

Increased risk of skin cancer in Japanese heterozygotes of xeroderma pigmentosum group A—Mutation carriers of an XP gene are at moderately elevated risk of developing skin cancer

Genetic disorders represent diseases caused by inactivation of certain genes. Xeroderma pigmentosum (XP), one such genetic disorder, is characterized by extreme sensitivity to sunlight (ultraviolet light) and by the development of multiple skin cancers starting in childhood. Those with this XP inherit an abnormal XP gene from each parent, with the disorder frequency being about 1 in 20,000 people. In contrast, the frequency of those who bear the mutant gene in only one of the two XP genes (called “mutation carriers,” who are thus not sensitive to sunlight) is much more prevalent, at around 1 in 30 people.

This study found that the XP mutation carriers are only at moderately elevated risk of developing skin cancer when older on sun-exposed areas of the body (e.g., face or arm) compared with non-carriers.

Recent studies indicate that we all bear several tens of mutated genes under such carrier conditions (where only one gene is mutated out of two). There are individual differences in susceptibility to disease development, and the above-described genetic differences may be involved, at least partly, in such differences.

* Xeroderma pigmentosum (XP) is a hereditary disease characterized by young onset of skin cancers on sun-exposed areas of the body at a frequency several-thousand times higher than in those without the disease. This is caused by inactivation of genes that function in repairing DNA damage induced by exposure to ultraviolet light.

RERF's objective with this brief outline is to succinctly explain our research for the lay public. Much of the technical content of the original paper has been omitted. For further details about the study, please refer to the full paper published by the journal.