

Laboratory Service Report

1-800-533-1710

Patient Name SAMPLEREPORT,BMPMS	Patient ID SA00057778	Age 28	Gender F	Order # SA00057778
Ordering Phys CLIENT, CLIENT		>		DOB 01/12/1985
Client Order # SA00057778	Account Information			Report Notes
Collected 05/24/2013 00:00	C7028846-DLMP Roches 3050 Superior Drive	ster		
Printed 06/10/2013 13:56	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

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. Mutation-specific testing for BMPR1A-related JPS is available at Mayo Medical Laboratories by ordering BMPR1A/61486 BMPR1A Gene, Known Mutation. Please contact the Molecular Genetics Laboratory at 1-800-533-1710 with questions about this test.

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.

Performing Site Legend on Last Page of Report

Patient Name	Collection Date and Time	Report Status
SAMPLEREPORT,BMPMS	05/24/2013 00:00	Final
Page 1 of 2		>> Continued on Next Page >>



Laboratory Service Report

1-800-533-1710

Patient Name SAMPLEREPORT,BMPMS	Patient ID SA00057778	Age 28	Gender F	Order # SA00057778
Ordering Phys CLIENT,CLIENT				DOB 01/12/1985
Client Order # SA00057778	Account Information			Report Notes
Collected 05/24/2013 00:00	C7028846-DLMP Rocheste 3050 Superior Drive	er		
Printed 06/10/2013 13:56	Rochester, MN 55901			

Test	Flag Results	Unit	Reference Value	Perform Site*
with testing. Call Mayo	from allogenic donors will interfere Medical Laboratories for g patients who have received a bone			
Laboratory developed te	st.			
Method				MCR
the presence of a mutatintron/exon boundaries comparative genomic hybror the presence of large	analysis was performed to test for ion in all coding regions and of the BMPRIA gene. Array ridization (aCGH) was used to test ge deletions and duplications. s based on GenBank accession number;			
NM_004329.2. Array Billed?	Yes.			MCR
-	olon Cancer CGH Array, for billing			11010
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	Lab Director: Franklin R. Cockerill, III, M.D.
INICIX	200 First St SW Rochester MN 55905	Lab Director. Franklin IX. Cockerill, III, IVI.D.

Patient Name	Collection Date and Time	Report Status
SAMPLEREPORT,BMPMS	05/24/2013 00:00	Final
Page 2 of 2		** End of Report **