THREE MUTATIONS THAT CAUSE DIFFERENT FORMS OF
CANINE NEURONAL CEROID LIPOFUSCINOSIS

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ABSTRACT

Neuronal ceroid-lipofuscinosis (NCL), also known as Battens disease, is really a group of inherited neurodegenerative diseases. A common feature of the ceroid lipofuscinoses is the deposition of autofluorescent cytoplasmic storage material in cells in the brain, retina, and many other tissues. The major symptoms are mental retardation, visual failure, loose of motor skills, seizures, and eventually premature death. In the European countries and USA, the disease affects one in 12,500 to 100,000 people. Usually children appear to be healthy at the birth and develop normally until onset of disease.

Canine NCLs have been reported in a variety of breeds where they are important as veterinary diseases and as potential models for the human NCLs. We have discovered that a missense mutation in the CLN8 gene causes NCL in English Setters, a missense mutation in the CTSD gene causes American Bulldog NCL, and frame shift mutation in the CLN2 gene causes Dachshund NCL.