Patient ID	Patient Name		Birth Date	Sex	Age
321	TESTRNV, IMPLEMENTATION		1968-02-19	M	54
Order Number	Client Order Number	Ordering Physician	Report Notes		
X100389346	X100389346	UNKNOWN,PROVIDER			
Account Information		Collected			
C7028846 DLMP Rochester		16 May 2022 11:00			

Galactosemia Mutation Panel

Result Summary

MCR

NEGATIVE

Result

MCR

None of the listed variants were identified.

Interpretation

1 MCR

In the absence of galactose-1-phosphate uridyltransferase (GALT) enzyme levels, these results do not exclude carrier or affected status for this individual. Enzyme testing is recommended. However, this test cannot be added on to the existing sample and a new sample would be needed to assess the GALT activity in this individual (GALT / Gal-1-P Uridyltransferase, RBC).

If a diagnosis of galactosemia is still suspected, additional genetic testing strategies, such as full gene analysis of the GALT gene (GALZ / Galactosemia, Full Gene Analysis), should be considered for identifying pathogenic variants that are not detected by this assay. Contact the Genomics Laboratory at 1–800–533–1710 for further discussion regarding this option.

A genetic consultation may be of benefit.

ADDITIONAL INFORMATION

CAUTIONS

This assay will not detect all of the known pathogenic variants causing galactosemia. Therefore, the absence of a detectable variant does not rule out the possibility that an individual is a carrier of or affected with this disease.

Many disorders may present with symptoms similar to those associated with galactosemia. Therefore, biochemical testing is recommended to establish the diagnosis of galactosemia prior to DNA analysis.

A negative result does not eliminate the risk of carrier status for any of the included conditions, due to the possibility that the patient carries a variant that is not interrogated with this assay or the rare chance of a false-negative result for a tested variant. For tested variants, the negative predictive value of this screen is greater than 98%. The patient's residual risk to be a carrier after a negative screen is dependent on ethnic background and family history.

A positive control was not available for all variants targeted on this panel. For more information regarding availability of a positive control for each variant see Targeted Variants Interrogated by Expanded Carrier Screen Panels in Special Instructions. The negative predictive value of these targets is unknown.

Rare polymorphisms exist that could lead to false-negative or false-positive results. If results obtained do not match the clinical findings, additional testing should be considered.

All detected variants are evaluated according to American College of Medical Genetics and Genomics recommendations. This assay was designed to specifically target known pathogenic or likely pathogenic variants. In rare cases, DNA variants of undetermined significance may be identified. The laboratory encourages health care providers to contact the laboratory at any time to learn how the status of a particular variant may have changed over time.

Multiple in-silico evaluation tools may have been used to assist in the interpretation of these results. Of note, the sensitivity and specificity of these tools for the determination of pathogenicity is currently unvalidated.

Test results should be interpreted in the context of clinical findings, family history, and other laboratory data. Misinterpretation of results may occur if the information provided is inaccurate or incomplete.

Bone Marrow transplants from allogenic donors will interfere with testing. Call Mayo Clinic Laboratories for instructions for testing patients who have received a bone marrow transplant.

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

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	Patient Name		Birth Date	Sex	Age
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deidentified 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.

Specimen

MCR

WB Whole Blood

Method

MCR

An array based assay was used to test for the presence of the following variants in the GALT gene (GenBank accession number NM_000155; build GRCh37 (hg19)): -119_-116delGTCA, 136_140del, L74P, D98N, S135L, T138M, R148Q, M142K, Q169K, F171S, Q188R, L195P, E203K, Y209C, K285N, N314D, A320T, R333G, R333W, E340*, Q344K, 253–2A>G, *380Rext*49 and 5 kb deletion.

Released By

MCR

LINDA HASADSRI

Received: 23 May 2022 12:24 **Reported:** 25 May 2022 10:54

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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