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Sequencing and characterisation of s2-casein gene (*CSN1S2*) in the Old World camels have proven genetic variations useful for the understanding of species diversification

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Abstract

The *CSN1S2* gene encodes s2-casein, the third most abundant protein in camel milk. Despite its importance in foals, human nutrition, and dairy processing, the *CSN1S2* gene in camels has received little attention. This study presents the first complete characterisation of the *CSN1S2* gene sequence in Old World camels (*Camelus bactrianus* and *Camelus dromedarius*). Additionally, the gene promoter, consisting of 752bp upstream exon 1, was analyzed. The entire gene comprises 17 exons, ranging in length from 24bp (exons 4, 8, 11 and 13) to 280bp (exon 17). Interesting was the identification of the exon 12 in both species. Promoter analysis revealed 24 putative binding sites in the Bactrian camel and 22 in dromedary camel. Most of these sites were typical elements associated with milk protein, such as C/EBP- , C/EBP- , Oct^{-1}, AP1. SNP discovery showed relatively high genetic diversity compared to other camel casein genes (*CSN1S1, CSN*$_2$*, CSN3*), with 34 polymorphic sites across the two species. Particularly noteworthy is the transition g.311G>A in the *CSN1S2* promoter, creating a new putative consensus binding site for a C/EBP- in the Bactrian camel. At exon level, two novel variants were found. One was detected in exon 6 of the bactrian camel (g.3639C>G), resulting in an amino acid replacement p.36Ile>Met. The second variant was found in non-coding exon 17 of dromedary *CSN1S2* (g.1511G>T). Although this mutation occurs in the 3’-UnTranslated Region, it represents the first example of exonic polymorphism in the *CSN1S2* for this species. This SNP also affects the binding sites of different microRNAs, including the seed sequence of the miRNA 4662a-3p, highlighting its role as a regulatory factor for *CSN1S2* gene. A PCR-RFLP was setup for genotyping a dromedary Tunisian population (n=157), and the minor allele frequency was found to be 0.27 for the G allele, indicating a potential yield improvement margin. Interspersed elements (INEs) analysis revealed 10 INEs covering 7.34 % and 8.14 % of the *CSN1S2* sequence in the Bactrian and dromedary camels, respectively. Furthermore, six elements (A, B, F, H, I and L) are shared among cattle and camels and are partially found in other ruminants, suggesting a common ancestral origin of these retrotransposons. Conversely, elements C, D, E and G are specific to camels.

Keywords: Bactrian camel, *CSN1S2*, s2-casein, dromedary camel

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