

## Border Collie goniodysgenesis and glaucoma project

If you submitted a sample from your dog for the research project we can supply you with preliminary results of testing our candidate gene, but please note that these results are still in development. They have not yet been reviewed or validated and cannot be considered definite. We are not a certified testing laboratory. Therefore the results are supplied for information and interest only. They are experimental in nature, do **not** constitute a diagnostic test and must **not** be used for diagnosis or treatment.

### Disclaimer

We make no representations of any kind, either expressed or implied, of accuracy or fitness for a particular purpose of these results and shall have no liability to you in relation to the supply of the results to you or use of or reliance on the results by you or any third party. Any result presented here should be validated by a certified commercial test once this becomes available.

You must be the registered owner of the dog to receive the results. By signing and submitting this form you declare that you are the registered owner of the dog for which you are requesting results. Please also supply official confirmation that you are the owner (for example Kennel Club registration papers). If ownership has been transferred please explain why the registration is in a different name.

To validate this research it is important to have accurate clinical and pedigree information. For some dogs we do not have complete information and we have not been able to use those samples for our analysis. Therefore could you please also include any information you have from gonioscopy testing for your dog (for example BVA report, or information verbally given to you by the vet). Could you also send details of the dog's pedigree, at least parents and grandparents, so that we can be sure of the family relationships.

*Please note that we have not been able to test all dogs submitted to the project. If your dog's test was not successful we would advise you to access a commercial test once it becomes available.*

Complete the details on **both** pages below. You can enter the details in the forms and the relevant information will be copied to appropriate boxes. Your name written in the signature box will be taken as a signature. Save the completed document as a .pdf. Check that you have included all the requested paperwork and send by email to:

**bc.research@outlook.com**

Results will be sent to you by email so please ensure that you supply a valid email address.

### Check list

1. Completed "Request for Results Form"
2. Completed "Results Form"
3. Proof of ownership
4. Eye test results
5. Pedigree (complete on the "Requests for Results Form" or send scan of printed pedigree)

**Request for Results Form**

**To: Border Collie Goniodysgenesis and Glaucoma Research Project**

Please supply me with the genetic test results from the goniodysgenesis project for:

..... (Dog's registered name)

I understand that these results are from a research project and are **not** suitable for use in diagnosis or treatment of goniodysgenesis and should not be relied upon for any purpose.

I have included any test results from gonioscopy testing and official confirmation that I am the owner of ..... (Dog's registered name)

I have attached the pedigree **OR** completed the pedigree details below for

..... (Dog's registered name)

Sire's Sire.....

Sire .....

Sire's Dam .....

Dam's Sire .....

Dam .....

Dam's Dam .....

My contact details are:

Name .....

E-mail address .....

Signature .....

Date .....

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### Results Form

Dog's registered name .....

Owner's name .....

#### Genotyping results (FOR OFFICE USE: to be completed by bc.research)

The genotype detected for your dog is :

AA Low risk, not a carrier	AB Carrier	BB High risk
Most dogs with this genotype did not have severe goniodysgenesis and none had glaucoma. Some dogs diagnosed with mild to moderate goniodysgenesis or with ungraded goniodysgenesis had this genotype.	Most dogs with this genotype did not have severe goniodysgenesis and none had glaucoma. Some dogs diagnosed with mild to moderate goniodysgenesis or with ungraded goniodysgenesis had this genotype.	Most dogs with this genotype had severe goniodysgenesis and many have progressed to glaucoma. Several ungraded dogs also had this genotype. However two dogs with severe disease had other genotypes and two apparently unaffected dogs had this genotype.

Genotype	Status			
	Glaucoma	Severe	Mild/mod/ungraded	Unaffected
AA	0%	0%	35%	48.5%
AB	0%	14%	24%	48.5%
BB	100%	86%	41%	3%
Number examined	9	14	29	60

#### Notes

1. We believe the BB genotype correlates strongly with severe goniodysgenesis with the risk of glaucoma. However, we do not yet know what determines progression to glaucoma in dogs with BB genotype.
2. Glaucoma is found in many dog breeds and other species. It develops at different ages and is associated with multiple different genes. Therefore a dog with any of the genotypes (including AA) may still develop glaucoma from other causes.
3. Dogs with low grade goniodysgenesis were generally AB or AA (ie **not** the high risk genotype). There may be mutations in other genes that contribute to this lower level goniodysgenesis. We do not know what these genes are or whether these mutations can also lead to glaucoma. However, all glaucoma cases tested here were BB.