Endocrine Cancer: Deletion/Duplication Panel

Test Code: MD202  
Turnaround time: 2 weeks  
CPT Codes: 81323 x1, 81403 x1, 81404 x1

**Condition Description**

Thyroid cancer is divided into several subcategories: (1) differentiated (follicular, papillary and Hurthle); (2) medullary; and (3) anaplastic (aggressive undifferentiated tumor). Medullary thyroid cancer (MTC) develops from the “C” or parafollicular cells of the thyroid gland which produce calcitonin. Approximately 80% of the cases of MTC are sporadic. The remaining inherited syndromes include multiple endocrine neoplasia (MEN) type 2A (also known as MEN 2A), MEN 2B, and familial MTC (FMTC). All three of these subtypes, MEN 2A, MEN 2B and FMTC, are inherited in an autosomal dominant pattern and involve an elevated risk for the development of medullary carcinoma of the thyroid. MEN 2A and MEN 2B have an increased risk for the development of pheochromocytoma. MEN 2A has an elevated risk for parathyroid adenoma or hyperplasia. Additional features in MEN 2B include distinctive facies with enlarged lips, mucosal neuromas of the lips and tongue, and ganglioneuromatosis of the gastrointestinal tract. MTC generally occurs in early childhood in MEN 2B, early adulthood in MEN 2A, and middle age in FMTC.

**References:**


**Genes**

CDC73, MAX, MEN1, PRKAR1A, PTEN, RET, SDHAF2, SDHB, SDHC, SDHD, TMEM127, TP53, VHL

**Indications**

The test is indicated for:

- Individuals with a clinical or suspected diagnosis of endocrine cancer.

**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**

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.
- Endocrine Cancer: Sequencing Panel.