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Identification of mutations in the bovine *KIT* gene, a candidate for the Spotted locus in cattle

M. Tazzoli^{1,2}, J. E. Beever², L. Fontanesi¹, V. Russo¹

¹ Dipartimento di Protezione e Valorizzazione Agroalimentare. Università di Bologna, Italy

² Department of Animal Sciences. University of Illinois, USA

Corresponding author: Luca Fontanesi. Dipartimento di Protezione e Valorizzazione Agroalimentare, Sezione di Allevamenti Zootecnici. Via F.lli Rosselli 107, 42100 Reggio Emilia, Italy - Tel. +39 0522 290516 - Fax: +39 0522 290523 - Email: luca.fontanesi@unibo.it

ABSTRACT

In mammals, abnormal migration of melanoblasts from the neural crest during embryonic development may be the reason of the piebaldism phenotype that is a mixture of pigmented and unpigmented areas in the coat. Several cattle breeds, like for example Holstein, show the piebald spotted coat colour phenotype, that, according to cross-breeding studies, is due to a recessive allele (*s*), member of the allele series of the *Spotted* (*S*) locus. Dominant alleles at this locus act as suppressors of the spotted pattern and produce uniformly pigmented animals while others determine the colour-sided pattern known, for example, in the Hereford breed. The bovine *v-kit* Hardy-Zuckerman 4 feline sarcoma viral oncogene (*KIT*) gene was localized in the region of chromosome 6 where the *Spotted* locus was mapped. *KIT* plays a major role during the embryonic development in directing the migration of the melanoblasts from the neural crest. Mutations in this gene cause different coat colour patterns in mouse and human. In pigs, the *KIT* gene is responsible for variations at the *White* (*I*) locus. Thus, this gene can be considered a strong candidate for the *Spotted* locus in cattle. With the final aim to identify DNA markers associated to different coat colour pattern distributions that could eventually be used as markers for breed traceability of dairy and beef products, we sequenced the 21 bovine *KIT* exons in cattle breeds showing different coat colour patterns. PCR primers were designed on available bovine *KIT* genomic sequence in order to amplify all coding exons and parts of the intronic regions using DNA extracted from 8 animals of either Holstein, Hereford and Angus breeds. Sequencing revealed 75 mutations including 72 single nucleotide polymorphisms and 3 indels of one, three and five bp, respectively. These mutations were organized in a few haplotypes that were not observed in all studied breeds. Further investigations are in progress to evaluate the association of the identified polymorphic sites with coat colour patterns in beef and dairy cattle breeds.