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Association of the Y402H Polymorphism in Complement Factor H Gene and Age-Related Macular Degeneration

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PURPOSE: Age-related macular degeneration (AMD), with its complex traits and multiple risk factors, is the leading cause of irreversible blindness in old people. A strong association between a coding variant, Y402H, in the complement factor H gene (CFH) and AMD has been identified in caucasian population. This study was conducted to investigate whether the same association between the Y402H polymorphism in CFH and AMD could be expected in Brazilian individuals, known constituted by a large mixed-race population.

Methods: Blood samples were collected from AMD patients (111) and controls counterparts (111) from the Ophthalmology Department/São Paulo Federal University. After plasma separation, genomic DNA was extracted, amplified by PCR technique and analyzed for the Y402H polymorphism, located in exon 9 of CFH and PCR-directed sequencing. The samples were analyzed by the department of Immunology/São Paulo University. Criteria for inclusion were age (more than 50 years) and the diagnosis of AMD without other ocular causes of neovascularization.

Results: 37.7% of the AMD patients were CC (H402) homozygous (14.3% control group); 38% of the AMD patients were CT heterozygous (43.9% control group) and 27.3% TT (T402) homozygous (41.8% control group).

Conclusion: The association of the Y402H polymorphism of the CFH gene to AMD susceptibility is more present in patients with AMD compared to the control group.