Test Definition: BMPRZ

BMPR1A Gene, Full Gene Analysis

Overview

Useful For

Confirmation of juvenile polyposis syndrome for patients with clinical features

Additional Tests

Test ID	Reporting Name	Available Separately	Always Performed
COLAB	Hereditary Colon Cancer CGH Array	Yes, (order FMTT)	Yes

Testing Algorithm

When this test is ordered, comparative genomic hybridization array will always be performed at an additional charge.

See Colonic Polyposis Syndromes Testing Algorithm in Special Instructions.

Special Instructions

- Molecular Genetics: Inherited Cancer Syndromes Patient Information
- Informed Consent for Genetic Testing
- Colonic Polyposis Syndromes Testing Algorithm
- Informed Consent for Genetic Testing (Spanish)

Method Name

Polymerase Chain Reaction (PCR) Amplification/DNA Sequencing

COLAB: Array Comparative Genomic Hybridization (aCGH)

NY State Available

Yes

Specimen

Specimen Type

Varies

Advisory Information

This test should be ordered only for individuals with symptoms suggestive of juvenile polyposis syndrome. Asymptomatic patients with a family history of juvenile polyposis syndrome **should not** be tested until a variant has been identified in an affected family member.

Shipping Instructions

Specimen preferred to arrive within 96 hours of collection.

Specimen Required

Patient Preparation: A previous bone marrow transplant from an allogenic donor will interfere with testing. Call 800-533-1710 for instructions for testing patients who have received a bone marrow transplant.

Specimen Type: Whole blood

Container/Tube:

Preferred: Lavender top (EDTA) or yellow top (ACD)

Acceptable: Any anticoagulant

Specimen Volume: 3 mL

Collection Instructions:

1. Invert several times to mix blood.

2. Send specimen in original tube.

Forms

1. **New York Clients-Informed consent is required.** Document on the request form or electronic order that a copy is on file. The following documents are available in Special Instructions:

-Informed Consent for Genetic Testing (T576)

-Informed Consent for Genetic Testing-Spanish (T826)

2. Molecular Genetics: Inherited Cancer Syndromes Patient Information (T519) in Special Instructions

Specimen Minimum Volume

1 mL

Reject Due To

All specimens will be evaluated at Mayo Clinic Laboratories for test suitability.

Specimen Stability Information

Specimen Type	Temperature	Time	Special Container
Varies	Ambient (preferred)		
	Frozen		
	Refrigerated		

Clinical and Interpretive

Clinical Information

Juvenile polyposis syndrome (JPS) is a rare hereditary cancer predisposition syndrome caused by alterations in the *SMAD4* or *BMPR1A* genes. JPS is characterized by the presence of multiple histologically defined juvenile polyps in the upper and/or lower gastrointestinal (GI) tract and an increased risk for GI cancers. Age of onset for cancer development is typically in the second or third decade of life, although some patients present with a more severe infantile-onset form of the disease. JPS is inherited in an autosomal dominant fashion, although a significant proportion of probands have no family history. Approximately 50% of patients with JPS have an identifiable alteration

in the *SMAD4* or *BMPR1A* genes.

Reference Values

An interpretive report will be provided.

Interpretation

All detected alterations are evaluated according to American College of Medical Genetics and Genomics (ACMG) recommendations.(1) Variants are classified based on known, predicted, or possible pathogenicity and reported with interpretive comments detailing their potential or known significance.

Cautions

Some individuals who are carriers or have a diagnosis of juvenile polyposis syndrome may have a variant that is not identified by this method (eg, promoter alterations). The absence of a variant, therefore, does not eliminate the possibility of positive carrier status or the diagnosis of juvenile polyposis syndrome. For carrier testing, it is important to first document the presence of a *BMPR1A* gene alteration in an affected family member.

It is strongly recommended that patients undergoing predictive testing receive genetic counseling both prior to testing and after results are available.

In some cases, DNA alterations of undetermined significance may be identified.

Rare alterations exist that could lead to false-negative or false-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. Errors in our interpretation of results may occur if information given is inaccurate or incomplete.

Clinical Reference

1. Richards S, Aziz N, Bale S, et al: Standards and guidelines for the interpretation of sequence variants: a joint consensus recommendation of the American College of Medical Genetics and Genomics and the Association for Molecular Pathology. Genet Med. 2015 May;17(5):405-424

2. Lammi L, Arte S, Somer M, et al: Mutations in *AXIN2* cause familial tooth agenesis and predispose to colorectal cancer. Am J Hum Genet. 2004;74:1043-1050

3. Liu W, Dong X, Mai M, et al: Mutations in *AXIN2* cause colorectal cancer with defective mismatch repair by activating beta-catenin/TCF signaling. Nat Genet. 2000;26:146-147

4. Mai M, Qian C, Yokomizo A, et al: Cloning of the human homolog of conductin (*AXIN2*), a gene mapping to chromosome 17q23-q24. Genomics. 1998;55:341-344

5. Dong X, Seelan RS, Qian C, et al: Genomic structure, chromosome mapping and expression analysis of the human *AXIN2* gene. Cytogenet Cell Genet. 2001;93:26-28

Performance

Method Description

Bidirectional sequence analysis is performed to test for the presence of a variant in all coding regions and intron/exon boundaries of the *BMPR1A* gene.(Unpublished Mayo method)

Additionally, array comparative genomic hybridization (aCGH) is used to test for the presence of large deletions and

duplications.(Aradhya S, Lewis R, Bonaga T, et al: Exon-level array CGH in a large clinical cohort demonstrates increased sensitivity of diagnostic testing for Mendelian disorders. Genet Med. 2012;14[6]:594-603)

PDF Report

No

Day(s) and Time(s) Test Performed

Performed weekly, Varies

Analytic Time

14 days

Maximum Laboratory Time

20 days

Specimen Retention Time

Whole Blood: 2 weeks (if available) Extracted DNA: 3 months

Performing Laboratory Location

Rochester

Fees and Codes

Fees

- Authorized users can sign in to Test Prices for detailed fee information.
- Clients without access to Test Prices can contact Customer Service 24 hours a day, seven days a week.
- Prospective clients should contact their Regional Manager. For assistance, contact Customer Service.

Test Classification

This test was developed and its performance characteristics determined by Mayo Clinic in a manner consistent with CLIA requirements. This test has not been cleared or approved by the U.S. Food and Drug Administration.

CPT Code Information

81479

Hereditary Colon Cancer CGH Array, additional test

81228

LOINC® Information

Test ID	Test Order Name	Order LOINC Value
BMPRZ	BMPR1A Gene, Full Gene Analysis	95769-6

Result ID	Test Result Name	Result LOINC Value
52479	Result Summary	50397-9
52480	Result	82939-0
52481	Interpretation	69047-9

Result ID	Test Result Name	Result LOINC Value
52482	Additional Information	48767-8
52483	Specimen	31208-2
52484	Source	31208-2
52485	Array Billed?	No LOINC Needed
52486	Released By	18771-6