Renal Cancer Panel

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Hereditary Renal Cancer

Genetic testing with the Renal Cancer Panel may be appropriate if your personal and/or family history is suggestive of a hereditary predisposition to renal (kidney) cancer. This includes:

- A personal or family history of renal cancer at a young age (i.e. ≤46 years)
- A personal or family history of bilateral renal cancer (disease in both kidneys) or multiple primary tumors in a single kidney
- Multiple renal cancers within a family
- A pattern of cancer in which the individuals with similar or related cancers (see table below) are on one side of the family spanning multiple generations

Your healthcare provider will determine if genetic testing is medically necessary for you.

Genes Included on the Renal Cancer Panel are Listed in the Table Below

High-Risk Genes Well-studied • Greater than 4-fold risk of developing one or more cancers • Can cause a moderate risk for other cancers • National or expert opinion guidelines for screening and prevention are established

Newer Genes Not as well-studied • Precise lifetime risks and tumor spectrum not yet determined • Guidelines for screening and prevention are limited or not available

Lifetime Cancer and/or Tumor Risks

	Gene	Lifetime Cancer and/or Tumor Risks*
High-Risk Genes	*EPCAM***	Colorectal (69-75%), Endometrial (12-55%), Ovarian, Gastric, Pancreatic, Biliary tract, Urinary tract-transitional cell, Small bowel, Brain, Sebaceous tumors, Prostate
	FH	Renal-type II papillary (10-19%), Paraganglioma/Pheochromocytoma, Leiomyomas-skin and uterine
	FLCN	Renal cancer and tumors (6-41%)
	MLH1	Colorectal (34-46%), Endometrial (18-54%), Ovarian (10-20%), Gastric (6-20%), Urinary tract-transitional cell (1-4%), Pancreatic (1-4%), Biliary tract (2-3%), Small bowel (4-12%), Brain, Sebaceous neoplasms, Prostate
	MSH2	Colorectal (37-48%), Endometrial (21-57%), Ovarian (10-24%), Urinary tract-transitional cell (8-20%), Gastric (<1-9%), Pancreatic (1-4%), Biliary tract, Small bowel (1%), Brain, Sebaceous neoplasms, Prostate
	*MSH6***	Colorectal (20-44%), Endometrial (16-71%), Ovarian (1-13%), Gastric, Pancreatic, Biliary tract, Urinary tract-transitional cell, Small bowel, Brain, Sebaceous neoplasms, Prostate
	*PMS2***	Colorectal (11-20%), Endometrial (12-26%), Ovarian, Gastric, Pancreatic, Biliary tract, Urinary tract-transitional cell, Small bowel, Brain, Sebaceous neoplasms, Prostate
	PTEN	Female breast (25-85%), Thyroid (3-38%), Endometrial (5-28%), Colorectal, Renal, Melanoma, Gastrointestinal polyps, Lhermitte-Duclos disease
	SDHB	Paraganglioma/Pheochromocytoma, Renal, Gastrointestinal stromal tumor (GIST)
	SDHD	Paraganglioma/Pheochromocytoma, Renal, Gastrointestinal stromal tumor (GIST)
	TP53	Female breast (85%), Soft tissue sarcoma, Osteosarcoma, Brain, Hematologic malignancies-Acute leukemias, among others, Adrenocortical carcinoma, among others. Overall risk for cancer: up to 95% in females, 88% in males
	TSC1	Renal cancer (5%) and tumors, Benign central nervous system tumors-subependymal nodules and subependymal giant cell astrocytomas, Hamartomatous tumors-cardiac rhabdomyomas and angiomyolipomas
	TSC2	Renal cancer (5%) and tumors, Benign central nervous system tumors-subependymal nodules and subependymal giant cell astrocytomas, Hamartomatous tumors-cardiac rhabdomyomas and angiomyolipomas
	VHL	Renal-clear cell (up to 69%), Hemangioblastomas-retinal and central nervous system (50-80%), Pheochromocytomas (11-19%), Pancreatic neuroendocrine tumors (8-17%), Endolymphatic sac tumors (up to 10%)
Newer Genes	*BAP1*	Renal, Melanoma-cutaneous and uveal, Mesothelioma, Basal cell carcinoma
	MET	Renal-type I papillary
	MITF	Renal, Melanoma
	SDHC	Paraganglioma/Pheochromocytoma, Renal, Gastrointestinal stromal tumor (GIST)

*Most commonly associated cancer/tumors listed; lifetime risks provided when available. Risks relate to carriers of a single pathogenic variant.
**Tumor spectrum is representative of Lynch syndrome; data are limited with regard to the association of certain cancers with pathogenic variants in *MSH6, PMS2* and *EPCAM*.

Possible Outcomes of Genetic Testing

+ Positive

- Pathogenic or likely pathogenic variant identified
- Medical management recommendations may be available
- Family member testing may be recommended

− Negative

- No significant genetic changes identified
- Medical management based on personal and/or family history

? Variant of Uncertain Significance (VUS)

- A genetic change identified, but its association with disease is unclear
- Medical management based on personal and/or family history

Medical Management Based on Genetic Test Results

Clinical guidelines may be available which provide options and recommendations for patients who have a positive (pathogenic or likely pathogenic variant) test result indicating an increased risk for cancer and/or tumors. Guidelines and recommendations for early detection and/or risk reduction are specific to the gene in which the pathogenic variant was found.

Recommendations May Include:

- Clinical exams, such as skin or eye exams
- Blood or urine analysis
- Imaging exams, such as a MRI, CT and/or ultrasound
- Screening procedures, such as a colonoscopy or endoscopy
- Risk-reducing surgery

In some cases, guidelines for screening and prevention are limited or not available for a positive result. Once your test results are available, a discussion with your healthcare provider is recommended to determine the most appropriate medical management options for you and your family.

Resources

General
American Cancer Society
www.cancer.org

GeneDx
www.genedx.com/oncology

National Cancer Institute
www.cancer.gov

Kidney Cancer
Kidney Cancer Association
www.kidneycancer.org

National Kidney Foundation
www.kidney.org

Find a Genetic Counselor
Canadian Association of Genetic Counsellors
www.cagc-accg.ca

National Society of Genetic Counselors
www.nsgc.org

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