

# Cancer researchers discover gene defect responsible for cancer syndrome

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University of Hawai'i Cancer Center researchers have discovered germline BAP1 mutations are associated with a novel cancer syndrome characterized by malignant mesothelioma, uveal melanoma, cutaneous melanoma and atypical melanocytic tumors. Germline mutations are hereditary gene defects that are present in every cell.

The study investigated two unrelated families with BAP1 defects and found an increase in the occurrence of mole-like melanocytic tumors that are non-cancerous flat or slightly elevated and pigmented [skin lesions](#). These benign skin lesions were found to carry the BAP1 mutation and it was concluded that people with this specific type of melanocytic lesion are at higher risk of developing melanoma and mesothelioma.

This discovery provides physicians with a visual marker in identifying individuals that may carry germline BAP1 mutations. People having this syndrome should reduce their exposure to [environmental risk factors](#) such as UV radiation for melanoma and avoid erionite and asbestos exposure for mesothelioma. It will also help identify individuals who are at higher risk for melanomas which is usually cured with timely detection and to assist in the early detection of mesothelioma which typically leads to better prognosis.

"Identifying this gene as a cause of several cancers can tell us who is at risk in a family before the cancer develops," said Michele Carbone, MD, PhD, director of the UH Cancer Center and professor of pathology, John

A. Burns School of Medicine. "We can advise patients to undergo routine exams and genetic testing for early diagnoses and treatment." Carbone is the leading author of the paper included in the August 30, 2012 issue of the [Journal of Translational Medicine](#), published by [BioMed Central](#).

This novel gene-testing has been patented by Carbone and colleagues and is performed exclusively at The Queen's Medical Center in Honolulu, Hawaii where they receive genetic samples from across the entire United States. Carbone previously discovered that individuals who carry BAP1 mutations are susceptible to developing mesothelioma and melanoma of the eye. This latest discovery builds on this and other research on the BAP1 mutation.

**More information:** [www.translational-medicine.com ...  
nt/10/1/179/abstract](http://www.translational-medicine.com/abstract/10/1/179/abstract)

Provided by University of Hawaii at Manoa

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