

Stockholm, Feb. 12, 2020

Hereditary Cataracts in the Russian Blue cat

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Cataracts were diagnosed in the Russian Blue breed of cat approximately 10 years ago in Finland by Dr. Sari Jalomäki. She contacted the undersigned (KN) and a collaborative study was designed. The aim was to elucidate whether the cataract observed was a genetic problem or not, describe the cataract clinically, evaluate the prevalence of cataract specifically in Russian Blue cats and, finally, find the specific gene defect/mutation for the disorder. Our hope was also to be able to provide a molecular genetic test for the defect. Dr. William Murphy, a molecular geneticist from the University of Texas A & M, USA, was engaged for the molecular genetic parts of the study.

Cats were evaluated with ophthalmic examinations mainly at one small animal hospital in Stockholm (Djurakuten) and another in Göteborg (Blå Stjärnans Djursjukhus), Sweden, using indirect ophthalmoscopy and biomicroscopy with slit lamp examinations after dilation of the pupils. Cases with cataracts were photographed and swabs with saliva for DNA-analysis were collected from affected and normal Russian Blue cats.

In total 66 Russian Blue cats were included in the study, whereof 22 were affected with cataracts.

The investigations showed that the disease was clearly hereditary and an autosomal simple recessive mode of inheritance for the disorder was postulated. The cataracts observed were bilateral in most cases and changes were most often found in the area between the posterior part of the nucleus and the anterior part of the posterior lens cortex, thus a very specific localization in affected animals. In some cases the changes were more extended. Minor changes did not cause visual problems, however, more extended opacifications of the lens and complete cataracts caused severe visual impairment or blindness.

Direct genotyping of DNA from affected and control animals as well as SNP-genotyping were performed. Also, molecular analysis was accomplished using genome wide association studies (GWAS). These investigations revealed some strong candidate genes that could be causative for the disorder. In order to proceed with expression studies Dr. Murphy proposed a qualitative study of lens tissue from affected and normal cats. This is the stage of the studies that we are in at the present time.

Further details in regards to ongoing studies

Clinical eye examinations

Clinical examination of the eye and its adnexa (surrounding structures) using specific instrumentation, such as an indirect ophthalmoscopy and slit lamp biomicroscopy after

dilation of the pupils, is of utmost importance in order to diagnose various defects and diseases of the eye. Thereby "new" diseases may be found, for example such as those that have not previously been described in the literature, and the prevalence of specific diseases of the eye in a population can be calculated. Further, affected animals can thereby be found and removed from the breeding stock in order to reduce the number of affected animals in the population.

It is recommended that eye examinations are performed regularly in cats used for breeding. The eye certificate with results from the examination should not be older than one year in regards to these cats. For non-breeding animals clinical eye examinations are recommended to be performed at least 3 times during the life-time of the animal, e.g. at approximately age 1, 3 and 7 years. Kittens can be examined for the first time as early as at age 8-16 weeks.

Lens tissue

Lens tissue is needed for further molecular genetic research from affected and non-affected Russian Blue cats. The tissue should be collected immediately after death and immersed into vials (with RNA-later). The vials are then sent by the investigator in Sweden to Dr. William Murphy, USA, for further studies.

Please note that lens tissue from the eye can be obtained from cats that are euthanized due to unrelated causes, such as renal or heart problems or just old age, of course with the owner's consent. Cat owners should contact Dr. Kristina Narfström's "eye nurses" at Djurakuten in Stockholm, phone: 08-301900 or Dr. Karin Nygren at Blå Stjärnans Djursjukhus in Göteborg, phone: 031-653500 in relation to these issues.

Blood samples

-In the future blood samples may be collected. At the present time they are not needed for the ongoing studies. Owners and breeders of Russian Blue cats will be notified in the future should such a need occur.

Literature

-Karin Nygren, Sari Jalomäki, Lena Karlstam and Kristina Narfström. Hereditary cataracts in Russian Blue cats. Journal of Feline Medicine and Surgery, 2018, Vol 20(12), 1105-1109.

-Website: www.ecvo.org/ Hereditary eye diseases/Manual/Breeds/Cat/Russian Blue

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See sections 5,7,8,9,10 and 10.1