

Van den Ende-Gupta Syndrome, (VDEGS)

In brief

A skeletal syndrome causing severe skeletal anomalies has been described in Wire Fox Terriers. The disorder causes multiple skeletal defects with the most common being severe patellar luxation and severe underbite. The mode of inheritance is autosomal recessive. The genetic defect is estimated to be relatively common within the breed. In one study, 22% of the control population were carriers of the disorder.

Clinical overview

The disorder causes multiple skeletal anomalies that are already evident in young puppies. Severe underbite is often noticed as 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 abnormal gait, 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.

References

Online database

Online Mendelian Inheritance in Animals, OMIA (<http://omia.angis.org.au/>). Faculty of Veterinary Science, University of Sydney; OMIA001033-9615.

Scientific articles

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 | DOI:10.1371/journal.pgen.1006037.

Disease severity

Considerable



Clinical signs

- Severe underbite
- Severe patellar luxation
- Other skeletal defects

Mode of Inheritance

Autosomal Recessive

Results of the genetic test are reported as follows:

- Clear
-  Carrier
-  At risk

Mutation(s) found in:

Fox Terrier - Wire