CLINICAL SUMMARY

# Testing for Hereditary Melanoma

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The number of new melanomas diagnosed in the United States is rising rapidly, with over 59,000 new cases diagnosed each year. A recent article confirms the importance of *CDKN2A* (which includes the *p16* and *p14* genes) screening of patients with multiple primary melanomas.

## Puig S, et al. Role of the *CDKN2A* Locus in Patients with Multiple Primary Melanomas. Journal of Clinical Oncology 2005;23:13.

### Purpose:

To determine the frequency of *CDKN2A* and *CDK4* mutations in patients with multiple primary melanomas.

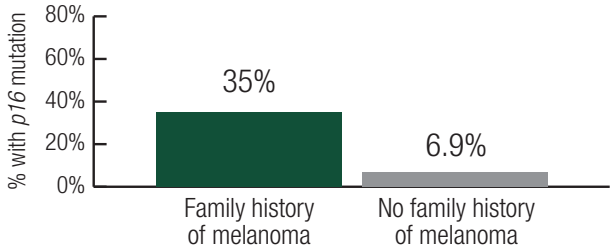
### Study Population:

One hundred four men and women with at least 2 primary melanomas were included in the study.

### Results:

Sixteen patients (15.4%) had a detectable mutation in *p16* and one patient (~1%) had a mutation in *p14ARF*. No mutations were identified in *CDK4*. The following table describes the likelihood of identifying a *p16* mutation based on the number of primary melanomas in an individual. The graph illustrates the likelihood of a *p16* mutation in multiple primary melanoma patients with and without family history of melanoma.

| # of melanomas diagnosed in patients | % with *p16* mutation |
|---|---|
| 2 | 8.7 |
| 3 - 7 | 39.1 |




- The age range in this study was from 9 to 83 years. The mean age of onset was 44 years. The mean age of onset was significantly lower in carriers of mutations compared with non-carriers.
- *p16* mutations accounted for 94% of all mutations identified.
- Melanoma patients with early age of onset, positive family history, and the presence of two or more melanomas are the most likely to harbor *CDKN2A* mutations.

### Bottom Line:

Patients with multiple primary melanomas, even in the absence of family history, should consider screening for *CDKN2A* mutations. These high-risk patients benefit from enhanced surveillance and early detection of melanoma.

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Clinical Support, Healthcare Information and Customer Services: 800-4-MYRIAD (800-469-7423)

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Myriad Genetic Laboratories, Inc.
320 Wakara Way, Salt Lake City, UT 84108

*For more information:*
**www.myriadpro.com** 800-4-MYRIAD (800-469-7423)