

VETERINARY TECHNICAL DATASHEET

Cerebral Dysfunction; mutation originally found in Frisian Stabyhoun



Mutation Found In :Frisian Stabyhoun (Pointer)

Disorder Type

- Nervous system

Disease Severity

- Severe

Background

Cerebral dysfunction is caused by altered function of the dopamine transporter in the brain and causes widespread mental and physical signs.

Key Signs

- Depression
- Circling
- Excessive sniffing
- Limb weakness

Clinical Description

The disease causes severe, progressing cerebral signs including mental depression, excessive sniffing and circling, walking backwards, and limb weakness. Affected puppies do not exhibit any abnormalities in routine blood work, cerebrospinal fluid analysis, or magnetic resonance imaging and are typically euthanized before 4 months of age.

Mode of Inheritance

- autosomal recessive

Gene Name

- SLC6A3

Next Steps

Treatment is supportive care. Humane euthanasia for affected dogs is often elected.

References

Arata S, Ogata N, Shimozuru M, Takeuchi Y, Mori Y. Sequences and polymorphisms of the canine monoamine transporter genes SLC6A2, SLC6A3, and SLC6A4 among five dog breeds. J Vet Med Sci. 2008 Sep;70(9):971-5.