GENOMIA
genetic laboratory

Result certificate #012345

Detection of mutations in LHX3 gene causing pituitary dwarfism in some dog breeds

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: 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 mutations c.622-37-31del a c.545_547dupACA in gene LHX3 causing pituitary dwarfism in German shepherds, Saarloos Wolfdogs, Czechoslovakian Wolfdogs and Tibetan Terriers were tested. The disease is characterised by degeneration of hypophysis (pituitary) resulting in deficiency of pituitary hormones. Common clinical manifestations are growth retardation, retention of secondary hairs (puppy coat) with signs of alopecia. The affected dogs can have normal size during the first weeks of their lives. Between the 3rd and the 4th month of age the differences are already evident.

Mutations that cause pituitary dwarfism are inherited as an autosomal recessive trait. That means the disease affects dogs with P/P (positive/positive) genotype only. The dogs with N/P (negative/positive) 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: SOP171-dwarfism, fragment analysis

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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