LABOKLIN
LABORATORY FOR CLINICAL DIAGNOSTICS

Newsletter | December 2019

GENETIC NEWSLETTER

Stargardt disease (STGD)
in Labrador Retriever

Stargardt disease is an inherited retinal degenerative disease that leads to visual impairment and blindness in both human and dogs. Photoreceptors are light-sensing cells found in the retina of the eye. There are two types of photoreceptors: rods and cones. Both rods and cones work together to detect light and convert it into electrical signals, which are then "seen" by the brain. Rods are found in the outer retina and help us see in dim and dark lighting. Cones are found in the macula, which is the part of the retina at the back of the eye and help us see fine visual detail and colour. In Stargardt disease both cones and rods degenerate, but for unclear reasons, cones are more strongly affected in most cases. The mutation responsible for Stargardt disease in Labrador retriever has been identified by researchers at The Swedish University of Agricultural Sciences and a test is now available at Laboklin.

More breeds added to the Lafora DNA test

The Lafora DNA test has recently been validated for more breeds and the test is now valid for: Basset, Beagle, Cardigan Welsh Corgi, Chihuahua, French Bull Dog, Miniature Wire haired Dachshund, Pembroke Welsh Corgi.

Progressive Retinal Atrophy (NECAP1 PRA / NECAP PRA5) *in Giant Schnauzer*

NECAP1 is a novel form of PRA implicating the Giant Schnauzer breed. The disease is caused by a mutation in the NECAP1 gene. Like other forms of PRA this disease leads to retinal degeneration and subsequent blindness. The variant was identified by scientists at the Animal Health Trust and it is thought to be autosomal recessive. A test is now available at Laboklin. Please note that Giant Schnauzer can also be affected by the prcd PRA form of PRA which is also available at Laboklin.

Hypophosphatasia (HPP) *in Karelian Bear Dog*

Canine hypophosphatasia (HPP) is a severe inherited skeletal disorder affecting the Karelian Bear Dog (KBD). Hypophosphatasia is a metabolic bone disease characterised by impairment in skeletal mineralization which leads to variable presentation of skeletal hypomineralization, growth retardation, seizures and movement difficulties.

Upper airway syndrome (UAS)
in Norwich Terrier

Upper airway Syndrome is an inherited disease caused by a mutation in the ADAMTS3 gene and characterised by airway oedema which leads to fluid retention in the tissue that lines the airways and makes it more likely that dogs with the mutation will develop breathing obstructions. Dogs homozygous for the mutation (UAS/ UAS) are at significantly higher risk of developing the disease than dogs which are heterozygous (N / UAS).

Copper storage disease /Copper toxicosis (CT)
** in Doberman and Labrador Retriever*

Copper homeostasis is (among other factors) regulated by the uptake of copper in the small intestine and the excretion of surplus copper via the biliary tract. In the breeds Labrador Retriever and Dobermann, dogs with copper toxicosis have a decreased ability to excrete copper resulting in excessive copper storage in the liver and other organs, leading to liver damage and cirrhosis. The disease is known to have a relatively late onset (middle aged or older dogs) with variable symptoms like weight loss, lethargy, weakness, vomiting, diarrhea, abdominal pain and neurological dysfunction.

Inflammatory Pulmonary Disease (IPD)
in Rough and Smooth Collies

We are pleased to announce that in cooperation with professor Leeb of the University of Bern in Switzerland, Laboklin is now able to offer a DNA test for the detection of the mutation which causes Inflammatory Pulmonary Disease (IPD) in Collies and the test is now available for ordering. Inflammatory Pulmonary Disease (IPD) is an inherited lung disease affecting the Collie breed and characterised by recurrent pneumonia, clinical symptoms were seen when affected puppies were only few days old and include foamy vomiting, shallow breathing, cough, increased breathing sounds and fever. Affected dogs responded to therapy with antibiotics and secretolytics, but tended to relapse quickly without antibiotic treatment.

Retinal dysplasia (RD) / Oculoskeletal Dysplasia (OSD 3) *in Northern Inuit and Tamaskan*

Oculoskeletal Dysplasia (OSD3) is a genetic disease characterised by Skeletal and ocular defects including cataracts, retinal detachment and primary achondroplasia (dwarfism).

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Coat Colour
I - Locus (phaeomelanin intensity)

I-Locus controls the intensity of the red pigment (phaeomelanin). Phaeomelanin is the tan pigment including all shades of red, gold, fawn, sable and cream pigments. The richness of the red colour varies in the different breeds and within a breed, from the very rich red of the Irish setter to cream. This intensity is controlled by the I-Locus which has recently been identified.The dominant I-allele stands for intense phaeomelanin (red, orange, yellow), the recessive i-allele is responsible for for cream, cream-white or white. The expression of the phaeomelanin on the dog body, is initially determined by the loci E, K and A locus. The I-Locus decides how bright or rich the red colour is. A test is now available at Laboklin.

update to Laboklin Coat Colour Bundle

The bundle now includes: A + B + D1 + E1 + I + K + S
In order to offer a comperhensive coat colour analysis we have bundled the most important coat colour DNA tests in one simple bundle which provides in depth analysis of your dog's coat colour genes. The following tests are now included: A Locus (Agouti AY, aw, at and a), Chocolate B Locus (bd, bc, bs and bd), Dilution (D1), E Locus (E1 cream, red, chocolate, apricot), I Locus (intensity new), K- Locus and S Locus (piebald, spotted white).

Raine Syndrome has been added to Laboklin Border Collie DNA bundle

The Border COllie bundle has now been updated and includes the DNA test for Raine Syndrome which is relevant to the breed. The bundle now includes: Collie Eye Anomaly (CEA) Option 1*, Cobalamin Malabsorption (Imerslund-Gräsbeck syndrome (IGS)), MDR1 Gene Varian / Ivermectin Sensitivity *, Sensory Neuropathy (SN), Neuronal Ceroid Lipofuscinosis (CL / NCL), Trapped Neutrophil Syndrome (TNS), Glaucoma and Goniodysgenesis (GGD), and Raine Syndrome.

new Portuguese Waterdog DNA Bundle

(GM1 + prcd PRA + Improper Coat)

- GM1-Gangliosidosis
- Progressive Retinal Atrophy (prcd-PRA) Option 1
- Furnishings and Improper Coat

new Bullterrier DNA bundle

(Bullterrier and miniature Bullterrier)

- Lethal Acrodematitis (LAD)
- Primary Lens Luxation (PLL)
- Larynx / Laryngeal Paralysis (LP)
- Polycystic Kidney Disease (PKD)

update to the Golden Retriever DNA bundle

Neuronal Ceroid Lipofuscinosis (CL / NCL) has now been added to the Golden Retriever DNA bundle. The bundle now includes:

- Progressive Retinal Atrophy (GR-PRA1) KC
- Progressive Retinal Atrophy (GR-PRA2) KC
- Progressive Retinal Atrophy (prcd-PRA) Option 1 KC
- Ichthyosis * KC
- Muscular Dystrophy (MD), and
- Neuronal Ceroid Lipofuscinosis (CL / NCL)

update to the Rottweiler DNA bundle

Hyperuricosuria (HUU / SLC) has been replaced with Leukoencephalomyelopathy (LEMP) which is more relevant to the breed. The bundle now includes:

- Degenerative Myelopathy / DM (Exon 2)
- Leukoencephalomyelopathy (LEMP)
- Juvenile Laryngeal Paralysis & Polyneuropathy (JLPP) KC
- Coat (hair) Length I,
- X-linked Myotubular Myopathy (XLMTM)

update to the Collie DNA bundle

Hyperuricosuria (HUU / SLC) has been replaced withInflammatory Pulmonary Disease (IPD), which is more relevant to the breed. The bundle now includes:

- Collie Eye Anomaly (CEA) Option 1* KC
- Degenerative Myelopathy DM (Exon 2) KC
- Inflammatory Pulmonary Disease (IPD)
- MDR1 Gene Variant KC
- Progressive Retinal Atrophy (rcd2-PRA) Option 1 KC

new Shiba Inu DNA bundle

(GM1 , GM2 , A Locus and E Locus)

- GM1-Gangliosidosis
- GM2 Gangliosidosis Variant 0 (Sandhoff Disease)
- Coat Colours: A-Locus Agouti (fawn, sable, black and tan/tricolor, recessive black), and
- Coat Colours: E-Locus E1 (yellow, lemon, red, cream and appricot)

new Australian Kelpie can now be tested for Collie Eye Anomaly (CEA)

The Collie Eye Anomaly (CEA) DNA test has now been validated for Australian Kelpie.

New Kennel Club DNA testing Schemes

- ☐ Neuronal Ceroid Lipofuscinosis (CL / NCL) *in English Setter and Saluki.*
- ☐ Progressive retinal atrophy (rcd4-PRA) / LOPRA *in English Setter and Standard Poodle*
- ☐ Pug Dog Encephalitis (PDE) / Necrotizing Meningoencephalitis (NME) *in Pug*
- ☐ Progressive Retinal Atrophy (prcd-PRA) *in Standard Poodle*
- ☐ Mucopolysaccharidosis type IIIb (MPS3b) *in Schipperke*
- ☐ Dilated cardiomyopathy (DCM) *in Standard Schnauzer*
- ☐ LEMP and LPN2 *in Leonberger*

** partner lab*