

Laboratory Service Report

1-800-533-1710

MCR

MCR

Patient Name SAMPLEREPORT,BMPMS	Patient ID SA00057777	Age 27	Gender M	Order # SA00057777
Ordering Phys CLIENT, CLIENT				DOB 06/15/1985
Client Order # SA00057777	Account Information			Report Notes
Collected 05/24/2013 00:00	C7028846-DLMP Rochester 3050 Superior Drive			
Printed 06/10/2013 13:52	Rochester, MN 55901			

Perform Reference Test Flag Results Unit Value Site*

BMPR1A Gene, Full Gene Analysis

RECEIVED: 05/24/2013 14:34 **REPORTED:** 06/10/2013 12:58

Reason For Referral

MCR Possible diagnosis of juvenile polyposis syndrome (JPS).

Test for the presence of a mutation in the BMPR1A gene.

Result

A mutation was NOT detected.

Interpretation MCR

This result decreases the likelihood but does not rule out the diagnosis of juvenile polyposis syndrome (JPS). We predict that some individuals who have a diagnosis of JPS and involvement of the BMPR1A gene may have a mutation that is not identified by the methods described above (e.g. promoter mutations, deep intronic mutations, etc.). In addition, this assay does not rule out mutations in other genes associated with JPS, such as SMAD4.

A genetic consultation may be of benefit.

Unless reported or predicted to cause disease, alterations found deep in the intron or alterations that do not result in an amino acid substitution are not reported. These and common polymorphisms identified for this patient are available upon request.

CAUTIONS:

Test results should be interpreted in context of clinical findings, family history, and other laboratory data. Misinterpretation of results may occur if the information provided is inaccurate or incomplete.

Rare polymorphisms exist that could lead to false negative or positive 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.

the presence of a mutation in all coding regions and

Laboratory developed test.

Method

Bi-directional sequence analysis was performed to test for

Performing Site Legend on Last Page of Report

Patient Name	Collection Date and Time	Report Status
SAMPLEREPORT,BMPMS	05/24/2013 00:00	Final
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^{*} Report times for Mayo performed tests are CST/CDT



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Test	Flag Results	Unit	Reference Value	Perform Site*
	of the BMPR1A gene. Array			
	ridization (aCGH) was used to test ge deletions and duplications.			
-	s based on GenBank accession number	r:		
NM 004329.2.	o succe on consum acception named	- /		
Array Billed?	Yes.			MCR
See COLDB, Hereditary Co	olon Cancer CGH Array, for billing			
information.				
Specimen	Blood			MCR
Reviewed By				MCR
Matthew John Ferber PhD				
Release Date	10 Jun 2013 12:56			MCR

* Performing Site:

MCR	Mayo Clinic Laboratories - Rochester Main Campus 200 First St SW Rochester, MN 55905	Lab Director: Franklin R. Cockerill, III, M.D.

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SAMPLEREPORT,BMPMS	05/24/2013 00:00	Final
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