Hereditary cerebellar abiotrophy in Australian Kelpie dogs

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A breeder of Australian Kelpie dogs has had several pups that show significant cerebellar disease. A prospective breeding trial was carried out. A bitch showing clinical disease was mated with her full brother, a clinically normal dog from the same litter. In the resulting litter of 8 pups, 2 males and 2 females were severely affected clinically. A fifth pup, a male, was thought "clumsy" by the owner.

Three pups showed clinical disease from 5 to 6 weeks of age (Table 1). When these were examined at 8 weeks of age, all were very bright and alert. There was significant ataxia, hypermetria, head tremor and truncal ataxia. There was no sign of weakness. There was decreased proprioception in all legs but this was particularly evident in both hind legs. Tactile placing was depressed. Each pup had varying degrees of change, one pup being unable to progress forward without tumbling over. Another pup had grand mal seizures.

A fourth pup was examined at 13 weeks of age. This pup was noticed to have clinical signs between 11 and 12 weeks of age. There was a decrease in proprioception in all limbs, the menace response was absent and there was a mild head tremor. The fifth pup was examined at 5½ months of age. The dog showed a decrease in hind limb proprioception and mild hypermetria when running. A sixth and seventh dog were examined at the same time and showed some decrease in hind limb proprioception only. Pup 7 was not necropsied and the last (Pup 8) was not available for examination but was reported to be normal.

In all dogs, lesions were confined to the brain. In Pup 5, there was moderate hydrocephalus, with attenuation of the corpous callosum. In Pup 6, the cerebellum was marginally smaller than that of Pup 5.

In Pups 1, 2, and 3, histological changes were very similar and confined to the cerebellum. There was regional loss of Purkinje cells. Associated with this there was a marked reduction in the granular cell density (Figure 1). There was mild spongiosis and Wallerian degeneration in the white matter tracts of affected folia. The lesion was most severe in the lingula, central and culmen lobules of the vermis. Changes were milder in the anterior ansiform lobules and medial hemispheres. In these areas, occasional necrotic Purkinje cells were seen. These had pyknotic nuclei and eosinophilic cytoplasm. Occasional empty glial baskets were present.

In Pup 4, no necrotic Purkinje cells were observed. However, there was loss of Purkinje cells and a paucity of granular layer in decline and folium lobules of the caudal vermis. There was associated mild Wallerian degeneration in the lobular white matter. Pup 5 had hydrocephalus observed grossly but no change was present in the cerebellum. Pup 6 showed a mild to moderate loss of Purkinje cells and granular cells in the decline folium of the vermis. Mild vacuolation and Wallerian degeneration was present in the white matter of the dentate nucleus. Mild white matter spongiosus was also present in the vestibular nuclei.

The lesion varied in intensity within each dog. The most severely affected areas were the anterior lobules of the vermis (Figure 2). The vermis was also the most commonly involved site. As the lesion progressed, the anteriolateral areas were affected. In one pup (Pup 4), the decline and folium lobules of the vermis were affected.

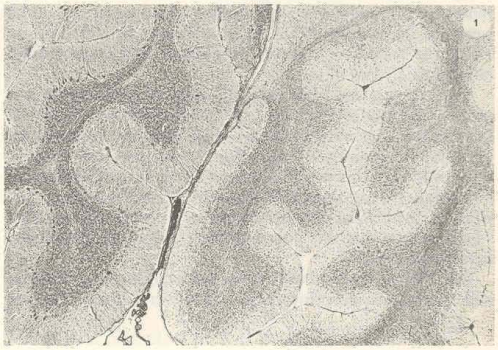

Figure 1. Cerebellum. Affected folium with paucity of granular cells, absence of Pirkinje cells and mild Wallerian degeneration adjacent to a normal folium. Haematoxylin and eosin x 40.

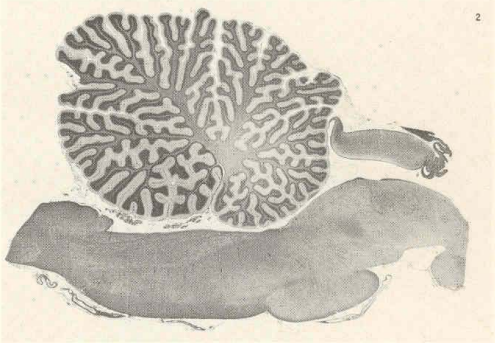

Figure 2. The anterior lobules of the cerebellar vermis are the most severely affected. Haematoxylin and eosin x 10.

TABLE 1
Clinical signs in Australian Kelpie dogs with cerebellar abiotrophy

Dog	Age at Examination	Age of onset of Clinical Signs	Clinical Signs	Pathological Findings
1	8 weeks	5-6 weeks	Ataxia, hypermetria, head tremor, truncal ataxia, proprioceptive deficit	Regional loss of Purkinje cells and granular cells. Necrotic Purkinje cells. Degeneration cerebellar white matter.
2	8 weeks	5-6 weeks	As above	As above
3	8 weeks	5-6 weeks	As above plus intermittent seizures	As above
4	13 weeks	11-12 weeks	Proprioceptive deficit, mild head tremor, absent menace response	Regional loss of cells and granular cells. Degeneration cerebellar white matter
5	5.5 months	Unknown	Proprioceptive deficit in hind limbs, mild hypermetria	Moderate hydrocephalus
6	5.5 months	Unknown	Proprioceptive deficit in hind limbs	Regional loss of Purkinje cells and granular cells. Degeneration cerebellar white matter and vestibular nuclei
7	5.5 months	—	Normal	Necropsy not performed

Cerebellar disease in dogs is uncommon. Cerebellar hypoplasia has been reported in Chow Chows (Knecht *et al* 1979), but the most commonly reported lesion in the dog is abiotrophy. Carmichael *et al* (1983) reports a degenerative cerebellar disease with hydrocephalus in Bull Mastiffs. Gordon Setters (Cork *et al* 1981), Rough coated Collies (Hartley *et al* 1978), Airedales (Cordy and Snelbaker 1952) and Border Collies (Gill 1980) are affected by abiotrophic processes in the cerebellar cortex. Cerebellar cortical and extrapyramidal nuclear abiotrophy has been reported in Kerry Blue Terriers (de Lahunta and Averill 1976; De Forest *et al* 1976; Montgomery and Storts 1983). In this disease, degeneration of Purkinje cells is the first change followed by a retrograde transynaptic neuronal degeneration in the olivary nucleus, caudate nucleus and substantia nigra.

Age of onset of clinical signs varies with the breed. In the report by Gill (1980) the clinical signs began between 6 and 8 weeks of age, whilst in the Gordon Setters (Steinberg *et al* 1981), the condition is slowly progressive with onset from 6 months to 2 years of age.

In the disease described here, the onset of degeneration appears to be prior to 6 weeks of age but the variation in clinical signs means that mildly affected dogs may not be identified until several months of age. Pup 3 was reported to have seizures though this was not noticed at the time of clinical examination. No histological lesions were present to explain this, either in the central nervous system or in other organs. This would suggest that this pup may have had an accompanying biochemical lesion.

The fifth pup showed only mild clinical signs of hypermetria and decreased proprioception. However, there was no evidence of cerebellar lesions histologically. The moderate hydrocephalus that was present may have been causing some cerebellar peduncle dysfunction. It is uncertain whether this lesion is associated with the abiotrophy seen in its littermates.

In the other dogs examined, the lesions appear similar to those seen in other breeds (Cork *et al* 1981; Gill 1980), varying only in the age of neuronal degeneration.

The pattern of inheritance has not been fully identified. However, it is not inconsistent with a single recessive autosomal gene which has been suggested in other breeds (Steinberg *et al* 1981). In the kelpies studied here, the putative gene appears to have been inherited from several dogs which have been prominent in sheep dog trials and have been widely used for breeding. Consequently, an increase in the incidence of cerebellar abiotrophy in working sheep dog strains can be expected.

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(Accepted for publication 6 April, 1989)