

|                                                       |                                          |                                             |                    |                  |
|-------------------------------------------------------|------------------------------------------|---------------------------------------------|--------------------|------------------|
| Patient ID<br><b>SA00148220</b>                       | Patient Name<br><b>TESTINGRNA, ATLAS</b> | Birth Date<br><b>1989-01-09</b>             | Gender<br><b>F</b> | Age<br><b>32</b> |
| Order Number<br><b>SA00148220</b>                     | Client Order Number<br><b>SA00148220</b> | Ordering Physician<br><b>CLIENT, CLIENT</b> | Report Notes       |                  |
| Account Information<br><b>C7028846 DLMP Rochester</b> |                                          | Collected<br><b>06 Oct 2021 12:00</b>       |                    |                  |

## Acute Porphyrria Gene Panel

### Interpretation

MCR

This result decreases the likelihood but does not rule out the involvement of the genes evaluated in this panel.

Individuals may have a pathogenic variant in one of the interrogated genes that is not detectable by the methods utilized. Additionally, the clinical phenotype that is observed in this individual and/or family may be due to a pathogenic variant or variants in another gene not targeted by this test.

This result should be interpreted in the context of clinical findings, family history, and other laboratory testing.

A genetic consultation may be of benefit.

### Result Summary

MCR

## Negative

### Result

MCR

No reportable variants were detected.

### Test Description

MCR

Evaluation of 4 genes associated with acute porphyria

### Specimen

MCR

WB Whole Blood

### Resources

MCR

Disease specific information, including current therapies may be available at:

1. GeneReviews: <https://www.ncbi.nlm.nih.gov/books/NBK1116/>
2. <https://www.fda.gov/drugs>

Information regarding clinical trials, if available, can be found at the following sites:

1. ClinicalTrials.gov: <http://clinicaltrials.gov/ct2/search/advanced>

### Method

MCR

Next generation sequencing (NGS) and/or Sanger sequencing was performed to test for the presence of variants in coding regions and intron/exon boundaries of the genes analyzed, as well as select regions that have known pathogenic variants. The human genome reference GRCh37/hg19 build was used for sequence read alignment. At least 99% of the bases are covered at a read depth >30X. Sensitivity is estimated at >99% for single nucleotide variants, >94% for indels up to 39 base pairs, >95% for deletions up to 75 base pairs and insertions up to 47 base pairs. NGS and/or a PCR-based quantitative method was performed to test for the presence of deletions and duplications in the genes analyzed. See the Genes Analyzed field for a list of genes tested.

There may be regions of genes that cannot be effectively evaluated for sequencing or deletion and duplication analysis as a result of technical limitations of the assay, including regions of homology, high GC content, and repetitive sequences. Confirmation of select reportable variants was performed by alternate methodologies based on internal laboratory criteria. See [www.mayocliniclabs.com](http://www.mayocliniclabs.com) APGP for details regarding genes with regions not routinely covered.

### Genes Analyzed

MCR

ALAD, CPOX, HMBS, and PPOX

### Disclaimer

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#### Clinical Correlations

An online research opportunity called GenomeConnect ([genomeconnect.org](http://genomeconnect.org)), a project of ClinGen, is available for the recipient of this genetic test. This patient registry collects de-identified genetic and health information to advance the knowledge of genetic variants. Mayo Clinic is a collaborator of ClinGen. This may not be applicable for all tests.

If testing was performed because of a clinically significant family history it is often useful to first test an affected family member. Detection of a reportable variant(s) in an affected family member

### 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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| Account Information<br><b>C7028846 DLMP Rochester</b> |                                          | Collected<br><b>06 Oct 2021 12:00</b>       |                    |                  |

would allow for more informative testing of at risk individuals.

To discuss the availability of further testing options or for assistance in the interpretation of these results, Mayo Clinic Laboratory genetic counselors can be contacted at 1-800-533-1710.

#### Technical Limitations

Next generation sequencing may not detect all types of genomic variants. In rare cases, false negative or false positive results may occur. The depth of coverage may be variable for some target regions, but assay performance below the minimum acceptable criteria or for failed regions will be noted. Given these limitations, negative results do not rule out the diagnosis of a genetic disorder. If a specific clinical disorder is suspected, evaluation by alternative methods can be considered.

There may be regions of genes that cannot be effectively evaluated for sequencing or deletion and duplication analysis as a result of technical limitations of the assay, including regions of homology, high GC content, and repetitive sequences. Confirmation of select reportable variants was performed by alternate methodologies based on internal laboratory criteria.

Additionally, low level mosaic variants may not be detected.

This test is not designed to differentiate between somatic and germline variants. If there is a possibility that any detected variant is somatic, additional testing may be necessary to clarify the

significance of results.

Genes may be added or removed based on updated clinical relevance. Please refer to the Targeted Genes and Methodology Details for the Acute Porphyria Gene Panel in the Special Instructions section of the Test Catalog for the most up to date list of genes included in this test.

#### Reclassification of Variants Policy

See [www.mayocliniclabs.com](http://www.mayocliniclabs.com)

APGP for information regarding the laboratory's policy for reclassification of variants.

#### Variant Evaluation

Variant curation is performed using published ACMG-AMP recommendations as a guideline. Other gene-specific guidelines may also be considered. Variants classified as benign or likely benign are not reported.

Results from in silico evaluation tools may change over time and should be interpreted with caution and professional clinical judgment.

#### Released By

Devin Oglesbee, Ph.D.

**Received:** 07 Oct 2021 16:12

**Reported:** 15 Oct 2021 10:35

MCR

#### Laboratory Notes

- 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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1-800-533-1710  
Acute Porphyria Gene Panel

APGP

|                                                                                                                                                                              |                                |                                  |               |                                                               |
|------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|--------------------------------|----------------------------------|---------------|---------------------------------------------------------------|
| PATIENT NAME<br><b>TESTINGRNV, ATLAS</b>                                                                                                                                     |                                |                                  |               | ORDER NUMBER<br><b>L107000375</b>                             |
| PATIENT ID<br>SA00148220                                                                                                                                                     | DATE OF BIRTH<br>01/09/1989    | AGE<br>32 Y                      | SEX<br>Female | REQUESTED BY<br>CLIENT CLIENT                                 |
| COLLECTED<br>10/6/2021, 12:00 PM                                                                                                                                             | RECEIVED<br>10/7/2021, 4:12 PM | REPORTED<br>10/15/2021, 10:35 AM |               |                                                               |
| The collected, received, and reported dates and times on the report are in the time zone of the performing location.<br>7028846<br>MCL RochesterCampus<br>Rochester MN 55901 |                                |                                  |               | CLIENT ORDER NUMBER<br>SA00148220<br>CLIENT MRN<br>SA00148220 |

**TEST DESCRIPTION**

Evaluation of 4 genes associated with acute porphyria

**SPECIMEN**

WB Whole Blood

**RESULT SUMMARY**

Negative

**RESULT**

No reportable variants were detected.

**INTERPRETATION**

This result decreases the likelihood but does not rule out the involvement of the genes evaluated in this panel.

Individuals may have a pathogenic variant in one of the interrogated genes that is not detectable by the methods utilized. Additionally, the clinical phenotype that is observed in this individual and/or family may be due to a pathogenic variant or variants in another gene not targeted by this test.

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A genetic consultation may be of benefit.

**RESOURCES**

Disease specific information, including current therapies may be available at:

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**METHOD**

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PATIENT NAME TESTINGRNV, ATLAS

21-44563



1-800-533-1710  
Acute Porphyría Gene Panel

APGP

|                                                                                                                                     |                                |                                  |               |                                   |
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| 7028846<br>MCL RochesterCampus<br>Rochester MN 55901                                                                                |                                |                                  |               | CLIENT MRN<br>SA00148220          |

Genes Analyzed field for a list of genes tested.

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**GENES ANALYZED**

ALAD, CPOX, HMBS, and PPOX

**DISCLAIMER**

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|              |                   |          |
|--------------|-------------------|----------|
| PATIENT NAME | TESTINGRNV, ATLAS | 21-44563 |
|--------------|-------------------|----------|



1-800-533-1710  
Acute Porphyria Gene Panel

APGP

|                                                                                                                                                                              |                                |                                  |               |                                                               |
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Methodology Details for the Acute Porphyria Gene Panel in the Special Instructions section of the Test Catalog for the most up to date list of genes included in this test.

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TEST CLASSIFICATION

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RELEASED BY

Devin Oglesbee, Ph.D.

Code : MCR Laboratory : Mayo Clinic Laboratories - Rochester Main Campus  
Lab Director : WILLIAM G MORICE, II MD, PhD CLIA Certificate : 24D0404292

Address : 200 FIRST STREET SW  
ROCHESTER MN 55905