GENOMIA
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Result certificate #012345

Detection of c.865_866delTC variant in SCARF2 gene causing Van den Ende-Gupta in Wirehaired Fox Terriers

Customer: Jan Novák, Dlouhá 1, 30000 Plzeň, Czech Republic
Sample:
Sample: 08-12345
Date received: 25.11.2008
Sample type: blood

Information provided by the customer
Name: Lassie DEMO
Breed: ---
Tattoo number: 1392013
Microchip: 123 456 789 012 345
Reg. number: REGQ12345
Date of birth: 31.12.1909
Sex: female
Date of sampling: 25.11.2008
The identity of the animal has been checked.

Result: Mutation was not detected (N/N)

Legend: N/N = wild-type genotype. N/P = carrier of the mutation. P/P = mutated genotype (individual will be most probably affected with the disease). (N = negative, P = positive)

Explanation

Presence or absence of c.865_866delTC in SCARF2 gene causing VDEGS in Wirehaired Fox Terrier was tested. The Van den Ende-Gupta Syndrome causes severe skeletal anomalies that are already evident in young puppies. Other characteristic features of the disorder are severe patellar luxation and overshot (distinctly shortened maxilla) that often is the first sign of this disease. The disorder can also cause elbow luxation, knee-joint swelling and abnormal structure of the nasal septum.

Variant c.865_866delTC that causes VDEGS is inherited as an autosomal recessive trait. That means the disease affects dogs with P/P genotype only. The dogs with N/P genotype are considered carriers of the disease (heterozygotes). In offspring of two heterozygous animals following genotype distribution can be expected: 25 % N/N, 25 % P/P and 50 % N/P.

Method: SOP171-VDEGS, fragment analysis

Date of issue: 30.11.2008
Date of testing: 25.11.2008 - 30.11.2008
Approved by: Mgr. Martina Šafrová, Laboratory Manager

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