Utilizing Liquid Biopsies for Prognostication and Disease Monitoring in Uveal Melanoma

**Introduction**

- Uveal melanoma (UM) is the most common primary intraocular tumor in adults and accounts for <5% of all melanomas.
- Approximately 50% of patients with early-stage UM will develop metastatic disease.
- There are no blood biomarkers currently that identify patients with the UM who are at higher risk of disease relapse.
- It is hypothesized that the presence of circulating tumor cells (CTCs) and cell-free DNA (cfDNA) prognosticate an increased risk of relapse.

**Objective**

- Determine factors associated with survival for early-stage and metastatic UM patients.
- Identify biomarkers (CTCs, cfDNA, circulating tumor DNA (ctDNA)) associated with relapse.

**Methods**

- **Review of data on patients:**
  - Presenting at MD Anderson with UM from 12/1/2014 to 7/26/2023
  - Enrolled in IRB Protocol LAB11-0314
- **Factors examined:**
  - Patient demographics
  - Clinicopathologic features
  - Treatment strategies
- **Biomarkers obtained from peripheral blood samples:**
  - CTCs using the CellSearch Circulating Melanoma Cell Assay®
  - cfDNA using MagMax™ cell-free DNA Isolation Kit, Oncomine™ Pan-Cancer Assay, and Ion Torrent™ Technology
- **Statistical analyses:**
  - Unpaired t-test, univariable and multivariable Cox regression modeling

**Results**

- **Total Patients (n = 157)**
  - Sex
    - Male: 72 (46%)
    - Female: 85 (54%)
  - Median age at diagnosis: 55 years (19-83 years)
- **Ethnicity**
  - Non-Hispanic: 147 (94%)
  - Hispanic or Latino: 4 (2%)
  - Unknown: 6 (4%)
- **Disease status at study enrollment**
  - Early-stage disease: 111 (71%)
  - Metastatic: 46 (29%)
- **Classification**
  - Class 1A: 25 (16%)
  - Class 1B: 16 (10%)
  - Class 2: 55 (35%)
  - Class 3: 61 (39%)
  - **PRAE**
    - Positive: 25 (16%)
    - Negative: 44 (28%)
    - Unknown: 88 (56%)
- **Tumor mutation analysis (n = 53)**
  - Wildtype: 2 (4%)
  - GNAQ: 28 (53%)
  - BAP1: 16 (30%)
  - GNA11: 20 (38%)
  - Other: 16 (30%)
- **Median time between diagnosis and first blood sampling**
  - Early-stage: 21.42 months
  - Metastatic: 10.58 months
  - Total Study follow-up: 63.83 months
- **ctDNA Tumor Panel Concordance**
  - MAP2K1, GNAQ, TP53: NO
  - GNA11: YES
  - GNAQ, MAP2K1: GNAQ YES
  - MAP2K1, GNAQ: GNAQ YES
  - RAF1: NO
  - PRAE, GNAQ: NO
  - GNAQ, MAP2K1: YES
  - TP53: YES

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**References**

Anand K et al. Cancers. 2019; 11(6):856