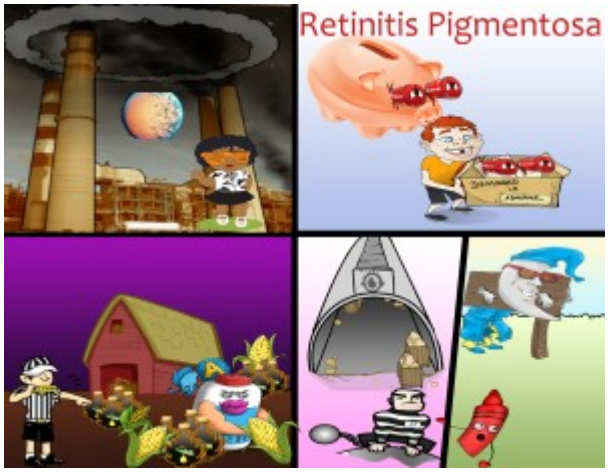
Retinitis Pigmentosa

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 fundoscopy, RP classically looks like bone spicule-shaped retinal deposits. Ultimately, this disease leads to permanent blindness.



PLAY PICMONIC

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.