



GENETIC VARIABILITY OF LIPOGENIC ENZYMES (DGAT2, SCD) AND GLYCOPROTEIN (BTN1A1) IN CZECH DAIRY GOAT POPULATION

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OBJECTIVE

The aim of this study was to determine genetic variant - genetic polymorphism in lipogenic enzyme:

DGAT2 – diacylglycerol acyltransferase 2

SCD - stearoyl-CoA desaturase

and glycoprotein

BTN1A1 - butyrophilin

MATERIAL AND METHODS

Samples

230 Czech mixed dairy goat breeds



Statistical analysis

PowerMarker data analysis software (Liu and Muse, 2005)

Methods

Table 1. Analyses applied to the DNA samples for genotyping at *BTN1A1*, *DGAT2* and *SCD* loci

Locus	Methods	Region	SNP	Product size	Reference
<i>BTN1A1</i>	PCR-RFLP	exon 4	218C>T, Gln >Lys	459bp	Qu et al., 2011*
<i>DGAT2</i>	PCR-RFLP	exon 4	234C>A, Leu>Met	344bp	An et al., 2011
<i>SCD</i>	PCR-RFLP	exon 3	15G>A, Val >Met	362bp	Chen et al., 2010
			68A>G		
		intron 3	55A>G		

* method has been optimized according to our conditions

DGAT2- Diacylglycerol acyltransferase2

- *DGAT* include 2 major gene families shown to encode DGATs (*DGAT1* and *DGAT2*)
- Both genes code proteins with DGAT activity
- plays a critical role in the synthesis of triacylglycerol (TAG)
- has high expression in liver, white adipose tissue and mammary gland

SNPs in goat *DGAT2* gene

Locus	Location	SNP	AA change	Reference	SeqRef-SNP
<i>DGAT2</i>	exon 3	A>G	Lys>Arg	Fang et al., 2012	NM_205793.2
	exon 4	234 C>A	Leu>Met	An et al., 2011	NC_007313, GU564264
	intron 5	182 C>T	No		JF438995, JF438996
	intron 6	301 G>T	No		

Result

- Detection polymorphisms in the exon 4 (SNP- C234A) by the PCR-RFLP
- no polymorphism – in Czech goat breeds,
- genotype CC is original and natural for the Czech goat breeds

SCD – *stearoyl-CoA desaturase*

- Stearoyl-CoA desaturase (*SCD*) is an integral membrane protein of the endoplasmic reticulum that catalyzes the rate-limiting step in the biosynthesis of monounsaturated fatty acids from saturated fatty acids (Ntambi and Miyazaki, 2004)
- plays a central role in MUFA synthesis (Ntambi and Miyazaki, 2004)
- mapped on 26 chromosome (Yahyaoui 2003; Yahyaoui et al. 2003)
- span a 12–15 kb region
- Consist : 6 exon, varying in size from 131 (third exon) to 4047 bp
5 introns, varying in size from 600 to 3700 bp (Bernard et al. 2001; Yahyaoui et al. 2002)

SNP in goat *SCD* gene

Locus	Location	SNP	AA change	Reference	SeqRef-SNP
	exon2	G>A	No	Crepaldi et al., 2013	rs155505578
	exon3	430G>A	Val>Met	Chen et al., 2011	AF325499-ex3_15G>A
		483A>G	Arg CGA/CGG	Chen et al., 2011	AF325499-ex3_68A>G
	intron3	585T>A,	No	Zhang et al., 2010	Ref.AF422168
SCD		601A<G	No	Zhang et al., 2010	Ref.AF422168-IVS3+55A>G
	intron4	719T>A	No	Zhang et al., 2010	Ref. AF422169
	exon5	C>T	No	Crepaldi et al., 2013	rs155505581
		904/325T>A	No	Yahyaoui et al., 2003	AF339909/AF422170
		942/363G>T	No	Yahyaoui et al., 2003	AF339909/AF422170
	exon 6	690 A>G	313 Tyr > Cys	Zhang et al., 2010	Ref. AF422171
		718 C>G	322 Phe > Leu	Zhang et al., 2010	Ref. AF422171
		802 A>C	350 Arg > Ser	Zhang et al., 2010	Ref. AF422171
	3'UTR	-/TGT		Bernard et al., 2001	AF325499

BTN1A1 – butyrophilin

- *BTN1A1* – milk fat globule membrane proteins
- it is a member of the immunoglobulin superfamily
- it may have a cell surface receptor function
- plays a key role in the secretion of milk lipid and production - effect on performance traits

SNP:

- missense mutation: CTT (Leu) → TTT (Phe) at position 377aa of the *BTN1A1* (526aa) (by Qu Y. et al., 2011)
- association study: milk traits – milk fat, protein total solid, solid-non fat, lactose (Qu Y. et al., 2011)

Result

- SNP in the exon 4 at position CTT (Leu) → TTT (Phe) at position 216 T → C (GU569967) – not confirmed by sequencing analysis in Czech goat breeds
- sequencing analysis – revealed a 2 new SNP in exon 4:
at position 214 T → C (GU59967)
and 218 G → A, Gln → Lys (GU569966)

Allele frequencies, genotype distribution, gene diversity and heterozygosity, PIC and Hardy – Weinberg equilibrium at the *BTN1A1*, *SCD* and *DGAT2* loci in Czech dairy goat populations

Marker	Genotype	Count	Freq	Allele	Count	Freq	He	He _(obs)	PIC	ChiSquare value
<i>BTN1A1</i>	MM	175	0.76	M	399	0.87	0.23	0.21	0.20	1.25
	MN	49	0.21	N	61	0.13				
	NN	6	0.03							
Total		230	1.00			1.00				
<i>SCD</i>	AA	3	0.02	A	44	0.15	0.30	0,25	0.27	43.89
	AB	32	0.21	B	250	0.82				
	AC	6	0.04	C	10	0.03				
	BB	109	0.72							
	CC	2	0.01							
Total		152	1.00			1.00				

CONCLUSSION

- described genetic polymorphism at the *SCD* gene in Chinese goat breeds exist also in Czech goat populations
- characterized SNPs at loci *DGAT2* and *BTN1A1* was not confirmed in Czech goat population
- revealed two new SNPs at locus *BTN1A1* in exon 4
- alleles and genotypes investigated at selected loci can be ancestral and typical for Czech goat population
- necessity for continued research for confirming characterized SNP and searching new SNPs at selected loci as well other candidate genes
- find out an association with production traits in order to improve performance

THANK YOU FOR ATTENTION

