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MACF1 gene variant rs2296172 is associated with type 2 diabetes susceptibility in the Bania population group of Punjab - India

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Abstract

Microtubule Actin Cross linking Factor 1 (*MACF1*) gene variant rs2296172 has been associated with Type 2 Diabetes (T2D). However, this variant has never been evaluated as such in Indian populations. We replicated this variant in pooled population of Northwest India and specifically in an endogamous caste group, Bania of Punjab, India. We genotyped variant rs2296172 by Taqman allele discrimination assay in 651 T2D patients and in 568 healthy controls from Northwest India. The association of the SNP with T2D was evaluated by case - control association study design. The SNP rs2296172 of *MACF1* was found to be significantly associated with T2D with p value = 0.009 in Northwest Indian population but allelic distribution was observed to be deviated from Hardy-Weinberg equilibrium (HWE). Assuming population stratification the most plausible cause, we further evaluated the samples belonging to Bania caste group from Punjab, India. We observed significant association of this SNP with T2D with OR = 1.71 (1.03-2.83) at 95%CI, (p =0.03) and sample set following HWE. *MACF1* variant rs2296172 was found to be associated with T2D in endogamous ethnic population group (Bania) of Punjab, India. Deviation from Hardy-Weinberg equilibrium in the pooled population group from Northwest India, underlines that Indian population sub structure exists and may have implications in association studies. Thus, ideal case - control association study design in Indian populations is to evaluate endogamous population groups rather than the conventional practice of pooling samples based on geography or linguistic affinities only.

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