

## Genetic Signature of Skin Aging: A Pilot Study

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The sun plays a major role in prematurely aging our skin. Our skin is at the mercy of many forces as we age: sun, harsh weather, and bad habits. But we can take steps to help our skin stay supple and fresh-looking. Sunlight is a major cause of skin aging. The increasing knowledge of the genetic bases of several common multifactorial diseases paves the way to personalized medicine that means preventive and therapeutic interventions that are tailored to individuals on the basis of their genetic profiles. The aging process, as well as multi factorial diseases, depends on a complex crosstalk between intrinsic (genetic and hormonal) and extrinsic (nutrition, lifestyle etc.) factors. Skin changes are the most visible signs of senescence process, and a field of increasing interest in a society that places more and more interest in appearance and beauty. Skin disease characterized by increased trans-epidermal water loss and skin barrier abnormalities and disruption. On this subject, numerous environmental events such as: chemical injuries, traumatic wounds, UV exposure and genomic characteristics can compromise the barrier activity. To date, the mutational spectrum of FLG gene comprises different variations that show an ethno-specific distribution profile, especially among north European and Mediterranean populations.

The study was conducted in 100 Italian volunteers, with an age between 21 and 66 years old (23 males and 77 females). All selected patients were Caucasian (people with European origin), belonging to Fitzpatrick skin type 2 and 3. Subjects underwent medical history and clinical examination; exclusion criteria included systemic diseases or presence of genetic diseases which were clinically evident. All patients signed written informed consent. The examination of each subject was conducted using a lifestyle questionnaire and, to evaluate the impact of

lifetime sun exposure (LSE), using the Sun Exposure and Behavior Inventory (SEBI).

For the aging process, like to multi-factorial and polygenic diseases, is known that single genetic polymorphism has only a modest effect since the interaction of each gene and its polymorphism with other ones (gene-gene interaction) and with environmental factors (gene-environment interaction) has a crucial role in the development of the pathology. Moreover, the diversity of ethnic background may be a possible bias in such research. In the light of these considerations we selected Caucasian, Italian subjects only, and we constructed a literature based genetic risk score for skin aging with the aim to evaluate the contribution of individual genetic variability to skin aging.

Prior skin aging studies have analyzed intrinsic and extrinsic skin aging parameters, believed to reflect genetic and environmental factors contributing to skin aging feature. Heritability analyses in twins have shown that genetic component of skin deterioration process accounts for about 60% [3]. In recent years, research on genetic polymorphisms indicate that Genetic Risk Scores (GRSs) allow the composite assessment of genetic risk in complex traits. Although some authors have expressed doubts as to whether a candidate gene approach can ever add significantly to risk prediction, because of the modest impact on risk, and the apparent inconsistency of effect, other authors demonstrate that, depending on the prevalence and heritability of the disease, few genetic variants may have a strong predictive power.

Our results indicate that the use of the genetic risk score including 8 single nucleotide polymorphisms

involved in aging process as previously described in literature, could be promising to predict skin properties evolution and address anti-aging and skin treatment against specific metabolic target.

Keywords: Aging skin; Skin elasticity