

SUMMARY

Genetic variability of fatty acid transformations genes in domestic sheep (*Ovis aries*)

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Adipose tissue plays an important role in regulating and maintaining the body's homeostasis. Through the secretion of hormones and proteins, it controls lipid and carbohydrate metabolism. Fatty acids, the main component of fats, are responsible for the permeability of cell membranes and participate in a number of metabolic reactions, such as thermoregulation or energy storage. In addition, they affect the quality of products of animal origin (meat and milk), which is related to the ratio of saturated/unsaturated fatty acids (SFA/UFA).

The genes directly or indirectly involved, through encoded proteins, in fatty acid transformations are: fatty acid binding protein 4 (*FABP4*), peroxisome proliferator-activated receptor alpha (*PPAR α*) and gamma (*PPAR γ*), and stearyl-CoA desaturase (*SCD*). Interactions between proteins and their genes are essential for the maintenance of energy balance and the proper functioning of the body. A key argument for conducting experiments focusing on selected genes is the role they play along with the protein products in the changes occurring in adipose tissue, as well as an unsatisfactory knowledge of the genetic structure of *FABP4*, *PPAR α* , *PPAR γ* and *SCD* in domestic sheep.

The research presented in this study was divided into two main parts including an analysis of the structure of selected fatty acid metabolism genes at the genome and transcriptome levels. The first part concerned the analysis of genetic variation in coding sequences, splice and regulatory sites in five breeds of sheep raised in Poland for different purposes: meat (Suffolk, Ile de France), dairy/prolific (Olkuska, Kołudzka) and primitive (Polish mountain sheep). The determinations were aimed to identify polymorphisms in gene sequences *FABP4*, *PPAR α* , *PPAR γ* and *SCD*, and to show possible relationships between the constitutional type and genetic variability in the studied *loci*.

The second part consisted of an analysis of transcript levels of *FABP4*, *PPAR α* , *PPAR γ* and *SCD* in muscle tissue and liver of sheep. Based on the results of Real-Time PCR, the investigation aimed to determine the effect of different factors such as age, breed or varied diet on changes in the expression of genes. Furthermore, fatty acid content of intramuscular fat was determined and basic parameters of growth and fatness were compared.

In the first part of this study, the identification of the genetic structure of the selected genes revealed six polymorphic changes: *FABP4* -c.73 + 13A>T and c.348 + 85G>A, *PPAR γ* -

c.391 – 66C>T (c.481 – 66C>T) and c.529 + 27G>C (c.619 + 27G>C), *SCD* – c.*945G>A and c.*1116A>G. Only in the case of SNP locus c. 1116 *> G was there an association between the body constitution and the productive type of investigated sheep. In the present work substitution c.*945G>A located in the regulatory region of the 3'-UTR of the *Ovis aries* *SCD* gene was described for the very first time.

The results obtained in the second part of the experiment show that age was a factor modifying the expression of *FABP4*, *PPAR α* , *PPAR γ* and *SCD* genes in the liver and muscle tissue. An age-dependent increase in transcripts of *FABP4* and *SCD* genes and a decrease of *PPAR α* and *PPAR γ* were found. Age-dependent changes in the expression of selected genes are not associated with changes in the fatty acid profile of intramuscular fat of sheep *longissimus lumborum* and *biceps femoris* muscle.

It has been shown that increasing the dietary fat intake and the introduction of compound feed rich in PUFA (C18:3 n-3) led to changes in the level of gene expression of *ad libitum* fed sheep. The higher level of gene expression of *FABP4*, *PPAR γ* and *SCD* in the *longissimus lumborum* muscle was associated with the increased content of n-3 and n-6 PUFA in the intramuscular fat. The semi-intensive feeding of sheep with increased fat intake and the introduction of PUFA-rich diets did not result in changes in gene expression in the liver and muscle tissue. It has been observed that introduction of feeds rich in linoleic acid C18:2 n-6 decreased the concentration of monounsaturated fatty acids in the intramuscular fat of *longissimus lumborum* muscle.

The analyses of the transcript level of the investigated genes, performed in the second part of this work, showed that the breed of sheep did not affect the expression level of the four tested genes in muscle and liver tissue or the fatty acid profile of intramuscular fat tissue.

It is concluded, based on the relationship demonstrated between body constitution and productive type, that further studies are needed to investigate the correlation between c.*1116A>G polymorphism and growth rate, slaughter value and meat quality of lambs. In the future this may lead to more effective selection of sheep. In addition, the performed analysis of the genetic structure and the demonstrated impact of factors such as age and varied nutrition on transcript levels of *FABP4*, *PPAR α* , *PPAR γ* and *SCD* genes are a valuable addition to the knowledge of genetics, or nutrigenomics.