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
(aggresive 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:


- American Thyroid Association Guidelines Task Force, Kloos RT, Eng

- Moline J, Eng C. GeneReviews™. Multiple Endocrine Neoplasia Type
  Accessed on October 5, 2013.

- Romei C, Cosci B, Renzini G, et al. RET genetic screening of
  sporadic medullary thyroid cancer (MTC) allows the preclinical diagnosis
  of unsuspected gene carriers and the identification of a relevant

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

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.
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.