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Dear pet owner,

in your hands you find our first issue of a newsletter dedicated especially to you - to pet owners, resp. breeders of dogs and cats.

With the newly installed newsletter we want to inform you about new tests and trends concerning animal genetics. You will find interesting data e.g. on availability of tests as well as on clinical relevance, handling procedures and turnaround times.

We hope you will find the newsletter both informative and helpful. Of course any comments or suggestions are most welcome - please send back an email to labogen@laboklin.de to let us know.

Kind regards,

Dr. Elisabeth Müller

### EIC - LABOKLIN acquires an exclusive license for European countries

Exercise Induced Collapse (EIC) is a neuromuscular disease affecting Labrador Retrievers and closely related breeds. Affected animals develop signs within 5 to 15 minutes after start of intensive training or stress. Typical signs are weakness of muscles and collapse thus sometimes resembling epileptic fits or even myasthenia gravis. Even though age of onset is reported mostly to be the early adult age EIC can stay a hidden disease as long as no intensive training is carried out and stress to the animal is limited. Up to now the mutation is detected in Labradors as well as in Chesapeake-Bay and in Curly-Coated Retrievers. Just recently a mutation in the DNM1 gene was recognized to be responsible for the disease. This work was done by Professor James Mickelson and his coworkers at the University of Minnesota. Professor Mickelson kindly agreed to pass on an exclusive license to LABOKLIN for Europe so we can offer the test now for all animals within the European countries.



### GM2 - Gangliosidosis

GM2 Gangliosidosis is a storage disease similar to Sandhoff disease in humans in which an enzymatic defect results in intracellular (lysosomal) accumulation of lipids. Affected animals will most prominently show clinical signs of neurologic disorders like head shaking, impaired coordination of leg movement resulting in paralysis. In GM2 Gangliosidosis an early onset is reported often beginning at the age of two months and characterized by rapid progression.

The mutation in the HEXB gene responsible for the impaired enzyme action in GM2 Gangliosidosis in Burmese cats is identified now as result of intensive work by Dr. Douglas Martin and his fellow scientists at Auburn University. Thus a genetic test to identify affected - and more important - carriers is at hand now. LABOKLIN has an exclusive license at hand to carry out the genetic tests throughout Europe. By now the test is performed routinely - please contact us in case you need more information.

### Buccal swabs- sufficient for genetic tests?

Buccal swabs are a preferable sampling device whenever you want to test e.g. very young animals. Quite often we are asked if there is any loss of reliability when tests are performed using buccal swabs. Indeed that is not the case. Evaluation of our data show, in 2008 we were able to achieve a result in 99,93% of buccal swabs from cats and > 92% of swab taken from cats. Thus buccal in our hands are appropriate samples. What can be affected, when swabs are taken instead of blood samples? This is a constant worry of pet owners as well. As all tests for hereditary diseases are based on analysis of DNA the sample has to contain sufficient DNA, that is sufficient number of cell nuclei. That is always the case in blood samples and almost always in swabs (see above). So, swabs are as good as/nearly as efficient as blood samples. In case we do not get sufficient amount of DNA to work with we will obtain no result at all. We will report that we could not obtain a result. So with swabs there is no insecurity in the obtained result at all - there is only a slight risk in having to send a second sample.



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### Highest possible quality standards performed by LABOKLIN

Highest possible quality standard has always been our goal. Consequently of course we work according to DIN EN ISO/IEC 17025, thus are subject to external control at regular intervals and all processes can be subject to inspection at any time. We top that by routinely processing our samples in duplicates. Only results approved by an independently performed second run will be reported to the customer. And of course we are member of the International Society of Animal Genetics (ISAG) and design and perform check studies with other labs to round up our control scheme.

Since we are member of the ISAG DNA profiles for identity and parentage tests are performed using the markers recommended by ISAG. This means: every profile established by us can be used for any comparison with any profile established by any other lab using ISAG markers. This is especially interesting when breeding across the borders results in parents with profiles run by different labs in different countries. Our special service: once we have run a profile we store DNA without additional costs for 10 years. So in case there is additional test demand (e.g. a new test for a hereditary disease is established) you do not need new samples to be taken. If needed DNA data bases can be established as well.

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### An update on MDR1 gene defect

Side effects to Ivermectin which is widely used for control of ectoparasites are known to be caused by a defect in the multidrug transporter MDR1 situated in many cell membranes. Due to a mutation in these animals the transporter loses its ability to hold back substances like Ivermectin, Loperamide just to name some of the drugs affected resulting in accumulation in the brain. While treated with these drugs affected animals will show signs of intoxication. Only intensive treatment and supportive care can prevent death. A list of substances that are suspect to be transported by the same carrier and thus will probably accumulate in the brain of genetically affected animals despite state of the art dosage is listed on our website [www.laboklin.de](http://www.laboklin.de). Also we have listed the breeds today known to be affected and the percentage of animals carrying the mutation. Of interest should be that as long as alternative therapeutic concepts are used even the affected animals can live a perfectly healthy life. Knowledge of the status in animals of these breeds will help choosing the appropriate drug in case of any disease. Furthermore there is evidence that even animals reported to be just carriers will have a reduced efficacy of the drug transporter MDR1 resulting in slightly impaired membrane barriers.

MDR for dogs

