SESSION 29

giusi.monteleone@senfimizo.unipa.it

IDENTIFICATION AND EVALUATION OF β-DEFENSIN POLYMORPHISMS IN VALLE DEL BELICE DAIRY SHEEP

G. Monteleone[1], D. Calascibetta[1,2], M. Scaturro[2], P. Galluzzo[1], M. Palmeri[2], B. Portolano[1,2]

[1]Dip. S.En.Fi.Mi.Zo.-Sez. Produzioni Animali, Università di Palermo, Italy;
[2]Consorzio Regionale di Ricerca Bioevoluzione Sicilia, Santa Margherita di Belìce (AG), Italy

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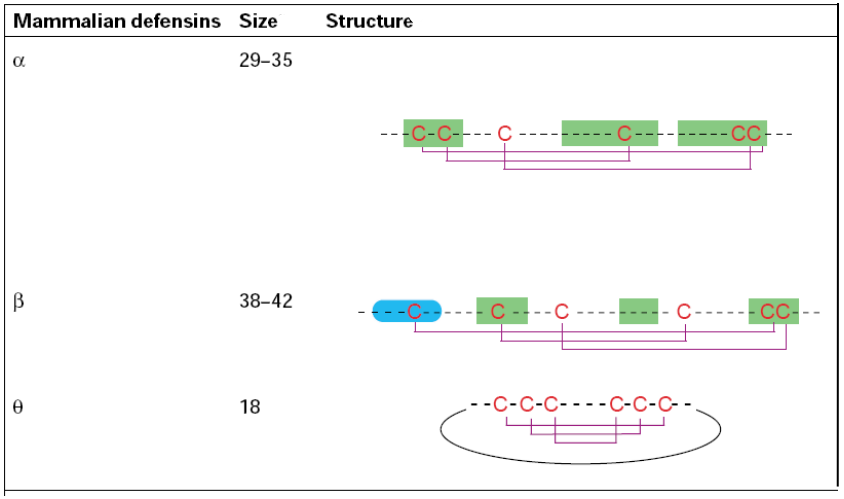
INTRODUCTION (1)

Defensins

- small peptides belonging to the antimicrobial peptides family;
- classified into α-, β-, and θ-defensins based on structure, size and disulfide bonds pattern;
- acting directly against bacteria, viruses and fungi;
- involved in the innate immunity mechanisms;
- expressed in epithelial cells lining various organs and in leukocytes.

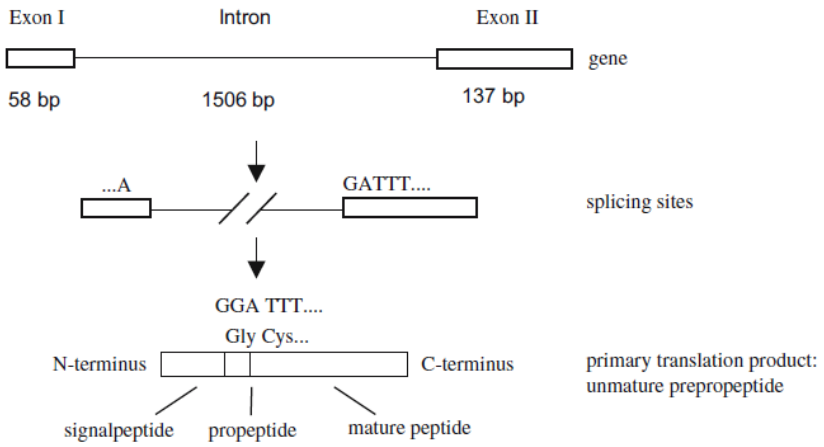


Classification, size and structure of mammalian defensins (Yang *et al.*, 2002)

INTRODUCTION (2)

Sheep β-defensin genes: SBD1 and SBD2

- mapped on chromosome 26;
- two exons and one intron of approximately 1500 bp;
- encode for the signal sequence, the pro-peptide and the mature peptide.



AIM

To identify, validate, and analyze polymorphisms on SBD1 and SBD2 genes in Valle del Belice dairy sheep

MATERIAL & METHODS

- 400 samples of Valle del Belice sheep from four flocks;
- genomic DNA extraction from whole blood;
- PCR reactions;
- sequencing and primer extension reactions;
- analysis of obtained data.

RESULTS & DISCUSSION (1)

	SNP position	Region	Genotypic frequencies		
			Wild type	Heterozygote	Mutated homozygote
SBD1	1747 A→G	3′-UTR	AA (0.78)	GA (0.16)	GG (0.06)
	1757 T→C	3′-UTR	TT (0.78)	TC (0.16)	CC (0.06)
SBD2	89 C→T	coding	CC (0.72)	CT (0.28)	TT (0)
	1659 G→A	coding	GG (0.39)	GA (0.56)	AA (0.05)
	1667 G→A	coding	GG (0.97)	GA (0.03)	AA (0)
	1750 G→A	3′-UTR	GG (0.49)	GA (0.51)	AA (0)
	1761 G→A	3′-UTR	GG (0.83)	GA (0.17)	AA (0)

RESULTS & DISCUSSION (2)

Analyses of SBD2 non-synonymous SNPs

- G→A at position 1659 determines the change Arg^{42} > Lys^{42}
- G→A at position 1667 determines the change Gly^{45} > Arg^{45}

PANTHER

may not have functional impact

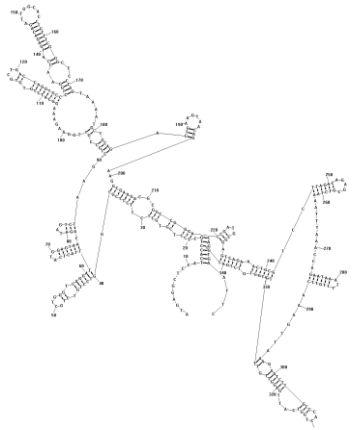
SIFT

do not affect protein function

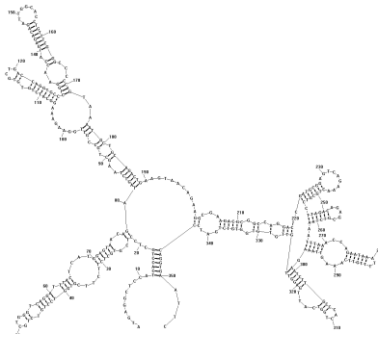
RESULTS & DISCUSSION (3)

Analysis of SBD2 3′-UTR SNPs

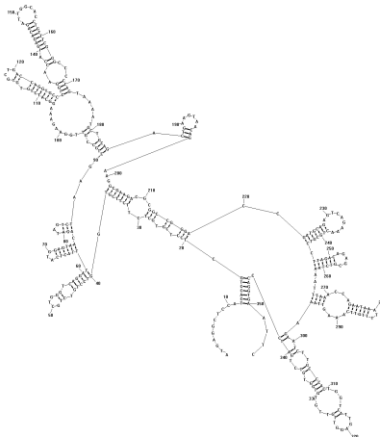
Wild Type SBD2 mRNA



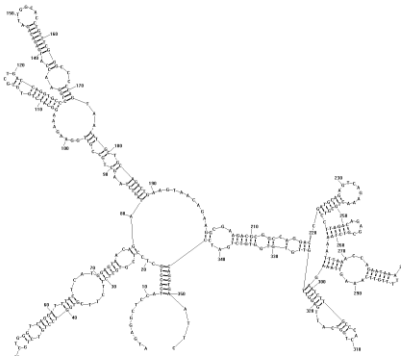
SNP 1750



SNP 1761



SNPs 1750 & 1761



CONCLUSIONS

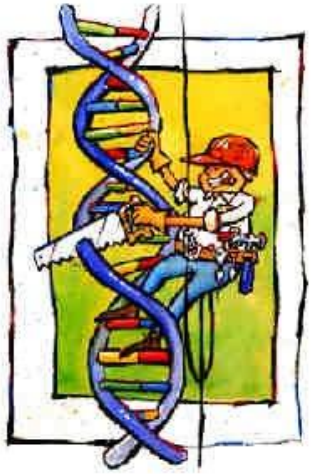
- Sheep β-defensin genes are characterized by polymorphisms;
- SBD2 coding SNPs determining an amino acid change may not have effect on protein function;
- 3′-UTR SNPs could determine changes in the secondary structure of SBD2 mRNA.

⇩

role in the modulation of immune response

FUTURE PERSPECTIVES

- Do SNPs in SBD2 determining amino acid change compromise protein function?
- Do SNPs in 3′-UTR affect post transcriptional events?
- Are these SNPs associated with milk production traits?



Thank you all

for the attention!