



## Curly Coat Syndrome and Episodic Falling in Cavalier King Charles Spaniels in the UK Mutation Frequency Study Report - June 2012

In 2012 scientists at the Kennel Club Genetics Centre at the Animal Health Trust undertook a study to measure the frequency of the mutations responsible for congenital keratoconjunctivitis sicca and ichthyosiform dermatosis (curly coat syndrome) and episodic falling in Cavalier King Charles Spaniels in the UK. A report describing the study, its conclusions and recommendations has been distributed to the UK Cavalier Breed Clubs and is available on the Animal Health Trust website:

http://www.aht.org.uk/cms-display/genetics\_success.html

We tested 280 DNA samples from Kennel Club registered Cavaliers and used the results to calculate the frequencies of these two mutations in the breed as a whole. Our results show that **19.1%** of Cavaliers are carriers of the episodic falling mutation and **10.8%** are carriers of the curly coat syndrome mutation. Both mutations are present in UK dogs of all four coat colours.

Almost 30% of UK Cavaliers of breeding age are carriers of episodic falling or curly coat syndrome, and a small number are carriers of both. Around 1-2% of Cavaliers carry two copies of the episodic falling and/or the curly coat syndrome mutation and are affected.

Our recommendations based on these results are as follows:

- All Cavalier King Charles Spaniels that are to be bred from should be DNA tested for both mutations prior to mating, regardless of colour or ancestry
- When planning a litter, breeders should choose a dog and bitch that cannot combine to produce affected puppies
- Carriers should not be excluded from breeding programmes until the mutation frequency within the breed falls below 0.01 (1%) to avoid reducing genetic diversity unduly
- Progress towards elimination of these two inherited diseases from the breed should be monitored by carrying out further mutation frequency checks every few years

The research team is grateful to the committees and members of all the Cavalier Breed Clubs for their help with our study. We hope that this research will be helpful to all Cavalier breeders and owners.

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