





Patient ID SA00064434	Patient Name TESTINGRNV, TALDOABNORMAL	Birth Date 2003-01-02	Gender M	Age 10
Order Number SA00064434	Client Order Number SA00064434	Ordering Physician Client, Client	Report Notes	
Account Information C7028846 DLMP Rochester		Collected 12 Nov 2013 00:00		

Polyols, QN, U

Erythritol

 **350 mmol/mol Cr**


MCR
Reference Value
<171

Interpretation (TALDO)

MCR

In this sample, the excretions of arabinitol, erythritol, ribitol and sedoheptulose are elevated. This pattern is suggestive of Transaldolase (TALDO) deficiency (OMIM 602063). If clinically indicated, consider further confirmation by molecular genetic analysis of the TALDO1 gene. Please contact the Biochemical Genetics consultant or genetic counselor on call (1-800-533-1710) if you have any questions.

ADDITIONAL INFORMATION

Gas Chromatography-Mass Spectrometry (GC-MS) Stable Isotope Dilution Analysis

Reviewed By

MCR

JANICE HELGESON

Received: 13 Nov 2013 10:24



Reported: 13 Nov 2013 11:18

Arabinitol

 **201 mmol/mol Cr**




MCR
Reference Value
<97

Ribitol

 **99 mmol/mol Cr**


MCR
Reference Value
<17

Sedoheptulose

 **500 mmol/mol Cr**


MCR
Reference Value
<28

Performing Site Legend

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