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In the search of the polled locus in a *Bos taurus* x *Bos indicus* population

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Abstract:

The polled phenotype is an autosomal dominant trait in cattle. There are many advantages of producing genetically polled cattle. For instance, they are safer to work with, and there are fewer events of injuries among animals with consequent risk of decrease in milk production as well as meat and leather quality. The objective in this study was to identify regions and SNPs associated with the polled/horned phenotype in a composite *Bos taurus* x *Bos indicus* breed (Canchim). A sample of 391 Canchim animals with phenotype information available (84 polled and 307 horned) was genotyped using the Illumina BovineHD BeadChip. The quality control filters (QC) included call rate (< 0.90) for samples and SNPs, minor allele frequency (MAF < 0.01), and Hardy-Weinberg disequilibrium (< 0.001). After applying the QC filters, 387 animals (83 polled and 304 horned) and 699,620 SNPs remained in the study. The genome-wide association analysis was conducted considering a case (polled) x control (horned) study using plink software, and the Bonferroni correction was used to adjust p-values for multiple tests. The adjusted p-values were then used to create a Manhattan plot for visualization and identification of significant peaks. According to the Manhattan plot a very distinct peak can be seen at the proximal end of chromosome (BTA) 1 (between 1.0 and 8.6Mb), while other peaks can be found on BTA 8 (76.86 to 86.93Mb), BTA 17 (34.19 to 40.30Mb), BTA 27 (1.57 to 2.25Mb), and a more disperse peak on BTA 10 (between 30.95 and 83.58Mb). Reducing the peak on BTA 1 to an interval between 1.38 to 2.43Mb, there were a total of 10 SNPs associated with gene coding regions; one was on the IFNGR2 gene, 7 on the C1H21orf63 gene (also known as EVA1C gene), and two on the URB1 gene, and one SNP was in the inter-genic region between the C1H21orf63 and URB1 genes. A polymorphism within the interferon gamma receptor 2 gene (IFNGR2) has been recently described as a candidate for polledness in Holstein cattle. The peak on BTA 8 is formed by 20 SNPs, of which three are within genes (DCAF12, WNK2, PHF2), and all others are in inter-genic regions, while the peak on B TA 10 was formed by 25 SNPs, with 11 being within genes (ATPBD4, BSC1, MYEF2, SLC24A5, SLC8A3), and the rest being in inter-genic regions. The peaks on BTA 17 and BTA 27 were formed by 27 and 8 SNPs, respectively, but none of them are within genes. So far we have not found any biological evidence for the peaks on BTA 8, 10, 17 and 27, which could indicate they are spurious signals due to population structure. The area around the peak on BTA 1 matches previous results for identifying the polled locus in cattle, and more refined analysis should be carried out to identify the causative polymorphism. However, all other peaks were not expected and do not seem to have a direct effect on the phenotype, requiring further analysis, including to account for population structure.

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