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Lack of association between SNP rs3914132 of the RELN gene and otosclerosis in India

S. Priyadarshi[1], K.C. Panda[2], A.K. Panda[2] and P.V. Ramchander[1]

[1]Institute of Life Sciences, Nalco Square, Chandrashekarpur, Bhubaneswar, India
[2]Capital Hospital, Unit VI, Bhubaneswar, India

Corresponding author: P.V. Ramchander
E-mail: pramchander@yahoo.com / ramchanderpv@ils.res.in

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ABSTRACT. Otosclerosis (MIM 166800) is primarily a metabolic bone disorder of the otic capsule, which leads to bony fixation of the stapedial footplate in the oval window; it is among the most common causes of acquired hearing loss. The etiology of this disease is largely unknown, although epidemiological studies suggest the involvement of both genetic and environmental factors. Recently, a reelin gene, SNP rs3914132, located in intron 2, was shown to be associated with otosclerosis in a European population. When we sequenced blood DNA samples of 85 individuals with otosclerosis and 85 controls, four SNPs of this gene: rs3914131 (P = 0.6463), rs3914132 (P = 0.1822), rs9641319 (P = 0.7371), and rs10227303 (P = 0.5669) were not significantly associated with this disease. In one familial case, a novel variant (C/T) at contig position 2923488 was found to be inherited by the proband and affected family members.

Key words: Otosclerosis; Reelin gene; SNP

