.02 by LC-MS/MS following methanolysis.

Diagnosis / Interpretative proficiency

Many different differential diagnoses were reported (see table below). Interestingly, MPS VII as a single possible diagnosis was reported by 7 participants. Altogether 25 participants included MPS VII in their differential diagnosis, but on the other hand, 24 participants concluded that this was a normal sample, i.e. not an MPS, and 12 participants reported 'no diagnosis'. Interpretative proficiency was 38%, i.e. much lower than analytical proficiency, which reflects subtle abnormalities observed upon GAG subtyping. In addition, quite a few participants may have no experience with MPS VII samples and perhaps do not include it in their differential diagnosis. This sample demonstrates that in the case of elevated CS, MPS VII must be considered.

Overall proficiency (based on points): 50%

This sample appeared to be particularly challenging with an overall proficiency of 50%. Because of the low proficiency, the Scientific Advisory board has discussed during its Autumn meeting whether this sample should be excluded from performance assessment. Finally, it was decided to score sample 2020.02, since including/excluding it from scoring did not affect performance assessment of any participant.

As mentioned in the 2018 annual report, 10 labs report results obtained by LC-MS/MS. While overall proficiency of sample 2020.02 was 50%, the proficiency of the 10 participants using LC-MS/MS methods was 95% for this sample. This striking difference may be partly explained by the power of LC-MS/MS analysis to detect GAG abnormalities.

Diagnosis	N	%
Normal	25	28,1
MPS IV	13	14,6
No Diagnosis	12	13,5
MPS VII	7	7,9
MPS III	5	5,6
MPS VI	4	4,5
MPS VI/MPS VII	4	4,5
MPS I	3	3,4
MPS I/MPS VII	2	2,2
MPS VII/Normal	2	2,2
MPS VII/No Diagnosis	1	1,1
MPS IV/No Diagnosis	1	1,1
MPS I/MPS II/MPS VII	1	1,1
No Diagnosis/Normal	1	1,1
MPS VII/To be entered	1	1,1
MPS I/MPS II/MPS III/MPS VI/MPS VII	1	1,1
MPS III/MPS VII	1	1,1
MPS IV/MPS VII	1	1,1

Diagnosis	N	%
MPS I/MPS II/MPS VI	1	1,1
MPS I/MPS II/MPS VI/MPS VII	1	1,1
MPS III/Normal	1	1,1
MPS IV/Normal	1	1,1
N results	89	100
N non-submitters	8	
N registered	97	

Scoring

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

9.5. Sample 2020.03 – MPS II

Patient details
This was an MPS II sample from an adult patient (44 y) not receiving ERT treatment.

Analytical performance
Clearly abnormal sample with strongly elevated total GAG for age. All but one of the 86 participants that submitted results of total GAG screening in this sample reported an elevated concentration (99%). The single lab that reported 'normal' for quantitative GAG screening commented that their adult reference range had not been developed yet, but that the GAG value appeared to be elevated for the patient's age. 98% of the participants (84/86) reported elevated DS, while increased HS was reported by 74% of the participants.

Diagnosis / Interpretative proficiency
The majority of the labs reported MPS I/II (VII) as the most likely diagnosis (50; see table below), while another 27 included MPS VI in the differential diagnosis (various combinations of MPS II with MPS I, VI and VII all scored 2). Three participants specifically reported MPS II as the diagnosis. In total, 77 labs (88%) mentioned MPS II among the various differential diagnoses. MPS I and/or MPS VI were reported by 6 laboratories. One participant reported 'normal' for this sample (critical error).
From the 27 participants that reported MPS I, II, VI (and VII) as a differential diagnosis, 10 fouind HS elevated. Increased HS is not expected in MPS VI urines, except in patients with a severe phenotype. Apparently, these labs do not differentiate between MPS I, II and VI.
Overall proficiency was 92%.

Diagnosis	N	%
MPS I/MPS II	26	29,5
MPS I/MPS II/MPS VII	21	23,9
MPS I/MPS II/MPS VI/MPS VII	16	18,2
MPS I/MPS II/MPS VI	11	12,5
MPS VI	3	3,4
MPS II	3	3,4
MPS I/MPS VI	2	2,3
MPS III	2	2,3
MPS VII	1	1,1
Normal	1	1,1
No Diagnosis	1	1,1

Diagnosis	N	%
MPS I	1	1,1
N results	88	100
N non-submitters	9	
N registered	97	

Scoring

- Analytical results: elevated (total) GAG and elevated DS were each scored 1 mark.
- Interpretation: MPS II with MPS I, VI or VII in various combinations: two points. MPS I and/or VI (and VII): 1 point
- Critical error: diagnosis 'normal' (n=1)

This sample was also circulated in 2015 (sample 2015.01).

9.6. Sample 2020.04 – MPS I (Scheie phenotype)

Patient details
MPS I, Scheie phenotype, female aged 27 y not receiving ERT when the urine sample was collected.

Analytical performance
In this sample 95% (80/84) of the participants reported increased total GAG and increased DS. Only 54% (45/84) of the participants reported increased HS in this sample. This confirms previous results in other MPS I samples and suggests that HS is generally less elevated in MPS I urine samples compared to MPS II samples (e.g. 2020.03 and 2020.06, both obtained from adult patients).

Diagnosis / Interpretative proficiency
In total 65 participants (75%) reported a differential diagnosis including MPS I in various combinations with MPS II, VI and VII. In total 12 participants concluded MPS VI (and VII), i.e. without mentioning MPS I, which was marked 1. This is explained by the relatively low number of labs that detected increased HS (54%), as described above. In previous years other MPS I samples (e.g. 2018.01 and 2016.03) gave similar results and this indicates the difficulty to distinguish MPS I from MPS VI on the basis of urine mucopolysaccharides analysis with traditional methods for GAG subtype analysis (electrophoresis, TLC).
Overall proficiency (based on points): 88%

Diagnosis	N	%
MPS I/MPS II/MPS VI/MPS VII	15	17,2
MPS I/MPS II/MPS VI	12	13,8
MPS VI	11	12,6
MPS I	9	10,3
MPS I/MPS VI	8	9,2
MPS I/MPS II/MPS VII	7	8,0
MPS I/MPS II	6	6,9
MPS I/MPS VII	5	5,7
MPS III	3	3,4
MPS I/MPS VI/MPS VII	3	3,4
No Diagnosis	3	3,4
MPS IV	1	1,1
MPS VII	1	1,1
MPS I/MPS IV/MPS VI	1	1,1
MPS I/MPS III	1	1,1

Diagnosis	N	%
MPS VI/MPS VII	1	1,1
N results	87	100
N non-submitters	9	
N registered	96	

Scoring

- Analytical results: elevated (total) GAG and elevated DS were each scored 1 mark.
- Interpretation: MPS I with MPS II, VI or VII in various combinations: two points. MPS VI (and VII): 1 point
- Critical error. Reporting 'normal' as the most likely diagnosis was considered a critical error in this sample (n=0).

9.7. Sample 2020.05 – MPS IV A

Patient details
Sample obtained from an MPS IV A patient with a severe phenotype.

Analytical performance
Abnormal GAG screening results were reported by 82/84 participants (98%). From the 77 participants that submitted a result for KS, 60 (78%) reported an elevated level of this GAG species. N-acetyl-galactosamine 6-sulfatase (galactose 6-sufatase) deficiency in MPS IV A may lead to storage of chondroitin-6-sulfate and indeed 36 labs (46%) reported elevated chondroitin sulfate.

Diagnosis / Interpretative proficiency
MPS IV was reported as the most likely diagnosis by 64 participants. Four participants reported MPS IV in combination with normal/no diagnosis. Four participants concluded this was a normal sample (critical error). Overall proficiency was 79%, which is similar to a previous sample from an MPS IV A patient (2019.03; overall proficiency 82%). The MPS IV A samples circulated in 2019 and 2020 were both obtained from patients with a severe phenotype. Overall proficiency in these years (79%, 82%) is much higher than achieved with MPS IV A samples obtained from patients with a more attenuated phenotype that were circulated in 2013, 2015 and 2017 with proficiencies of 61-65%. This is in line with the idea that attenuated MPS IV A is easily missed in urine MSP analysis.
Overall proficiency was 79%.

Diagnosis	N	%
MPS IV	64	73,6
Normal	3	3,4
MPS III	3	3,4
MPS IV/MPS VII/No Diagnosis	2	2,3
MPS VI	2	2,3
MPS IV/No Diagnosis	1	1,1
MPS VI/MPS VII	1	1,1
MPS IV/MPS VI	1	1,1
MPS III/MPS IV/MPS VI	1	1,1
MPS I/MPS II/MPS VII	1	1,1
MPS IV/MPS VII	1	1,1
MPS I/MPS II/MPS VI/MPS VII	1	1,1
No Diagnosis/Normal	1	1,1
MPS VII/No Diagnosis	1	1,1
No Diagnosis	1	1,1

Diagnosis	N	%
MPS I/MPS VI/MPS VII	1	1,1
MPS IV/MPS VII/Normal	1	1,1
MPS VII/Normal	1	1,1
N results	87	100
N non-submitters	9	
N registered	96	

Scoring

- Analytical results: elevated (total) GAG and elevated KS were each scored 1 mark.
- Interpretation: MPS IV: score 2. MPS IV/normal: 1 point
- Critical error: Reporting 'normal' as the most likely diagnosis was considered a critical error in this sample (n=4).

9.8. Sample 2020.06 – MPS II

Patient details

A sample from an adult MPS II patient (42 y) not receiving ERT treatment.

Analytical performance

From the 84 participants that submitted results of total GAG screening in this sample 82 (98%) reported elevated GAG concentration. Also, most labs reported abnormal test results of GAG electrophoresis or TLC: 94% (77/82) reported elevated DS, while 77% (63/82) found elevated HS. These results are similar to the other MPS II sample circulated this year (2020.03)

Diagnosis / Interpretative proficiency

The majority of the labs reported MPS I/II (VII) as the most likely diagnosis (50/87; 57%), while another 28 (32%) included MPS VI in the differential diagnosis. In total, 78 (90%) mentioned MPS II among the correct possible diagnoses. None of the participants reported 'normal' as the most likely diagnosis in this sample.

Overall proficiency (based on points) 92%.

Diagnosis	N	%
MPS I/MPS II	29	33,3
MPS I/MPS II/MPS VI/MPS VII	19	21,8
MPS I/MPS II/MPS VII	15	17,2
MPS I/MPS II/MPS VI	9	10,3
MPS II	6	6,9
MPS IV	2	2,3
No Diagnosis	2	2,3
MPS I/MPS III/MPS VII	1	1,1
MPS I/MPS II/MPS IV/MPS VI	1	1,1
MPS VI	1	1,1
MPS I/MPS IV/MPS VI	1	1,1
MPS I/MPS III	1	1,1
N results	87	100
N non-submitters	9	
N registered	96	

Scoring

- Analytical results: elevated (total) GAG and elevated DS were each scored 1 mark.
- Interpretation: MPS II with MPS I, VI or VII in various combinations: two points. MPS I and/or VI (and VII): 1 point
- Critical error. Reporting ‘normal’ as the most likely diagnosis was considered a critical error in this sample (n=0).

This sample was also circulated in 2016 (sample 2016.01). The proficiencies of the 2 different MPS II samples included in the 2020 surveys (2020.03 and 2020.06) were very similar: both 92%.

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.

Detailed scores – **Round 1**

Lab n°	Sample 1 Normal sample			Sample 2 MPS VII			Sample 3 MPS II			
	A	I	Total	A	I	Total	A	I	Total	Total
1	2	1	3	1	0	1	2	2	4	8
2	2	2	4	1	2	3	2	1	3	10
3	2	2	4	2	1	3	2	2	4	11
4	2	2	4	1	2	3	2	2	4	11
5	2	2	4	0	0	0	2	2	4	8
6	2	2	4	2	1	3	2	2	4	11
7	2	2	4	2	2	4	2	2	4	12
8	2	2	4	1	2	3	2	2	4	11
9	2	2	4	2	2	4	2	2	4	12
10	2	2	4	2	0	2	2	1	3	9
11	--	--	--	--	--	--	--	--	--	0
12	2	2	4	1	0	1	2	2	4	9
13	2	2	4	2	1	3	2	1	3	10
14	2	2	4	1	0	1	2	2	4	9
15	2	2	4	2	2	4	2	2	4	12
16	2	2	4	1	0	1	2	2	4	9
17	2	2	4	2	1	3	2	0	2	9
18	2	2	4	1	2	3	2	2	4	11
19	2	2	4	2	2	4	2	2	4	12
20	2	2	4	2	2	4	2	1	3	11
21	2	2	4	2	2	4	2	2	4	12
22	2	2	4	2	1	3	2	2	4	11
23	2	2	4	1	2	3	2	2	4	11
24	1	0	1	2	1	3	1	0	1	5
25	2	2	4	1	0	1	2	2	4	9
26	2	2	4	1	0	1	2	2	4	9
27	2	2	4	1	0	1	2	2	4	9

Lab n°	Sample 1 Normal sample			Sample 2 MPS VII			Sample 3 MPS II			
	A	I	Total	A	I	Total	A	I	Total	Total
28	2	2	4	1	1	2	2	2	4	10
29	2	2	4	2	1	3	2	2	4	11
30	2	2	4	1	1	2	2	2	4	10
31	2	2	4	1	1	2	2	2	4	10
32	2	2	4	1	0	1	2	2	4	9
33	--	--	--	--	--	--	--	--	--	0
34	2	2	4	1	0	1	2	2	4	9
35	2	2	4	2	1	3	2	2	4	11
36	2	2	4	1	1	2	2	2	4	10
37	1	0	1	2	0	2	2	2	4	7
38	2	2	4	2	2	4	2	2	4	12
39	2	2	4	0	0	0	2	2	4	8
40	2	2	4	0	0	0	2	2	4	8
41	2	2	4	0	0	0	2	2	4	8
42	2	2	4	2	1	3	2	2	4	11
43	2	2	4	0	0	0	2	2	4	8
44	2	2	4	2	2	4	2	2	4	12
45	2	2	4	1	0	1	2	2	4	9
46	2	0	2	1	0	1	2	2	4	7
47	2	2	4	1	0	1	2	2	4	9
48	2	2	4	1	0	1	2	2	4	9
49	1	1	2	2	0	2	2	2	4	8
50	0	1	1	1	0	1	1	0	1	3
51	2	2	4	1	2	3	2	2	4	11
52	2	0	2	1	0	1	2	2	4	7
53	1	0	1	0	0	0	2	2	4	5
54	2	2	4	2	2	4	2	2	4	12
55	1	1	2	1	0	1	2	2	4	7
56	2	2	4	2	1	3	2	2	4	11
57	2	2	4	1	0	1	2	2	4	9
58	2	2	4	2	0	2	2	2	4	10
59	1	2	3	2	2	4	2	2	4	11
60	2	2	4	1	0	1	2	2	4	9

Lab n°	Sample 1 Normal sample			Sample 2 MPS VII			Sample 3 MPS II			
	A	I	Total	A	I	Total	A	I	Total	Total
61	2	2	4	2	0	2	2	2	4	10
62	2	2	4	2	2	4	2	1	3	11
63	--	--	--	--	--	--	--	--	--	0
64	2	2	4	2	2	4	2	2	4	12
65	--	--	--	--	--	--	--	--	--	0
66	2	2	4	1	0	1	2	2	4	9
67	2	2	4	0	0	0	2	2	4	8
68	2	2	4	1	1	2	2	2	4	10
69	2	2	4	1	1	2	2	2	4	10
70	2	2	4	1	0	1	2	2	4	9
71	2	2	4	0	0	0	2	2	4	8
72	2	2	4	2	2	4	2	1	3	11
73	2	2	4	1	0	1	2	2	4	9
74	2	2	4	1	0	1	2	2	4	9
75	0	1	1	2	2	4	2	2	4	9
76	1	2	3	1	0	1	2	2	4	8
77	2	2	4	2	1	3	2	2	4	11
78	2	2	4	2	2	4	2	2	4	12
79	0	0	0	0	0	0	2	2	4	4
80	2	2	4	2	2	4	2	2	4	12
81	2	2	4	1	2	3	2	2	4	11
82	2	2	4	1	0	1	2	2	4	9
83	2	2	4	0	1	1	2	2	4	9
84	1	0	1	2	1	3	2	2	4	8
85	2	2	4	1	0	1	2	2	4	9
86	2	2	4	1	0	1	1	0	1	6
87	2	2	4	0	0	0	1	0	1	5
88	2	2	4	2	1	3	2	2	4	11
89	2	2	4	1	1	2	2	2	4	10
90	2	2	4	0	0	0	2	2	4	8
91	2	2	4	1	0	1	2	2	4	9
92	2	2	4	2	2	4	2	2	4	12
93	2	2	4	2	1	3	1	0	1	8

Lab n°	Sample 1 Normal sample			Sample 2 MPS VII			Sample 3 MPS II			
	A	**I**	**Total**	**A**	**I**	**Total**	**A**	**I**	**Total**	**Total**
94	2	2	4	1	0	1	1	0	1	6
95	2	2	4	0	0	0	2	2	4	8
96	--	--	--	--	--	--	--	--	--	0
97	--	--	--	--	--	--	--	--	--	0

Detailed scores – Round 2

Lab n°	Sample 4 MPS I (Scheie phenotype)			Sample 5 MPS IV A			Sample 6 MPS II			
	A	I	Total	A	I	Total	A	I	Total	Total
1	2	2	4	2	2	4	2	2	4	12
2	1	0	1	2	2	4	2	2	4	9
3	2	2	4	2	1	3	2	2	4	11
4	--	--	--	--	--	--	--	--	--	0
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	1	0	1	1	2	3	8
8	2	1	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	2	2	4	1	0	1	2	2	4	9
12	2	2	4	1	0	1	2	2	4	9
13	2	2	4	2	2	4	2	2	4	12
14	1	1	2	1	2	3	1	0	1	6
15	2	2	4	2	2	4	2	2	4	12
16	2	2	4	2	2	4	2	2	4	12
17	2	1	3	2	2	4	2	2	4	11
18	2	2	4	2	2	4	2	2	4	12
19	2	2	4	1	0	1	2	2	4	9
20	2	2	4	1	2	3	2	2	4	11
21	2	2	4	2	2	4	2	2	4	12
22	2	1	3	1	2	3	2	2	4	10
23	2	2	4	2	2	4	2	2	4	12
24	2	2	4	1	0	1	2	2	4	9
25	2	2	4	1	1	2	2	2	4	10
26	2	2	4	2	2	4	2	2	4	12
27	2	1	3	2	2	4	2	2	4	11
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 4 MPS I (Scheie phenotype)			Sample 5 MPS IV A			Sample 6 MPS II			
	A	I	Total	A	I	Total	A	I	Total	Total
31	2	2	4	1	0	1	2	2	4	9
32	2	2	4	1	0	1	2	2	4	9
33	--	--	--	--	--	--	--	--	--	0
34	2	2	4	2	2	4	2	2	4	12
35	2	1	3	2	2	4	2	2	4	11
36	--	--	--	--	--	--	--	--	--	0
37	2	2	4	2	2	4	2	2	4	12
38	2	2	4	2	2	4	2	2	4	12
39	2	1	3	2	2	4	2	2	4	11
40	2	2	4	2	2	4	2	2	4	12
41	2	2	4	1	0	1	2	2	4	9
42	2	2	4	2	2	4	2	2	4	12
43	2	2	4	2	2	4	2	2	4	12
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	2	2	4	12
48	2	2	4	2	2	4	2	2	4	12
49	2	2	4	2	2	4	2	2	4	12
50	--	--	--	--	--	--	--	--	--	0
51	2	2	4	2	2	4	2	2	4	12
52	2	2	4	2	2	4	2	2	4	12
53	1	0	1	1	0	1	2	2	4	6
54	2	2	4	2	2	4	2	2	4	12
55	2	2	4	2	2	4	2	2	4	12
56	2	2	4	2	2	4	2	2	4	12
57	2	2	4	2	2	4	2	2	4	12
58	2	2	4	2	2	4	2	2	4	12
59	2	2	4	1	0	1	2	2	4	9
60	2	2	4	1	2	3	2	2	4	11
61	2	2	4	2	2	4	2	2	4	12
62	2	2	4	1	0	1	1	0	1	6
63	--	--	--	--	--	--	--	--	--	0

Lab n°	Sample 4 MPS I (Scheie phenotype)			Sample 5 MPS IV A			Sample 6 MPS II			
	A	I	Total	A	I	Total	A	I	Total	Total
64	2	2	4	2	2	4	2	2	4	12
65	--	--	--	--	--	--	--	--	--	0
66	2	1	3	1	1	2	2	2	4	9
67	2	1	3	2	2	4	2	2	4	11
68	2	2	4	1	1	2	2	2	4	10
69	2	2	4	2	2	4	1	2	3	11
70	--	--	--	--	--	--	--	--	--	0
71	2	2	4	2	2	4	1	2	3	11
72	2	2	4	2	2	4	2	2	4	12
73	2	1	3	2	2	4	2	2	4	11
74	2	2	4	2	2	4	2	2	4	12
75	0	0	0	1	2	3	2	2	4	7
76	2	2	4	2	2	4	2	2	4	12
77	2	1	3	2	1	3	2	0	2	8
78	2	2	4	2	2	4	2	2	4	12
79	2	2	4	2	2	4	2	2	4	12
80	2	2	4	1	0	1	1	0	1	6
81	2	1	3	2	2	4	2	2	4	11
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	1	3	2	2	4	2	2	4	11
86	1	0	1	1	0	1	1	0	1	3
87	1	0	1	1	0	1	1	0	1	3
88	2	1	3	1	0	1	2	2	4	8
89	2	2	4	2	2	4	2	2	4	12
90	2	2	4	2	2	4	2	2	4	12
91	2	1	3	2	2	4	2	1	3	10
92	2	2	4	1	2	3	2	2	4	11
93	1	0	1	1	0	1	1	0	1	3
94	1	0	1	1	0	1	1	0	1	3
95	2	2	4	1	1	2	2	2	4	10
96	1	0	1	1	0	1	1	0	1	3

Lab n°	Sample 4 MPS I (Scheie phenotype)			Sample 5 MPS IV A			Sample 6 MPS II			
	A	**I**	**Total**	**A**	**I**	**Total**	**A**	**I**	**Total**	**Total**
97	--	--	--	--	--	--	--	--	--	0

Total scores

Lab n°	1	2	3	4	5	6	Cumulative score	Cumulative score (%)	Critical error
1	3	1	4	4	4	4	20	83	
2	4	3	3	1	4	4	19	79	
3	4	3	4	4	3	4	22	92	
4	4	3	4	--	--	--	11	46	
5	4	0	4	4	4	4	20	83	
6	4	3	4	4	4	4	23	96	
7	4	4	4	4	1	3	20	83	
8	4	3	4	3	4	4	22	92	
9	4	4	4	4	4	4	24	100	
10	4	2	3	3	4	4	20	83	
11	--	--	--	4	1	4	9	38	
12	4	1	4	4	1	4	18	75	
13	4	3	3	4	4	4	22	92	
14	4	1	4	2	3	1	15	62	
15	4	4	4	4	4	4	24	100	
16	4	1	4	4	4	4	21	88	
17	4	3	2	3	4	4	20	83	
18	4	3	4	4	4	4	23	96	
19	4	4	4	4	1	4	21	88	
20	4	4	3	4	3	4	22	92	
21	4	4	4	4	4	4	24	100	
22	4	3	4	3	3	4	21	88	
23	4	3	4	4	4	4	23	96	
24	1	3	1	4	1	4	14	58	
25	4	1	4	4	2	4	19	79	
26	4	1	4	4	4	4	21	88	
27	4	1	4	3	4	4	20	83	
28	4	2	4	4	4	4	22	92	
29	4	3	4	4	4	4	23	96	
30	4	2	4	4	4	4	22	92	
31	4	2	4	4	1	4	19	79	
32	4	1	4	4	1	4	18	75	

Lab n°	1	2	3	4	5	6	Cumulative score	Cumulative score (%)	Critical error
33	--	--	--	--	--	--	0	0	
34	4	1	4	4	4	4	21	88	
35	4	3	4	3	4	4	22	92	
36	4	2	4	--	--	--	10	42	
37	1	2	4	4	4	4	19	79	
38	4	4	4	4	4	4	24	100	
39	4	0	4	3	4	4	19	79	
40	4	0	4	4	4	4	20	83	
41	4	0	4	4	1	4	17	71	
42	4	3	4	4	4	4	23	96	
43	4	0	4	4	4	4	20	83	
44	4	4	4	4	4	4	24	100	
45	4	1	4	4	4	4	21	88	
46	2	1	4	4	4	4	19	79	
47	4	1	4	4	4	4	21	88	
48	4	1	4	4	4	4	21	88	
49	2	2	4	4	4	4	20	83	
50	1	1	1	--	--	--	3	12	
51	4	3	4	4	4	4	23	96	
52	2	1	4	4	4	4	19	79	
53	1	0	4	1	1	4	11	46	
54	4	4	4	4	4	4	24	100	
55	2	1	4	4	4	4	19	79	
56	4	3	4	4	4	4	23	96	
57	4	1	4	4	4	4	21	88	
58	4	2	4	4	4	4	22	92	
59	3	4	4	4	1	4	20	83	
60	4	1	4	4	3	4	20	83	
61	4	2	4	4	4	4	22	92	
62	4	4	3	4	1	1	17	71	
63	--	--	--	--	--	--	0	0	
64	4	4	4	4	4	4	24	100	
65	--	--	--	--	--	--	0	0	
66	4	1	4	3	2	4	18	75	

Lab n°	1	2	3	4	5	6	Cumulative score	Cumulative score (%)	Critical error
67	4	0	4	3	4	4	19	79	
68	4	2	4	4	2	4	20	83	
69	4	2	4	4	4	3	21	88	
70	4	1	4	--	--	--	9	38	
71	4	0	4	4	4	3	19	79	
72	4	4	3	4	4	4	23	96	
73	4	1	4	3	4	4	20	83	
74	4	1	4	4	4	4	21	88	
75	1	4	4	0	3	4	16	67	
76	3	1	4	4	4	4	20	83	
77	4	3	4	3	3	2	19	79	
78	4	4	4	4	4	4	24	100	
79	0	0	4	4	4	4	16	67	
80	4	4	4	4	1	1	18	75	
81	4	3	4	3	4	4	22	92	
82	4	1	4	4	4	4	21	88	
83	4	1	4	4	0	4	17	71	
84	1	3	4	4	4	4	20	83	
85	4	1	4	3	4	4	20	83	
86	4	1	1	1	1	1	9	38	
87	4	0	1	1	1	1	8	33	
88	4	3	4	3	1	4	19	79	
89	4	2	4	4	4	4	22	92	
90	4	0	4	4	4	4	20	83	
91	4	1	4	3	4	3	19	79	
92	4	4	4	4	3	4	23	96	
93	4	3	1	1	1	1	11	46	
94	4	1	1	1	1	1	9	38	
95	4	0	4	4	2	4	18	75	
96	--	--	--	1	1	1	3	12	
97	--	--	--	--	--	--	0	0	

Overall Proficiency

Sample	Diagnosis	Analytical (%)	Interpretation (%)	Total (%)
MPS 2020.01	Normal sample	92	90	91
MPS 2020.02	MPS VII	63	38	50
MPS 2020.03	MPS II	96	89	92
MPS 2020.04	MPS I (Scheie phenotype)	94	83	88
MPS 2020.05	MPS IV A	83	75	79
MPS 2020.06	MPS II	94	89	92

11. Tentative schedule for 2021

Sample distribution	February 2021
Start of analysis of Survey 2021-1; Website open	March 29, 2021
Survey 2021-1 - Results submission deadline	April 26, 2021
Survey 2021-1 – Interim reports available	May-June 2021
Start of analysis of Survey 2021-2; Website open	August 30, 2021
Survey 2021-2 – Results submission deadline	September 27, 2021
Survey 2021-2 – Interim reports available	October-November 2021
Annual Report 2021 available	December 2021 - January 2022

In line with other interpretative ERNDIM schemes, the Scientific Advisory Board (SAB) is planning to raise the score required for adequate performance in the Urine MPS scheme from 15 points (62%) to 17 points (71%). This change will be implemented in the 2021 survey. Please note that each year the SAB will review the minimal score required in its autumn meeting and that the minimal score required may be adjusted depending on the overall results achieved.

Date of report, 2021-03-08

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