GENOMIA
genetic laboratory

Result certificate #012345

Detection of g.52737379delC mutation in NIPAL4 gene causing congenital ichthyosis in American bulldog and American Bully

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 g.52737379delC mutation in NIPAL4 gene causing congenital ichthyosis in American bulldog and American Bully was tested. Congenital ichthyosis is an inherited skin disorder. In the affected dogs, the outermost layer of the skin is not formed correctly. The typical signs are recognizable as early as 1 to 2 weeks of age. Compared to normal littermates, the affected dogs have a dishevelled fur and the glabrous skin of the abdomen is reddened. Complications due to infection caused by fungi of Malassezia genus. Together with the growth of the fungi, the inflammation of acoustic duct and itching infection of skin folds or paws develop and even hyperkeratosis may develop in adult dogs.

Mutation that causes congenital ichthyosis 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 (healthy non-carriers), 25 % P/P (affected), and 50 % N/P (healthy carriers).

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

verified and certified document GENOMIA genetic laboratory

Genomia is accredited in compliance with ISO/IEC 17025:2018 under #1549
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