What can we expect in the future?

Much research is being performed in the area of RP, and at RCTX, we are passionate about finding a treatment.

WHY IS GENETIC TESTING IMPORTANT?

Since RP can be caused by one of many different possible genes, testing can help narrow down the exact source. It can also:

- Predict a possible future prognosis
- Identify the impact on family members such as siblings, cousins, children, and grandchildren
- Open up the possibility of future research, clinical trials, and gene therapy

WHAT RESEARCH IS BEING DONE?

Gene therapy is the study of replacing or modifying mutated genes in the retina to help them resume normal function.

Gene and cell therapies are the current focus of RP-related research. In 2017, the FDA approved the first gene therapy treatment called Luxturna for a specific inherited retinal disease. Since then, additional efforts have focused on developing therapies that target specific genes or cells to help slow the progression of disease, and even improve symptoms.

Optogenetics is another significant area of research. It uses gene editing to bypass damaged retinal cells and give other parts of the retina the ability to detect light.

These treatments are currently being studied in clinical trials underway at RCTX.

If you are a patient with RP and are interested in a clinical trial or a research opportunity, please let your doctor know.

RETINA CONSULTANTS OF TEXAS IS PASSIONATE ABOUT FINDING A TREATMENT FOR RETINITIS PIGMENTOSA (RP) AND IS PERFORMING RESEARCH IN THIS AREA. IF YOU HAVE RP AND ARE INTERESTED IN PARTICIPATING IN A CLINICAL TRIAL, PLEASE LET YOUR DOCTOR KNOW.

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Scan here to watch a video featuring our own Dr. Kenneth Fan sharing helpful video that covers inherited retinal diseases such as Retinitis Pigmentosa.

Retina Consultants of Texas

RCA Research

Retina Consultants of Texas

RCA Research

RETINITIS PIGMENTOSA

What is retinitis pigmentosa (RP)?

Retinitis Pigmentosa (RP) is an inherited retinal disease that most patients have from birth. It most commonly causes problems with night vision and peripheral vision.

There are many different types and genetic causes. Some patients develop symptoms early in childhood, but some will not have symptoms until later in life. Because of the genetic cause, patients may sometimes have a strong family history with parents, grandparents, or siblings also affected.

How RP affects vision

Light enters your eye and is detected by the **retina**, light-sensing nerve tissue lining the back of your eye. The information it receives is transmitted through the **optic nerve** to the brain, where it is interpreted as the images you see.

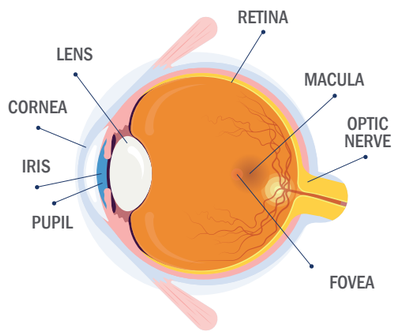
The retina has two types of cells (called **photoreceptors**) that gather light: **rods** and **cones**. RP causes degeneration of the rods and cones, as well as the retinal pigmented epithelium (RPE), which results in vision loss. These changes in the RPE result in classic examination findings called **bone spicules**.

The **rods** are around the outer ring of the retina and are active in dim light. Most forms of RP affect the rods first. You may lose your side (peripheral) vision and the ability to see at night.

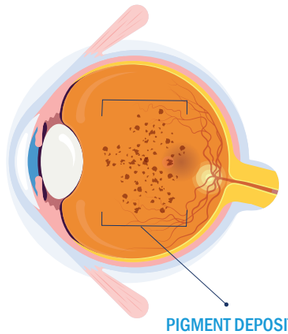
In rare cases, RP can affect your cones and central vision as well. Cones are mostly in the center of your retina. They help you see color and fine detail. When RP affects cones, you slowly lose your central vision and your ability to see color.

Some patients may have other medical issues, such as problems with hearing or kidney function.

HEALTHY EYE



AFFECTED EYE



Houston: 800.833.5921 San Antonio: 210.903.1046 retinaconsultantstexas.com

Risk factors for retinitis pigmentosa

RP is a genetic condition. This means that it was caused by genes passed down from one or both parents. This does NOT always mean that a patient will pass the disease on to their children.

Over 100 different genes can cause RP. When one of these genes is not functioning properly, key parts of the visual cycle in the retina may be missing, which may affect your vision. It is important to speak to a genetic counselor or your inherited retinal disease specialist about any questions or concerns.

Most common genes that can cause RP:

- USH2A
- CRB1
- CEP290
- PDE6B
- EYS

Symptoms of retinitis pigmentosa

- Loss of night vision (usually starting in childhood; parents may notice that children have trouble moving around in the dark or adjusting to dim light)
- Loss of side (peripheral) vision, so you have trouble seeing things out of the corners of your eyes. Over time, your field of vision narrows until you only have some central vision (aka **tunnel vision**).
- Sensitivity to bright light
- Loss of color vision

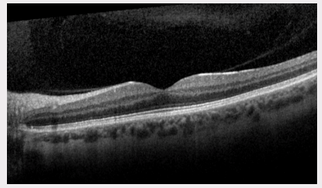
How Your Field of Vision Changes as RP Progresses

Examination and diagnostic testing

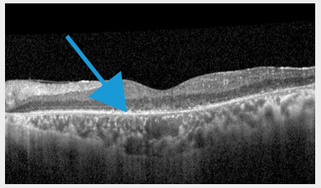
RP is diagnosed by your retina specialist or eye care specialist. It is diagnosed using a combination of a thorough medical history, physical exam, diagnostic testing and possible genetic testing. Diagnostic testing includes **optical coherence tomography** (OCT), **fundus photography**, **fundus autofluorescnce** (FAF) and **fluorescein angiography** (FA).

WHAT YOUR DOCTOR SEES

On OCT

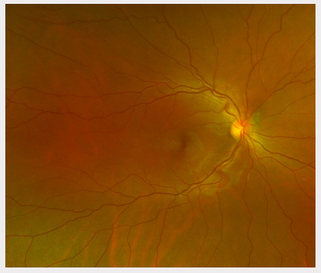


Normal macula

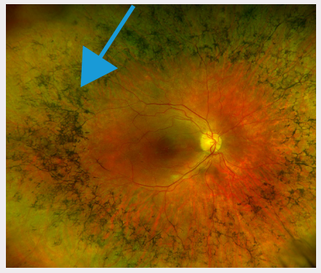


Example OCT with loss of photoreceptors and RPE atrophy in a patient with retinitis pigmentosa

On Fundus Photography



Normal macula



Example retina color photograph in a patient with retinitis pigmentosa demonstrating bone spicules, vascular attenuation, and retinal atrophy

Treating retinitis pigmentosa

Although there is much research and development being performed, there is presently no cure for retinitis pigmentosa. Clinical trials are currently being performed in the areas of gene therapy, optogenetics, and even stem cell treatments (see back page for more information). Aside from newer research opportunities, low vision aids and occupational rehabilitation/training can help make the most of your vision. In addition, please ask your retina specialist about:

- Vitamins and supplements
- Protective eye wear
- Eye drops for cystoid macular edema