UROAMP

Urinary Comprehensive Genomic Profiling Test for Urothelial Cancer

Patient ID:

Comprehensive Genomic Profiling Report

PATIENT

Name: Sample Patient
DOB: 1963-03-29
Age & Gender: 60, Male
MRN: GWO117
Collection Date: 2022-10-20
Received Date: 2022-10-22
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PHYSICIAN

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Practice Name: Sample Clinic
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Report Highlights

- Risk factors for BCG resistance; gene identified associated with BCG response failure.
- Genes identified a higher risk of tumor invasion/metastatic disease and/or recurrence.
- There is an FDA-approved drug for mutation identified. See the Drug Therapy Associated Findings section.

Drug Therapy Associated Findings

Genomic Findings	FDA-Approved Therapies
FGFR3	BALVERSA® (erdafitinib)

Prognostic Findings

Genomic Findings	Prognostic Considerations
TP53	10x increased risk for high-grade cancer and 6x increased risk for tumor invasion. Considerations for repeat biopsy, more frequent surveillance, and continued BCG therapy, if clinically appropriate.
ARID1A	Increased risk of BCG refractory cancer.

Treatment decisions should not be made on this information alone and should also leverage a complete assessment of clinical history, pathology risk factors, treating physician judgment and patient preferences.

Mutated Genes Identified

Gene	Description	Mutation Fraction	Variant Loci (Protein Change)	References
ARID1A	In multiple studies, ARID1A is associated with worse prognosis, higher grade, and diagnosis at later stage. ARID1A has also been associated with a lack of response to BCG therapy.	26%	Chr1:27087458 (Q678*)	2
FGFR3	Mutations in FGFR3 are most common in papillary tumors and less frequent in CIS and invasive cancers. Erdafitinib (Balversa) is an FDA-approved drug indicated in patients with later stage tumors and FGFR3 mutations.	24%	Chr4:1803568 (S249C)	2
TP53	TP53 mutations are more common in clinically and histologically-advanced disease. Literature has found that TP53 mutations are an unfavorable prognostic factor. Alterations in this gene are associated with the development of urothelial carcinoma or recurrence. In a UroAmp validation study, presence of TP53 mutations were associated with a 9x increased risk for HG cancer and 6.6x increased risk for invasive carcinoma.	20%	Chr17:7578534 (K132N)	3

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Report Status: **FINAL**
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Test Validation and Studies

Next-generation DNA sequencing analysis was performed using the UroAmp assay to assess over 250,000 locations in the genome, across 60 recurrent urothelial cancer genes. The UroAmp variant caller has a validated sensitivity of 98% and specificity of 100%.[1]

For additional validation and studies scan or visit convergentgenomics.com/validation

References

1 Bicocca, V. et al., Urinary comprehensive genomic profiling correlates urothelial carcinoma mutations with clinical risk and efficacy of intervention. J. Clin. Med. 2022, 11(19), 5827.

2 Pietzak EJ et al., Next-generation sequencing of nonmuscle invasive bladder cancer reveals potential biomarkers and rational therapeutic targets. Eur. Urol 2017 Dec;72(6):952-959.

3 Eich, ML. et al., Incidence and distribution of UroSEEK gene panel in a multi-institutional cohort of bladder urothelial carcinoma. Mod Pathol 32, 1544–1550 (2019).

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CONVERGENT Genomics

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This test was developed and its performance characteristics were determined by Convergent Genomics. The Lab is regulated under CLIA88 as quaified to perform high-complexity clinical testing. This test has not been cleared nor approved by the FDA; FDA clearance or approval is not required. This test is used for clinical purposes and clinical correlation of its results are recommended. A copy of this form shall be as valid as the original.

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