GERMLINE BAP1

Background: Germline BAP1 mutations are associated with a predisposition to uveal melanoma and malignant mesothelioma. Uveal melanoma is a rare ocular cancer that affects the uveal tract, comprising the iris, ciliary body, and choroid. Mesothelioma is a cancer that occurs in the tissue that lines internal organs in the chest and abdomen. It is often associated with exposure with asbestos. Other cancer risks that may be associated with a BAP1 germline mutation include cutaneous melanoma, atypical melanocytic tumors, renal and lung cancers. The full spectrum of cancer types and likeliness of developing those cancers is not currently well defined.

BAP1 testing is recommended for individuals who have a personal/family history of uveal melanoma, malignant mesothelioma, and/or other cancers included in the BAP1 spectrum.

Assay: Sequencing: Sanger sequencing of coding exons of BAP1.

Deletion/Duplication: Our lab offers a custom comparative genomic hybridization and single nucleotide polymorphism (CGH + SNP) array designed by Agilent technologies. This high-density array is designed to detect exonic and intronic copy number changes as small as 400 bp and 1.5kb, respectively, in the targeted gene(s). Additionally, the array can detect copy neutral loss of heterozygosity. The analysis of the targeted gene(s) is performed using Cytogenomics software (Agilent Technologies). These results may be confirmed by qPCR.

Utility: Clinical management

Sensitivity: The clinical sensitivity of BAP1 germline mutations in individuals is not currently established.


