GENOMIA
genetic laboratory

Result certificate #073052:

Detection of c.622-37-31del and c.545_547dupACA mutations in LHX3 gene causing pituitary dwarfism in German shepherds, Saarloos and Czechoslovakian Wolfdogs by fragment analysis

Sample
Sample: 16-00327
Name: Ianek Srdcerváč
Breed: Czechoslovakian Wolfdog
Microchip: 203 098 100 387 850
Reg. number: CMKU/CSV/4406/15
Date of birth: 27.11.2015
Sex: male
Date received: 04.01.2016
Sample type: blood
The identity of the animal has been checked.

Customer
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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 mutations c.622-37-31del a c.545_547dupACA in gene LHX3 causing pituitary dwarfism in German shepherds, Saarloos Wolfdogs and Czechoslovakian Wolfdogs 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: SOP160

Report date: 06.01.2016
Responsible person: Mgr. Martina Šafrová, Laboratory Manager

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