Photoreceptive Abiotrophy of the Retina in the Elkhound

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Idiopathic degeneration of the retinal photoreceptive layer, here termed photoreceptive abiotrophy of the retina, is that hereditary abnormality which in human beings is familiarly known as retinitis pigmentosa. In mice and rats, the animal species in which it has been most thoroughly documented, the disease begins within the first few weeks of life and progresses to blindness within a month or two. In dogs, where it is usually known as progressive retinal atrophy, it has been reported to produce blindness at a few weeks of age in some breeds and at 1-2 years in other breeds. It has occurred in Gordon setters, Irish setters, Labrador retrievers, and possibly in poodles. In man retinitis pigmentosa has a variable onset and course but usually begins during adolescence with night blindness and leads to blindness in early adult life.

Photoreceptive abiotrophy is transmitted as a recessive mendelian trait, not necessarily linked with other hereditary abnormalities, and produces clinical symptoms and pathologic changes most like those of severe vitamin A deficiency. Yet there is no substantial evidence of vitamin deficiency in man or animals with this disease. It has been suggested, therefore, that the pathogenesis resides in the peripheral utilization of vitamin A in the visual cycle. To explore such a possibility it is necessary to study the condition in a species with sufficiently large eyes for appropriate clinical, pathological, and biochemical investigation. The chance occurrence of photoreceptive abiotrophy in elkhounds gave us an opportunity to explore this possibility and the present paper is a report of our initial observations on the clinical and pathological characteristics of the disease.
The chief clinical signs in dogs have been gradual blindness and extinction of the electroretinogram\textsuperscript{14} (ERG), while the chief pathologic change has been degeneration of the outer retinal layer. Electron microscopy of retinas with this condition has not been previously reported for dogs but has been reported for rats\textsuperscript{5} and for human beings\textsuperscript{15}.

The family of dogs which constitute the substance of this report (Fig. 1) were elkhounds in which the propositus (A1) was one of two affected female siblings (out of a litter of six) which became blind at about one year of age. One of these two was disposed of without clinical or pathologic investigation but a description of the other constitutes a part of the present paper. Blindness is said to have occurred in previous generations and in collateral lines but the details could not be further documented.

The surviving blind female (A1) was bred to one of her normally sighted male sons. This resulted in a litter of four in which two males

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\includegraphics[width=\textwidth]{figure1}
\caption{Chart indicating genetic relationship of affected dogs and (in parentheses) the times at which they were sacrificed for histologic study.}
\end{figure}

\textit{Abb. 1.} Die Tabelle zeigt die genetische Verwandtschaft der erkrankten Hunde sowie die Zeiten, zu denen sie für die histologische Untersuchung getötet wurden.
(A2a and A2b) developed the abiotrophy while one male and one female did not. Attempts to inbreed further by mating one of the affected males with his affected mother so as to produce a pure strain of dogs with the abiotrophy have so far been successful in producing only two pregnancies with two puppies each time but all these puppies died within a few days after birth. We have, however, obtained a family of elkhounds related to the propositus in which four of the puppies were affected. This report is therefore based on the clinical examination of eight affected dogs and on the pathologic examination of five.

Case Reports

Case A1. When first examined this female dog was 2 years old and had been blind for at least 6 months. The owner had observed...
Fig. 3. Fundus photograph of dog A1 showing clumping of pigment in non-tapetal area. (This clumping is not seen in the normal dog nor in the affected dog during the first year of the disease).


an abnormally gray reflex from the eyes but it was not apparent whether this was attributable to a primary color change in the eyes or to the fact that the pupils were dilated.

On examination the dog was found to be completely blind with dilated pupils that constricted slightly when bright light was shone on the temporal side of the retinas. The lenses were clear except for a few peripheral opacities at the equator. The vitreous of both eyes showed the white, reflectile particles known as asteroid hyalitis. The fundi showed normally protuberant discs but extreme narrowing of the arteries and veins (Fig. 2). Toward the periphery and away from the tapeta the arteries were converted into thin white lines. The tapeta had approximately normal appearances but the edges may have been unusually fragmented. The pigment in the periphery of the
fundi also appeared abnormally clumped and mottled (Fig. 3), in comparison with normal elkhound fundi, but there was none of the bone-corpuscle pigmentation nor invasion of the retina such as we see in human retinitis pigmentosa.

By retinoscopy the eyes were found to be no more than one diopter myopic. Electrorretinography gave no response whereas control observations on normal dogs gave good responses.

No change in appearance of the fundi occurred over an observation period of two years. The animal was disposed of at the age of four years for histologic, histochemical, and electron microscopic study. Portions of the retinas (and other parts of the eyes) were fixed in formalin or alcohol for paraffin embedding while other portions were fixed with osmium for epoxy embedding and still other portions were used fresh for study of dehydrogenases and glycogen synthesis according to methods we have previously used16–18.

Sections stained with hematoxylin and eosin (Fig. 4) or PAS-hematoxylin showed complete loss of the rods and cones. The retina was best preserved adjacent to the nerve head but even here the photoreceptors and outer nuclear layer were completely absent and the ganglion cells were reduced in number (Fig. 5). Away from the disc the retina showed extensive gliosis with distortion, rarefaction, and loss of identity of the bipolar and ganglion cell layers although individual ganglion cells were recognizable even in the most gliotic areas (Fig. 6). In places, the entire retina was thinned to a tenuous glial strand or to actual hole formation. These holes were often bridged by an accessory strand of pigment epithelial cells or, over the tapetum, by a lamina or two of non-pigmented epithelial cells (Fig. 8). PAS stains of these bridging cells showed them to be abnormally filled with glycogen granules. Toward the periphery of the retina pigment epithelial cells had migrated into the retina forming islands of single cells or clusters and sometimes free extracellular pigment (Fig. 7). Similarly non-pigmented cells had migrated into the retina overlying the tapetum and could be differentiated from glia by the sheet-like arrangement.

Along with its migration into the retina and apparent focal proliferation the pigment epithelium had disappeared from much of its normal position behind the retina or had been so severely attenuated in places that it was no longer recognizable. The choriocapillaris was also unrecognizable except in the region about the disc. The optic nerve appeared normal and the tapetum was normal.
Fig. 4. Posterior portion of eye through optic foramen showing intact but thinned retina; dog A1. The outer retinal layers are missing. H. & E.

Fig. 5. Area adjacent to nerve head of same showing intact nerve fiber layer, preservation of some ganglion cells, an intact but distorted bipolar layer, and an absent outer nuclear and photoreceptive layer. H. & E.
Flat mounts of the retinal vessels, prepared by trypsin digestion, showed severe acellularity and occlusion of all the smaller vessels with extensive pigment ensheathing of the veins (Fig. 9).

Lactic acid diaphorase activity was high in the gliotic tissue and showed the large clumps of nitro blue tetrazolium such as we have associated with non-mitochondrial enzyme localization. Succinic diaphorase activity was also high but showed the granular type of activity such as we have associated with mitochondrial activity. Thus there appeared in these retinas no dearth of either mitochondrial or non-mitochondrial activity—in fact, these appeared greater than in normal retinas.

On the other hand, glycogen was abnormally scanty, as determined by the periodic acid-Schiff staining of alcohol-fixed tissue, and the retina was lacking in its capacity to synthesize glycogen when incubated in media containing glucose or uridine diphosphoglucose (UDPG).

Electron microscopy has its usual limitations in that only a portion of the retina and pigment epithelium could be examined. The areas selected were from the posterior portions of the globe but not in the region of the tapetum. Sections were stained with uranyl acetate and lead. Those from a normal retina are shown in Figures 13 and 14. The pigment epithelium appeared normal except that it contained more glycogen particles than normal and its villi lacked the orientation usually provided by the photoreceptors (Fig. 19). Bruch's membrane was abnormally thick but less dense than usual. The choriocapillaris was absent. The retina was very much thinned and showed complete loss of photoreceptors with no vestige of rod or cone segments (Fig. 20). The remainder of the retina consisted chiefly of glial cells with occasional desmosomes and in the outer portion of the retina a stratum of terminal bars that were interpreted as remains of the outer limiting membrane.

Cases A2a and A2b. These were the two male offspring (of Case A1) which ultimately developed the abiotrophy. One was sacri-
Fig. 6. Peripheral portion of retina overlying edge of the tapetum; dog A1. Noteworthy are the ganglion cells, the distorted and rarified bipolar layer, and the total absence of photoreceptive layer. The choriocapillaris and pigment epithelial layers are not recognizable. H. & E.
Fig. 8. “Hole” in retina bridged by a double layer of epithelium; dog A1. The granular material giving rise to the dark stain in the epithelium is glycogen. PAS-hematoxylin.


Fig. 7. Peripheral area showing severely gliotic retina invaded by pigment cell clusters and free pigment; dog A1. The underlying pigment epithelium is intact. H. & E.


ficed at age 15 months and the other at 32 months. One male and one female sibling did not develop the disease and will not be described further although they were also examined periodically and served as controls for the affected dogs.

At three months and six months of age the visual function of all four dogs in the sibship appeared equal and normal and the vessels, tapeta, and retinal periphery were normal. The pupils showed full responsiveness to photic stimulation. At eight months of age, however, the retinal arteries of the two affected puppies were pathologically narrow and the owner of the dogs observed an abnormally gray reflex from the fundus (as compared to that from the normal siblings) although this was not apparent to the examiner. The pupils at this stage were reactive to light. The tapeta had an equivocally yellow and washed-out appearance in comparison with the normal. Electroretinography showed diminished or absent responses whereas the two controls showed good responses to flashes of light.

Periodic observations up to 15 months of age showed progressive narrowing of the retinal vessels but the vitreous, tapeta, retinal periphery, and optic nerves remained normal. At this stage one of the affected dogs (A2a) was disposed of and the eyes studied histologically in the same manner as described in the foregoing case.

Case A2a. Cross sections of these eyes showed complete loss of all rods and cones but the outer nuclear layer was still partially intact about the disc (Fig. 10). Elsewhere it was also spottily present but

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**Fig. 9.** Flat mount of retinal vessels showing extensive acellularity of the small vessels and adherent pigment clumps; dog A1. PAS-hematoxylin.

**Fig. 10.** Dog A2a (15 months). Cross section of retina overlying the tapetum in the vicinity of the nerve head. The ganglion cell layer and bipolar layer are intact but the outer nuclear layer is greatly rarified and the photoreceptors are completely absent. The underlying pigment epithelium is normal. H. & E.

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**Abb. 9.** Retina, flach präpariert, die starke Zellverminderung der kleinen Gefäße und anhaftende Pigmentklumpen aufweist; Hund A1. PAS-Hämatoxylin.

greatly distorted by gliosis and clefts. The bipolar cell layer was also thinned or absent in places but ganglion cells were widely identified. The irregular loss of these nuclear layers resulted in great variations in the thickness of the whole retina. The pigment epithelium and the non-pigmented epithelium over the tapetum were present and appeared normal but the choriocapillaris was not recognized. The epithelial cells had not migrated into the retina. The optic nerves were normal.

The retinal vessels visualized as flat mounts showed moderate capillary acellularity, but much less than in the previous case, and unaccompanied by pigment ensheathing (Fig. 11).

Some glycogen was present (identified by periodic acid-Schiff-amylase) in the inner retinal layers but less than in the normal eyes and much less than would be expected in view of the extensive damage to the outer layers. Nor were we able to demonstrate glycogen synthesis by means which we had previously employed for the retina and which were successful in showing glycogen synthesis in the normal control.

Case A2b. The second of the affected dogs (A2b) in this litter was sacrificed at 32 months of age, that is 17 months after the disposal of its affected sibling. At this time the dog was completely blind and the ERG was completely extinguished. The retinal arteries were severely narrow and the periphery showed a splotchy pigment but the nerve head showed its normal protuberance. The retinas and other ocular structures were prepared in a manner similar to those of the previous cases.

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Fig. 11. Flat mount of retinal vessels showing only moderate acellularity of the capillaries and no adherent pigment; dog A2a. PAS-hematoxylin.

Fig. 12. Dog A2b (32 months). Cross section of peripheral retina showing a thinned and severely gliotic retina but partial preservation of the ganglion cells and bipolar cells. The rods and cones have disappeared completely but the pigment epithelium is intact. H. & E.

Abb. 11. Retina, flach präpariert, die nur geringe Azzellularität der Kapillaren und kein anliegendes Pigment aufweist (Hund A2a, PAS-Hämatoxylin).


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Cross sections of the retinas showed complete loss of the rods and cones from the entire retina and loss of the external nuclear layer from the entire retina except in the region about the disc where it was partially preserved. The bipolar and ganglion cell layers were intact and normal about the disc but became progressively distorted by gliosis away from the disc (Fig. 12). Except in the immediate peripapillary area the retina was considerably thinner than normal and in addition there were foci in which the retina was reduced to a tenuous glial strand or to actual hole formation. At these latter sites pigment epithelial cells had migrated into the retina and sometimes formed bridges connecting two edges of the retina. Similarly non-pigmented epithelial cells sometimes formed bridges or clusters of glycogen rich cells within the retina overlying the tapetum.

The pigment epithelium was everywhere intact and the retina was nowhere fused with the choroid. The choriocapillaris was present. Flat mounts of the retinal vessels showed severe acellularity of all but the larger vessels and extensive ensheathing of the vessel walls by pigment.

Electron microscopy showed many of the same changes as noted in the first case. No trace of the photoreceptors was present and the retina was reduced to a thin membrane consisting chiefly of glial cells. Although these glial cells appeared to be arranged in a haphazard manner terminal bars were identified forming a lamina that corresponded to the outer limiting membrane (Fig. 18). The ultrastructure of the pigment epithelium was normal although the villi, lacking the usual arrangement which they have with the photoreceptors, were much folded on themselves. Bruch’s membrane and the choriocapillaris were normal.

While the foregoing studies were taking place and our interest in the disease became known, we were shown four night blind Elkhounds from a litter of nine distantly related to the foregoing family. They were six months old; one was a female and three were males. Night blindness had been observed for a few weeks. Clinical examination revealed suggestive narrowing of the retinal vessels but no other ophthalmoscopic abnormality. Electroretinography could not be done at that time for technical reasons.

Case B1a. One of the night blind dogs was sacrificed and the tissue prepared in accordance with the preceding methods. Light microscopy revealed no abnormality. Specifically the retinas including
Fig. 13. Normal elkhound retina. Outer segments of photoreceptors with their characteristically laminated plates. C, outer segments of cones. The other laminated structures are outer segments of rods.

Electron micrographs showing successive stages in the development of retinal abiotrophy in the elkhound. The marker (—) in all pictures indicates one micron.

Fig. 14. Normal elkhound retina. Inner segments of photoreceptors packed with mitochondria. C, cones, R, rods.

Fig. 15. Abiotrophy in dog 6 months old (B1a). The outer segments of the photoreceptors show fragmentation and beginning disorientation of their laminae. The pigment epithelium and epithelial processes are normal. Os, outer segments; PE, pigment epithelium.

Abb. 15. Abiotrophie bei einem 6 Monate alten Hund (B1a). Die Aussensegmente der Lichtrezeptoren weisen Fragmentation und beginnende Desorientierung ihrer Plättchen auf. Das Pigmentepithel und die epithelialen Anteile sind normal; Os, Aussensegmente. PE, Pigmentepithel.
Fig. 16. Inner segments of the photoreceptors in the same specimen showing reduced concentration of mitochondria and unusually conspicuous cilia. Heavy arrows indicate the terminal bars representing external limiting membrane; light arrows indicate the cilia.

Abb. 16. Innensegmente der Lichtrezeptoren desselben Präparates mit schwächerer Konzentration der Mitochondrien und aussergewöhnlich deutlichen Zilien. Fett gedruckte Pfeile zeigen die Schlussleisten, die die äussere Grenzmembran darstellen; fein gedruckte Pfeile zeigen die Zilien.
Fig. 17. Abiotrophy in dog 11 months old (B1b). The outer segments of the photoreceptors are severely fragmented and their laminated platelets are disoriented. The inner segments showed marked reduction in mitochondria. The pigment epithelium was normal except for pleating of its inner surface.
Is, inner segments; Os, outer segments; PE, pigment epithelium; heavy arrows, external limiting membrane.

Is, Innensegmente; Os, Aussensegmente; PE, Pigmentepithel; fette Pfeile: äussere Grenzmembran.
**Fig. 18.** Abiotrophy in dog 32 months old (A2b). The photoreceptors have disappeared entirely and the external limiting membrane, consisting of a series of terminal bars, lies directly apposed to the folded inner surface of the pigment epithelium. M, Müller cells in the retina; PE, pigment epithelium; heavy arrows, external limiting membrane.

the rods and cones were normal. The pigment epithelium, choriocapillaris and optic nerves were normal. Flat mounts of the retinal vessels showed normal cellularity. Tetrazolium diaphorase was normal using lactate DPN and succinate substrates and the glycogen content as well as the glycogeneogenesis were normal.

The electron microscopic findings were, however, interpreted as abnormal. The laminated discs in the outer segments of the rods and cones were disarranged (Fig. 15) in comparison to the regular lamination of the controls and the cilia connecting the outer and inner segments were unusually conspicuous and extended anomalously to the base of the photoreceptors (Fig. 16). The mitochondria in the inner segments were also less abundant than normal. Within the retina proper the only abnormality was an increased pleating of the glial membranes. On the other hand the pigment epithelium and choriocapillaris were normal.

Two of the night blind siblings of the foregoing case were examined when they were 11 months of age. In the meantime they had become progressively night blind (as had the third sibling not examined) and the retinal arteries were interpreted as having become narrower. Electroretinography at that time showed no recordable response in one dog and a minimal response in another while a normal control dog showed a good response.

Case B1b. One of these siblings was sacrificed (11 months of age) and the tissues examined by light and electron microscopy.

Hematoxylin and eosin stains showed no definite abnormality and periodic acid-Schiff stains showed minimal glycogen. Sections incubated in glucose, however, showed glycogen synthesis in Müller's cells that was thought to be approximately normal. Tetrazolium preparations incubated with lactate-DPN and succinate were also interpreted as either normal or only slightly less than normal.

Electron microscopy showed advanced disorganization of the outer segments of the rods and cones (Fig. 17). Portions of these outer segments appeared to be detached fragments lying in the subretinal space with irregular arrangement of their lamination. The cilia were perhaps thickened but were not as elongated as in the preceding specimen. The pigment epithelium was present but showed excessive pleating of its inner surface. The synaptic organs in the outer plexiform layer were suggestively decreased in number and the Müller cells showed a non-specific packing and parallel stratification about the blood vessels (such as we have seen in retinas with gliosis).
Fig. 19. Abiotrophy in dog 4 years old (A1). The retina (upper half of picture) is thin and gliosed. The photoreceptors are entirely absent but a single terminal bar represents a portion of the external limiting membrane. The pigment epithelium is normal although its villous processes project freely in the subretinal space since the photoreceptors are lacking. A pigment epithelial cell has invaded the retina (right middle). PE, pigment epithelium; heavy arrow, terminal bar; double arrows, internal limiting membrane.
Comments

The name photoreceptive abiotrophy is suggested as most effectively designating the entity which we are describing in the present report. Photoreceptive indicates the predominant involvement of the photoreceptors while abiotrophy refers to the degeneration of a tissue which has matured normally and then undergone secondary degeneration. The name seems to us preferable to progressive retinal atrophy which would etymologically have to include several types of primary progressive retinal degeneration in addition to the photoreceptive type, and it is certainly preferable to the term retinitis pigmentosa which is a misnomer by all standards. This latter name is established in human nosology for historical reasons but use of it can only lead to confusion if applied to the analogous abnormality in animals.

The photoreceptive abiotrophy here described appears to be a mendelian recessive abnormality which produces signs in dogs at about the sixth to eighth month of life. It thus comes on later than the comparable abnormality reported in mice but much earlier than that occurring typically in man. From these few observations it would appear that the rapidity of onset and course parallel the life span of the species.

Like retinitis pigmentosa of man and the comparable condition in mice the characteristic changes in dogs consist of early extinction of the electroretinogram, predominant degeneration of the rods and cones, and a paradoxical narrowing and obliteration of the retinal vessels—paradoxical because these vessels do not supply the photoreceptive layer of the retina. The migration of pigment into the retina which is such a conspicuous feature in man occurs also in the dog but in our cases did so only after the condition had been present 1–2 years. The asteroid spots in the vitreous found in one of the dogs does not occur significantly with retinitis pigmentosa of man but is a non-

Fig. 20. Inner portion of retina of same specimen showing portions of glial cells and part of a single ganglion cell. Nuc, nucleus of ganglion cell; er, ergastoplasm; ly, lysosomes; double arrows, internal limiting membrane.

specific and relatively frequent finding with various types of retinal degeneration in dogs. The preservation of the protrusion of the disc despite extensive retinopathy and blindness is significant in indicating that the optic nerve and ganglion cell layer are preserved.

The present series of five cases permits a first-order attempt to reconstruct the sequence of morphologic changes occurring in the retina. At the onset of symptomatic night blindness the retina shows no morphologic abnormality by light microscopy nor by histochemical studies of lactate and succinate diaphorase or glycogeneogenesis. Electron microscopy, however, reveals disarrangement of the laminated plates in the outer segments of the rods and cones, fragmentation of the outer segments and suggestive abnormalities in the cilia of the photoreceptors. Similar disarrangement of the outer segments were reported by Dowling in rats with retina atrophy or with vitamin A deficiency.

After the night blindness has been present for 1–2 years all the photoreceptors will have disappeared but remnants of the outer nuclear layer will still be present about the disc and spottily present throughout the rest of the retina. The entire peripheral retina will have become thinned and in places it will be reduced to a tenuous glial membrane. No pigment has migrated into the retina at this stage. By one or two years after the onset of the night blindness, the gliosis will have become so severe as to destroy the nuclear lamination of the retina throughout the periphery although scattered ganglion cells are still present and the inner layers are still identifiable about the disc. At this stage pigment epithelial cells in the periphery have migrated into the retina. These form brown clumps that can be seen with the ophthalmoscope and cell clusters that can be seen with the microscope. Such clumps are particularly apt to be associated with areas of thinning or rupture of the retina and to surround the blood vessels. Similar clusters of non-pigmented epithelial cells occur in the retina overlying the tapetum. The absence of pigment in these latter cells permits one to detect their pathologic loading with glycogen granules.

Except for migration of its cells into the retina the pigment epithelium may remain normal even late in the disease. The optic nerves also remain histologically normal. The smaller retinal blood vessels become occluded but there is no histologic evidence for this at a stage when the arterial narrowing is first noted clinically.

Since photoreceptive abiotrophy occurs in mice, dogs, and man it probably will be found eventually in other species as well. One paper
describes the condition in cats\textsuperscript{80} and we incidentally processed the 
eyes of a cat which had identical pathologic changes as in the dogs but we were unable to obtain genealogic or clinical data to corroborate a possible abiotrophic etiology. We also had occasion to examine the 
eyes of several dogs (and human beings) showing loss of photoreceptors in the retinal periphery as a senile manifestation. Evidence does not permit a conclusion as to whether these senile changes do or do not have an hereditary basis but they are obviously different from the present abiotrophy which begins at an early age and involves the entire retina. Virus infections are frequently cited as causing retinopathy in dogs with blindness similar to that of the inherited disease but the viral cases we have had an opportunity to study showed patchy involvement of all layers of the retina, and unlike the abiotrophy, they had produced acute symptoms within a few days’ time. Finally, there are several retinotoxic agents in man and animals which produce a degeneration of the photoreceptors comparable to those of an abiotrophy. Best documented are chloroquine and Mellaril\textsuperscript{®} (Thioridazine) in human beings and iodoacetic acid in animals (rabbits). These instances are cited to emphasize the importance of making a diagnosis of photoreceptive abiotrophy only when there is clearcut evidence of hereditary transmission and typical clinicopathologic findings.

To date it cannot be said that any lead points unequivocally to the pathogenesis of photoreceptive abiotrophy in man or animals. The finding of obliteration of the choriocapillaris in some cases was not borne out in other cases. One might logically expect a missing enzyme in the vitamin A-retinene cycle but none has been reported. Graymore\textsuperscript{41} has found a specific and early defect in a lactic acid dehydrogenase isoenzyme of rat retinas with photoreceptive abiotrophy. A suggestive finding in the present cases was the unexpected paucity of glycogen in some of the affected retinas and the absence of the capacity for glycogen synthesis by the retinal glia but this was not evident early in the disease.

\textit{Summary}

An idiopathic degeneration involving predominantly the photoreceptors of the retina occurs in the elkhound similar to that which has been reported in other breeds of dogs and analogous to that which occurs in mice and men. It is suggested that the term applied to the comparable condition in man, retinitis pigmentosa, is inappropriate. A preferred name is photoreceptive abiotrophy of the retina.
Photoreceptive abiotrophy in the present eight cases appeared to be a mendelian recessive trait producing signs at 6–8 months of age consisting of night blindness, narrowing of the retinal vessels and extinction of the electroretinogram. Pathologically it consists of degeneration of the photoreceptive cells and paradoxic obliteration of the retinal capillaries. Electron microscopy reveals disorganization and fragmentation of the outer segments of the rods and cones and pleating of the inner surface of the pigment epithelium, long before any abnormality is detectable by light microscopy.

The pathogenesis of photoreceptive abiotrophy is obscure but in the early case here reported no abnormality was detected in diaphorase activity nor in glycogeneogenesis although there was a paucity of glycogen in the retinal glia and an absence of the normal capacity for glycogen synthesis at a later stage.

Zusammenfassung


Die Pathogenese der Abiotrophie der Lichtrezeptoren ist unbekannt; jedoch wurde bei einem der hier beschriebenen frühen Fälle weder eine Anomalie der Diaphoraseaktivität noch der Glykogenogenese entdeckt, obgleich zu einem späteren Zeitpunkt eine Glykogenausfällung der Retina Glia und Fehlen der normalen Fähigkeit der Glykogensynthese feststellbar war.

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