MAYO CLINIC
Mayo Medical Laboratories

Laboratory Service Report 1-800-533-1710

Patient Name	Patient ID	Age	Gender	Order #
SAMPLEREPORT,LDLM	0000180982	46	F	0000180982
Ordering Phys				**DOB** 06/10/1966
Client Order # 0000180982	**Account Information**			**Report Notes**
Collected 03/14/2013 22:27	C7028846-DLMP Rochester 3050 Superior Drive Rochester, MN 55901			
Printed 07/01/2013 11:15				

Test	Flag	Results	Unit	Reference Value	Perform Site*

LDLR Large Del/Dup
RECEIVED: 03/14/2013 22:27 **REPORTED:** 07/01/2013 11:11

Interp MCR

These results do not rule out the diagnosis of Familial Hypercholesterolemia (FH) or Familial Defective ApoB-100 (FDB) in this patient. Some individuals who have features of FH or FDB and involvement of LDLR or APOB may have a mutation that is not identified by the methods used here. In addition, some individuals with features of FH or FDB may have involvement of a gene other than LDLR or APOB (e.g. PCSK9).
Large deletion/duplication analysis of a portion of the promoter and all 18 exons of the LDLR gene (GenBank number NM_000527.3) was performed via multiplex ligation-dependent probe amplification (a PCR-based method).

A genetic consultation may be of benefit.

CAUTIONS:
Test results should be interpreted in the context of clinical findings, family history, and other laboratory data. Rare polymorphisms could interfere with test results. If results obtained do not match the clinical findings, additional testing should be considered.

Bone marrow transplants from allogenic donors will interfere with testing. Call Mayo Medical Laboratories for instructions for testing patients who have received a bone marrow transplant.
For research use only.

Reviewed By Jennifer Herman MCR

Result MCR

A mutation was not detected in LDLR.

* Performing Site:

MCR	Mayo Clinic Laboratories - Rochester Main Campus 200 First St SW Rochester, MN 55905	Lab Director:

Patient Name	Collection Date and Time	Report Status
SAMPLEREPORT,LDLM	03/14/2013 22:27	Final
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* Report times for Mayo performed tests are CST/CDT