Renal Cancer: Deletion/Duplication Panel

Test Code: MD206  
Turnaround time: 2 weeks  
CPT Codes: 81300 x1, 81323 x1, 81294 x1, 81297 x1, 81403 x1, 81404 x1, 81405 x1, 81406 x1

Condition Description

Renal cancer is a multifarious and heterogeneous disease with a varied spectrum of malignant subtypes and clinical presentation. A number of gene mutations have been reported in the literature. Renal cell carcinoma (RCC) tumor subtypes include clear cell or conventional (70-80%); papillary type 1 and type 2 (10-15%); chromophobe (3-5%) and collecting duct (1%). The general population's lifetime risk to develop RCC is 1.5%. RCC is the seventh and eighth most common cancer in men and women respectively. Approximately 3-5% of RCC cases are hereditary and occur as a result of an inherited mutation. Unlike sporadic RCC cases, hereditary RCC is often categorized by earlier disease onset and/or multifocal or bilateral tumors.

References:

Genes

BAP1, BUB1B, CDC73, CDKN1C, FH, FLCN, GPC3, MET, MLH1, MSH2, MSH6, PALB2, PTEN, SDHB, SDHC, SDHD, SMARCB1, TP53, TSC1, TSC2, VHL, WT1

Indications

The test is indicated for:
- Individuals with a clinical or suspected diagnosis of hereditary renal 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: Whole Blood (EDTA)**

**Specimen Requirements:**
- EDTA (Purple Top)
  - Infants and Young Children (2 years of age to 10 years old): 3-5 ml
  - Older Children & Adults: 5-10 ml
- Autopsy: 2-3 ml unclotted cord or cardiac blood

**Specimen Collection and Shipping:**
Ship sample at room temperature for receipt at EGL within 72 hours of collection. Do not freeze.

**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.
**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.
- Renal Cancer: Sequencing Panel.