

## VETERINARY TECHNICAL DATASHEET

Progressive Retinal Atrophy Type III, (PRA type III); mutation originally found in Tibetan Spaniel and Tibetan Terrier



Mutation Found In :Tibetan Spaniel, Tibetan Terrier

### 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. Causative gene mutations have been identified in several breeds but some of them are still unidentified. PRA type III is encountered in the Tibetan Spaniel and the Tibetan Terrier. The disorder is inherited in an autosomal recessive manner.

### Key Signs

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

### Clinical Description

The onset of clinical signs of PRA type III is typically at 5 years of age with initial loss of vision in dim light (night blindness) which gradually progresses to total blindness. The first signs are caused by loss of rod photoreceptor cells required for vision in dim light followed by reduction of the visual field and blindness as the cone cells of the retina also degenerate.

### Mode of Inheritance

- autosomal recessive

### Gene Name

- FAM161A

### 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

Downs LM, Mellersh CS. An Intronic SINE Insertion in FAM161A that Causes Exon-Skipping Is Associated with Progressive Retinal Atrophy in Tibetan Spaniels and Tibetan Terriers. PLoS ONE 9(4): e93990 2014.