

## VETERINARY TECHNICAL DATASHEET

Rod-Cone Dysplasia 1, (rcd1); mutation originally found in Irish Setter



Mutation Found In :Irish Setter, Irish Red and White Setter

### Disorder Type

- Eye

### Disease Severity

- Moderate

### Background

Rod-cone dysplasia 1 (rcd1) in Irish Setters and Irish Red and White Setters is an inherited eye disorder that causes photoreceptor degeneration that results in blindness. It is inherited in an autosomal recessive manner.

### Key Signs

- Night blindness
- Loss of vision
- Blindness

### Clinical Description

The first clinical signs of rcd1 include night blindness from rod cell degeneration which begin by 6 to 8 weeks of age. The disorder will progress as the cone cells also degrade, impairing day vision, and eventually the disorder will result in complete blindness. rcd1 is fast progressing, causing blindness by one year of age.

### Mode of Inheritance

- autosomal recessive

### Gene Name

- PDE6B

### 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

Suber ML, Pittler SJ, Qin N, Wright GC, Holcombe V, Lee RH, Craft CM, Lolley RN, Baehr W, Hurwitz RL. Irish setter dogs affected with rod/cone dysplasia contain a nonsense mutation in the rod cGMP phosphodiesterase beta-subunit gene. Proc Natl Acad Sci U S A 1;90:3968-3972, 1993.