



THE WORKING KELPIE COUNCIL OF AUST INC.

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CEREBELLAR ABIOTROPHY (ATAXIA) IN AUSTRALIAN WORKING KELPIES

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Cerebellar abiotrophy (CA) is a developmental disorder affecting the brain of a number of dog breeds, including Kelpies. Specifically, it affects the region of the brain that controls co-ordination, and as a result control of muscles fails to develop normally. This leads to the appearance of incoordination that is generally referred to as ataxia.

With financial support from the Working Kelpie Council, the Australian Companion Animal Health Foundation, and The University of Sydney, we undertook a study of the pathology and genetic predisposition to CA in Working Kelpies. This work formed the subject of a PhD project by Dr Annie Pan, and was supervised by Professors Rosanne Taylor and Claire Wade, and myself. The project expanded an earlier study conducted by Dr Alan Wilton (deceased) and colleagues.

The main objectives of the project were to (1) understand the neuropathology of CA in Australian Working Kelpies, and (2) identify CA-associated gene regions and causative mutations for CA in this breed. The rationale was that identification of the genetic basis of CA in Kelpies could provide a foundation for the development of reliable DNA-based testing for this disorder.

DNA analysis

Two genetic variants associated with increased risk of CA were identified in Australian samples. These two variants correlated with the timing of clinical signs of ataxia. A third variant was found in one family of Kelpies from Germany, but we have not seen cases of CA with this variant in Australia. Based on our statistical analysis, we consider these gene variants to be very strong risk factors for developing CA when two copies are inherited.

Recently, Dr Kao Castle and her company Dog Breeding Science developed their own DNA tests based on Dr Pan's PhD work. These tests were made available to Kelpie owners and breeders on a fee for service basis. The DNA tests report results for three markers based on the two variants found in Australian dogs and the third from the German family. They named the tests as NUP153, LINGO3 and VMP1. A marker test accurately tracks a DNA sequence that is close to the cause of the disorder. Many DNA tests for genetic health and improvement are marker tests.

Dog Breeding Science reports the results for each of these tests in one of three ways:

- "Negative" = clear of that risk gene; has no copies of the risk gene.
- "Carrier" = positive for one copy of the risk gene. This copy could have come from either parent. These dogs will not have any signs of CA but may give rise to pups with a higher risk of developing CA if bred with another carrier or with a dog with two copies of the marker.
- "Positive" or "Double Carrier" = positive for two copies of the risk gene. It would have received a copy from both parents. These dogs have a much greater risk of developing CA.