SNP discovery in the ovine ABCG2 gene

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INTRODUCTION

Breast Cancer Resistance Protein (ABCG2)

- A member of the ATP-binding cassette (ABC) transporters superfamily, involved in the transport of xenobiotics from cells
- Its expression is strongly induced in the mammary gland during lactation.
- A SNP in the bovine *ABCG2* gen (exon 14) → suggested as a QTN influencing milk production traits.

OBJETIVE

Search for variability in the ovine *ABCG2* gene in order to assess the possible influence of this gene on milk production traits in sheep.

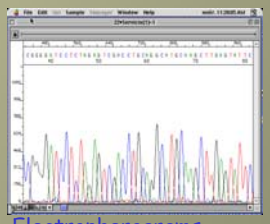
MATERIAL & METHODS

12 unrelated Spanish Churra rams

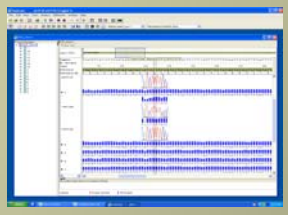
Genomic DNA was extracted from blood samples

Thirteen primer pairs were designed to amplify 5'-UTR, exons 1 to 11 and 3' downstream of *ABCG2*

Cycle Sequencing
ABI Prism 3130 Genetic Analyzer



Electropherograms



Sequences assembly and polymorphisms detection with Applied- Biosystems SeqScape® software v2.5

RESULTS

Table 1: Minor allele frequency (MAF) and location of the *ABCG2* gene SNPs identified in this work

SNP_ID	MAF	REGION	TYPE OF CHANGE
LongTranscriptT>C	0.091	5'-upstream	Transition
c.356_-195C>A	0.125	Intron2	Tranversion
c.356_-129T>G	0.25	Intron2	Transversion
c.356_-75A>T	0.375	Intron2	Transversion
c.416_-72T>G	0.25	Intron2	Transversion
c.356_-32G>A	0.25	Intron2	Transition
c.356_+85T>C	0.188	Intron3	Transition
c.416_+108G>T	0.437	Intron3	Transversion
c.416_+128G>A	0.437	Intron3	Transition
c.417_-29G>A	0.2	Intron3	Transition
c.417_-23C>A	0.15	Intron3	Transversion
c.417_-8T>A	0.2	Intron3	Transversion
c.711G>A	0.417	**Exon6**	Transition
c.842_+41C>T	0.333	Intron6	Transition
c.936A>G	0.333	**Exon7**	Transition
c.945A>C	0.333	**Exon7**	Transversion
c.954T>C	0.333	**Exon7**	Transition
c.978T>G	0.389	**Exon7**	Transversion
c.986G>A	0.444	**Exon7**	Transition
c.989A>C	0.375	**Exon7**	Transversion
c.993T>A	0.444	**Exon7**	Transversion
c.994_+50A>C	0.222	Intron7	Transversion
c.1128C>T	0.5	**Exon9**	Transition
c.1434_-98A>G	0.333	Intron10	Transition
c.1434_-84A>G	0.333	Intron10	Transition
c.1434_-81C>A	0.333	Intron10	Transversion
c.1434_-9A>G	0.333	Intron10	Transition
c.1523_+112G>C	0.333	Intron11	Transversion
c.1523_+124G>A	0.333	Intron11	Transition
c.2186_+138G>T	0.125	3'-downstream	Transversion
c.2186_+137A>G	0.125	3'-downstream	Transition
c.2186_+14T>G	0.208	3'-downstream	Transversion

- We sequenced ~ 5.4 Kb of the ovine *ABCG2* gene including the 5'-UTR, exons 1 to 11 and the 3'-UTR
- A total of 32 nucleotide polymorphisms (SNPs) were detected.
- Positions indicated in Table 1 refer to the bovine *ABCG2* gene reference sequence (ENSBTAG00000017704).
- Nine of SNPs identified were located in the coding region (exons 6, 7 and 9). None of them causes an aminoacid change.
- Exon 7 was the most variable region with 7 SNPs in only 152 pb.
- Minor allele frequency (MAF) range: 0.091 - 0.5, for the SNPs in the 5´upstream and Exon 9, respectively.

FUTURE WORK

Future association analysis will assess the relation between some of the allelic variants reported herein and milk production traits recorded in a commercial population of Churra sheep.

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