

Patient ID SA00122959	Patient Name TESTINGRNV, SAMPLEREPORT	Birth Date 1982-06-29	Gender F	Age 37
Order Number SA00122959	Client Order Number SA00122959	Ordering Physician CLIENT,CLIENT	Report Notes	
Account Information C7028846 DLMP Rochester		Collected 15 Sep 2019 10:00		

Purines and Pyrimidines Panel, U

Interpretation (PUPYU)

1 MCR

POSITIVE In this urine sample, the concentrations of xanthine, hypoxanthine, and s-sulfocysteine were elevated, and uric acid was low. These findings are suggestive of a biochemical diagnosis of molybdenum cofactor deficiency (MOCD). Consider molecular analysis of GPHN, MOCS1, and MOCS2 for confirmation of type of MOCD. Please contact the Biochemical Genetics consultant on call (1-800-533-1710) if you have any questions.

ADDITIONAL INFORMATION

Liquid Chromatography-Tandem Mass Spectrometry (LC-MS/MS)

Uracil

20 mmol/mol Cr

MCR

Reference Value
≤ 20

Thymine

3 mmol/mol Cr

MCR

Reference Value
≤ 3

Adenine

3 mmol/mol Cr

MCR

Reference Value
≤ 3

Hypoxanthine

 422 mmol/mol Cr

MCR

Reference Value
≤ 30

Xanthine

 364 mmol/mol Cr

MCR

Reference Value
≤ 20

Orotic Acid

4 mmol/mol Cr

MCR

Reference Value
≤ 5

Dihydroorotic

3 mmol/mol Cr

MCR

Reference Value
≤ 3

Uric Acid

 51 mmol/mol Cr

MCR

Reference Value
70–700

Deoxythymidine

3 mmol/mol Cr

MCR

Reference Value
≤ 3

Deoxyuridine

3 mmol/mol Cr

MCR

Reference Value
≤ 3

Thymidine

3 mmol/mol Cr

MCR

Reference Value
≤ 3

Uridine

3 mmol/mol Cr

MCR

Reference Value
≤ 3

Deoxyadenosine

3 mmol/mol Cr

MCR

Reference Value
≤ 3

Deoxyinosine

3 mmol/mol Cr

MCR

Reference Value
≤ 3

Deoxyguanosine

3 mmol/mol Cr

MCR

Reference Value
≤ 3

Adenosine

3 mmol/mol Cr

MCR

Reference Value
≤ 3

Performing Site Legend

Code	Laboratory	Address	Lab Director	CLIA Certificate
MCR	Mayo Clinic Laboratories - Rochester Main Campus	200 First Street SW, Rochester, MN 55905	William G. Morice M.D. Ph.D	24D0404292

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Inosine	MCR	Dihydrouracil	MCR
3 mmol/mol Cr	Reference Value ≤ 3	6 mmol/mol Cr	Reference Value ≤ 6
Guanosine	MCR	Dihydrothymine	MCR
3 mmol/mol Cr	Reference Value ≤ 3	3 mmol/mol Cr	Reference Value ≤ 3
AICAR	MCR	N-carbamoyl-beta-alanine	MCR
3 mmol/mol Cr	Reference Value ≤ 3	10 mmol/mol Cr	Reference Value ≤ 10
Succinyladenosine	MCR	N-carbamoyl-beta-aminoisobutyric Acid	MCR
3 mmol/mol Cr	Reference Value ≤ 3	3 mmol/mol Cr	Reference Value ≤ 3
S-Sulfocysteine	MCR	Reviewed By	MCR
 89 mmol/mol Cr	Reference Value ≤ 5	Amy Breitlow	
		Received: 16 Sep 2019 10:43	Reported: 16 Sep 2019 13:05

Laboratory Notes

- 1 This test was developed and its performance characteristics determined by Mayo Clinic in a manner consistent with CLIA requirements. This test has not been cleared or approved by the U.S. Food and Drug Administration.

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