Hyperparathyroidism/Endocrine Tumor Panel

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Hereditary Hyperparathyroidism/Endocrine Tumors

Genetic testing with the Hyperparathyroidism/Endocrine Tumor Panel may be appropriate if your personal and/or family history is suggestive of a hereditary predisposition to cancer and/or tumors. **This includes:**

- A personal history of two or more endocrine tumors or disorders including hyperparathyroidism, pituitary adenoma, thyroid cancer, benign thyroid disease, carcinoid tumors, or neuroendocrine tumors of the pancreas or gastrointestinal tract
- A personal history of parathyroid carcinoma
- A personal history of multi-glandular hyperparathyroidism
- Multiple relatives on the same side of the family diagnosed with endocrine tumors and/or hyperparathyroidism

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

Genes Included on the Hyperparathyroidism/Endocrine 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

Moderate-Risk Genes Well-studied • Approximately 2- to 4-fold risk of developing one or more cancers • May increase risk for other cancers • Limited guidelines for screening and prevention

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

	Gene	Lifetime Cancer and/or Tumor Risks*
High-Risk Genes	*APC*	Colorectal (up to 93%), Duodenal or periampullary (4-12%), Gastric, Thyroid (up to 3%), Pancreatic, Brain-medulloblastoma, Liver-hepatoblastoma, Desmoid tumors, Gastrointestinal polyps
	CDC73	Hyperparathyroidism, Parathyroid cancer and tumors, Jaw tumors-ossifying fibromas, Renal tumors, Uterine tumors
	MEN1	Hyperparathyroidism, Parathyroid tumors (95%), Neuroendocrine tumors of the gastro-entero-pancreatic (GEP) tract (up to 80%), Anterior pituitary tumors (20-65%), Carcinoid tumors, Adrenal tumors (pheochromocytomas and adrenocortical tumors), and other tumors
	PRKAR1A	Myxomas-cardiac (20-40%) and cutaneous, Testicular tumors-large-cell calcifying Sertoli cell tumors, Pituitary tumors (10-20%), Thyroid (10%), Schwannomas-psammomatous melanotic (up to 10%), Primary pigmented nodular adrenocortical disease (25-60%)
	PTEN	Female breast (25-85%), Thyroid (3-38%), Endometrial (5-28%), Colorectal, Renal, Melanoma, Gastrointestinal polyps, L'hermitte-Duclos disease
	RET	Thyroid-medullary (greater than 90%), Pheochromocytoma (up to 50%), Hyperparathyroidism (up to 30%)
Moderate-Risk Genes	*CHEK2*	Female breast, Male breast, Colorectal, Gastric, Prostate, Thyroid
Newer Genes	*AIP*	Pituitary adenomas (benign pituitary tumors)
	CASR	Hyperparathyroidism, Parathyroid tumors
	CDKN1B	Hyperparathyroidism, Pituitary tumors, Gastro-entero-pancreatic neuroendocrine tumors, Parathyroid tumors
	DICER1	Lung tumors-pleuropulmonary blastoma, Thyroid tumors-multinodular thyroid goiter and cancer, Renal tumors-cystic nephroma, Ovarian tumors-Sertoli-Leydig, Embryonal rhabdomyosarcoma-cervix, Pituitary blastoma, Pineoblastoma

*Most commonly associated cancer/tumors listed; lifetime risks provided when available. Risks relate to carriers of a single pathogenic variant.

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 dental, skin, hearing or eye exams
- Blood and/or urine analysis
- Imaging exams, such as an MRI, CT and/or ultrasound
- 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

Hyperparathyroidism/Endocrine Resources
National Institute of Diabetes and Digestive and Kidney Diseases (NIDDK)-Primary Hyperparathyroidism
www.niddk.nih.gov/health-information/endocrinediseases/primary-hyperparathyroidism

The American Association of Endocrine Surgeons (AAES)
endocrinediseases.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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