

# VETERINARY TECHNICAL DATASHEET

Spinal Dysraphism



Mutation Found In :Weimaraner

## Disorder Type

- Nervous system

## Disease Severity

- Moderate

## Background

Spinal dysraphism is a malformation of the spinal cord that occurs as a result of abnormal prenatal development of the neural tube. The disorder is encountered in multiple breeds but the genetic cause for spinal dysraphism has only been identified in the Weimaraner. The mode of inheritance is still unclear but an autosomal recessive mode is believed to be likely.

## Key Signs

- "Bunny-hopping" hind limb gait
- Paraparesis
- Crouching stance
- Scoliosis of the lumbar spine
- Kinked tail

## Clinical Description

In hereditary spinal dysraphism, abnormalities are usually located in the lumbar (lower back) region of the spinal cord. Clinical signs usually become evident when puppies begin to walk. Characteristic signs of spinal dysraphism are a "bunny-hopping" hind limb gait, weakness in the hind limbs, and a crouching stance. Affected dogs can also have a kinked tail and abnormal hair patterns on the back and neck as well as scoliosis of the lumbar spine. The condition is not progressive and is not painful.

## Mode of Inheritance

- autosomal recessive

## Gene Name

- NKX2-8

## Next Steps

Though an affected dog may have an abnormal gait and appearance of the lumbar spine, the condition is not expected to markedly negatively impact their quality of life. Therapy is limited to general supportive care. There is no cure.

## References

Safra N, Bassuk AG, Ferguson PJ, Aguilar M, Coulson RL, Thomas N, Hitchens PL, Dickinson PJ, Vernau KM, Wolf ZT, Bannasch DL. Genome-wide association mapping in dogs enables identification of the homeobox gene, NKX2-8, as a genetic component of neural tube defects in humans. PLoS Genet 9:e1003646, 2013.