# Cost should not be a barrier to genetic testing in patients with paragangliomas and pheochromocytomas

Shana L Merrill, MS, LCGC; Jill S Dolinsky, MS, CGC; Sarah Witherington, MS, CGC; Jennifer Thompson, BS, Lauren Fishbein MD, PhD; Katherine Nathanson, MD

Paragangliomas and pheochromocytomas (PGL/PCCs) are tumors of the autonomic nervous system that often occur secondarily to underlying genetic predispositions. In retrospective European and US clinical cohorts with confirmed diagnoses of PGL/PCC, ~20-40% of patients have detectable germline mutations. Identifying a genetic predisposition significantly impacts the medical management of the proband and family members. Thus, consideration of genetic testing has been recommended for all patients with a PGL/PCC, regardless of family history. However, anecdotally and based on published assessments of barriers to testing, patients and physicians view cost as a barrier. To our knowledge, there has been no systematic assessment of this barrier for PGL/PCC genetic testing. We hypothesize that for the majority of patients with insurance, out-of-pocket cost is not a barrier to genetic testing for PGL/PCCs. We retrospectively reviewed data from 562 samples that were submitted to Ambry Genetics for either a PGL panel (*MAX, SDHAF2, SDHB, SDHC, SDHD, TMEM127* ± *SDHA, VHL, RET*) or PGL single gene analysis between August 2011 and April 2014. Samples submitted for *RET* or *VHL* single gene analysis only or for known familial mutation testing were excluded. A pathogenic alteration was identified in ~25% of total samples tested, consistent with previously published mutation detection rates. Of the samples submitted, 32 were cancelled prior to initiation of testing due to financial reasons, including 10 with genetic testing exclusions on the insurance plan, 4 denied coverage based on lack of medical necessity, and 18 with an out-of-pocket cost higher than the patient was willing to pay. Therefore, 5.7% of samples analyzed were cancelled due to financial reasons. This descriptive study confirms that at least 1 in 4 patients undergoing genetic testing for PGL/PCC have a clinically significant result and provides reassurance that it is unlikely financial barriers will hinder the adoption of genetic testing as standard of care for all patients with PGL/PCC.