

Patient ID SA00141550	Patient Name TESTINGRNA, ATLAS	Birth Date 1980-01-01	Gender M	Age 41
Order Number SA00141550	Client Order Number SA00141550	Ordering Physician CLIENT, CLIENT	Report Notes	
Account Information C7028846 DLMP Rochester		Collected 14 Jan 2021 15:37		

Test Description

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Evaluation of 28 genes associated with glycogen storage disease

Specimen

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WB Whole Blood

Result Summary

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Pathogenic Variants Detected

Result

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Gene (Transcript)	Variant	Zygosity	Classification
GAA (NM_000152.3)	c.-32-13T>G Chr17(GRCh37):g.78078341	Homozygous	Pathogenic

The following homozygous PATHOGENIC alteration was detected:
GAA (NM_000152.3), c.-32-13T>G, Chr17(GRCh37):g.78078341

No additional reportable variants were detected within all other tested genes. See the Genes Analyzed section for a complete list of genes evaluated by this assay.

Interpretation

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GAA, c.-32-13T>G, PATHOGENIC

The homozygous c.-32-13T>G variant in the GAA gene (mim:606800) is an established pathogenic variant that is typically associated with late-onset Pompe disease. This result is consistent with a diagnosis of Pompe disease. Clinical correlation is recommended.

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

Consultation with a genetics professional is recommended for interpretation of this result and to determine whether familial testing may be of benefit to this family. Genetic testing for family members is available by ordering FMTT / Familial Mutation, Targeted Testing for the specific variant(s) detected. Please contact the laboratory at 1-800-533-1710 or the online test catalog at www.mayocliniclabs.com for information about FMTT.

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 SA00141550	Patient Name TESTINGRNV, ATLAS	Birth Date 1980-01-01	Gender M	Age 41
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Resources

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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

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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. 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 amplified 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 specific details on regions not routinely covered.

Genes Analyzed

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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

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Clinical Correlations

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.

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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: 18 Jan 2021 08:58

Reported: 29 Jan 2021 13:57

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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