

Result certificate #012345

Detection of c.10_11insG mutation in COL9A3 gene causing Oculoskeletal dysplasia 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.

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.10_11insG mutation in COL9A3 gene causing Oculoskeletal dysplasia (OSD) in Labrador Retrievers was tested. Affected dogs have shorter forelimbs and their radius and ulna forelimb bones exhibit morphological abnormalities. Other symptoms include ocular defects such as cataracts, retinal detachment or vitreous dysplasia. Beside the recessive homozygotes (dogs carrying two mutated alleles of COL9A3 gene - each allele inherited from one parent - test result P/P), this disease affects even heterozygotes (dogs carrying only one mutated allele - test result N/P), however the homozygotes do not exhibit any skeletal malformations. Dogs with N/N result are not in risk of OSD.

Method: SOP172-OSD1, direct DNA sequencing

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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