

# NEWSLE<sub>2720</sub>T<sub>2</sub>ER

Coat colour expert Dr. Anna Laukner now in team LABOKLIN

MLS in Beagles

Research project about PHA, vWD and Haemophilia

TNS and dwarfism – now available on site

Controlled breeding using genetic tests?

#### **MLS in Beagles**

**The Musladin-Luëke Syndrome** (MLS) was first described in the early 1970's in beagles with a 2-3% incidence in the British and Australian populations. Later the disease would be named after the beagle breeders Musladin and Luëke.

This genetic disease is characterised by severe skin and joint fibrosis. The affected dogs are easily identifiable on hand external features such as their small build and tight skin as well as a broad skull with widely spread, slanted eyes and folded ears. The strongly reduced motility of their legs is noticeable, especially through the hopping, ballerina-like gait.

The first symptoms appear at the age of three weeks and worsen throughout the first year of life. After that the disease stabilises.

Recently, the research group of Bader (Lerner Research Institute, Cleveland) and Neff (University of California, Davis) determined the causative mutation. It is a point mutation in the ADAMTSL2 gene to which a strong association with MLS has been attributed.

Since the mode of inheritance is recessive only dogs that carry the mutation on both alleles get sick. In the mating of two heterozygous carriers, 25% of the offspring will be affected. Knowing the genetic status of dogs can insure a controlled breeding that prevents MLS-affected puppies from being born.

## Coat colour expert Dr. Anna Laukner now in team LABOKLIN

We are happy to announce to you that the renowned coat colour expert, Dr. Anna Laukner, has joined in collaboration with us at LABOKLIN. She is available to our customers immediately for any specific questions regarding fur colour in dogs and cats. As your contact person, she may be reached at labogen@laboklin.de.

### Research project about PHA, vWD and Haemophilia

#### Your help is needed!

After successfully undertaking and completing various projects to identify Haemophilia in numerous dog breeds, the genetic research in this field naturally continues. Therefore, we still gratefully accept samples from dogs with Haemophilia A or B as well as vWD (von Willebrand disease). Using the respective factor test, we verify the deficiency and search for the genetic origin.

Additionally, we have started a project about the Pelger-Huët anomaly (PHA). For this, we are also looking for affected animals and ones related to such animals.

Through our research, we hope to find the causative mutations. Tests developed using such information would allow breeders to screen animals and eliminate the disease via controlled breeding. If you would like to support us in our pursuits please contact us beforehand to discuss further procedures (labogen@laboklin.de or 0971-72020, Mrs Dipl.-Biol. A. Kehl).



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



NEWSLE<sub>120</sub>T<sub>2</sub>ER

Coat colour expert Dr. Anna Laukner now in team LABOKLIN

MLS in Beagles

Research project about PHA, vWD and Haemophilia

TNS and dwarfism – now available on site

Controlled breeding using genetic tests?

## TNS and dwarfism – now available on site

For many years now, we have offered the genetic analysis of Trapped Neutrophil Syndrome (TNS) in Border Collies as well as dwarfism in German Shepherds, Saarloos and Czechoslovakian Wolfhounds. Until now, these tests were performed by a partner lab of ours. Since the unveiling of the relevant mutations, the genetic tests have, at last, been established in our laboratory. Therefore, we are now able to provide you with an expedited and lower-cost processing.

#### Controlled breeding using genetic tests?

A major benefit of genetic tests is the ability to identify clinically sound carrier animals: something that is usually not able to be achieved using other lab test, such as determining the von Willebrand factor. The identification of genetically affected animals is possible even before the first signs of disease. This allows for selective monitoring and controlled feeding and keep, if necessary. How can one then use this knowledge for controlled breeding and to possibly eliminate the disease?

In general, it is essential to distinguish between recessive and dominant modes of inheritance. In dominant modes of inheritance, animals with only one mutated allele are affected, i.e. even heterozygous animals (N/X) fall ill. For example, this is the case in PKD in the Persian cat or Hereditary Cataracts in Australian Shepherds.

In a recessive mode of inheritance, only the homozygous animals are affected (X/X). This type of inheritance has been determined for most mutations. The clinically sound carrier animals (N/X) pass the mutated allele to 50% of their offspring. If both parents are carriers, clinically ill animals can result from the breeding of two clinically healthy animals. Therefore, controlled breeding can prevent such matings and the carrier animal (N/X) with a genetically healthy animal (N/N) results in further carrier animals, but no clinically ill animals. In order to counteract a shrinking gene pool, it is not advisable to eliminate all the heterozygous animals from the pool. If this did occur, it could promote the development of other diseases and the entire health situation of the breed could be compromised. Also, under certain conditions (avoiding pain breeding), even clinically ill animals can be bred; if they are paired with genetically healthy animals, only clinically healthy offspring will arise. In this way, desired traits of the affected animals can be kept in the breeding line.

In X-linked recessive diseases the special characteristic to note is that male animals only have one X chromosome (upon which the respective gene is located) and, statistically, half of the male offspring of a carrier female will fall ill, whereas, half of the female offspring are clinically healthy carrier animals.

**A good example of successful screening** and controlled breeding is PKD, a dominantly inherited kidney disease in Persian cats and in the homozygous form is embryonically lethal. In 2005, LABOKLIN identified 22% of the examined cats as heterozygous. This number declined continuously and by 2012 amounted to only 2% of examined animals. This shows that through genetic testing a considerable health improvement in pure-breds can be achieved.

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

www.LABOKLN.com

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