Sunnybrook
ODETTE CANCER CENTRE
A Cancer Care Ontario Partner

Hereditary Breast, Ovarian, Prostate and Gastrointestinal Cancer Gene Panel (36 genes)

Everyone has two copies of each gene listed below. Normally, these genes help to protect us from getting cancer. If a person inherits a change, called a pathogenic variant (mutation) from one of their parents, their chance of getting cancer is increased.

GENES*	LIFETIME CANCER and/or TUMOUR RISK(S)
APC	Colon, small intestine, stomach, thyroid, pancreatic, brain, liver, gastrointestinal polyps
ATM**	Breast; Possible pancreatic, prostate and colon
BMPR1A SMAD4	Colon, stomach, small intestine, pancreatic, gastrointestinal polyps
BARD1	Breast; Possible ovarian
BRCA1	Breast, ovarian/fallopian tube/primary peritoneal, prostate, male breast, pancreatic
BRCA2	Breast, ovarian/fallopian tube/primary peritoneal, prostate, male breast, pancreatic; Possible melanoma (form of skin cancer)
BRIP1	Ovarian; Possible breast and prostate
CDH1	Stomach/diffuse gastric, breast, colon
CDKN2A	Melanoma, pancreatic
CHEK2**	Breast, male breast, prostate, colon; Possible thyroid, ovarian and kidney
CTNNA1	Stomach/diffuse gastric
GALNT12	Colon polyps
GREM1	Colon, colon polyps of mixed histology (common founder Ashkenazi Jewish mutation)
HOXB13	Prostate
MLH1	Colon, uterine, ovarian, stomach, small intestine, pancreas, prostate, urinary tract, bladder, hepatobiliary tract; Possible brain
MSH2 EPCAM	Colon, uterine, prostate, ovarian, bladder, urinary tract, stomach, small intestine; Possible pancreas and brain
MSH6	Colon uterine, ovarian, urinary tract, prostate, stomach, small intestine, bladder; Possible hepatobiliary tract and pancreas
PMS2	Colon uterine; Possible prostate, ovarian, urinary tract, stomach, small intestine, hepatobiliary tract, brain
MLH3	Colon; possible other Lynch syndrome associated risk
MSH3	Colon, colon polyps (adenomas). Note: cancer risk only associated with having two MSH3 mutations
MUTYH	Colon, small intestinal, gastrointestinal polyps; Possible breast. Note: cancer risk only associated with having two MUTYH mutations (called biallelic).
NTHL1	Colon, colon polyps (adenomas). Note: cancer risk only associated with having two NTHL1 mutations
PALB2	Breast, pancreatic, male breast; Possible ovarian
POLD1	Colon, endometrial (lining of uterus), colon polyps
POLE	Colon, gastrointestinal polyps
PTEN	Breast, thyroid, uterine, kidney, colon, melanoma (form of skin cancer), gastrointestinal polyps
RAD51C	Ovarian; Possible breast and prostate
RAD51D	Ovarian; Possible breast and prostate
RPS20	Colon polyps
RNF43	Colon polyps
SDHB	Paraganglioma/pheochromocytoma, kidney, gastrointestinal stromal tumour (GIST)

Last updated: August 2021

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SDHD	Paraganglioma/pheochromocytoma, kidney, gastrointestinal stomal tumour (GIST), thyroid
STK11	Colon, breast, pancreatic, stomach, small intestine, ovarian tumours, lung, cervix, uterine, testicular tumours
TP53	Breast, sarcoma (bone and soft tissue), brain, adrenocortical, leukemia and others

*****Cancer syndromes connected to above genes: Familial Adenomatous Polyposis (APC), Juvenile polyposis syndrome (BMPR1A/SMAD4), Hereditary breast and ovarian cancer (BRCA1 and BRCA2), Hereditary diffuse gastric cancer (CDH1), Familial atypical multiple mole melanoma (CDKN2A), Hereditary mixed polyposis syndrome (GREM1/SCG5), Lynch syndrome (MLH1, MSH2, EPCAM, MSH6 and PMS2), MSH3-associated polyposis (2 MSH3 mutations), MUTYH-associated polyposis (2 MUTYH mutations), NTHL1-associated polyposis (2 NTHL1 mutations), Polymerase proofreading-associated polyposis (POLD1 and POLE), Cowden syndrome (PTEN), Peutz-Jeghers syndrome (STK11) and Li Fraumeni syndrome (TP53).
****** Risks may vary based on family history and/or specific gene mutation.

Genetic Testing:

Genetic Testing
Blood sample (or other DNA sample) is tested for mutations in the above gene(s).

○ **Predictive:** Testing for the known mutation previously found in your family.
○ **Comprehensive:** Full testing of the above 36 genes (cancer gene panel).

Possible Genetic Test Results:

RESULTS	EXPLANATION
Positive	Cancer Risk Increased • A mutation was found in one of the genes tested • Increased risk for cancer specific to the gene that has a mutation • Gene-specific cancer screening and prevention recommendations • Offering genetic testing to adult at-risk relatives for the specific mutation is recommended
Negative	Hereditary Cancer Risk Reduced • No mutations were found in any of the genes tested • Cancer risk(s) are based on personal and family history • Cancer screening and prevention recommendations based on family history • There still may be mutations in genes that were not tested • There still may be mutations in other family members that you did not inherit
Uncertain	Hereditary Cancer Risk Not Fully Defined • A genetic change (called a variant of uncertain significance) was found however the cancer risk is unclear • Cancer risk(s) are based on personal and family history • Cancer screening and prevention recommendation based on family history • Keep in touch every year, for updates on variant classification