

Neuroaxonal Dystrophy; mutation originally found in Spanish Water Dog

In brief

Neuroaxonal dystrophy in Spanish Water Dogs is a rare neurodegenerative disorder. Clinical signs appear between six and eleven months of age. Affected dogs are presented with progressing neurological signs such as gait abnormalities, abnormal vocalisation, incontinence, and behavioural changes. The disease is pathologically characterised by spheroid formation in the central nervous system. The inheritance pattern is autosomal recessive. Clinical signs are progressive and usually result in euthanasia over quality of life concerns.

Clinical overview

Clinical signs of the disease are progressive in nature and include gait abnormalities; behavioural changes such as dullness, nervousness, or abnormal vocalisation; incontinence; and uncontrolled defecation. Affected dogs may also show compulsory pacing, visual deficits, nystagmus, tilting of the head and neurological deficits, predominantly related to sensory localisation. Neurological examination may show a mild head tilt, generalised cerebellar ataxia, hypermetria of the thoracic limbs, a depressed patellar reflex, proprioceptive deficits, decreased menace, visual deficits, positional nystagmus, and decreased muscle tone. Due to the progressive nature of the condition, clinical signs typically lead to euthanasia before two years of age. A characteristic pathologic feature for the disease is the development of spheroids in the central nervous system. Spheroid formation can be observed in the grey matter of the cerebral hemispheres, the cerebellum, the brain stem, and in the sensory pathways of the spinal cord.

References

Online database

Online Mendelian Inheritance in Animals, OMIA (<http://omia.angis.org.au/>). Faculty of Veterinary Science, University of Sydney.

Scientific articles

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.

Disease severity

Severe

Clinical signs

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

Mode of Inheritance

Autosomal Recessive

Results of the genetic test are reported as follows:

- Clear
- 🚩 Carrier
- 🚩 At risk

Mutation(s) found in:

Spanish Water Dog