

Abstract of the doctoral thesis of MSc Magdalena Jankowska entitled:

“Application of molecular biology techniques in the karyotype evaluation among young horse populations in Poland”

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Date of abstract preparation: 28.05.2021

In Poland, horse breeding has a centuries-old tradition. It resulted in shaped and valuable breeds, such as horses of native conservative breeds: the Polish Konik horse, Hucul horse, Wielkopolska horse, Malopolska horse, Silesian horse and Polish Coldblood horse (at the sokolski and sztumski type), as well as purebred polish Arabian horses kept, among others, in studs in Janów Podlaski, Michałów and Białka.

Reproduction is an essential element of horse breeding. Horses are relatively late-maturing animals and the assessment of the breeding value for a mare takes up to 5 years, depending on the breed. Additionally, compared to other livestock species, horses exhibit a poor reproduction rate. The percentage of foalings in mares is estimated at around 50-60% per season. It has been shown that sex chromosomal abnormalities are the main, non-infectious causes of decreased fertility and are widely associated with foaling problems in horses. Moreover, carriers of chromosomal aberrations often do not show any phenotypic or behavioral disorders, which shows the need for the application of cytogenetic techniques to diagnose them.

To date, there has been only one screening study presenting the karyotype abnormalities in the horse population in Poland, showing the incidence of anomalies in 2% of the tested animals. However, the above study was performed using classical cytogenetic techniques with a limited number of analyzed metaphases that could result in the underestimation of the number of affected animals. To increase this identification rate of chromosomal aberrations, including those with a lower frequency of occurrence, the screening in the population of young horses in Poland was undertaken using molecular cytogenetic techniques.

The presented study showed karyotype abnormalities in 3.8% of the studied young horse population. The cytogenetic analysis revealed sex chromosome abnormalities, including X chromosome monosomy, X chromosome trisomy, sex reversal syndrome and

the loss of X chromosome fragments. Additionally, a translocation between chromosome 1 and chromosome X was identified in the Polish horse population for the first time.

In horses, which are characterized by a low rate of reproduction, proper cytogenetic diagnosis is especially essential to reduce or even avoid an unnecessary and expensive fertility treatment.