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LABOGEN

GENETIC news

Old and new in HCM

Hypertrophic cardiomyopathy (HCM) is the most common heart disease in cats. Affected breeds include Maine Coon, Persian, Ragdoll and British Shorthair, as well as the domestic cat. Breeding studies using echocardiography are particularly meaningful in 2 years and older and in females older than 3-4 years. In younger animals, the significance of such studies is limited, because they might develeop HCM later. As of recently, there is the possibility to determine NT-proBNP and Troponin I in blood. Both are suitable biomarkers for the presence of heart disease, because the values are elevated in HCM, but they offer no replacement for an ultrasound examination.

For the breeds Ragdoll and Maine Coon there are additional genetic tests for HCM, which can be carried out in very young animals using blood or a cytobrush.
While in the Ragdoll a very high correlation between genetic disposition and disease is found, the research results in the Maine Coon vary. In 2005, Dr. Kathryn Meurs (North Carolina State University) identified a mutation in the MYBPC3 gene, which she associated with the development of HCM in the Maine Coon. However, in a subsequent study, Dr. Gerhard Wess from the University of Munich could not confirm a significant correlation between this mutation and the occurrence of HCM.
It is possible that this is due to the different Maine Coon - populations (USA and Germany) that were examined as well as the small number of samples that were available. As far as the population in this country is concerned it is unclear whether the observed genetic change in the U.S. has an influence on heart disease. Certain is, however, that the mutation in the gene MYBPC3 changes one amino acid resulting in a changed protein. What impact this has on the development of HCM is not fully understood. There are certainly other factors that positively or negatively influence the development of cardiomyopathy.

In conclusion:
Even if the genetic predisposition is only one component in the development of HCM, the risk of this serious illness should be minimized through the examination of the parent animals. Therapies can only alleviate the symptoms; no cure is possible.

New diagnostics for FIP (feline infectious peritonitis)

1. The disease:

FIP is one of the most common infectious causes of death in cats. The disease is caused by **mutated feline coronaviruses**. So far there is no effective therapy; in some animals life can merely be prolonged using certain medications. The primary and purely diarrhea causing coronaviruses are found worldwide, however, FIP occurs only in a few individuals when the harmless coronaviruses mutate within the cat into the deadly FIP virus.
Clinically, two different forms are manifest, which may, however, fade into one another. The most common form is the “wet” FIP, which is characterized by an accumulation of fluid in body cavities, especially in the abdomen. In the “dry” form no free liquid is built this effusion-free form causes inflammatory

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nodular changes in various organs such as the kidney and liver. Cats with a clinically manifested FIP also often show weight loss, very high fever and anaemia with jaundice.
Previously, there was hardly a sure diagnosis for FIP. As of recently, however, it is possible to specifically **identify the viruses containing specific mutations associated with the formation of a FIP disease**.

Thereby, finally, distinction between the harmless coronaviruses and the FIP-causing mutants is possible. The test method used is PCR: a molecular biological diagnostic method in which parts of the genetic material (DNA) of a pathogen can be isolated. Thus if one of the mutations is identifiable, either in the effusion liquid in a wet FIP or in blood in the dry FIP, a definite diagnosis of FIP can be made. If none of the described mutations can be located, however, one can not rule out FIP completely.

2. Detection of coronavirus-shedders:

Cats that excrete coronaviruses with the faeces represent a source of infection for other animals in the household. The more viruses are shed, the more will be ingested and therefore, the likelihood increases that virus replication will occur in the cat and disease-causing mutants will be formed. **Coronaviruses found in faeces do not possess the described mutations.** Thus, a transfer of the mutant viruses through the faeces from one cat to another is not possible. However, a **repeated faecal examination via PCR** still remains a useful diagnostic tool **for the identification of chronic coronavirus carriers**, especially in multi-cat households. We recommend an examination of samples from three consecutive days and we evaluate the amount of viruses found semiquantitatively (low, medium, high) in order to better estimate the threat of infection.

Hereditary Nasal Parakeratosis (HNPK) in Labrador Retrievers

The mutation responsible for causing Hereditary Nasal Parakeratosis (HNPK) was first described by Prof. Dr. Tosso Leebs' research group, University of Bern.
LABOKLIN was able to acquire an **exclusive license** for the genetic test of the HNPK mutation which is about to be patented. The genetic test is from now on performed by

us inhouse, which permits **substantially shortened testing times.** Among tested dogs to date about 7% were mutation carriers and 3% homozygous for the mutated gene. The genetic test is required to achieve a specific breeding selection, which can eventually lead to the elimination of the harmful gene variant.

We now offer the following tests for the Labrador Retriever:

- Degenerative myelopathy (DM)
- Exercise Induced Collapse (EIC)
- Coat Colour (E-, B- and D-locus)
- Hereditary Nasal Parakeratosis (HNPK)
- Myopathy (cnm)
- Narcolepsy
- Progressive rod cone degeneration
- Progressive Retinal Atrophy (prcd-PRA)
- Pyruvate kinase deficiency (PK)
- Skeletal dysplasia (SD2).

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