

VETERINARY TECHNICAL DATASHEET

X-Linked Progressive Retinal Atrophy 2, (XLPRA2)



Mutation Found In :Mixed breed

Disorder Type

- Eye

Disease Severity

- Moderate

Background

Progressive retinal atrophy (PRA) is an eye disorder encountered in several dog breeds and is characterized by retinal degeneration and progressive loss of vision leading eventually to blindness. Many of the causative mutations behind different forms of PRA have been identified but some of them have not. X-linked progressive retinal atrophy 2 (XLPRA2) is an eye disorder causing early onset retinal degeneration in mixed breed dogs, and is due to the degeneration of rod photoreceptors. It is inherited in an X-linked manner.

Key Signs

- Night blindness
- Blindness

Clinical Description

Clinical signs such as night blindness and “tunnel vision” are observed typically 6 to 7 weeks of age. The disease is progressive and after onset will result in blindness by 2 years of age.

Mode of Inheritance

- X-linked

Gene Name

- RPGR

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 (such as cliffs, sharp points on furniture, moving vehicles).

References

Zhang Q, Acland GM, Wu WX, Johnson JL, Pearce-Kelling S, Tulloch B, Vervoort R, Wright FA, Aguirre GD. Different RPGR exon ORF15 mutations in Canids provide insights into photoreceptor cell degeneration. Hum Mol Gen 11(9):993-1003, 2002.

Zangerl B, Johnson JL, Acland GM, Aguirre GD. Independent Origin and Restricted Distribution of RPGR Deletions causing XLPRA. J Hered 98(5):526-530, 2007.