

Cerebellar ataxia and the AHT's contribution to polymyopathy research

Cerebellar ataxia

The Animal Health Trust (AHT) announced at the Hungarian Vizsla Health day (November 6th) that a mutation for a rare cerebellar ataxia (cerebellar cortical degeneration) affecting the Hungarian Vizsla had been discovered, after work carried out in collaboration with Joe Fenn at the Royal Veterinary College. Clinical signs of progressive ataxia (gait incoordination) are apparent between 2-3 months of age, and a marked head tremor has been seen in one case. There is no treatment or cure and affected dogs are euthanized on welfare grounds due to the progressive nature of the disease.

Although this rare condition has only been formally reported in two cases, a DNA test will be launched early in 2016 to avoid the potential for the condition to become more common in the future, and with the aim of completely eradicating this genetic form of the disease. The disease is caused by a recessive mutation, so all carriers can be safely bred from, provided they are bred to a DNA-tested clear mate.

The AHT is currently randomly screening a subset of DNA samples submitted to the research department for the mutation, to estimate the number of carriers in the UK Vizsla breed population.

Polymyopathy research

The AHT has made a contribution to the polymyopathy research by genome sequencing DNA from a polymyopathy case which was seen in the neurology clinic at the AHT. Genome sequencing is a method of determining all the 2.4 billion letters of DNA code needed to make a dog – this is a huge amount of data!

Lorna Kennedy at the University of Manchester is currently performing a genome-wide association study comparing cases of polymyopathy with healthy control dogs. This approach uses genotyping arrays with DNA markers which give a snapshot of the DNA at around 170 thousand places in the genome, or one every 14 thousand letters of DNA. The approach is used to look for areas of the DNA that are similar in the cases, but different in the controls i.e. places that are likely to contain the mutations that cause the disease of interest. If significant regions of the genome are identified, then the genome sequencing data can be used to look through every letter of DNA to search for a fault that could potentially cause disease.

We believe this data is a valuable resource for the research into polymyopathy and we are happy to share the data with Lorna and collaborators.