



Patient ID SA00064685	Patient Name TESTINGRNV, OLIWBABNORMAL	Birth Date 1985-02-11	Gender F	Age 28
Order Number SA00064685	Client Order Number SA00064685	Ordering Physician Client, Client	Report Notes	
Account Information C7028846 DLMP Rochester		Collected 21 Nov 2013 00:00		

Oligosaccharidoses Screen, WBC

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MCR

In this specimen, alpha-N-acetyl-galactosaminidase activity is reduced. This result is consistent with a diagnosis of Schindler disease (OMIM 609241), a rare autosomal recessive lysosomal storage disorder. If clinically indicated, consider further confirmatory testing by molecular genetic analysis of the NAGA gene. A genetics consultation is recommended. 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 FIA-MS/MS

Reviewed By

MCR

JANICE HELGESON

Received: 22 Nov 2013 11:31

Reported: 22 Nov 2013 11:42

Performing Site Legend

Code	Laboratory	Address
MCR	Mayo Clinic Dept. of Lab Med and Pathology	200 First Street SW, Rochester, MN 55905