

# Identification of Pathogenic Variants in Patients with Melanoma who Meet NCCN Criteria for Hereditary Breast and Ovarian Cancer and Lynch Syndrome Testing

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## INTRODUCTION

Currently, *CDKN2A* and *CDK4* have primarily been implicated in familial melanoma. However, pathogenic variants (PVs) in these genes represent only a small percentage of familial melanoma cases, indicating that other genes may be involved.

## METHODS

We evaluated the results of a multi-gene hereditary cancer panel among individuals tested between 9/2013 and 7/2019 who met NCCN testing criteria according to melanoma history.

- Personal history (PHx) and/or family history (FHx, first degree relatives only) of melanoma (N=27,946)
- No PHx or FHx of melanoma (N=581,224)

## RESULTS

Figure 1. Cancer History for Patients with Personal History of Melanoma

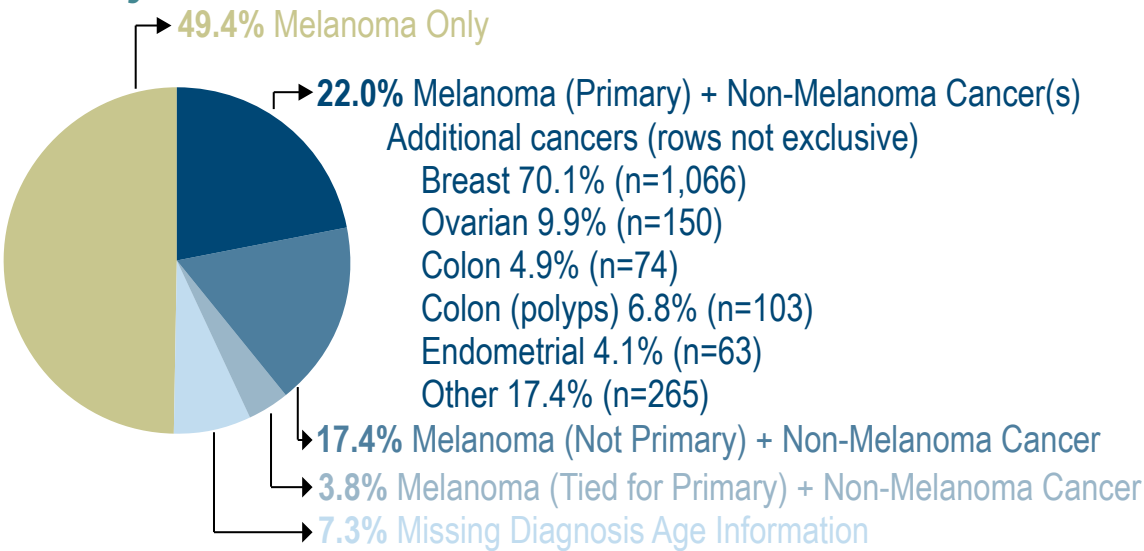
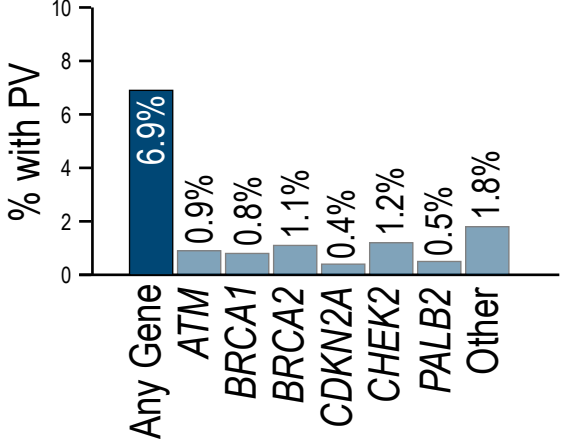


Figure 2. Distribution of Pathogenic Variants Among Tested Genes

### PHx and/or FHx of Melanoma

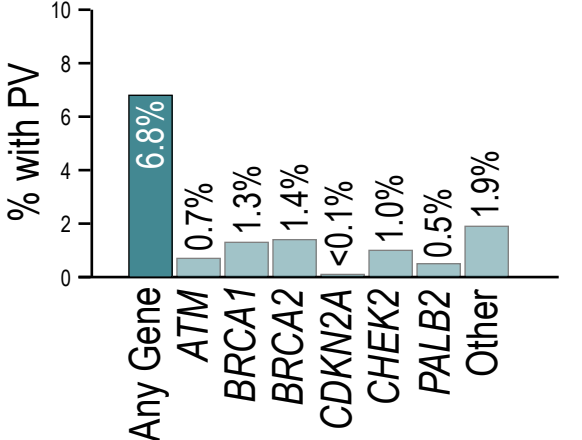


Among individuals with a PHx and/or FHx of melanoma, PVs were most common in *ATM*, *BRCA1*, *BRCA2*, *CHEK2*, and *PALB2*.

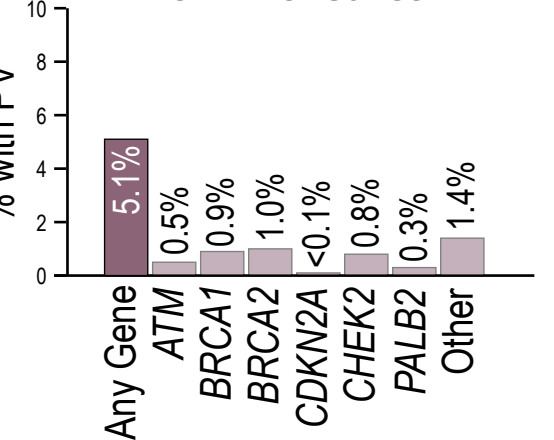
The prevalence of PVs in *ATM*, *CHEK2*, and *PALB2* was similar to that observed for individuals with a history of a non-melanoma cancer.

**PV Positive Rate by Melanoma History:** PHx and FHx = 8.9% (108/1,208); PHx Only = 7.7% (534/6,907); FHx Only = 6.8% (1,506/22,247)

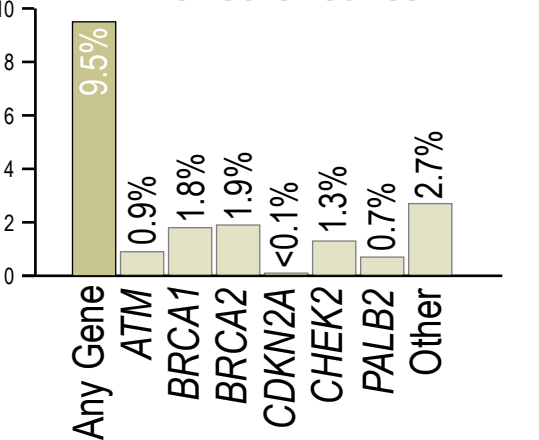
### No PHx/FHx of Melanoma



### No PHx of Cancer



### PHx of Other Cancer



## CONCLUSIONS

Early identification of PV-carrying patients with melanoma would present provider and patient with an opportunity to prevent the occurrence of a second primary cancer through increased surveillance or risk-reducing surgeries.

Identification of patients with concerning family cancer histories eligible for hereditary cancer testing according to NCCN guidelines during dermatology appointments may lead to positive health outcomes by preventing future cancers.