

# VETERINARY TECHNICAL DATASHEET

Spondylocostal Dysostosis



Mutation Found In :Schnauzer (Miniature)

## Disorder Type

- Skeletal

## Disease Severity

- Severe

## Background

Spondylocostal dysostosis is a severe developmental disorder of the Miniature Schnauzer. The disease leads to stillbirth or death soon after birth and is inherited in an autosomal recessive manner.

## Key Signs

- Stillbirth or death soon after birth
- Short trunk
- Skeletal malformations
- Rib anomalies

## Clinical Description

Spondylocostal dysostosis causes severe malformations of the axial skeleton. Affected puppies are stillborn or die soon after birth. This is presumably caused by impaired respiratory function resulting from truncal shortening and rib fusion. Affected puppies have a comma-like appearance due to reduction of the hindquarters, malformations and fusions of the ribs, and reduced body length. Some affected puppies have other developmental anomalies upon exam.

## Mode of Inheritance

- autosomal recessive

## Gene Name

- HES7

## Next Steps

Humane euthanasia for affected dogs is often elected.

## References

Willet C, Makara M, Reppas G, Tsoukalas G, Malik R, Haase B, Wade C. Canine Disorder Mirrors Human Disease: Exonic Deletion in HES7 Causes Autosomal Recessive Spondylocostal Dysostosis in Miniature Schnauzer Dogs. PLoS ONE 10(2): e0117055, 2015.