

## **RESEARCH INTO SYRINGOMYELIA (UK and USA)**

**Carol Fowler**

**I have attempted to collect and summarise the various research projects which have been undertaken, or are now in process, or proposed, for Syringomyelia in Cavalier King Charles Spaniels.**

**I hope this will show that although things are bad for our breed at present, with so much veterinary attention on the problem, the future for the CKCS breed will be a much healthier one.**

*Collection and archiving of Cavalier King Charles Spaniel DNA, to identify the mode of inheritance and genes responsible for Syringomyelia, MVD and Epilepsy.*

**CLARE RUSBRIDGE BVMS, Dip ECVN, MRCVS, European and RCVS Specialist in Veterinary Neurology, and PENNY KNOWLER BSc**  
The Stone Lion Veterinary Centre, Wimbledon, London, UK

**Clare Rusbridge first described the disease when a resident at the Royal College of Veterinary Surgeons in 1997. She later established that the disease was inherited. Over the past 8 years she has continued to research this disease, focusing on the genetics, pathogenesis and treatment, while undertaking a PhD on Occipital Hypoplasia / Syringomyelia, based at Utrecht University. She is currently collaborating with Dr Guy Rouleau, MD PhD FRCP(C), at the Centre for the Study of Brain Diseases, CHUM Research Centre, Montreal, and Dr Berge Minassian, MD CM FRCP(C), Centre for Applied Genomics, at the Hospital for Sick Kids, Toronto, Canada.**

**Their research aims to include the identification and characterisation of genetic factors predisposing to Chiari 1 malformation, to increase the understanding of the pathophysiology of SM, for better diagnosis, clinical management and ultimate prevention. The initial stage of this undertaking is nearing completion with the collection and archiving of Cavalier King Charles Spaniel DNA. This has been achieved by the support of the Cavalier Club UK, the UK DNA archive, Boehringer Ingelheim Limited, CKCS Club USA Health Foundation, veterinarians, pet owners and breeders worldwide.**

**Publications to date:**

***Syringomyelia in Cavalier King Charles Spaniels. RUSBRIDGE, MACSWEENEY, DAVIES, et al***

**J Am Anim Hosp Assoc 2000 36: 34-41**

***Dorsal dens angulation and a Chiari malformation in a Cavalier King Charles Spaniel. BYNEVELT, RUSBRIDGE, BRITTON***

**Veterinary Radiology and Ultrasound, 2000, 41**

*Hereditary aspects of occipital bone hypoplasia and syringomyelia (Chiari type 1 malformation) in Cavalier King Charles Spaniels. RUSBRIDGE, KNOWLER*  
The Veterinary Record, July 26 2003  
*Syringomyelia RUSBRIDGE*  
UK Vet Vol 8 No 8 November 2003  
*Inheritance of occipital bone hypoplasia (Chiari 1 malformation) in Cavalier King Charles Spaniels. RUSBRIDGE, KNOWLER*  
Journal of Veterinary Internal Medicine 2004, 18  
*Neurological Diseases in Cavalier King Charles Spaniels. RUSBRIDGE*  
Journal of Small Animal Practice 2005, 46  
*Inherited occipital hypoplasia/syringomyelia in the Cavalier King Charles Spaniel – experiences in setting up a worldwide DNA collection. RUSBRIDGE, KNOWLER, ROULEAU, MINASSIAN, ROTHUIZEN*  
Awaiting publication in Journal of Heredity, 2005

*Neurological signs and results of magnetic resonance imaging in 40 cavalier King Charles spaniels with Chiari type 1-like malformations.*

**D. LU, C LAMB, D PFEIFFER, M TARGETT**  
The Royal Veterinary College, Hertfordshire, UK  
Veterinary Record, August 30, 2003

The study found that the dogs had a wide variety of neurological signs, but there was no apparent correlation between the neurological signs and the severity of cerebellar herniation, syringohydromyelia, or hydrocephalus.

*Cavalier King Charles Spaniel Health Survey*

**DR LARRY GLICKMAN**  
Purdue University School of Veterinary Medicine, West Lafayette, Indiana, USA

The study aims to identify health trends in the breed along with diseases and disorders prevalence. To retain geneticist(s) and a canine genetics counsellor to address each concern carefully, reviewing them with each generation. Develop a university based clearing house for MRI, Cardiac and DNA research.

*The Study of Caudal Occipital Malformation Syndrome (COMS) in Cavalier King Charles Spaniels*

**CARLY ABRAMSON, DVM, Assoc Prof. and PHIL MARCH, DVM**  
The Ohio State University School of Veterinary Medicine, USA

Caudal Occipital Malformation Syndrome (COMS), similar to Chiari malformation in humans, is a recognised problem affecting the Cavalier King Charles Spaniel breed, which can lead to the development of neurological disease. Currently,

treatment recommendations are based on the presence of progressive neurological deterioration. There are no objective parameters to evaluate affected patients which are at risk for developing neurological disease prior to clinical deterioration. Researchers hypothesise that Magnetic Resonance Imaging (MRI) and ultrasound parameters can be used to predict dogs at risk, and objectively evaluate efficacy of therapy for this disorder.

The objective of this study is to evaluate a family of CKCS that includes clinically affected and clinically normal dogs. Measurements of ventricle size, foramen magnum size, and spinal cord dilation, as well as cerebrospinal fluid (CSF) flow velocity studies at the foramen magnum, will be evaluated on MRI. Basilar artery resistive index obtained with ultrasound will be assessed. The dogs will be monitored over 2 years to determine the ability of these objective parameters to predict clinical disease progression. For those patients receiving medical or surgical treatment of COMS, each diagnostic test will be applied post-treatment to evaluate their ability to monitor success of treatment.

*The Effect of Chiari Type 1 Malformations on CSF Flow in Cavalier King Charles Spaniels*

**DR NATASHA OLBY and DR SOPHIA CERDA-GONZALEZ**  
North Carolina State University Veterinary School, USA, May 2005

This study aims to compare CSF flow patterns and velocity at the level of the foramen magnum between three groups of Cavalier King Charles Spaniels: normal CKCS, symptomatic CKCS, and asymptomatic CKCS with Chiari malformations. Researchers will also begin to establish the incidence of asymptomatic Chiari malformations in a group of CKCS, as well as investigate the genetics of this disorder.

The pattern and velocity of CSF flow can be measured using a specialised, non-invasive MRI technique called PVC MRI. The researchers' aims are to determine the site of peak flow and turbulence (which will aid in determining the most appropriate surgical approach to treating the disease), and to determine what CSF flow velocity is the threshold for developing clinical signs. The study will also allow the screening of potential breeding dogs for the presence of the disease, and will allow researchers to estimate the incidence of asymptomatic disease in CKCS in North Carolina and surrounding regions.

*Proposed Sequential MRI Study of CKCS Litters*

**DR CURTIS DEWEY**  
Long Island Veterinary Specialists, New York, USA

Dr Dewey plans to start a project of sequentially MRI scanning litters of CKCS pups, in order to identify the prevalence of the malformation and the progression of the disorder. He plans to focus on early surgical intervention in symptomatic

**puppies, to determine whether or not such intervention is more likely to lead to disease resolution, than operating at adulthood. Dr Dewey's emphasis is on finding the most effective treatment options for dogs affected with COMS.**

**To the best of my knowledge, the above information is accurate and correct, at the time of writing.**

**Carol Fowler, May 2005**