



Both paragangliomas and pheochromocytomas can be associated with germline pathogenic variants. Although these neuroendocrine tumors are relatively rare, the identification of patients and families with germline risk enables the implementation of surveillance programs to decrease the morbidity and mortality associated with these tumors. Individuals with germline risk require lifelong screening, which is implemented as early as age 5 years. In addition to ensuring that surveillance protocols are implemented, nurses provide education about symptoms that require prompt evaluation.

#### AT A GLANCE

- Paragangliomas and pheochromocytomas are neuroendocrine tumors that are often the result of a germline pathogenic variant.
- Individuals with a germline pathogenic variant associated with paragangliomas and pheochromocytomas require extensive monitoring starting at a young age and continuing throughout their life.
- Extensive education and support are provided by nurses so that patients understand the importance of surveillance and reporting of any symptoms suggestive of excessive catecholamine release.

#### KEYWORDS

paraganglioma; genetic testing; pheochromocytoma; hereditary neuroendocrine tumor

#### DIGITAL OBJECT IDENTIFIER

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# Hereditary Neuroendocrine Tumors

Providing comprehensive care for individuals who have a germline pathogenic variant associated with paragangliomas and pheochromocytomas

Suzanne M. Mahon, DNS, RN, AOCN®, AGN-BC, FAAN

**P**heochromocytomas and paragangliomas are neuroendocrine tumors of the adrenal medulla (inner part of the adrenal gland) and sympathetic and parasympathetic ganglia. Although rare, with an estimated 8 per 100,000 people, paragangliomas and pheochromocytomas can be associated with significant morbidity and mortality (Huang et al., 2018). An estimated 30% to 40% of paragangliomas and pheochromocytomas are the result of germline pathogenic variants (Fishbein, 2019). The early identification of a germline pathogenic variant enables an individual to implement a lifelong screening program to detect and monitor for paragangliomas and pheochromocytomas. If a paraganglioma or pheochromocytoma is growing or changing, early surgical intervention may ultimately decrease the associated morbidity and mortality. This article will describe the clinical characteristics of paragangliomas and pheochromocytomas, associated germline pathogenic variants, screening recommendations for individuals at increased risk, and implications for clinical care.

#### Clinical Pathophysiology and Features

Paragangliomas and pheochromocytomas are tumors that arise from chromaffin

cells (also called pheochromocytes) in neuroendocrine tissue extending from the paravertebral axis at the base of the skull to the pelvis (Huang et al., 2018). The presentation of symptoms can be variable (Else et al., 2018). Typically, parasympathetic paragangliomas are not associated with an excess catecholamine release and are primarily located near the skull. Symptoms of paragangliomas, particularly in the head and neck area, can result in pain from an enlarging mass, difficulty swallowing, coughing, or hearing loss in one ear. Pheochromocytomas are paragangliomas in the adrenal gland and are usually associated with excess catecholamine release. Sympathetic paragangliomas may also cause an excess catecholamine release and are located primarily in the abdomen and pelvis. Symptoms of excess catecholamine release are reflected as sustained or paroxysmal elevations in blood pressure, headache, episodic profuse diaphoresis, forceful palpitations, pallor, and an overwhelming sense of apprehension, panic, or anxiety.

Most paragangliomas and pheochromocytomas are benign. However, they can be associated with a high morbidity and mortality rate because of the hypersecretion of catecholamines and metanephrines, resulting in uncontrolled hypertension that can be sustained, episodic, or orthostatic, as well as cardiovascular disease