




Patient ID <b>M0002252</b>	Patient Name <b>SAMPLEREPORT, STER A</b>	Birth Date <b>1971-09-28</b>	Gender <b>F</b>	Age <b>40</b>	
Order Number <b>M0002252</b>	Client Order Number <b>M0002252</b>	Ordering Physician <b>,</b>	Report Notes		
Account Information <b>C7028846 DLMP Rochester</b>		Collected <b>27 Jun 2012 01:28</b>			

**Sterols, P**

**Reason For Referral**

Reason for referral not provided.

**Desmosterol**

 **69.3 mg/L**  
High

**Lathosterol**

**5.1 mg/L**

**Campesterol**

**4.8 mg/L**

**Sitosterol**

**2.2 mg/L**

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Reference Value  
0.0-5.0

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Reference Value  
0.0-7.0

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Reference Value  
0.0-7.0

MCR

Reference Value  
0.0-5.0

**Interpretation**

MCR

In this sample, the concentration of desmosterol was elevated. Desmosterolosis is a condition caused by deficiency of 3-beta hydroxysterol-delta24-reductase, which presents with multiple malformations often resembling Smith-Lemli-Opitz syndrome and plasma levels of desmosterol >15 mg/L (see: FitzPatrick et al Am J Med Genet 1998, 75: 145-152). Consider sending another sample to repeat the analysis.

**ADDITIONAL INFORMATION**

Gas Chromatography-Mass Spectrometry (GC/MS)

**Reviewed By**

MCR

JANICE HELGESON

**Received:** 27 Jun 2012 01:28

**Reported:** 29 May 2013 12:40

**Performing Site Legend**

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