What You Should Know About Retinitis Pigmentosa

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Retinitis pigmentosa is the name given to a whole family of diseases in which the primary symptoms are difficulty seeing under low light conditions and loss of side vision. Because so much is misunderstood by the lay public about this group of diseases, this pamphlet attempts to demystify them and help patients and their families to understand the situation better.

The Retina

The retina is the lining of the back of the eye upon which the light is focused. It has two types of cells called cones and rods. The cones are responsible for color vision and fine central vision (the perception of details, reading, etc.) The rods are responsible for peripheral vision and night vision. Figure 1 shows that most of the rods are in the peripheral retina. Most of the cones are in the central retina. Cones work under conditions of bright light. The rods work in dim light. In retinitis pigmentosa, generally the rods are more severely diseased, accounting for the prominent problems with night vision and side vision. Figure 2 shows what a person with advanced “tunnel vision” (loss of side vision) might see, compared to normal. The cones are not normal either, but they are usually affected less severely and later in the course of the disease.

Genetics of Retinitis Pigmentosa

Retinitis pigmentosa is a disease caused by a gene defect. That is, one is born with the disease and cannot acquire it from the environment. It is possible to develop the gene defect in two ways.

Fig. 1

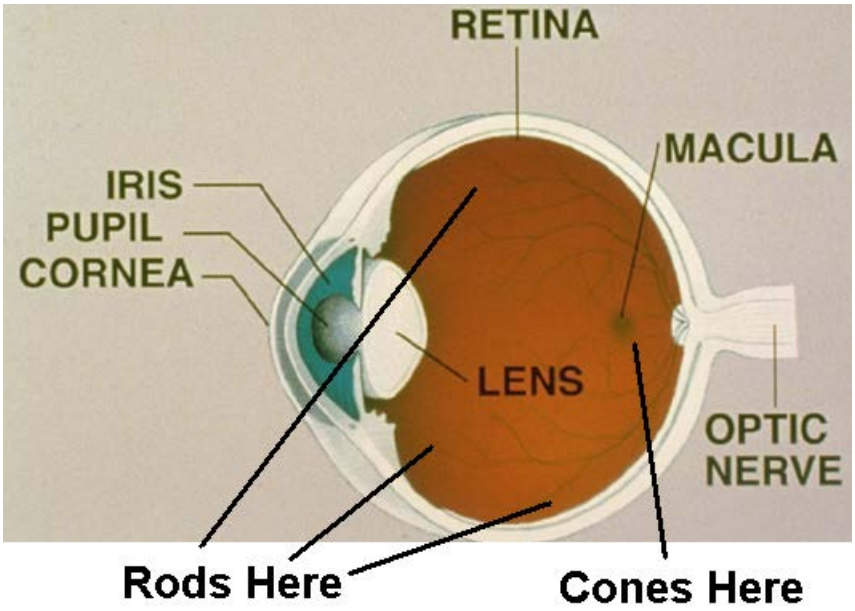
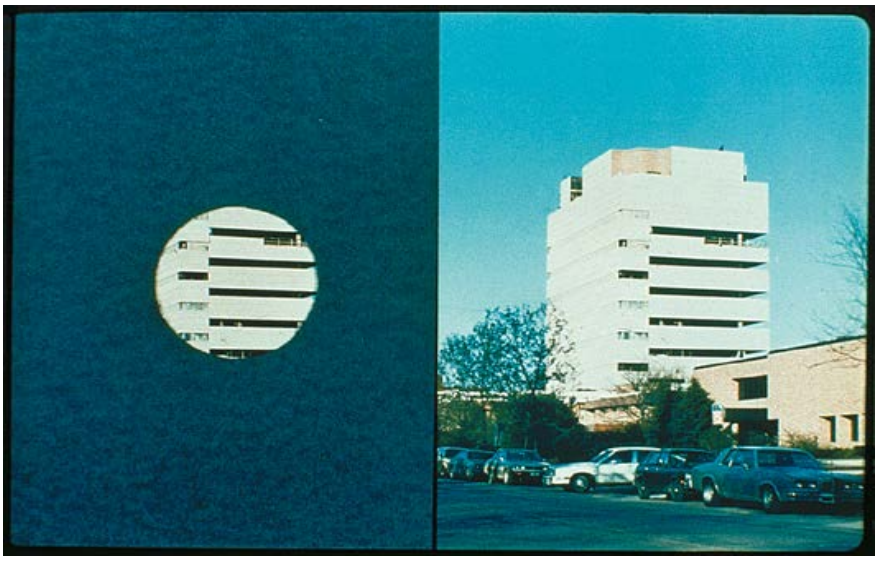


Fig. 2

Tunnel Vision Normal View



Some patients develop the genetic defect because the sperm or the egg from their parents suffered a mutation. This mutation is then the cause of the disease. Patients who develop the disease in this way will not have a family history of the disease. They will be the first in their families to have retinitis pigmentosa. They will be able to pass the genetic defect on to their children, however. Whether they will be able to pass the disease, as opposed to the genetic defect, will depend on whether the genetic defect is dominant, recessive, or sex linked. We will discuss what these terms mean below. In other patients, one or both parents have a genetic defect, which by chance, is passed down to the affected patient. In these cases a family history of the disease may be found which is why ophthalmologists place great importance on obtaining an accurate family history when the question of retinitis pigmentosa arises.

The most common genetic defect which can cause retinitis pigmentosa is a dominant one. This type of gene need only be contributed by one parent or the other to be able to express itself as the disease retinitis pigmentosa. Patients with the form of the disease have a 50% chance with each child they have of passing the disease to the child. Of patients in whom the pattern of inheritance can be determined, 46% have the dominant form.

Less commonly the genetic defect will occur on the X chromosome, which is one of the chromosomes determining sex. The Y chromosome is the other sex chromosome. If a person has two X chromosomes, that person is female. If a person has an X and a Y chromosome, that person is male. If the genetic defect for retinitis pigmentosa occurs on the X chromosome, it will be masked by the other, normal X chromosome in a female. In a male with the defective X chromosome, however, there is no second, normal X chromosome present to mask the defective one and thus the disease is expressed. For this reason, only males carrying the affected X chromosome will have the disease. Females can carry the defective gene but not have the disease. This is called the sex- linked form of retinitis pigmentosa. A woman who carries the defective gene has a 50% chance with each daughter that the daughter will also be a gene carrier (but will not have the disease). Each of the woman’s sons has a 50%

chance of having the disease. No son of an affected male will have the disease, but 100% of an affected male's daughters will be gene carriers (but not have the disease). Of patients in whom the pattern of inheritance can be determined, 8% have the sex linked form.

The last form of the disease is the recessive form. In this case, a person must have two defective genes to have the disease. In other words, each parent must contribute a defective gene to the affect patient. Since this would be extremely rare, the doctor seeks a history of consanguinity between parents of the affected person. That is, are the parents perhaps distantly related as a way of accounting for the fact that they both have the same defective gene? If an affected patient marries an unrelated spouse, the chances of any child having the disease are nearly zero. On the other hand, all the children would carry the recessive gene. Of patients in whom the pattern of inheritance can be determined, 20% have the recessive form.

Diagnosing Retinitis Pigmentosa

A detailed history of symptoms and a detailed family history are the basis for the ensuing complete eye examination, which requires dilation of the pupils and use of the slit lamp, headlight, and various lenses. All patients require visual field testing to determine the status of the peripheral vision. All patients require photographs of the retina to serve as objective baselines for comparison of disease progression in the future. The fundus looks different in retinitis pigmentosa than in a normal eye. Compare fig. 3, which shows a normal fundus, to fig. 4, which shows a fundus of a patient with retinitis pigmentosa. Finally, all patients require an electroretinogram for accurate diagnosis and information on the relative involvement of the cones as well as the rods. These are the minimum and some patients require other tests such as hearing evaluations and various blood tests and special types of photography involving the injection of dye in a vein.

Fig. 3 Normal Fundus

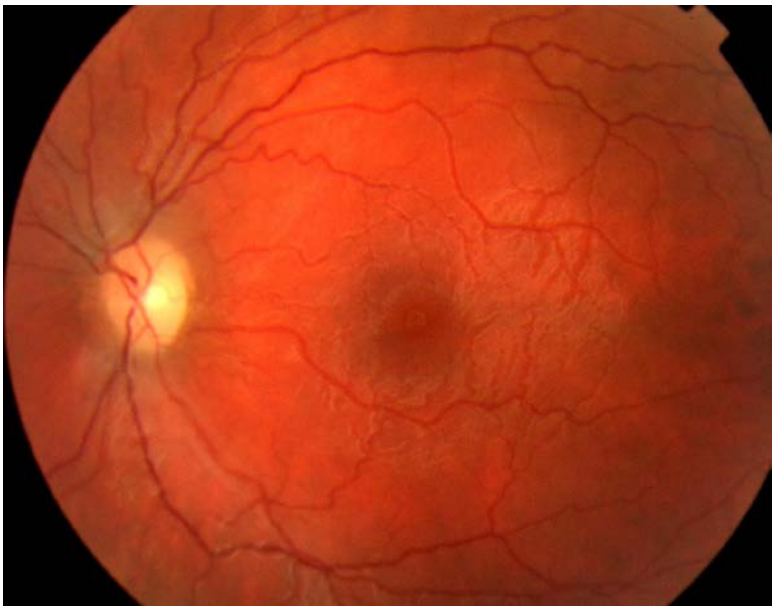
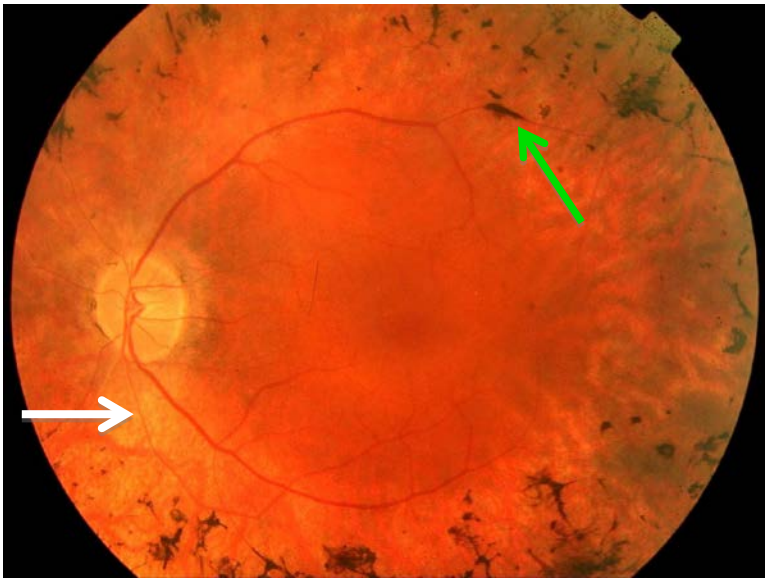


Fig. 4 Fundus of a Patient with Retinitis Pigmentosa



Legend: Note the thin arterioles (white arrow) and bone spicule pigment (green arrow) typical of this disease.

Associated Diseases

Certain types of retinitis pigmentosa are associated with hearing disturbances, irregular heart rhythms, blood disorders, and neurological disorders. Questions are asked to elicit these possible associations and if any clinical suspicion arises the appropriate tests to provide an answer are undertaken.

Treatment

At the present time there is no commonly applicable treatment for retinitis pigmentosa that is recognized by a consensus of ophthalmologists as effective, although active research is ongoing. There are experiments being conducted on gene therapy and retinal cell transplants, but despite promising pilot studies they are not ready for conventional use. Retinal prosthetic implants are being developed for end-stage cases, but these are not used for patients with less than very advanced disease. One study has concluded that 15,000 units per day of vitamin A palmitate will slow the rate of deterioration of the electroretinogram, but this study did not show a benefit in visual acuity or visual field for the patient; therefore the recommendation to use this much vitamin A is somewhat controversial. It is probably advisable to wear UV and blue wavelength blocking sunglasses for outdoor use since these wavelengths of light may be harmful in general to the retina. Patients should be put in touch with the National Society to Prevent Blindness (Schaumburg, IL) and the National Retinitis Pigmentosa Foundation (Baltimore, MD) for educational materials. Low vision aids are useful in advanced cases including magnifiers, closed circuit TVs, “night scopes”, and talking books. The frequently associated problems of cataract and macular edema are treated as they arise- cataracts with implant surgery and macular edema with Diamox and rarely laser therapy.

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