

## CANINE FUCOSIDOSIS

The Aims of this document are two-fold:

- To provide details of the disease and its outcomes
- To provide details of the procedure to be followed for requesting Fucosidosis Testing

**Canine Fucosidosis** is a severe, progressive and ultimately fatal disease that affects the English Springer Spaniel. It is an inherited condition in the breed and has been reported in the UK, Europe and Australia. It is inherited through an autosomal recessive trait.

The disease affects young adults, usually between 18 months and 4 years of age, and is characterised by nervous signs that progress over a period of several months. These signs include inco-ordination and ataxia (loss of control of movement), change in temperament, loss of learned behaviour, loss of balance, apparent deafness, visual impairment and varying degrees of depression. The inco-ordination and ataxia affects all four legs and is mostly evident when affected animals are walking on slippery surfaces or attempt more complicated movements such as turning. In addition, affected dogs lose weight and may suffer from swallowing difficulties and sometimes regurgitation of food.

The disease is caused by the absence of an enzyme called alpha-L-fucosidase. This enzyme is one of many required to break down complex compounds into simple molecules that the body can use. When the enzyme is absent, the pathway is blocked and the more complex compounds build up in the cells of the affected animal. These substances accumulate in lymph nodes, liver, pancreas, kidney, lungs and bone marrow, but it is the accumulation in the brain and peripheral nerves that is most important since it interferes with normal function, giving rise to the clinical signs described above, eventually resulting in death.

Fortunately, the defective gene responsible for this disease was identified in the mid 1990's. Since 1997 a DNA blood test has been available through the Animal Health Trust in Newmarket, making it possible to determine with complete accuracy the genetic status of each dog as either a CLEAR, CARRIER or AFFECTED. Prior to that, since around 1983, we had to rely on examination of blood samples to detect the activity of the enzyme, alpha-L-fucosidase, in both plasma and extracted white blood cells (leukocytes). Although these earlier enzyme blood tests made it possible to diagnose affected animals and those which were clearly either CARRIERS or NORMAL, there was a degree of overlap between normals and carriers (the 'grey' area), and problems could therefore potentially arise with those dogs whose results lay within this 'grey' borderline area. For these dogs (if still in the breeding pool) or their descendants, DNA testing would provide reassurance and prevent further uncertainty.

Using the results of the original enzyme testing and current DNA testing, breeders can plan their breeding programmes accordingly, and in time it should be feasible to completely eradicate this fatal disease from the breed. For the time being, should they wish to use a CARRIER in order not to sacrifice excellent breed points which it might have, this is OK provided it is never mated to

another CARRIER or to an untested dog. Likewise, two untested dogs should never be mated together. By mating a CARRIER to a CLEAR, we will not be producing AFFECTED dogs - we will just be prolonging the production of CARRIERS. Although not an ideal situation, it is acceptable until breeders find the 'Clear' dog that also proves to be their ideal stock to carry on their breeding lines.

**Breeders wishing to have any of their stock DNA tested can do so through:**

**Dr Nigel Holmes/Dr Cathryn Mellersh**

Canine DNA Testing  
The Animal Health Trust  
Lanwades Park  
Newmarket  
Suffolk CB8 7UU

Telephone: 01638 750659 Fax: 01638 750794 Email: [nigel.holmes@ah.org.uk](mailto:nigel.holmes@ah.org.uk) or  
[cathryn.mellersh@ah.org.uk](mailto:cathryn.mellersh@ah.org.uk)

**The procedure for requesting a 'DNA Fucosidosis' test is as follows:**

- ❑ The Animal Health Trust charges £50 (inclusive of VAT) for each sample to be DNA Fucosidosis tested.
- ❑ You should make application to the AHT for the 'Fucosidosis' test to be carried out on the AHT specified form
- ❑ The form i.e. 'Request for DNA Testing' can be downloaded from this site. A separate form should be completed for each blood sample to be tested.
- ❑ You should arrange for your Vet to take a Blood sample of 5mls in an EDTA tube, from the dog or bitch you wish to be tested.
- ❑ You will most likely be charged separately by your Vet to do this.
- ❑ After your Vet has taken the blood sample, ask them to send the sample to the address given above, with your completed '**Request for DNA Testing**' form and the appropriate fee.
- ❑ Cheques should be made payable to The Animal Health Trust.
- ❑ The AHT will usually post the results to you within 4 - 6 weeks of receipt of sample

**If you have any queries or would like any further information, please contact**

Health Co-Ordinators: **Louise Scott** (Tel: 020 8427 3396 or Email: [louise@goldcliffe1.freeserve.co.uk](mailto:louise@goldcliffe1.freeserve.co.uk)) or  
**Lesley Bloomfield** (Tel: 01923 823579 or Email: [lesley@fernl.in.free-online.co.uk](mailto:lesley@fernl.in.free-online.co.uk))