Patient ID	Patient Name		Birth Date	Gender	Age
SA00148250	TESTINGRNV, ATLAS		1989-01-01	F	32
Order Number	**Client Order Number**	**Ordering Physician**	**Report Notes**		
SA00148250	SA00148250	CLIENT,CLIENT			
Account Information		**Collected**			
C7028846 DLMP Rochester		06 Oct 2021 12:00			

Glycogen Storage Disease 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 28 genes associated with glycogen storage disease

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 GSDGP for details regarding genes with regions not routinely covered.

Genes Analyzed MCR

AGL, ALDOA, ENO3, EPM2A, FBP1, G6PC, GAA, GBE1, GYG1, GYS1, GYS2, LAMP2, LDHA, NHLRC1, PFKM, PGAM2, PGK1, PGM1, PHKA1, PHKA2, PHKB, PHKG2, PRKAG2, PYGL, PYGM, RBCK1, SLC2A2, and SLC37A4

Disclaimer 1 MCR

Clinical Correlations
An online research opportunity called GenomeConnect (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.

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

MAYO CLINIC LABORATORIES

Patient ID	Patient Name		Birth Date	Gender	Age
SA00148250	TESTINGRNV, ATLAS		1989-01-01	F	32
Order Number	**Client Order Number**	**Ordering Physician**	**Report Notes**		
SA00148250	SA00148250	CLIENT,CLIENT			
Account Information		**Collected**			
C7028846 DLMP Rochester		06 Oct 2021 12:00			

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 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 Glycogen Storage Disease 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
GSDGP 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 MCR

Devin Oglesbee, Ph.D.

Received: 07 Oct 2021 16:13 **Reported:** 15 Oct 2021 11:59

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

MAYO CLINIC

1-800-533-1710

Glycogen Storage Disease Gene Panel

GSDGP

PATIENT NAME				ORDER NUMBER
TESTINGRNV, ATLAS				**L107000421**
PATIENT ID SA00148250	DATE OF BIRTH 01/01/1989	AGE 32 Y	SEX Female	REQUESTED BY CLIENT CLIENT
COLLECTED 10/6/2021, 12:00 PM	RECEIVED 10/7/2021, 4:13 PM	REPORTED 10/15/2021, 11:59 AM		
The collected, received, and reported dates and times on the report are in the time zone of the performing location. 7028846 MCL RochesterCampus Rochester MN 55901				CLIENT ORDER NUMBER SA00148250 CLIENT MRN SA00148250

TEST DESCRIPTION

Evaluation of 28 genes associated with glycogen storage disease

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.

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

A genetic consultation may be of benefit.

RESOURCES

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

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

PATIENT NAME				ORDER NUMBER
TESTINGRNV, ATLAS				**L107000421**
PATIENT ID SA00148250	DATE OF BIRTH 01/01/1989	AGE 32 Y	SEX Female	REQUESTED BY CLIENT CLIENT
COLLECTED 10/6/2021, 12:00 PM	RECEIVED 10/7/2021, 4:13 PM	REPORTED 10/15/2021, 11:59 AM		
The collected, received, and reported dates and times on the report are in the time zone of the performing location. 7028846 MCL RochesterCampus Rochester MN 55901				CLIENT ORDER NUMBER SA00148250 CLIENT MRN SA00148250

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 GSDGP for details regarding genes with regions not routinely covered.

GENES ANALYZED

AGL, ALDOA, ENO3, EPM2A, FBP1, G6PC, GAA, GBE1, GYG1, GYS1, GYS2, LAMP2, LDHA, NHLRC1, PFKM, PGAM2, PGK1, PGM1, PHKA1, PHKA2, PHKB, PHKG2, PRKAG2, PYGL, PYGM, RBCK1, SLC2A2, and SLC37A4

DISCLAIMER

Clinical Correlations
An online research opportunity called GenomeConnect (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 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.

Glycogen Storage Disease Gene Panel

GSDGP

PATIENT NAME				ORDER NUMBER
TESTINGRNV, ATLAS				**L107000421**
PATIENT ID SA00148250	DATE OF BIRTH 01/01/1989	AGE 32 Y	SEX Female	REQUESTED BY CLIENT CLIENT
COLLECTED 10/6/2021, 12:00 PM	RECEIVED 10/7/2021, 4:13 PM	REPORTED 10/15/2021, 11:59 AM		

The collected, received, and reported dates and times on the report are in the time zone of the performing location.

7028846
MCL RochesterCampus
Rochester MN 55901

CLIENT ORDER NUMBER
SA00148250

CLIENT MRN
SA00148250

Genes may be added or removed based on updated clinical relevance. Please refer to the Targeted Genes and Methodology Details for the Glycogen Storage Disease 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 GSDGP 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.

TEST CLASSIFICATION
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.

RELEASED BY

Devin Oglesbee, Ph.D.

Code : MCR Laboratory : Mayo Clinic Laboratories - Rochester Main Campus Address : 200 FIRST STREET SW
Lab Director : WILLIAM G MORICE, II MD, PhD CLIA Certificate : 24D0404292 ROCHESTER MN 55905