



CAVALIER KING CHARLES SPANIEL CLUB HEALTH INFORMATION SHEET

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DNA SAMPLING

COLLECTING DNA FROM CAVALIERS

Over the past few years DNA samples have been, and are being, collected for various projects relating to Cavaliers. These are:

- LUPA – Heart project led by Simon Swift.
- SM Genome Scan – led by P Knowler and Dr C Rusbridge.
- EBV Scheme – led by Dr Sarah Blott.
- KC DNA Profiling – parentage profiling.

These schemes are totally un-connected and samples are required for each scheme independently.

WHY TAKE SAMPLES OF DNA?

The functions of DNA are vital for inheritance, coding for proteins and the genetic blueprint of life. Given the enormity of DNA's functions in the body and its responsibility for the growth and maintenance of life, it is not surprising that the discovery of DNA has led to such a great number of developments in treating disease. DNA holds the instructions for an organism's development and reproduction - ultimately, its survival.

DNA samples are taken for three basic purposes:

- DNA Profiling.
- Genetic Screening.
- Genome Scanning.

HOW ARE SAMPLES TAKEN?

The amount of DNA material required for use by the Researchers is dependant upon how the sample is to be used. Samples are generally taken in one of two ways:

- By blood sample – provides the most DNA material. Blood contains white cells from which DNA can be extracted and even small volumes of blood will provide lots of DNA. The problem, of course, is that this is invasive and there are issues of the legality of blood sampling without an appropriate Home Office Licence.
- By Buccal Swabs (Cheek swabs). This involves removing the cells that line the inside of the dog's cheek, called buccal cells, with a small brush or swab. DNA can be extracted from these cells, though nothing like the amounts of DNA made from blood can be produced from these swabs. However, technological advances have meant that research scientists can work with vanishingly small amounts of DNA, making mouth swabbing a convenient and non-invasive source of DNA. The only difference between blood sampling and mouth swabbing is in the amount of DNA that is eventually produced

In the UK, a veterinary practitioner generally takes blood samples. The dog's owner can generally take buccal swabs.

DNA PROFILING

Each dog has a unique DNA signature that is referred to as its DNA profile or fingerprint, which can be likened to a biological microchip because each DNA profile is unique to an individual dog. The DNA profile can therefore be used to uniquely identify a particular dog. Unlike the microchip, however, the DNA profile can provide more information than this. Since a dog inherits half of its DNA from its dam and the other from its sire, this means that half of a dog's DNA profile is inherited from its dam and the other half from its sire. Therefore a dog's DNA profile can be used to verify that its parents are in fact its biological parents provided DNA profiles are available from both parents.

As only a small amount of DNA material is required, profiling is generally carried out using Buccal swabs. Some laboratories, however, require a small amount of blood – generally 1 to 3 ml.

DNA profiling takes 5 to 10 days and is therefore not instantaneous. As such it cannot be used as a method of permanent identification for health screening tests where the dog needs to be identified at the time that it is presented for examination. Permanent identification should be done using microchip or tattoo.

DNA profiling **is not** used to identify any genetic defects in the dog.

GENETIC SCREENING

Once the genetic make up, and therefore the genetic carriers, for a specific disease have been identified, DNA can be used to identify whether a dog is genetically clear, a carrier or is affected by that disease.

As only a small amount of DNA material is required, screening is generally carried out using Buccal swabs. Some laboratories, however, require a small amount of blood – generally 1 to 3 ml.

To date there are no hereditary diseases identified for the Cavalier for which there are DNA tests available.

GENOME SCANNING

Where the mode of inheritance for a specific disease is not known, Researchers make use of a protocol known as Genome Scanning to identify the genes responsible for the transmission of the disease. This is a complex operation and is beyond the scope of this information sheet. Simplistically a researcher examines segments of DNA material from affected animals and compares them to segments from known clear animals. Differences in the DNA make up may lead to the identification of the gene or genes responsible for the disease.

This type of testing requires the maximum amount of material from which the DNA can be extracted and is normally obtained from blood samples – generally up to 8 ml at a time.

This type of process is currently being carried out for both Syringomyelia and Mitral Valve Disease in the Cavalier.

The research to try to identify the genes, and the subsequent DNA markers, responsible for syringomyelia is being carried out at The University of Montreal, Canada and for mitral valve disease by The Royal Veterinary and Agricultural University, Denmark.

COLLECTION OF DNA AS PART OF THE EBV PROGRAMME

Cheek swab samples are being taken as part of the EBV activities from Cavaliers for DNA extraction and storage.

Once the DNA markers for syringomyelia have been identified by The University of Montreal, Canada and for mitral valve disease by The Royal Veterinary and Agricultural University, Denmark then the AHT teams will be able to screen the EBV donated samples and determine their disease genotypes and thus the status of the donor dog. The resultant information will then be added to the EBV program and this will aid in the more precise estimation of breeding values.

SHARING OF DNA

Many owners have asked whether the various centres share the DNA information. This is not possible for several reasons.

- Generally the amount of material collected at any one time is only enough for the purpose for which it has been taken.
- Not all the DNA information is extracted at any one time, for example the information for a specific disease is held in a known location within the DNA string and this is the only section that requires to be examined.
- There is difficulty in transferring DNA from location to location.
- There is difficulty in identifying DNA from specific dogs.
- The various projects do not have the time, money or staff to enable the sorting of DNA samples for other researchers.