

VETERINARY TECHNICAL DATASHEET

Dystrophic Epidermolysis Bullosa; mutation originally found in Golden Retriever



Mutation Found In :Golden Retriever

Disorder Type

- Skin

Disease Severity

- Moderate

Background

DEB is characterized by a dysfunctional collagen protein that causes separation of the of the layers within the skin - the epidermis separates from the underlying dermis.

Key Signs

- Blistering of the skin
- Lesions in oral cavity and upper digestive tract
- Growth retardation

Clinical Description

Dystrophic epidermolysis bullosa (DEB) results in fragile skin caused by the dysfunctional collagen protein. Therefore, areas of high friction such as footpads, groin, and oral cavity tend to exhibit the characteristics blisters. Puppies may be smaller than littermates, likely as a result of eating less due to the discomfort this poses.

Mode of Inheritance

- autosomal recessive

Gene Name

- COL7A1_Golden Retriever

Next Steps

Treatment is pain management, supportive care, and symptomatic depending on the severity of the dog's clinical signs.

References

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Palazzi X, Marchal T, Chabanne L, Spadafora A, Magnol JP, Meneguzzi G. Inherited dystrophic epidermolysis bullosa in inbred dogs: A spontaneous animal model for somatic gene therapy. J Invest Dermatol. 115(1):135-137, 2000.