



Uveal Melanoma

Prognostic Genetic Test

Test Description

- Copy number testing using multiplex ligation-dependent probe amplification (MLPA) on chromosomes 1, 3, 6 and 8 to detect monosomy, disomy and trisomy.
- Microsatellite analysis (MSA) on chromosome 3 to detect chromosome copy loss and/or isodisomy.
- Sequencing of *GNAQ*, *GNA11*, *SF3B1* and *EIF1AX* to detect frequently occurring mutations in UM tumors.

Benefits of Genetic Testing

Prognosis: Factors contributing the survival prognosis of a patient with uveal melanoma include clinical tumor stage, histologic grade of malignancy and genetics of the tumor. These factors considered together provide the best indication of whether metastatic disease will develop and when this is likely to happen [Eleuteri, A. et al. 2012. *Int. J. Biom Eng and Tech*].

Individualized multivariant survivorship prediction: Genetic tumor information is combined with individual patient tumor histology and demographics. The Liverpool Uveal Melanoma prognostic algorithm (LUMPO) is used to produce a personalized survivorship prediction.

Patients want to know: Irrespective of result, patients report a desire for prognostic information in large part because they find it difficult to cope with uncertainty regarding their prospects for future health and survival. Neither mental health nor quality of life scores differ significantly based on test result [Beran, T. et al. 2009. *J. Genet Counsel*]. Prognostication may also contribute to treatment selection, possibly encouraging more aggressive therapy if the tumor is predicted to have a high risk of metastasis.

Sampling after radiotherapy: If sufficient tumor is still present, the full benefits of the test can be realized. Radiotherapy does not significantly affect the quality of samples for DNA analysis by Impact Genetics.

Confidence in good prognostic result: In many cases, this test will confirm that tumor was sampled. This is critical when the test result indicates good prognosis.

Metastatic disease surveillance: Reliable prognostication allows screening to be tailored to each patient, according to the level of risk. This would prevent patients with a good prognosis from being subjected to excessive investigations while targeting special care at high-risk individuals.

Sample requirements:

- A single FNAB pass usually provides sufficient tumor for testing, however a minimum of two passes is preferred to address tumor heterogeneity.
- Tumor and buccal samples for DNA analysis can be safely sent with up to 7 days in transit at room temperature, and can be safely stored for months.
- A biopsy may be taken from an enucleated eye at the time of plaque insertion or on the last day of proton beam radiotherapy.

Sample collection material is available on request.



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Choose excellence and expertise

Comprehensive multivariant testing: Impact Genetic's Uveal Melanoma Prognostic Genetic Test combines multiple techniques in order to provide the most comprehensive result available.

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

Re-tests: We bank any remaining DNA. In the future, when new science or test methods are available, we will contact referring specialists to offer to re-test. For any new findings, we re-issue our report at no added charge.

Certified lab: Our lab is fully accredited and certified: College of American Pathologists (CAP) and CLIA '88, SO 15189 Plus and Ontario Lab Accredited (OLA), New York State CLEP, European Molecular Genetics Quality Network.

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

Logistics: Impact provides genetic testing to customers around the world. Samples are routinely and reliably shipped with the support of our dedicated customer service team.

Ordering: Please visit impactgenetics.com for details

Turnaround time: 3-6 weeks

