VETERINARY TECHNICAL DATASHEET

Hyperekplexia or Startle Disease

Mutation Found In :Irish Wolfhound

Disorder Type

- Nervous system

Disease Severity

- Severe

Background

Hyperekplexia or startle disease is a rare congenital disorder encountered in Irish Wolfhounds. The first clinical signs of hyperekplexia can be seen in very young puppies. Affected puppies suffer from muscle stiffness and tremors when handled, and are usually euthanized before three months of age.

Key Signs

- Muscle stiffness
- Tremor
- Difficulty or inability to stand
- Rigid posture
- Cyanosis

Clinical Description

The first clinical signs of hyperekplexia can be observed in one week old puppies. The affected puppies respond to handling with rigid limbs and tremors. Relaxation or sleeping eases the clinical signs. Affected puppies have difficulties standing and they develop a stiff posture with all four limbs extended. Cyanosis or bluish discoloration of mucous membranes is also characteristic of hyperekplexia. Cyanosis can be observed especially when an affected puppy is nursing. The condition is severe and progressive. Affected puppies are usually euthanized before three months of age.

Mode of Inheritance

- autosomal recessive

Gene Name

- SLC6A5

Next Steps

The wellbeing of affected puppies should be closely monitored. Because of the severity of the clinical signs, affected puppies are usually euthanized on welfare grounds before three months of age.

References

Gill JL, Capper D, Vanbellinghen JF, Chung SK, Higgins RJ, Rees MI, Shelton GD, Harvey RJ. Startle disease in Irish wolfhounds associated with a microdeletion in the glycine transporter GlyT2 gene. Neurobiol Dis 43(1):184-189, 2011.