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Genomic Variant Hotspots in Nelore Cattle Revealed By Missing Genotypes

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SNP genotyping with High Density (HD) chips presents a relative low cost when compared to genotyping by re-sequencing using Next Generation Sequencing technologies, which is a significant advantage when a large number of individuals is under study. Currently, there are commercial HD chips for several economically important species, for a very reasonable price per sample. The Illumina BovineHD BeadChip includes more than 777.000 SNPs designed from sequence data derived from two subspecies, *Bos taurus* and *Bos indicus*, and several composite breeds, selected for beef and/or dairy production. Although those breeds share most part of their genomic sequence, some SNP probes designed based on one breed may not be fully compatible to others, yielding lower call rates when compared to most loci on the panel. Those genotypes are usually removed from datasets as low quality data during quality control procedures. Among this discarded data, some SNP probes do not produce any genotype due the presence of genomic variations within their flanking regions. In order to investigate whether or not the "missing" genotypes actually reveal genomic variant hotspots, we examined BovineHD missing genotypes from a total of 1,709 Nelore DNA samples and used sequence data from eight of them to identify putative polymorphisms flanking the assayed SNPs. We only used information from markers "missing" in all samples, and located in exon, and manually curated those related to non-synonymous mutations. Functional annotation of these SNP-containing genes revealed some interesting categories, including biological regulation, response to stimuli, signaling, immune system process, growth and reproduction, which are involved in phenotypic differences that

have already been described between taurine and zebuine cattle.
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