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DNA sequence analysis of estrogen receptor alpha and steroid 5-alpha-reductase type 2 genes in Holstein cattle affected by hypospadias and congenital anomalies of the female genital tract

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Abstract

This study aimed to detect DNA sequence variations of estrogen receptor alpha (ESR1), steroid 5-alpha-reductase type 2 (SRD5A2) genes involved in hormonal pathways and investigate their associations between hypospadias and congenital anomalies of the female genital tract. Total five blood samples were collected from cattle affected by one hypospadias, two female genital tract anomalies and two clinically normal cattle. 400 bp fragment in exon 1 of ESR1 gene and 567 bp fragment including exon 2 region of SRD5A2 genes were amplified by PCR and PCR products were directly sequenced in both directions. In this study, while first hypospadias case was reported for cattle raised in Turkey, it was also investigated very rare congenital anomalies of the female genital tract. As a result of sequence analysis an A/C transversion was detected at nucleotide position 344 of the obtained sequence from the amplification of ESR1 gene. Five different nucleotide substitutions A/G, G/A, A/C, G/T and G/A were detected at nucleotide position 74, 313, 365, 445 and 468 of the sequence obtained from the amplification of SRD5A2 gene, respectively. The A/C transversion on the exon 1 of the ESR1 gene was not associated with hypospadias and other congenital anomalies due to the fact that it also observed in one sample of the clinically normal cattle and previously reported as variation between individuals in different cattle breeds. As a result of sequence analysis of SRD5A2 gene two different single nucleotide substitutions were observed as specific to one of the female genital system anomalies although it was not detected any mutations associated with hypospadias. The effects of these nucleotide substitutions on female genital system should be investigated in more samples.

Keywords: Cattle, DNA, SRD5A2, ESR1, mutations

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