Renal Cancer: Deletion/Duplication Panel

Test Code: MD206
Turnaround time: 2 weeks
CPT Codes: 81294 x1, 81297 x1, 81300 x1, 81323 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, SMARCBI, 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.

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.

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.
- Renal Cancer: Sequencing Panel.