# VETERINARY TECHNICAL DATASHEET

Lagotto Storage Disease, (LSD)

Mutation Found In :Lagotto Romagnolo

## Disorder Type

- Nervous system

## Disease Severity

- Moderate/severe

## Background

Lagotto storage disease (LSD) is a progressive neurological disorder characterized by cerebellar ataxia. Histological examination reveals neuronal vacuolization in the peripheral and central nervous systems. Aggregation of vacuoles can also be seen in several other tissues, but only vacuolization of the nerve cells seems to be relevant. Lagotto storage disease affects the Lagotto Romagnolo breed.

## Key Signs

- Ataxia
- Episodic nystagmus
- Behavior changes

## Clinical Description

The onset of clinical signs and pattern of progression vary considerably between individuals. The first signs can be seen at the age of 4 months to 4 years. Typically the first observable sign of LSD is usually progressive ataxia (uncoordinated movements), which may not be noticed by the owner. Some of the affected dogs also suffer from episodic nystagmus (involuntary eye movement). In some cases, nystagmus is the first sign noticed. Lagotto storage disease is a progressive condition leading to behavioral changes such as restlessness, depression, and aggression. The life expectancy of affected dogs depends on the progression of the disorder and the severity of the signs. Some dogs can live for several years with mild signs, but dogs with severe clinical signs are usually euthanized earlier.

## Mode of Inheritance

- autosomal recessive

## Gene Name

- ATG4D

## Next Steps

Progression of clinical signs in affected dogs should be carefully monitored. Owners should be advised on how to best support their pets as clinical sign worsen, such as avoiding unfamiliar circumstances and offering reassurance. Affected dogs with deteriorating quality of life due to disease progression are usually euthanized on welfare grounds.

## References

Kyöstilä K, Syrjä P, Jagannathan V, Chanddrasekar G, Jokinen TS, Seppälä EH, becker D, Drögemuller M, Dietschi E, Drögemuller C, Lang J, Steffen F, Rohdin C, Jäderlund KH, Lappalainen AK, Hahn K, Wohlsein P, Baumgärtner W, Henke D, Oevermann A, Kere J, Lohi H, Leeb T. A missense change in the ATG4D gene links aberrant autophagy to a neurodegenerative vacuolar storage disease. PLoS Genet 2015 Apr 15;11(4):e1005169.