Hereditary Retinal Degeneration in a Siamese Cat

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Abstract: A 5-year-old Siamese cat was presented at the Veterinary Medical Teaching Hospital in Seoul National University with visual impairment. The cat was blind and its pupillary light reflexes were sluggish OU. While there were no remarkable findings in the anterior segment, generalized tapetal hyperreflectivity and retinal vascular attenuation were observed in the posterior segment of both eyes. Any historical significance and clinical symptoms suggestive of infection or inflammation were absent. All of the above findings strongly indicated a diagnosis of hereditary retinal degeneration in the cat. This clinical case report demonstrates the possibility of the presence of feline hereditary retinal degeneration in Korea. Awareness of the condition should be raised to prevent the spread of hereditary retinopathy in the Korean cat population.

Key words: blindness, cat, hereditary, retinal degeneration, Siamese.

Introduction

Most unsighted cats have a similar fundic appearance of generalized retinal atrophy (3), because clinical detection of the disease tends to take place later in cats than in dogs, which makes it more difficult to determine the specific etiology (3). Clinicians usually attribute feline bilateral retinal degeneration to one of following etiologies: nutrition, drugs, or hereditary retinopathy.

Nutritional retinopathy caused by taurine deficiency has become rare due to increased awareness of the necessity for cat-specific food (4). An evaluation of cardiac function may also aid in the diagnosis of taurine deficiency (4). In addition, careful history taking and a meticulous review of medical records can establish the possibility of drug-induced retinal degeneration. Although genetic studies are needed to confirm its hereditary basis, hereditary retinopathy can be diagnosed clinically through the exclusion of other etiologies.

Case

A five-year-old spayed female Siamese cat was presented at the Veterinary Medical Teaching Hospital in Seoul National University with vision loss. The owner had noticed her visual impairment 6 months prior to the visit. There was no history of drug administration or anesthetic/surgical events, and the cat was fed only commercially available cat food. A complete physical examination was unremarkable. But the cat was blind OU, and incomplete and sluggish pupillary light reflexes was observed OU (Fig 1). Tear production (Schirmer tear test®; Intervet, Summit, NJ, USA) measured 22 mm/min OD and 19 mm/min OS, and the intraocular pressures (TonoVet® tonometer; Icare, Finland) measured 19 mmHg OD and 22 mmHg OS. A slit lamp biomicroscopic examination (SL-D7, Topcon, Japan) showed no remarkable findings in the anterior segment. Indirect ophthalmoscopy was performed to evaluate the posterior segment. Generalized hyperreflectivity of the tapetal fundus was observed OU and the retinal vasculature was severely attenuated OU. Funduscopic changes were documented using a digital fundus camera (Fig 2).

The lack of historical significance and clinical symptoms suggestive of infection or inflammation, in combination with the progressive nature of bilateral vision loss, most likely indicates a case of hereditary retinal degeneration. Additional examinations were refused by the client when she was informed that there is no specific treatment for retinal diseases.

Discussion

Feline retinal degeneration has been reported less frequently than canine cases, and the approach used by clinicians for differential diagnosis should differ to the canine diagnostic approach.

First of all, nutritional retinopathy caused by taurine deficiency must be ruled out before hereditary retinopathy is diagnosed. Recently, the incidence of nutritional retinopathy has become rare due to an increased awareness of the necessity for cat-specific food. However, cats living with dogs often eat dog food without their owners’ knowledge. Additionally, even though they have been fed cat-specific food, taurine deficiency may develop if they have inherent amino acid absorption failure. In these cases, it is helpful to measure plasma taurine concentrations in order to confirm taurine deficiency (4). Generally, however, taurine-deficient retinopathy shows a characteristic oval hyperreflective lesion along the area centralis and is also accompanied by cardiomyopa-
thy, which enables clinicians to differentiate it through careful ophthalmic examination and cardiac function evaluation (4,7,10).

Secondly, enrofloxacin associated retinal toxicity is one of the possible etiologies of feline diffuse retinal degeneration. One of the characteristics of the clinical course of this disease is the rapid progression of retinal degeneration, resulting in acute blindness (4,7,10). Research has proven that rod degeneration precedes cone degeneration. Risk factor analysis has shown that old age and hepatic or renal dysfunction could reduce drug clearance, thereby increasing the risk of fluoroquinolone retinal toxicity (4).

Thus, hereditary retinopathy can be considered when clinicians have ruled out nutritional, toxic, and inflammatory/infectious causes. Breeds identified to have a hereditary basis include the Abyssinian cat and the Somali cat (5). Of those breeds, many genetic and phenotypic studies have long been performed on the Abyssinian cat (7,10).

Two types of hereditary retinal dystrophy have been identified in Abyssinian cats. Early-onset retinal dystrophy, also known as cone-rod dystrophy (CoRD), is a primary photoreceptor disorder with an autosomal dominant mode of inheritance. Affected kittens show sluggish pupillary light reflexes and slightly dilated pupils by 4 to 5 weeks of age, leading to complete vision loss within the first 4 months of life, which is in contrast to this case. Unlike CoRD, retinal degeneration in Abyssinian cats (rdAc) is considered to be a late-onset disease caused by autosomal recessive inheritance of the mutant rdAc allele. Studies show that rod degeneration precedes cone degeneration ultrastructurally by 4 to 5 months of age. Clinically, however, the fundic appearance of homozygous (affected) cats is normal until 1.5 to 2 years of age. The end stage of blindness is usually reached by 3 to 7 years of age, implying that there is a great degree of phenotypic variation in rdAc disease under the influence of the uniform genetic defect (5,6).

Although most molecular genetic studies have focused on Abyssinian cats, there are growing concerns about the presence of hereditary retinopathy in Siamese cats (1,2). Breed predilection for the Siamese cat was observed in a retrospective study reported in 1999 (3). The mean age ± SD of the Siamese cats in this study was 12 ± 2.8 years, which is much greater than the age of the patient described in this case. Luckily, the owner in this case detected her cat’s visual impairment earlier than most because she used to entertain her cat with a laser pointer and realized that her cat had become reluctant to look up at the moving laser beam.

Typically, cats’ exceptional ability to adapt to progressive blindness makes their owners believe that they are visually normal until their retina becomes completely atrophied (3). In the initial stages of progressive retinal atrophy in dogs, nyctalopia is commonly observed early by their owners (10). This early clinical symptom, however, tends not to be noticed by cat owners, making a timely diagnosis difficult in feline patients. Although the most reliable method for achieving an early diagnosis of rdAc is to perform full-field flash elec-

Fig 1. Weak pupillary light reflexes were found in OU. A. The right eye. B. The left eye.

Fig 2. Funduscopic photographs showed marked retinal atrophy, generalized tapetal hyperreflectivity and severe retinal vascular attenuation OU. A. The right eye. B. The left eye.
troretinograms (ERGs) at approximately 8 months of age (5), most cats diagnosed late in the clinical course with complete retinal atrophy are not suitable candidates for the evaluation of retinal function, making ERGs useless in general practice.

The prevalence of affected (homozygous) and carrier (heterozygous) animals in the Abyssinian cat population was found to be 45% and 44%, respectively (5). This unusually high incidence of rdAc in Abyssinian cats is the result of frequent close inbreeding among purebred Abyssinian cats. Recent reports have demonstrated that there is potential for the rdAc mutation to be present in breeds other than Abyssinian and Somali cats (5). One representative breed is the Siamese cat, where rdAc allele frequency is approaching 34% (6,9). The rdAc mutant allele has been known to be transmitted to other popular cat breeds through crossbreeding and the use of popular sires. The fact that rdAc is highly suspected in this Siamese case suggests that such genetic input may have already occurred in the Korean Siamese population. Therefore, extensive genetic and phenotypic studies need to be performed in Korea, and in order to prevent the spread of hereditary retinal disease in Korea, every cat breed should be tested whether or not it carries the gene mutation.

Conclusion

This clinical case report demonstrates the possibility of the presence of hereditary retinal degeneration in the Korean Siamese population. To address this issue, cat breeders and veterinary ophthalmology communities in South Korea need to be alert to the potential of hereditary retinopathy.

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References