

Variation in the coding sequence of Janus Kinase 2 (*JAK2*) gene in association with meat utility features and reproduction parameters of selected breeds of cattle and sheep

Abstract

The breeding progress of livestock is based on the improvement of individuals in successive generations. Selection is an important element of breeding to consolidate and improve features with utility value. In addition to selection based on phenotypes, discoveries in molecular genetics have allowed the refinement of breeding in terms of desired genotypes. Therefore, there is a need to search for and identify genes whose variability may be directly or indirectly related to the performance characteristics of sheep and cattle.

Janus kinase belongs to the group of protein tyrosine kinases (PTKs) involved in catalyzing the phosphorylation of proteins and determining signaling pathways from various cytokines (e.g. growth hormone, erythropoietin, interleukin, interferon). Together with other genes, it creates somatotrophic axis in which polymorphisms are directly and indirectly related to the phenotype, mainly in relation to the composition and synthesis of milk, meat and carcass properties and reproduction.

The aim was to validate and search for new polymorphic sites in the *JAK2* gene in cattle and sheep. Another objective was to determine the allele frequencies and genotypes of polymorphic sites in *JAK2* and estimation of a possible relationship between selected genetic variants and meat performance characteristics and some parameters shaping the reproductive performance of cattle and sheep.

Using molecular biology methods (PCR, Sanger sequencing, PCR-RFLP, PCR-ACRS), a total of 6 polymorphic sites were analyzed, 3 in cattle (exon 16, 20 and 23) and 3 in sheep (two in exon 6 and one in 24). Then, association analyses were performed with selected meat and reproductive performance characteristics.

In the first paper [D-1], the polymorphism in exon 20 (rs110298451) of the *JAK2* gene was analysed in three breeds of beef cattle: Angus, Hereford and Limousine depending on production capacity. In the case of the Limousine breed, the most favorable features were noted in individuals with the *AA* genotype, i.e. higher birth weight, higher average daily gains and higher body weight at day 210 compared to individuals with the *GG* genotype. Different

results were obtained for the Hereford breed, in which the *GG* genotype determined the highest birth weight, daily gains and body weight at weaning, compared to individuals with the *AA* genotype. In Angus cows, heterozygotes were characterized by the highest body weights and daily gains.

The sequencing of individuals in the **D-2** work allowed the validation of existing and the discovery of new polymorphic sites in the sheep *JAK2* gene. In the case of the first polymorphic site (rs160146162) located in exon 6, the *AA* genotype was rare in the Pomeranian breed (15%) and not identified in the Suffolk breed. The *GG* genotype was most often identified in the Suffolk breed, while in the Pomeranian breed, heterozygous individuals were the most common. In the second polymorphic site (rs160146160), also located in exon 6, the *AG* genotype was most frequently identified in both breeds, and the *GG* genotype the least frequently. In the case of the third polymorphic site (rs429445187), located in intron 24, the highest share of heterozygotes was noted.

The third publication [**D-3**] was an extension and a summary of earlier work. Hereford and Limousine cows with *AA* (e16/*Rsa*I) and *AA* (e23/*Hae*III) genotypes were characterized by the highest body weight and better daily gains ($P \leq 0.05$). In the case of sheep, regardless of breed, in the studied periods, the highest body weight and daily gains were characterized by individuals with genotypes *AA* (e6/*Ear*I), *GG* (e6/*seq*) and *AA* (e24/*Hpy*188III) ($P \leq 0.01$). The same individuals of the Pomeranian breed were also characterized by better fertility and survival of lambs ($P \leq 0.01$).

These are the first association studies for all polymorphic sites identified in the coding part of the *JAK2* gene. Single nucleotide polymorphisms in the *JAK2* gene can serve as genetic markers of growth and development traits and selected reproductive traits in ruminants, if, for example, they are further studied in subsequent populations and analyzed in haplotype and/or combined genotype systems.

Nicola Oster 5.10.23r.