



Patient ID SA00062392	Patient Name SAMPLEREPOR, ARSB A	Birth Date 1975-09-30	Gender F	Age 37
Order Number SA00062392	Client Order Number SA00062392	Ordering Physician Client, Client	Report Notes	
Account Information C7028846 DLMP Rochester		Collected 17 Sep 2013 10:01		

Arylsulfatase B, Fibroblasts

Arylsulfatase B, Fibroblasts



0.79 nmol/min/mg Prot

Reference Value
≥6.08

Low

MCR

Reviewed By

Daniel Kraft

MCR

Received: 17 Sep 2013 11:03

Reported: 17 Sep 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.

ADDITIONAL INFORMATION

Colorimetric Enzyme Assay

Cryopreserve for Biochem Studies

MCR

Cryopreserve for Biochem Studies was cancelled on 09/17/2013 at 11:03; Test not performed.

Received: 17 Sep 2013 11:03

Reported: 17 Sep 2013 11:03

Fibroblast Culture

MCR

Fibroblast Culture was cancelled on 09/17/2013 at 11:03; Test not performed.

Received: 17 Sep 2013 11:03

Reported: 17 Sep 2013 11:03

Performing Site Legend

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