



ERNDIM Urine MPS

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Urine Mucopolysaccharides Interim survey report 17-03-MPS

Report prepared by Dr. G.J.G. Ruijter

Laboratory N°: 392 (ERNDIM Urine MPS)

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Date of survey deadline: 2017-05-01

Date of report: 23 juni 2017

Results of samples and evaluation of reporting

17-03-MPS Sample : MPS 2017.01

Lab 392

Patient information: M 18.0 year

Total Score : 4

Diagnosis: MPS IIIA attenuated phenotype

Comments on proficiency:

This sample of a mild MPS IIIA patient was challenging with a proficiency of 57%. MPS III was reported by 56 participants, while 28 considered this as a normal urine. Four participants concluded MPS III or normal.

Creatinine (mmol/L)

N	96
Average	7,97
SD	0,45
Median	8,01
Your result	7.25
Z-Score	-1,60

GAG quantitative (mg/mmol creat)

Method	DMB colorimetric
N	67
Average	5,95
SD	2,13
Median	6,00
Your result	8.51
Z-Score	1,20

GAG excretion according to reference values

N registred	102
N results	95
Normal (%)	48 (51 %)
Increased (%)	47 (49 %)
Not detected	-- --
Your result	Increased

Score : 1

Scientific advisor comment:

Results of samples and evaluation of reporting

17-03-MPS Sample : MPS 2017.01

Lab 392

Patient information: M 18.0 year

Total Score : 4

Diagnosis: MPS IIIA attenuated phenotype

Chondroitinsulfate

N registred	102
N results	84
Normal (%)	69 (82 %)
Increased (%)	3 (4 %)
Not detected	12 (14 %)
Your result	Normal

Dermatansulfate

N registred	102
N results	89
Normal (%)	30 (34 %)
Increased (%)	5 (6 %)
Not detected	54 (61 %)
Your result	Not detected

Heparansulfate

N registred	102
N results	90
Normal (%)	12 (13 %)
Increased (%)	58 (64 %)
Not detected	20 (22 %)
Your result	Increased

Keratansulfate

N registred	102
N results	81
Normal (%)	23 (28 %)
Increased (%)	1 (1 %)
Not detected	57 (70 %)
Your result	Not detected

Score : 1

Scientific advisor comment:

Diagnosis

N registred = 102

N results = 97

MPS III	56 (58 %)
Normal	28 (29 %)
No Diagnosis	7 (7 %)
MPS IV	2 (2 %)
MPS I/MPS II/MPS VII	1 (1 %)
MPS I/MPS II	1 (1 %)
MPS VI	1 (1 %)
MPS I/MPS II/MPS III/MPS VI	1 (1 %)

Your result: MPS III

Score : 2

Scientific advisor comment:

Results of samples and evaluation of reporting

17-03-MPS Sample : MPS 2017.02

Lab 392

Patient information: M 3.0 year

Total Score : 4

Diagnosis: MPS II

Comments on proficiency:

A straightforward MPS II sample with high GAG concentration. Total proficiency was 86%

Creatinine (mmol/L)

N	97
Average	2,17
SD	0,19
Median	2,20
Your result	1.86
Z-Score	-1,61

GAG quantitative (mg/mmol creat)

Method	DMB colorimetric
N	68
Average	51,64
SD	16,38
Median	53,96
Your result	43.06
Z-Score	-0,52

GAG excretion according to reference values

N registred	102
N results	95
Normal (%)	3 (3 %)
Increased (%)	92 (97 %)
Not detected	-- --
Your result	Increased

Score : 1

Scientific advisor comment:

Results of samples and evaluation of reporting

17-03-MPS Sample : MPS 2017.02

Lab 392

Patient information: M 3.0 year

Total Score : 4

Diagnosis: MPS II

Chondroitinsulfate

N registred	102
N results	84
Normal (%)	65 (77 %)
Increased (%)	16 (19 %)
Not detected	3 (4 %)
Your result	Increased

Dermatansulfate

N registred	102
N results	94
Normal (%)	3 (3 %)
Increased (%)	90 (96 %)
Not detected	1 (1 %)
Your result	Increased

Heparansulfate

N registred	102
N results	92
Normal (%)	15 (16 %)
Increased (%)	74 (80 %)
Not detected	3 (3 %)
Your result	Increased

Keratansulfate

N registred	102
N results	80
Normal (%)	22 (28 %)
Increased (%)	3 (4 %)
Not detected	55 (69 %)
Your result	Not detected

Score : 1

Scientific advisor comment:

Diagnosis

N registred = 102

N results = 96

MPS I/MPS II	32 (33 %)
MPS I/MPS II/MPS VII	28 (29 %)
MPS I/MPS II/MPS VI/MPS VII	11 (11 %)
MPS I/MPS II/MPS VI	10 (10 %)
MPS II	4 (4 %)
MPS VI	2 (2 %)
MPS III	2 (2 %)
MPS II/MPS VII	1 (1 %)
MPS VII	1 (1 %)
MPS I/MPS II/MPS III/MPS VI	1 (1 %)
MPS IV	1 (1 %)
MPS I/MPS II/MPS III/MPS VI/MPS VII	1 (1 %)
No Diagnosis	1 (1 %)
Normal	1 (1 %)

Your result: MPS II

Results of samples and evaluation of reporting

17-03-MPS Sample : MPS 2017.02

Lab 392

Patient information: M 3.0 year

Total Score : 4

Diagnosis: MPS II

Score : 2

Scientific advisor comment:

Results of samples and evaluation of reporting

17-03-MPS Sample : MPS 2017.03

Lab 392

Patient information: M 46.0 year

Total Score : 4

Diagnosis: MPS II attenuated

Comments on proficiency:

The proficiency, 85%, of this sample from an MPS II patient with an attenuated phenotype was almost identical to sample 2017.02, which was from a severe MPS II patient.

Creatinine (mmol/L)

N	96
Average	6,30
SD	0,48
Median	6,34
Your result	5.66
Z-Score	-1,33

GAG quantitative (mg/mmol creat)

Method	DMB colorimetric
N	67
Average	23,27
SD	6,20
Median	24,00
Your result	20.8
Z-Score	-0,40

GAG excretion according to reference values

N registred	102
N results	94
Normal (%)	0 (0 %)
Increased (%)	94 (100 %)
Not detected	-- --
Your result	Increased

Score : 1

Scientific advisor comment:

Results of samples and evaluation of reporting

17-03-MPS Sample : MPS 2017.03

Lab 392

Patient information: M 46.0 year

Total Score : 4

Diagnosis: MPS II attenuated

Chondroitinsulfate

N registred	102
N results	85
Normal (%)	66 (78 %)
Increased (%)	4 (5 %)
Not detected	15 (18 %)
Your result	Normal

Dermatansulfate

N registred	102
N results	94
Normal (%)	1 (1 %)
Increased (%)	92 (98 %)
Not detected	1 (1 %)
Your result	Increased

Heparansulfate

N registred	102
N results	92
Normal (%)	21 (23 %)
Increased (%)	65 (71 %)
Not detected	6 (7 %)
Your result	Increased

Keratansulfate

N registred	102
N results	79
Normal (%)	21 (27 %)
Increased (%)	1 (1 %)
Not detected	57 (72 %)
Your result	Not detected

Score : 1

Scientific advisor comment:

Diagnosis

N registred = 102

N results = 96

MPS I/MPS II	30 (31 %)
MPS I/MPS II/MPS VII	20 (21 %)
MPS I/MPS II/MPS VI	16 (17 %)
MPS I/MPS II/MPS VI/MPS VII	13 (14 %)
MPS VI	7 (7 %)
MPS I	5 (5 %)
MPS III	2 (2 %)
MPS II/MPS VII	1 (1 %)
MPS II	1 (1 %)
MPS III/MPS VI/MPS VII	1 (1 %)

Your result: MPS I/MPS II

Score : 2

Scientific advisor comment: