

Detection of c.2083C>T mutation in ITGA10 gene causing Canine Chondrodysplasia in Norwegian Elkhound and Karelian Bear Dog

Customer: Jan Novák, Dlouhá 1, 30000 Plzeň, Czech Republic

Sample:

Sample: 08-12345

Date received: 25.11.2008

Sample type: blood

Information provided by the customer

Name: Lassie DEMO

Breed: ---

Tattoo number: 1392013

Microchip: 123 456 789 012 345

Reg. number: REGQ12345

Date of birth: 31.12.1909

Sex: female

Date of sampling: 25.11.2008

The identity of the animal has been checked.

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.2083C>T mutation in ITGA10 gene causing Canine Chondrodysplasia in Norwegian Elkhound and Karelian Bear Dog was tested. Chondrodysplasia is a genetic disorder of cartilage and bone development. The affected dogs show shortened limbs compared to healthy dogs. Further, it may occur skeletal abnormalities such as shorter protruding toes on the outer side of the front paws, bowed forelimbs, shortened vertebral bodies and substantially delayed ossification of carpal bones.

Mutation that causes chondrodysplasia 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-chondrodysplasia, direct DNA sequencing

Date of issue: 30.11.2008

Date of testing: 25.11.2008 - 30.11.2008

Approved by: Mgr. Martina Šafrová, Laboratory Manager



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