

# Interpretation of the results

## How to read the scientific report ?

In genetic, results are always expressed in the following way :

**+ / +** If the animal is "normal" thus carrier of the normal shape of the gene, we say that it carries two copies of the "normal" allèle or "**wild type**" ("**wt**") **in english**, He is thus " homozygote normal " (Generally, we say that its screening is negative because absence of the affected allèle),

**+ / -** If the animal is carrier of a normal allèle (or "**sauvage**" ou "**wt**" ou "**+**" ) and of a affected allèle "-"; he is said heterozygote (we say that the screening is positive because presence of the affected allèle),

**- / -** If the animal is carrier of two affected allèles "-" or forms moved by a gene, is said moved homozygote.

As for the PKD, can exist only the forms " + / + " thus "normal" or " + / - " thus " carrier and affected ". In that case handbook, the shape "- / -" does not exist because animals are not viable.

Both boards below have not only for objective to illustrate our comments but also to inform you about the consequences of the use of a carrier cat for the reproduction.

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

			Probabilités			
		Père	Mère	Sain	Atteint	Non viable
S t a t u t	Sain	Sain	Sain	100%	0%	0%
	Porteur	Sain	Sain	50%	50%	0%
	Sain	Porteur	Porteur	50%	50%	0%
	Porteur	Porteur	Porteur	25%	50%	25%



## PKD and breeding (Source : [www.lorien-cats.com](http://www.lorien-cats.com))

« It is completely possible to make reproduce a positive cat (carrier) the lineage and/or the exceptional qualities of which we do not want to lose : It will be necessary to marry this cat with a (healthy) negative cat. In statistics, 50 % of kittens will be negative (healthy). Indeed on as soon as will be obtained the (healthy) negative quality kittens it is deeply advised to keep them and to sterilize the positive parent (carrier). As breeder, I advise you to test all your cats with genetics tests, including those born at home. Echographies gave rise regrettably to many errors.

## Screening of Polycystic Kidney Disease

### POLYCYSTIC KIDNEY DISEASE (PKD)

The **polycystic kidney disease** is a frequent genetic disease on different felines races, in particular the Persian and the similar races. Frequency of the genetic abnormality was esteemed for four races : Persian (26%), Exotic Shorthair (37%), British Shorthair (12%), Maine Coon (1%), but rest still unknown for the other races of cats.

This affection is translated by the progressive development of cysts in loins, by the destruction of the renal tissue, by the incapacity renal being able to provoke the premature death of the animal.

The renal dysfunction appears between 2 and 10 years according to Growth rate of the renal cysts. The opposite photo watches the loins of a Persian Colourpoint male cat of 7 years after its death for cause of PKD. The size of loins is 5 times superior to the normal.



The renal polykystose is a monogenic disease which is passed on according to an autosomal mode dominant. That is it implies a single gene, that is it is not connected to the sex (as color-blindness at the man) and that the presence of a single copy of the defective gene is enough to provoke the disease.

**Before the existence of a genetic test, the PKD was generally diagnosed by allowing ultrasound the detection of the disease at a relatively early stage.** This method visualizes by imaging the presence of cysts on loins. This technique is effective on the condition of being realized by an experimented staff and with an adapted equipment.

The **Dr Leslie LYONS's team of the University of Californie in Davis** (Not far from Sacramento in the North of California) identified the responsible gene then the transfer provoking the genetic defect. A DNA test was developed and validated with the aim of determining, from the birth, if a cat is healthy or achieves of PKD from a simple oral smear or from a blood test (reliability superior to 99 %).

Two situations are then possible :

- **+ / +** : Homozygote normal, the cat carries two normal copies of the gene : he is healthy, does not develop the disease and does not pass on the genetic abnormality in his descent,
- **- / +** : The cat is said "heterozygote" that is it carries a normal copy and a defective copy of the gene (transfer responsible for the disease): He's affected, develops the disease more or less prematurely and the probability of transmission in its descent is 50 %.

The early screening of a cat achieves of PKD (before it develops the clinical signs) allows to adapt his food and to apply possibly certain preventive treatments to delay and limit the development of the renal insufficiency. The early screening also allows to select the breeders, to adapt the crossings to avoid producing kittens affected in the descent and propagating the disease in the breeding or the race.

### Bibliographic References :

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