Inhera™ Colorectal Cancer

Inhera™: Colorectal Cancer Final Report
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Intermountain® Precision Genomics

Patient

Patient Name: Donald Jones
Date of Birth: 04/25/1958
Accession ID: 11644-29-876-4032
Age: 62
Sex: Male

Specimen

Specimen Type: Buccal Swab
Collection Date: 11/11/2020
Received Date: 11/16/2020

Ordering Provider

Provider: THOMAS D. WISE
Report Date: December 17, 2020

Indication for Testing

C18.9 – malignant neoplasm of colon, unspecified

Test Result

Negative Result No pathogenic/likely pathogenic variants associated with the indication for testing were detected.

Test results reviewed and approved by:

Jeremy Wallentine, MD

December 17, 2020

Test Disclaimer

The following genes were evaluated by sequencing (*, including CNVs): *APC*, ATM*, BLM, BMPR1A*, CDH1*, CHEK2*, EPCAM*, MLH1*, MSH2*, MSH6*, MUTYH*, PMS2*, POLD1, POLE, PTEN*, SMAD4*, STK11*, TP53**. Positive CNV calls are confirmed by MLPA. Results are negative unless stated in the Test Results section of the report. Note: A negative result, meaning no DNA variants of consequence were detected, does not entirely exclude a diagnosis of a specific hereditary condition as some DNA variants (i.e., some intronic variants, large deletions and duplications, and chromosomal rearrangements) associated with the condition may not always be detected. Benign and likely benign variants are not included in this report. Variants in other genes not included in this study and nongenetic factors, including diet and lifestyle, may also influence this risk. We strongly recommend that you discuss these results with a genetic counselor and/or your health care provider.

Test Methodology

The Inhera™-Colorectal Cancer provides comprehensive coverage of 18 genes (*APC, ATM, BLM, BMPR1A, CDH1, CHEK2, EPCAM, MLH1, MSH2, MSH6, MUTYH, PMS2, POLD1, POLE, PTEN, SMAD4, STK11, TP53*) associated with an increased risk of inherited colorectal cancer. Genomic DNA (gDNA) is isolated from a patient specimen. The quality/quantity of the gDNA is assessed prior to Next Generation Sequencing (NGS) library preparation. NGS library preparation utilizes unique indexes to generate targeted libraries of approximately 500 base pairs (bp). The enrichment workflow enriches 350-650 bp centered symmetrically around the midpoint of the probe, providing coverage of exons (coding regions) and up to 50 bp of flanking intronic (non-coding) regions. All targeted regions are sequenced by NGS on Illumina's MiSeq or NextSeq platform. Reads are aligned to human genome reference sequence (GrCh37) using our in-house bioinformatics pipeline Hereditary Cancer Panel Pipeline version 3.3. During assay validation, Intermountain Precision Genomics (IPG) established the mean depth of coverage for all targeted regions to be > 500x. Sequence changes are identified using a clinical bioinformatics pipeline, and the clinical pathogenicity of each genetic variant is established based on the American College of Molecular Genetics (ACMG) scoring criteria[1]. To generate the low coverage table, we calculate the percentage of the gene that is covered below 30X for the regions covered by the assay. Low coverage is reported on exons that contribute to more than 5% of the gene covered below 30X. The bioinformatics pipeline includes a copy number variant (CNV) caller to detect large deletions and/or duplications in the genes listed above in bold. Any positive CNV calls are confirmed by MLPA (Multiplex Ligation-dependent Probe Amplification) analysis before reporting.

Regulatory Disclosures

This test was developed by Intermountain Precision Genomics (IPG). Its performance characteristics are determined by IPG. The methods have not been cleared or approved by the U.S. Food and Drug Administration. IPG is certified under CLIA-88 and accredited by the College of American Pathologists as qualified to perform high-complexity testing. This test is used for clinical purposes and should not be regarded as investigational or research.

References

Richards S, Aziz N, Bale S, Bick D, et al. Genetics in medicine : official journal of the American College of Medical Genetics. 2015, May. 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. (PMID: 25741868)