

Result certificate #078861

Detection of c.289G>T COL6A1 gene causing muscular dystrophy in Landseer

Sample

Sample: 16-14432 Name: Fame Alvemia Breed: Landseer (European Continental type) Microchip: 203 098 100 338 235 Reg. number: CMKU/LS/665/13 Date of birth: 18.11.2013 Sex: female Date received: 02.06.2016 Sample type: buccal swab The identity of the animal has been checked by MVDr. Petr Pohořalý Customer Renata Cepková U Kostela 166 28141 Ratboř Czech Republic

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.289G>T mutation in COL6A1 gene causing muscular dystrophy in Landseer was tested. Muscular dystrophy is an inherited disease causing progressive muscle weakness.

Mutation that causes muscular dystrophy in Landseer 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: SOP172-MDL, direct DNA sequencing

Report date: 13.06.2016 Responsible person: Mgr. Martina Šafrová, Laboratory Manager

Jafurra

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