Polysaccharide storage myopathy (PSSM) in the domestic horse

Summary

Polysaccharide storage myopathy is a hereditary disease occurring in horses of various breeds and ages. It is characterized by abnormal accumulation of glycogen and polysaccharide derivatives in the skeletal muscles. The most common symptoms of the disease are exercise intolerance, muscle stiffness, progressive lameness of the rear limbs, increased breathing rate, sweating, and muscle pain. Proper diagnostics can identify individuals carrying the mutant gene and eliminate them from breeding, so that the population with the mutant allele of the *GYS1* gene remains under control. The aim of the analyses was to design a suitable restriction enzyme that will recognize the mutation site of the gene *GYS1*, mutations of which are responsible for the occurrence of PSSM in the domestic horse. A bioinformatic analysis of sequence of the gene was performed. The use of the newly designed primers should result in an amplified sequence 244 bp in length (210 bp without primers). The enzyme *CviAII* enzyme was used to cut it in silico to obtain fragments of 35 bp and 175 bp. Owing to these differences, greater potential for molecular diagnostics of the *GYS1* gene mutation responsible for PSSM in the domestic horse was demonstrated. The disease is not treated pharmacologically Analgesics are administered to reduce pain, but they do not alleviate the disease symptoms. The only treatment is a diet low in starch and rich in fat in combination with regular, gradually increasing exercise.

KEY WORDS: Polysaccharide storage myopathy (PSSM), *GYS1* gene, domestic horse