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4th May 2023

George Sofronidis
Orivet
PO Box 110 St Kilda
Victoria 3182 Australia

Re: DNA tests on Orivet Border Collie panel for PLL, Cystinuria, vWII, MH

Dear Mr Sofronidis,

The National Border Collie Council Health Subcommittee has been reviewing DNA and health tests for Border Collies.
We have identified a number of tests that Orivet currently includes on the BC panel that are not appropriate and/or relevant to the breed.
We believe that inclusion of these tests is misleading and can and does lead to people assuming their dogs are clear for a particular disease when in fact they could be a carrier or clinically affected.

These tests are:

Primary Lens Luxation – the test used is not the causal mutation for border collies. There are cases of affected dogs in the UK where the parents were bred together after relying on a clear result on the Orivet panel. BVA ophthalmologists are aware of this issue and recommend regular eye testing as part of the KC recommended testing for BCs. Refer to article by Dr Sheila Crispen “Hereditary Eye Diseases in Dogs” 2018, BVA and KC.
A single nucleotide substitution in the ADAMTS17 gene has been shown to be the cause of PLL in 17 breeds. This mutation has been excluded from involvement in the Border Collie and the Shar Pei, indicating other mutations are the cause in these breeds.

Cystinuria – the test used is not the causal mutation for Border Collies.
There are confirmed cases of cystinuria III diagnosed by clinical testing in the US where DNA testing has shown the dogs to be clear for the cystinuria mutation that Orivet is using. Research is ongoing at the University of Pennsylvania to discover the mutation responsible for cystinuria III in BCs. They are the leaders in cystinuria research, having discovered the mutations for forms I and II.

Myotonia congenita/hereditaria (cattle dog) – we are concerned that this test has not been validated for border collies. There is currently no published research to support this test for border collies. If you are aware of studies validating this test in Border Collies could you please forward the references to us.

Von Willebrands Disease type II – the mutation that this test is based on is a non-causal mutation in any breed. This was published in 2018 as per the reference below and extract from the paper below. Cornell University is the leader in research on von Willebrand's disease in dogs. Border Collies appear to be affected sporadically by vWIII and there is no DNA test for this form in border collies.

Donner J, Anderson H, Davison S, Hughes AM, Bouirmane J, Lindqvist J, et al. (2018) Frequency and distribution of 152 genetic disease variants in over 100,000 mixed breed and purebred dogs. PLoS Genet 14(4): e1007361. https://doi. org/10.1371/journal.pgen.1007361
"Given the large number of additional breeds (Table 3 and [11]) discovered to tentatively carry vWD type 2, we also submitted samples from dogs homozygous for the originally published putative variant (VWF c.4937A>G; [32]) for vWF:Ag testing (Table 4). The results of all tested dogs were either borderline or within the reference range specified by the clinical testing laboratory, lending further support to the notion [23] that the originally published variant is in fact not the causal variant in the gene."

The National Border Collie Council is concerned that breeders may be relying on these incorrect tests to make breeding decisions and potentially putting pups at risk of being affected by one of these diseases.

We request that Orivet remove these tests from the Border Collie Panel as soon as possible and would appreciate it if you could confirm when these tests have been removed from the Border Collie panel.

Yours sincerely,

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