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GS12 - KO11210

PDP1 Test (Sussex and Clumber spaniels)

Name	Breed	Result
Spice Twice Brother Lustig	Clumber Spaniel	Normal
Reg. No FI10287/10	Chip 985121017355107	*Microchip verified.

The test results will be reported as follows:

Normal: The dog carries two copies of the normal PDP1 gene and has therefore no increased risk of suffering from exercise intolerance.

Carrier: The dog carries one mutant and one normal copy of the PDP1 gene. The dog does not develop the syndrome but can transfer the defect to approximately 50% of its offspring.

Affected: The dog carries two mutant copies of the PDP1 gene and will develop the disease.

Exercise intolerance syndromes are well known to be associated with inborn errors of metabolism affecting glycolysis (phosphorylase and phosphofructokinase deficiency) and fatty acid oxidation (palmitoyl carnitine transferase deficiency). A few years ago, Cameron et al. identified a canine model for profound exercise intolerance caused by a deficit in PDP1, the phosphatase enzyme that activates the pyruvate dehydrogenase complex (PDHc). The mutation is present in 20% of the dogs in Clumber and Sussex Spaniel breeds. Homozygosity for the mutation produces severe exercise intolerance.

On behalf of Genosoper Oy

Helsinki 23.5.2012

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