VETERINARY TECHNICAL DATASHEET

Cone-Rod Dystrophy, mutation originally found in Standard Wirehaired Dachshund, (crd SWD)

Mutation Found In :Dachshund (Wirehaired), Dachshund (Miniature Wirehaired)

Disorder Type

- Eye

Disease Severity

- Moderate

Background

The onset of this disorder involves the reduction of cone cells required for day vision and continues with the reduction of rod cells necessary for night vision.

Key Signs

- Progressive vision loss leading to blindness

Clinical Description

Ocular fundus changes, such as an initial cellophane-like increase in the tapetal area and pigment migration from the non-tapetal region, are observed. The age of onset of crd SWD varies from 10 months to 3 years, although affected puppies can be recognized as early as 5 to 10 weeks of age by the presence of pinpoint sized pupils upon examination with focal light. The disease leads to blindness within 6 years from the time signs are first noted.

Mode of Inheritance

- autosomal recessive

Gene Name

- NPHP4

Next Steps

Early on, affected dogs should not be exposed to bright light as it can be irritating or even painful. The diagnosis of crd SWD can be confirmed by performing a fundic exam to evaluate the retina. Treatment is supportive.

References

Ropstad EO, Bjerk's E, Narfström K. Clinical findings in early onset cone-rod dystrophy in the Standard Wire-haired Dachshund. Vet Ophthalmol 10(2):69-75, 2007.

Wiik AC, Wade C, Biagi T, Ropstad EO, Bjerk's E, Lindblad-Toh K, Lingaas F. A deletion in nephronophthisis 4 (NPHP4) is associated with recessive cone-rod dystrophy in standard wire-haired dachshund. Genome Res 18(9):1415-1421, 2008.