

Abstract of the doctoral thesis of mgr Anna Miksza-Cybulska entitled:

**„The use of SNP microarrays to determine the genetic diversity of the national mountain sheep population”**

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The tradition of mountain sheep-keeping in Poland has a long history. Mountain sheep are an integral element of the pastoral economy, not only being the basis for the existence of the local community of the Polish Carpathians, but also because of the role they now play in the culture of highlanders and shaping the mountain landscape. Mountain sheep currently include three breeds: the Podhale Zackel (CP), the Polish mountain sheep (POG) and the Polish colored mountain sheep (POGB). These versatile breeds provide milk for the production of traditional cheeses, wool, hides and very tasty meat. The breeds are perfectly adapted to the harsh climatic conditions of the mountains. Cultural grazing of mountain sheep is of particular importance for the preservation of mountain landscapes and the biodiversity of plant communities in protected areas of national and landscape parks. Taking into account the threat to the existence of mountain sheep, and at the same time their high utility values, perfect adaptation to the environmental conditions of the Polish Carpathians and special importance for the cultural heritage of these regions, the breeds were included in the genetic resources protection program.

The aim of the study was to characterize the genetic structure with the use of SNP microarrays of three mountain sheep breeds including the assessment of the size, nature and biological significance of genetic differences between them and to determine the possibility of using SNP microarrays to monitor genetic variability of native sheep breeds and the usefulness of the obtained data in the implementation of conservation programs populations at risk.

The experimental material was blood taken from randomly selected 300 ewes belonging to three native mountain sheep breeds: CP, POG and POGB, coming from two different herds in each breed group. Molecular analyzes were carried out in the laboratory of the Department of Molecular Animal Biology of the National Research Institute of Animal Production in Balice. Genomic DNA was isolated from whole blood and genotyped using the OvineSNP50 BeadChip microarray (Illumina, San Diego, CA, USA). Microarray scanning was performed using the HiSanSQ system (Illumina). Genotypes were tagged using the GenomeStudio software (Illumina). Statistical analyzes included the assessment of the level of genotyping errors, estimation of genetic variation indicators, visualization of genetic variation and differentiation, identification of differential selection signatures between the studied breeds, identification of directional selection signatures and analysis of genome regions under the influence differential or directional selection in terms of the genes encoded by them.

The analyzes allowed for the identification of differences in the genetic structure of the studied populations, despite a certain group of individuals with a similar genetic profile. The level of heterozygosity was similar for all analyzed breeds, while the effective population size of the Polish colored mountain sheep was the lowest among the evaluated mountain sheep. The value of the inbreeding factor of the studied populations was low, which proves that there is no risk of inbreeding and that the breeding work is properly conducted. Comparing the genetic distances of  $F_{ST}$  between the studied breeds, the highest values were noted between the POG and POGB, and the lowest between the CP and the POG. Detailed studies of the genome revealed the presence of differential selection signals, including 92 genes in the Podhale Zackel, 90 in the Polish mountain sheep and 100 in the Polish colored mountain sheep. In addition, a common genome region under selective pressure was detected, in which two genes were found (ISPD – a gene associated with the development of the nervous system, and SOSTDC1 – a gene associated with the morphogenesis of hair follicles).

The possibility of obtaining comprehensive information on the genetic diversity of the studied populations indicates the usefulness of SNP microarrays in determining the genetic variability of native sheep breeds. Genomic information can be used in the monitoring and management of endangered populations, which are an essential element of the proper implementation of sheep genetic resources conservation programs.