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Indel Polymorphisms in Some Genes In Cattle

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Abstract
*Insertion–Deletion (InDel) polymorphisms are structural genetic variations resulting from the insertion or deletion of one or more base pairs in the DNA sequence. In cattle (*Bos taurus *and* Bos indicus*), these polymorphisms are widely distributed across the genome and can occur in promoters, exons, introns, and 5′ or 3′ untranslated regions (UTRs), where they may influence gene expression, mRNA stability, protein structure, and ultimately phenotypic traits. Recent research has demonstrated that InDel polymorphisms play significant roles in economically important traits in cattle, including growth performance, body conformation, carcass composition, milk yield, reproductive traits, and disease resistance. Therefore, InDels represent a promising category of molecular markers for modern cattle breeding programs, supporting precision breeding and sustainable livestock production.*

Key Words*: indel polymorphism, cattle genetics*, *molecular markers, cattle*

Introduction

Insertion–Deletion (InDel) polymorphisms are structural genetic variations caused by the insertion or deletion of one or several base pairs within a DNA sequence (Mills et al., 2006). Typically observed in short sequences ranging from 1 to 100 base pairs, InDels can occur in promoters, exons, introns, or 5′/3′ untranslated regions (UTRs), where they may influence gene expression, protein structure, and metabolic pathways (Xu et al., 2018; Zhou et al., 2019; Jin et al., 2018). Studies in cattle (Bos taurus and Bos indicus) have shown that insertion–deletion (InDel) polymorphisms are associated with numerous economically important traits, including growth, carcass composition, milk production, body measurements, and reproductive performance. These findings indicate that InDel polymorphisms can serve as valuable molecular markers in cattle breeding programs, enabling marker-assisted selection (MAS) to improve growth, productivity, and carcass traits. Additionally, differences in allele frequencies among breeds and populations provide insights into geographic adaptation and the genetic architecture of cattle.

Indel Polymorphism in cattle

Lan et al. (2023) investigated a novel indel in the bovine SEPT7 gene and its association with ovarian length. In an analysis of 408 samples, three loci were genotyped, and two loci (L1 and L5) were found to be polymorphic, with minimum allele frequencies of 0.417 and 0.094, respectively. The L1 indel of SEPT7 was significantly associated with ovarian length ($p < 0.05$), with individuals carrying II and ID genotypes exhibiting longer ovaries than those with the DD genotype. This study suggests that SEPT7 can serve as a molecular marker for animal fertility and supports the application of genetic techniques in cattle breeding.

Yang et al. (2022) investigated the association of two insertion/deletion (indel) variants in the VISFATIN gene with growth traits in Chinese cattle breeds. In the study, a 35-bp insertion in intron 4 and a 6-bp deletion in intron 5 were identified. The indel in intron 4 showed moderate polymorphism across all samples, while the indel in intron 5 exhibited low polymorphism in some breeds. Association analysis revealed that the intron 4 indel was significantly associated with chest girth, rump length, and body weight in Ji'an (JA) cattle, whereas the intron 5 indel affected rump length in Jinnan (JN) cattle. This study is the first to demonstrate that indels in the VISFATIN gene are associated with growth traits in these two Chinese indigenous cattle breeds and suggests that the gene could serve as a molecular marker for cattle breeding.

Liu et al. (2022) investigated two insertion/deletion polymorphisms (Indel-3 and Indel-4) in the bovine FoxO1 gene and their association with growth traits in three Chinese cattle breeds. Both indels are located in the second intron, and four haplotypes (H1: D3D4, H2: I3D4, H3: D3I4, H4: I3I4) were identified, with the D allele, DD genotype, and D3D4 haplotype having the highest frequency. In Nanyang cattle, individuals with the DI genotype (D3I3, D4I4, or H1H4/H2H3) showed significantly better phenotypic traits than other genotypes, indicating hybrid vigor. These results suggest that these two indels in the FoxO1 gene can be used as genetic markers for early selective breeding and marker-assisted selection (MAS) in cattle.

Guo et al. (2022) identified a novel 8-bp indel and a copy number variation (CNV) in the SPAG17 gene and investigated their association with growth traits in cattle. The indel was analyzed in 1,520 individuals from eight breeds, and the CNV was studied in 355 individuals. In Xianan cattle, individuals with the ID genotype showed superior performance in body height ($p = 0.038$) and body slanting length ($p = 0.041$) compared to other genotypes. For the CNV, different copy numbers were significantly associated with body height in Qinchuan cattle ($p = 0.045$) and body weight in Xianan cattle ($p = 0.036$). Additionally, a significant difference between the 8-bp indel and copy number loss was observed in Xianan cattle ($p < 0.01$). These findings suggest that variations in the SPAG17 gene can serve as effective DNA molecular markers for beef cattle breeding.

Wang et al. (2022) investigated two novel insertion/deletion (InDel) variants in the bovine PSAP gene and their association with growth traits at different developmental stages. The study, conducted across eight cattle breeds, identified the P5 and P8 InDels. Analyses revealed that the dominant genotype for daily gain and body weight varied across developmental stages in both NY and JX cattle. PSAP gene expression analysis showed high expression during the middle stage of adipocyte differentiation, suggesting a role in fat development. The study demonstrates that InDels can affect phenotypes at different developmental stages depending on the host gene's expression pattern and tissue-specific function. These findings provide a new approach for molecular marker studies in cattle breeding and genetics.

Ju et al. (2021) investigated eight polymorphic insertion/deletion (InDel) variants in the FHIT gene and their association with milk traits in Xinjiang Brown cattle (XJBC). Analysis of 388 healthy and unrelated individuals revealed that all eight InDel loci were polymorphic, with polymorphism information content ranging from 0.061 to 0.375. Correlation analysis showed that all eight InDels were significantly associated with six different milk traits ($p < 0.05$). Specifically, P2-23bp was associated with 305-day milk yield in the sixth parity ($p = 0.005$), P3-24bp with milk fat yield in the third parity ($p = 0.009$), P5-21bp with somatic cell score in the first parity ($p = 0.001$) and milk protein percentage in the sixth parity ($p = 0.002$), and P7-26bp with somatic cell score in the sixth parity ($p = 0.003$). These findings suggest that FHIT InDel genotypes can be used in marker-assisted selection (MAS) to improve milk production traits in cattle.

Jakaria et al. (2021) investigated single-nucleotide polymorphisms (SNPs) and an 11-bp indel in the myostatin (MSTN) gene and their association with the double-muscled phenotype in Belgian Blue (BB), Peranakan Ongole (PO), and BB × PO crossbred cattle. Analysis of 86 blood samples revealed four SNPs in the coding region of MSTN in PO and crossbred cattle; however, these SNPs could not distinguish between normal and double-muscled phenotypes. Additionally, an 11-bp deletion in exon 3 of MSTN was identified in BB cattle. Using PCR-RFLP with the NmuCI (Tsp45I) restriction enzyme, the genotypes +/+, +/del.11, and del.11/del.11 were successfully observed. This study indicates that the 11-bp indel in MSTN can be used to identify the double-muscled phenotype in cattle.

Peng et al. (2020) detected insertion/deletion (InDel) variations in seven candidate genes and investigated their association with phenotypic traits in Xinjiang Brown, Red Steppe, and Yunling cattle. The analysis revealed that the genotypic and allelic distributions of these seven genes differed significantly among the three breeds ($p < 0.05$ or $p < 0.01$). Furthermore, InDel variations in SREBP1c and PAX7 were significantly associated with eight phenotypic traits in Xinjiang Brown cattle ($p < 0.05$ or $p < 0.01$), suggesting that these InDels can serve as useful DNA markers for economically important traits.

Zhou et al. (2019) investigated the distribution of a 19-bp insertion/deletion (indel) in the PLAG1 gene and its association with growth traits in Chinese cattle breeds. The indel, located in intron 1, was found to be polymorphic in Qinchuan, Pinan, Xianan, and Jiaxian Red cattle and correlated with body height. Analysis of 37 cattle breeds (1,354 individuals) revealed three genotypes and two alleles (W, 366 bp; D, 347 bp). From northern to southern breeds, the frequency of the W allele gradually decreased while the D allele increased, consistent with regional differences in cattle height. In Yunling cattle, the indel was significantly associated with body height, cross height, and chest circumference ($p < 0.05$). This study provides evidence that the 19-bp indel of PLAG1 is an effective molecular marker for cattle breeding.

Zhou et al. (2019) investigated the distribution of a 19-bp insertion/deletion (indel) in intron 1 of the PLAG1 gene and its association with growth traits in Chinese cattle breeds. Analysis of 1,354 individuals from 37 breeds identified three genotypes and two alleles (W, 366 bp; D, 347 bp). From northern to southern breeds, the frequency of the W allele gradually decreased while the D allele increased, consistent with regional differences in cattle height. In Yunling cattle, the 19-bp indel was significantly associated with body height, cross height, and chest circumference ($p < 0.05$). These results provide new evidence that the PLAG1 19-bp indel is an effective trait marker and a potential candidate molecular marker for cattle breeding.

Wu et al. (2019) investigated insertions and deletions (indels) in the MSRB3 gene and their association with growth traits in four Chinese indigenous cattle breeds (Luxi, Qinchuan, Nanyang, and Jiaxian Red). Four indels were identified, and three of them were significantly associated with growth traits ($P < 0.05$). Specifically, the DD genotype of P1 was associated with body length in Nanyang cattle; the II and/or DD genotypes of P6 were associated with enhanced growth traits in Qinchuan cattle; and the II genotype of P7 was associated with hucklebone width in Luxi cattle. These findings indicate that polymorphisms in the MSRB3 gene are significantly associated with growth traits and could serve as candidate loci for marker-assisted selection (MAS) in cattle breeding.

Zhang et al. (2019) characterized the genomes of three Chinese native cattle breeds, identifying approximately 34.3 million SNPs and 3.8 million InDels through whole-genome resequencing. On average, 10.4 million SNPs were shared among the three breeds, while 3.0 million, 4.9 million, and 5.8 million SNPs were specific to LQ, WN, and WS breeds, respectively. Gene ontology (GO) analysis revealed four immune response-related GO terms overrepresented across all samples, with two immune signaling pathways significantly overrepresented in WS cattle. Immune-related genes such as PGLYRP2, ROMO1, FYB2, CD46, and TSC1 were identified in all three breeds. This study provides insights into the genetic basis of Chinese indicine adaptation to tropical and subtropical environments and offers a valuable resource for further investigation of the genetic characteristics of these breeds.

Jiang et al. (2019) investigated indel variations in 11 candidate genes and their associations with milk composition traits in a Chinese Holstein population. In 769 cows, the indel in FCGR2B was significantly associated with milk yield, protein yield, and protein percentage ($P = 0.0041–0.0297$). Several indels in CENPE and one in MAP3K1 were significantly related to milk yield, fat yield, and protein yield; polymorphisms in RETSAT were associated with milk yield, fat yield, protein yield, and protein percentage; the ACSBG2 indel affected fat yield and protein percentage; one indel in TBC1D1 was associated with fat

and protein percentage; indels in NLK were related to protein yield and protein percentage; and a variant in UGDH was associated with milk yield. Exonic indels in FCGR2B and CENPE were predicted to alter mRNA and protein secondary structures, potentially causing protein dysfunction. This study provides the first evidence linking eight functional genes with milk yield and composition traits in dairy cattle.
Xu et al. (2018) identified a 19-bp indel in the PLAG1 gene and investigated its association with growth traits in Chinese cattle. Using Pool-Seq and agarose gel electrophoresis, three genotypes and two alleles (W, 142 bp; D, 123 bp) were detected, with WW genotype and W allele being predominant. Association analysis showed that the 19-bp indel was significantly related to hip width and rump length in Pinan cattle, heart girth and cannon bone circumference in Xianan cattle, and body height, chest depth, hip width, rump length, sacrum height, and shoulder width in Jiaxian cattle ($P < 0.05$ or $P < 0.01$). These findings suggest that the PLAG1 indel can serve as a candidate molecular marker for cattle breeding.

Xu et al. (2018) investigated a 19-bp insertion/deletion (indel) in the PLAG1 gene and its association with growth traits in 566 cattle. Sequencing of pooled DNA and agarose gel electrophoresis identified three genotypes and two alleles: 142 bp (W) and 123 bp (D), with WW genotype and W allele being predominant. The 19-bp indel was significantly associated with growth traits across breeds, including hip width and rump length in Pinan cattle ($P < 0.05$), heart girth and cannon bone circumference in Xianan cattle ($P < 0.01$ or $P < 0.05$), and heart girth, hip width, hucklebone width, rump length, height at sacrum, and chest depth in Jiaxian cattle ($P < 0.05$). These results suggest that the PLAG1 indel can serve as a candidate molecular marker for cattle breeding.

Jin et al. (2018) investigated novel insertion/deletion (indel) variants in the SIRT1, SIRT2, and SIRT3 genes and their association with body measurement traits in 955 cattle from five breeds. The study identified a 12-bp indel in SIRT1, a 7-bp indel in SIRT2, and a 26-bp indel in SIRT3 for the first time. Association analysis showed that indels in SIRT1 and SIRT2 were significantly associated with body measurement traits, including body weight, chest circumference, hip cross height, hip width, and body height ($P < 0.05$ or $P < 0.01$). These findings suggest that the novel SIRT1 and SIRT2 indels could serve as potential molecular markers for growth traits in cattle breeding and selection programs.

Conclusion

Collectively, the reviewed studies demonstrate that insertion/deletion (indel) polymorphisms represent an important source of genetic variation influencing economically valuable traits in cattle. Indels identified in key functional genes such as SEPT7, VISFATIN, FoxO1, SPAG17, PSAP, FHIT, PLAG1, MSRB3, SIRT1/2/3, and MSTN were significantly associated with a wide range of phenotypes, including reproductive performance, growth and body measurements, muscle development, milk yield and composition, and adaptive traits. Many of these loci showed breed-specific or developmental stage specific effects, highlighting the complex genetic architecture underlying cattle productivity. Furthermore, several indels, particularly those in PLAG1, FoxO1, SPAG17, and MSTN, demonstrated strong and consistent associations across studies, emphasizing their potential utility as reliable molecular markers. These findings reinforce the value of indel polymorphisms as cost-effective and informative genetic markers that can enhance the accuracy and efficiency of marker-assisted selection (MAS) in cattle breeding programs. Future research integrating genomic, transcriptomic, and functional analyses will further clarify the biological mechanisms through which indels influence phenotypic variation and will support their broader application in precision livestock breeding.

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