



Canine Genetics Progress Report

Breed: Large Munsterlander

Condition: Hereditary Cataract

Date: 01.07.2008

Recent / Current Funding:

Funding Body: Kennel Club Charitable Trust

Amount: £250,000

Start Date: 01.03.2006, duration 36 months

The above grant is specifically to study Hereditary Cataract (HC) and Progressive Retinal Atrophy (PRA) in the Golden Retriever, the Tibetan Spaniel and the American Cocker Spaniel but it is our intention to also study any breeds for which we can collect sufficient samples.

Sample Collection

Sample Collection has progressed well over the past year, although it has slowed a little since the last report. We have now collected DNA samples from 127 Large Munsterlanders. Of these samples 21 are from dogs affected with hereditary cataract (HC) and two more are from dogs that may be affected with HC. We also have samples from several dogs with opacities of the lens, but not necessarily inherited forms. These samples from dogs with slightly unusual forms of cataract will contribute to the later stages of the research, and will help understand which forms of cataract are inherited, but they will be classed as ‘unknown’ (as opposed to affected or unaffected) for the early stages of the research. The sample collection includes samples from many of the parents and littermates of affected dogs, which adds considerable strength to the sample collection.

In summary, sample collection has gone very well and we now have the minimum number of affected dogs that are required to initiate DNA studies.

Studies In Other Breeds

The AHT has initiated DNA analyses of other breeds with HC, for which substantial sample collections already exist at the AHT, and relevant findings from those studies will be applied to the Large Munsterlander, as and when they become available. These studies involve whole-genome scans to identify regions of the genome that contain markers associated with HC. We are analysing over 22,000 genetic markers in samples from around 300 dogs, so these experiments are extensive and the analysis involved is considerable. It is possible the Large Munsterlander shares an HC mutation with other breed(s) that develop similar

types of hereditary cataracts (the Golden retriever for example) and if that is the case DNA tests we develop, based on our research in these other breeds, will directly benefit the Large Munsterlander.

We have had some preliminary results that suggest at least one region of the genome that is associated with HC in Golden retrievers. We are in the process of following up that result now. Once the result has been confirmed in Golden retrievers we will genotype a number of affected Large Munsterlanders with genetic markers from the same region, to see if the same genomic region is associated with HC in the Large Munsterlander. If it is then we will combine the results from the two breeds to narrow the region, identify candidate genes and finally identify the mutation responsible for HC. We anticipate having completed that initial work by October, and will know by then whether or not we have identified a region associated with HC in the Large Munsterlander or if we will need to progress to a whole genome scan.

In the previous reported we noted that the DNA yield from the Large Munsterlanders had been lower in general than for other breeds we are studying. As we have extracted DNA from many dogs now, all from swabs taken by many different owners, it is unlikely it is an 'owner-specific' effect and is more likely to be a breed-specific effect (perhaps Large Munsterlanders produce more or less saliva than other breeds, or are simply more active and dislike having swabs taken!). If we need to progress to a whole-genome scan we will need more DNA from some of the dogs, both affected and unaffected, and will therefore be contacting some owners in the near future to request additional DNA samples from their dogs; this will ensure we have sufficient DNA to conclude the studies. This by no means implies that the DNA samples we already have are not going to be useful, nor will they lead to inaccurate result; we are merely trying to ensure we have sufficient DNA from all the crucial dogs to ensure success.

Thank You

The AHT would like to thank the very many Munsterlander owners and breeders who have offered us samples and information about their dogs. Without this kind of co-operation we would not be in a position to investigate HC in this breed; with your continued co-operation we will continue to make progress.

We would also like to thank everybody who has raised and donated money for the research effort at the Animal Health Trust. As a charity we rely heavily on donations and they really do make a significant difference to the work we can do.