Medical Xpress

Neurological disorder in golden retriever dogs caused by a mutation in mitochondrial DNA

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Sensory ataxic neuropathy (SAN) is a recently identified neurological disorder in Golden Retriever dogs with onset during puppyhood. Affected dogs move in an uncoordinated manner and have sensory deficits. Researchers from the Swedish University of Agricultural Sciences, Uppsala University and the Karolinska Institutet have now revealed that SAN is caused by a mutation in mitochondrial DNA. The study is published May 29 in the open-access journal *PLoS Genetics*.

The researchers were able to trace back all affected offspring on the maternal side, over more than 10 generations, to a female that lived during the 1970s. This implied a maternal inheritance, which was confirmed by the identification of a one base pair deletion in the mitochondrial tRNA-Tyr gene. Further analyses revealed that the mutation leads to mitochondrial dysfunction, which in turn causes a progressive loss of neurons.

The researchers showed that about five percent of the current Swedish Golden Retriever population carries the mutation causing SAN. The identification of the mutation now allows genetic screening tests to identify carriers and prevent the mutation being transmitted to further generations.

"This is a good example of how a close collaboration between clinicians and geneticists led to a rapid detection of a harmful mutation that can

now be eliminated from this dog population to reduce suffering and disease," said co-author Karin Hultin Jäderlund. The study also provides a new animal model for similar mitochondrial disorders in humans, said co-author Izabella Baranowska, and could potentially be used for testing therapeutic approaches.

More information: Baranowska I, Jäderlund KH, Nennesmo I, Holmqvist E, Heidrich N, et al. (2009) Sensory Ataxic Neuropathy in Golden Retriever Dogs Is Caused by a Deletion in the Mitochondrial tRNATyr Gene. *PLoS Genet* 5(5): e1000499. doi:10.1371/journal.pgen.1000499, www.plosgenetics.org/article/i … journal.pgen.1000499

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