VETERINARY TECHNICAL DATASHEET

L-2-Hydroxyglutaric aciduria, (L2HGA); mutation originally found in Staffordshire Bull Terrier

Mutation Found In :Staffordshire Bull Terrier

Disorder Type

- Nervous system

Disease Severity

- Severe

Background

L-2-hydroxyglutaric aciduria (L2HGA) is an inherited inborn error of intermediary metabolism found primarily in Staffordshire Bull Terriers. The mutation causes the enzyme that breaks down L-2-hydroxyglutaric acid (L-2-hydroxyglutarate dehydrogenase) to be defective and therefore concentrations in the body increase; high concentrations are particularly toxic to tissues in the central nervous system.

Key Signs

- Ataxia
- Muscle stiffness during exercise or excitement
- Altered behavior
- Epileptic seizures

Clinical Description

Accumulation of L-2-hydroxyglutaric acid levels in the body leads to damage to the central nervous system in affected dogs. Typically, the disease presents itself between six months and one year of age but can present as late as seven years of age. Neurological clinical signs include ataxia, muscle stiffness during exercise or excitement, altered behavior, or epileptic seizures.

Mode of Inheritance

- autosomal recessive

Gene Name

- L2HGDH

Next Steps

While there is no cure, palliative treatment for the clinical signs associated with the disease can improve the dog's quality of life. Seizures do generally respond to standard anti-epileptic therapy, such as phenobarbital.

References

Penderis, J, Calvin, J, Abramson, C, Jakobs, C, Pettitt, L, Binns,MM, Verhoeven, NM, O'Driscoll, E, Platt, SR, Mellersh, CS. L-2-hydroxyglutaric aciduria: characterization of the molecular defect in a spontaneous canine model. J Med Genet 44:334-40, 2007.