

SYRINGOMYELIA NEWS

Winter/Spring 2005

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)

It is a year since we started to produce these Newsletters with the aim of updating breeders and owners of the research developments into the cause and treatment of syringomyelia. It has been quite a year and we feel very fortunate to have come into contact with so many CKCS lovers. We would welcome other contributions to this newsletter from other pet lovers, vets or researchers.

The DNA collection continues to grow with well over 800 samples, some spanning 6 generations. Our knowledge of the disease has been advanced particularly by the help of owners who have MRI'd their dogs from UK and USA. However, we are particularly indebted to the Dutch breeders who have undertaken a nationwide MRI screening at considerable personal expense. They take the view that selection for health is every bit as important as selection for temperament and appearance. Dana Schuller-Kuyper organised a talk by Clare in Utrecht, Netherlands in late January.



The talk was extremely well received. The participants, some travelling from Germany, included breeders and members of the veterinary and human medical profession.

We are very grateful for the hard work and commitment that has been shown from around the world. Pat Barrington and the health committee of the Canadian Cavalier Club spearheaded a series of very successful blood clinics, one of which totalled a record 33 dogs. The breeders travelled long distances to take part and enjoyed the social opportunity meeting up with their puppy owners. These collections were undertaken with super efficiency, each dog being examined by a vet who gave their services free and forwarded the completed paperwork of phenotype forms, medical notes and pedigrees.

Thanks also to Isabelle Barthes in France, who has successfully completed a DNA collection there. The attention to completed paperwork, especially 5 generation pedigrees was particularly appreciated by Penny. This has enabled DNA from related dogs from different countries to be linked. Veterinary Neurologist Dr. Laurent

Cauzinille helped translate the documents, wrote a review article for the French Veterinary profession and gave a presentation on syringomyelia to the CENA (French Cavalier & King Charles Club).

Syringomyelia/occipital hypoplasia (Chiari malformation) does occur in other breeds. We are keen to obtain DNA from as many of these affected dogs as possible – not just CKCS. We have been helped to locate some volunteer donors from an online support group set up by Angela Baker. Apart from offering support for the distress that syringomyelia causes to owners and dogs alike, the group has provided a forum for debate and a means of disseminating information. Members have a wide range of expertise and experiences and the website provides the opportunity to share this in a constructive manner. http://uk.groups.yahoo.com/group/ArnoldChiari_dogs/. Angela founded the Support Group in 2003 and membership currently stands at 308. There have been 5859 messages posted. Membership is free.



The UK Archive will be shipping over 450 samples to Canada in the next few weeks. We are grateful to Wendy Hallows and Francine Jury who has been assisting in this hard work. Archiving is now done at the Centre for Integrated Genomic and Medical Research (CIGMR) Division of Epidemiology and Health Sciences at Manchester University, M13 1BJ.

As some of you may have already heard, the Rouleau laboratory will be relocating to the University of Montreal Health Centre (CHUM) situated at the Notre Dame Hospital in Montreal on March 7, 2005. The new Centre for the Study of Brain Diseases is an integrated research unit headed by Dr. Guy Rouleau that comprises 4 clinical scientists with an interest in the genetics of various neurological diseases. The new facilities include state of the art genetic equipment that will enable the processing of a high volume of DNA samples as well as cell culture and genotype analysis. Melanie Benard who is co-ordinating the project in Canada is also moving. Details of the new address and information for sending samples from out-with the UK can be obtained from Clare or Penny (address above) or from www.thecavalierclub.co.uk.

We are delighted to receive a grant for \$8,000 from the Cavalier Health Foundation (USA) towards the archiving of DNA which has been undertaken by Dr Berge Minassian at the Hospital for Sick Children in Toronto. Some owners and breeders (not all CKCS) have expressed the wish to raise funds for the DNA research in North America we have arranged deposit facilities for both Canadian and USA dollars to be sent to a designated Syringomyelia Research Fund directed by Dr Minassian (again details from www.thecavalierclub.co.uk or from us).

We are also delighted that we can report the success of Clare's collaboration with Dr

Berge Minassian team in Toronto in identifying a unique canine gene responsible for Lafora disease (a form of epilepsy) (Science 2005; 307: 81) The experience gained by Clare and Penny in this research inspired the 'DNA for Healthy Cavaliers' project.

Reminders

If the medical status of the dog changes simply fill in another form and send or fax to Clare marking 'UPDATE' clearly on the form. This is especially important for MVD when dogs often go for regular checkups.

1. • DNA from our priority dogs (particularly MRI clear)
2. • If possible send a copy of CD of MRI to Clare (or she can return it)
3. • accurately filled in phenotype forms, the provision of medical notes if available and a pedigree. This enables the geneticist to make full use of the DNA.

Notes in the Diary -- upcoming lectures by Clare Rusbridge

ACKCSC National Specialty in Lancaster, Pennsylvania May 5th 2005

National Meeting/Show of the Cavalier King Charles Spaniel Club USA May 12th 2005

Canadian Cavalier King Charles Spaniel Club, Ontario, May 29th 2005

American College Veterinary Internal Medicine; Neurology Forum, Baltimore, Maryland, 3rd June 2005 (veterinary surgeons only)