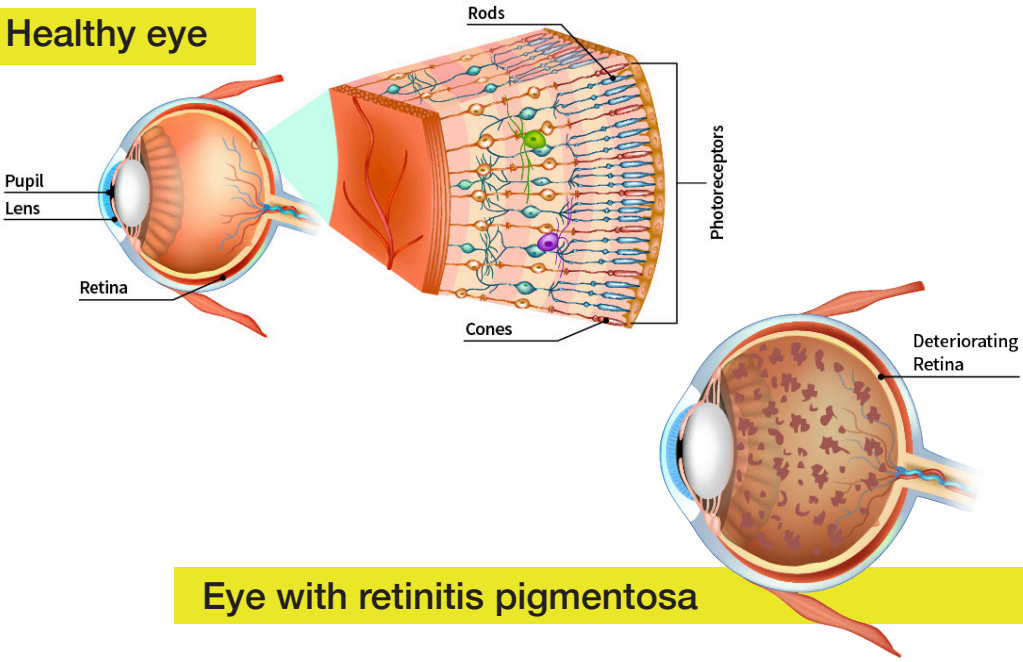
Retinitis Pigmentosa (RP)

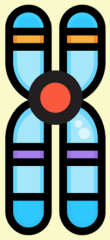
What is Retinitis Pigmentosa (RP)?

Retinitis pigmentosa (reh·tuh·nai·tuhs pig·muhn·tow·suh), also known as RP, is a group of inherited retinal diseases (IRDs) that affect how we see. RP affects the **retina**, which is in the back of the eye. The retina captures images and sends them to the brain to help us see. In people with RP, the cells of the retina that allow us to see images, called **photoreceptors**, slowly die off causing vision loss. How much and fast RP progresses differs from person to person. Many people living with RP lose their vision slowly over decades, typically starting with the peripheral vision. RP affects about 80,000–110,000 people in the United States.



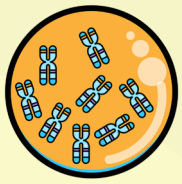
What are genes?

Cells are the basic building blocks of all living things.



Chromosomes are made up of **DNA**—the body's hereditary material.

A **gene** is a small section of DNA that contains the instructions for a specific molecule in the body, which is passed from parent to child.



The command center of each cell is called the **nucleus**, and it contains **chromosomes**.

Each gene contains the information required to build specific **proteins** needed in the body, for example, proteins build bones, determine eye color, allow muscles to move, control digestion, and keep your heart beating.

If there is a change in a gene's DNA sequence, it is called a **variant**. Not all variants cause disease. A disease-causing variant is called a pathogenic variant or **mutation**.

What causes RP?

RP is a genetic condition. This means that there is a gene that is not working properly causing someone to have a health problem such as RP. It's important to identify the specific gene causing your RP because over 79 genes have been found to cause retinitis pigmentosa. Different mutations can affect the retina in various ways. Genetic information about your RP is very helpful because it provides important details to your eye doctor for managing the disease and providing information to your family.

What are the symptoms of RP?

RP may develop in childhood or as an adult. First symptoms of RP are often:

- Difficulty seeing at night or in low light
- Loss of peripheral (side) vision

As the condition gets worse, you may notice:

- Difficulty with glare in bright conditions, depth perception, and color identification
- Further reduction of peripheral vision (it feels like you are looking through a tunnel)
- Changes in your central vision (central vision is necessary for tasks such as reading, driving, and recognizing faces)

Symptoms will vary depending on your age, stage of the disease, and genetic cause of disease. Some people encounter different speeds of vision loss, while others may keep their central vision longer than others.

Involve your family members in your RP journey. This will help them understand what is happening to you when your RP progresses. Your vision loss can limit important day-to-day activities, such as reading, driving, playing sports, and household chores. Having a reliable and strong support system and finding resources can help you keep your quality of life.

How is RP diagnosed?

RP may take longer to diagnose than other, more commonly occurring vision conditions. The diagnosis typically starts with an eye examination with an eye doctor (an optometrist or ophthalmologist). If the eye doctor thinks that you have an IRD, such as RP, they will ask you to see a retina specialist or an eye doctor more experienced in diagnosing and treating IRDs. The retina specialist will conduct further testing to confirm your diagnosis. They will conduct a very detailed examination including the steps listed below.

The goals of the detailed examination are to:

- Establish the specific diagnosis
- Determine the amount of vision loss
- Identify relevant treatments or interventions

Initial and follow-up examinations. May include the following:

- **Patient history:** The doctor will ask about your current vision issues, medical history, and medication use.
- **Family history:** This will include putting together a family medical history to show genetic relationships and medical disorders that occur in your family. The family medical history can help the healthcare team understand the diagnosis and estimate who else in your family may have a chance for developing the RP.

- **Clinical eye examination.** The eye examination includes:
    - **Visual acuity:** A measure of your ability to see detail up close and in the distance.
    - **Slit lamp:** A non-invasive procedure that uses a microscope and bright light to look at different parts of your eye.
    - **Dilation:** Eye drops are put into your eye to widen the opening on the front of your eye, called the pupil. This allows the eye doctor to examine the health of your retina.
    - **Indirect ophthalmoscopy:** A noninvasive tool worn by the eye doctor that provides magnification, allowing them to examine your retina in the back of your eye through the dilated pupil.
- **Imaging:** The eye doctor will take a series of pictures to view different parts of your eye, including:
    - **Retinal photographs:** A photo taken of your retina, inside of the eye.
    - **Optical Coherence Tomography (OCT):** An imaging device that takes cross-sectional pictures of the retina. This allows your ophthalmologist to map and measure the thickness of your retina.
    - **Fundus autofluorescence:** A non-invasive imaging test that can map in the retina, highlighting areas of your retina that have been affected by RP.
    - **Visual Field Testing:** A test that measures central and side or peripheral vision.
    - **Electroretinography (ERG):** A test to measure the electrical response of your eye's light-sensitive cells, called rods and cones. To avoid discomfort during the test, the eye doctor will place drops in your eyes.

Why is Genetic Testing Important?

Identifying the genetic cause of disease is an important part of care for patients with Inherited Retinal Diseases (IRDs). Many times, the exact type of IRD a person has can be difficult to determine based only on tests conducted in the eye doctor's office.

Results from genetic testing can lead to a more accurate diagnosis if a genetic cause of disease is identified. Having a genetic diagnosis may help to:

- Identify potential treatment options or clinical trial options for you
- Determine the chance for other family members to develop the condition
- Identify the potential chance for other health concerns to develop (in some cases, other parts of the body may be affected)
- Inform you about what will happen to their vision over time

A health care provider will order the genetic test, collect the sample, and review the results with the patient. Ideally, the health care team would include a genetic counselor to guide the patient and their family through the results of the genetic testing, discuss the impact on other family members, and assist couples who have questions around family planning decisions. It is often necessary to have genetic testing done if you are interested in joining a clinical trial, or if it has been several years since your last genetic test was completed.

- **Genetic testing:** You may meet with a genetic counselor (someone that specializes in the genetic testing process and educating patients) to determine the testing approach that is best based on the other test results. People living with RP are often asked to provide a blood or saliva sample that will be sent to a lab for analysis. The genetic test confirms or disputes the diagnosis of a specific IRD, so that correct information is provided to the patient and family. Genetic testing is often a requirement for participation in IRD-related clinical research trials.

Once the diagnosis is established, in addition to providing any relevant medical management, the doctor may:

- Connect you to supportive services such as a genetic counselor (someone that specializes in the genetic testing process and educating people living with RP) and vision rehabilitation
- Help your family members understand their risk for the disease
- Provide information to you about clinical trials and new therapies in development

What is the treatment for RP?

There are currently no FDA-approved treatments for RP. There is one exception for a rare type of RP caused by the RPE65 gene that can be treated by gene therapy. However, there are several clinical trials in process that may lead to treatment options. Continue to see your eye doctor every 1-2 years or as recommended. It is important to check your eyes regularly as other eye health conditions can occur. If at any time you feel there has been a change in your vision, you should see an eye doctor and have a dilated eye examination. It is also important to have a current record of an eye exam if you would like to join a clinical trial.

Those with RP usually develop cataracts at younger age than other people do who do not have RP. A cataract is a haziness or opacification of the natural lens in the eye, and may be treated with cataract surgery.

People with RP are also more likely to develop swelling or edema in the retina (cystoid macular edema), which can be treated with an eye drop or possibly an oral pill to decrease the swelling.

How can I manage my RP?

There are steps you can take now to protect your eyes and slow vision loss:

- Find vision rehabilitation providers who can help you and your family in finding ways and resources to navigate daily life with greater independence
- Wear a hat and sunglasses to protect your eyes from sunlight when you go outside
- Find a supportive community where you can connect with other people who are living with RP
- Consider talking to a mental health professional, who can help you process this diagnosis and find ways to keep you engaged in your goals

- Get protective eyewear when playing sports (PreventBlindness.org/tips-for-buying-sports-eye-protectors) or working in or around potentially dangerous conditions (PreventBlindness.org/preventing-eye-injuries-at-work). This includes work in manufacturing, construction, landscaping, auto repair, plumbing, woodworking, agriculture, mining, metal fabrication and health caree
- Ask your eye doctor about participating in a clinical trial (PreventBlindness.org/clinical-trials-for-eyediseases-and-vision)
- Do not smoke and avoid secondhand smoke exposure
- Try to incorporate green leafy vegetables, fish or fish oil, and antioxidant rich foods into your diet

Find more information about RP

- **National Eye Institute - Retinitis Pigmentosa:** www.nei.nih.gov/learn-about-eye-health/eye-conditions-and-diseases/retinitis-pigmentosa
- **Foundation Fighting Blindness:** www.fightingblindness.org/diseases/retinitis-pigmentosa
- **Prevent Blindness Videos:**
    - Webinar - Inherited Retinal Diseases and Children's Vision and Eye Health: What You Need to Know: youtu.be/OZpdT_8Phk4?si=lYA8ICvOtxsafnKz
    - Interview - Inherited Retinal Diseases and Retinitis Pigmentosa: youtu.be/5uhvIEIFlQQ?si=ZL0g-V7V58wQvSud

Resources for those with RP

- **Vision rehabilitation center:** Find a vision rehabilitation center so you can learn how to maximize the use of the vision do have to help you work, do daily activities, and do the things you enjoy. (lowvision.preventblindness.org/finding-low-vision-rehabilitation-services)
- **Living Well with Low Vision:** Prevent Blindness offers resources and help finding low vision clinics near you and clinical trials for individuals with vision loss on the Living Well with Low Vision website. (LowVision.PreventBlindness.org)
- **Blind Life:** Blind Life website (TheBlindLife.net) and YouTube (YouTube.com/c/theblindlife) channel provides videos, tips, and insight for those living with vision loss.

"Being diagnosed with RP in my early 30's was a devastating blow. But getting connected with the valuable resources available has allowed me to not only advocate for myself, but guide others to do the same. Now I realize that this diagnosis has provided me so many opportunities, connections, and blessings that I might otherwise have not experienced."

– Brenda N., Certified ADA coordinator and individual living with RP

- **Thriving Blind Academy:** Thriving Blind Academy is dedicated to promoting literacy, employment and financial awareness in the blind community. Free and paid programs are available for people of all ages. (ThrivingBlindAcademy.org)
- **Hadley:** Hadley offers practical help, connection and support free of charge to anyone with visual impairment, their families and professionals supporting them. (HadleyHelps.org)
- **Guide Dogs:** Many patients with RP disease and vision loss benefit from use of a guide dog. A guide dog is a specially trained service animal that helps an individual with vision loss or blindness. Information on guide dog services in the United States can be found at:
  - www.guidedogsofamerica.org
  - www.guidedogs.com/client-programs/guide-dog-program

Resources for being/staying active in the workforce

- **Rehabilitation Services Administration (RSA):** RSA State Liaisons, Project Officers, and State Teams may respond to questions or provide help to the public, individuals with disabilities, and other stakeholders. Contact your state liaison: rsa.ed.gov/about/people/state-liaisons
- **National Assistive Technology Act Technical Assistance and Training (AT3) Center:** The mission of the AT3 is to increase access to assistive technologies for those with disabilities across their lifespan. Your state's AT program can help you find the right device: at3center.net/state-at-programs

  They might:
  - Give you free information
  - Let you try out devices
  - Lend you equipment to test at home

Questions to ask your doctor:

- Should I speak to a genetic counselor and get genetic testing?
- How often do you recommend getting an eye exam?
- Is there anything I can do to prevent my vision from worsening?
- Can you suggest a vision rehabilitation center in my area to help me?
- Are there low-vision aids that can help me as I lose my vision?
- Can you suggest resources to help me cope with my vision loss?
- Are there any treatments that might help me to see better?
- Are there other eye problems that can happen in association with RP?
- Am I eligible to take part in a clinical trial? Where is the closest center that conducts research and clinical trials for RP?
- What accommodations or resources are available for me as a student? As an employee?
- Can I continue to work, play sports, and do my favorite activities?

Participate in a support group for patients with RP or connect to an online community

- **Global Genes:** GlobalGenes.org
- **MD Support:** MDSupport.org
- **Eye2Eye:** Eye2Eye is is a free, phone-based, peer support program, designed to assist adults who are blind or visually impaired and their families: shp.rutgers.edu/psychiatric-rehabilitation/eye2eye

Complete genetic testing

- Learn more about IRDs and benefits of genetic testing at FightingBlindness.org/genetic-testing
- **Find a provider to help you access genetic testing:** The Foundation Fighting Blindness's list of Retina Doctors makes it easy to find a genetic testing specialist or health care provider in your area of the United States who can discuss your testing options and assist you in learning more about your IRD. (FightingBlindness.org/retinal-specialists)

Stay up to date on clinical trials

- Stay up to date on clinical trials. Learn more about clinical trials for RP at ClinicalTrials.gov.
  - Learn about current clinical trials for RP disease: fightingblindness.org/news/retinitis-pigmentosa-research-advances-899
  - Search studies in the U.S. that are currently active or recruiting people who have RP: clinicaltrials.gov/search?cond=Retinitis%20Pigmentosa&aggFilters=status:rec&locStr=United%20States&country=United%20States