



Alpha-L-Fucosidase Deficiency

The fucosidosis is a **lysosomale disease** (Disease of red blood cells) inherited from storage due to a deficiency of the protein called **Alpha-L-Fucosidase**. The deficiency pulls an accumulation of fucoses containing sphingolipides, glycoproteins, and mucopolysaccharides (glycosamonioglycans) in lysosomes (red blood cell). This disease pulls neuromuscular disorders, deteriorates the infections which the animal can catch and affects gradually the nervous function. It exists at the human and was put in evidence to the pedigree dog Springer Anglais. The responsible gene is present on a region of the DNA which contains a large number of Cytosine Guanine. The DNA test is the best means to check the disease in the race and to eradicate it of the race of English Springer.

To realize the DNA screening of this disease, a simple oral smear or a blood test allows us to make the analysis. On simple request of your part, we send you a free of charge kit of oral or blood taking. At reception of your taking in the laboratory, only 10 working days are enough so that you have your results by email.

For more information, do not hesitate to contact us !

PhosphoFructoKinase Deficiency

The PhosphoFructoKinase Deficit is an **autosomic recessive genetic disease which prevents the metabolism of the glucose in available energy**. This one pulls weaknesses and muscular cramps, discolored urine, anaemia and jaundice. The deficiency in PFK also destroys red blood cells to the affected dogs, leading to the anaemia. The gene of the deficiency of frequency PFK in Cocker spaniels is estimated at 10 % of the population. Because the disease is autosomic recessive, the dogs which are carriers of the disease present no sign of deficiency in PFK, but are statistically 50 % of chance to pass on the gene in their descent. There is an important frequency of this defect, so the only effective treatment for the deficit in PFK is the screening and the selection.

Based on searches led to the University of Pennsylvania, GENINDEXE developed a genetic test based on the DNA. The used method is the PCR, it is an extremely reliable method, that allows to define if the dog is not carrier, carrier or affected.



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