

RISING DERM STARS

Non-Invasive Buccal Swab Gene Testing for Skin Cancer Risk

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Background: The awareness of skin cancer is increasing, and there are new commercial kits on the market which advertise the ability to provide a patient with a skin cancer risk profile. These tests are based on research findings that individual single nucleotide polymorphisms (SNPs) are associated with increased risk of melanoma and non-melanoma skin cancer (NMSC). However, to our knowledge, there is no current study in the literature that examines the prevalence of an extended set of identified SNPs within the skin cancer and environmental control patient population. In this pilot study, we aimed to examine whether previously identified melanoma and non-melanoma associated SNPs which were found to be present in a relatively heterogeneous population with a history of skin cancer versus an age and environmental matched controls. The undertaking of this project serves to further the current understanding of the genetic profile for those at higher risk for developing skin cancer.

Methods: Patients with biopsy-proven NMSC were recruited from a private Mohs micrographic surgical practice in Southern California. Nineteen NMSC patients and their age-matched and environmental control spouses underwent genotyping of 7 previously discovered SNPs associated with melanoma and NMSC via buccal swabs.

Results: In a random, heterogeneous population in Southern California, SNP's Chr1, PAD16, PIGU, TDG had a similar association with NMSC previously reported in prior studies. Due to small trial size, no conclusions or observable associations could be drawn from the SNPs MC1R, TP53, and XRCC1.

Conclusion: This data supports that 4 of the 7 SNP's studied had similar associations and could be a predictive tool of NMSC risk. The remaining three SNP's did not have a definitive association with malignancy. Previous trials predominantly studied more homogeneous patient populations (Icelandic, English) or people who lived in locations with more inclement weather. A better understanding of the genetic contributions that can affect DNA damage will help to prevent DNA damage and target with precision medicine DNA repair therapeutics in the future. Larger studies are needed to further elucidate the specific roles of these SNPs collectively and ultimately to develop a genetic profile and prevention techniques for those patients at increased risk of developing skin cancer.