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MOLECULAR DIAGNOSTICS OF SELECTED HORSE DISEASES OF THE MUSCULAR AND NERVOUS SYSTEM

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Animal genetic diseases are a significant problem for breeders. The implementation of veterinary tests identifying the genetic predisposition can improve breeding and change clinical decisions and animal care. We present the optimization of molecular diagnostics of selected genetic horses' diseases. One of the more common horse diseases, inherited in a dominant manner, is Hyperkalemic Periodic Paralysis (HYPP), manifested by muscle weakness and tremor and cardiac arrhythmia. HYPP in horses is associated with a change in c .4248C> G (p.F1416L) in the SCN4A gene coding for the subunit of the skeletal muscle sodium channel. Chronic Para spinal myositis (PSSM) is manifested by impaired glycogen metabolism and excessive deposition of sugars in the muscles, resulting in motor failure, tremor and muscle stiffness. Type 1 PSSM is linked with the c.926G>A (p.R309H) mutation in the GYS1 gene, coding the glycogen synthase. The Cerebellum Abiotrophy (CA) syndrome is a neurological disease and most often affects purebred Arabian horses. It is inherited in a recessive way and connected with the degeneration of the cerebellar cell of Purkinje. The symptoms are ataxia, head tremor and balance disorders. The diagnosis of CA is difficult due to symptoms similar to other neurological diseases. CA is associated with the mutation c.284G>A (p.R95H) in the TOE1 gene. The aim was to develop tests to identify horses' genetic predisposition to these diseases. PCR primers were designed, and the amplification conditions were optimized. The products were sequenced using the ABI PRISM® 3700 sequencer. The developed DNA tests can be the basis for the routine diagnostics of these diseases. They enable unambiguous diagnosis and the possibility of their implementation immediately after birth allows for the implementation of appropriate veterinary care. In the case of CA, they enable the detection of asymptomatic mutations carriers and thus eliminate them from breeding lines.

Recent Publications

1. Scott EY, Penedo MCT, Murray JD, Finno CJ (2017) Defining trends in global gene expression in Arabian horses with cerebellar abiotrophy. The Cerebellum. 16:462–472.
2. Brault LS, Famula TR, Penedo MCT (2011) Inheritance of cerebellar abiotrophy in Arabians. Am. J. Vet. Res. 72:940–944.
3. Brault LS, Cooper CA, Famula TR, Murray JD, Penedo MCT (2011) Mapping of equine cerebellar abiotrophy to ECA2 and identification of a potential causative mutation affecting expression of MUTYH. Genomics. 97:121–129.
4. Brosnahan MM, Brooks SA, Antczak DF (2010) Equine clinical genomics: A clinician's primer. Equine Vet. J. 42:658–670.
5. Mccue ME, Valberg SJ, Jackson M, Borgia L, Lucio M, Mickelson JR (2009)Polysaccharide storage myopathy phenotype in quarter horse-related breeds is modified by the presence of an RYR1 mutation. Neuromuscul. Disord. 19:37–43.

Biography

Karolina Lisiak - Teodorczyk has her expertise in molecular diagnostics. She is focusing on searching for genetics wariants responsible for hyperkalemic periodic paralysis, the cerebellum abiotrophy (CA) syndrome or chronic Para spinal myositis. She is currently being involved in developed new tests which will bring enormous benefits to animal husbandry.

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