

Glycogen Storage Disease type 4 (GSDIV)

What's GSDIV ?

Glycogen Storage are disease of metabolism, that means that is they result from a defect in the chain of chemical reactions which transform sugars into useful energy. These sugars or carbohydrates are stored in cells in the form of glycogen. During the physical exercise, the glycogen is normally transformed into useful energy by muscles, thanks to a series of biochemical reactions which bring in several enzymes. If one of them lack, the glycogen accumulates in cells without being able to be used.

Symptoms :

In the most common shape, kittens die in the birth or after bus they are incapable to produce enough glucose necessary for the birth and at the first hours of the life. More rarely, kittens can live normally until 5 months but the disease quickly leads to neuromuscular degenerations, to muscular atrophies, to heart failures and to death of the animal before 15 months.

Transmission :

For the Norwegian forests cat, the glycogénose of type IV is a hereditary illness monogenic (a single implicated gene) autosomale (males as females are concerned) recessive (the kitten has to inherit from the allèle of the gene moved by each of his two relatives to be affected).

Diagnosis :

Pr John Fyfe of the university of Michigan (USA) identified the gene and the transfer implicated in this disease (Fyfe and al. 2007). According to him, GENINDEXE propose screening test of the disease based on the detection of the transfer of the gene GBE1. de la maladie. According to the result of the test, 3 situations are possible : wild Homozygote (+ / +), Heterozygote (+/-) or moved Homozygote (-/-).



Interpretation of results :

In genetics the code to indicate the wild copy (allèle) of a gene is "+" and on the contrary the code to indicate the copy (allèle) moved by a gene is "-".

So, after a screening test GSDIV, the status of a cat can be or:

- + / + **Homozygote normal - not carrier of GSDIV, will never pass on the transfer**
- / + **Heterozygote - Carrier of GSDIV, the probability of transmission of the transfer is 50 %**
- / - **Moved homozygote - Affected by GSDIV, passes on the transfer in 100 % of the cases**

To optimize the organization of your reproduction, please consult the chessboard of crossing below :

		Father						
		Not carrier		Carrier		Affected		
		+	+	+	-	-	-	
Mother	Not carrier	+	+/+	+/+	+/+	+/-	+/-	+/-
		-	Not carrier	Not carrier	Not carrier	Carrier	Carrier	Carrier
	Carrier	+	+/+	+/+	+/+	+/-	+/-	+/-
		-	Not carrier	Not carrier	Not carrier	Carrier	Carrier	Carrier
	Affected	+	+/-	+/-	+/-	-/-	-/-	-/-
		-	Carrier	Carrier	Carrier	Affected	Affected	Affected

References

Fyfe JC, Kurzhals RL, Hawkins MG, Wang P, Yuhki N, Giger U, Van Winkler TJ, Haskins ME, Patterson DE & Henthorn PS. (2007) A complex rearrangement in GBE1 causes both perinatal hypoglycemic collapse and late-juvenile-onset neuromuscular degeneration in glycogen storage disease type IV of Norwegian forest cats. *Mol Genet Metab.* 2007 Apr;90(4):383-92.