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

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### 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.

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

- **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**

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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.