Goniodysgenesis & Glaucoma in the Border Collie

Affected breeds: Border Collie

Goniodysgenesis is a developmental defect at the front of the eye which is often associated with glaucoma and blindness.

Goniodysgenesis is caused by a recessive genetic mutation. This means that dogs which carry the mutation ("CARRIERS") are normal but will pass the mutation on to an average of 50% of their offspring. Puppies which inherit two copies of the mutation will develop Goniodysgenesis, and will often also be affected by glaucoma ("AFFECTED").

Since the mutation is fairly common, it is recommended that breeders continue to include carriers in their breeding plans so as to keep the gene pool as wide as possible.

This test is particularly useful for breeders:

- To identify carriers among their breeding stock so that they can avoid CARRIER X CARRIER mating combinations which would risk AFFECTED puppies.
- To conclusively confirm Goniodysgenesis in an affected dog

This test will be reported as:

CLEAR : no evidence of the Goniodysgenesis mutation
CARRIER : carries one copy of the defect, which will be passed to 50% of offspring
AFFECTED : carries two copies of the defect, causing Goniodysgenesis, and often glaucoma

The genetic status of dogs can be used to predict breeding outcomes when different combinations are mated:

CLEAR X CLEAR = 100% CLEAR
CARRIER X CLEAR = 50% CARRIER, 50% CLEAR
CARRIER X CARRIER = 25% AFFECTED, 50% CARRIER, 25% CLEAR

References

Pugh CA, Farrell LL, Carlisle AJ, et al. Arginine to Glutamine Variant in Olfactomedin Like 3 (OLFML3) Is a Candidate for Severe Goniodysgenesis and Glaucoma in the Border Collie Dog Breed. G3 (Bethesda). 2019;9(3):943–954. Published 2019 Mar 7. doi:10.1534/g3.118.200944

Oliver, JAC, Wright, H, Massidda, PA, Burmeister, LM, Mellersh, CS. A variant in OLFML3 is associated with pectinate ligament abnormality and primary closed-angle glaucoma in Border Collies from the United Kingdom. Vet Ophthalmol. 2020; 23: 25–36.