

## **Ceroid Lipofuscinosis**

This is an inherited disease, which is not contagious, but it is fatal and cannot be treated. It affects the nervous system including the brain. Ceroid Lipofuscinosis is known as Batten's Disease in Humans.

CL has been found in other breeds of dogs i.e. Cocker Spaniels, Dachshunds, English Setters, Miniature Schnauzers, Rough Collies, and Salukis as well as in Devon Cattle, South Hampshire Sheep and in Siamese Cats

The occurrence of CL in Border Collies is not the fault of any one person or group. The defective gene was carried by an imported dog at a time when the disease was unidentified. Since then, CL has occurred sporadically, as most Australian bred Border Collies are descended from that dog. Therefore, no breeder can be 100% sure that their stock does not carry the defective gene.

Affected animals appear normal until aged approx 15 months. From that age any or all of the following signs may be noted:

- Unreasonable apprehension or fear of familiar objects/surroundings
- sight disturbance,
- abnormal gait – is unsteady on feet and has difficulty in climbing or jumping, tends to prop or goose step,
- demented behaviour,
- mania,
- hyperactivity,
- rage,
- disorientation,
- fixations,
- loss of toilet training,
- strange or abnormal behaviour

The progress and effect of the symptoms will steadily continue to deteriorate and medication cannot improve the condition. Affected animals have all been euthanased by the age of 3½ years. CL symptoms can be confused with other brain disorders.

## **History**

Early this century, F.E. Batten studied and described the disease in children, hence the name Batten's Disease. This is the same disease now being diagnosed in dogs and known as Ceroid Lipofuscinosis. It is hoped research being conducted on the disease in dogs will benefit the children suffering from Batten's Disease. As in dogs, there is no cure and no treatment yet available. The life span of children with Batten's Disease is approximately 7 years.

In June 1980, a 17 month old Border Collie bitch was referred to the University of Melbourne suffering from 'fits' and 'sight disturbance'. In December that year another dog, a 19 month old male was also studied. After enquiries were made at the Eye and Ear Hospital, Melbourne, it was suggested that the dog was affected by Ceroid Lipofuscinosis. The dog was put down and the first case re-opened and compared. It was considered that the two animals suffered from the same condition.

Cases from 3 separate litters were diagnosed in 1985/86 and others were discovered in 87/88. Since then other litters have been found to have affected animals, in all (from 1980 to 1998) a total of 18 litters were diagnosed as having produced CL.

The Border Collie Club of Victoria published in 1989 a series of articles and in March 1989, Dr. R. Mitten delivered a lecture at the club which drew a large audience. A sub-committee was then elected to deal with all aspects of CL in Victorian Border Collies.

The Border Collie Club of NSW Inc. then formed a sub-committee which acts in conjunction with the Victorian Club to verify disease related information. All Border Collie Clubs in Australia liaise to share information. Newsletters carry articles concerning CL and pedigrees of PROVEN carriers have been published with the very generous permission of the owners /breeders of affected or carrier animals.

## **Veterinary Aspects & Inheritance of CL**

Ceroid Lipofuscinosis is one of a group of Metabolic Storage Diseases. It is characterised by the accumulation of Ceroid Lipofuscinosis, a wax like liquid waste product of cell metabolism, which is normally removed by body enzymes. In cases of CL one of the enzymes is missing. Research has determined that CL has an autosomal recessive mode of inheritance, i.e. both the sire and dam of an affected dog must be either carriers or affected themselves. A mating of a carrier animal to a clear (non carrier) animal can produce carrier offspring.