

Patient ID SA00122962	Patient Name TESTINGRNA, SAMPLEREPORT	Birth Date 1982-06-29	Gender F	Age 37
Order Number SA00122962	Client Order Number SA00122962	Ordering Physician CLIENT, CLIENT	Report Notes	
Account Information C7028846 DLMP Rochester		Collected 15 Sep 2019 10:00		

Purines and Pyrimidines Panel, P

Interpretation (PUPYP)

In this plasma sample, the concentration of adenine was elevated, which is consistent with a biochemical diagnosis of adenine phosphoribosyltransferase (APRT) deficiency and observed in patients on treatment. Please contact the Biochemical Genetics consultant on call (Tel. 1-800-533-1710) if you have any questions.

ADDITIONAL INFORMATION

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

Uracil

2 nmol/mL

MCR

Reference Value
≤2

Thymine

2 nmol/mL

MCR

Reference Value
≤2

Adenine

 28 nmol/mL

High

MCR

Reference Value
≤3

Hypoxanthine

15 nmol/mL

MCR

Reference Value
≤15

Xanthine

3 nmol/mL

MCR

Reference Value
≤3

Dihydroorotic

2 nmol/mL

MCR

Reference Value
≤2

Uric Acid

257 nmol/mL

MCR

Reference Value
150-500

Deoxythymidine

2 nmol/mL

MCR

Reference Value
≤2

Deoxyuridine

2 nmol/mL

MCR

Reference Value
≤2

Uridine

9 nmol/mL

MCR

Reference Value
≤9

Deoxyinosine

2 nmol/mL

MCR

Reference Value
≤2

Deoxyguanosine

2 nmol/mL

MCR

Reference Value
≤2

Inosine

2 nmol/mL

MCR

Reference Value
≤2

Guanosine

2 nmol/mL

MCR

Reference Value
≤2

Dihydrouracil

3 nmol/mL

MCR

Reference Value
≤3

Dihydrothymine

2 nmol/mL

MCR

Reference Value
≤2

N-carbamoyl-B-alanine

2 nmol/mL

MCR

Reference Value
≤2

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



Patient ID SA00122962	Patient Name TESTINGRNA, SAMPLEREPORT	Birth Date 1982-06-29	Gender F	Age 37
Order Number SA00122962	Client Order Number SA00122962	Ordering Physician CLIENT,CLIENT	Report Notes	
Account Information C7028846 DLMP Rochester		Collected 15 Sep 2019 10:00		

N-carbamoyl-B-aminoisobutyric acid

2 nmol/mL

MCR

Reference Value
≤2

Reviewed By

Amy Breitlow

MCR

Received: 16 Sep 2019 10:45

Reported: 16 Sep 2019 10:59

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.

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