Patient Name	Patient ID	Age	Gender	Order #
SAMPLEREPORT,BMPKM	SA00057780	37	F	SA00057780
Ordering Phys CLIENT,CLIENT				**DOB** 06/12/1975
Client Order # SA00057780	**Account Information**			**Report Notes**
Collected 05/24/2013 00:00	C7028846-DLMP Rochester 3050 Superior Drive Rochester, MN 55901			
Printed 06/28/2013 15:11				

Test	Flag	Results	Unit	Reference Value	Perform Site*

BMPR1A Gene, Known Mutation
RECEIVED: 05/24/2013 14:56 **REPORTED:** 06/10/2013 13:00

Reason For Referral — MCR

Family history of juvenile polyposis syndrome (JPS). Test for the presence of a mutation in the BMPR1A gene.

Result — MCR

The following deletion was detected:
Exon: 1
DNA change: c.44_47delTGTT
Amino acid change: p.L15SfsX20 (Leu15SerfsX20)
Classification: DELETERIOUS

Interpretation — MCR

This alteration is a known deleterious mutation.

This result is consistent with a diagnosis of juvenile polyposis syndrome (JPS) for this individual. Appropriate screening procedures and/or prophylactic measures should be considered.

Since a mutation has been identified, testing of at risk family members is possible.

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.

Performing Site Legend on Last Page of Report

MAYO CLINIC
Mayo Medical Laboratories

Laboratory Service Report 1-800-533-1710

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Test	Flag	Results	Unit	Reference Value	Perform Site*
Laboratory developed test.					
Method					MCR
DNA sequence analysis was used to test for the presence of the c.44_47delTGTT mutation in exon 1 of the BMPR1A gene. Testing was performed for this specific mutation because it was previously identified in an affected family member of this individual. Mutation nomenclature is based on GenBank accession number; NM_004329.2.					
Specimen		Blood			MCR
Reviewed By					MCR
Matthew John Ferber PhD					
Release Date		10 Jun 2013 12:58			MCR

* Performing Site:

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

* Report times for Mayo performed tests are CST/CDT