From a mystery to a hereditary muscular dystrophy – where are we at?

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Associate Professor Maria Kaukonen, DVM PhD, presented our annual spring meeting with an update on muscular dystrophy. In 2018 this disease was a mystery to us all but it has now been identified as a form of muscular dystrophy.

First we learned where it all started. 7 years ago in 2018 first contact was made with Maria Kaukonen, describing the symptoms and discussing the role of genetics. Kaukonen suspected the disease to be a polygenic trait, meaning that the disease was caused by the collaboration of multiple genes. Genetic testing isn't possible for polygenic traits but the investigation moved forward nonetheless. As the research continued, it was revealed that the disease was caused by just one genetic flaw, meaning its monogenic.

The affected dogs could be divided into two groups based on the bloodwork. The first group had severely elevated CK, moderately elevated ASAT, mildly elevated ALAT and normal AFOS. The second group had severe elevation in liver values such as ALAT, ASAT, AFOS, but CK was normal. A few unfortunate individuals fit in both groups with all the values elevated. The first group represents true muscular dystrophy, whereas the second group represents some form of liver disease, likely polygenic.

Different types of muscular dystrophies

There are numerous different muscular dystrophies, and they are classified according to location of the symptoms, genetic background, age of onset etc. Hereditary muscular dystrophies present in multiple breeds. For further resources in Finnish you can read the licentiate thesis of LVM Ida-Lotta Jakonen titled "Koirien perinnölliset lihasdystrofiat – esimerkkinä schapendoes". None of the previously documented muscular dystrophies in other breeds match with ours.

In the lecture we were shown the diagnostics and typical symptoms of muscular dystrophies. Most important diagnostic tool is the bloodwork. Other ways are EMG (electromyography) other imaging techniques and histopathological tissue biopsies. Kaukonen doesn't recommend biopsies lightly since the process is painful and the diagnostic value is small. So far biopsy samples have been collected from euthanised dogs.

Over 300 samples

At the moment the research team has access to 327 genetic samples of our breed. The samples have been given voluntarily for overall genetic research, not just concerning the muscular dystrophy so not all dogs are provided with bloodwork. 76 of these 327 dogs are known to have elevated CK with 44 of them having a severe elevation. Approximately half of them have serious symptoms. 9 affected dogs have been autopsied and sampled post mortem, which has provided valuable insight to the disease. The samples must be taken as fresh as possible in Helsinki with special arrangements, so unfortunately foreign dogs can't participate in this way.

Mode of inheritance

When investigating muscular dystrophies, researcher use pedigree analysis and genome wide association studies (GWAS) where the whole genome is sequenced. The pedigree analysis has shown that the mode on inheritance is most likely autosomal and recessive. This means it doesn't relate to gender and the dog needs to get a faulty allele from both parents to be affected. This is similar to gPRA and HUU. GWAS has located the allele to a certain part of a certain chromosome which confirms the mode of inheritance and the fact that we can most likely get a genetic test for the disease.

Almost 70 % carriers or affected

Kaukonen believes the genetic variant responsible for this muscular dystrophy has finally been found through GWAS but confirmations are ongoing. No further info can be released before the findings have been scientifically published.

At this point the genetic variant has been scanned in 290 samples. 90 of these dogs represent wild type, i.e. free from disease, 120 dogs are carriers and 80 are homozygotes, i.e. genetically affected. Approximately 10 % of the homozygotes have normal boodwork but the rest have a high CK. Approximately 10 % of carriers have mildly or moderately elevated CK and 7 % of wild types have a mildly elevated CK. At this point all the dogs with CK over 550 are homozygotes.

New samples are welcome

We are still hoping to receive information from abroad also. If your dog has had bloodwork done, mainly CK whether it be normal or elevated, you can participate by sharing the results in our database. The form is available in Finnish and English at www.schapendoes.fi/jalostus/lihasdystrofia/veriarvot/. You can also send samples for the genetic study with the bloodwork results, more information at www.koirangeenit.fi.

There will be a scientific article published once the confirmation is done for the genetic variant. At that point commercial laboratories can start providing the test and we will keep you updated. Dogs participating in the study will get the result from the research team automatically.