ISSN 0972-3757
International Journal of
HUMAN GENETICS


PRINT: ISSN 0972-3757 ONLINE: 2456-6360
Int J Hum Genet, 11(3): 193-198 (2011)
DOI: 10.31901/24566330.2011/11.03.07

# Distribution of COL9A2 and COL9A3 Gene Polymorphism in Male Chinese Singaporean – A Pilot Observational Study

**Edwin C.W. Lim[1,2]*, W.P. Wong[1], Gavin B.P. Ng[3], L.L. Chan[4], S.B. Tan[5], P.B. Tow[5] and Y. Zhao[6]**

*[1]*Department of Physiotherapy, Singapore General Hospital, Outram Road, Singapore 169608, Singapore*
*[2]Centre of Clinical Research Excellence: Spine, Division of Physiotherapy, School of Health and Rehabilitation Sciences, University of Queensland, St Lucia, QLD 4067, Australia*
*[3]Bioprocessing Technology Institute, Agency for Science Technology and Research, 20 Biopolis Way, #06-01 Centros, Singapore, 138668, Singapore*
*[4]Department of Diagnostic Radiology, Singapore General Hospital, Outram Road, Singapore*
*[5]Department of Orthopaedic Surgery, Singapore General Hospital, Outram Road, Singapore*
*[6]Department of Clinical Research, Singapore General Hospital, Outram Road, Singapore*

**KEYWORDS** COL9A2. COL9. Val2. Degenerative Disc Disease

**ABSTRACT** The association between allelic variants and lumbar disc degenerative disease (DDD) has been investigated in Europe and Northern Asia. However, this has not been investigated in Southeast Asia. This observational study aims to compare the distribution of COL9A2 and COL9A3 gene polymorphism among male Chinese Singaporeans with and without lumbar DDD. COL9A2 gene polymorphism was investigated in p326 (tryptophan 2, Trp2 and glutamine 2, Gln2, alleles) and p335 (valine 2 or Val2 allele). COL9A3 gene polymorphism was investigated in p17 (glycine 3 or Gly3 allele) and p103 (tryptophan 3 or Trp3 allele). The Val2 allele was significantly decreased in the group with lumbar DDD ($p < 0.05$). No significant difference in allelic distributions of Trp2, Gln2 and Gly3 was found. The Trp3 allele was absent from all the subjects. The presence of at least one Val2 allele appears to have a protective effect against DDD. However, these should be interpreted with caution, given the limitations. Further investigations are warranted in order to verify such genetic predisposition prior to the potential development of preventative or therapeutic strategies in the near future.