Animal Health Trust
Canine Genetics Research Group
Lanwades Park
Kentford
Newmarket
Suffolk CB8 7UU

Tel: 01638 751000 x1284
Fax: 01638 555643
Email: louise.burmeister@aht.org.uk
Web: www.aht.org.uk

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UPDATE: Keeshond Primary Hyperparathyroidism Research

The Animal Health Trust has been committed to finding (and publishing in a scientific peer-reviewed journal) the genetic cause of Primary Hyperparathyroidism (PHPT) seen amongst the Keeshond population.

Thanks to the continued support from the Keeshond community, we have been able to make significant steps toward finding the mutation which causes the disease. To date, we have compared PHPT affected and unaffected Keeshond DNA samples (some of which are part of the Give a Dog a Genome initiative) to point towards specific regions of DNA in the dog genome that we believe contains the mutation.

Recently, we have discovered a particular region of DNA which is very interesting; however, it is a very complex region to investigate in detail and our investigations have been limited by the technologies that are currently available. We have therefore not yet been able to pinpoint the exact mutation but are confident that it is located within this region. Excitingly, we now have access to some very promising technologies which are hoped to resolve the challenges that we have faced so far.

These exciting new technologies, however, don't offer a quick, easy or cheap fix and our research will likely continue through the year before we can potentially provide concrete conclusions. Because of this, we believe that it would best serve the Keeshond community if we were to develop and provide a linkage test of our own in the meantime.

A linkage test is a type of DNA test that looks at one or more DNA markers located very close to, and inherited with, the disease mutation. They are used when the disease mutation has not been completely defined or cannot be reliably tested for technical reasons. This will allow us to accurately predict how the PHPT mutation is inherited. To our knowledge, the causal genetic mutation for PHPT is not known by anyone, and therefore any DNA tests currently available commercially must also be linkage tests.

Robust and reliable linkage tests are just as useful as mutation-based tests, so we would still be able to offer genetic advice on how to avoid introducing the PHPT mutation into breeding lines based on the test results. We would look to work with the UK Kennel Club to ensure that dogs can still be assigned 'hereditary clear' status for PHPT as long as the current or future Kennel Club testing requirements are met.

We hope to be able to offer this test by the summer of 2020.

As mentioned, research is ongoing to identify the exact mutation that causes PHPT. An important component of this research is further investigation of the clinical aspects of the disease, being undertaken by Dr Barbara Skelly at Cambridge University. We suspect that due to the nature of the disease, some dogs that are affected by PHPT (and have the disease mutation) might not show any obvious clinical signs of being ill. In these circumstances, specialised blood and urine tests can give more accurate diagnoses and insights into how PHPT manifests. We therefore encourage the Keeshond community to continue to support us and Dr Skelly with future requests for information or participation in the research. We can of course assure the community that any communications with us are completely confidential.

Canine Genetics Research Group,
The Animal Health Trust