



Patient ID <b>SA00062771</b>	Patient Name <b>TESTINGRNV, LDLM NORM</b>	Birth Date <b>1980-01-21</b>	Gender <b>F</b>	Age <b>33</b>
Order Number <b>SA00062771</b>	Client Order Number <b>SA00062771</b>	Ordering Physician <b>Client, Client</b>	Report Notes	
Account Information <b>C7028846 DLMP Rochester</b>		Collected <b>24 Sep 2013 08:59</b>		

## LDLR Large Del/Dup

### 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).

### ADDITIONAL INFORMATION

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:

Rare variants may be present that could lead to false negative or positive results. If results obtained do not match the clinical findings, additional testing should be considered.

Test results should be interpreted in the context of clinical

findings, family history, and other laboratory data. Misinterpretation of results may occur if the information provided is inaccurate or incomplete.

Samples may contain donor DNA if obtained from patients who received heterologous blood transfusions or allogeneic blood or marrow transplantation. Results from samples obtained under these circumstances may not accurately reflect the recipient's genotype. For individuals who have received blood transfusions, the genotype usually reverts to that of the recipient within 6 weeks. For individuals who have received allogeneic blood or marrow transplantation, a pre-transplant DNA specimen is recommended for testing.

### Reviewed By

MCR

Jamie Bruffat

### Result

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A mutation was not detected in FBN1.

**Received:** 25 Sep 2013 14:01

**Reported:** 25 Sep 2013 14:48

### Performing Site Legend

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