Endocrine Cancer: Deletion/Duplication Panel

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

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: DNA, Isolated

Specimen Requirements:

- Microtainer
- 3µg Isolation using the Perkin Elmer™Chemagen™ Chemagen™ Automated Extraction method or Qiagen™ Puregene kit for DNA extraction is recommended.

Specimen Collection and Shipping:

Refrigerate until time of shipment in 100 ng/µL in TE buffer. Ship sample at room temperature with overnight delivery.

Type: Whole Blood (EDTA)

Specimen Requirements:
<table>
<thead>
<tr>
<th>EDTA (Purple Top)</th>
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<tbody>
<tr>
<td>Infants and Young Children ( 2 years of age to 10 years old): 3-5 ml</td>
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<tr>
<td>Older Children &amp; Adults: 5-10 ml</td>
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<tr>
<td>Autopsy: 2-3 ml unclotted cord or cardiac blood</td>
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**Specimen Collection and Shipping:**
Ship sample at room temperature for receipt at EGL within 72 hours of collection. Do not freeze.

**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.