GENOMIA
genetic laboratory

Result certificate #012345

Detection of c.143G>C mutation in COL11A2 gene causing Skeletal Dysplasia 2 in Labrador Retrievers

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.

SAMPLE

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.143G>C mutation in COL11A2 gene causing Skeletal Dysplasia 2 (SD2) in Labrador Retrievers was tested. The dogs affected by SD2 have typical short legs and normal length and width of the body. Male affected dogs mostly had shoulder heights of less than 55 cm and female affected dogs mostly had shoulder heights of less than 50 cm, while the international breed standards (FCI) call for 56-57 cm in males and 54-56 cm in females. No secondary health problems have been observed. The hearing and the eyesight of the dogs are not affected und they have no other problems with joints.

Mutation that causes SD2 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.

The test does not exclude presence of mutation causing different type of skeletal dysplasia.

Method: SOPAgriseq_canine, ngs

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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