

# Genetics of goniodysgenesis and glaucoma in the Border Collie

Roslin Institute Update - 2 Feb 2017

We have currently been able to perform genetic analysis on 92 animals. Of these, 45 have at some time been given a diagnosis of goniodysgenesis. We have focussed our attention on 11 of these with a diagnosis of **severe goniodysgenesis** and seven who have **lost eyes due to glaucoma**. We are comparing these with 47 animals who have been **tested clear**, some on more than one occasion.

We have two approaches to finding the gene or genes responsible for this problem.

- Firstly, we do a scan of all the DNA (the whole genome) looking at about 100,000 sites where there is known to be variability. This enabled us to locate a genetic region that seems to be associated with severe goniodysgenesis. All but one of the severely affected dogs have the same pattern in this region. Some of the ones with lower grade or ungraded goniodysgenesis also have this pattern but some do not. This may be for a number of reasons.
  - It may relate to the diagnostic uncertainty.
  - Mild goniodysgenesis may in fact be normal variation for this breed and unrelated to the genetic factor causing the serious form.
  - Different veterinary ophthalmologists may record the results of the gonioscopy differently.
  - There may be two genetic regions, one for severe goniodysgenesis/glaucoma and one for mild goniodysgenesis.

We have also found that a couple of the clear dogs have the same genetic pattern. This does not mean that they are likely to develop glaucoma; they may not have the DNA change that is actually causing the problem.

- Secondly, to find this causal DNA change we have done the full genetic sequence of 10 dogs, six with severe goniodysgenesis or glaucoma and four unaffected animals. This will help us to narrow down the region where the problem is likely to lie. Within this region there is no obvious gene or DNA change that would be a strong candidate, and we are continuing to examine this region in detail to test whether any of the changes we see could be involved.

We understand the Kennel Club is currently advising that goniodysgenesis in the Border Collie is a recessive condition. Our results do not rule this out; we will be more comfortable talking about the mode of inheritance once we have discovered the precise DNA change that is associated. This would also allow us to develop a test.

To help us work out the genetic basis, at present the most valuable samples are from animals who have a diagnosis of severe goniodysgenesis (especially if followed by glaucoma), and their close relatives who have been tested clear, or who have reached an advanced age without developing glaucoma.

**Any information that we release or publish will be anonymous and the pedigrees will be disguised so that the animals or kennels can't be identified.**

We realise that this is a difficult time for Border Collie breeders and owners and we are very grateful to everyone who has provided us with samples for DNA extraction and information on pedigrees. It all helps towards finding a test and eliminating this distressing condition.