

## Retinitis Pigmentosa

Retinitis pigmentosa (RP) is a disease that results in dystrophic tissue architecture of the retina and/or retinal pigment epithelium. Retinal degeneration occurs in a hereditary fashion. Clinical features include night blindness, defects in color and contrast vision, and tunnel vision (peripheral visual field loss). RP is associated with abetalipoproteinemia, Kearns-Sayre syndrome, and Refsum disease. On funduscopy, RP classically looks like bone spicule-shaped retinal deposits. Ultimately, this disease leads to permanent blindness.



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### Characteristics

#### Hereditary Retinal Degeneration

[Hair-Red-Kid with Red-Tins-Damaged](#)

RP is a hereditary disease that causes retinal degeneration. Several genes may cause RP, but mutations in the rhodopsin gene that code for an important G-protein coupled receptor in photoreceptor cells is implicated in the majority.

### Clinical Features

#### Night Blindness

[Nightgown and Blinds-over-eyes](#)

Night blindness, or nyctalopia, is due to the degeneration of rods. It can be assessed by measuring the time delay in dark adaptation or slow waves on electroretinogram.

#### Contrast and Color Defects

[Contrasting-Convict and Colors-Broken](#)

As the degeneration of rods proceeds, contrast sensitivity decreases. Finer details like shadows and depth become more difficult to perceive. Colors also start appearing less distinct.

#### Tunnel Vision

[Tunnel View](#)

In late or advanced stages, peripheral retinal degeneration can result in peripheral visual field loss, or tunnel vision.

### Associations

#### Abetalipoproteinemia

[A-beta-fish-lips-protein](#)

Abetalipoproteinemia is a malabsorption disease that results from mutations in the microsomal triglyceride transfer protein (MTTP) gene. It causes deficiencies of apolipoproteins B-48 and B-100.

## Kearns-Sayre Syndrome

### [Corn Syrup](#)

Kearns-Sayre syndrome is classified as an inborn error of metabolism disease. It is a mitochondrial myopathy characterized by ophthalmoplegia, retinitis pigmentosa, and cardiac defects.

## Refsum Disease

### [Referee](#)

Refsum disease is a hereditary motor and sensory neuropathy. It is due to an accumulation of phytanic acid and can cause retinitis pigmentosa.

## Diagnosis

### Bone Spicule-shaped Retinal Deposits

#### [Bone Towers with Black-smoke and Spiculated-moon](#)

On fundoscopy, several bone spicule-shaped retinal deposits may be seen. They progress from the periphery to the macula.

## Considerations

### Permanent Blindness

#### [Permed-Hair with Blinds-Over-Eyes](#)

Ultimately, this disease leads to permanent blindness as there is no cure.