VETERINARY TECHNICAL DATASHEET

Osteogenesis Imperfecta, (OI); mutation originally found in Beagle

Mutation Found In :Beagle

Disorder Type

- Skeletal

Disease Severity

- Severe

Background

Osteogenesis imperfecta (OI) is a severe inherited skeletal disorder encountered in multiple dog breeds, though this mutation was first identified in the Beagle. Osteogenesis imperfecta is characterized by defective collagen resulting in fragile bones and loose joints.

Key Signs

- Bones that break easily
- Weak teeth
- Dentinogenesis imperfecta
- Hyperlaxity

Clinical Description

The clinical signs of osteogenesis imperfecta include pain, factures and lameness due to brittle bones, joint laxity, and brittle, opalescent teeth. Other possible signs are loss of hearing, stunted growth, and blue tinted sclera. The clinical signs are already evident in puppyhood. Bones of affected dogs fracture easily, for example during the course of normal puppy play.

Mode of Inheritance

- autosomal dominant

Gene Name

- COL1A2

Next Steps

Treatment is supportive care and activity restriction to prevent fractures. Affected puppies are typically smaller than littermates and are often euthanized by 3 months of age due to welfare concerns.

References

Campbell BG, Wootton JAM, Macleod JN, Minor RR. Canine COL1A2 mutation resulting in C-terminal truncation of pro-alpha 2(I) and severe osteogenesis imperfecta. Journal of Bone & Mineral Research16:1147-1153, 2001.