Papillary Renal Carcinoma: MET Gene Deletion/Duplication

Test Code: UY
Turnaround time: 2 weeks
CPT Codes: 81228 x1

Condition Description

Papillary renal tumors, which account for 15 to 20% of renal carcinomas, occur in both sporadic and familial forms. Hereditary papillary renal carcinoma (HPRC) is an autosomal dominant hereditary cancer syndrome in which affected individuals are at risk of developing bilateral, multifocal type 1 papillary renal carcinoma, often at a late age of onset (50 to 70 years). To date, the kidney is the only organ to be affected in HPRC patients. The tumors are most often well differentiated; however, they are malignant and can metastasize. HPRC is a highly penetrant disease in which affected individuals are highly likely to develop bilateral, multifocal type 1 papillary kidney cancer. In the early reports, this disease was described as having a late onset; however, recently an early onset form of this disease has been described.

Germline mutations in the MET gene on chromosome 7 were identified in a hereditary form of papillary renal carcinoma. MET belongs to the family of tyrosine kinases, the members of which play important roles in transmitting signals from the cellular surface to the nucleus. Missense mutations in the tyrosine kinase domain of the Met proto-oncogene at 7q31 are responsible for constitutive activation of the MET protein in HPRC.

References


Click here for the OMIM summary on this condition.

Genes

MET

Indications

This test is indicated for:

- Confirmation of a clinical diagnosis of hereditary papillary renal carcinoma in individuals who have tested negative for sequence analysis
- Individuals at-risk for hereditary papillary renal carcinoma due to family history who have tested negative for sequence analysis

Methodology

DNA isolated from peripheral blood is hybridized to a CGH array to detect deletions and duplications. The targeted CGH array has overlapping probes which 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

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

- Preferred specimen type: Whole Blood

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: Refrigerate until time of shipment. Ship sample within 5 days of collection at room temperature with overnight delivery.

Type: Saliva

Specimen Requirements:

Oragene™ Saliva Collection kit (available through EGL) used according to manufacturer instructions.

Specimen Collection and Shipping: Store sample at room temperature. Ship sample within 5 days of collection at room temperature with overnight delivery.

Special Instructions

Submit copies of diagnostic biochemical test results with the sample, if appropriate. Contact the laboratory if further information is needed.

Sequence analysis is required before deletion/duplication analysis by targeted CGH array. If sequencing is performed outside of EGL Genetics, please submit a copy of the sequencing report with the test requisition.

Related Tests

- Sequencing analysis of the MET gene is available (UX) and is required before deletion/duplication analysis.
- Prenatal testing is available to individuals who are confirmed carriers of mutations. Please contact the laboratory genetic counselor to discuss appropriate testing prior to collecting a prenatal specimen.