

Detection of c.590G>A mutation in OLFML3 gene related with Goniodysgenesis and Glaucoma in Border Collies

Sample

Sample: 18-40829
Name: Flower of Old Hill Not Just a Banana
Breed: Border Collie
Microchip: 900108000883765
Reg. number: CMKU/BOC/7636/-14/14/15
Date of birth: 14.03.2014
Sex: female
Date received: 28.05.2018
Sample type: buccal swab
The identity of the animal has been checked by Mgr. Markéta Dajbychová, Genomia

Customer

Martina Paulino
Mezi Rolemi 11
158 00 Praha 5
Czech Republic

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: SOP172, direct DNA sequencing

Report date: 05.06.2018

Responsible person: Mgr. Martina Šafrová, Laboratory Manager



Genomia s.r.o, Janáčkova 51, 32300 Plzeň, Czech Republic
www.genomia.cz, laborator@genomia.cz, tel: +420 373 749 999