

# VETERINARY TECHNICAL DATASHEET

Cavalier King Charles Spaniel Muscular Dystrophy, (CKCS-MD)



Mutation Found In :Cavalier King Charles Spaniel

## Disorder Type

- Muscle

## Disease Severity

- Severe

## Background

Duchenne or dystrophin muscular dystrophy (DMD) is caused by several different mutations in the dystrophin gene; this variant is associated with causing the condition in Cavalier King Charles Spaniels. 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

Duchenne muscular dystrophy results from dysfunction of the sarcolemma (cell membrane of muscle fiber cells) which causes degeneration and necrosis of muscle tissue. It is a progressive condition that eventually leads to muscle fibrosis (formation of excess fibrous connective tissue). First signs of disease, such as 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

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

## References

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