

### Episodic Falling in Cavalier King Charles Spaniel

Prof. Dr. Robert Harvey working at the London School of Pharmacy has recently identified the underlying genetic defect causing Episodic Falling in Cavalier King Charles Spaniels. LABOKLIN recently implemented the genetic test for this disease under licence from the London School of Pharmacy. This test is currently patented within Europe, with Laboklin as the testing laboratory.

Episodic Falling is a neurological disorder found in Cavalier King Charles Spaniels. Episodes are triggered by exercise, stress or excitement and are characterised by stiffness throughout thoracic and pelvic limbs resulting in a characteristic 'deer-stalking' position and/or collapse. Clinical symptoms are variable in onset (between fourteen weeks and four years of age) and severity.

The new DNA test enables the identification of clinically normal carrier animals and will enable confirmatory diagnosis of affected animals.

### NCL in Tibetan Terriers

Neuronal ceroid lipofuscinoses (NCL) are a part of the group of inherited progressive and mostly fatal neurological storage diseases. In affected animals ceroid lipofuscin, a degradation product of metabolism, is deposited in the connective tissue and retina of the eyes as well as the nerve cells of the brain leading to cell death. NCL is inherited in an autosomal recessive manner, which means that only animals which have inherited the mutation from both parents and have the mutation on both chromosomes will have the disease.

NCL in Tibetan Terriers was first described in 1992. In this breed there is a late form of the disease where the first symptoms occur at an age of approximately 6 years. Affected animals show in particular a change in character, such as a state of anxiety and/or aggressive behavior. Permanent restlessness, uncleanliness, coordination difficulties and deterioration in vision are characteristic features. Progression of the disease leads mostly to euthanasia at an age of 8-10 years. The genetic cause of the disease has been known in Border Collies, American Bulldogs, English Setters, Dachshunds and Australian shepherds for a long time. Research has also been carried in Tibetan Terriers out for many years. The research group under Gary Johnson was recently able to demonstrate the connection between a mutation and the disease. LABOKLIN can now offer a test for the detection of this mutation whereby even clinically unobtrusive carriers can be detected. This makes it possible to allow targeted breeding to eliminate this disease.

### Dilution: LABOKLIN has acquired the patent rights

The dilution gene causes a paling of the basic color, e.g. from black to blue in Dobermans, but the test is also interesting for a variety of other breeds. LABOKLIN has acquired the patent rights for color dilution in dogs (so-called dilution test) from the Veterinary University Hannover and now has the sole rights for this test.

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### JEB in German Short-Haired Pointer

Junctional epidermolysis bullosa (JEB) is a skin disease which leads to blister formation between individual skin layers after minimal friction or bumping due to defects in skin regeneration. Erosion and scab formation occur around the ball of the paws, at pressure points of the extremities, such as the knee, elbow, ankles, paw root joints, hip and inside the ears as well as in the gums, tongue and lips. Some dogs also develop grainy tooth enamel. JEB is also inherited in a recessive fashion. Clinically unremarkable carriers of this disease can be detected by a gene test so that by selective breeding no affected pups will be born.

### NEW: Episodic Falling in Cavalier King Charles Spaniel

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### JEB in German Short-Haired Pointer

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### Smears as sample material

### MDR1-new pharmaceutical list

### Hemophilia A in dogs

### Smears as sample material

Smears from the inside of the mouth using a cotton wool swab are commonly preferred for extraction of DNA for various gene tests because they can be less stressful than taking a blood sample.

However, the following points must be considered: recent results have shown that young suckling puppies should be separated from the mother for at least 2 hours before sampling can be done to avoid contamination by maternal DNA.

The amount of DNA on a cotton swab is relatively low compared to a blood sample and a swab is only sufficient for one DNA preparation procedure. In order to guarantee the greatest test safety whenever possible we always carry out two separate tests on the submitted samples and only report the results of the tests when there is concordance. We would therefore again request that either an EDTA blood sample or two mouth swabs should be submitted as samples from each animal for carrying out genetic testing. Only then can the customer enjoy the luxury of a confirmatory test free of charge.

### MDR1-new pharmaceutical list

In addition to a rapid processing and a qualitatively high-grade evaluation of your samples, LABOKLIN offers you an abundance of sources of information. In particular, on the topic of MDR1 you will find a large amount of additional information on our home page. A continuously updated list of pharmaceuticals indicates medications which are problematic or easy to administer if a mutation in the MDR1 gene is present. Here you can find exact instructions on which medications can be administered to dogs of which genotype.

### Tests for hemophilia A in dogs

**We request your assistance! After successfully carrying out various projects on the recognition of hemophilia in some breeds of dogs, we now need your cooperation.**

It is known that a deficiency of factor VIII leads to hemophilia. The sequence of this gene has already been confirmed and can be used for a comparison with test results. Within the framework of a doctoral thesis a veterinarian investigated samples in our laboratory where there was a suspicion of hemophilia. For this thesis we now need samples from dogs affected by hemophilia A so that we can analyze their factor VIII gene. Through these investigations we hope to find the causative mutation in each individual case. The tests developed can allow the respective breeder to eliminate the disease by testing and selective mating. Breeds which we are focusing on are Great Danes and Poodles.

Do you have an affected Great Dane or Poodle or do you know a breeder who has a dog (normally male dogs are affected) which bled to death for no known reason? If you would be willing to support our project please contact us to discuss further details. We would be pleased to receive an e-mail from you at [labogen@laboklin.de](mailto:labogen@laboklin.de) or give us a call under the telephone number 0049-(0)-971-72020 and ask for Frau Kehl or Frau Dr. Kühnlein.