Paraganglioma-Pheochromocytoma: Deletion/Duplication Panel

**Test Code:** MD203  
**Turnaround time:** 2 weeks  
**CPT Codes:** 81228 x1, 81403 x1

### Condition Description

The estimated prevalence of pheochromocytomas (PHEOs) is 1:4500 and 1:1700 for paragangliomas (PGLs), with an annual incidence of 3 to 8 cases per 1 million a year in the general population. The age of onset for PGL individuals is between 15 and 45 years of age, with early symptoms typically consisting of local swelling and cranial nerve injury. Although the tumors are benign in nature, only 4-16% shows malignant degeneration, giving rise to swelling causing cranial nerve damage, facial asymmetry, deafness, or hoarseness. PGLs are composed of pheochromocytomas (PCC, IPGLs, PH, or PHEOs) and extra-adrenal PGLs, and are characterized by the tumor production and secretion of catecholamines arising from the adrenal medulla (pheochromocytoma) or sympathetic nervous ganglia. Diagnosis for PGLs is determined by a biochemical assay, tumor location, function, inheritance, and gene mutation. If the tumor has metastasized the overall five year survival rate in patients is 40-72%. Size and location of the primary tumor has been correlated with risk for metastasis and the duration of survival.

**Reference:**

### Genes

**PH, MAX, RET, SDHAF2, SDHB, SDHC, SDHD, TMEM127, VHL**

### Indications

The test is indicated for:
- Individuals with a clinical or suspected diagnosis of pheochromocytomas and paragangliomas.

### Methodology

**Deletion/Duplication Analysis:** DNA isolated from peripheral blood is hybridized to a gene-targeted CGH array to detect deletions and duplications. The targeted CGH array has overlapping probes that cover the entire genomic region.

Please note that a "backbone" of probes across the entire genome are included on the array for analytical and quality control purposes. Rarely, off-target copy number variants causative of disease may be identified that may or may not be related to the patient's phenotype. Only known pathogenic off-target copy number variants will be reported. Off-target copy number variants of unknown clinical significance will not be reported.

### Detection

**Deletion/Duplication Analysis:** Detection is limited to duplications and deletions. The CGH array will not detect point or intronic mutations. Results of molecular analysis must be interpreted in the context of the patient's clinical and/or biochemical phenotype.

### Specimen Requirements

Submit only 1 of the following specimen types

**Type: Whole Blood**

Specimen Requirements:

In EDTA (purple top) tube:
- Infants (2 years): 3-5 ml
- Older Children & Adults: 5-10 ml.

**Specimen Collection and Shipping:** Ship sample at room temperature with overnight delivery.

**Type: Isolated DNA**

Disclaimer: This information is confidential and subject to change without notice. It may not be reproduced in whole or part unless authorized in writing by an authorized EGL representative.
Specimen Requirements:

In microtainer: 10 ug

Isolation using the Qiagen™ Puregene kit for DNA extraction is recommended.

Specimen Collection and Shipping: Refrigerate until time of shipment in 100 ng/ul of TE buffer. Ship sample at room temperature with overnight delivery.

Special Instructions

This test is for germline mutation analysis. DNA isolated from FFPE tumor samples is not suitable for this test.

Related Tests

- Hereditary Cancer Syndrome: Sequencing Panel.
- Paragangioma-Pheochromocytoma: Sequencing Panel.