
GENETIC

NEWS

Cystinuria: News for what we know

It has been known, for many years, that Cystinuria is an inherited metabolic disease, which leads to urinary stones and urinary retention. Currently, it has been described in 70 breeds. In accordance to its heterogeneity, in respect to inheritance, genetic cause, frequency, severity, therapy and course of disease, the following classifications of disease have been recognized:



- **Type IA - autosomal recessive inheritance: Newfoundland, Landseer, Labrador**

Already, in 2002 genetic testing was developed for the Newfoundland and Landseer. An evaluation, of all animals (more than 1000 dogs) tested by LABOKLIN since 2006, resulted in the following genotype distribution: 75% wildtype homozygous, 24% carriers and 1% affected homozygous. In the more than 150 Landseers tested, only the wildtype genotype was detected. Genetic testing for Labradors wasn't available until 2013 and it is believed that mutation is quite rare.

- **Type 1B - autosomal dominant inheritance: Miniature Pinscher**

The underlying mutation was first found in dogs from Germany and neighbouring countries.

- **Type IIA - dominant inheritance: Australian Cattle Dog**

A genetic test has been available since 2013. However, as of yet, no data in regards to the frequency of the causative mutation has been compiled.

- **Type III - androgen-dependent expression: e.g. Mastiff, Bulldogs, Kromfohrlander and Irish Terrier**

In addition to stone analysis, a urinalysis for cystine crystals, casts and COLA content may be conducted in every breed. Additionally, genetic testing can be conducted for Type I and Type II in the before mentioned breeds.

In cooperation with Professor Dr. Giger

Prof. Dr. Urs Giger and Prof. Dr. Paula Henthorn of the University of Pennsylvania have recently been able to prove that a cystinuria Type III exists in Irish Terriers and the related Kromfohrlanders, as well as Mastiffs and Bulldogs. This type only affects non-castrated adult male dogs. Neutering an affected animal can reverse the cystinuria completely, thereby, healing the animal of disease. It is recommended that all intact male dogs be checked regularly for COLA content via urinalysis.

In a current study, Prof. Dr. Giger would like to examine to what extent, as well as when and how long "chemical castration" has a positive effect on COLA elimination. We are pleased to assist him in this endeavour by gathering urine samples before and periodically after "castration", as well as collecting respective blood and information about the dogs. Please contact us, in advance, if you are interested in contributing to this study, since we can only accept animals that are tested before and after chemical castration and from which we are able to obtain all other required information. Thank you!

Online application to LABOKLIN

Who is familiar with the difficulty of deciphering another persons' handwriting? It is easy enough to confuse a V for a U. Illegible handwriting has been a continuous source of errors at LABOKLIN, causing the names of animals or studbook numbers to be false and requiring post-dated corrections to occur. In order to eliminate this problem, we have redesigned our online request forms, so that they may be filled out electronically and printed afterwards. Reading and consequent written errors should soon be a thing of the past!



Scan the QR-code to get directly to the page with submission forms.

LABOKLIN on facebook

As of now, you are also able to find us on the social media network "Facebook". Our screenname there is: „Labogen – Die Genetik von Laboklin.“ Simply, click the "Like" button and you will not only be informed about developments in our laboratory, but also, you will be able to see photos from various trade fairs, as well as read answers to frequently asked questions.

