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

In brief
Duchenne or dystrophin muscular dystrophy (DMD) is a severe X-linked disorder that causes muscle degeneration and formation of excess connective tissue. DMD is characterised by spinal curvature and a crouched posture. 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 euthanised at a young age because of the severity of the disorder.

Clinical overview
Duchenne muscular dystrophy results from dysfunction of sarcolemma (cell membrane of striated muscle fiber cells) which causes degeneration and necrosis of muscle tissue. DMD is a progressive condition that eventually leads to muscle fibrosis (formation of excess fibrous connective tissue). First signs of disease, such as bunny-hopping gate, 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 characterised 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. Affected puppies are usually euthanised at a young age because of the severity of the disorder.

Disease severity
Severe

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

Mode of Inheritance
X-linked Recessive

Results of the genetic test are reported as follows:
- Clear
- Carrier
- At risk

Mutation(s) found in:
Golden Labrador Retriever
Golden Retriever
Goldendoodle
Mixed breed
References

Online database
Online Mendelian Inheritance in Animals, OMIA (http://omia.angis.org.au/). Faculty of Veterinary Science, University of Sydney.

Scientific articles


