VETERINARY TECHNICAL DATASHEET

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

Mutation Found In :Dachshund (Shorthaired), Dachshund (Miniature Shorthaired), Dachshund (Wirehaired), Dachshund (Miniature Wirehaired), Dachshund (Longhaired), Dachshund (Miniature Longhaired)

Disorder Type

- Skeletal

Disease Severity

- Severe

Background

Osteogenesis imperfecta (OI) is a severe skeletal disorder that affects multiple breeds, though this mutation was originally found in the Dachshund, where it is known as Brittle Bone Disease. This disease results in defective collagen, which causes 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, and weak or underdeveloped, slightly pink teeth. Other possible signs are impaired hearing, dwarfism, and blue tinted sclera. Clinical signs are already visible at a young age. Bones of affected dogs fracture easily, for example during the course of normal puppy play.

Mode of Inheritance

- autosomal recessive

Gene Name

- SERPINH1

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

Drögemüller C, Becker D, Brunner A, Haase B, Kircher P, Seeliger F, Fehr M, Baumann U, Lindblad-Toh K, Leeb T. A missense mutation in the SERPINH1 gene in Dachshunds with osteogenesis imperfecta. PLoS Genet 5:e1000579, 2009.