VETERINARY TECHNICAL DATASHEET

Progressive Retinal Atrophy, (PAP1_PRA); mutation originally found in Papillon and Phalne

WISDOM HEALTH™

Mutation Found In :Papillon, Continental Toy Spaniel

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 affecting the Papillon and Phalne breeds is caused by a mutation in the CNGB1 gene. 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

In the Papillon and Phalne breeds, the onset of clinical signs of PRA is typically at 5-6 years of age with initial loss of vision in dim light (night blindness) that gradually progresses to total blindness. Because PRA in the Papillon and Phalne progresses slowly, some affected dogs often maintain adequate daylight vision throughout their natural lifespan. The onset of clinical signs is caused by loss of rod photoreceptor cell function, followed by degeneration of cone cells.

Mode of Inheritance

- autosomal recessive

Gene Name

- CNGB1

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

Ahonen SJ, Arumilli M, Lohi H. A CNGB1 Frameshift Mutation in Papillon and Phalne Dogs with Progressive Retinal Atrophy. PLoS ONE 8:e72122, 2013.