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LABOGEN

genetic news

Hereditary skin diseases in dogs

Apart from many acquired and allergic dermatological diseases, there are many hereditary ones as well. In some instances the genetic cause is known: **Ichthyosis** is a congenital disorder of the normal desquamation of the skin and is caused by a defect in the keratinization of the epidermis. The name Ichthyosis is derived from Greek „ichthys“ meaning „fish“. This is due to the fact that affected dogs shed abnormally large, thickened, pigmented and scale-like areas of skin. Additionally, the skin itself can be variably pigmented. The first symptoms of the disease are visible within the first few weeks of life. Thus far, there is no known treatment. However the abnormal desquamation can decrease with age. The responsible mutation for **ichthyosis in the Golden Retriever** is known. Since the pathogenic gene variant shows a high prevalance in the dogs we have investigated (38% homozygous affected animals and 44% carrier animals) mating must be planned very precisely in this breed. A total exclusion of carrier animals from the breeding pool would extremely reduce the genetic diversity of the breed. However one of the mates should be free of the mutation. We can forward your samples for testing to our partnerlab.

Hereditary nasal parakeratosis (HNPK) in the Labrador Retriever causes the dog‘s nose to dry out. Especially on the dorsal part of the nose a dry, bark-like thickened layer of skin is formed that cannot be removed. This layer often cracks, becomes irritated and secondary bacterial infection is common. HPNK is not as prevalent as ichthyosis: 28% of the animals we have examined are carriers, 4% homozygous affected with the mutation. By the use of genetic testing successful eradication of this disease would be possible.

Dry eye and curly coat syndrome appears to be a problem unique to **Cavalier King Charles Spaniels**. It manifests in the form of a keratoconjunctivitis sicca (conjunctival / corneal inflammation due to a lack of tear production), as well as unusually rough and curly fur with hair loss which leads to pruritus. Abnormalities of the skin on the footpads and of the claws cause pain and lameness. The teeth are also affected. In addition, affected puppies appear smaller than their littermates. With 8% carrier animals and less than 1% homozygous affected dogs in the animals we have tested so far, this disease is not too common and should be controlled easily with genetic testing.

Progressive retinal atrophy in cats

The progressive retinal atrophy (PRA) affects the retina in the eyes of cats. A progressive degradation of the receptor cells leads to blindness in affected animals. To date several types are known in the cat:

The **rdAc-PRA** occurs among many cat breeds, including **Abyssinian, American Curl, American Wirehair, Bengal, Colorpoint, Cornish Rex, Munchkin, Ocicat, Peterbald, Siamese and relatived breeds, Singapura, Somali and Tonkanese**. The first symptoms appear between 1.5 and 2 years of age and lead to total blindness by the age of three to five years. This PRA form is an autosomal recessive trait, in contrast to rdy-PRA, which is inherited autosomal-dominantly. With **rdy-PRA** the first symptoms are already noticeable at about three weeks and affected cats go blind after about 7 weeks of age. This variant has been observed in **Abyssinian and Somali** cats.

Recently the research team led by Prof. Lyons and Dr. Gandolfi at the University of Missouri was able to find the genetic cause of PRA in **Persians**, also called **pd-PRA**. The degradation of photoreceptors starts at about 5 weeks of age, and the cats go blind by about 16 weeks of age. It‘s mode of inheritanc is autosomal-recessive, therefore only homozygous animals are affected. Genetic testing is possible in **Persians and related breeds**.

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

CH-4002 Basel • Riehenring 173 • Postfach
Tel. +41 61 3196060 • Fax +41 61 3196065
labor.basel@laboklin.ch

A-4040 Linz • Rosenstraße 1
Tel. +43 732 7172420 • Fax +43 732 717322
labor.linz@laboklin.at