Henry Ford Health System
CENTER FOR PRECISION DIAGNOSTICS

Hereditary Renal / Urinary Tract Cancer Panel (26 genes)

BAP1	*BUB1B*	*CDC73*	*CDKN1C*	*DICER1*	*DIS3L2*	*EPCAM*	*FH*	*FLCN*	*GPC3*
MET	*MLH1*	*MSH2*	*MSH6*	*PALB2*	*PMS2*	*PTEN*	*SDHB*	*SDHC*	*SDHD*
SMARCB1	*TP53*	*TSC1*	*TSC2*	*VHL*	*WT1*				

Indication

This Hereditary Renal / Urinary Tract Cancer Panel is a comprehensive 26-gene panel that identifies inherited risks for developing cancers of the urinary tract (kidneys, renal pelvis, ureters, bladder and urethra). This panel includes genes responsible for rare hereditary cancer syndromes that have been linked to increased risk for urinary tract cancers, such as Lynch syndrome (*MLH1*, *MSH2*, *MSH6*, *PMS2*, or *EPCAM*), Li-Fraumeni syndrome (*TP53*), Cowden syndrome (*PTEN*), Tuberous sclerosis complex (*TSC1*, *TSC2*), Von Hippel-Lindau syndrome (*VHL*), hereditary paraganglioma-pheochromocytoma syndromes (*SDHB*, *SDHC*, *SDHD*), WT1-related Wilms tumor (*WT1*), hereditary papillary renal cell carcinoma (*MET*), hereditary leiomyomatosis and renal cell carcinoma (*FH*), Birt-Hogg-Dubé syndrome (*FLCN*), DICER1 syndrome (*DICER1*), BAP1 tumor predisposition syndrome (*BAP1*), CDC73 related disorders (*CDC73*), Perlman syndrome (*DIS3L2*), Simpson-Golabi-Behmel syndrome (*GPC3*), and Rhabdoid tumor predisposition syndrome (*SMARCB1*). These syndromes have been associated with increased lifetime risk for multiple types of cancer, including renal cancer, and are also characterized by other clinical features and cancer types specific for each syndrome. This panel also includes other genes linked to renal cancer predisposition (*PALB2*, *CDKN1C*, *BUB1B*).

Hereditary cancer syndrome is a genetic predisposition to develop certain types of cancers, often at an early age. Hereditary cancer risk assessment is performed to identify patients and families who may be at risk. Clues that a hereditary cancer syndrome may be present include the following:

- Kidney cancer diagnosed at an unusually young age
- Several different types of cancer or two or more primary kidney cancers in the same person
- Multiple close family members with kidney and neuroendocrine tumors, especially when on the same side of the family
- Unusual presentation of a specific type of cancer
- The presence of birth defects that are known to be associated with inherited cancer syndromes
- Occurrence of certain types of adult cancer in which the probability of harboring a hereditary cancer syndrome is high

Testing method

Next Generation Sequencing (NGS) provides coverage of all coding exons and noncoding DNA in exon-flanking regions (on average 50 bp) enriched using hybrid capture Illumina TruSight Cancer Sequencing Panel. Single base pair (point) mutations, small insertions/deletions (1-25 bp), complex insertions and deletions, or larger deletions and duplication (<100 bp) are detected using a combination of clinically validated computational data analysis methods for sequence variant calling, filtering, and annotation. Gross deletions and duplications at each targeted gene and exon are evaluated through comparative

depth of coverage analysis of NGS targeted sequencing data using clinically-validated analysis algorithm. All reportable copy number variants are confirmed by independent methodology using gene-specific Multiplex Ligation-dependent Probe Amplification (MLPA) or genome-wide SNP microarray assay. If a pathogenic mutation or deletion is detected in exons 13, 14 or 15 of *PMS2*, confirmatory send out testing will be performed to determine if this variant is located in the *PMS2* gene or pseudogene, *PMS2CL*.

Turnaround time

5-10 business days

Sample requirements

3 ml peripheral blood in EDTA (lavender) top tube
Specimen stability: Ambient - 72 hours; Refrigerated - 1 week

CPT codes

81292, 81295, 81298, 81307, 81317, 81321, 81404 (X2), 81405 (X5), 81406, 81407, 81438, G0452

References

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Ship Specimens to:

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