

Patient Name SAMPLEREP, FUCT A	Patient ID SA00062408	Age 26	Gender F	Order # SA00062408
Ordering Phys CLIENT, CLIENT				DOB 05/22/1987
Client Order # SA00062408	Account Information			Report Notes
Collected 09/17/2013 10:27	C7028846-DLMP Rochester SDSC 2 - Client Support Rochester, MN 55901			
Printed 10/14/2013 14:34				

Test	Flag	Results	Unit	Reference Value	Perform Site*
Cryopreserve for Biochem Studies					MCR
RECEIVED: 09/17/2013 11:15 REPORTED: 09/17/2013 11:15 Cryopreserve for Biochem Studies was cancelled on 09/17/2013 at 11:15; Test not performed.					
Fibroblast Culture					MCR
RECEIVED: 09/17/2013 11:15 REPORTED: 09/17/2013 11:15 Fibroblast Culture was cancelled on 09/17/2013 at 11:15; Test not performed.					
Alpha-Fucosidase, Fibroblasts	L	0.11	nmol/min/mg Prot	>=0.41	MCR
RECEIVED: 09/17/2013 11:15 REPORTED: 09/17/2013 13:37 Interpretation (FUCT)					
In this specimen, fucosidase activity is reduced. This is consistent with a diagnosis of fucosidosis (OMIM 230000), a rare autosomal recessive lysosomal storage disorder. If clinically indicated, consider further confirmatory testing by molecular genetic analysis of the FUCAL 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.					
Fluorometric Enzyme Assay Reviewed By Daniel Kraft					
					MCR

* Performing Site:

MCR	Mayo Clinic Laboratories - Rochester Main Campus 200 First St SW Rochester, MN 55905	Lab Director:
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Patient Name SAMPLEREP, FUCT A	Collection Date and Time 09/17/2013 10:27	Report Status Final
Page 1 of 1	** End of Report **	

* Report times for Mayo performed tests are CST/CDT