VETERINARY TECHNICAL DATASHEET

Van den Ende-Gupta Syndrome, (VDEGS)

Mutation Found In :Fox Terrier (Wire)

Disorder Type

- Skeletal

Disease Severity

- Moderate/severe

Background

The skeletal syndrome described in Wire Fox Terriers, also known as Van den Ende-Gupta Syndrome, causes severe skeletal anomalies. The disorder causes multiple skeletal defects with the most common being severe patellar luxation and severe underbite. The genetic defect is estimated to be relatively common within the Wire Fox Terrier breed. In one study, 22% of the control population were carriers of the disorder. The mode of inheritance is autosomal recessive.

Key Signs

- Severe underbite
- Severe patellar luxation
- Other skeletal defects

Clinical Description

The disorder causes multiple skeletal anomalies that are already evident in young puppies. A severe underbite is often the first sign of the disorder. Another characteristic feature of the disorder is severe patellar luxation. Patellar luxation causes alteration in gait, hind-limb-lameness, and "bouncing" of the affected leg. Bilateral patellar luxation can sometimes only be seen as an abnormal gait or a reluctance to move, jump, or climb stairs. The disorder can also cause many other skeletal changes, such as elbow luxation, abnormal structure of the nasal septum, rib changes, spinal changes, and abnormal bone mineralization and ossification. Affected dogs can also have abnormally small eyes and thin sclera. Affected dogs are treated according to their condition and the severity of symptoms. Surgical correction of some of the skeletal abnormalities could reduce the degree of discomfort an affected dog experiences but there is no curative treatment for the disorder.

Mode of Inheritance

- autosomal recessive

Gene Name

- SCARF2

Next Steps

On presentation clinical signs of this disease, a full assessment of the skeletal features should be performed. Surgical correction of some of the bone structures may be possible to improve the body's biomechanics. Arthritis secondary to altered biomechanics later in life is a possibility.

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.