Wilms Tumor: Sequencing Panel

| Test Code: MM207 | Turnaround time: 4 weeks | CPT Codes: 81405 x1, 81479 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, idioopathic 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**

**Next Generation Sequencing:** In-solution hybridization of all coding exons is performed on the patient's genomic DNA. Although some deep intronic regions may also be analyzed, this assay is not meant to interrogate most promoter regions, deep intronic regions, or other regulatory elements, and does not detect single or multi-exon deletions or duplications. Direct sequencing of the captured regions is performed using next generation sequencing. The patient's gene sequences are then compared to a standard reference sequence. Potentially causative variants and areas of low coverage are Sanger-sequenced. Sequence variations are classified as pathogenic, likely pathogenic, benign, likely benign, or variants of unknown significance. Variants of unknown significance may require further studies of the patient and/or family members.

**Detection**

**Next Generation Sequencing:** Clinical Sensitivity: Unknown. Mutations in the promoter region, some mutations in the introns and other regulatory element mutations cannot be detected by this analysis. Large deletions/duplications will not be detected by this analysis. Results of molecular analysis should be interpreted in the context of the patient's clinical/biochemical phenotype.

Analytical Sensitivity: ~99%.

**Specimen Requirements**

Submit only 1 of the following specimen types

**Type:** DNA, Isolated

**Specimen Requirements:**
- Microtainer
- 8µg
- Isolation using the Perkin Elmer™Chemagen™ Chemagen™ Automated Extraction method or Qiagen™ Puregene kit for DNA extraction is recommended.

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.
Specimen Collection and Shipping:
Refrigerate until time of shipment in 100 ng/µL in TE buffer. Ship sample at room temperature with overnight delivery.

Type: Saliva

Specimen Requirements:
Oragene™ Saliva Collection Kit
Oragene™ Saliva Collection Kit used according to manufacturer instructions. Please contact EGL for a Saliva Collection Kit for patients that cannot provide a blood sample.

Specimen Collection and Shipping:
Please do not refrigerate or freeze saliva sample. Please store and ship at room temperature.

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.

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
- Wilms Tumor: Deletion/Duplication Panel