VETERINARY TECHNICAL DATASHEET

Duchenne or Dystrophin Muscular Dystrophy, (DMD); mutation originally found in Golden Retriever

Mutation Found In :Golden Retriever

Disorder Type

- Muscle

Disease Severity

- Severe

Background

Duchenne or dystrophin muscular dystrophy (DMD) is a severe X-linked disorder that causes muscle degeneration and formation of excess connective tissue. Duchenne muscular dystrophy results from sarcolemma (cell membrane of striated muscle fiber cells) dysfunction which causes degeneration and necrosis of muscle tissue. Due to the X-linked recessive mode of inheritance, mainly males are affected, although some female carriers were suggested to suffer from less severe muscle weakness. The disorder is caused by several different mutations in the dystrophin gene and affected puppies are usually euthanized at a young age because of the severity of the disorder.

Key Signs

- Muscle fibrosis
- High serum creatine kinase concentration
- Spinal curvature
- Crouched posture
- Thick tongue base
- Excessive salivation

Clinical Description

DMD is a progressive condition that eventually leads to muscle fibrosis (formation of excess fibrous connective tissue). First signs of disease, such as a bunny-hopping gait, can be observed in eight to ten weeks old puppies. Affected puppies have a thick tongue base and are unable to open the mouth properly which causes eating difficulties and excess drooling. Duchenne muscular dystrophy is characterized by crouched posture caused by spinal curvature and bending of the back. Serum creatine kinase concentrations can be over 300 times higher than normal levels.

Mode of Inheritance

- X-linked

Gene Name

- Dystrophin

Next Steps

Because of the severity of the clinical signs, affected puppies are usually euthanized at a young age on welfare grounds.

References

Sharp NJH, Kornegay JN, Vancamp SD, Herbstreith MH, Secore SL, Kettle, S, Hung WY, Constantinou CD, Dykstra MJ, Roses AD, Bartlett RJ. An Error in Dystrophin Messenger RNA Processing in Golden Retriever Muscular Dystrophy, an Animal Homologue of Duchenne Muscular Dystrophy. Genomics 13:115-121, 1992.