Haplotype variation at the POLLED locus in the South African Bonsmara cattle breed

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Summary

The Bonsmara breed plays a major role in the South African red meat industry and contributes 15.3% to the market share. An increased awareness of animal welfare necessitates the breeding of genetically polled animals, especially since more than 70% of South African cattle are rounded off in commercial feedlots. Over the past two decades, commercial beef producers and feedlots realized the advantages of polled cattle. The aim of this study was to characterize the POLLED locus in the South African Bonsmara beef cattle breed, using PCR-based genotyping data for the Celtic mutation, as well as 150k SNP genotypic data. Here we confirmed that the Celtic allele is responsible for the polled phenotype in Bonsmara cattle and demonstrated that this mutation was introgressed from a very limited number of founders. Our results will enable the development of a haplotype test to manage the genetic variability around the polled locus.

Keywords: beef cattle, haplotypic frequency, polledness

Introduction

In South Africa, the red meat industry plays a major role in livestock production, with more than 70% of all beef cattle slaughtered in the formal sector originating from commercial feedlots (Scholtz et al., 2008). The Bonsmara contributes a large percentage to the red meat industry, having a 15.3% market share. At present the Bonsmara is the most numerous beef cattle breed in South Africa, contributing approximately 81000 registered cows to the national beef population (Van Marle-Köster et al., 2013). In Bonsmara herds the polled trait was either inherited from the Shorthorn/Hereford ancestors from which the breed was developed or from more recent crosses through the upgrading of Red Poll and Red Angus cows to Bonsmara stud status. It is also possible that some “polled” strains originated from spontaneous mutations, as it has been described in the Charolais breed (Capitan et al., 2011; Allais-Bonnet et al., 2013).

Since the domestication of cattle, selection practices were focused on adapted animals and aesthetic traits, such as coat colour, body composition and the presence of horns, which were related to male fertility. However, over the last few decades the focus has shifted towards sustainable animal production, with an increased awareness of animal welfare. Horns in cattle are a major cause of bruising, hide and carcass damage, as well as other injuries, but the practice of dehorning cattle has serious welfare implications. Breeding genetically polled animals would provide a long term and welfare friendly solution to dehorning. Commercial
beef producers and feedlots in South Africa are well aware of the advantages of polled cattle, with Bonsmara stud breeders indicating a preference for polled animals.

The POLLED locus has been mapped to the centromeric region of BTA1 (e.g. Georges et al., 1993; Medugorac et al., 2012; Seichter et al., 2012) and three distinct causative variants have been identified at the POLLED locus namely the Celtic, Friesian and Mongolian alleles (Medugorac et al., 2012; Allais-Bonnet et al., 2013, Medugorac et al., 2017). The Celtic allele (PC) is responsible for polledness in most of the European Bos taurus breeds. The Friesian allele (PF) is responsible for the polled phenotype predominantly in the Holstein Friesian breed (and a few other breeds that have been introgressed with Holstein genetics). Finally, the Mongolian allele (PM) has been described only in East Asian Bos taurus and Bos grunniens breeds. None of these mutations are located in known coding or regulatory regions, thus adding to the complexity of the molecular basis of polledness.

The aim of this study was to characterize the genetic variability around the POLLED locus in the South African Bonsmara beef cattle breed, using PCR-based genotyping data for the Celtic mutation, as well as 150k SNP genotypic data.

Material and methods

Hair samples from 92 South African Bonsmara animals, with a phenotypic record for horn status (horned, polled, or scurs: small abnormal horny growth), were received from two different breeders, with consent from the SA Bonsmara breed society. Genomic DNA was extracted with a Zymogen Tissue kit in the Department of Animal and Wildlife Sciences (University of Pretoria). Animals were genotyped for the Celtic mutation (PC) by PCR and agarose-ethidium bromide electrophoresis using primers CELT-Fw: GAAGTGTGGCCGGTAGAAAA and CELT-Rv: ATCAAGGACACCTCCCCACAC, as described in Allais-Bonnet et al. (2013). This screening allows the identification of carriers of the Celtic mutation, as well as the identification of homozygous and heterozygous polled animals.

Sixteen homozygous polled animals (PC/PC) were genotyped with the 150k GGP bovine SNP chip at the Agricultural Research Council – Biotechnology Platform. Individual and sample based quality controls were performed using Plink (Purcell et al., 2007). These individuals had a call rate > 85%. SNP markers were excluded based on the following criteria: call rate below 90%, minor allele frequency (MAF) below 2% and Mendelian error rate of more than 10%. After quality control, 107 395 SNPs remained in the dataset.

A 1.2 Mb interval on BTA1 centered around the localisation of the PC allele (1.6 Mb ± 600 kb on the UMD3.1 assembly) was extracted to study haplotype diversity around this mutation. The SNP positions of this 1.2 Mb region were updated according to the UMD3.1 assembly using SNPCovert (Nicolazzi et al., 2016). Haplotype analysis was performed with Haplovieview (Barret et al., 2005).

Results and discussion

A total of 92 Bonsmara animals were genotyped for the Celtic mutation at the POLLED locus. It was possible to distinguish between homozygous and heterozygous polled animals on a genotypic level, and a total of 16 homozygous polled (PC/PC), 60 Heterozygous polled (PC/p) and 16 horned (pp) animals were detected (Table 1). All of the 60 polled and 16 scurred animals carried at least one PC allele, suggesting that the Celtic allele is the most frequent or
even unique polled allele segregating in this population. Interestingly, we identified 16 homozygous \( \text{P}_C/\text{P}_C \) animals which were all “clean” polled, whereas 27% (16/60) of the \( \text{P}_C/\text{p} \) animals were scurred. This result suggests an additive effect of the \( \text{P}_C \) allele preventing the development of scabs or small horny growth in homozygous individuals.

Table 1. The total number of animals for each phenotype and respective genotypes for the Celtic locus

<table>
<thead>
<tr>
<th>Phenotype</th>
<th>Number of animals</th>
<th>Celtic (( \text{P}_C )) genotype</th>
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<tbody>
<tr>
<td>Polled</td>
<td>60</td>
<td>PP 16, Pp 44, pp 0</td>
</tr>
<tr>
<td>Scurs</td>
<td>16</td>
<td>PP 0, Pp 16, pp 0</td>
</tr>
<tr>
<td>Horned</td>
<td>16</td>
<td>PP 0, Pp 0, pp 16</td>
</tr>
<tr>
<td>Total</td>
<td>92</td>
<td>PP 16, Pp 60, pp 16</td>
</tr>
</tbody>
</table>

A subsequent analysis of the SNP Chip genotyping data from the 16 \( \text{P}_C/\text{P}_C \) Bonsmara animals revealed a reduced genetic diversity around the Celtic allele, with only two haploblocks observed in these animals in the window \( 1.0-2.2 \) Mb (56 SNP markers, Table 2).

Table 2. Haplotype frequencies for the two haploblocks detected among 16 \( \text{P}_C/\text{P}_C \) animals in the window \( 1.0-2.2 \) Mb.

<table>
<thead>
<tr>
<th>Haploblock</th>
<th>Map position (bp)</th>
<th>Haplotype frequencies</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td></td>
<td>CGCAAAG 0.969</td>
</tr>
<tr>
<td>Haploblock 1</td>
<td>1 264 369 – 1 336 351</td>
<td>AAACGGA 0.031</td>
</tr>
<tr>
<td></td>
<td></td>
<td>AGAGAG 0.625</td>
</tr>
<tr>
<td></td>
<td></td>
<td>GAGAAA 0.281</td>
</tr>
<tr>
<td></td>
<td></td>
<td>AAAGGG 0.094</td>
</tr>
</tbody>
</table>

One of these haploblocks of six SNPs encompassed the Celtic mutation and presented only three distinct alleles with major differences in terms of frequencies. This result suggests that the Celtic allele was introgressed in the Bonsmara breed from a very limited number of founders. A low haplotype diversity combined with an intense selection on the polled phenotype in the Bonsmara breed can result in the selection of deleterious alleles linked with the Celtic mutation through a hitchhiking mechanism. Therefore, it is of primary importance to maintain some genetic variability around the Celtic allele. Our results will enable the development of a haplotype test to manage this problem.

Conclusion

In conclusion we demonstrate that the Celtic allele is responsible for the polled phenotype in SA Bonsmara cattle and that this mutation was introgressed from a very limited number of founders. Our results will enable the development of a haplotype test to manage the genetic variability around the polled locus.

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List of References


