

Result certificate #012345

Detection of c.1775T>C mutation in COL11A1 gene causing Multiocular defects in Bobtails

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: Mutation was not detected (N/N)

Explanation

Presence or absence of c.1775T>C mutation in COL11A1 gene was tested. It causes multiple eye defects for example cataracts, bilateral vitreous degeneration, macrophthalmia, changes in lens shape, retinal detachment or inflammatory deposits on the eye.

Mutation that cause multiple eye defects is inherited as an autosomal dominant trait. Only one copy of the mutated gene is sufficient for development of clinical signs (result N/P (negative / positive) or P/P (positive / positive)). In case of affected animal the risk of transfer to the offsprings is 50%.

Method: SOP188-MPS-canine, MPS

Date of issue: 06.02.2021

Date of testing: 01.02.2021 - 06.02.2021

Approved by: Mgr. Martina Šafrová, Laboratory Manager



Genomia s.r.o, Republikánská 6, 31200 Plzeň, Czech Republic www.genomia.cz, laborator@genomia.cz, tel: +420 373 749 999