

Customer: Dr. Josette Debrincat, 20, Animal Doctors, Guze Bajada, HMR 2131 Hamrun, Malta**Sample:**

Sample: 24-26623

Date received: 01.10.2024

Sample type: blood

Information provided by the customer

Name: Scott Dell'Antica Contrada**Breed:** Border Collie

Microchip: 380 206 044 889 554

Reg. number: MKC BOR/14/2024

Date of birth: 18/12/2022

Sex: male

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: 13.10.2024

Date of testing: 01.10.2024 - 13.10.2024

Approved by: Mgr. Martina Šafrová, Laboratory Manager



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