ANSWERS TO YOUR QUESTIONS
ABOUT USHER'S SYNDROME
(RETINITIS PIGMENTOSA WITH HEARING LOSS)

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Hearing loss
Pattern of hearing loss in Type II, showing a greater loss of the higher frequencies:

What is Usher Syndrome?

Usher syndrome is an inherited disorder with hearing loss, progressive loss of vision due to retinitis pigmentosa and in some cases, balance problems. There are two main types of Usher syndrome.

Type I Profound hearing loss present at birth, balance problems, and retinitis pigmentosa.

Type II Moderate to severe hearing loss, no balance problems, and retinitis pigmentosa.

About 90% of all Usher syndrome patients have either Type I or Type II.

Balance problems

There are three senses we use to keep our balance; vision, proprioception (we feel where our bodies and limbs are in space) and vestibular function (we have a sense of motion and changes in speed or direction). One of these senses, the vestibular, does not work in Type I Usher syndrome. Although a baby with Type I may be slow to sit and walk, they learn to compensate by using the other two senses.

Retinitis pigmentosa (RP) is the eye disease that causes a loss of vision. Changes take place in the retina, or back of the eye. These changes cause the eye to become less able to adjust to low light, resulting in night blindness. As the RP progresses, the visual field narrows until only central vision remains. The narrowing of vision is referred to as “tunnel vision”. Most people will retain at least some central vision.

Hearing loss

Pattern of hearing loss in Type 1, showing a profound loss of hearing at all frequencies

WHAT IS USHER'S SYNDROME?

Usher's syndrome is an inherited disorder characterized by hearing loss, present at birth or shortly thereafter, and a progressive loss of vision. This loss of vision is caused by retinitis pigmentosa, a pigmentary degeneration of the retina. The
retina, a delicate layer of cells located in the back of the eye, acts like the film in a camera. It picks up the picture and transmits it to the brain, where actually occurs. In retinitis pigmentosa (RP), the retina begins to degenerate, causing vision to diminish. Since there are other medical conditions which also involve the retina and the ear and are not Usher's syndrome, expert medical diagnosis is important.

Of all people who are born with profound hearing impairments, an estimated three to six percent have Usher's syndrome.

WHAT ARE THE SYMPTOMS OF Usher's SYNDROME?

Most people with Usher's syndrome are born with a profound hearing loss, while others have milder hearing losses. One of the earliest visual symptoms of Usher is difficulty seeing at night or in dimly lit places (night blindness). Night blindness usually begins during the teenage years. Later there is a loss of side (peripheral) vision, resulting in “tunnel vision”. The visual symptoms of Usher syndrome generally worsen over the years. Some patients will also eventually lose their central vision, although RP usually does not cause complete blindness. When Usher affects several members of a family, the degree is of hearing loss is typically similar within that family.

WHAT CAUSES Ushers SYNDROME?

We know that certain cells in the retina—the rods and cones—die in Usher syndrome. Cells also die in the ear. However, we do not know what causes this cell death.

Current scientific evidence suggests there may be at least two genetic (inherited) types of Usher's syndrome: people born with a profound hearing loss and RP (Type 1), and people born with a mild to moderate hearing loss and usually a less severe forms of RP (Type 11). The biochemical or other defect that causes Usher is still unknown.

WHO IS DOING THE RESEARCH ON Ushers Syndrome?

Through the efforts of the RP Foundation Fighting Blindness, the number of highly qualified scientific investigators working full time on Usher's syndrome and related diseases continues to grow. Research efforts in this type of retinal-ear disease are difficult because there is no means of safely removing living tissue from the delicate eye or well-protected ear.

WHAT ABOUT RESEARCH CENTERS?
The RP Foundation Fighting Blindness supports extensive research at a number of major medical centers and hospitals around the world. The purpose is to find the causes, means of prevention and treatment of retinal degenerative diseases such as retinitis pigmentosa, Usher's syndrome and macular degeneration. Some of these centers also provide clinical evaluations. They may perform special tests requested by a patient's ophthalmologist or otologist (ear specialist). Even with these evaluations, it is still important for a patient with Usher's syndrome to remain in the care of his or her own physicians.

IS THERE A TREATMENT TO ARREST THE PROGRESSION OF USHER'S SYNDROME?

Currently there is no way to halt the degeneration of the retina or to alter the hearing loss, although many speculative attempts at "treatments" have been undertaken. No convincing scientific evidence has demonstrated any benefits from these so-called treatments for individuals with Usher syndrome. Yet our scientists always follow up even the most remote possibilities while continuing a vigorous search for a retinal therapy.

CAN USHER'S SYNDROME CAUSE BLINDNESS?

Blindness, to the layman, indicates a complete loss of sight. Although some Usher patients with advancing age become blind in this sense, most retain at least some residual vision, but are "legally blind". Each individual case differs.

WHAT IS MEANT BY THE TERM "LEGAL BLINDNESS"?

Legally blind individuals are those whose visual sharpness or acuity (with glasses, if needed) is 20/200 or worse in the better eye or whose visual field, regardless of acuity, is restricted to a 20 degree diameter or a 10 degree radius.

HOW QUICKLY WILL MY VISION DIMINISH?

Each person will experience a different rate of visual loss. However, most Usher patients have a very gradual progression of symptoms which can help in adjustment to their visual handicap.

CAN USHER'S SYNDROME BE EASILY DIAGNOSED?

In addition to a complete eye examination, other tests are sometimes required to determine the precise state of the retina, such as electroretinography and visual field. In advanced stages there is a characteristic change in the appearance of the retina. The hearing loss part of Usher's syndrome is easily diagnosed by audiometric tests, which may include air and bone condition and speech reception threshold tests.
E ARE VISUAL AND HEARING PROBLEMS THE ONLY CONDITIONS ASSOCIATED WITH USHIP'S SYNDROME?

Many people with Usher's syndrome also have a mild balance problem. Other medical symptoms may be present, although these are not related to Usher's syndrome.

WHAT ABOUT CATARACTS AND USHER'S SYNDROME?

It is not unusual for an individual with Usher's syndrome to develop a cataract, which is a haziness of the lens of the eye. Cataracts that significantly interfere with vision may need to be surgically removed. Whether or not surgery improves the vision often depends on the extent of retinal changes. Because surgery is not for everyone, it is necessary to discuss its advisability with an ophthalmologist.

DOES HAVING NIGHT BLINDNESS MEAN A PATIENT HAS USHER'S SYNDROME?

No. Night blindness can be a symptom of a number of retinal disorders. It requires expert evaluation.

CAN AN EYE TRANSPLANT BE PERFORMED?

The retina (the layer of cells in the eye affected by RP) is connected to the brain, and, within the limits of current scientific knowledge, cannot be transplanted. Although you may hear people talk about 'eye transplants', they are actually referring to corneal transplants, which have no relationship to RP.

WHAT ARE THE EFFECTS OF VITAMIN TREATMENTS?

As far as our scientific advisers can ascertain, vitamin treatments are not helpful in treating Usher's syndrome. There is no proven vitamin deficiency or shortage in patients with Usher's syndrome. Thousands of people affected by RP and Usher's have taken large quantities of vitamins, especially vitamin A, without obvious beneficial effect to the retina or to the ear. However, scientists are continuing to study this matter.

DOES LIGHT AFFECT THE VISUAL LOSS IN USHER'S SYNDROME?

Although there is no scientific evidence that normal levels of light increase visual loss, many people with Usher's syndrome are more comfortable avoiding bright lights. As a precaution, individuals with Usher's syndrome and other retinal degenerations are encouraged to protect their eyes from long-term exposure to bright sunlight until more is learned. A pair of good quality sunglasses is useful
for bright days outdoors. They may use their eyes in ordinary light without restriction.

THERE ARE TIMES WHEN MY VISION SEEMS BETTER AND TIMES WHEN IT SEEMS WORSE. DOES THIS HAPPEN TO OTHERS?

Yes, it does. A number of factors might account for good days or bad days for someone with Usher’s syndrome. Some people feel they see better on cloudy days; others feel they do not see as well on cloudy days. Fatigue and emotional stress may also temporarily affect vision. All people with Usher’s syndrome have days when, for no apparent reason, they seem to function better or worse than others.

DOES PREGNANCY AFFECT USHER’S SYNDROME?

Some women have reported that their vision changed during pregnancy, although it has not been systematically studied to date.

IS USHER’S SYNDROME INHERITED?

-Usher's syndrome is inherited as a recessive disorder, meaning that a double dose of the Usher's gene is required for a person to have the syndrome. In most cases, a single copy of the gene has been received from the mother and one copy from the father. When two carriers marry, each child has a one in four chance of having Usher's syndrome. An individual who has one does of the gene is referred to as carrier, but has no symptoms of the disorder. There is presently no way to detect carriers of the gene. If a person with Usher's marries-a person without the syndrome, it is highly unlikely that the children will have Usher's, but they will be carriers.