

Cats' eye diseases genetically linked to diseases in humans

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Dr. Narfstrom examines cat with eye disease. Credit: Photo courtesy of Dr. Kristina Narfstrom

About one in 3,500 people are affected with retinitis pigmentosa (RP), a disease of the retina's visual cells that eventually leads to blindness. Now, a University of Missouri researcher has identified a genetic link between cats and humans for two different forms of RP. This discovery will help scientists develop gene-based therapies that will benefit both cats and humans.

"The same genetic mutations that cause retinal blindness in humans also cause retinal blindness in cats," said Kristina Narfstrom, the Ruth M. Kraeuchi-Missouri Professor in Veterinary Ophthalmology in the MU

College of Veterinary Medicine. "Now, cats with these mutations can be used as important animal models to evaluate the efficiency of gene therapy. In addition, the eye is an ideal organ to use as we examine the potential of gene replacement intervention because it offers an accessible and confined environment."

Researchers examined the genetic mutations in two groups of cats; one with a congenital form of RP and another with a late-onset form and were able to identify the genes responsible for both forms of the disease in cats. In the study, researchers found that cats with the late-onset form of the disease have a mutation in the CEP290 gene, which is the same mutation found in humans with Joubert syndrome and Leber's congenital amaurosis. In both of these diseases, the genetic mutations result in changes in the function and structure of the photoreceptors. A photoreceptor is a nerve cell found in the eye's retina that is capable of phototransduction, or the process by which light is converted into electrical signals. The changes in the photoreceptors result in cell death, which lead to blindness.

"Cats are excellent models because they have relatively large eyes that are comparable to those of human babies. The retinal changes that occur and the progression to blindness in cats is similar to what happens in the human disease," Narfstrom said. "As a surgeon, I can use the same treatment methods and tools in cats that they use in humans."

Human autosomal recessive RP is among the most common cause of retinal degeneration and blindness, with no therapeutic intervention available. Initially it leads to night blindness, then loss of peripheral vision and, with progression, there is also a loss of central vision.

Like humans, Abyssinian cats with the CEP290 mutation have normal vision at birth but develop early changes in the interior of their eyes by the time they are approximately 2 years old. The cats with the congenital

form of the disease are blind from birth with severe changes in the interior of their eyes after only a couple of months.

More information: In May, Narfstrom will present her latest findings during the Association for Research in Vision and Ophthalmology 2009 Annual Meeting in Fort Lauderdale, Fla. An earlier study, "Mutation in CEP290 Discovered for Cat Model of Human Retinal Degeneration," was published in the Journal of Heredity.

Source: University of Missouri-Columbia

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