

Result certificate #012345

Detection of c.155G>C mutation in FAM83G gene causing HFH in Irish Terriers and Kromfohrländers

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 c.155G>C mutation in FAM83G gene causing palmoplantar hyperkeratosis (HFH) in Irish Terriers and Kromfohrländers was tested. HFH leads to cornification of the skin, particularly on the pads of the feet and the nose; it can also occur on the elbows and other parts of the body where it causes reddening of the skin, scaling and scabbing, which tend to be sites of entry for infection, cracking, bleeding and suppuration. Clinical signs are evident in puppies as young as 4-5 months of age.

Mutation that causes HFH 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: SOPAgriseq_canine, ngs, accredited method

Date of issue: 06.02.2021

Date of testing: 01.02.2021 - 06.02.2021

Approved by: Mgr. Martina Šafrová, Laboratory Manager



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