

# Clinicians Guide to: Genetic Testing for Hereditary Cancer



Fulgent offers both comprehensive and focused genetic testing for over 15 types of hereditary cancer. Results of this testing can help establish or confirm a diagnosis, guide personalized medical management, and identify risk for other cancers or conditions – while also informing close biological relatives of their own potential risk factors.

## WHO IS THIS TEST FOR?

Patients with a personal or family history suggestive of a hereditary cancer syndrome. Red flags for hereditary cancers can include onset of cancer before age 50, more than one primary cancer in a single person, or multiple affected people within a family. This test is designed to detect germline pathogenic variants and is not validated to detect somatic mutations.

# SHOULD I ORDER A FOCUSED PANEL OR A COMPREHENSIVE PANEL?

**Fulgent Focus Cancer** panels cover up to 50 genes associated with high/moderate risk for hereditary cancer syndromes and established clinical management guidelines for pathogenic variants. These tests maximize the chances of identifying a pathogenic cancer susceptibility variant, while minimizing the number of variants of uncertain clinical significance found.

**Fulgent Comprehensive Cancer** panels cover up to 154 genes associated with potential risk for hereditary cancer syndromes. These panels include well-established genes as well as candidate genes that may have been recently discovered or for which additional research is needed. These tests maximize the chances of identifying a pathogenic cancer susceptibility variant, while providing a comprehensive review of candidate genes to give you information you can use now and potentially in the future.

## **TEST SPECIFICATIONS**

#### Sample Requirements (kits available upon request):

- Blood, two 4-mL EDTA tubes lavender top (preferred)
- Extracted DNA, 3 µg in TE buffer
- Buccal swab or saliva

Coverage\* ~99% at 50x Turnaround Time\*\* 2-3 weeks

\* Full gene sequencing and deletion/duplication, including detection of MSH2 inversion for select panels, BRCA2 Alu variant, and PMS2 (including exons 1-5 and 12-15), as well as sequencing of noncoding regions for selected genes. Please contact us for more details.

\*\*Rush order available upon request

# WHICH PANEL SHOULD I ORDER?

Hereditary cancer testing can establish or confirm a diagnosis, identify risks for other cancers and health conditions, help define the best management plan for patients with a positive test result (including increased surveillance, pharmacological treatments, and prophylactic surgeries), and determine whether close family members (children, siblings, and parents) may also be at risk. If a pathogenic variant is identified in your patient, their close relatives can have as high as a 50 percent risk of carrying the same variant and therefore also being at risk of developing cancer.



# **AVAILABLE PANELS**

#### Full Focus Cancer (50 Genes)

APC, ATM, AXIN2, BAP1, BARD1, BMPR1A, BRCA1, BRCA2, BRIP1, CDH1, CDKN1B, CDKN2A, CHEK2, EPCAM, FH, FLCN, GREM1, HOXB13, MAX, MBD4, MEN1, MET, MLH1, MLH3, MSH2, MSH3, MSH6, MUTYH, NF1, NTHL1, PALB2, PMS2, POLD1, POLE, PTEN, RAD51C, RAD51D, RET, RNF43, SDHA, SDHB, SDHC, SDHD, SMAD4, STK11, TMEM127, TP53, TSC1, TSC2, VHL

<b>Custom Focus Cancer (Up to 50 Genes)</b> Choose any combination of genes from the Focus Cancer Panel Gene list.	
Breast and Ovarian Cancer Focus (20 Genes)	Lynch Focus (5 Genes)
ATM, BARD1, BRCA1, BRCA2, BRIP1, CDH1, CHEK2, EPCAM, MLH1, MSH2, MSH6, MUTYH, NF1, PALB2, PMS2, PTEN, RAD51C, RAD51D, STK11, TP53	EPCAM, MLH1, MSH2, MSH6, PMS2
Breast Focus (13 Genes)	Ovarian Focus (14 Genes)
ATM, BARD1, BRCA1, BRCA2, CDH1, PTEN, STK11, TP53, CHEK2, PALB2, NF1, RAD51C, RAD51D	BRCA1, BRCA2, BRIP1, EPCAM, MLH1, MSH2, MSH6, MUTYH, PALB2, PMS2, RAD51C, RAD51D, STK11, TP53
Breast Cancer STAT Panel (9 Genes)	Prostate Focus (13 Genes)
The Breast Cancer STAT Panel examines 9 genes strongly associated with an increased risk for hereditary breast cancer and provides a more rapid turnaround time for surgical interventions.	ATM, BRCA1, BRCA2, CHEK2, EPCAM, HOXB13, MLH1, MSH2, MSH6, NBN, PALB2 PMS2, TP53
Genes: ATM, BRCA1, BRCA2, CDH1, CHEK2, PALB2, PTEN, STK11, TP53	
<ul> <li>Turnaround Time: 10 days (additional confirmatory testing may affect turnaround time)</li> </ul>	
Colorectal Focus (23 Genes)	
APC, AXIN2, BMPR1A, CDH1, CHEK2, EPCAM, GREM1, MBD4, MLH1, MLH3, MSH2, MSH3, MSH6, MUTYH, NTHL1, PMS2, POLD1, POLE, PTEN, RNF43, SMAD4, STK11, TP53	-

### Full Comprehensive Cancer (154 Genes)

ABRAXAS1, AIP, AKT1, ALK, ANKRD26, APC, ATM, ATR, AXIN2, BAP1, BARD1, BLM, BMPR1A, BRCA1, BRCA2, BRIP1, BUB1B, CASR, CDC73, CDH1, CDK4, CDKN1B, CDKN1C, CDKN2A, CEBPA, CEP57, CHEK2, CTC1, CTNNA1, CTRC, CYLD, DDB2, DDX41, DICER1, DIS3L2, DKC1, EGFR, EGLN1, ELANE, ENG, EPCAM, ERCC1, ERCC2, ERCC3, ERCC4, ERCC5, EXO1, EXT1, EXT2, EZH2, FAN1, FANCA, FANCB, FANCC, FANCD, FANCE, FANCF, FANCG, FANCI, FANCL, FANCM, FH, FLCN, GALNT12, GATA2, GEN1, GPC3, GREM1, HOXB13, HRAS, IKZF1, KIF1B, KIT, LZTR1, MAX, MBA4, MEN1, MET, MITF, MLH1, MLH3, MRE11, MSH2, MSH3, MSH6, MUTYH, NBN, NF1, NF2, NHP2, NOP10, NTHL1, PALB2, PALLD, PAX5, PDGFRA, PHOX2B, PIK3CA, PMS2, POLD1, POLE, POLH, POT1, PRKAR1A, PRS51, PTCH1, PTCH2, PTEN, RAD50, RAD51C, RAD51D, RB1, RECQL, RECQL4, REST, RET, RHBDF2, RNF43, RPS20, RUNX1, SAMD9L, SBDS, SDHA, SDHAF2, SDHB, SDHC, SDHD, SLC45A2, SLX4, SMAPA4, SMARCB1, SMARCE1, SPINK1, SRP72, STK11, SUFU, TERC, TERT, TGFBR1, TINF2, TMEM127, TP53, TRIP13, TSC1, TSC2, TYR, VHL, WRAP53, WRN, WT1, XPA, XPC, XRCC2

Custom Comprehensive Cancer (Up to 154 Genes) Choose any combination of genes from the Comprehensive Cancer Panel Gene list.	
Breast Comprehensive (37 Genes) ABRAXAS1, AKT1, ATM, BARD1, BLM, BRCA1, BRCA2, BRIP1, CDH1, CHEK2, CTNNA1, DICER1, EPCAM, FANCC, FANCM, MLH1, MRE11, MSH2, MSH6, MUTYH, NBN, NF1, NTHL1, PALB2, PIK3CA, PMS2, PTEN, RAD50, RAD51C, RAD51D, RECQL, SDHB, SDHD, SMARCA4, STK11, TP53, XRCC2	Ovarian Comprehensive (20 Genes) ATM, BARD1, BRCA1, BRCA2, BRIP1, CDH1, EPCAM, MLH1, MRE11, MSH2, MSH6, MUTYH, NBN, PALB2, PMS2, RAD51C, RAD51D, SMARCA4, STK11, TP53
Breast and Ovarian Comprehensive (37 Genes)	Pancreatic Comprehensive (24 Genes)
ABRAXAS1, AKT1, ATM, BARD1, BLM, BRCA1, BRCA2, BRIP1, CDH1, CHEK2, CTNNA1, DICER1, EPCAM, FANCC, FANCM, MLH1, MRE11, MSH2, MSH6, MUTYH, NBN, NF1, NTHL1, PALB2, PIK3CA, PMS2, PTEN, RAD50, RAD51C, RAD51D, RECQL, SDHB, SDHD, SMARCA4, STK11, TP53, XRCC2	APC, ATM, BMPR1A, BRCA1, BRCA2, BUB1B, CDK4, CDKN2A, EPCAM, FANCA, FANCC, MEN1, MLH1, MSH2, MSH6, NF1, PALB2, PMS2, SMAD4, STK11, TP53, TSC1, TSC2, VHL
Colorectal Comprehensive (29 Genes)	Paraganglioma-Pheochromocytoma Comprehensive (14 Genes)
APC, ATM, AXIN2, BLM, BMPR1A, CDH1, CHEK2, CTNNA1, EPCAM, FAN1, GALNT12, GREM1, MBD4, MLH1, MLH3, MSH2, MSH3, MSH6, MUTYH, NTHL1, PMS2, POLD1, POLE, PTEN, RNF43, RPS20, SMAD4, STK11, TP53	EGLN1, FH, KIF1B, MAX, MEN1, NF1, RET, SDHA, SDHAF2, SDHB, SDHC, SDHD, TMEM127, VHL
Endometrial Comprehensive (15 Genes)	Prostate Comprehensive (20 Genes)
BRCA1, BRCA2, CHEK2, EPCAM, MLH1, MSH2, MSH6, MUTYH, PMS2, POLD1, POLE, PTEN, SMARCA4, STK11, TP53	ABRAXAS1 ATM, ATR, BRCA1, BRCA2, BRIP1, CHEK2, EPCAM, GEN1, HOXB13, MLH1, MRE11, MSH2, MSH6, NBN, PALB2, PMS2, RAD51C, RAD51D, TP53
Gastric Comprehensive (22 Genes)	Renal/Urinary Comprehensive (30 Genes)
APC, BMPR1A, BRCA1, BRCA2, CDH1, CTNNA1, EPCAM, KIT, MLH1, MSH2, MSH6, NF1, PDGFRA, PMS2, PTEN, SDHA, SDHB, SDHC, SDHD, SMAD4, STK11, TP53	BAP1, CDC73, CDKN1C, CHEK2, DICER1, DIS3L2, EPCAM, FH, FLCN GPC3, MET, MITF, MLH1, MSH2, MSH6, MUTYH, PMS2, PTEN, REST, SDHA, SDHB, SDHC, SDHD, SMARCA4, SMARCB1, TP53, TSC1, TSC2, VHL, WT1
Hematologic Malignancy Comprehensive (22 Genes)	Sarcoma Comprehensive (30 Genes)
ATM, BLM, CEBPA, DDX41, EPCAM, GATA2, HRAS, IKZF1, MLH1, MSH2, MSH6, NBN, NF1, PAX5, PMS2, RUNX1, SAMD9L, SRP72, TERC, TERT, TP53	APC, BLM, CDKN1C, DICER1, EPCAM, EXT1, EXT2, FH, HRAS, KIT, MLH1, MSH2, MSH6, NBN, NF1, PDGFRA, PMS2, POT1, PRKAR1A, PTCH1, PTEN, RB1, RECQL4, SDHA, SDHB, SDHC, SDHD, SUFU, TP53, WRN
Melanoma Comprehensive (14 Genes)	Thyroid Comprehensive (10 Genes)
BAP1, BRCA2, CDK4, CDKN2A, CHEK2, MITF, MUTYH, POT1, PTEN, RB1, SLC45A2 TERT, TP53, TYR	AKT1, APC, CHEK2, DICER1, MEN1, PIK3CA, PTEN, PRKAR1A, RET, TP53
Nervous System/Brain Comprehensive (32 Genes)	
AIP, ALK, APC, ATM, CDKN1B, CDKN2A, DICERI, EPCAM, HRAS, KIF1B, LZTR1, MEN1, MLH1, MSH2, MSH6, NBN, NF1, NF2, PHOX2B, PMS2, POT1, PRKAR1A, PTCH1, PTEN, SMARCA4, SMARCB1, SMARCE1, SUFU, TP53, TSC1, TSC2, VHL	-

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