GENOMIA
genetic laboratory

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.590G>A mutation in OLFML3 gene related with Goniodysgenesis and Glaucoma in Border Collies was tested. Goniodysgenesis is a hereditary disorder characterized by development abnormalities of anterior chamber. Due to abnormal development of intraocular fluid egress channels inside the eye the iridocorneal angle, through which the excessive chamber fluid is filtered and drained, get narrower or closed. Goniodysgenesis is significantly associated with the glaucoma and blindness.

Goniodysgenesis occurs in severe and mild forms. Severe goniodysgenesis potentially leading to glaucoma is connected with homozygosis for c.590A allele of OLFML3-gene which indicates autosomal recessive mode of inheritance. The vast majority of dogs with severe goniodysgenesis and glaucoma are homozygous for the mutation mentioned, however there are some cases of heterozygotes affected with this disease. The exact mode of inheritance has not been elucidated yet.

Result options: N/N healthy dog, N/P carrier of disposition to goniodysgenesis, P/P dog in risk of goniodysgenesis development.

Method: SOPAgriseq_canine, ngs, accredited method

Date of issue: 06.02.2021
Date of testing: 01.02.2021 - 06.02.2021
Approved by: Mgr. Martina Šafrová, Laboratory Manager