LABOKLIN
LABOR FÜR KLINISCHE DIAGNOSTIK GMBH & CO. KG
NEWSLETTER 4/2012

Genetic diseases in the dermatology

A genetic disposition for certain skin diseases is prevalent in different breeds. For some of these diseases the causative mutations have recently been identified.

For **Ichthyosis** in the Golden Retriever, Norfolk Terrier and Jack Russell Terrier the responsible genetic modification is known. The causal gene variant of the disease and thus affected animals occur only sporadically in the two Terrier breeds. Since the pathogenic gene variant in Golden Retrievers, 30% in sick animals and 40% in carrier animals is so widely spread in France it is necessary to carefully control the mating of these animals. A total exclusion of carrier animals would extremely reduce the genetic diversity of the breed. However, one of the mated animals should always be free from the mutation.

The **Hereditary Nasal Parakeratosis (HNPK)** leads to dehydration of the dogs' nose. Especially on the upper side of the nose, dry, thickened layers of skin build up, which are irremovable. These can crack and secondary bacterial infections can take hold. The genetic origin of the disease in the Labrador Retriever was discovered by researchers in Prof. Dr. Tosso Leeb (Institute of Genetics of the Vetsuisse Faculty at the University of Bern) and Prof. Dr. Hannes Lohi (University of Helsinki) work groups. Through the use of the genetic testing for HNPK in Labrador Retrievers the occurrence of new cases can be prevented.

Atopic dermatitis is a problem in many breeds. Since this is a complex disease, with both genetic factors as well as environmental factors, no genetic testing can be expected in the foreseeable future.

Still relevant is the **Colour Dilution Alopecia (CDA),** which is associated with the mutant dilution gene (d/d). Histological studies suggest that dogs with lighter coat colours are predisposed for CDA. However, the alopecia is triggered by currently unknown factors. Additionally, the correlation between the genotype d/d and CDA is breed specific. In this respect up to 93% of Pinschers with a dilute coat colour (genotype d/d) suffer from alopecia, whereby hardly any cases of CDA have been described in French bulldogs.

A further autosomal recessive inherited skin disorder, for which LABOKLIN offers a genetic test, is the so-called **Dry Eye Curly Coat Syndrome** in the Cavalier King Charles spaniel. For detailed information on this see our newsletter 2/2012.

During the 58th annual congress of the German Veterinary Society an event under the theme "experts explain" designed specifically for breeders took place for the first time. In this newsletter we offer you a small selection of interesting topics from the veterinarians', as well as the breeders' event.

GLEANINGS

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Genetic diseases in the neurology

Also in the field of neurology there is a considerable amount of research, some of which has already contributed to the development of genetic tests:

Prof. Fredholms' team was able to identify a point mutation in the NDRG1 gene as the cause of **Polyneuropathy** in the Alaskan Malamute, whereby a deletion in the same gene causes Hereditary Neuropathy in Greyhounds. In Leonbergers, the situation is more complicated: the early form which begins at an age between 18 and 24 months has a genetic origin, while the etiology of forms later in life are still being researched (Drogemuller, University of Bern). For some accumulation diseases, which also clinically appear as neuropathies, the genetic causes were successfully identified. These include the Globoid Cell Leukodystrophy (Krabbe) in the West Highland White and Cairn Terrier, Fucosidosis in English Springer Spaniels and GSD IV in the Norwegian Forest Cat.

Epilepsy has been an important topic for a long time in many breeds and the genetics have been examined for many years. Partial success has been achieved: genetic tests have been developed for the Neuronal Ceroid Lipofuscinosis, which can cause epileptic episodes, in English Setters, Tibet Terriers, Border Collies, Australian Shepherds, American Bulldogs and Dachshund. Additionally, the genetics of the Neonatal Encephalopathy (NEWS) in the Standard Poodle as well as the Myoclonus Epilepsy in the Miniature Wire-haired Dachshund and Basset hound have been determined. The recent elucidation of the genetics of the Idiopathic-genetic Epilepsy in Belgian Shepherds and the Benign Juvenile Epilepsy in Lagotto Romagnolo has provided the science with further valuable findings regarding the origin of these diseases. Further research will be necessary to identify the etiology in other breeds.

New at LABOKLIN: IGS in the Border Collie

The **Imerslund-Gräsbeck-Syndrome (IGS)** occurs due to a selective cobalamin malabsorption, this means vitamin B12 cannot be absorbed through the intestinal epithelium. Chronic vitamin B12 deficiency leads to changes in the blood, such as anemia, as well as irreversible neurologic damage. A life-long vitamin B12 substitution is required in order to reduce the symptoms. Using genetic testing and directed breeding the number of cases displaying this autosomal recessive disease can be minimized.

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