



THE WORKING KELPIE COUNCIL OF AUST INC.

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HISTORY OF WKC'S RESEARCH WITH ATAXIA (CA) BEFORE ITS TRANSFER TO SYDNEY UNIVERSITY. - By Dr Don Robertson.

Genes for cerebellar abiotrophy (CA) have probably been in the Kelpie population for many generations, but were first brought to notice of the Working Kelpie Council in 1987 when Dr Don Robertson told the WKC about an affected litter. The investigation was published in the Australian Veterinary Journal in 1989 by Jan Thomas of Murdoch University and Don Robertson of Muresk, a branch of Curtin University.

At that time, it was presumed to be the same inherited ataxia previously reported in many other breeds. It showed the same autosomal recessive inheritance pattern as in these other breeds. However, subsequent research has shown that the Kelpie forms of CA have only been found in Kelpies and the related Border Collie and Koolie breeds, which have a history of occasional crossbreeding.

The Working Kelpie Council immediately commissioned research by DNA specialists at Curtin University to establish the incidence of the disease and to find DNA tests to identify carriers. The molecular geneticists, Professor John Wetherall and Associate Professor David Growth, were confident that they would quickly locate the mutation, but it proved elusive.

A video clip of affected pups and dogs, paid for by a grant from Curtin University, was widely disseminated by the WKC with detailed description of symptoms. Breeders, owners and veterinarians were encouraged to report suspected cases to the WKC. Confidentiality was guaranteed because diagnosis could not be certain and the WKC wanted no impediment to reporting.

As a result, veterinarians from Australia and overseas sought advice from WKC.

Where practicable, affected pups, dogs or samples were collected and sent for analysis by pathologists. This was funded sometimes by breeders, sometimes by WKC and often by volunteer WKC members concerned to assist the research. Nancy Withers, Jim Marshall, Tony Rutter and many others located and transported affected dogs for the research.

Reports were accumulated and collated suggesting that the incidence of ataxia was not widespread but had risen recently because a few carrier sires had been used very widely.

By 1995 the Curtin University researchers had a substantial collection of samples with known pedigrees but had to admit that the hoped-for breakthrough was not imminent. They handed the work to a new team at Murdoch University. This group, headed initially by Professor Clive Huxtable, continued to receive samples through the work of the WKC volunteers but were not able to make progress.

Seeking a fresh approach, the WKC sent slides from affected dogs to Professor Colin Masters, medical neuropathologists at the University of Melbourne, to determine whether knowledge of similar conditions in humans might be useful. National Health and Medical Research funding was a possibility. No parallels with human conditions were found.

The WKC then approached Dr Alan Wilton at the University of New South Wales because of his successful work with dingo DNA and tests for some recessive defects in Border Collies. He was optimistic that his group would have similar success with CA in Kelpies. The samples from Curtin University were delivered to him. Unfortunately, Murdoch University had not kept its collection.

Dr Wilton's group revived the original videos and added to them. The WKC distributed this second video to its members with exhortations to continue supporting the research with reports and samples.

Among those responding, dogs were provided that had a form of ataxia differing from that previously studied. It usually becomes evident later in a pup's life and gets progressively worse, unlike early-onset ataxia, which is typically apparent at six weeks of age and does not get worse with age. This eventually proved to be critically important information for the researchers, but for a couple of years it caused confusion because it was not recognised that separate genes were involved. Jeremy Shearman researched this phase as part of his PhD.

WKC made substantial annual contributions in funding and in kind to this research at the University of New South Wales, which was also supported with generous funding from Terry Snow, a philanthropist, who had bought a dog from a breeder of show kelpies that were also producing ataxic pups. Terry Snow also paid for a third educational video filmed with Kathy Christian's dogs and featuring WKC representative, Kevin Howell, which was distributed by WKC.

Annie Pan took up the challenge working on this project as her Master's thesis with Alan Wilton at the University of New South Wales.

Terry Snow offered further funding for this next phase but made it conditional upon the WKC collecting a \$25 fee for CA research for every dog registered by the WKC. This was unacceptable to the WKC Board so Terry Snow's funding lapsed. WKC funding continued.

When Alan Wilton died in 2011, the research was delayed until it was taken up by Professors Peter Williamson, Claire Wade and Rosanne Taylor at the University of Sydney. Annie Pan completed her thesis and relocated to the University of Sydney.

The WKC transferred its annual financial and in-kind support to the Sydney University team.