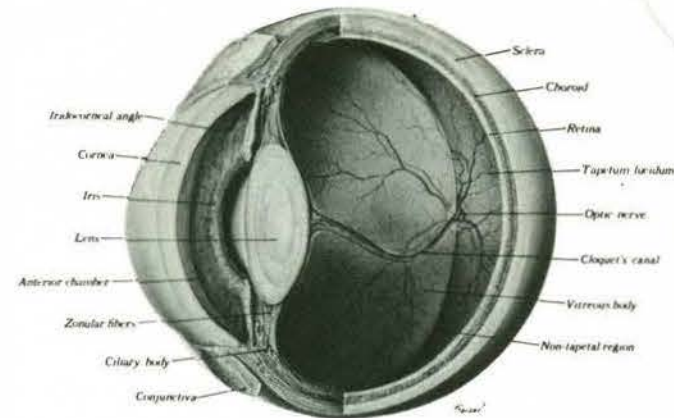




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A Clinic Devoted to Inherited Eye Diseases

A CLINIC DEVOTED TO INHERITED EYE DISEASES



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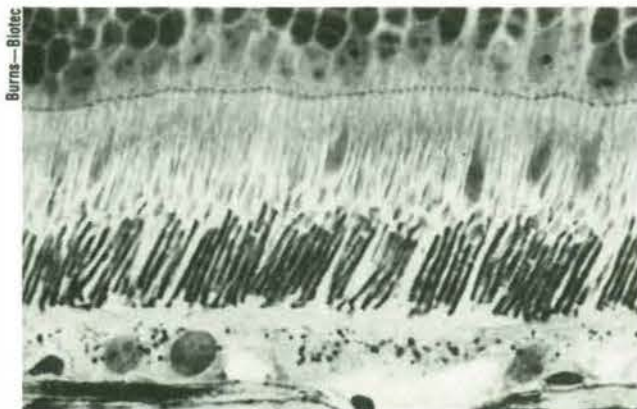
A UNIT WHICH WILL PROVIDE CLINICAL EXAMINATIONS AND GENETIC COUNSELING

Inherited eye diseases occur in animals. They frequently are disabling and present an expensive and frustrating problem to the owner or breeder. To study such disorders in great depth, the School of Veterinary Medicine, University of Pennsylvania, established a new service for the diagnosis and prevention of inherited eye diseases (Inherited Eye Disease Studies Unit). The clinic, headed by Dr. Gustavo D. Aguirre, associate professor of ophthalmology, is part of the Section of Medical Genetics at the School. "It is a unique clinic," explained Dr. Aguirre. "It is totally devoted to inherited eye diseases in cats, dogs, horses, and other species."

The Inherited Eye Disease Studies Unit will provide clinical examination and genetic counseling as well as continue to engage in extensive research. This work will be done in collaboration with members of the Section of Medical Genetics, and with Drs. Gregory Acland and L. Stramm, both of the Scheie Eye Institute, University of Pennsylvania. "We draw upon the many resources here at the School," Dr. Aguirre said. "We can provide various levels of examination, ranging from a routine ophthalmological exam to an elec-

troretinogram. We can do pedigree analysis, laboratory studies for detailed examinations of the cells of the eye, as well as tissue culture studies and electron microscopy."

Within this new service Dr. Aguirre plans to study the course of the many different inherited eye diseases and their mode of inheritance. This information may help in the development of early detection methods. It can also be used to devise a selective breeding program to eliminate or reduce the incidence of inherited eye diseases.



Section of retina showing major regions that can be affected by inherited diseases

Now inherited canine eye diseases are frequently diagnosed at screening clinics organized by dog clubs. Here breeders and owners bring puppies and adults to be examined by veterinary ophthalmologists. Such clinics have been very important in eliminating affected animals from breeding programs in cases where the disease is apparent in young dogs. But inherited eye diseases are complex and may require more specialized equipment for early identification of affected animals. They vary in ophthalmological manifestation from breed to breed. It is possible to detect retinal disorders by means of an ophthalmological exam in Irish setters, and collies by the age of six months. In Norwegian elkhounds such an exam does not reveal the disease until the dog is one year of age or older. In poodles no signs are evident until the animals are between three to five years old. This late detection poses a serious problem. Animals are reared at great expense, they become family companions, show careers may be started, and some dogs may have been incorporated into a breeding program.

"Many eye diseases are inherited recessively," Dr. Aguirre said. "Some animals are carriers and when two such dogs are mated, some of the offspring will show the disease, some will be carriers, and some will be genetically normal. Inherited eye diseases can affect any part of the eye. They may occur in the retina, the cornea, the optic nerve, or other parts of the organ."

Much research has been done on diseases affecting the retina. Commonly these are referred to as progressive retinal atrophy (PRA). The term encompasses a large number of outer retinal degenerations affecting a wide variety of purebred and mixed breed dogs. According to Dr. Aguirre, the name defines the end-stage retinal disease, when the ophthalmologic abnormalities can be seen. The inheritance of PRA has been studied in Irish setters, Norwegian elkhounds, miniature and toy poodles, collies, and cocker spaniels, and in each breed was found to be inherited as simple recessive. Some years ago, Dr. Aguirre and his associates here at the School perfected a test to detect the disease at an early age by means of electroretinography (ERG).

Here the electric impulses emanating from the rods and cones of the eye are measured. Rods and cones are photoreceptive cells in the retina and are essential for vision. In dogs with PRA these cells develop abnormally and then degenerate. Researchers found that electric impulses in those dogs which will later develop the disease are different from impulses in normal dogs. This difference can be detected in very young animals, long before any ophthalmological signs appear. ERG diagnosis for PRA can be made by six weeks of age for Irish setters, Norwegian elkhounds, collies, and miniature schnauzers. In the poodle and the American and English cocker spaniel, the disease can be detected at a later age, closer to nine or ten months. "The ERG helps us identify the afflicted animals early," Dr. Aguirre said. "Then they will not be incorporated into breeding programs. The test also helps to find animals which can be used for test breeding to identify other carriers." Currently Dr. Aguirre and Dr. Gregory Acland, assistant professor in the Department of Ophthalmology, School of Medicine, are working on developing a procedure by which late onset PRA in poodles and other breeds can be identified at an age earlier than ten months. Electrophysiology is Dr. Acland's special interest and he has designed the computer program at the School used to evaluate and interpret the ERG results.

Studies by Drs. Aguirre and Acland have found that PRA is not one disease but a different one in each breed. Its course, during the maturation of the eye, is distinct for each breed studied. When a dog is born, its eyes are not fully developed. The retina is immature at birth and it develops during the first six weeks of life. By age six weeks it is similar in function and structure to that of an adult. The ERG can detect abnormalities in the rod and cone structure while the eye is maturing and this helps to identify dogs with the disease. It was found that in afflicted Irish setters the rods and cones do not develop normally; it was also found that affected animals had a metabolic abnormality within the rods which resulted in the death of the visual cells. In the miniature poodle rods and cones appear to develop normally though their response to light stimuli appears to be at lower levels. It is thought that PRA in other breeds also represents distinct diseases.

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The study of PRA has implications not only for dogs but also for humans. It appears to be similar to retinitis pigmentosa, an eye disease affecting people. Here too, the disease varies and appears to be a number of diseases rather than a single one. Dr. Aguirre and his colleagues work closely with the Scheie Eye Institute, a part of the Ophthalmology Department at the Medical School where he is on the staff and has a laboratory.

Other inherited or potentially inherited eye diseases in dogs affect the eyelids (entropion, ectropion, distichiasis), cornea (dystrophy, endothelial decompensation), iris (persistent pupillary membrane), lens (cataract), and vitreous

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(persistent tunica vacuolosa). Even glaucoma, a disease of defective intraocular pressure regulation, can be inherited. Not every dog breed is affected with these many different types of eye diseases. Instead, certain diseases specifically affect some of the breeds and are known as breed related ocular diseases.

Cats, too, have inherited eye diseases, though it appears to be not as common as in dogs. Retinal degeneration is a potential concern to Abyssinian breeders since the disease has been shown

to affect a large number of the breeding stock in some European countries. On the other hand, central retinal degeneration (FCRD) which is found in many cat breeds, has been shown by Dr. Aguirre and associates not to be inherited. The disease is the result of a deficiency of a specific nutrient (taurine) that must be present in cat foods.

Inherited eye diseases are a complex problem. The new clinic will be of great help to veterinarians, breeders, and owners. It can provide researchers with many more cases from which the different conditions can be studied on all levels, ranging from ophthalmologic examinations to the study of tissues, the study of electric impulses, the study of cell types and cell culture, in the hope of finding a way to identify carriers of the diseases, and as a means to identify afflicted animals early in life.

The clinic will see patients on Tuesdays; appointments can be made by calling (215) 898-4680.

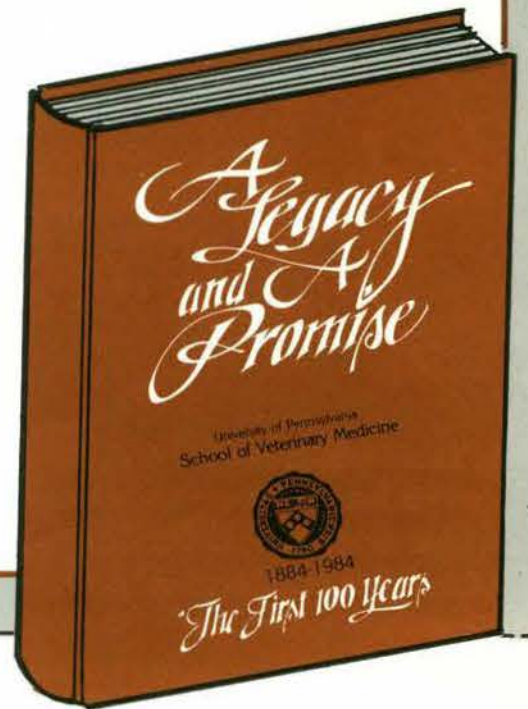
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