Clinical Utility of a Custom Next-Generation Sequencing Panel in the Diagnosis of Needle Biopsies from Renal Masses

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INTRODUCTION

Image-guided, percutaneous biopsy of kidney tumors is increasingly utilized, particularly in patients at higher risk of adverse outcomes. Despite improved biopsy techniques, low yield and disrupted tissue architecture may make histologic diagnosis challenging. Selected patients may avoid expirative treatment if benign or indolent tumors are determined accurately by biopsy.

Specific mutations and copy number changes have been identified in kidney tumors which could be utilized in subtyping and outcome prediction. Simultaneous evaluation of mutation and copy number could be highly useful in providing both diagnostic and prognostic information from minimal biopsy material.

MATERIALS & METHODS

Patient Characteristics

48 percutaneous 18-22 gauge core needle biopsies prospectively collected from 45 patients (11/2011 – 1/2014)

28 Men, 20 Women

Median age (years): 72 (IQR: 63, 74)

Median Mass (cm): 2.7 (IQR: 1.0, 14.0)

DNA Yield: 20 ng – 21 ug

Molecular Analyses

- **aCGH**: 256 ng – 1 pg DNA
- **Blinded copy number–based molecular classification**: n=40 diagnostic
- **NGS with leftover DNA**: 220 ng – 260 ng
- **Copy number–based molecular classification**: n=41 diagnostic

RESULTS

**Materials and Methods**

- **Tumor Biopsy**
- **Copy Number-Based Subtyping**
- **Histology**
- **Specimen Histology**
- **Copy Number-Based Subtyping**

**Histology and Copy Number-Based Subtyping**

ccRCC (n=16):

- **Benign**
- **pRCC**
- **Non-D**

ccRCC (n=17):

- **Benign Neoplasms/Non-Diagnostic (n=15):**
- **Benign Fibrosis**
- **Poorly diff./HG RCC**

ccRCC (n=13):

- **Benign RCC**
- **Other Malignant RCC**

Other Malignant RCC (n=12):

- **pRCC**
- **ccRCC**
- **Non-D**

Benign Neoplasms/Non-Diagnostic (n=15):

- **Benign Fibrosis**
- **Poorly diff./HG RCC**

**Abbreviations**

- UroGenRA:
- UroGen: corporate partners: Illumina, Affymetrix, Roche, Focus Diagnostics, BioMap, Assure, Biorad, QIAGEN, Overlapping Regions: subdivision into 75
- OT: ≥50%, N=50%

**CONFLICTS OF INTEREST**

B.G., M.J., L.J., and J.H. are full-time employees and stock/stock option holders of Cancer Genetics, Inc. R.S.K.S. is a Board Member, paid consultant and stock/stock option holder for Cancer Genetics, Inc.