

Uveal Melanoma

Background: Melanotic lesions are the most common primary intraocular tumors. There are two categories of lesions: benign nevi and malignant melanomas. Race is a strong risk factor, with both occurring 15 times more often in Caucasians than in African-Americans.

While a benign choroidal nevus is a common fundus finding, a malignant choroidal melanoma is only seen in six cases per million.

Conventional cytogenetic and comparative genomic hybridization (CGH) assays have revealed non-random aberrations. Monosomy for chromosome 3 has been found in 50% of tumors and is a significant predictor of metastatic disease and very poor overall prognosis. It is possible that adjuvant therapy can improve the prognosis if monosomy for 3 can be detected early enough.

Assay: DNA micro-satellite marker based testing for presence or absence of two copies of chromosome 3.

Utility: Prognostic evaluation, clinical management

Sensitivity: If the patient has complete monosomy for chromosome 3, the testing will detect the alteration >99% of the time and the overall prognosis is poor in most of these patients. It is also possible to detect a partial monosomy for chromosome 3 and the prognostic significance of a partial monosomy is being studied but has yet to be determined for risk of metastatic disease. Disomy for chromosome 3 is usually associated with a good prognosis. In the event that there is admixture of the tumor sample with normal cells, the sensitivity can be compromised.

Turn around: 8-10 weeks

Fee: \$1000

CPT codes: 83891, 83900x4, 83901x9, 83907, 83909x3, 83912

Uveal Melanoma Testing INSTRUCTIONS FOR SAMPLE SUBMISSION

Documentation: Each sample must be accompanied by:

1. A requisition for DNA analysis completed by the physician, nurse or genetic counselor requesting screening. ***Please note: ICD-9 code is required for billing purposes. If ICD-9 code is unknown, please provide patient's clinical symptom(s) or family history that prompted testing.***
2. An informed consent signed by the patient and the professional obtaining the consent. Please have the patient initial at the top of each page and send **all** pages of the consent.
3. A completed registration form with check, money order, credit card authorization or information for billing the referring institution.

IN THE EVENT THAT ALL PROPERLY COMPLETED FORMS DO NOT ACCOMPANY THE SPECIMEN, YOU WILL BE NOTIFIED, AND TESTING WILL BE HELD UNTIL PAPERWORK IS COMPLETE.

Preparing Sample:

- Fine needle aspirates of biopsy samples collected in HBSS before plaque therapy, stored at 4 degrees C. and shipped on ice.
- 2 EDTA tubes of blood (lavender top) to send with tumor aspirates (at least 4mL in each tube).
- Label each collected sample with the patient's name and date sample was obtained.

Shipping Sample: Ship at room temperature via Federal Express or other overnight courier that guarantees AM delivery to arrive Monday-Friday. There is no one in the laboratory evenings and weekends to receive samples. If sample is drawn on a Friday, please refrigerate it until shipment on the following business day.

Shipping Address: Genetic Diagnostic Laboratory
University of Pennsylvania
415 Anatomy-Chemistry Building
3620 Hamilton Walk
Philadelphia, PA 19104

GENETIC DIAGNOSTIC LABORATORY
UNIVERSITY OF PENNSYLVANIA SCHOOL OF MEDICINE
DEPARTMENT OF GENETICS
Tel: (215) 573-9161 • Fax: (215) 573-5940
CLIA ID: 39D0893887

REQUEST FOR UVEAL MELANOMA TESTING

PATIENT FIRST NAME _____ PATIENT LAST NAME _____

BIRTH DATE _____ SEX _____ RACE _____ ETHNICITY _____

STREET ADDRESS _____

CITY _____ STATE _____ ZIP _____ HOME PHONE _____

Does the patient have IRIS ___ or CHOROIDAL ___ MELANOMA ? Which EYE is affected? LEFT ___ RIGHT ___
PATIENT'S IRIS COLOR:
BLACK ___ BROWN ___ BLUE ___ OR GREEN ___ ?

ARE THERE ANY FAMILY MEMBERS WITH
DERMAL MELANOMA ___ No ___ Yes, relationship _____
BASAL CELL CARCINOMA ___ No ___ Yes, relationship _____
SQUAMOUS CELL CARCINOMA ___ No ___ Yes, relationship _____
DOES PATIENT HAVE DERMAL NEVI? ___ No ___ YES, HOW MANY _____

WHAT IS THE PATIENT'S OCCUPATION: (or former occupation if retired)? _____
WHAT PRESCRIPTION MEDICATIONS IS THE PATIENT TAKING?

Does the patient have a history of smoking? ___ No ___ Yes, How many years? _____

REFERRING PHYSICIAN _____ PHONE _____ FAX _____

GENETIC COUNSELOR _____ PHONE _____ FAX _____

EMAIL ADDRESS FOR COUNSELOR OR PHYSICIAN _____
INSTITUTION and DEPARTMENT _____

STREET ADDRESS _____

CITY _____ STATE _____ ZIP _____ COUNTRY _____

ICD-9 CODE _____

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Patient Registration Form

Please provide the following information. We cannot perform your test without ALL of this information. PLEASE PRINT ALL ANSWERS

PATIENT INFORMATION

LAST FIRST MI

Date of Birth: _____

Gender: ___ Male ___ Female

Patient

Address: _____
STREET/APT NO.

CITY, STATE, ZIP COUNTRY IF OUTSIDE UNITED STATES HOME TELEPHONE

REFERRING PHYSICIAN

NAME MD DO OTHER INSTITUTION

STREET ADDRESS CITY, STATE, ZIP

TELEPHONE FAX

PAYMENT OPTIONS (must choose one) [a receipt will be mailed to patient for self-pay options]

_____ I have enclosed a check payable to the "Genetic Diagnostic Laboratory" for \$ _____

_____ Please charge my credit card for the amount of \$ _____
___ VISA ___ Master Card ___ Discover ___ American Express

Card Number: _____ Exp date: _____

Name as it appears on card: _____

_____ I have Pennsylvania Medicaid. A copy of my Medicaid card is attached.

_____ INSTITUTIONAL BILLING: The Institution where my testing originated has agreed to pay all charges for the testing. INCLUDE Billing Address, Person Authorizing Payment, Telephone, and Fax below:

BILLING ADDRESS

BILLING ADDRESS

PERSON AUTHORIZING PAYMENT PHONE FAX

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Informed Consent for Molecular Testing of Uveal Melanoma Tumor

I, _____, hereby request testing for **monosomy of chromosome 3**, using a molecular test. I understand that a sample of my frozen tumor tissue or fine needle aspirate biopsy (FNAB) will be used along with analysis of a blood sample. The results of the genetic analysis can indicate a risk for developing metastatic disease from the uveal melanoma.

I understand that:

1. In some cases the molecular test directly detects an abnormality, called monosomy of chromosome 3, in the tumor/FNAB specimen. The test is >99% accurate. There is a chance that the tumor has an alteration that will not be detected by this testing, or that an alteration of unknown significance may be identified.
2. The molecular analysis performed at the University of Pennsylvania Genetic Diagnostic Laboratory in no way guarantees my health.
3. These tests are relatively new and are subject to change periodically to improve or expand the utility of the test. The tests are not considered research but are considered to be the best and newest laboratory service available. This testing is often complex and utilizes specialized materials so that there is a small possibility that the test will not work properly or that an error will occur. My signature below acknowledges my voluntary participation in this testing, but in no way releases the laboratory and staff from their professional and ethical responsibility to me.
4. Because of the complexity of molecular based testing and the important implications of the test results, results will be reported to me only through the physician who requested the testing. The results are confidential; they will only be released to other medical professionals or other parties with my written consent. Participation in molecular testing is completely voluntary.
5. I understand that any blood or tissue specimens obtained for the purposes of this genetic testing become the exclusive property of the Genetic Diagnostic Laboratory. After the specific tests requested have been completed and reported, the Laboratory may dispose of, retain, or preserve these specimens and may use these specimens for research. I understand that my identity will be protected and that research results will not be provided to me or to any other party. If use of this genetic material results in a scientific publication, it will not contain any identifying information. I understand that the Genetic Diagnostic Laboratory is not a DNA banking facility and my DNA or tumor sample may not be available for future clinical studies.

Physician's Statement: I have explained molecular testing for uveal melanoma to this individual. I have addressed the limitations outlined above, and I have answered this individual's questions.

Signature _____ Print Name _____

Date _____

I agree to undergo the genetic analysis, and I have had the opportunity to ask questions about the testing.

Patient's Signature _____ Print Name _____

Patient's Date of Birth _____ Date Signed _____