Association between the angiotensin-converting enzyme (ACE) insertion/deletion (I/D) polymorphism and pterygium in Sardinian patients

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Pterygium is a common ocular surface disorder characterized by proliferation, inflammatory infiltrates, fibrosis, angiogenesis and extracellular matrix breakdown. Epidemiological studies indicate exposure chronic to UVB light as the most important risk factor for the development of pterygium. The Angiotensin Converting Enzyme (ACE) is the major component of the Renin-angiotensin system (RAS). It converts the inactive decapptide Angiotensin I (Ang I) to the active octapeptide Angiotensin II (Ang II). Ang II is the most potent vasoconstrictor and stimulant of the aldosterone release. Recent discoveries have demonstrated that Ang II is also involved in cell proliferation, apoptosis, angiogenesis and tissue fibrosis. Moreover, it acts as growth factor and participates in inflammatory responses. The gene encoding ACE is mapped on chromosome 17q23; it contains 25 introns and 26 exons and shows a polymorphism characterized by the presence (insertion, I) or absence (deletion, D) of a 287-bp Alu sequence of DNA in intron 16. The presence or absence of Alu sequence in the ACE gene leads to the D/D, I/D and I/I genotypes. Novel studies have reported that the absence or presence of specific ACE I/D polymorphisms within ACE gene in several illnesses, such as cardiovascular diseases and breast cancer, can confer increased risk to develop the pathologies. Due to these evidences and the pterygium features, the aim of our study is to evaluate the ACE I/D gene polymorphism type in a group of Sardinia pterygium patients and to establish the possible correlation between ACE-type polymorphism and the development of pterygium in a case-control study.

Keywords: pterygium, RAS, Angiotensin II, ACE I/D polymorphism