Test Description

Genes tested

*BAP1*

Sequence analysis

Our lab performs sequence analysis of all *BAP1* coding exons (1-17) and flanking intronic regions. Our sequence analysis is able to detect mosaic variants at a level of 12.5% or greater. We consider reported polymorphisms when designing our sequencing assays to ensure the accuracy of our sequencing results.

Copy number changes

Our lab uses Multiplex Ligation-dependent Probe Amplification (MLPA) to look for whole-exon and multi-exon deletions and duplications in the *BAP1* gene.

Benefits of genetic testing

- **Develop tailored surveillance:** *BAP1-TPDS* carriers are at increased life-long risk for additional cancers. Recommended management has been established for *BAP1* carriers.

- **Bundle Testing:** Germline analysis can be ordered in conjunction with uveal melanoma prognostic testing.

- **Other family members:** By finding a familial *BAP1* pathogenic variant, other at-risk family members can determine their carrier status.

*BAP1-TPDS* is a uniquely variable condition. Examination and personal history may not be enough to determine if family members have inherited the familial mutation. The only way to determine with certainty if at-risk family members are carriers is genetic testing for a known familial *BAP1* variant.
BAP1-TPDS (BAP1 Tumor Predisposition Syndrome)

Genetic Test

Choose Impact: Excellence and expertise

- **BAP1 Expertise**: As contributing members of the ClinVar BAP1 working group, we review variants from BAP1-TDPS cases from around the world.

- **Ascribing value to variants**: When a variant is identified, thorough analysis is performed using the recommended guidelines [Richards et al 2015 PMID: 25741868]. Where needed to define a causative variant, we request family member samples to test at no added charge.

- **Reports**: We strive to provide as much useful information as possible. World-renowned medical experts contribute to cases involving complex interpretation.

- **Re-tests**: If no causative BAP1 variant is found, we bank any remaining DNA and re-test in the future when new science or test methods are available. For any new findings, we re-issue our report at no added charge.

- **Clinically appropriate turn-around time**:
  - Proband turn-around time is 3-9 weeks.
  - Known familial variant turn-around time is 2-3 weeks.

- **Certified lab**: Our lab is fully accredited and certified:
  - College of American Pathologists (CAP) and CLIA '88
  - Institute for Quality Management in Healthcare (IQMH) ISO 15189

- **Service excellence**: Impact is committed to exceptional customer service. Our team happily provides test order support so you can spend more time with your patients.

- **Logistics**: Impact provides genetic testing services to over 25 countries and samples are routinely shipped from across the world without disruption. We provide the necessary paperwork and recommend a courier service that will reliably deliver samples.