Progressive Retinal Atrophy (rdAc)

About the disease

A mutation (rdAc) in the *CEP290* gene produces a defective protein that is associated with progressive retinal atrophy (PRA) in Abyssinian and related cats. The disease is characterized by progressive degeneration of the photoreceptors (rods and cones) in the retina. Affected cats typically develop late onset blindness; having normal vision at birth but developing clinical signs at 1 to 2 years of age (detectable by eye examination) and blindness by 3 to 5 years. However, age of disease onset and progression can be variable and some affected cats may not develop clinical signs, and eventually blindness, until later in life (6+ years).

About the test

At the Molecular Diagnostic Unit, we have developed a PCR-based pyrosequencing assay to quickly and accurately identify the rdAc mutation known to cause PRA in Abyssinian and related breeds.

This assay will allow owners and breeders to identify Affected and Carrier cats and will enable them to inform breeding programs to reduce PRA (rdAc) in their kittens.

Interpretation of results

A **Normal** result on the PRA (rdAc) genetic assay means that the cat does not have the rdAc genetic mutation. It is possible that some cats may go on to develop retinal atrophy due to other, as yet unidentified, genetic mutations.

A **Carrier** result on the PRA (rdAc) genetic assay means that the cat carries one copy of the rdAc genetic mutation. This cat is a Carrier of PRA.

An **Affected** result on the PRA (rdAc) genetic assay means that the cat carries two copies of the rdAc genetic mutation and will be affected by PRA.

Each certificate we issue will specify whether the cat is Normal, Carrier or Affected for the PRA (rdAc) mutation.

The gene test will help breeders decide whether or not to use cats for breeding. Generally Affected cats should not be used for breeding because they are certain to pass on the genetic

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mutation. There is a 25% probability of two Carrier cats producing Affected kittens. Breeding Carrier and Normal cats will produce around 50% Normal and 50% Carrier kittens.

This strategy can be used as part of a breeding programme to gradually eliminate the defective gene from the affected populations.

FAQs

What breeds are at risk?

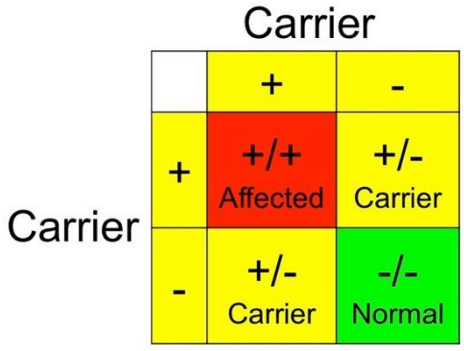
Abyssinian	American Curl	American Wirehair	Balinese
Bengal*	Colourpoint Shorthair	Cornish Rex	Javanese
Ocicat	Oriental Shorthair	Siamese	Singapura
Somali	Tonkinese		

* See below for more details about Bengal PRA

Does PRA affect Bengals?

It is possible that Bengals may have the rdAc mutation since Abyssinians were used in the foundation of the breed, but if present this is likely to be at a low prevalence. Recently a specific PRA mutation (PRA-b) was found in the Bengal breed and this mutation likely accounts for the majority of Bengal PRA cases. Please see the Bengal PRA factsheet for further information.

What are the genetics of breeding?



Autosomal Recessive
PRA is inherited as an autosomal recessive trait, meaning that two copies of the defective gene are required in a cat to produce the disease.

Carrier cats (those with a single copy of the defective gene) are not affected and have normal vision. However, breeding from two Carrier cats has a 25% chance of producing Affected kittens and a 50% chance of producing more Carrier kittens. Therefore, it is NOT recommended to breed two Carrier cats together, since this can produce Affected kittens.

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What do I do with a Carrier?

		Carrier	
		+	-
Normal	-	+/- Carrier	-/- Normal
	-	+/- Carrier	-/- Normal

Breeding is still possible
It is possible to continue to use Carrier cats in breeding programmes to retain important breeding lines and to avoid reducing the size of the gene pool.

As long as Carrier cats are mated to Normal cats no Affected kittens will be produced. This mating is likely to produce kittens that are Carriers, which can be identified by genetic testing and, if necessary, future matings arranged with Normal cats.