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## Progressive Retinal Atrophy in Felines

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**Introduction**

In Abyssinian, Somali and some Ocicat breeds, an inherited late-onset blindness condition has been identified and is characterized by progressive degeneration of the photoreceptors (rods and cones) in the retina. This disease has been designated "rdAc".

Cats affected with this form of blindness have normal vision at birth, with degeneration first detected by electroretinographic (ERG) exam at about seven months of age. Vision loss progresses slowly and is variable, with most cats becoming blind by usually 3-5 years of age. There is no treatment available for the condition. This is an autosomal recessive condition, thus the disease is not associated with gender and two copies of the mutation are required for the cats to lose their vision. Carriers, cats that have one copy of the mutation, are not affected and have normal vision.

A single nucleotide mutation in the gene called *CEP290* produces a defective protein which is associated with this progressive retinal atrophy (PRA) in the cat. In addition to Abyssinian, Somali and Ocicat, a survey of 43 cat breeds showed presence of the *CEP290* mutation in many other breeds including, American Curl, American Wirehair, Bengal, Balinese/Javanese, Colorpoint Shorthair, Cornish Rex, Munchkin, Oriental Shorthair, Peterbald, Siamese, Singapura and Tonkinese. The high frequency of the *CEP290* mutation in Siamese (about 33%) and related breeds (Oriental Shorthair, Balinese/Javanese, Colorpoint Shorthair, Peterbald) poses a significant health risk in the Siamese breed group.

A different form of blindness called "rod cone dysplasia", or "Rdy" has also been identified in Abyssinian and Somali cats. The mutation is a single base pair deletion in a different gene, *CRX*, which also results in a defective protein that is critical for eye development.

Cats carrying one copy of this mutation have retarded development and degeneration of photoreceptor cells, which leads to early-onset blindness by 7 weeks of age. Current information suggests that the "Rdy" mutation is restricted to the Abyssinian and Somali breeds. The "Rdy" mutation is inherited as a dominant trait. Cats that have one or two copies of the mutation will be affected. The Rdy mutation is rare.

To assist owners and breeders in identifying affected and carrier cats, the VGL offers DNA tests for the two mutations known to cause the two different forms of PRA in cats - rdAc (*CEP290*) and Rdy (*CRX*). The tests use DNA collected from buccal swabs. Breeders can use these tests as tools to avoid breeding two carriers of rdAc which would produce 25% affected offspring, or to test for the presence of "Rdy" in the CRX gene of suspected affected cats.

Since both forms of blindness are found in the Abyssinian breed, all associated breeds may have a concern for these diseases. The two mutations (rdAc and Rdy) have been tested in a novel form of blindness in the Bengal cat, and both have been excluded from causing the Bengal cat blindness (LA Lyons, personal communication). Since Bengal cats have had Abyssinian breedings in their ancestry, these genetic tests may be warranted in some Bengal lines.

**Procedure for collecting a feline DNA sample****ORDER TEST | PRICE LIST****Allow 2-6 business days for results.****Results reported as:**

| Test Result | CEP290 PRA-rdAc Status   |
|-------------|--|
| N/N         | Normal, cat does not have rdAc mutation*   |
| N/rdAc      | Carrier, cat has one copy of rdAc mutation. Breedings between carriers will be expected to produce 25% affected kittens. |
| rdAc/rdAc   | Affected   |

**\* This test only detects the mutation in the *CEP290* gene known to cause PRA in Abyssinian, Somali, Ocicat and other breeds mentioned above.**

| Test Result | CRX PRA-Rdy Status   |
|-------------|--|
| N/N         | Normal, cat does not have Rdy mutation**   |
| N/Rdy       | Affected, cat has one copy of the Rdy mutation. This cat will produce affected kittens 50% of the time when bred to a normal cat, or 75% of the time when bred to another cat with one copy of the Rdy mutation. |
| Rdy/Rdy     | Affected, cat will always produce affected kittens.  |

**\*\* This test only detects the mutation in the *CRX* gene known to cause PRA in Abyssinian and Somali breeds.**

References:

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Menotti-Raymond M, David VA, Pflueger S, Roelke ME, Kehler J, O'Brien SJ, Narfström K. Widespread retinal degenerative disease mutation (rdAc) discovered among a large number of popular cat breeds. *Vet J.* 2009 Sep 9. [Epub ahead of print] PubMed PMID: 19747862.

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