UVEAL MELANOMA-RELATED GENE PANEL: GNAQ, GNA11 AND BAP1

Background: Uveal melanoma (UM) is a rare ocular cancer that affects the uveal tract, comprising the iris, ciliary body, and choroid. UM is the most common primary malignancy of the eye with an incidence of ~1,200 new cases diagnosed per year in the United States. Clinical features associated with increased risk for developing metastatic disease, most commonly to the liver, include large tumor diameter, ciliary body involvement, and epithelioid cell type. Chromosomal aberrations, particularly monosomy for chromosome 3 and amplification of chromosome 8q, or Class 2 classification by gene expression profile, predict a >50% risk for developing metastasis within 5 years of the initial diagnosis.

Initiating events in a normal uveal melanocyte toward becoming a nevus include activating mutations in GNAQ or GNA11. Melanomas undergo additional genetic changes. Mutations of BAP1, located at 3p21, and loss of the other copy of chromosome 3 have been identified in metastatic UMs.

BAP1 germline analysis is warranted in patients with ocular melanoma and a strong personal or family history of cancers.


Utility: Prognostic evaluation, clinical management, and prediction of metastasis-free survival.

Sensitivity: Testing targets specific gene mutations and does not detect mutations that are outside of the targeted area. The testing will detect alterations >99% of the time and the overall prognosis is poor in most patients who are found to have a GNAQ or GNA11 mutation in conjunction with bi-allelic loss of BAP1. In the event that there is admixture of the tumor sample with normal cells, the sensitivity can be compromised.

Turn around: 4-6 weeks

Fees: $1,200

CPT codes: 81403, 81404, 81406

