GENOMIA
genetic laboratory

Result certificate #012345

Detection of c.831dup mutation in TNR gene causing DAS in Weimaraners

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.831dup mutation in TNR gene causing paroxysmal dystonia-ataxia syndrome (DAS) in Weimaraners was tested. The syndrome combines the symptoms of dystonia (increased muscle contractions leading to abnormal posture, limb twisting and stiffness) and ataxia (movement coordination disorder) with hypermetria (overshooting movements). Symptoms occur in episodes that occur several times a day for 5 to 15 minutes, often in response to emotional arousal or physical activity, and may result in collapse. The age of onset of clinical symptoms is 3 to 7 months.

Mutation that causes DAS 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: SOP188-MPS-canine, MPS, 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

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