

Patient Name SAMPLEREP, ARSB A	Patient ID SA00062392	Age 37	Gender F	Order # SA00062392
Ordering Phys CLIENT, CLIENT				DOB 09/30/1975
Client Order # SA00062392	Account Information			Report Notes
Collected 09/17/2013 10:01	C7028846-DLMP Rochester SDSC 2 - Client Support			
Printed 10/14/2013 09:33	Rochester, MN 55901			

Test	Flag	Results	Unit	Reference Value	Perform Site*
Arylsulfatase B, Fibroblasts	L	0.79	nmol/min/mg Prot	>=6.08	MCR

RECEIVED: 09/17/2013 11:03 REPORTED: 09/17/2013 13:11

Interpretation (ARSB)

MCR

In this specimen, arylsulfatase B is decreased. This result indicates that this individual may be affected with mucopolysaccharidosis VI (Maroteaux-Lamy syndrome; OMIM 253200), or multiple sulfatase deficiency (OMIM 272200). Typically these disorders can be distinguished based upon clinical presentation. If clinically indicated, consider molecular genetics analysis of the ARSB and/or SUMF1 genes. Enzyme replacement therapy is available for individuals with mucopolysaccharidosis VI. Please contact the Biochemical Genetics consultant or genetic counselor on call (1-800-533-1710) if you have any questions.
Colorimetric Enzyme Assay

Reviewed By

Daniel Kraft

MCR

Cryopreserve for Biochem Studies

MCR

RECEIVED: 09/17/2013 11:03 REPORTED: 09/17/2013 11:03

Cryopreserve for Biochem Studies was cancelled on 09/17/2013 at 11:02; Test not performed.

Fibroblast Culture

MCR

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Fibroblast Culture was cancelled on 09/17/2013 at 11:02; Test not performed.

* 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, ARSB A	Collection Date and Time 09/17/2013 10:01	Report Status Final
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* Report times for Mayo performed tests are CST/CDT