



Genindexe

La génétique à votre service

Spinal Muscular Atrophy (SMA)

The **Spinal Muscular Atrophy** is an hereditary neuropathy. It is exprimed by the degeneration of the motoneurone which are used for the voluntary actions as fact of walking, of crawling, to maintain the head or to swallow. It is exprimed by the degenerescence of the motoneurons which are used for the voluntary actions as fact of walking, of crawling, to maintain the head or to swallow. The first cases of SMA to Maine Coon were identified in 1997.

The SMA also exists at the man's where it is present under several types according to the age of appearance and the gravity of the symptoms . The SMA of type I appears at the newborn child's who cannot hold the head and quickly die while the SMA of type IV appears to the adulthood and evolves slowly. **The SMA of type III can be diagnosed after 12 months and even until the adolescence : the individual can walk but with frequent falls.** It is the shape of SMA to Maine Coon which is similar the SMA revealing to Maine Coon..

Symptoms

On Maine Coon, first symptoms appears **towards the age of 3 months** : the cat meat some difficulties to **walk and to jump and can present shivers of the posterior members**. To certain kittens, the breath is more difficult and accelerated by moment. The development of the disease varies according to the individual. After a phase of fast evolution (7 in 12 months), the muscular atrophy stabilizes and evolves slowly with sometimes phases of forgiveness in the progress. The ex-members not being got by the muscular atrophy the kitten continues to move by crawling by means of its legs before. Certain cats can live, with a more or less severe handicap, up to the age of 9 years. Various examinations can be realized to confirm a suspicion of SMA: blood examination (increase of the rate of CPK), electromyography, biopsy (atrophy and denervation myofibers). A screening DNA test is now available allowing not only to confirm a SMA diagnosis , but also to identify the carrier cats of the transfer (and to avoid so accidental marriages of carrier cats).

Mode of transmission and DNA test

SMA is a disease which is passed on according to the mode recessive autosomic. That is males as females are concerned autosomic, and that is a kitten has to inherit from the copy (allèle) of the affected gene by each of his two relatives to be affected.

Moreover, this recessive transfer mutation recessive is at complete insertion : A homozygous cat for the affected allele will necessarily develop the disease. GENINDEXE propose you a genetic test after having validated it on Maine Coon race no carrier (homozygote normal), carriers (heterozygotes) and affected (homozygotes muted).

Expression of results and meaning

In genetique the code to design the normal copy(allele) of a gene is « + » and in contrary the code to design the affected copy (allele) of a gene is « - ».

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

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

To avoid the distribution of this pathology to Maine Coon, we recommend to test the cats the pedigree of which arises from lineages known to be got by the SMA as well as the big breeders. 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	+	+/+	+/+	+/+	+/-	+/-	+/-
		-	Carrier	Carrier	Carrier	Affected	Affected	Affected
	Affected	+	+/-	+/-	+/-	Affected	Affected	Affected
		-	Carrier	Carrier	Carrier	Affected	Affected	Affected

