Association of COL1A1 Sp1 Gene Polymorphism with Degenerated Intervertebral Disc Prolapse from a Subset of Indian Population: A Case Control Study

Shailendra Deepak Anjankar MBBS; Subodh Raju

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Introduction

Degenerated Disc Disease (DDD) is a very common disorder responsible for increased morbidity in productive age group. Its etiology is multifactorial and genetic factors have been predominantly implicated. Disc prolapse results due to tear in annulus, which is a fibrous structure composed largely of type I Collagen. Functional polymorphism at the Sp1 site of the COL1A1 (Collagen I alpha 1) gene has shown positive association with DDD in Dutch and Greek populations.

Methods

50 clinically and radiologically proven patients of disc prolapse requiring surgery were included as cases and 50 healthy, age matched volunteers served as controls. After isolating DNA from their blood sample, genotyping for COL1A1 polymorphism (rs1800012) was performed and identified as GG, GT and TT.

Learning Objectives

By the conclusion of this session, participants should be able to:
1) Identify the gene which is responsible for defective development of Collagen 1 which can cause disc prolapse.
2) Understand the fact that multiple genes, gene - gene interaction, gene - environment interaction may be the reason for disc prolapse

Results

The mean age and body mass index in cases and controls were similar. 76% of the patients were males. Commonest site of disc degeneration was L4-5 (36%), followed by L5-S1 (34%). Homozygous – GG, heterozygous GT and homozygous TT genotypes were seen in 38(76%), 10 (20%) and 2(4%) cases respectively, controls had similar percentage of the genotypes as well. The alleles in cases and control group showed no significant difference (p=0.6744) and followed the Hardy-Weinberg Equilibrium in the study

Conclusions

The COL1A1 (rs1800012) is in Hardy Weinberg equilibrium in the present subset of Indian population. But taken as a single factor, it was not found to be associated with DDD in this preliminary study. Disc degeneration is multifactorial and also anticipated to be a result of multiple genes involvement and gene – gene interaction.

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