GSD4 test

Glycogen Storage Disease

in Norwegian Forest Cat

The glycogen storage disease, a metabolic disease

The glycogen storage disease type IV (GSD IV) is an inherited disorder of glucose metabolism. In Norwegian Forest Cat, GSD IV is due to an inherited deficiency of an enzyme of glycogen synthesis called GBE (Glycogen Branching Enzyme). In affected cats, an abnormal form of glycogen (a polymer of glucose) is stored in most tissues.

Most affected kittens die at birth or soon after birth because they are not able to produce enough glucose during the birth process and the first hours of life. Rarely, affected kittens could live normally until 5 months of age, but the disease leads quickly to neuromuscular degeneration, to severe muscular atrophies, to cardiac failures and to death before 15 months of age.

The glycogen storage disease, an inherited disorder

The GSD IV is inherited as an autosomal recessive trait in the Norwegian Forest Cat. The DNA test supplies an answer among three possible situations:

DNA test result	The cat is	Genetic status	Will develop the disease?	Will transmit the genetic anomaly?
Normal homozygous	Clear	2 normal copies of *GBE1* gene	NO	NO
Heterozygous	Carrier	1 normal copy and 1 defective copy of *GBE1* gene	NO	YES statistically to 50% of its progeny
Mutated homozygous	Affected	2 defective copies of *GBE1* gene	YES neonatal mortality, potential survival until 15 months	Not able to reproduce

Using this DNA test, breeders are able to detect very early carriers cats, to select breeding animals and to adapt breeding strategies in order to reduce neonatal mortality due to GSD IV, to avoid producing affected kittens, to avoid spreading the genetic defect in their lines or in the breed.

The GSD4 test, a reliable and validated test

Pr John Fyfe from the University of Michigan (USA) identified the GBE1 gene and the mutation causing GSD IV in Norwegian Forest Cat (Fyfe et al. 2007). In collaboration with Pr John Fyfe, ANTAGENE developed a DNA test and validated it on carriers and affected Norwegian Forest Cats.

The GSD4 test detects directly the causative mutation in the GBE1 gene.

The frequency of GSD IV carriers is 12,2% in Europe (data from ANTAGENE, October 2007) and around 15% in the USA.

The DNA test is reliable, easy to do from a cheek swab sampling (cytobrush), can be done at any age even in young kittens and is done only one time in the animal life.

Ordering

Request by email sampling kits, which are sent by post mail. Sample is realized and validated by a veterinarian in order to deliver a genetic certificate. Samples are sent to ANTAGENE.

For more information on our services and researches on dogs and cats, please consult our web site: www.antagene.com

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