Usher syndrome

What is Usher syndrome?

Usher syndrome is the most common genetic condition that affects both vision and hearing. The major symptoms of Usher syndrome are hearing loss and vision loss from an eye disorder called **retinitis pigmentosa**, or RP.

Vision loss from RP can begin anywhere from early childhood to adolescence.

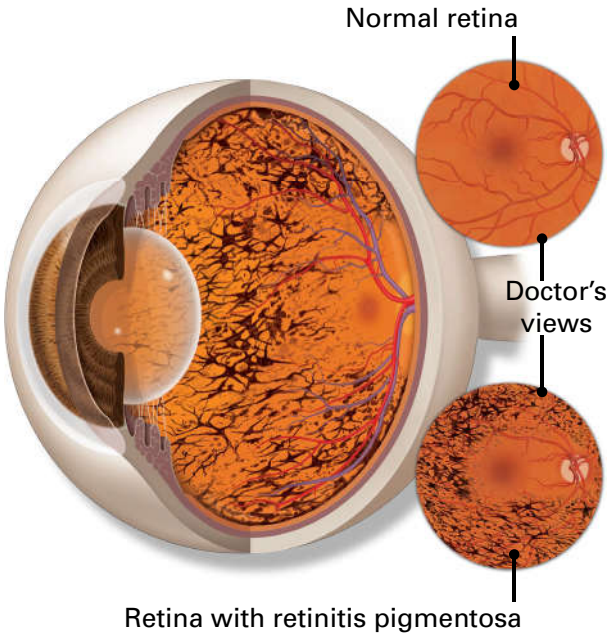
Who is at risk for Usher syndrome?

Usher syndrome is a recessive disorder. This means that a person has to inherit the change in the gene from both parents. A person who inherits the changed gene from one parent does not have the syndrome but is a carrier for it. When two carriers of the same Usher syndrome gene have a child together, there is a one-in-four chance that the child will have Usher syndrome.

What are symptoms of Usher syndrome?

The main visual symptom of Usher syndrome is loss of vision from RP.

In people with RP, the light-sensing cells in the **retina** slowly degenerate (die). These cells are called **rods** and **cones**. This causes a gradual loss of vision. With most forms of RP, rods start dying first. This causes a loss of side and night vision. When the cones begin dying, the result is loss of color perception and central (reading) vision. Generally, after many years, blindness results.



Eye Words to Know

Retina: Layer of nerve cells lining the back wall inside the eye. This layer senses light and sends signals to the brain so you can see.

Rods: One of the two types of "photoreceptor" (light sensing) cells in the retina. These cells are found in the side of the retina. They help us with our peripheral or side vision. They also help us see in low light or at night.

Cones: These are the other type of photoreceptor cells in the retina. They are found in the middle of the retina and give us our central vision. The also help us read and see colors.

Retinitis pigmentosa: An inherited disease that causes the death of the light-sensitive cells in the retina. This can lead to blindness over time.

How is Usher syndrome diagnosed?

Usher syndrome affects hearing, balance, and vision. So diagnosis of the disorder usually includes the evaluation of all three senses.

An ophthalmologist evaluation of the eyes may include:

- a visual field test to measure peripheral (side) vision
- an electroretinogram (ERG) to measure the response of the eye's light-sensitive cells
- and a retinal examination to observe the retina and other structures in the back of the eye.

How is Usher syndrome treated?

Currently there is no known cure for Usher syndrome or for retinitis pigmentosa. The best treatment involves early diagnosis so that educational programs can begin as soon as possible, depending on the severity of the vision loss and the age and ability of the child.

Treatment may include:

- instruction on reading with Braille
- and learning to use low-vision devices and techniques.

Some research has shown that vitamin A may slow the progression of certain forms of RP, but there is concern that high intake of vitamin A may lead to worsening of other eye conditions. Your ophthalmologist can advise you about the risks and benefits of Vitamin A and how much you can safely take. Taking too much vitamin A can be harmful, and evidence of vitamin A's effect on RP progression is not substantial.

Recently, inherited diseases similar to RP and Usher syndrome have been successfully treated with genetic therapy. This is when scientists create a gene in a lab. This corrected gene is then inserted into the body. Hopefully, the corrected gene replaces the bad gene that caused the disease. It is likely that such treatment may be available for Usher's soon as well.

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Summary

Usher syndrome is a condition that is inherited from your parents. It causes hearing and vision loss. Vision loss happens because of an associated disease called retinitis pigmentosa. This is when light-sensitive cells in the retina die, causing blindness over time. There is no cure for Usher syndrome but early detection is important. Treatment can include learning Braille and how to use low-vision devices and techniques. Studies have shown that taking vitamin A may slow progression of the disease in some cases.

If you have any questions about your eyes or your vision, speak with your ophthalmologist. He or she is committed to protecting your sight.

Get more information about Usher syndrome from EyeSmart—provided by the American Academy of Ophthalmology—at **aao.org/usher-link**.

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