



Distribution of functional variants within Runs of Homozygosity in four Italian cattle breeds

Journal:	<i>Italian Journal of Animal Science</i>
Manuscript ID	Draft
Manuscript Type:	Abstract Submission
Date Submitted by the Author:	n/a
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Abstract:	<p>Several methods have been used to identify genomic regions subjected to selective sweeps. An alternative method can make use of runs of homozygosity (ROH), defined as stretches of homozygous regions in a genome. The aim of the present study was to detect selective sweeps using ROH in four cattle breeds. Individuals of Cinisara (71), Modicana (72),</p>

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	<p>Reggiana (168) and Italian Holstein (96) were genotyped with the Illumina Bovine50SNP v2 BeadChip. To identify genomic regions that were most commonly associated with ROH within each breed, the percentage of occurrences of a single nucleotide polymorphism (SNP) in ROH was calculated across animals. The genomic regions most commonly associated with ROH were identified by selecting the top 1% of the SNPs most commonly observed in ROH in each breed. This approach resulted in the identification of 11 genomic regions in the Cinisara and Italian Holstein, and 8 in Modicana and Reggiana showing increased frequency of ROH. Generally, ROH patterns differed between breeds. There were two common genomic regions between breed pairs, and in particular one in BTA6 between Modicana and Reggiana and one in BTA10 between Cinisara and Italian Holstein. A highly homozygous region (> 45% of individuals with ROH) was found only in Modicana breed in BTA6 (6:37,019,972-39,069,719) within a QTL affecting milk fat and protein concentration. In these genomic regions we identified from 126 to 347 genes for each breed. According to Panther and KEGG database, a majority of the genes was involved in multiple signaling and signal transduction pathways in a wide variety of cellular and biochemical processes. Several of these genes were also comprised in a list of genes related to phenotypes for which cattle breeds have been subjected to strong positive selection. For most genes associated with ROH islands, a biological link to traits such as milk yield and composition, reproduction, immune response, coat colour, genetic disorders and resistance/susceptibility to infectious and diseases, which are known to be under selection, can be hypothesized. These results showed that selective sweeps detected with ROH approach are shared among breeds and that scanning the genome for ROH might be an alternative or complementary strategy to detect selective sweep related with important economically traits.</p>
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Distribution of functional variants within Runs of Homozygosity in four Italian cattle breeds

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Several methods have been used to identify genomic regions subjected to selective sweeps. An alternative method can make use of runs of homozygosity (ROH), defined as stretches of homozygous regions in a genome. The aim of the present study was to detect selective sweeps using ROH in four cattle breeds. Individuals of Cinisara (71), Modicana (72), Reggiana (168) and Italian Holstein (96) were genotyped with the Illumina Bovine50SNP v2 BeadChip. To identify genomic regions that were most commonly associated with ROH within each breed, the percentage of occurrences of a single nucleotide polymorphism (SNP) in ROH was calculated across animals. The genomic regions most commonly associated with ROH were identified by selecting the top 1% of the SNPs most commonly observed in ROH in each breed. This approach resulted in the identification of 11 genomic regions in the Cinisara and Italian Holstein, and 8 in Modicana and Reggiana showing increased frequency of ROH. Generally, ROH patterns differed between breeds. There were two common genomic regions between breed pairs, and in particular one in BTA6 between Modicana and Reggiana and one in BTA10 between Cinisara and Italian Holstein. A highly homozygous region (> 45% of individuals with ROH) was found only in Modicana breed in BTA6 (6:37,019,972-39,069,719) within a QTL affecting milk fat and protein concentration. In these genomic regions we identified from 126 to 347 genes for each breed. According to Panther and KEGG database, a majority of the genes was involved in multiple signaling and signal transduction pathways in a wide variety of cellular and biochemical processes. Several of these genes were also comprised in a list of genes related to phenotypes for which cattle breeds have been subjected to strong positive selection. For most genes associated with ROH islands, a biological link to traits such as milk yield and composition, reproduction, immune response, coat colour, genetic disorders and resistance/susceptibility to infectious and diseases, which are known to be under selection, can be hypothesized. These results showed that selective sweeps detected with ROH approach are shared among breeds and that scanning the genome for ROH might be an alternative or complementary strategy to detect selective sweep related with important economically traits.