



## **ERNDIM Urine MPS**

Dep. Clinical Genetics Ee2422  
Erasmus MC  
P.O. Box 1738  
3000 DR Rotterdam  
The Netherlands

### **Urine Mucopolysaccharides Interim survey report 17-08-MPS**

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

#### **Laboratory N°: 392 (ERNDIM Urine MPS)**

Contact: Tijen Tanyalcin

E-mail: [tanyalcinlab@gmail.com](mailto:tanyalcinlab@gmail.com)

Date of survey deadline: 2017-10-02

Date of report: 25 October 2017

## Results of samples and evaluation of reporting

17-08-MPS Sample : MPS 2017.04

Lab 392

Patient information: F 14.0 year

Total Score : 4

**Diagnosis:** MPS IV A.

Diagnosis was confirmed by enzyme testing and mutation analysis.

### Comments on proficiency:

Total proficiency was 65% for this sample, which is similar to previous MPS IV samples. Detection of keratan sulfate resulted in the correct diagnosis.

MPS IV was reported by 60 participants (from a total of 97 reports), while 21 considered this as a normal urine sample. Three participants concluded MPS IV or normal.

### Creatinine (mmol/L)

N	92
Average	5,59
SD	0,31
Median	5,60
Your result	5.04
Z-Score	-1,78

### GAG quantitative (mg/mmol creat)

Method	DMB colorimetric
N	64
Average	10,39
SD	4,20
Median	10,33
Your result	10.46
Z-Score	0,02

### GAG excretion according to reference values

N registred	102
N results	92
Normal (%)	31 (34 %)
Increased (%)	61 (66 %)
Not detected	-- --
Your result	Increased

**Score : 1**

**Scientific advisor comment:**

## Results of samples and evaluation of reporting

17-08-MPS Sample : MPS 2017.04

Lab 392

Patient information: F 14.0 year

Total Score : 4

**Diagnosis:** MPS IV A.

Diagnosis was confirmed by enzyme testing and mutation analysis.

### Chondroitinsulfate

N registred	102
N results	84
Normal (%)	60 (71 %)
Increased (%)	16 (19 %)
Not detected	8 (10 %)
Your result	Increased

### Dermatansulfate

N registred	102
N results	85
Normal (%)	23 (27 %)
Increased (%)	3 (4 %)
Not detected	59 (69 %)
Your result	Not detected

### Heparansulfate

N registred	102
N results	86
Normal (%)	33 (38 %)
Increased (%)	5 (6 %)
Not detected	48 (56 %)
Your result	Not detected

### Keratansulfate

N registred	102
N results	82
Normal (%)	4 (5 %)
Increased (%)	55 (67 %)
Not detected	23 (28 %)
Your result	Increased

**Score : 1**

**Scientific advisor comment:**

### Diagnosis

**N registred = 102**

**N results = 94**

MPS IV	59 (63 %)
Normal	20 (21 %)
No Diagnosis	4 (4 %)
MPS III	4 (4 %)
MPS IV/Normal	3 (3 %)
MPS I/MPS II	1 (1 %)
No Diagnosis/Normal	1 (1 %)
MPS I/MPS II/MPS III/MPS IV/MPS VI/MPS VII/Normal	1 (1 %)
MPS I/MPS III/MPS IV/MPS VI	1 (1 %)

Your result: MPS IV

**Score : 2**

**Scientific advisor comment:**

## Results of samples and evaluation of reporting

17-08-MPS Sample : MPS 2017.05

Lab 392

Patient information: F 12.0 year

Total Score : 4

**Diagnosis:** Normal control

### Comments on proficiency:

Most participants (n=87) concluded 'normal' in this sample.

Total proficiency was 93%.

### Creatinine (mmol/L)

N	93
Average	6,16
SD	0,43
Median	6,20
Your result	5.66
Z-Score	-1,17

### GAG quantitative (mg/mmol creat)

Method	DMB colorimetric
N	64
Average	5,86
SD	2,59
Median	5,75
Your result	7.64
Z-Score	0,69

### GAG excretion according to reference values

N registred	102
N results	93
Normal (%)	89 (96 %)
Increased (%)	4 (4 %)
Not detected	-- --
Your result	Normal

**Score : 1**

**Scientific advisor comment:**

## Results of samples and evaluation of reporting

17-08-MPS Sample : MPS 2017.05

Lab 392

Patient information: F 12.0 year

Total Score : 4

**Diagnosis:** Normal control

### Chondroitinsulfate

N registred	102
N results	85
Normal (%)	78 (92 %)
Increased (%)	2 (2 %)
Not detected	5 (6 %)
Your result	Normal

### Dermatansulfate

N registred	102
N results	85
Normal (%)	23 (27 %)
Increased (%)	2 (2 %)
Not detected	60 (71 %)
Your result	Not detected

### Heparansulfate

N registred	102
N results	85
Normal (%)	32 (38 %)
Increased (%)	2 (2 %)
Not detected	51 (60 %)
Your result	Not detected

### Keratansulfate

N registred	102
N results	81
Normal (%)	19 (23 %)
Increased (%)	1 (1 %)
Not detected	61 (75 %)
Your result	Not detected

**Score : 1**

**Scientific advisor comment:**

### Diagnosis

**N registred = 102**

**N results = 94**

Normal	85 (90 %)
No Diagnosis	3 (3 %)
MPS III	2 (2 %)
MPS VII/Normal	1 (1 %)
MPS I/MPS II	1 (1 %)
MPS I/MPS II/MPS VI	1 (1 %)
MPS IV/No Diagnosis	1 (1 %)

Your result: Normal

**Score : 2**

**Scientific advisor comment:**

## Results of samples and evaluation of reporting

17-08-MPS Sample : MPS 2017.06

Lab 392

Patient information: F 19.0 year

Total Score : 4

**Diagnosis:** MPS VI.

Diagnosis confirmed by enzymatic and genetic analysis.

### Comments on proficiency:

All participants concluded that this was a sample from an MPS patient. GAG was grossly elevated and 98% of the participating labs found elevated DS. Surprisingly, 27% found increased HS. This may be secondary to DS storage and is possibly due to compromised lysosomal function.

Total proficiency was 88%.

### Creatinine (mmol/L)

N	94
Average	2,79
SD	0,24
Median	2,80
Your result	2.3
Z-Score	-2,07

### GAG quantitative (mg/mmol creat)

Method	DMB colorimetric
N	64
Average	50,50
SD	16,62
Median	52,35
Your result	62.01
Z-Score	0,69

### GAG excretion according to reference values

N registred	102
N results	93
Normal (%)	1 (1 %)
Increased (%)	92 (99 %)
Not detected	-- --
Your result	Increased

**Score : 1**

**Scientific advisor comment:**

## Results of samples and evaluation of reporting

17-08-MPS Sample : MPS 2017.06

Lab 392

Patient information: F 19.0 year

Total Score : 4

**Diagnosis:** MPS VI.

Diagnosis confirmed by enzymatic and genetic analysis.

### Chondroitinsulfate

N registred	102
N results	83
Normal (%)	65 (78 %)
Increased (%)	11 (13 %)
Not detected	7 (8 %)
Your result	Normal

### Dermatansulfate

N registred	102
N results	89
Normal (%)	2 (2 %)
Increased (%)	87 (98 %)
Not detected	0 (0 %)
Your result	Increased

### Heparansulfate

N registred	102
N results	88
Normal (%)	26 (30 %)
Increased (%)	24 (27 %)
Not detected	38 (43 %)
Your result	Not detected

### Keratansulfate

N registred	102
N results	80
Normal (%)	15 (19 %)
Increased (%)	4 (5 %)
Not detected	61 (76 %)
Your result	Not detected

**Score : 1**

**Scientific advisor comment:**

### Diagnosis

**N registred = 102**

**N results = 92**

MPS VI	31 (34 %)
MPS I/MPS VII	1 (1 %)
MPS I/MPS II/MPS IV	1 (1 %)
MPS I/MPS III/MPS VI	1 (1 %)
MPS IV	1 (1 %)
MPS I/MPS VI/MPS VII	1 (1 %)
MPS III	2 (2 %)
MPS VI/MPS VII	2 (2 %)
MPS VII	3 (3 %)
MPS I/MPS II/MPS VII	5 (5 %)
MPS I/MPS II	5 (5 %)
MPS I/MPS VI	7 (8 %)
MPS I/MPS II/MPS VI	12 (13 %)
MPS I/MPS II/MPS VI/MPS VII	16 (17 %)
MPS I	3 (3 %)
MPS I/MPS II/MPS III	1 (1 %)

Your result: MPS I/MPS VI

## Results of samples and evaluation of reporting

---

**17-08-MPS Sample : MPS 2017.06**

**Lab 392**

Patient information: F 19.0 year

Total Score : 4

**Diagnosis:** MPS VI.

Diagnosis confirmed by enzymatic and genetic analysis.

---

**Score : 2**

**Scientific advisor comment:**