

Detection of inversion in ADAMTS17 gene
causing POAG in Petit Basset Griffon
Vendéen

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 inversion in ADAMTS17 gene causing primary open angle glaucoma (POAG) in Petit Basset Griffon Vendéen was tested. The initial clinical signs occur in 3 to 4-year-old dogs. The so-called aphakic crescent (half-moon) connected with lens subluxation occurs in approximately one third of the affected dogs. In the late stage of the disease, globe enlargement develops. Retina degeneration and cupping deformation of the optic papilla can be seen only in late disease. Pain is not a feature of this type of disease and so the owners of the affected dogs become aware of the presence of POAG when either the globe enlargement or a vision problem becomes noticeable.

Mutation that causes POAG in Petit Basset Griffon Vendéen is inherited autosomally recessively which means that the disease develops only in those dogs who inherit mutated allele from both parents; 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-POAG-PBGV, 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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