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RP1 gene polymorphisms and Retinitis pigmentosa: A possible genotype-phenotype correlation?

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Introduction: Retinitis pigmentosa (RP) is a genetic disease involving the retina, the eye back portion, photosensitive and appointed to focus light signals towards the optical nerve and brain, after photo transduction. It is an uncommon condition affecting about 1/4,000 people in the USA and 1-5/10,000 in Italy. The term “pigmentosa” deals with the characteristic appearance of abnormal areas of pigment into the retina. Degeneration induces a slow and progressive death in photoreceptors and retinal pigment epithelium, losing ability to transmit brain the visual information. Today about 50 RP causative genes are known. We focused our attention on RP1, encoding a microtubule associated protein (MAP) needed for molecule trafficking between inner and outer segment of rods. We chose this gene due to its pathways correlations with congenital adrenal hyperplasia (CAH) ones, pathology that affects the proband of a Sicilian RP affected family (9 members). We carried out a case-control study regarding this family vs. a control group of 200 healthy Sicilian donors. The association of 5 single nucleotide polymorphisms (SNPs) of RP1 exon 4 and its 3' UTR with RP family phenotype was investigated.

Material & Methods: Genotyping was performed by PCR-RFLP (Polymerase Chain Reaction- Restriction Fragment Length Polymorphism) and direct sequencing.

Results & Conclusions: Data obtained highlight the fundamental role of a haplotype, inferred by all analyzed RP1 polymorphisms suggesting a possible genotype-phenotype correlation between these SNPs coexistence and query disease.

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