



**Genindexe**

La génétique à votre service

## Pyruvate Kinase Deficit (PK Def.)

### What is Pyruvate Kinase ?

Pyruvate kinase (PK) is an essential enzyme to the production of energy in the erythrocytes (red blood cells). If the erythrocytes are deficient in PK they don't succeed in maintaining the metabolism of the normal cells and, consequently, are prematurely destroyed. This deficiency shows itself as a hemolytic anaemia of variable gravity.

### Pyruvate Kinase Deficiency :

The deficiency in PK was described on Abyssin and Somali races. The feline disease differs from the canine disease by the fact that the got cats have a normal life expectancy, presents an anaemia only in an occasional way, and do not seem to develop either osteosclerose or hepatic incapacity.

The clinical signs of the disease reflect the inanimate state of the animal and include an intolerance in the physical exercise, the generalized weakness, the cardiac breath and the splenomegaly (increase of the volume of the spleen).

### Transmission :

PK Def is transmitted according to a recessive autosomic mode. Heterozygotes (carriers) don't present any clinical signs of the disease and live a normal life. However, they can transfer the mutation in population and it is more important that these animals are revealed before the reproduction.

The deficit in PK can be detected by means of the tools of the molecular genetics.

These DNA tests identify at the same time affected animals and carriers animals.

### Interest of the DNA test :

The genetic defect leading to the disease was identified. The genetic anomaly directly responsible of this disease is detected by the DNA test. This method allows a very big precision and the analysis can be made at any age. It offers the possibility to make the distinction between no carriers animals and affected but especially to identify carriers. It is an essential information to control the disease because carriers are capable of propagating the transfer in the population, but cannot be identified by leaning on the only clinical signs.

### Expression of results and meaning :

affectedIn genetics the code to indicate the normal copy ( allèle ) of a gene is "+" and on the contrary the code to indicate the affected copy ( allèle ) 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 :

