Clinical Prognostication Test In Uveal Melanoma

Tech ID: 30376 / UC Case 2019-286-0

BACKGROUND

Uveal melanoma commonly known as ocular or choroidal melanoma, is a rare cancer of the eye. It is an intraocular malignancy that arises from melanocytes of the choroid, ciliary body, and iris of the eye. Ocular melanoma is diagnosed in approximately 2,000-2,500 adults annually in the United States. In both the U.S. and Europe, this equates to about 5 - 7.5 cases per million people per year and, for people over 50 years old, the incidence rate increases to around 21 per million per year. While the primary tumor is highly treatable, about half of the patients will develop metastasis —typically to the liver. Metastatic disease is universally fatal. While traditional staging methods such as tumor size and location, still play a role in assessing metastatic risk, they are rarely used to individualize patient management plans. Newer methods include chromosomal gene expression analysis, yet these methods have their technical limitations. Clearly, what is needed is a better, cheaper and reproducible prognostic test.

TECHNOLOGY DESCRIPTION

Researchers at UC San Diego have developed a method to predict patient survival by examining the methylation of a specific gene(s) that can predict patient survival and can be used to estimate prognostic classes of the disease. Furthermore, researchers demonstrate using a published cohort that a gene set or methylation data of a single gene can accurately predict survival in patients with primary uveal melanoma.

APPLICATIONS

The methodology can be applied to biopsy specimens from fine needle aspiration or enucleation, paraffin-embedded or frozen specimens.

ADVANTAGES

The methylation data from our gene of interest is highly predictive of survival and would be performed at a much lower cost and faster turnaround time compared to the current gene panel testing. Sample handling would also be easier as DNA is much more stable than RNA.

STATE OF DEVELOPMENT

Experimental stage, validated in a published cohort. The methylation signature was analyzed on 80 uveal specimens. These underwent whole exome sequencing, RNA sequencing and have associated clinical findings including survival. This allowed us to isolate a set of genomic loci where methylation is highly associated with poor survival. We will continue to validate it in additional cohorts.

INTELLECTUAL PROPERTY INFO

The technology is patent pending and available for licensing

PATENT STATUS

Patent Pending

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OTHER INFORMATION

KEYWORDS

Cancer, diagnostic, prognostication, uveal melanoma, methylation

CATEGORIZED AS

- **Medical**
  - Disease: Cancer
  - Disease: Ophthalmology and Optometry
  - Other
  - Screening

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