



LABORATORY REPORT

Example Client, XYZ123
1234 Warde Road
Ann Arbor MI 48108

EXAMPLE, REPORT W
WX0000003826 F 12/05/1988 34 Y

Referral Testing

Collected: 09/29/2023 13:49 Received: 09/29/2023 13:49

<u>Test Name</u>	<u>Result</u>	<u>Flag</u>	<u>Ref-Ranges</u>	<u>Units</u>	<u>Site</u>
Galactokinase - Blood	See Below				WMMR

Test	Result	Flag	Units	Ref Range
Galactokinase, B	2.1		nmol/h/mg Hb	≥0.7

Interpretation (GALK)
In this specimen, the activity of galactokinase (GALK) is not reduced. This result is not consistent with galactosemia due to GALK deficiency. However, if the reason for referral was bilateral cataracts, consider sending plasma to rule out cerebrotendinous xanthomatosis (CTX; MCL test CTXP). CTX is a multisystem disorder of bile acid synthesis which may first present with cataracts. 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
Dietrich Matern, M.D.

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.

Test(s) performed at
Mayo Clinic Laboratories - Rochester Main Campus
200 First Street SW, Rochester, MN 55905

Performing Site:
WMMR: MAYO CLINICAL LABS 3050 Superior Drive NW Rochester MN 55901

Reported Date: 2023.09.29 13:50

LAB: L - LOW, H - HIGH, AB - ABNORMAL, C - CRITICAL, . - NOT TESTED

F329000022 Ordered By: KAJAL SITWALA, MD, PhD
WX0000003826 WX00000000002353
Printed D&T: 09/29/23 13:50

Kajal V. Sitwala, MD, PhD - Medical Director
Form: MM RL1
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