

Miniature & Bull Terrier Complaint and Genealogy (mBTcg)

Health - English Only => Eye issues => Topic started by: ChaosEnergy on April 21, 2008, 12:47:34 PM

Title: **AHT: Canine Genetics Progress Report Oct 2007**

Post by: **ChaosEnergy on April 21, 2008, 12:47:34 PM**

Source: <http://www.lancashireheelers.org/AHTupdate02.php>

Breed: Lancashire Heelers
Condition: Primary Lens Luxation (PLL)
Date: 01.10.2007

Recent / Current Funding:

1. Funding Body: Kennel Club Health Foundation Fund
Amount: £83,281 (including £8000 from Miniature Bull Terrier Breed Club) this grant was to study four inherited conditions, one of which was PLL Start Date: March '03, 24 months
2. Funding Body: Kennel Club Health Foundation Fund
Amount: £49,823 (including £2000 from Lancashire Heeler Breed Club) Start Date: March '05, 24 months
3. Funding Body: Canine Health Foundation (American Kennel Club)
Amount: \$9586
Start Date: January '05, 12 months
4. Funding Body: Canine Health Foundation (American Kennel Club)
Amount: \$12927
Start Date: February '07, 12 months

The Primary Lens Luxation research project is currently a collaboration between Cathryn Mellersh (AHT), David Sargan (University of Cambridge) and David Gould (Davies Veterinary Specialists).

Progress Update

During 2006 we identified which one of the dog's 38 chromosomes harbours the PLL mutation and by July 2007 we had reduced the search to approximately 600,000 of the 3,000,000,000 letters or nucleotides of DNA that make up the dog genome.

Analysis of DNA samples from different breeds indicated the same region of the same chromosome was associated with PLL in several breeds, including the Miniature Bull Terrier, the Tibetan Terrier, the Lancashire Heeler and the Jack Russell Terrier.

Over the last three months we have continued to reduce the size of the region in which we know the PLL mutation is located –the so- called `PLL critical region'. We have done this by analysing additional closely spaced genetics markers from within the PLL critical region in large number of dogs affected with PLL. We know PLL is a recessive condition, so all dogs affected with PLL will carry two copies of the PLL mutation (i.e. be homozygous for the mutation). Likewise, we expect affected dogs to also carry two identical copies of, or be homozygous for, genetic markers located very close to the mutation.

If an affected dog is heterozygous for (i.e. carries different copies of) a given genetic marker then we know that marker must be located some way from the mutation and therefore be outside the PLL critical region.

Focusing on approximately 150 dogs affected with PLL we have been able to narrow the PLL region to approximately 300,000 nucleotides. We are continuing to narrow the PLL region. We have identified a gene within the region that is a very good candidate for PLL that we would now like to sequence (i.e. read letter by letter). Unfortunately the sequence and structure of the DNA in this region makes it technically challenging to sequence so we are currently investigating different methods of analysing the DNA surrounding this gene. Once we have found a way in which the DNA can be successfully analysed we will sequence the candidate gene to see if it harbours the PLL mutation.

Since the last report we have had a dedicated member of staff working on this project, and that continues to be the situation. In addition, the principal investigators meet on a regular basis, to monitor progress and discuss results. Identifying the mutation responsible for this condition continues to be a major focus for all concerned with this project.

Sample Collection

The research has progressed sufficiently well that we are now only targeting samples from dogs, of any breed, that are affected with PLL. Samples from additional affected dogs will continue to play a valuable role in the research right up until the point at which we find the mutation and can develop a DNA test. We would also like to thank everybody who has made a financial donation to support our research studies. As a charity the AHT relies heavily on donations, whilst all research performed at the University of Cambridge is also funded solely through external donations and competitive grants, and not through support from the higher education funding system. All donations to support our research are truly appreciated by both organisations.