

## VETERINARY TECHNICAL DATASHEET

Cleft Lip and Palate with Syndactyly (CLPS); originally found in Nova Scotia Duck Tolling Retriever



Mutation Found In :Nova Scotia Duck Tolling Retriever

### Disorder Type

- Skeletal

### Disease Severity

- Moderate/severe

### Background

A cleft palate (CP) is an abnormal hole in the roof (palate) of the mouth which results in an opening between the nasal passages and the oral cavity through which milk passes when an affected puppy is nursing. Though CP can occur in many breeds, this mutation, found in the Nova Scotia Duck Tolling Retriever is associated with additional birth defects. This more complex form of CP can also include cleft lip and syndactyly (joined digits) in this breed.

### Key Signs

- Cleft palate
- Cleft lip
- Joined digits

### Clinical Description

Cases of this more complex syndrome will exhibit a CP but may also have a cleft lip and evidence of fused digits, as the condition's name suggests. The palate is the roof of the mouth separating the oral cavity and nasal passages. A CP occurs when the two sides of the palate fail to come together and fuse during embryonic development. This forms an opening between the mouth and nasal passages allowing milk to flow into the nasal passages and either spill out of the nostrils while nursing or the puppy may try to gag out the milk from its lower respiratory tract. These feeding difficulties lead to decreased growth and frequent chronic infections with a greatly increased risk of developing aspiration pneumonia.

### Mode of Inheritance

- autosomal recessive

### Gene Name

- ADAMTS20

### Next Steps

Puppies suffering from severe CP are unlikely to survive through puppyhood without proper treatment. A CP can be operated on when a puppy is 3 to 4 months old. Puppies with severe clinical signs may need tube feeding to survive until the operation can be performed.

### References

Wolf ZT, Brand HA, Shaffer JR, Leslie EJ, Arzi B, Willet CE, Cox TC, McHenry T, Narayan N, Feingold E, Wang X, Sliskovic S, Karmi N, Safra N, Sanchez C, Deleyiannis FW, Murray JC, Wade CM, Marazita ML, Bannasch DL. Genome-wide association studies in dogs and humans identify ADAMTS20 as a risk variant for cleft lip and palate. PLoS Genet. Mar 23;11(3):e1005059, 2015.