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Association between the rs820218 Variant within the SAP30BP Gene and Rotator Cuff Rupture in an Amazonian Population

Background

Rotator cuff disease is one of the leading causes of musculoskeletal pain and disability, and its etiology is most likely multifactorial but remains incompletely understood. Therefore, the objective of this research was to investigate the relationship of the single-nucleotide rs820218 polymorphism of the SAP30-binding protein (SAP30BP) gene with rotator cuff tears in the Amazonian population.

Methods

The case group consisted of patients who were operated on due to rotator cuff tears in a hospital in the Amazon region between 2010 and 2021, and the control group was composed of individuals who were selected after negative physical examinations for rotator cuff tears. Genomic DNA was obtained from saliva samples. For the genotyping and allelic discrimination of the selected single nucleotide polymorphism (rs820218) in the SAP30BP gene, real-time PCR was performed.

Results

The frequency of the A allele in the control group was four times as high as that in the case group (AA homozygotes); an association of the genetic variant rs820218 of the SAP30BP gene with rotator cuff tears was not established ($p = 0.28$ and 0.20), as the A allelic frequency is ordinarily low in the general population.

Conclusions

The presence of the A allele indicates protection against rotator cuff tears.

Keywords

rotator cuff tears; Amazonian population; SAP30BP; rs820218; single-nucleotide polymorphism; genetic susceptibility

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Biography

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