

# Characterization and Validation of SNPs in Exon 19 of CACNA2D1 Gene and its Relationship with Mastitis Traits in Indigenous Cattle

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**Abstract**—Calcium channel, voltage-dependent, alpha-2/ delta subunit 1 (CACNA2D1) gene plays an important role in excitation-contraction coupling. The CACNA2D1 gene may be one of the candidate genes related with some phenotypic traits due to its location in QTLs associated with SCS and mastitis. The dbSNP database which is a public repository for genetic variation within and across different species was investigated in this study (<http://www.ncbi.nlm.nih.gov/SNP/>). As of build 144 which was made available on June 2015, dbSNP has amassed over 600 million submissions representing many organisms, including *Homo sapiens*, *Equus caballus*, *Felis catus*, and many other organisms. All the polymorphisms reported in exon 19 of the CACNA2D1 gene were retrieved. A total of 120 Sahiwal cattle were selected to characterize exon 19 of CACNA2D1 gene to identify polymorphism and its association with mastitis susceptibility/resistance. A 249 bp PCR fragment of CACNA2D1 gene encompassing the exon 19 and partial intronic region was amplified and digested with *Hae III* to screen the SNP which was significantly associated with SCS. Genotype analysis using PCR-RFLP revealed a monomorphic banding pattern. Sequencing was also carried out to explore other SNPs which are deposited in dbSNP in the nucleotide sequence of a particular region. The result indicates highly conserved DNA sequence in Sahiwal cattle. The phylogenetic tree revealed that *Bos indicus* is closer to *Bos taurus* cattle, *Bos mutus* (Yak) and *Bison bison* (American buffalo) compared to other species. This study provides preliminary information that the targeted region of CACNA2D1 gene in indigenous cattle has no significant association with mastitis resistance which maybe a breed specific characteristic. Since present study has formulated the results based on a relatively small sample, further studies are required to study these SNPs in large samples to establish the role of these SNPs in CACNA2D1 gene in conferring resistance against mastitis.

**Keywords:** Sahiwal, Polymorphism, CACNA2D1, Exon , Mastitis