Retinitis Pigmentosa

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Retinitis Pigmentosa
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Introduction
Retinitis pigmentosa can be diagnosed and measured by:

Genetic testing. This test looks at a sample of your blood or other tissues to see if you have certain genes that are associated with a disease. It can also help determine the likely course or severity of a disease.

Electroretinography. This test measures the electrical activity in the retina, or how well the retina responds to light. It works much like the EKG, which measures the activity of the heart.

Visual field testing. Retinitis pigmentosa can affect your peripheral (side) vision. Visual field testing helps measure your side vision and find any blind spots that may be developing.

Optical coherence tomography. Also known as OCT, this imaging test takes special, highly detailed pictures of your retina. It can help diagnose RP and find out how it is affecting your retina. (Campenhout, 2018)

Pathophysiology
Retinitis pigmentosa is a genetic condition that can be passed down in families. The type and speed of vision loss from retinitis pigmentosa varies from person to person. It depends on the type of the condition

Diagnosis & Testing
What is Retinitis Pigmentosa (RP)?
Retinitis pigmentosa, also known as RP, refers to a group of inherited diseases causing a degeneration of the retina. Worldwide, RP affects 15,000 people. The retina is a thin piece of tissue lining the back of the eye. It converts light into electrical signals that the brain interprets as vision. People with RP experience a gradual decline in their vision, because photoreceptor cells in the retina degenerate. (Latham, 2017)

Since retinitis pigmentosa begins as rod degeneration, the patient first notices increasing difficulty in night vision, followed by difficulty seeing in the periphery.

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Topic Selection
“I chose this topic because of a family member who is currently living with retinitis pigmentosa. Not knowing much about the disease, I wanted to educate myself so that I would have a better understanding of the subject and be able to be a resource to him and other family members. Since it is a genetic mutation, I want to be informed in the instance that my husband or son develop symptoms of the disease”

Treatment & Prognosis
There is no single treatment, and no cure for retinitis pigmentosa.

Research shows that taking certain vitamins, including vitamin A palmitate, may help some people with retinitis pigmentosa.

Some patients develop swelling of the retina and may be helped by a certain type of drug called a “retinal detachments.”

Cataracts or clouding of the eye’s natural lens, may be helped by a certain type of lens implant.

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Signs & Symptoms
• Loss of side (peripheral) vision
• Loss of central vision (in advanced cases)
• Decreased vision at night or in low light
• Eye fatigue
• Difficulty seeing in dim light

Implications for Care
• Patients should have annual examinations, including visual field testing and periodic (every 5 years) ERG evaluations (Levinson, 2017)
• Low-vision specialists can provide magnifying devices and feeding-expanding lenses for patients with RP who have poor central vision.
• Genetic Counseling
• Optimal Safety

Significance of Pathophysiology
Retinitis pigmentosa can be inherited in one of three ways:

Autosomal recessive inheritance
In autosomal recessive inheritance, it takes two copies of the mutant gene to give rise to the disorder. An individual with a recessive gene mutation is known as a carrier. When two carriers have a child, there is 1 in 4 chance the child will have the disorder. In 2 chances the child will be a carrier. In 4 chances the child will neither have the disorder nor be a carrier.

Autosomal dominant inheritance
In this inheritance pattern, it takes just one copy of the gene with a disorder-causing mutation to affect another person. When a parent has a dominant gene mutation, there is a 1 in 2 chance that any children will inherit this mutation and the disorder.

X-linked inheritance
In this form of inheritance, mothers carry the mutated gene on one of their X chromosomes and pass it to their sons. Because females have two X chromosomes, the effect of a mutation on one X chromosome is offset by the normal gene on the other X chromosome. If a mother is a carrier of an X-linked disorder there is:
• In 2 chance of having a son with the disorder
• In 2 chance of having a daughter who is a carrier

(Levinson, 2017)

References

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