

## VETERINARY TECHNICAL DATASHEET

Progressive Retinal Atrophy, (CNGA1-PRA); mutation originally found in Shetland Sheepdog



Mutation Found In :Shetland Sheepdog

### Disorder Type

- Eye

### Disease Severity

- Moderate

### Background

Progressive retinal atrophy (PRA) comprises a group of genetically inherited diseases affecting dogs of various breeds. PRA is characterized by retinal degeneration and progressive loss of vision culminating in blindness. PRA is known to affect over 100 breeds. The genetic background varies highly among breeds and populations. In Shetland Sheepdogs, Collie Eye Anomaly may cause PRA but another causative mutation for PRA has been found as well. All dogs homozygous for the CNGA1 mutation have PRA but not all PRA cases in Shetland Sheepdogs can be explained by this mutation. The mode of inheritance is autosomal recessive.

### Key Signs

- Degeneration of the photoreceptor cells of the retina
- Night blindness
- Vision loss
- Blindness

### Clinical Description

Clinical signs of the disease typically appear only when the vision of the dog is already impaired. The dog may be reluctant to walk in a dark environment or up or downhill. Affected dogs may also have dilated pupils with an abnormal shine. The disease does not cause pain and there is no curative therapy. The age of onset and age of diagnosis vary between 2 to 11 years of age.

### Mode of Inheritance

- autosomal recessive

### Gene Name

- CNGA1

### Next Steps

A blind dog tends to adapt well to the loss of vision. However, some dogs may exhibit a tentativeness when introduced to unknown environments because their vision is compromised. Occasionally, they may react abruptly (snapping) if they are startled so caution and use of verbal queues should be taken when handling a blind dog. Caretakers should take precautions to protect the blind dog from threats it cannot detect (ex. cliffs, sharp points on furniture, moving vehicles).

### References

Wiik A, Ropstad E, Ekesten B, Karlstam L, Wade C, Lingaas F. Progressive retinal atrophy in Shetland sheepdog is associated with a mutation in the CNGA1 gene. Anim Genet. Oct;46(5):515-21, 2015.