

VETERINARY TECHNICAL DATASHEET

Osteochondrodysplasia; mutation originally found in Miniature Poodle



Mutation Found In :Poodle (Miniature)

Disorder Type

- Skeletal

Disease Severity

- Moderate/severe

Background

Osteochondrodysplasias are inherited disorders of bone and cartilage development. Miniature Poodles suffer from a form of osteochondrodysplasia characterized by stunted growth, misshapen legs, and abnormal movement.

Key Signs

- Stunted growth
- Misshapen limbs
- Abnormal movement
- Abducted hind limbs
- Enlarged joints
- Flattened rib cage
- Shortened and bent long bones
- Deformed paws
- Underbite

Clinical Description

The signs of osteochondrodysplasia can typically be observed in puppies as young as 3 weeks of age. The clinical signs of osteochondrodysplasia in Miniature Poodles include a flattened ribcage, deformed paws, abducted hind limbs, enlarged joints, and an underbite. Long bones of the limbs are shortened and bent. Affected dogs are smaller in size compared to their unaffected littermates. Affected dogs can live for several years, but they often suffer from arthritis caused by misshapen limbs. Abnormal structure of the ribcage can cause breathing difficulties.

Mode of Inheritance

- autosomal recessive

Gene Name

- SLC13A1

Next Steps

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

References

Jackson GC, Mittaz-Crettol L, Taylor JA, Mortier GR, Spranger J, Zabel B, Le Merrer M, Cormier-Daire V, Hall CM, Offiah A, Wright MJ, Savarirayan R, Nishimura G, Ramsden SC, Elles R, Bonafe L, Superti-Furga A, Unger S, Zankl A, Briggs MD. Pseudoachondroplasia and multiple epiphyseal dysplasia: a 7-year comprehensive analysis of the known disease genes identify novel and recurrent mutations and provides an accurate assessment of their relative contribution. Hum Mutat 33:144-57, 2012.

