

# VETERINARY TECHNICAL DATASHEET

Neuroaxonal Dystrophy; mutation originally found in Spanish Water Dog



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Mutation Found In :Spanish Water Dog

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## Disorder Type

- Nervous system

## Disease Severity

- Severe

## Background

Neuroaxonal dystrophy in Spanish Water Dogs is a rare neurodegenerative disorder. Affected dogs are presented with progressing neurological signs such as gait abnormalities, abnormal vocalization, incontinence, and behavioral changes. At the cellular level, the disease is characterized by spheroid formation in the central nervous system. Clinical signs are progressive and usually result in euthanasia over quality of life concerns.

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## Key Signs

- Progressive neurological signs
- Gait abnormalities
- Behavioral deficits
- Incontinence

## Clinical Description

Clinical signs appear between six and eleven months of age. The signs are progressive in nature and include gait abnormalities; behavioral changes such as dullness, nervousness, or abnormal vocalization; incontinence; and uncontrolled defecation. Affected dogs may also show compulsory pacing, visual deficits, nystagmus, head tilt and neurological deficits, predominantly related to sensory localization. Neurological examination may show a mild head tilt, generalized cerebellar ataxia, hypermetria of the thoracic limbs, a depressed patellar reflex, proprioceptive deficits, decreased menace, visual deficits, positional nystagmus, and decreased muscle tone. A characteristic pathologic feature for the disease is the development of spheroids in the grey matter of the cerebral hemispheres, the cerebellum, the brain stem, and in the sensory pathways of the spinal cord.

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## Mode of Inheritance

- autosomal recessive

## Gene Name

- TECPR2

## Next Steps

Treatment is supportive care, however, due to the progressive nature of the condition, clinical signs typically lead to euthanasia before two years of age on welfare grounds.

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## References

Hahn K, Rohdin C, Jagannathan V, Wohlsein P, Baumgärtner W, Seehusen F, Spitzbarth I, Grandon R, Drögemüller C, Jäderlund K. TECPR2 Associated Neuroaxonal Dystrophy in Spanish Water Dogs. PLoS ONE 10(11): e0141824, 2015.