



ERNDIM Urine MPS

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Urine Mucopolysaccharides Interim survey report 18-03-MPS

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

Laboratory N°: 392 (ERNDIM Urine MPS)

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Date of survey deadline: 2018-04-30

Date of report: 20 June 2018

Results of samples and evaluation of reporting

18-03-MPS Sample : MPS 2018.01

Lab 392

Patient information: F 38.0 year

Total Score : 4

Diagnosis: MPS I attenuated phenotype (M. Scheie)

Comments on proficiency:

In this urine sample of an adult MPS I Scheie patient 97% of the participants reported elevated total GAG. GAG subtype analysis and its interpretation was challenging. Increased dermatan sulfate was reported by 92%, but only 49% found increased heparan sulfate. MPS I was included in the differential diagnosis (in combination with MPS II, VI and/or VII) by 64 participants (70%). MPS VI (in combination with MPS II and/or VII, but not MPS I) was mentioned by 17 labs (18%). Two participants concluded this was a normal sample. Overall proficiency was 84%.

Creatinine (mmol/L)

N	93
Average	3,52
SD	0,31
Median	3,59
Your result	3.01
Z-Score	-1,65

GAG quantitative (mg/mmol creat)

Method	DMB colorimetric
N	64
Average	12,49
SD	4,47
Median	11,97
Your result	21.42
Z-Score	2,00

GAG excretion according to reference values

N registred	100
N results	91
Normal (%)	3 (3 %)
Increased (%)	88 (97 %)
Not detected	-- --
Your result	Increased

Score : 1

Scientific advisor comment:

Results of samples and evaluation of reporting

18-03-MPS Sample : MPS 2018.01

Lab 392

Patient information: F 38.0 year

Total Score : 4

Diagnosis: MPS I attenuated phenotype (M. Scheie)

Chondroitinsulfate

N registred	100
N results	82
Normal (%)	62 (76 %)
Increased (%)	4 (5 %)
Not detected	16 (20 %)
Your result	Normal

Dermatansulfate

N registred	100
N results	87
Normal (%)	2 (2 %)
Increased (%)	80 (92 %)
Not detected	5 (6 %)
Your result	Increased

Heparansulfate

N registred	100
N results	85
Normal (%)	25 (29 %)
Increased (%)	42 (49 %)
Not detected	18 (21 %)
Your result	Normal

Keratansulfate

N registred	100
N results	78
Normal (%)	21 (27 %)
Increased (%)	0 (0 %)
Not detected	57 (73 %)
Your result	Not detected

Score : 1

Scientific advisor comment:

Diagnosis

N registred = 100

N results = 92

MPS VI	15 (16 %)
MPS I/MPS III	1 (1 %)
MPS I/MPS VI/MPS VII	1 (1 %)
MPS I/MPS II/MPS III/MPS VI/MPS VII	1 (1 %)
MPS VI/Normal	1 (1 %)
MPS I/MPS IV/MPS VI	1 (1 %)
Normal	2 (2 %)
MPS III	2 (2 %)
MPS VII	1 (1 %)
MPS I/MPS VII	2 (2 %)
MPS I/MPS VI	8 (9 %)
MPS I/MPS II/MPS VI	8 (9 %)
MPS I/MPS II	9 (10 %)
MPS I/MPS II/MPS VI/MPS VII	11 (12 %)
MPS I/MPS II/MPS VII	12 (13 %)
MPS I	13 (14 %)
No Diagnosis	3 (3 %)
MPS VI/MPS VII	1 (1 %)

Your result: MPS I/MPS VI

Results of samples and evaluation of reporting

18-03-MPS Sample : MPS 2018.01

Lab 392

Patient information: F 38.0 year

Total Score : 4

Diagnosis: MPS I attenuated phenotype (M. Scheie)

Score : 2

Scientific advisor comment:

Results of samples and evaluation of reporting

18-03-MPS Sample : MPS 2018.02

Lab 392

Patient information: M 4.0 year

Total Score : 4

Diagnosis: Normal sample

Comments on proficiency:

The majority of the participants reported normal GAG results in this sample. Total proficiency based on scores was 93%

Creatinine (mmol/L)

N	93
Average	3,38
SD	0,30
Median	3,40
Your result	2.83
Z-Score	-1,82

GAG quantitative (mg/mmol creat)

Method	DMB colorimetric
N	64
Average	9,17
SD	4,14
Median	9,41
Your result	22.62
Z-Score	3,25

GAG excretion according to reference values

N registred	100
N results	91
Normal (%)	86 (95 %)
Increased (%)	5 (5 %)
Not detected	-- --
Your result	Normal

Score : 1

Scientific advisor comment:

Results of samples and evaluation of reporting

18-03-MPS Sample : MPS 2018.02

Lab 392

Patient information: M 4.0 year

Total Score : 4

Diagnosis: Normal sample

Chondroitinsulfate

N registred	100
N results	87
Normal (%)	83 (95 %)
Increased (%)	1 (1 %)
Not detected	3 (3 %)
Your result	Normal

Dermatansulfate

N registred	100
N results	90
Normal (%)	29 (32 %)
Increased (%)	3 (3 %)
Not detected	58 (64 %)
Your result	Not detected

Heparansulfate

N registred	100
N results	90
Normal (%)	43 (48 %)
Increased (%)	1 (1 %)
Not detected	46 (51 %)
Your result	Normal

Keratansulfate

N registred	100
N results	82
Normal (%)	23 (28 %)
Increased (%)	0 (0 %)
Not detected	59 (72 %)
Your result	Not detected

Score : 1

Scientific advisor comment:

Diagnosis

N registred = 100

N results = 94

Normal	84 (89 %)
No Diagnosis	4 (4 %)
MPS VI/Normal	1 (1 %)
MPS I/MPS II/MPS VI/MPS VII	1 (1 %)
MPS I/MPS II/MPS VII	1 (1 %)
MPS IV/Normal	1 (1 %)
MPS VI	1 (1 %)
MPS IV/MPS VI/Normal	1 (1 %)

Your result: Normal

Score : 2

Scientific advisor comment:

Results of samples and evaluation of reporting

18-03-MPS Sample : MPS 2018.03

Lab 392

Patient information: M 5.0 year

Total Score : 4

Diagnosis: MPS II

Comments on proficiency:

With 99% of the participants reporting elevated GAG, this was a clear MPS sample. Results reported of GAG subtype analysis were: elevated DS, 98% and elevated HS 93%. MPS II was included in the differential diagnosis by 89% of the participants. Overall proficiency was 86%.

Creatinine (mmol/L)

N	93
Average	4,28
SD	0,34
Median	4,30
Your result	3.54
Z-Score	-2,18

GAG quantitative (mg/mmol creat)

Method	DMB colorimetric
N	65
Average	67,42
SD	27,10
Median	71,30
Your result	78.57
Z-Score	0,41

GAG excretion according to reference values

N registred	100
N results	91
Normal (%)	1 (1 %)
Increased (%)	90 (99 %)
Not detected	-- --
Your result	Increased

Score : 1

Scientific advisor comment:

Results of samples and evaluation of reporting

18-03-MPS Sample : MPS 2018.03

Lab 392

Patient information: M 5.0 year

Total Score : 4

Diagnosis: MPS II

Chondroitinsulfate

N registred	100
N results	82
Normal (%)	62 (76 %)
Increased (%)	16 (20 %)
Not detected	4 (5 %)
Your result	Normal

Dermatansulfate

N registred	100
N results	89
Normal (%)	0 (0 %)
Increased (%)	87 (98 %)
Not detected	2 (2 %)
Your result	Increased

Heparansulfate

N registred	100
N results	89
Normal (%)	4 (4 %)
Increased (%)	83 (93 %)
Not detected	2 (2 %)
Your result	Increased

Keratansulfate

N registred	100
N results	78
Normal (%)	19 (24 %)
Increased (%)	2 (3 %)
Not detected	57 (73 %)
Your result	Not detected

Score : 1

Scientific advisor comment:

Diagnosis

N registred = 100

N results = 92

MPS I/MPS II	33 (36 %)
MPS I/MPS II/MPS VII	27 (29 %)
MPS I/MPS II/MPS VI/MPS VII	10 (11 %)
MPS I/MPS II/MPS VI	5 (5 %)
MPS II	4 (4 %)
MPS VI	4 (4 %)
MPS III	2 (2 %)
MPS I	2 (2 %)
MPS I/MPS II/MPS III	1 (1 %)
MPS I/MPS II/MPS IV/MPS VI/MPS VII	1 (1 %)
MPS I/MPS VI	1 (1 %)
MPS VII	1 (1 %)
MPS II/MPS III	1 (1 %)

Your result: MPS II

Results of samples and evaluation of reporting

18-03-MPS Sample : MPS 2018.03

Lab 392

Patient information: M 5.0 year

Total Score : 4

Diagnosis: MPS II

Score : 2

Scientific advisor comment: