

奶牛产奶性状与乳房炎相关基因CpG含量及分布特征的比较分析

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摘要 DNA甲基化是表观遗传学的重要组成部分, 基因启动子区及第一外显子区的CpG甲基化通常抑制该基因的表达, 而去甲基化则促进基因表达。已有的研究发现荷斯坦牛的乳房炎指标SCC(Somatic cell count)与产奶量呈较强负相关。文章分析并比较了这两类性状的相关基因的启动子区、第一外显子、下游2 000 bp序列中CpG含量及分布特征。结果表明, 乳房炎相关基因的启动子、第一外显子中CpG含量显著低于产奶性状相关基因, 而两类型状基因下游2 000 bp序列中CpG含量无显著性差异。另外, 文中提出了两个量化基因序列中CpG特征的指标, 一个是CpG平均距离, 用来衡量序列中的CpG分布; 另一个是条件概率 $p(G|C)$, 用以量化序列中二核苷酸CpG随碱基C出现的可能性, 并对两类基因的启动子和第一外显子区域的这两个指标做了统计检验。研究结果对产奶性状与乳房炎相关基因的DNA甲基化调控研究奠定了基础。

关键词: 产奶性状相关基因 乳房炎相关基因 CpG座位 DNA甲基化 条件概率

Abstract: DNA methylation is a major part of epigenetics. DNA methylation on the CpG sites in gene promoter and the first exon often represses gene expression, but demethylation activates gene expression. Previous research has shown that a negative correlation was found between mastitis index (somatic cell count, SCC) and milk production traits in Holsteins. The content and distribution of CpG dinucleotide sites in different regions of the candidate genes related to milk production traits and mastitis were studied in the present study. The regions contained promoter (2000 bp upstream of transcriptional start site), exon 1, and 2000 bp downstream of transcriptional end site. The CpG number of promoter and exon 1 in the mastitis-related genes was significantly less than that of the milk production-associated genes. However, the CpG number of 2000 bp downstream of the genes for the two traits was not significantly different. Two new index quantified CpG characterizations were proposed. One is the CpG distance, which can measure the distribution of CpG. The other is the conditional probability $p(G|C)$, which is used to quantify the probability of CpG in a nucleotide sequence along with C. The two indexes of promoter and exon 1 in the two types of genes and their statistic analysis were carried out. This study sets the basis for DNA methylation regulation of milk production traits- and mastitis-related genes.

Keywords: milk production traits-associated genes, mastitis-related genes, CpG site, DNA methylation, conditional probability

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