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ASSOCIATION OF LUMBAR DISC DEGENERATION WITH TYPE IX COLLAGEN POLYMORPHISMS IN CHINESE

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Introduction: Intervertebral disc degeneration is usually thought to be related to aging and spinal loading. This study is the first to investigate on collagen IX allelic variants in the Southern Chinese population, and their contributions as genetic predisposing factors to intervertebral disc degeneration. In particular, we examined the association of Gln326Trp in the a2-chain (Trp2) and Arg103Trp in the a3-chain (Trp3) of collagen IX.

Methodology: Lumbar DDD was defined by MRI on 804 Southern Chinese volunteers between 18-55 years, and presence of annular tears, disc and end-plate herniations were noted. These were correlated with the frequencies of Trp2 and Trp3 alleles. Additionally all three collagen IX genes were scanned for mutations.

Results: The Trp3 allele was absent, while the Trp2 allele was present in 20% of the population. Between 30-39 years of age, Trp2 was associated with a 4-fold increase in the risk of developing annular tears, and between 40-49 years, with a 2.4-fold increase in risk of developing DDD and end-plate herniations. Affected Trp2 individuals had a tendency towards more severe degeneration. No additional mutations were found in the collagen IX genes.

Discussion: This is the largest-scale population study to date using MRI to precisely define DDD. For the first time, we demonstrated that the Trp2 allele is a significant age-dependent risk factor. Collagen IX is an extracellular matrix molecule thought to be important in the structural integrity and function of the intervertebral disc. Alteration in its structure by the presence of the amino acid tryptophan within collagen IX is likely to make the intervertebral disc more susceptible to mechanical damage, which is manifested as age-related annular tears, end-plate herniatons and nucleus degeneration.