

Urothelial Carcinoma by UroVysion FISH

Indications for Ordering

- May aid in diagnosis of urothelial carcinoma in individuals with hematuria
- Monitor for tumor recurrence in patients previously diagnosed with urothelial carcinoma

Test Description

Fluorescence in situ hybridization/computer-assisted analysis/microscopy

Tests to Consider

Primary test

[UroVysion FISH 2001181](#)

- Detects
 - Amplifications of chromosomes 3, 7, 17
 - Deletions of the 9p21 locus

Related tests

[Bladder Tumor Associated Antigen 2000183](#)

- Aid in management of bladder cancer patients in conjunction with cystoscopy
- Qualitative assay detects bladder tumor-associated antigen in urine of patients diagnosed with bladder cancer

[Cytology, Non-Gynecologic 2000623](#)

[NMP22, Urine 0080281](#)

- Aid in diagnosis of urothelial carcinoma in conjunction with standard diagnostic procedures and monitoring for tumor recurrence

Disease Overview

Incidence

- ~143,000 new cases of cancer in urinary system/year in U.S. (American Cancer Society, 2016)
 - Bladder cancer – ~77,000
 - Kidney and renal pelvis – ~63,000
 - Ureter and other urinary organs – ~3,500
- Caucasians:African Americans = 2:1
- Males:females = 4:1

Age of onset – ≥65 years

Symptoms

- Hematuria
- Irritative voiding

Screening/diagnosis issues

- Individuals complaining of mild hematuria traditionally tested for the presence of neoplastic lesions by
 - Cytology
 - More sensitive to high-grade lesions
 - May miss low-grade papillary tumors
 - Cystoscopy
 - Can detect low-grade papillary tumors
 - May miss high-grade carcinoma in situ
- Urothelial carcinoma shows high recurrence rate
 - ~70% of cases show recurring lesions after initial treatments
 - Necessitates ongoing, lifelong surveillance
- Bladder cancer can be detected by
 - Enumeration of morphologically abnormal cells for aneuploidy of chromosomes 3, 7, 17
 - Loss of both chromosomal 9p21 segments
- Molecular testing can serve as primary or adjuvant tests
 - Noninvasive when compared to cystoscopy
 - May be used to diagnose urothelial carcinoma
 - May be used to monitor recurrence of urothelial carcinoma
- UroVysion detects chromosomal abnormalities associated with urothelial cell carcinoma from voided urine specimens

Genetics

Variants

- Amplifications of chromosomes 3, 7, 17
- Deletions of the 9p21 locus

Test Interpretation

Sensitivity/specificity

- Clinical sensitivity – 68-81%
- Clinical specificity – 79-96%

Results

- Positive
 - One or more numeric chromosomal abnormalities commonly associated with urothelial carcinoma detected
 - ≥ 4 cells show gains for 2 or more chromosomes (3, 7, or 17) in the same cell, or
 - ≥ 12 cells have zero 9p21 signals
 - In the absence of clinical documentation of urothelial carcinoma within the bladder, positive result suggests
 - Possibility of urothelial carcinoma or other urological malignancy
 - Ureter
 - Urethra
 - Kidney
 - Prostate
 - Further clinical evaluation to exclude these tissues as a source of abnormal cells is recommended
- Negative
 - Lack of evidence for presence of numeric chromosomal abnormalities commonly associated with urothelial carcinoma within cells collected in specimen

Limitations

- Some urothelial cancers will not be detected
- Negative results in the presence of other symptoms/signs of urothelial carcinoma may suggest possibility of false-negative test result
- Not detected
 - Gene variants or defects other than
 - Amplification of chromosomes 3, 7, or 17
 - Deletion (loss) of 9p21 locus