

SYRINGOMYELIA NEWS

Autumn/Winter 2004

A research update

By Clare Rusbridge and Penny Knowler

Stone Lion Veterinary Centre, 41 High Street, Wimbledon, London, SW19 5AU

neuro.vet@btinternet.com (CR) Confidential Fax: 020 87860525

penny.knowler@ntlworld.com (SPK)

We have made amazing progress thanks to the wonderful foresight from committed owners, breeders and vets from around the world. After only one year of collecting DNA we have over 500 samples from a database of 8,000 cavaliers. This includes DNA from over 50 confirmed cases and over 60 related unconfirmed cases. A breed genetics complexity survey is being undertaken by the Mammalian Genotyping Service at the Marshfield Clinic, Wisconsin, USA, identifying 800 DNA markers. Analysis of this data by Dr Guy Rouleau's team in Montreal will take 1-2 months. Meanwhile he has asked that we continue to collect as many cases of Syringomyelia and MVD as possible. All DNA/Blood is now being centralized at the McGill University Health Centre, Montreal -- see address below. We are extremely grateful to the Canadian team in helping to achieve this exciting first stage in less time than one Cavalier generation. The difficulty of the task ahead cannot be overestimated. Never before has a complex inheritance such as Syringomyelia or MVD been addressed and we are breaking new ground. A poster presentation of our achievements and those of the cavalier community worldwide was given by Clare at the Advances in Canine and Feline Genomics 2nd International Conference in Utrecht, October 14, 2004. It aroused a great deal of interest. As a direct result, she has already helped set up a collection for a number of other disorders!

Our second research paper Inheritance of occipital bone hypoplasia (Chiari 1 malformation) in Cavalier King Charles spaniels has been published in the American Journal of Veterinary Medicine (JVIM 2004; 18: 673-678). Reprints are being sold in aid of SM research and can be obtained from Penny Knowler (address/email at top of page) It outlines the observations made from a pedigree analysis of the family tree as it existed in July 2003, containing 120 cases of SM and 40 cases of idiopathic epilepsy. A consistent observation with the present time is that all affected dogs have 3-4 grandparents descended from lines with known carriers in them. The degree of syringomyelia is quite variable. The most severe cases have considerable spinal cord damage and are significantly disabled by 12 months of age. In contrast, some have no clinical signs and a small syringomyelia which is only detectable by MRI or post mortem. The disease appears to be more severe and have an earlier onset with increased inbreeding. Geneticists agree that an understanding about the inheritance cannot be based on pedigree analysis alone.

The health committee of the UK Cavalier Club has been working extremely hard and is always on hand to answer queries and to offer support to its members and to us. Thanks to the sponsorship of Boehringer Ingelheim (producers of Vetmedin®), they were able to arrange health clinics for heart & eye testing in different parts of the country. They also took the opportunity to arrange a blood donation session at the clinic at Banwell in September. Twenty two dogs over 4 years old volunteered, including some oldies with clear hearts (which are helpful for the MVD research), and

one or two younger dogs who were picked by the Cardiologist as being of particular interest. Simon Swift, the 'Club Cardiologist', kindly helped the owners fill in the heart and syringomyelia details required, while the rest of the paperwork was completed with the assistance of the veterinary nurse. At the end of the day Simon took the blood & forms back to his clinic and posted the phenotype forms to Clare Rusbridge before the forms & blood were sent off to the UK DNA Archive.

The Fund has already benefited from the proceeds from a sponsored weight loss and a short sponsored walk. Christmas Cards with an exclusive design featuring all four colours of cavaliers are also on sale. All proceeds will go to the Research Fund.

Support continues to come from all over the world. The UK Cavalier Club has received some extremely generous donations for the Research Fund including 2000 Australian dollars from the Cavalier KCS Club of Victoria. We are also extremely delighted to be receiving blood from two more European countries, Germany and France. The people involved are so determined to help the cavalier breed that they have taken the time and trouble and expense to arrange their own clinics and collections.

Dedicated cavalier breeder Dana Schuller-Kuyper in Holland has opted to have a diagnostic MRI performed on her breeding dogs in order to ensure the best selection for health. DNA from these cavaliers has enormous value to the research because SM status is confirmed.



Positioning the cavalier carefully is important to obtain the best possible image from the MRI scan.



Dutch vet, Hayo van Geervliet, draws blood from the jugular vein of a very accommodating cavalier held by Pirkko.

Details of the UK collection can be found on <http://www.thecavalierclub.co.uk>.

Blood Donations are needed from our target groups SM and MVD.

Please send a copy of phenotype form and pedigree to Clare Rusbridge address above
This is needed to provide an ID# for use by the UK Archive and the researchers in
Canada.

Include any medical notes for MRI or Cardiologists reports.

It is essential to have accurate information, please update your Phenotype Forms if the
status of your dog has changed, (E.g. confirmation of diagnosis) – send to Clare!