Newsletter genetic | No. 03 / 2014
A service by LABOKLIN - Laboratory for clinical diagnostics
www.laboklin.com

LABOGEN

genetic news

Genetics and more

Here, in this category of our newsletter, we would like to take the opportunity to touch on some distinguishing facts about our laboratory beyond genetic testing. Today we want to inform you about accreditation. LABOKLIN is an accredited laboratory according to DIN EN ISO / IEC 17025. This means that our tests are always executed according to reliable and verifiable procedures. Thereby, we are not only subject to strict daily controls, but also regular external controls in the form of ring trials.
We have also implemented fixed testing, controlling and training procedures in our laboratory that are regularly reviewed on-site by an accreditation agency. Thus, you are trusting your samples to a lab partner, who is committed, with expertise and heart, to providing the highest possible quality of processing and consultancy.

ilac-MRA DAkkS Deutsche Akkreditierungsstelle D-PL-13186-01-00

Bleeding disorders in dogs - vWD and Co

Should it come to vessel injury and loss of blood from a wound, urgency to stop the bleeding is called for. After all, massive blood loss would lead to death. However, inappropriate coagulation must also be prevented, as this may also lead to death by embolism or infarction. For this purpose, the vertebrate body has a sophisticated system for the clotting of blood: a cascade-like process, in which platelets and clotting factors are involved. If one of these factors is missing it results in a so-called bleeding disorder. The diseases are named after the affected element of the cascade (see table).

The symptoms of the diseases barely differ from one another, only the severity of each varies somewhat. Unusually heavy bleeding occurs, for example, after injury or during dentition; affected dogs tend to form hematomas and often exhibit lameness due to bleeding into joints and muscles.

Bleeding disorders can occur genetically. If the underlying mutation is known, genetic testing can identify the, for the most part symptomless, carrier animals. As of yet, the known genetic changes are breed specific, i.e. there are different genetic variants in different breeds, but they result in the same symptoms. Therefore, genetic testing only makes sense in breeds for which the presence of the mutation has been correlated to the formation of disease (see table).

The following should be noted in regards to **inheritance**:

For each trait there are two copies in the genome. In each animal one copy is received from the sire and one from the dam. If a trait for disease is inherited as autosomal dominant, an animal may already become ill when it has only one copy of the altered gene. The **von Willebrand Disease Type 1** follows this pattern of inheritance, however, with a twist: the severity of the symptoms vastly varies in heterozygous animals (ones that only have one modified copy), as

D-97688 Bad Kissingen • Steubenstraße 4
Tel. +49 971 72020 • Fax +49 971 68546
info@laboklin.de

CH-4058 Basel • Riehenring 173
Tel. +41 61 3196060 • Fax +41 61 3196065
labor.basel@laboklin.ch

A-4040 Linz • Rosenstraße 1
Tel. +43 732 717242 • Fax +43 732 717322
labor.linz@laboklin.at

Disease	Altered factor	Inheritance	Breeds for wich genetic testing is available
von Willebrand Disease Type 1	von Willebrand Factor (vWF)	Autosomal dominant with incomplete penetrance and variable expressivity	Bernese Mountain Dog, Coton de Tulear, German Pinscher, Doberman, Drentsche Patrijshond, Kerry Blue Terrier, Manchester Terrier, Papillon, Poodle, Stabyhound, Welsh Corgi
von Willebrand Disease Type 2	von Willebrand Factor	Autosomal recessive	German Wirehaired Pointer, German Shorthaired Pointer
von Willebrand Disease Type 3	von Willebrand Factor	Autosomal recessive	Kooikerhondje, Scottish Terrier, Shetland Sheepdog
Factor VII Deficiency	Factor VII	Autosomal recessive	Airedale Terrier, Alaskan Klee Kai, Beagle, Giant Schnauzer, Scottish Deerhound
Haemophilia A	Factor VIII	X - linked recessive	Havanese
Haemophilia B	Factor IX	X - linked recessive	Rhodesian Ridgeback
Thrombopathy	Thrombocytes	Autosomal recessive	Basset Hound, Landseer

the causative mutation influences the expression of, i.e. the production of, the vWF protein. In heterozygous animals, a normal amount of vWF is expressed at times and the animals have no symptoms, but at other times there is a lack of vWF, and thus at these times bleeding tendency is present. Homozygous affected animals (ones that have two modified copies) always show symptoms.

The mode of inheritance for **Haemophilia A and B is X-linked recessive**, because the genes for Factor VIII and IX are located on the X chromosome. Males (XY) will suffer, as a carrier of the mutated gene, from the disease and transmit the disease to 100% of their female offspring. Male offspring of affected sires can only inherit the disease from the dam. In females (XX) inheritance is analogous to an autosomal recessive inheritance. Female carriers of the disease are clinically normal, but the mutated gene is passed to the next generation with a 50% probability, so the disease affects 50% of male offspring.

The **von Willebrand Disease Type 2** and **3** as well as **Thrombopathy** and **Factor VII Deficiency** are autosomal recessive disorders. Carriers pass the hereditary trait onto their offspring with a probability of 50%. When mating two carriers, there is the risk that 25% of the offspring will be affected. The existence of carriers in a healthy population increases genetic variability of the entire gene pool, which is why they should not be categorically excluded from breeding. However, breeding of carriers should only occur with mutation-free animals, so no homozygous, thus affected, animals will be born.

The tick season has begun

Did a tick bite? Than your animal may be in danger of being infected with Borrelia, Anaplasma or Ehrlichia. In this case, one can examine the tick directly using PCR for these pathogens to assess the risk of infection.