LABOKLIN
LABORATORY FOR CLINICAL DIAGNOSTICS

Newsletter | February 2019

GENETIC NEWSLETTER

Crufts 2019

We are very pleased to announce that we will be at Crufts this year, which is always a fantastic event that we look forward to. It is always pleasure meeting breeders whether they come to drop off a sample, to have a chat about a test, suggest a new bundle, enquire about coat colour inheritance or just to say hi. Crufts is always a great opportunity to to talk to breeders about their testing requirements and to see what we can do to help. Again our ever popular 10% Crufts discount will be available this year too. Visit us at Crufts 2019, Hall 3 Stand 7A.

More breeds added to the Lafora DNA test

The Lafora DNA test has recently been validated for two more breeds: Basset and Chihuahua. The test is currently part of the Kennel Club DNA testing scheme with Laboklin in Beagle and Miniature Wirehaired Dachshund.

Mycobacterium Avium Complex sensitivity (MAC) *Miniature Schnauzer*

While dogs are generally resistant to Mycobacterium avium infections, this mutation has been found to be associated with a genetic predisposition to infection with systemic avian tuberculosis in Miniature Schnauzers (referred to mycobacterium avium complex or MAC). The mutation is likely to cause impairment to the dog's immunodeficiency. Common symptoms of affected dogs are: lethargy, inappetence, weakness, nasal discharge, conjunctivitis, diarrhea, lymphadenopathy, hepatomegaly and splenomegaly. The age of onset varies between one and eight years. Because of the underlying immunodeficiency, the infections are poorly responsive to treatment and often recur. Since MAC is a zoonotic disease, humans with a suppressed immune system could be also at risk for an infection. A test is now available at Laboklin.

Long coat in French Bulldog

Two tests are now required to determine the genotype of coat length in Frencg Bull Dogs. We offer both tests either separately or as a bundle Coat Length I + II (test number 8737).

Chondrodystrophy (CDDY with IVDD Risk) and Chondrodysplasia (CDPA)

in many breeds

The test checks for two mutations: CDDY with IVDD Risk, and CDPA.

Chondrodystrophy CDDY (FGF4-18) which causes short legs and the risk of developing Intervertebral Disc Disease (IVDD).

Chondrodysplasia CDPA (FGF4-12), which causes the short legged phenotype in a number of breeds.

Chondrodystrophy (CDDY with IVDD Risk) is a trait that is common to many dog breeds and it is characterised by shorter legs due to shorter long bones. CDDY can also be associated with Intervertebral Disc Disease (IVDD) due to premature degeneration of the intervertbral disc. The intervertebral disc lie between the vertebrae and it is made of a cartilage which separate vertebrae from each other, absorb shocks and allow slight movement of the vertebrae. In affected dogs, premature calcification of part of the disc at early age (from birth to 1 year of age) results in degeneration of all discs in young dogs. These abnormal discs are susceptible to herniation into the spinal canal where the inflammation, and hemorrhage can cause severe pain and neurological dysfunction. CDDY is inherited as a semi-dominant trait which means that dogs with 2 copies of the mutation are smaller than dogs with only 1 copy. As for IVDD, the inheritance follows a dominant mode, meaning that 1 copy of CDDY mutation is sufficient to predispose dogs to IVDD.

The CDDY mutation has been found in breeds such as: Basset Hound, Beagle, Bichon Frise, Cardigan Welsh Corgi, Cavalier King Charles Spaniel, Chesapeake Bay Retriever, Chihuahua, American Cocker Spaniel, Coton de Tulear, Dachshund, Dandie Dinmont Terrier, English Springer Spaniel, French Bulldog, Havanese, Jack Russell Terrier, Nova Scotia Duck Tolling Retriever, Pekingese, Pembroke Welsh Corgi, Poodle (Miniature and Toy), Portuguese Water Dog, Scottish Terrier, Shih Tzu.

The second mutation CDPA explains the short-legged phenotype known as chondrodysplasia (CDPA) in breeds such as Basset Hound, Pembroke Welsh Corgi, Dachshunds, West Highland White Terriers and Scottish Terriers. CDPA inheritance is considered to follow an autosomal dominant mode.

In some breeds both mutations are present and so breeders will be able to plan breeding to reduce occurrence of CDDY, while retaining the short-legged phenotype CDPA.

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new breeds added to prcd PRA

Bearded Collie, Jack and Parson Russell Terriers

The prcd PRA mutation has been found in three more breeds: Bearded Collie, Jack Russell Terrier and Parson Russell Terrier. We currently don't have information about the prevalence of the mutation in these breeds.

Neuroaxonale Dystrophie (NAD)

is now valid for Rottweiler in addition to Papillon and Spanish Water Dog

Neuroaxonal Dystrophy is an uncharacterized juvenile-onset genetic disorder. Affected dogs exhibits various neurological deficits including gait abnormalities and behavioral deficits. Symptoms include slowly progressing neurological signs starting between six and eleven months of age. Owners of affected dogs reported gait abnormalities, behavioral changes and incontinence alone or in combination with uncontrolled defecation.

more DNA breed specific bundles

Laboklin DNA bundles are becoming increasingly popular and highly demanded by breeders. Each bundle includes a selection of the most important and relevant tests for the breed at a discounted price. We have just added 5 more bundles and more will follow.

new Australian Cattle Dog DNA bundle

(DM Exon 2 + NCL + PLL + prcd PRA 1 + rcd4 PRA)

- Degenerative Myelopathy DM (Exon 2)
- Neuronal Ceroid Lipofuscinosis
- Primary Lens Luxation (PLL)
- Progressive Retinal Atrophy (prcd-PRA) Option 1 KC
- Progressive retinal atrophy (rcd4-PRA) / LOPRA

new Dachshund DNA bundle

(CDDY&CDPA + OI + NCL + Cord 1 PRA + crd PRA)

- Chondrodystrophy (CDDY with IVDD Risk) and CDPA.
- Brittle Bone Disease (Osteogenesis Imperfecta)
- Neuronal Ceroid Lipofuscinosis (CL/NCL)
- Progressive Retinal Atrophy (cord1- PRA/crd4 PRA) KC
- Progressive Retinal Atrophy (crd PRA)

new English Springer Spaniel DNA bundle

(AMS+ FN + SPS + Fuco + PFKD + Cord 1)

- Acral Mutilation Syndrome (AMS) KC
- Familial Nephropathy (FN)
- Hypomyelination (Shaking Puppy Syndrome) SPS
- Fucosidosis KC
- PFK Deficiency KC (Phosphofructokinase deficiency)
- Progressive Retinal Atrophy KC (cord1- PRA / crd4 PRA)

new Welsh Corgi DNA bundle

(Short tail + CDDY&CDPA + DM Exon 2 + rcd3 PRA + vWD I)

- Brachyury (Bobtail Gene / Short Tail)
- Chondrodystrophy(CDDY with IVDD Risk) and CDPA.
- Degenerative Myelopathy DM (Exon 2)
- Progressive Retinal Atrophy (rcd3 PRA) KC
- von Willebrand disease Type I (vWD I).

new Cocker Spaniel DNA bundle

*AMS + FN + prcd PRA option 1**

- Acral Mutilation Syndrome (AMS) KC
- Familial Nephropathy (FN) / Hereditary Nephropathy * KC
- Progressive Retinal Atrophy (prcd-PRA) Option 1* KC

Another mutation added to the B-Locus coat colour test panel

In addition to the three known genetic variants for the brown coat colour, another mutation has now been identified and has already been added to our test panel. The new mutation is named b4 and so far has only been found in Australian Shepherd. Our panel will check for this variant in all breeds although we yet have no information about its prevalence in other breeds. Please note that there still unknown mutation(s) that cause the brown coat colour in some breeds such as French bulldog which cannot yet be tested.

prcd PRA option 1 and CEA option 1

are now accepted by the Kennel Club

We can now send results of our partner lab DNA tests: prcd PRA option 1 and CEA option 1 to the Kennel Club to be recorded and published as part of the official DNA testing scheme.

New Kennel Club DNA testing Schemes

- ☐ Sensory Neuropathy (SN) *in Border Collie*
- ☐ Neuroaxonale Dystrophy (NAD) in Papillon

* partner lab