Personalized Medicine testing recommendations for:

# Melanoma

## **Genetic Risk**, Cutaneous

**No germline biomarker testing is recommended by NCCN with the exception of risk stratification. Genetic counseling and testing if any of the following are indicated:**

**When and what should be tested**?

- Upon or before presentation
- Peripheral blood, saliva or buccal mucosa swab

**Who should be tested?**

- Consider genetic counseling referral for p16/CDKN2A mutation testing in the presence of 3 or more invasive cutaneous melanomas, or a mix of invasive melanoma, pancreatic cancer, and/or astrocytoma diagnoses in an individual or family.
- Multigene panel testing that includes CDKN2A is recommended for patients with invasive cutaneous melanoma who have a first-degree relative diagnosed with pancreatic cancer
- Presence of germline mutations or polymorphisms predisposing to melanoma (eg, CDKN2a, CDK4, MC1R, BAP1 [especially for uveal melanoma], TERT, MITF, PTEN, and potential other genes).
- Family history of cutaneous melanoma (especially if multiple); pancreatic, renal, and/or breast cancer; astrocytoma; uveal melanoma; and/or mesothelioma.
- CDKN2a, CDK4, MC1R, BAP1 (including uveal), TERT, MITF, PTEN, xeroderma pigmentosum

**FDA/NCCN Approved**

- CDKN2a
- CDK4
- MC1R
- BAP1
- TERT
- MITF
- PTEN

**Emerging**

Multigene panel testing for p16/CDKN2A mutation testing

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Personalized Medicine testing recommendations for:

# Melanoma

## **Genetic Risk**, Uveal

**No germline biomarker testing is recommended by NCCN with the exception of risk stratification. Genetic counseling and testing if any of the following are indicated:**

**When and what should be tested**?

- Upon or before presentation
- Peripheral blood, saliva or buccal mucosa swab

**Who should be tested?**

- Early age at diagnosis (<30 years of age)
- History of other primary cancers in the patient
- Family or personal history of other cancers known to be associated with a hereditary syndrome:
  - BAP1: RCC, basal cell, HCC, mesothelioma, cutaneous melanoma, cholangiocarcinoma, meningioma
  - BRCA, PALB2: breast, ovarian, or pancreatic cancers

**FDA/NCCN approved – Risk of distant metastasis**

Low Risk

- Disomy 3
- Gain of function chromosome 6p
- *EIF1AX* mutation
- PRAME(-)

Medium Risk

- *SF3B1* mutation
- PRAME(+)

High risk

- Monosomy 3
- Gain of chromosome 8q
- *BAP1*

**FDA/NCCN approved – Risk of development of uveal melanoma**

- *BAP1*
- *MBD4*
- *PALB-2*

**Emerging**

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Personalized Medicine testing recommendations for:

# Melanoma

## Cutaneous (somatic)

**When and what should be tested?**

- Treatment eligible
- Metastatic disease
- At time of initial diagnosis or recurrence
- Tissue or liquid

### FDA/NCCN approved stage III

- *BRAF*

### FDA/NCCN approved stage IV

- BRAF V600E, BRAF V600K, BRAF V600R,M,D,G
- NTRK 1,2,3
- TMB
- dMMR
- KIT
- NRAS
- ALK
- ROS1
- RET
- HER2
- MRD

### Emerging

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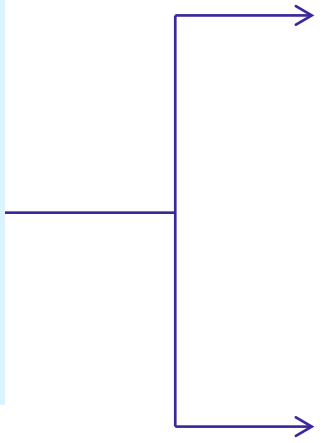
Personalized Medicine testing recommendations for:

# Melanoma

## Uveal (somatic)

**When and what should be tested?**

- Treatment eligible
- Unresectable or metastatic disease
- At time of initial diagnosis or recurrence
- Tissue or liquid (liquid only at recurrence unless another tissue biopsy obtained)



**FDA/NCCN approved**

- HLA-A *02:01
- For metastatic disease:
  - BRAF V600E
  - NTRK 1/2/3
  - RET
  - TMB
  - dMMR
  - MRD

**Emerging**

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