GENOMIA
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Result certificate #012345

Detection of SINE insertion in FAM161A gene causing PRA3 disease in Tibetan Terriers and Tibetan Spaniels

Customer: Jan Novák, Dlouhá 1, 30000 Plzeň, Czech Republic
Sample:
Sample: 21-12345
Date received: 01.02.2021
Sample type: blood

Information provided by the customer
Name: Lassie DEMO
Breed: Plemeno
Tattoo number: 1392013
Microchip: 123 456 789 012 345
Reg. number: REGQ12345
Date of birth: 1.1.2020
Sex: female
Date of sampling: 01.02.2021
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 SINE insertion in FAM161A gene causing Progressive Retinal Atrophy type 3 (PRA3) in Tibetan Terriers and Tibetan Spaniels was tested. The Progressive Retinal Atrophy is inherited retinal diseases characterized by degeneration of photoreceptor cells. The disease starts with rod degeneration leading to night blindness and loss of spatial vision. The first symptoms are worse coordination in darkness and bumping into objects. The subsequent progressive degeneration of cones leads to cataract and complete blindness. The onset of PRA3 is relatively late at approximately 5 years of age.

Mutation that causes PRA3 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), they are healthy but they can transmit the mutation on their offspring. In offspring of two heterozygous animals following genotype distribution can be expected: 25 % N/N (healthy non-carriers), 50 % N/P (healthy carriers) and 25 % P/P (affected).

Method: SOP171-PRA3, fragment analysis

Date of issue: 06.02.2021
Date of testing: 01.02.2021 - 06.02.2021
Approved by: Mgr. Martina Šafrová, Laboratory Manager

verified and certified document
GENOMIA genetic laboratory

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