ZARIYÃ SITARÃ DE LA MAGIE DES LIC...

SOMALI

Microchip number: 250269591809760
ID kit: FLKPWFR

Test date: 2025-09-08
French Cat Club

Health conditions known in the breed

Progressive Retinal Atrophy (Discovered in the Abyssinian)	Gene	Risk Variant	Copies	Inheritance	Result
	CEP290	T>G	0	AR	**Clear**

Information about the genetic condition

Progressive Retinal Atrophy (PRA), in the rdAc form, follows the typical pattern where functional loss of rod photoreceptors occurs first, followed by loss of function of cone photoreceptors. Age of onset for this form of PRA is typically late, with the first ophthalmoscopic signs of affected cats seen at one to two years of age. These signs may include a slight grayish discoloration along the central fundus progressing to the entire tapetal fundus, a hyper-reflective tapetum and attenuated blood vessels. The disorder is progressive, causing increasing levels of vision loss and eventual blindness by three to seven years of age. Early indications of visual compromise may include disorientation and lack of awareness of changes to the surroundings, especially in low light conditions. Affected cats may accidentally bump into things and become more vocal.

Breeder recommendation

This disease is autosomal recessive meaning that two copies of the mutation are needed for disease signs to be shown. A carrier cat with one copy of the PRA mutation can be safely bred with a clear cat with no copies of the PRA mutation. About half of the kittens will have one copy (carriers) and half will have no copies of the PRA mutation. Kittens in a litter which is expected to contain carriers should be tested prior to breeding. Carrier to carrier matings are not advised as the resulting litter may contain affected kittens. Please note: It is possible that disease signs similar to the ones caused by the PRA mutation could develop due to a different genetic or clinical cause.

Pyruvate Kinase Deficiency	Gene	Risk Variant	Copies	Inheritance	Result
	PKLR	G>A	0	AR	**Clear**

Information about the genetic condition

Pyruvate Kinase (PK) Deficiency presents as a chronic, intermittent, hemolytic anemia. The disorder has a high variability of age of onset and severity of clinical signs. The age of onset of clinical signs varies from six months to five years of age. Clinical signs of the disorder are highly variable but may include lethargy, weakness, diarrhea, pale mucous membranes, anorexia, poor coat quality, weight loss, icterus (jaundice), splenomegaly, and ascites in severe cases. The severity of clinical signs also varies greatly with some cats maintaining adequate quality of life and others requiring euthanasia. The disorder has been reported in multiple cat breeds.

Breeder recommendation

This disease is autosomal recessive meaning that two copies of the mutation are needed for disease signs to be shown. A carrier cat with one copy of the Pyruvate Kinase Deficiency mutation can be safely bred with a clear cat with no copies of the Pyruvate Kinase Deficiency mutation. About half of the kittens will have one copy (carriers) and half will have no copies of the Pyruvate Kinase Deficiency mutation. Kittens in a litter which is expected to contain carriers should be tested prior to breeding. Carrier to carrier matings are not advised as the resulting litter may contain affected kittens. Please note: It is possible that disease signs similar to the ones caused by the Pyruvate Kinase Deficiency mutation could develop due to a different genetic or clinical cause.

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Inheritance Mode Key

Autosomal Recessive (AR)

The trait is only expressed when both alleles (inherited from mother and father) contain the detrimental mutation.
Regarding to the presence of mutations dogs are classified into three groups:
• Affected (mut/mut)- both alleles carry mutation, disease could be clinically expressed
• Carrier (mut/normal)- one of two alleles carry mutation (heterozygotes), disease is not clinically expressed
• Clear (normal/normal)- mutation is not detected, normal genotype, healthy animal for the trait
Heterozygotes in this case are the carriers of mutation since they do not express the disease (unwanted trait). It is especially important to test such animals for mutations, since mutated alleles are “silently” (without seeing unwanted phenotype) carried through the population.

Autosomal Dominant (AD)

The trait is expressed when one of the alleles (inherited either from mother or father) is damaged (contains detrimental mutation). Only one single mutated allele already could cause the disease. The importance for genetic testing of such animals is primarily in early diagnostics of the disease and identification of animals before they mate because most of diseases with autosomal dominant mode of inheritance have an onset later in animals life.

X-linked Recessive (SR)

The trait is carried on a sex chromosome and that a trait is expressed only when both alleles (inherited from mother and father) are damaged (contain detrimental mutation). Males carry only a single copy of the gene, inherited from mother, since male sex chromosome Y does not contain full DNA sequence as female X chromosome does. Females on the other hand contain two X chromosomes. Heterozygotes in this case are the carriers of mutation since they do not express the disease (unwanted trait). Males carry only one copy of a gene: they could be normal homozygote or affected homozygote.

X-linked Dominant (SD)

The trait is carried on a sex chromosome and the trait is expressed when one of the alleles (inherited from mother or father) is damaged (contains detrimental mutation). Only one single mutated allele already could cause the disease (unwanted trait). Males carry only a single copy of the gene, inherited from mother, since male sex chromosome Y does not contain full DNA sequence as female X chromosome does. Females on the other hand contain two X chromosomes. Homozygotes in this case may be at higher risk or show a more severe form of the disease than heterozygotes. Males carry only one copy of a gene: they could be normal homozygote or affected homozygote.

Mitochondrial (MT)

Rather than genomic DNA, the trait is associated with mitochondrial DNA (mtDNA) of which there are thousands within each cell of the body. For disease (unwanted trait) to occur, a certain ratio of mtDNA, inherited only from mother, must contain the detrimental mutation compared to normal mtDNA.

Modifier (MO)

Genetic modifiers do not cause disease (unwanted trait) on their own. It is only when inherited in combination with specific detrimental mutations, the trait expression can be further influenced by the presence of a genetic modifier–either increasing likelihood of disease or the severity of a disease. It is dependent on the genetic modifier as to if heterozygotes or homozygotes will influence the trait expression.