

Laboratory Service Report

1-800-533-1710

Patient Name SAMPLEREPORT,FSHOX	Patient ID SA00049769	Age 46	Gender M	Order # SA00049769
Ordering Phys		·		DOB 05/25/1966
Client Order # SA00049769	Account Information			Report Notes
Collected 10/14/2012 13:00	C7028846-DLMP ROCHESTER 3050 SUPERIOR DRIVE			
Printed 10/15/2012 13:04	ROCHESTER,MN 559	901		

Test Flag Results Unit Value Site*

REPORTED 10/15/2012 11:38

SHOXDNAdx

Y03

Molecular Genetic Assessment for SHOX Deficiency using Mutation Detection and SNP-Based Detection of Whole Gene Deletions by DHPLC.

Genomic DNA is extracted and PCR is used to amplify the SHOX coding exons and selected intragenic markers, SNPs, using custom-designed primers. Heteroduplexes are formed by heating and cooling the PCR-products. These products are then subjected to DHPLC. DHPLC chromatograms from heterozygous mutations, small deletions and small insertions are identified and then confirmed by DNA sequencing (Note that exon 6B is evaluated only by direct DNA sequencing).

Results:

SHOX No mutation detected

Interpretation:

No indication of SHOX deficiency was detected.

Comments

This test detects small mutations and whole gene deletions. This test does not identify partial SHOX gene deletions, most regulatory mutations, or mosaicism of the SHOX gene. These results should be considered in the context of this individual's clinical and family histories. Genetic counseling is recommended. Please contact the laboratory if you have any questions regarding this test or these results.

This test was developed and its performance characteristics determined by Labcorp. It has not been cleared or approved by the Food and Drug Administration. The FDA has determined that such clearance or approval is not necessary.

* Performing Site:

Y036	Esoterix Endocrinology 4301 Lost Hills Road Calabasas Hills, CA 91301	Lab Director:	

Patient Name	Collection Date and Time	Report Status
SAMPLEREPORT,FSHOX	10/14/2012 13:00	Final
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^{*} Report times for Mayo performed tests are CST/CDT