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Case Report

Association between an *ACAN* gene variable number tandem repeat polymorphism and lumbar disc herniation: a case control study

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ABSTRACT. We investigated the association between an aggrecan gene (*ACAN*) polymorphism and lumbar disc herniation (LDH). This was a case-control study with quinquennial age and gender groups. The study comprised 119 men and women aged between 20 and 60 from Goiânia (Brazil). Of these, 39 were allocated to the case group (Ca) and 80 to the control group (Ct). We gathered sociodemographic and clinical data, and peripheral blood samples. DNA was isolated for genotyping the *ACAN* variable number tandem repeat (VNTR) via conventional polymerase chain reaction (PCR). Data were statistically

analyzed using the chi-square test, multiple comparison analysis, the Student *t*-test, and odds ratios, with a level of significance set at 5% ($P \leq 0.05$). The groups were homogenous in terms of sociodemographic, anthropometric, and life style variables. The allele score for the *ACAN* VNTR was significantly lower in volunteers with LDH; the *A22* allele was significantly more prevalent in this same group; the Ca group presented greater frequency of short alleles *A13*-*A25*, whereas the Ct group presented a higher frequency of long alleles. However, this difference was not statistically significant. In both groups, the most common alleles were *A28*, *A27*, and *A29*, and the *A26/A26* genotype was significantly more common in the Ca group. The results showed an association between short alleles and LDH among the investigated adults (Ca), corroborating the hypothesis that aggrecan with shorter repeat lengths can lead to a reduction in the physiological proteoglycan function of intervertebral disc hydration and, consequently, increased individual susceptibility to LDH.

Key words: Lumbar disc herniation; Variable number tandem repeat; Intervertebral disc degeneration; Genetic polymorphism; Proteoglycans; *ACAN*