

## Neuronal Ceroid Lipofuscinosis

**Affected breeds:** Border Collie, Tibetan Terrier

Neuronal Ceroid Lipofuscinosis (NCL) is a severe inherited disease which causes a gradual degeneration of the nervous system. The disease is caused by different mutations in several different breeds; Animal DNA Diagnostics Ltd provides tests for NCL in the Border Collie and in the Tibetan Terrier. The disease varies in nature in different breeds, as does the severity, and the age at which the disease becomes apparent. In Border Collies NCL may become obvious from about 15 months of age, although in the Tibetan Terrier the onset is later at 5-7 years. Affected puppies may develop psychological abnormalities such as increased aggression, hallucinations, hyperactivity and epileptic fits (Border Collies), and retinal atrophy leading to blindness (Tibetan Terrier). In addition affected dogs lose their coordination and may have difficulty in walking, eating and house-training.

This is a serious, fatal disease caused by a recessive genetic mutation. This means that dogs which carry the mutation ("CARRIERS") are healthy but will pass the mutation on to an average of 50% of their offspring. Puppies which inherit two copies of the mutation will suffer from TNS ("AFFECTED").

### **This test is particularly useful for breeders:**

- To identify carriers among their breeding stock so that they can avoid CARRIER X CARRIER mating combinations which would risk AFFECTED puppies.
- To conclusively diagnose Neuronal Ceroid Lipofuscinosis

### **This test will be reported as:**

**CLEAR** : no evidence of the Neuronal Ceroid Lipofuscinosis mutation  
**CARRIER** : carries one copy of the mutation, which will be passed to 50% of offspring  
**AFFECTED** : carries two copies of the mutation, and will develop NCL

### **The genetic status of dogs can be used to predict breeding outcomes when different combinations are mated:**

CLEAR X CLEAR = 100% CLEAR  
CARRIER X CLEAR = 50% CARRIER, 50% CLEAR  
CARRIER X CARRIER = 25% AFFECTED, 50% CARRIER, 25% CLEAR

### **References**

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