Muscle disorder in the Schapendoes

In 2018 University of Helsinki started to study a muscle disorder in the schapendoes, due to the initiative of the Finnish Schapendoes Club and concerned dog owners. This is a completely new disorder in veterinary science, and it only seems to occur in our breed. Dogs that are considered affected, have extremely high CK values (creatine kinase) and possibly other elevated values (ALT and AST). Clinically dogs can vary from asymptomatic to having severe symptoms leading to euthanasia. Clinical symptons in this condition usually are weight loss, chronic gastrointestinal symptoms (e.g. vomiting and diarrhea), muscle weakness and/or tension, muscle loss, fatigue, tremor and heat and exercise intolerance.

In our breed there have been individuals with extreme high liver enzyme levels in the past also. Extreme high values meaning in this case 10–20 -fold increase compared to reference values what it comes to liver. The dogs are usually clinically completely normal. This has been previously investigated in Utrecht University the Netherlands and in Norway, with no clear explanation so far and no certainty if these are linked with this muscle disorder.

At the moment there are 6 dogs in Finland that have been euthanized because of this condition and studied post mortem to help with the research. Additionally less than 10 dogs have died or been euthanized with more or less similar symptoms with no bloodwork or autopsy. Main autopsy findings are chronic or recurring degenerative changes in skeletal muscle leading to scarring of the muscles. Muscle findings are the connecting element between these dogs and no consistent liver changes were found. Thus the disorder has now been located in the skeletal muscles rather than the liver.

The Canine Genetics Group in University of Helsinki has now over 300 DNA-samples of our dogs. Starting 2020 they did whole-genome sequencings between 20 affected and 16 healthy individuals finding that the disorder is genetic and the pattern is autosomal recessive (similar to gPRA and HUU). They have located a rough problem area in the genes but unfortunately not yet the mutation itself. If the specific gene can’t be found by spring 2024, they will look into creating a genetic marker test for the breed, while still continuing the search for the affected gene. The marker test would be a bit less specific test than testing the actual gene but could be of help to us before finding the single gene. Both group leader, Professor Hannes Lohi, and the researcher in charge, Maria Kaukonen, are quite positive, that it will be found eventually.

Resources:
Differential diagnosis for hepatic disease as the cause of serum alt increase; myopathy of the schapendoes breed dog (P. Syrjä, T. Jokinen, M. Speeti, M. Kaukonen, H. Lohi 2023) LINK

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About the enzymes

CK – creatine kinase
Creatine kinase is found as various isoenzymes in skeletal muscles, heart muscle, brain, and to a small extent in some other organs. The concentration of creatine kinase increases rapidly in various muscle injuries and, as a result, of intense physical exertion. Also, as the lesion heals, the concentration drops rapidly. Creatine kinase is used in the diagnosis of muscle injuries and diseases, as well as in the monitoring of racehorse training, for example. In dogs, the half-life of creatine kinase in the blood is notably short, about 2 hrs.

ALT – alanine aminotransferase
ALT is found in the liver, muscle (cardiac and skeletal), kidneys, and erythocytes (in some species). ALT is fairly liver specific in dogs, cats, rabbits, rats and primates. It is a fairly specific indicator of liver damage, in which case blood activity may increase tenfold and remain elevated for 1–2 weeks. Some increases are possible in severe muscle diseases of the dog and cat due to release of enzyme from this tissue (in this situation, higher increases in AST are typically seen).

AST – aspartate aminotransferase
Aspartate aminotransferase belongs to the same group of enzymes as ALT, but it is found in several tissues. It's located in the cytosol and mitochondria of cells, so it is released relatively easily in cases of cell and tissue damage in acute diseases. Both AST and ALT levels increase with liver damage, but ALT is more liver specific. The increase in AST concentration is also often caused by muscle damage (heart or skeletal muscle), but it should be noted that in muscle damage, changes in concentration occur more slowly than, for example, in creatine kinase (CK). The effect of potentially heavy and repetitive physical activity on increasing concentrations should be taken into consideration, especially when examining the results of racehorses and race dogs.

Resources:
Movet Laboratories, Finland
eClinPath, Cornell University College of Veterinary Medicine

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Q&A

What are the blood values in this muscle disorder?
As the name says, the most critical value is muscle enzyme CK. Liver values (mainly ALT) can also be high, due to the changes in the muscles rather than actual liver disease. Secondary liver disease is also possible, but the autopsy results don't show consistent changes in the liver.

What makes these values high?
CK and ALT are high because of the processes this disorder generates in skeletal muscle. At the moment, unfortunately no one knows where the disorder comes from.
Tremor, which is one of the clinical symptoms in this disorder, has been known to be in this breed for decades. This could explain some of the elevations in CK but the levels have not been tested on these dogs in the past.

How do I know if my dog has the muscle disorder?
If your dog doesn't have any symptoms, you have no way of telling if they're affected. Bloodwork should be done at least for CK, ALT and AST. If your dog is considered to be used for breeding, check these values and ask the other partner to do so too.

I have the results, what's next?
With your dog's test results, there are three options:

a) No symptoms, normal blood values

Your dog most likely doesn't have this muscle disorder.

No symptoms, elevated blood values

Keep an eye on possible symptoms. The condition is progressing, and if you see clear signs of symptoms, check blood values again, in order to know where you are at the moment. Even if your dog has elevated blood values but no clinical symptoms, they may very well live a normal life. For example, one of the authors have two dogs with high values, tested 4,5 years ago, and both still very much alive and kicking with no symptoms at all.

b) Symptoms and elevated blood values

Unfortunately your dog most likely has this disorder. The condition is progressing and may lead to saying goodbye to your dog.

My dog has high CK/ALT/AST values. What can I do and who can I contact to get the right information?
The diagnosis is confirmed mainly by excluding other conditions so please consult your veterinarian. The Finnish Schapendoes Club has collected most recent information on their website. For further information (owners, breeders, veterinary professionals...) please contact the board at hallitus@schapendoes.fi.

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