GENOMIA
genetic laboratory

Result certificate #012345

Detection of c.1333_1338dup mutation in PK-LR gene causing PK deficiency in West Highland White Terriers

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.1333_1338dup mutation in PK-LR gene causing deficiency of pyruvate kinase (PK deficiency) in West Highland White Terriers was tested. The deficiency of this enzyme causes insufficient production of ATP, which results in erythrocyte lysis or their premature destruction in the spleen. The increased destruction of erythrocytes expresses itself clinically as anaemia.

Mutation that causes PK deficiency in west highland white terriers 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.

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

Genomia is accredited in compliance with ISO/IEC 17025:2018 under #1549
Genomia s.r.o, Republikánská 6, 31200 Plzeň, Czech Republic
www.genomia.cz, laborator@genomia.cz, tel: +420 373 749 999

ilac-MRA L 1549

Report verification code is: 12AB-CD34-GENO-MIA0-EFGH. You can verify report online at www.genomia.cz
Without a written consent by the lab, the report must not be reproduced unless as a whole.
The result refers only to the tested sample, as received. Genomia is not responsible for the accuracy of the information provided by the customer.