August 2018

Retinitis Pigmentosa

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Introduction

What is Retinitis Pigmentosa (RP)?

Retinitis pigmentosa, also known as RP, refers to a group of inherited diseases causing retinal degeneration. Worldwide, RP affects 1:5,000 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.

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 their form of the condition

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"

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Diagnosis & Testing

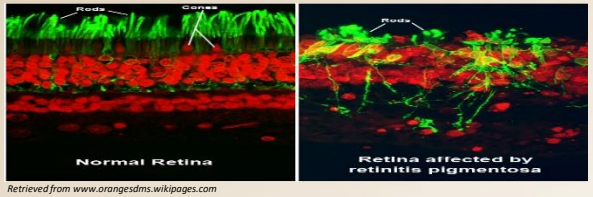
Retinitis pigmentosa can be diagnosed and measured by:

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 and health 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. (Campochiaro, 2018).



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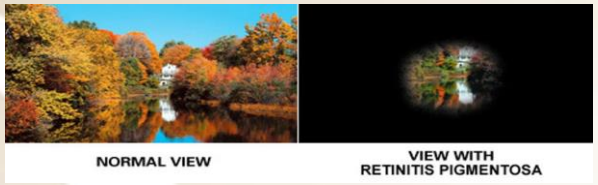

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Treatment & Prognosis

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 eye drop.

Cataracts or clouding of the eye's lens may also develop and surgery to treat this might be helpful for some patients.

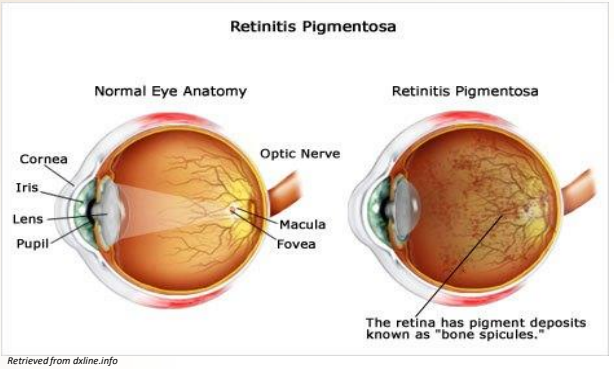
There is also an "artificial retina" called the ARGUS II implant, which may be helpful for some patients with severe vision loss due to retinitis pigmentosa (Ahuja, 2013)

Pathophysiology

RP is an inherited disorder that results from harmful changes in any one of more than 50 genes. These genes carry the instructions for making proteins that are needed in cells within the retina, called photoreceptors. Some of the changes, or mutations, within genes are so severe that the gene cannot make the required protein, limiting the cells function. Other mutations produce a protein that is toxic to the cell. Still other mutations lead to an abnormal protein that doesn't function properly (Hafler, 2016).

RP is characterized by changes in pigment and arteriolar attenuation, often with some degree of optic nerve atrophy. Post-mortem examination has shown that the pigmentation is caused by cells from the pigment epithelium budding off and settling within the layers of the neural retina. In the late stages of RP a thinning of the retinal blood vessels is seen, probably resulting from the loss of many retinal cells reducing the need for blood (Yang, 2018).

A variety of retinal molecular pathway defects have been matched to multiple known RP gene mutations. Mutations in the rhodopsin gene, which is responsible for the majority of autosomal-dominantly inherited RP cases, disrupts the rod-opsin protein essential for translating light into decipherable electrical signals within the phototransduction cascade of the central nervous system (Yang, 2018).



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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 field-expanding lenses for patients with RP who have poor central vision.
- Genetic Counseling
- Optimize Safety

Significance of Pathophysiology

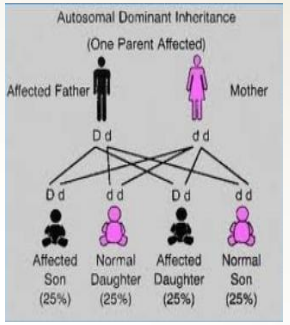
RP can be inherited in one of three ways:

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 a:
•1 in 4 chance the child will have the disorder
•1 in 2 chance the child will be a carrier
•1 in 4 chance 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 bring about the disorder. 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 a:
•1 in 2 chance of having a son with the disorder
•1 in 2 chance of having a daughter who is a carrier

(Levinson, 2017)



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Conclusion

- Retinitis Pigmentosa is a genetic disease with no current cure.
- Earliest sign is night blindness
- There are treatments out there, however it is only to slow the death rate of rod and cone cells
- Finding a cure is still years away, however researchers have already started human clinical trials
- Some people develop RP even without a family history
- Genetic testing is available to those who think they may have or carry the trait.
- Safety is priority #1 for patients with RP

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