

David Miskelly has kindly reviewed a very recent paper on PRA. David is, with his wife Louise a very active breeder of Abyssinians as well as a Vet

Progressive Retinal Atrophy in Abyssinians

Progressive Retinal Atrophy (PRA) or as it is also known Retinal Degeneration was first recognised in the Abyssinian and Somali breed in 1983 in Sweden. Further work showed the condition to be an inherited autosomal recessive condition. The incidence at that time in Scandinavian cats was 45% of cats affected with a similar level of carrier cats, a very high level. Cats affected by the disease are normal as kittens but usually develop a progressive blindness by 3-5 years of age. The defective gene has been identified and there is now a test for the “rdAc” mutation which is located in the CEP290 gene. This is excellent news for breeders as it will allow early identification of affected or carrier animals which can then be removed or used judiciously in breeding programmes.

Recently a further study has been carried out on a larger population of cats in the USA and Sweden. In an attempt to predict the frequency of the gene on a worldwide basis, buccal swabs and blood from Abyssinian and Somali cats in both the United Kingdom and Australia were also included in the study.

The key element of this study was to correlate the results of genetic tests with ophthalmoscopic examinations. A total of 87 cats were included in the study conducted in Sweden and the USA . The results obtained showed excellent concordance between the genetic profile and the retinal changes seen by the veterinary ophthalmologists on examination. Eighty two of the cats followed exactly the disease pattern predicted by their genetic profile. The five cats that did not fully follow the criteria included :

- An 18 month old male aby with 2 copies of the defective gene (affected) that was normal on ophthalmoscopic exam. However the animal unfortunately dropped out of the study and was not re-examined. It was likely that the young age at examination meant subtle changes may not have been visible.
- A 7 year old aby with 2 copies of the mutation (affected) was normal at age 6 but had generalised retinal degeneration at age 7. This was a much later onset than expected.
- 3 other cats which were either genetically normal or carriers developed PRA between the ages of 4 and 7. It is highly likely that these cats were affected by non-hereditary forms of retinal degeneration such as Feline Central Retinal Degeneration (FCRD) or retinal disease caused by toxins or inflammatory processes.

The variability of the onset in disease was also seen in affected cats included in the study with some cats showing retinal changes as young as 4 months and others as late as 3 years old . Interestingly the younger the disease was seen the more rapidly the condition progressed also. This could be caused by other factors such modifying

genes or environmental conditions being involved. It demonstrates that ophthalmoscopic exams alone will not pick up all cases early enough.

Another aspect of the study was that several other breeds were screened in relatively small number, but both an Ocicat (not surprising given the origins of this breed) and an eight year old Siamese with clinical signs of PRA were shown to have the affected gene. Until now this had been thought to be solely a condition of Abyss and Somalis but it may now be the case that other breeds have the condition. The paper is bringing this to the attention of vets at large and possibly further cases in other breeds will be identified.

Of particular interest to breeders in the UK are the results from the 34 cats included in the population study:

Normal	22
Carrier	10
Affected	2

Based on this relatively small numbers the genetic population study has predicted a prevalence of the mutation to be 4.2% in our breed in the UK. This compares with Scandinavia at 3.8% and Australia at 1.1% .

The take home message for UK Abyssinian breeders is that in the 1983 study the level in Scandinavia was 45% and it is now less than 4% so this shows what can be done by being selective when mating our cats and with the ready availability of highly reliable genetic tests even more progress can be made.

With the turmoil caused last year by the media in the pedigree dog world it would be prudent for us to be able to demonstrate that we are being responsible as breeders and using the genetic tests available to further our wonderful breed.

Veterinary Ophthalmology (2009) 12, 5, 285–291

Retinal degeneration in the Abyssinian and Somali cat (rdAc): correlation between genotype and phenotype and rdAc allelefrequency in two continents

Kristina Narfstroöm,* Victor David,† Oswald Jarret,‡ Julia Beatty,§ Vanessa Barrs,§ David Wilkie,¶
Stephen O'Brien† and Marilyn Menotti-Raymond†

*Department of Veterinary Medicine and Surgery, College of Veterinary Medicine, University of Missouri, Columbia, MO, USA; †Laboratory of Genomic Diversity, National Cancer Institute-Frederick, Frederick, MD, USA;

‡Institute of Comparative Medicine, University of Glasgow, Faculty of Veterinary Medicine, Bearsden, Glasgow G61 1QH, Scotland, UK; §Faculty of Veterinary Science, The University of Sydney, NSW 2006, Australia;

¶Department of Veterinary Clinical Sciences, The Ohio State University, Columbus, OH, USA