Wilms Tumor: Deletion/Duplication Panel

**Test Code:** MD207  
**Turnaround time:** 2 weeks  
**CPT Codes:** 81228 x1

### Condition Description

Despite the number of genes that appear to be involved in the development of Wilms tumor, hereditary Wilms tumor is uncommon, with approximately 2% of patients having a positive family history for Wilms tumor. Siblings of children with Wilms tumor have a low likelihood of developing Wilms tumor. The risk of Wilms tumor among offspring of persons who have had unilateral (sporadic) tumors is less than 2%.

Children with a significantly increased predisposition to develop Wilms tumor (e.g., most children with Beckwith-Wiedemann syndrome, WAGR syndrome, Denys-Drash syndrome, idiopathic hemihypertrophy, or sporadic aniridia) should be screened with ultrasound every 3 months at least until they reach age 8 years. Approximately 10% of patients with Beckwith-Wiedemann syndrome will develop a malignancy, with either Wilms tumor or hepatoblastoma being the most common, although adrenal tumors can also occur. Children with hemihypertrophy are also at risk for developing liver and adrenal tumors. Screening with abdominal ultrasound and serum alpha-fetoprotein is suggested until age 4 years; after age 4, most hepatoblastomas will have occurred, and imaging may be limited to renal ultrasound, which is quicker and does not require the child to fast for the exam.

**References:**

### Genes

- **CDKN1C, WT1**

### Indications

The test is indicated for:
- Individuals with a clinical or suspected diagnosis of Wilms tumor.

### 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:** Detection is limited to duplications and deletions. The CGH array will not detect point or intronic pathogenic variants. 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

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

- Wilms Tumor: Sequencing Panel
- Hereditary Cancer Syndrome: Sequencing Panel