Association of COL1A1, COL5A1, MMP1 gene polymorphisms with skin metabolism in a Caucasian Greek population

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Chart 1. Comparative chart with genotypes incidence rates of polymorphisms rs1800012, rs12722 and rs1799750 in Greek, Global and European population.

Background

Collagens are the most abundant proteins in the human body. Collagen molecules are constituted of three alpha (α) polypeptidic chains. COL1A1 and COL5A1 genes encode α1 chains of collagen type I and type V, respectively. In this study, we will focus on the variants rs1800012 of COL1A1 gene and rs12722 of COL5A1 gene, which interfere in the collagen structure and the rs1799750, a variant of the MMP1 gene which induces the collagen metabolism. The rs1800012 (G/T) showed association with skin thickness and keloid scars. The rs12722 (C/T) was associated with a connective tissue disorder, the Ehlers-Danlos classic type Syndrome. The rs1799750, an insertion polymorphism in the MMP1 gene (-/G) was associated with dystrophic epidermolysis bullosa, a skin fragility disorder.

Purpose

Through this study we aim to reveal the genetic penetrance of the variants as well as to estimate the susceptibility of developing dermopathies in a sample of 1402 Caucasian volunteers. Finally we hope that our findings can contribute to the utilization of personalized therapy for a more efficient treatment of the aforementioned pathological condition.

Materials and Methods

DNA extraction from oral epithelial cells, followed by real time PCR assays led to the determination of the individuals' genotype, which were processed thus enabling the calculation of the variants frequencies.

Poly-morphism	Alleles	Frequency	%	Geno-types	Frequency	%
rs1800012 (COL1A1)	G	2236	79,74	GG	896	63.91
	T	568	20,26	TG	444	31,67
				TT	62	4,42
rs12722 (COL5A1)	C	1227	43,45	CC	275	19,61
	T	1597	56,55	CT	677	48,29
				TT	450	32,10
rs1799750 (MMP1)	1G	1416	50,50	1G1G	365	26,03
	2G	1388	49,50	1G2G	686	48,93
				2G2G	351	25,04

Table 1. Frequency distribution of alleles and genotypes of polymorphisms rs1800012, rs12722 and rs1799750 .

Results

For rs1800012 the frequency of the G allele in our study group was 79,74% and of the mutated T, 20,26%. For rs12722 the 43,45% of population carried the C allele and the 56,55% the mutated T. For rs1799750 the 49,50% of the volunteers presented a G insertion in the MMP1 gene.

Conclusions

The comparison of our results with the frequency distribution of studied genetic variants in the European population, presented no significant differences between the two populations. These findings coincide with our expectations due to the fact that our volunteers are characterized as South-Eastern Europeans, despite the influence of populations with different origins.

Sources

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