impact genetics

BAP1-TPDS (*BAP1* Tumor Predisposition Syndrome)

Genetic Test

Molecular testing expertise ⁞ Rare disease experts ⁞ Exceptional service

Test Description

Genes tested

BAP1

Sequence analysis

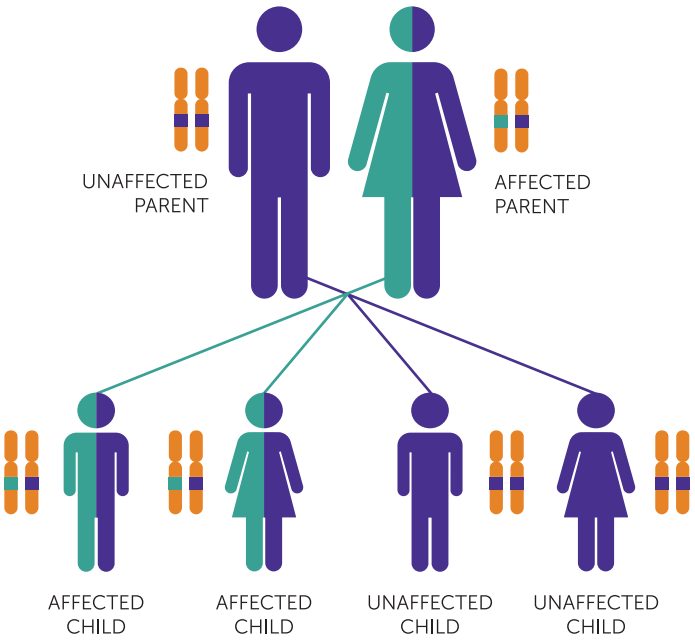
Our lab performs sequence analysis of the all *BAP1* coding exons (1-17) and flanking intronic regions. Our sequence analysis is able to detect mosaic variants at a level of 12.5% or greater. We consider reported polymorphisms when designing our sequencing assays to ensure the accuracy of our sequencing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

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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.