UroVysion FISH for Urothelial Carcinoma

Urothelial carcinoma shows a high recurrence rate; therefore, ongoing, lifelong surveillance is necessary. Bladder cancer can be identified by enumerating morphologically abnormal cells with aneuploidy of chromosomes 3, 7, 17, or by the loss of both chromosomal 9p21 segments.

Molecular testing can be used in conjunction with other standard procedures for diagnosis or to monitor patients for recurrence of urothelial carcinoma. Compared with cystoscopy, molecular testing is noninvasive. The UroVysion FISH test is designed to detect chromosomal abnormalities associated with urothelial cell carcinoma in voided urine specimens.

Disease Overview

Incidence

The following are the estimated numbers of new urinary system cancer cases in the United States per year:\(^1\):

- Urinary bladder cancer: approximately 81,400
- Kidney and renal pelvis: approximately 73,750
- Ureter and other urinary organs: approximately 3,970

Symptoms

Two primary symptoms associated with urothelial carcinoma are:\(^2\):

- Hematuria
- Irritative voiding

Screening/Diagnosis Issues

Individuals complaining of mild hematuria are traditionally tested for the presence of neoplastic lesions with the following tests:

- Cytology\(^2\)
  - More sensitive to high-grade lesions
  - May miss low-grade papillary tumors
- Cystoscopy\(^3\)
  - Can detect low-grade papillary tumors

Featured ARUP Testing

UroVysion FISH 2001181

**Method:** Fluorescence in situ Hybridization (FISH)/Computer Assisted Analysis/Microscopy

- May aid in diagnosis of urothelial carcinoma in individuals with hematuria
- Use to monitor for tumor recurrence in patients previously diagnosed with urothelial carcinoma
- Use to detect amplification of chromosomes 3, 7, 17, and deletion of the 9p21 locus
Genetics

Variants

- Amplification of chromosomes 3, 7, 17
- Deletion of the 9p21 locus

Test Interpretation

Sensitivity/Specificity

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

Results

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| Positive | Detection of ≥1 of the following numeric chromosomal abnormalities commonly associated with urothelial carcinoma:
- ≥4 cells show gains for ≥2 chromosomes (3, 7, or 17) in the same cell
- ≥12 cells have no 9p21 signals |
| Negative | Lack of evidence for the presence of numeric chromosomal abnormalities commonly associated with urothelial carcinoma within the cells collected in specimen |

In the absence of clinical documentation of urothelial carcinoma within the bladder, a positive result suggests the possibility of urothelial carcinoma or other urologic malignancy in the ureter, urethra, kidney, or prostate. Further clinical evaluation to exclude these tissues as a source of abnormal cells is recommended.

Source: Abbott, 2014

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
- Gene variants or defects other than amplification of chromosomes 3, 7, or 17, and deletion (loss) of 9p21 locus are not detected

References


Related Information

Bladder Cancer