Sunnybrook
ODETTE CANCER CENTRE
A Cancer Care Ontario Partner

Kidney (Renal) Cancer Gene Panel (15 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) UP TO AGE 70
BAP1	Kidney (10%), melanoma, mesothelioma, basal cell carcinoma
FH	Kidney (10-18%), paraganglioma/pheochromocytoma (up to 50%), leiomyomas-skin and uterine
FLCN	Kidney cancer and tumours (6-41%), lung cysts, pneumothorax, skin findings
MET	Papillary type I kidney (nearly 100%)
MITF	Kidney (up to 5 fold), melanoma (2-8 fold)
PTEN	Breast (25-85%), thyroid (up to 38%), uterine (up to 28%), kidney (34%), colon (9%), melanoma, form of skin cancer (up to 6%), gastrointestinal polyps
SDHA	Paraganglioma/pheochromocytoma (low malignant potential), kidney, gastrointestinal stromal tumour (GIST)
SDHAF2	Paraganglioma/Pheochromocytoma (up to 100%, paternal inheritance, low malignant potential)
SDHB	Paraganglioma/pheochromocytoma (77-100%), kidney (up to 14%), gastrointestinal stromal tumour (GIST)
SDHC	Paraganglioma/pheochromocytoma (low malignant potential), kidney, gastrointestinal stromal tumour (GIST)
SDHD	Paraganglioma/Pheochromocytoma (86-90%, paternal inheritance), kidney, gastrointestinal stromal tumor (GIST)
TP53	Breast, sarcoma (bone and soft tissue), brain, adrenocortical, leukemia and others. Overall risk for cancer: nearly 100% for women, 74% for men
TSC1	Kidney (2-5%), tumours: kidney, brain and spine, skin, liver, lung, heart; hamartomatous tumours
TSC2	Kidney (2-5%), tumours: kidney, brain and spine, skin, liver, lung, heart; hamartomatous tumours
VHL	Kidney (25-70%), pheochromocytoma (10-26%, <5% malignant potential), hemangioblastomas (brain and spine), pancreatic neuroendocrine tumours, endolymphatic sac tumours

*Cancer syndromes connected to above genes: Hereditary leiomyomatosis and renal cell carcinoma (FH), Birt-Hogg-Dubé (FLCN), Hereditary papillary renal carcinoma (MET), Cowden syndrome (PTEN), Hereditary PGL/PCC (SDHA, SDHAF2, SDHB, SDHC, SDHD), Li Fraumeni syndrome (TP53), Tuberous sclerosis complex (TSC1 and TSC2) and Von Hippel-Lindau disease (VHL).

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 15 genes (cancer gene panel).

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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 two to three years, for updates on variant classification