

## INVITAE HEREDITARY CANCER GENE PANEL TESTS

Invitae offers the broadest oncology testing menu, including guidelines-based panels for breast, gynecologic, and colorectal cancers; breast cancer STAT panels; and other tests for hereditary risk of brain, breast and gynecologic, endocrine, gastrointestinal, genitourinary, hematologic, pediatric, and skin cancers.

### CLINICAL AREA: HEREDITARY CANCER

#### FREQUENTLY ORDERED PANELS

##### CROSS-CANCER PANELS

Invitae Multi-Cancer Panel (80 genes)

##### Primary panel (80 genes)

ALK	APC	ATM	AXIN2	BAP1	BARD1	BLM	BMPR1A	BRCA1	BRCA2	BRIP1	CASR
CDC73	CDH1	CDK4	CDKN1B	CDKN1C	CDKN2A	CEBPA	CHEK2	DICER1	DIS3L2	EGFR	EPCAM
FH	FLCN	GATA2	GPC3	GREM1	HOXB13	HRAS	KIT	MAX	MEN1	MET	MITF
MLH1	MSH2	MSH6	MUTYH	NBN	NF1	NF2	PALB2	PDGFRA	PHOX2B	PMS2	POLD1
POLE	POT1	PRKAR1A	PTCH1	PTEN	RAD50	RAD51C	RAD51D	RB1	RECQL4	RET	RUNX1
SDHA	SDHAF2	SDHB	SDHC	SDHD	SMAD4	SMARCA4	SMARCB1	SMARCE1	STK11	SUFU	TERC
TERT	TMEM127	TP53	TSC1	TSC2	VHL	WRN	WT1				

Invitae Common Hereditary Cancers Panel (Breast, Gyn, GI) (42 genes)

##### Primary panel (42 genes)

APC	ATM	AXIN2	BARD1	BMPR1A	BRCA1	BRCA2	BRIP1	CDH1	CDKN2A	CHEK2	DICER1
EPCAM	GREM1	KIT	MEN1	MLH1	MSH2	MSH6	MUTYH	NBN	NF1	PALB2	PDGFRA
PMS2	POLD1	POLE	PTEN	RAD50	RAD51C	RAD51D	SDHA	SDHB	SDHC	SDHD	SMAD4
SMARCA4	STK11	TP53	TSC1	TSC2	VHL						

#### STAT TURNAROUND TIME

Invitae Breast Cancer STAT Panel (up to 9 genes)

##### Primary panel (7 genes)

BRCA1	BRCA2	CDH1	PALB2	PTEN	STK11	TP53
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##### Add-on ATM gene (1 gene)

ATM

##### Add-on CHEK2 gene (1 gene)

CHEK2

Invitae BRCA1 and BRCA2 STAT Panel (2 genes)

##### Primary panel (2 genes)

BRCA1	BRCA2
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#### CANCER PANELS BY ORGAN SYSTEM

##### BREAST CANCER

Invitae Breast Cancer Guidelines-Based Panel (11 genes)

##### Primary panel (11 genes)

ATM	BRCA1	BRCA2	CDH1	CHEK2	NBN	NF1	PALB2	PTEN	STK11	TP53
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Invitae Breast Cancer Panel (up to 26 genes)

##### Primary panel (14 genes)

ATM	BARD1	BRCA1	BRCA2	BRIP1	CDH1	CHEK2	NBN	NF1	PALB2	PTEN	RAD50
STK11	TP53										

##### Add-on preliminary-evidence genes (12 genes)

AKT1	FAM175A	FANCC	MRE11	MUTYH	PIK3CA	RAD51C	RAD51D	RINT1	SDHB	SDHD	XRCC2
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##### BREAST AND GYNECOLOGIC CANCERS

Invitae Breast and Gyn Cancers Guidelines-Based Panel (19 genes)

##### Primary panel (19 genes)

ATM	BRCA1	BRCA2	BRIP1	CDH1	CHEK2	EPCAM	MLH1	MSH2	MSH6	NBN	NF1
PALB2	PMS2	PTEN	RAD51C	RAD51D	STK11	TP53					

Invitae Breast and Gyn Cancers Panel (up to 35 genes)

##### Primary panel (23 genes)

ATM	BARD1	BRCA1	BRCA2	BRIP1	CDH1	CHEK2	DICER1	EPCAM	MLH1	MSH2	MSH6
NBN	NF1	PALB2	PMS2	PTEN	RAD50	RAD51C	RAD51D	SMARCA4	STK11	TP53	

##### Add-on preliminary-evidence genes (12 genes)

AKT1	CDC73	FAM175A	FANCC	MRE11	MUTYH	PIK3CA	POLD1	RINT1	SDHB	SDHD	XRCC2
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##### COLORECTAL CANCER

Invitae Colorectal Cancer Guidelines-Based Panel (up to 18 genes)

##### Primary panel (12 genes)

APC	BMPR1A	EPCAM	MLH1	MSH2	MSH6	MUTYH	PMS2	PTEN	SMAD4	STK11	TP53
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##### Add-on guidelines genes with emerging data genes (6 genes)

BLM	CHEK2	GALNT12	GREM1	POLD1	POLE
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Invitae Colorectal Cancer Panel (up to 25 genes)

##### Primary panel (18 genes)

APC	AXIN2	BMPR1A	CDH1	CHEK2	EPCAM	GREM1	MLH1	MSH2	MSH6	MUTYH	PMS2
POLD1	POLE	PTEN	SMAD4	STK11	TP53						

##### Add-on preliminary-evidence genes (7 genes)

ATM	BLM	BUB1B	ENG	FLCN	GALNT12	MLH3
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#### ADDITIONAL ORGAN SYSTEMS

Invitae Gastric Cancer Panel (up to 19 genes)

##### Primary panel (18 genes)

APC	BMPR1A	CDH1	EPCAM	KIT	MLH1	MSH2	MSH6	NF1	PDGFRA	PMS2	SDHA
SDHB	SDHC	SDHD	SMAD4	STK11	TP53						

##### Add-on preliminary-evidence gene (1 gene)

CTNNA1



## INVITAE CANCER GENE PANEL TESTS (continued)

### PEDIATRIC ONCOLOGY (continued)

Invitae Pediatric Nervous System/Brain Tumors Panel (up to 32 genes)	<b>Primary panel (24 genes)</b>											
	ALK	APC	DICER1	EPCAM	HRAS	MEN1	MLH1	MSH2	MSH6	NF1	NF2	PHOX2B
	PMS2	PRKAR1A	PTCH1	PTEN	RB1	SMARCB1	SMARCE1	SUFU	TP53	TSC1	TSC2	VHL
	<b>Add-on hereditary paraganglioma pheochromocytoma genes (8 genes)</b>											
	MAX	RET	SDHA	SDHAF2	SDHB	SDHC	SDHD	TMEM127				

### INDIVIDUAL HEREDITARY CANCER CONDITIONS

Invitae Ataxia-Telangiectasia Test (1 gene)	<b>Primary panel (1 gene)</b> ATM											
Invitae BAP1 Hereditary Cancer Predisposition Syndrome Test (1 gene)	<b>Primary panel (1 gene)</b> BAP1											
Invitae Basal Cell Nevus Syndrome Test (up to 3 genes)	<b>Primary panel (2 genes)</b> PTCH1 SUFU											
	<b>Add-on preliminary-evidence gene (1 gene)</b> PTCH2											
Invitae Birt-Hogg-Dubé Syndrome Test (1 gene)	<b>Primary panel (1 gene)</b> FLCN											
Invitae Bloom Syndrome Test (1 gene)	<b>Primary panel (1 gene)</b> BLM											
Invitae Carney Complex Test (1 gene)	<b>Primary panel (1 gene)</b> PRKAR1A											
Invitae CASR-Related Conditions Test (1 gene)	<b>Primary panel (1 gene)</b> CASR											
Invitae CDC73-Related Conditions Test (1 gene)	<b>Primary panel (1 gene)</b> CDC73											
Invitae Chronic Pancreatitis Panel (5 genes)	<b>Primary panel (5 genes)</b> CASR CFTR CTRC PRSS1 SPINK1											
	<b>Primary panel (5 genes)</b> EPCAM MLH1 MSH2 MSH6 PMS2											
Invitae Costello Syndrome Test (1 gene)	<b>Primary panel (1 gene)</b> HRAS											
Invitae DICER1 Syndrome Test (1 gene)	<b>Primary panel (1 gene)</b> DICER1											
Invitae Familial Acute Myeloid Leukemia with Mutated CEBPA Test (1 gene)	<b>Primary panel (1 gene)</b> CEBPA											
Invitae Familial Adenomatous Polyposis Test (1 gene)	<b>Primary panel (1 gene)</b> APC											
Invitae Familial Gastrointestinal Stromal Tumor Syndrome Panel (7 genes)	<b>Primary panel (7 genes)</b> KIT NF1 PDGFRA SDHA SDHB SDHC SDHD											
	<b>Primary panel (2 genes)</b> ALK PHOX2B											
Invitae Familial Neuroblastoma Panel (up to 3 genes)	<b>Add-on preliminary-evidence gene (1 gene)</b> KIF1B											
	<b>Primary panel (1 gene)</b> RUNX1											
Invitae Fanconi Anemia Panel (17 genes)	<b>Primary panel (17 genes)</b> BRCA2 BRIP1 ERCC4 FANCA FANCB FANCC FANCD2 FANCE FANCF FANCG FANCI FANCL FANCM PALB2 RAD51C SLX4 XRCC2											
	<b>Primary panel (1 gene)</b> GATA2											
Invitae Hereditary Breast and Ovarian Cancer Syndrome Panel (2 genes)	<b>Primary panel (2 genes)</b> BRCA1 BRCA2											
Invitae Hereditary Diffuse Gastric Cancer Syndrome Test (1 gene)	<b>Primary panel (1 gene)</b> CDH1											
Invitae Hereditary Leiomyomatosis and Renal Cell Cancer Test (1 gene)	<b>Primary panel (1 gene)</b> FH											
Invitae Hereditary Papillary Renal Cell Carcinoma Test (1 gene)	<b>Primary panel (1 gene)</b> MET											
Invitae Juvenile Polyposis Syndrome Panel (2 genes)	<b>Primary panel (2 genes)</b> BMPR1A SMAD4											
Invitae Li-Fraumeni Syndrome Test (1 gene)	<b>Primary panel (1 gene)</b> TP53											
Invitae Lynch Syndrome Panel (5 genes)	<b>Primary panel (5 genes)</b> EPCAM MLH1 MSH2 MSH6 PMS2											

## INVITAE CANCER GENE PANEL TESTS (continued)

### INDIVIDUAL HEREDITARY CANCER CONDITIONS (continued)

Invitae Melanoma-Pancreatic Cancer Syndrome Panel (2 genes)	Primary panel (2 genes) CDK4 CDKN2A
Invitae Multiple Endocrine Neoplasia Type 1 Test (1 gene)	Primary panel (1 gene) MEN1
Invitae Multiple Endocrine Neoplasia Type 2 Test (1 gene)	Primary panel (1 gene) RET
Invitae MUTYH-Associated Polyposis Syndrome Test (1 gene)	Primary panel (1 gene) MUTYH
Invitae Neurofibromatosis Type 1 Test (up to 2 genes)	Primary panel (1 gene) NF1  Add-on Legius syndrome gene (1 gene) SPRED1
Invitae Neurofibromatosis Type 2 Test (up to 2 genes)	Primary panel (1 gene) NF2  Add-on schwannomatosis gene (1 gene) SMARCB1
Invitae Nijmegen Breakage Syndrome Test (1 gene)	Primary panel (1 gene) NBN
Invitae Oligodontia-Colorectal Cancer Syndrome Test (1 gene)	Primary panel (1 gene) AXIN2
Invitae Perlman Syndrome Test (1 gene)	Primary panel (1 gene) DIS3L2
Invitae Peutz-Jeghers Syndrome Test (1 gene)	Primary panel (1 gene) STK11
Invitae PTEN-Related Disorders Test (1 gene)	Primary panel (1 gene) PTEN
Invitae RECQL4-Related Disorders Test (1 gene)	Primary panel (1 gene) RECQL4
Invitae Retinoblastoma Test (1 gene)	Primary panel (1 gene) RB1
Invitae Rhabdoid Tumor Predisposition Syndrome Panel (2 genes)	Primary panel (2 genes) SMARCA4 SMARCB1
Invitae Schwannomatosis Test (up to 2 genes)	Primary panel (1 gene) SMARCB1  Add-on neurofibromatosis type 2 gene (1 gene) NF2
Invitae Simpson-Golabi-Behmel Syndrome Test (1 gene)	Primary panel (1 gene) GPC3
Invitae Small Cell Carcinoma of the Ovary Hypercalcemic Type Test (1 gene)	Primary panel (1 gene) SMARCA4
Invitae Tuberous Sclerosis Complex Panel (2 genes)	Primary panel (2 genes) TSC1 TSC2
Invitae von Hippel-Lindau Syndrome Test (1 gene)	Primary panel (1 gene) VHL
Invitae Weaver Syndrome Test (1 gene)	Primary panel (1 gene) EZH2
Invitae Werner Syndrome Test (1 gene)	Primary panel (1 gene) WRN
Invitae Wilms Tumor Panel (4 genes)	Primary panel (4 genes) CDKN1C DIS3L2 GPC3 WT1
Invitae WT1-Related Disorders Test (1 gene)	Primary panel (1 gene) WT1



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