PGL/PCC Panel

BioReference LABORATORIES | genpath | GeneDx
OPKO Health Companies

Hereditary Paragangliomas (PGL) and Pheochromocytomas (PCC)

Paragangliomas and pheochromocytomas are tumors found in neuroendocrine tissue. These tumors may secrete excess adrenaline (in the form of epinephrine and norepinephrine) and most are not malignant (cancerous).

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

- A personal and/or family history of one or more paragangliomas or pheochromocytomas
- Multiple relatives diagnosed with the same or related cancers (see table below) on the same side of the family and spanning multiple generations

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

Genes Included on the Melanoma 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	*FH*	Renal-type II papillary (10-19%), Paraganglioma/Pheochromocytoma, Leiomyomas-cutaneous and uterine
	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
	NF1	Neurofibromas, Optic nerve gliomas (15%), Pheochromocytomas (1-13%), Malignant peripheral nerve sheath tumors (6-16%), Brain tumors (2-3%), Female breast (up to 26%), Gastrointestinal stromal tumor (GIST)
	RET	Thyroid-medullary (greater than 90%), Pheochromocytoma (up to 50%), Hyperparathyroidism (up to 30%)
	SDHB	Paraganglioma/Pheochromocytoma, Renal, Gastrointestinal stromal tumor (GIST)
	SDHD	Paraganglioma/Pheochromocytoma, Renal, Gastrointestinal stromal tumor (GIST)
	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	*MAX*	Paraganglioma/Pheochromocytoma
	SDHA	Paraganglioma/Pheochromocytoma, Gastrointestinal stromal tumor (GIST)
	SDHAF2	Paraganglioma/Pheochromocytoma
	SDHC	Paraganglioma/Pheochromocytoma, Renal, Gastrointestinal stromal tumor (GIST)
	TMEM127	Paraganglioma/Pheochromocytoma

*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:

- Blood or urine analysis
- Imaging exams, such as a MRI, CT and/or ultrasound

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

PGL/PCC Resources
Pheo Para Troopers
www.pheoparatroopers.org

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

National Society of Genetic Counselors
www.nsgc.org