

# VETERINARY TECHNICAL DATASHEET

Dental Hypomineralization; mutation originally found in Border Collie



Mutation Found In :Border Collie

## Disorder Type

- Dental

## Disease Severity

- Mild/moderate

## Background

Dental hypomineralization is a hereditary dental disease that is caused by abnormal mineralization of teeth during dental development. The disease causes abnormal tooth wear, pulpitis, and tooth loss. The disorder has been described in Border Collies. The genetic defect is estimated to be relatively common within the breed with approximately 11% of the breed population being carriers. The mode of inheritance is autosomal recessive.

## Key Signs

- Abnormal tooth wear
- Pulpitis
- Teeth loss
- Severe tooth hypomineralization

## Clinical Description

Clinical signs include brownish dental discoloration and abnormal wear of teeth. As the teeth wear, the biting surfaces of the teeth darkens, become dark brown in color; the enamel layer may also show a light brown discoloration and appear dull. The disorder causes severe tooth wear leading to pulp exposure, chronic inflammation of the pulp, and pulpal necrosis. Histologically, dentin of affected dogs has an abnormal structure and the enamel can be slightly hypoplastic.

## Mode of Inheritance

- autosomal recessive

## Gene Name

- FAM20C

## Next Steps

Affected dogs require regular dental treatment.

## References

Hytönen M, Arumilli M, Lappalainen A, Owczarek-Lipska M, Jagannathan V, Hundi S, Salmela E, Venta P, Sarkiala E, Jokinen T, Gorgas D, Kere J, Nieminen P, Drögemüller C, Lohi H. Molecular Characterization of Three Canine Models of Human Rare Bone Diseases: Caffey, van den Ende-Gupta, and Raine Syndromes. PLOS Genetics. 2016 May 17;12(5):e1006037.