MAYO CLINIC LABORATORIES

1-800-533-1710

GAL14

Galactosemia Gene Analysis

Patient ID	Patient Name		Birth Date	Gender	Age
SA00007953	SAMPLEREPORT, GCTABNORMAL		1966-06-10	F	49
Order Number	**Client Order Number**	**Ordering Physician**	**Report Notes**		
SA00007953	SA00007953	CLIENT, CLIENT			
Account Information		**Collected**			
C7028846 DLMP Rochester		07 Apr 2016 00:00			

Galactosemia Gene Analysis

Result Summary

MCR

POSITIVE

Result

MCR

The following heterozygous alterations were identified:
DNA change: c.–119_-116delGTCA (g.34646583_34646586)

AND

Amino Acid change: p.N314D (Asn314Asp)
DNA change: c.940A>G (g.34649442)
Together these sequence changes are consistent with the Duarte (D2) allele and are associated with reduced GALT activity.

AND

Amino Acid change: p.Q188R (Gln188Arg)
DNA change: c.563A>G (g.34648167)
Classification: PATHOGENIC

Interpretation

1 MCR

Biochemical and molecular test results are in agreement. The observed GALT enzyme activity in red blood cells (3.5 nmol/h/mg Hb) and the presence of c.–119_-116delGTCA and p.N314D, consistent with the Duarte (D2) variant, and p.Q188R suggest that this individual is affected with Duarte variant galactosemia. If clinically indicated, consider measuring galactose-1-phosphate (GAL1P / Galactose-1-Phosphate, RBC). Some individuals with Duarte variant galactosemia are treated with a low galactose containing diet during infancy.

Since mutations have been identified, genetic testing of at risk family members could be considered.

A genetic consultation may be of benefit.

ADDITIONAL INFORMATION

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.

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.

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.

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.

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.

Specimen

MCR

WB EDTA

Method

MCR

The multiplex PCR-based assay utilizing Agena Mass Array platform was used to test for the presence of the following mutations in the GALT gene (GenBank accession number NM_000155; build GRCh37 (hg19)): -119_-116delGTCA, D98N, S135L, T138M, M142K, F171S, Q188R, L195P, Y209C, K285N, N314D, Q344K, 253–2A>G, and 5 kb deletion.

Released By

MCR

Devin Oglesbee, Ph.D.

Received: 08 Apr 2016 08:59 **Reported:** 08 Apr 2016 11:00

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	Gender	Age
SA00007953	SAMPLEREPORT, GCTABNORMAL		1966-06-10	F	49
Order Number	**Client Order Number**	**Ordering Physician**	**Report Notes**		
SA00007953	SA00007953	CLIENT, CLIENT			
Account Information		**Collected**			
C7028846 DLMP Rochester		07 Apr 2016 00:00			

Galactosemia Reflex, B

Gal-1-P Uridyltransferase, RBC

MCR

⚠ 3.5 nmol/h/mg Hb

Low

Reference Value
≥24.5

Interpretation (GALT)

MCR

The galactose-1-phosphate uridyltransferase (GALT) activity in this sample is reduced and most consistent with the Duarte variant galactosemia (partial GALT deficiency; DG). Consider a galactose-free diet until the diagnosis is verified by molecular genetic testing. Molecular genetic test results to follow. Please contact the Biochemical Genetics consultant or genetic counselor on call (1–800–533–1710) if you have any questions.

ADDITIONAL INFORMATION

Enzyme reaction followed by LC-MS/MS

Reviewed By

MCR

Wanda Barber

Received: 08 Apr 2016 08:59 **Reported:** 08 Apr 2016 09:00

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