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PROGRESSIVE SENSORY NEUROPATHY WITH IN GOLDEN RETRIEVER DOGS

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Mitochondria are organelles present in almost all eukaryotic cells (except erythrocytes), are crucial for the production of cellular energy and play an important role in maintaining life and regulating cell death. They play a key role in various metabolic pathways, such as oxidative phosphorylation, fatty acid oxidation, the Krebs cycle, the urea cycle, gluconeogenesis and ketogenesis. Mitochondrial diseases are a group of genetically determined diseases that involve mitochondrial dysfunction, manifested by energy deficit in the cell. This group includes, for example, progressive sensory neuropathy with ataxia (SAN), a disease inherited in the maternal line, occurring in Golden Retriever dogs. This disease has not been observed in representatives of other breeds of domestic dogs.

The aim of the literature research was to identify the causative mutations of SAN in domestic dogs, as well as possible therapies.

This disease is caused by a deletion of adenine at position 5304 (mutation ΔT5304) in the mitochondrial gene encoding tRNA tyrosine (*tRNATyr/MT-TY*). tRNA plays a key role in the process of protein synthesis, serving as a link between the mRNA molecule and the elongating amino acid chain that will make up the protein. Its function is to read information from the mRNA (by binding the tRNA anticodon to the mRNA codon) and deliver the appropriate amino acid to the ribosome. Genes encoding tRNAs constitute about 9% of the mitochondrial genome, but mutations in these genes are responsible for more than half of mitochondrial diseases. The *tRNATyr* gene is responsible for encoding tRNA that delivers tyrosine to the ribosome. Progressive sensory neuropathy with ataxia in Golden Retrievers affects both the central and peripheral nervous systems. Symptoms of the disease most often manifest between 2 and 8 months of age. In rare cases, sick dogs may not have any clinical symptoms. The main symptoms of the disease are a slowdown in the rate of ATP production in the mitochondria of the muscle cells and a decrease in the activity of the respiratory chain. The most common symptoms also include ataxia, abnormal gait, and joint laxity. As the disease progresses, balance disorders, loss of muscle control and motor coordination may occur. Males often urinate without lifting their legs, and problems with holding urine may occur in both sexes. There is no muscle atrophy, and despite the fact that the disease significantly reduces the standard of living, there have been no animal behaviors that would indicate that the animals

felt pain. Heteroplasmy occurs to a small extent. It has been found that in sick individuals, unmutated mtDNA molecules are at a level of only about 3%.

Currently, there are no medications or therapies that would be used in sick individuals. Veterinarians recommend only rehabilitation of sick dogs, while in severe cases of this disease, dogs are put to sleep.

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