ATOPIC ECZEMA/DERMATITIS

ATOPIC ECZEMA: GENETIC ANALYSIS OF COL6A5, COL8A1 AND COL10A1 IN MEDITERRANEAN POPULATIONS

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Introduction: Atopic Eczema (AE) affects 1-3% of the adult population and 15-20% of children, depending on population and geographical localization. The variable prevalence across the world might reflect the influence of different UV exposure with respect to latitude and geographic localization. This variability may be responsible of a different genetic adaptation to local environment which ultimately determine a population-specific susceptibility to AE. The genes which have been identified so far are essentially implicated in the immune and inflammatory responses and in the maintenance of skin barrier. Little is known about the possible relationship between genes modulating Extra-Cellular Matrix (ECM) and AE etiopathogenesis.

Objective: the present study investigated the presence of susceptibility biomarkers localized within COL6A5 (rs12488457, A/C), COL8A1 (rs13081855, G/T) and COL10A1 (rs3812111, A/T) genes.

Materials and Methods: 1470 Mediterranean subjects (285 Italians, 87 Egyptians, 56 Greek and 992 controls) were genotyped by Real Time-PCR and subjected to biostatistic analysis using chi-square test and evaluation of ORs. The potential pathogenetic impact of the associated genes was then investigated by bioinformatic tools.

Results: biostatistical analysis revealed a positive association between rs13081855 and AE in Egyptian [p=0.03, OR(T)=1.77 (1.04 - 2.99)] and Italian [p=1.32*10-6, OR(T)=2.20 (1.58 - 3.05)] cohorts, but not among Greek patients. The rs12488457 and rs3812111 did not reveal any significant association among Mediterranean populations. Bionformatic results

showed that COL8A1 is involved in angiogenesis and ECM organization, explaining thereby its association with AE. On the other hand, the cellular and molecular function of COL6A5 (collagen metabolism, cell adhesion, ECM interactions) and COL10A1 (ECM organization, bone metabolism) explained the lack of association with AE.

Conclusions: The study highlighted that the susceptibility to the disease depends not only on latitude and geographical localization but also on the heterogenous distribution of genetic variants among populations exposed to similar environmental factors.