impact genetics

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Patient name:	--	**Referring specialist:**	--
		Contact person:	--
Date of birth:	--		
Medical record #:	--	**Institution:**	--
Patient history:	*Uveal Melanoma*		*Address:*
Family history:	*Positive family history*		*Fax:*
Date of report:	*MMM DD, YYYY*		
Family #:	--		
Patient #:	--	**Copies to:**	*Name*
			Fax:

Test requested: *BAP1 tumor predisposition syndrome (BAP1-TPDS) - proband*

Test results

A *BAP1* pathogenic variant has been identified.

Gene	Result
BAP1	c.1882_1885delTCAC, p.(Ser628ProfsTer8)
Methods used: Sequence analysis and multiplex ligation-dependent probe amplification (MLPA)	

Interpretation

- This patient is suspected to be affected with *BAP1*-TPDS. He is reported to have uveal melanoma (UM) and a family history of renal cell carcinoma and malignant mesothelioma.
- Sequence analysis of this patient's blood sample identified a heterozygous *BAP1* c.1882_1885delTCAC, p.(Ser628ProfsTer8) deletion. This deletion has been reported in the literature in two unrelated families with multiple affected individuals with UM and other *BAP1*-TPDS-related cancers (Pilarski et al 2014 PMID:24243779; Ohar et al 2016 PMID:26719535).
- **This deletion is predicted to be pathogenic and supports a diagnosis of *BAP1-TPDS.***

Guidance and recommendations

- Clinical correlation and genetic counseling is recommended. Genetic counseling services are available through LabCorp Integrated Genetics, on request (call 1-855-422-2557) [US only].
- The risk of inheriting this variant is 50% for each of this patient's children.
- Targeted testing for this variant can be offered to all at risk relatives.

These results and the interpretation, including guidance and supplemental information, were reviewed and approved by:

Electronically signed by Hilary Racher, PHD FCCMG DABMGG, Laboratory Director, on MMM DD, YYYY at <time>
Electronically signed by Brenda Gallie, MD FRCSC, Medical Director, on MMM DD, YYYY at <time>

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Details of samples tested

Patient Sample	Sample #	Collected	Received	Authorized/Test Started
Blood	*##-####*	*MMM DD, YYYY*	*MMM DD, YYYY*	*MMM DD, YYYY*

Supplemental information

Test Methods

- Sequence analysis of the all *BAP1* coding exons and flanking intronic regions.
- Gross deletion/duplication analysis was performed using multiplex ligation-dependent probe amplification (MLPA, SALSA P417-B1 BAP1 MRC Holland).

All *BAP1* variants reported to date are detectable by sequence analysis (Rai et al 2016 PMID: 26096145); the proportion of variants detectable only by copy number analysis methods is currently unknown.

Whenever possible, Impact Genetics banks DNA for future tests when a causative variant is not found. Outside of human error, our variant detection strategy shows 100% specificity.

Reporting: Non-pathogenic (benign) variants may not be included on reports but are available upon request. Classification of DNA variants may change over time as new information becomes available and where possible, reports will be re-issued if appropriate.

Test method limitations: Very low level mosaic variant carriers may not be detected by our methods. Our methods can detect mutant levels as low as 12.5% mutant DNA for most variant types. Most translocations or gross intronic re-arrangements cannot be detected by our methods. Deep intronic splice variants cannot be detected by conventional DNA analysis.

Notation: Variants are described using HGVS (v15.11) guidelines using the RefSeq accession # NM_004656.3 (*BAP1*). For coding DNA sequences, the A of the ATG initiator codon is denoted as nucleotide 1.

General disclaimer: This test was developed and its performance characteristics determined by Impact Genetics. It has not been cleared or approved by the Food and Drug Administration. Each of Impact Genetics' molecular tests use a direct method of variant detection and analysis is based on current knowledge of the genes. Characterization of a variant in a family does not preclude the remote possibility that a second, unidentified variant occurs in an individual patient. Moreover, it is possible for two relatives to have different gene variants.

Additional Information for clinicians and patients regarding the test performed and *BAP1*-TPDS is available at impactgenetics.com.