Huntsman Cancer Institute Leads International Consortium to Develop First Ever Genetic Testing Guidelines for Melanoma

October 8, 2009, Salt Lake City—An international coalition of physicians and scientists, led by Huntsman Cancer Institute (HCI) at the University of Utah, has proposed guidelines for the first time concerning genetic testing for melanoma patients.

The guidelines were developed from analysis of worldwide data on patients harboring a mutation in a gene known as CDKN2A. “Mutations in this gene are associated with an increased risk for both melanoma and pancreas cancer,” says Sancy Leachman, M.D., Ph.D, author of the large, multi-center study.

Approximately 5-10 percent of melanomas may be hereditary in nature, and about 2 percent of melanomas can be attributed to gene mutations. The percentage of families that carry a CDKN2A mutation varies depending on the number of family members who have had melanoma, the number of melanomas per patient, and whether a family member has had pancreatic cancer.

But that’s just part of the story. Researchers already knew there were variables based on the country of origin, but the new research takes this information a step further. “Rates of CDKN2A mutations vary
from country to country, but our research identifies which patients fit the criteria for referral for melanoma genetic testing based on their country of origin,” says Leachman. “For instance, a criterion that applies to Australia is not the same that would apply to Northern Europe, or the United States.” The data is significant because it led researchers to the conclusion that certain people are good candidates for genetic counseling and outlined guidelines for the first time. “Genetic counseling is now commonly used to identify individuals with hereditary colorectal, breast and ovarian cancer,” says Leachman. “But genetic counseling for a CDKN2A mutation is not part of routine practice, despite the fact that commercial tests are available. We maintain that clinicians can help identify individuals at greater risk by referring them to a genetic counseling specialist,” she says. “Identifying the risk may help prevent melanomas from occurring through screening and earlier detection.”

The research took two years to complete and involved the cooperation of physicians and scientists throughout the world. According to Leachman, there are so many variables surrounding this gene mutation, it is not possible to develop a single guideline for genetic testing that would be appropriate worldwide. “Our study provides a framework that clinicians can use to identify appropriate candidates for genetic evaluation based on where they live and their clinical features,” Leachman says.

Leachman is the director of the Melanoma and Cutaneous Oncology Program at HCI and an associate professor in the Department of Dermatology at the University of Utah. The study is published in the October issue of *Journal of the American Academy of Dermatology*. 