COLON CANCER (PROBAND)

London Health Sciences Centre | St. Joseph's Health Care London

Pathology and Laboratory Medicine

Orderable – E-order/Requisition

Turnaround Time: 4-6 weeks **STAT: 4 weeks**

Laboratory:
Molecular Diagnostics Lab

Requisition:
MOLECULAR DIAGNOSTIC REQUISITION

Method of Analysis:
All coding exons and 20 bp of flanking intronic sequence are enriched using an LHSC custom targeted hybridization protocol (Roche Nimblegen), followed by high throughput sequencing (Illumina). Sequence variants and copy number changes are assessed and interpreted using clinically validated algorithms and commercial software (SoftGenetics: Nextgene, Geneticist Assistant, Mutation Surveyor; and Alamut Visual). All exons have >300x mean read depth coverage, with a minimum 100x coverage at a single nucleotide resolution. This assay meets the sensitivity and specificity of combined Sanger sequencing and

Alternate Name(s):

Hereditary Colorectal/Gastric Cancer

Specimen:

Whole Blood-3 x 4 mL Lavender EDTA top Vacutainer tube

Collection Information:

Blood samples must be maintained at room temperature.

Reference Ranges:

See report

Interpretive Comments:

The Hereditary Colorectal/Gastric Cancer Panel (HCP-Colorectal/Gastric) is a genetic test designed to help assess the cancer predisposition risk for colorectal, pancreatic, melanoma, prostate, and endometrial cancers. This panel includes the 23 genes listed above. Mutations in these genes are associated with clinically actionable results which will directly impact medical management recommendations and disease risk assessment.

Comments:

Genes Tested (hg19;HGVS nomenclature):APC and5'UTR(NM_001127510.2), BMPR1A(NM_004329.2), CDH1(NM_004360.3), CDK4(NM_000075.3), CHEK2(NM_007194.3), CTNNA1(NM_001903.2), EPCAM(NM_002354.2:3' large del only), FLCN(NM_144997.5), GREM1(NM_013372.6), MLH1 and5'UTR(NM_000249.3), MSH2(NM_000251.2), MSH3(NM_002439.4), MSH6(NM_000179.2), MUTYH(NM_001128425.1), NTHL1(NM_002528.5), PMS2(NM_000535.5),

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POLD1(NM_001256849.1), POLE(NM_006231.2), PTEN and5'UTR(NM_000314.4), SDHB(NM_003000.2), SMAD4(NM_005359.5), STK11(NM_000455.4), TP53(NM_000546.5)

Critical Information Required:

Restricted to cancer genetics only.
Pedigree required.

Storage and Shipment:

Must be received in testing laboratory within 48 hours of collection, shipped at room temperature by courier/overnight delivery.

MLPA copy number analysis. All variants interpreted as either ACMG category 1, 2, or 3 (pathogenic, likely pathogenic, VUS; PMID: 25741868) are confirmed using Sanger sequencing, MLPA, or other assays. ACMG category 4 and 5 variants (likely benign, benign) are not reported, but are available upon request. This assay has been validated at a level of sensitivity equivalent to the Sanger sequencing and standard copy number analysis (>99%; PMID: 27376475,28818680).

Test Schedule:
As required,
Monday to Friday 0800-1600 hours