

# SYMPTOMS, COMPLICATIONS, AND PREVENTION OF THE GENETIC DISEASE XERODERMA PIGMENTOSUM

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## Abstract

This article is about what xeroderma pigmentosum is, its symptoms, treatment, and prevention. Early detection of this disease can protect you from skin cancer. Xeroderma pigmentosum (XP) is a rare genetic disease. It causes hypersensitivity to ultraviolet (UV) light, eye problems, and neurological symptoms. Xeroderma pigmentosum is caused by a gene change (mutation) inherited from your parents. There is no cure for XP, but treatment can reduce the risk of skin cancer and other complications.

**Keywords:** Xeroderma pigmentosum, ultraviolet (UV) rays, skin cancer, actinic keratosis, genetic testing.

## Introduction

Xeroderma pigmentosum (XP) is a rare genetic disorder that causes high sensitivity to ultraviolet (UB) rays. Symptoms usually affect parts of your body that are regularly exposed to the sun, such as your face, hands, and lips. People with XP usually start showing symptoms in early childhood. They can get bubble sunburns even after a few minutes in the sun. This condition mainly affects the eyes and areas of the skin that are exposed to the sun. Xeroderma pigmentosum is associated with an increased risk of UVR-induced cancers. People with this condition often experience premature aging. Some affected people also have problems with their nervous system. Xeroderma pigmentosum is not cancerous, but it can increase the risk of developing skin cancer.

## The Main Part

People with Xeroderma pigmentosum have easier sunburn and a higher risk of developing skin cancer. Symptoms of xeroderma pigmentosum usually appear in infancy or early childhood. About half of the affected children develop severe sunburn after spending a few minutes in the sun. Sunburn causes redness and blisters, which can last for weeks. However, some children with xeroderma pigmentosum may darken normally.

By age 2, almost all children with xeroderma pigmentosum have sepsis on the skin in areas exposed to the sun (e.g., face, hands, and lips); Sepia of this type are rarely found in young juveniles without the disease. In affected individuals, sunlight often causes dry skin (xeroderma) and skin discoloration (pigmentation). The combination of these characteristics gives the case its name.



Compared to those at moderate risk, people with xeroderma pigmentosum have:

The risk of developing non-melanoma skin cancers, such as basal cell carcinoma or squamous cell carcinoma, is 10,000 times higher.

The risk of developing melanoma is 2,000 times higher.

Some reports suggest that people with xeroderma pigmentosum are more likely to develop cancers such as:

- Astrocitoma.
- Ko'krak bezi saratoni.
- Glioblastoma.
- Buyrak saratoni.
- Leukemia.
- O'pka saratoni.
- Pancreatic cancer.
- Oshqozon saratoni.
- Moyak saratoni.
- Thyroid cancer.
- Bachadon saratoni.

Xeroderma pigmentosum is rare. Experts estimate that 1 in every 1 million people in the United States and Europe has a chromoderma pigmentosum.

It is more common in other parts of the world, with 1 in every 22,000 people in Japan. It is also more common in North Africa and the Middle East.

The symptoms of Xeroderma pigmentosum can vary, but generally affect your skin, eyes, and nervous system.

Xeroderma pigmentosum skin markings. Skin symptoms may include:

- Bubble sunburn.
- Dry, thin skin (xerosis).
- Sepcillary up to 2 years old (lentigos).
- The increase and decrease of skin pigment (poikiloderma).
- Thinning of the skin (atrophy).
- Red stripes on the skin as a result of increased dilation of blood vessels (telangiectasia).

People with Xeroderma pigmentosum have an increased risk of precancerous skin lesions called actinic keratosis.

**Symptoms of the disease.** Xeroderma pigmentosum eye marks. Eye symptoms usually affect people who have been infected with xeroderma pigmentosum before the age of 10. These symptoms may include:

- Dry eye.
- Degeneration of the eyelids (atrophy).
- Inflammation of the cornea (keratitis).
- Lack of transparency in the outer layer of the eyes (blurring of the cornea).
- Sensitivity to light (photophobia).



- Loss of eyelashes.

Over time, these symptoms can lead to vision loss or even blindness. People infected with Xeroderma pigmentosum are also at a higher risk of developing eye cancer.

Neurological signs of xeroderma pigmentosum. Xeroderma pigmentosum can also affect your nervous system. 1 in 4 people with Xeroderma pigmentosum develop neurological symptoms such as:

- Dysphagia (difficulty swallowing).
- Loss of reflexes.
- Poor muscle control (ataxia) and spasticity.
- Progressive loss of thinking (cognitive) skills.
- Progressive hearing loss as a result of damage to the inner ear nerve.
- Smaller size of the head (microcephaly).
- Vocal cord paralysis.

Experts believe that these symptoms develop due to the loss of nerve cells in your brain.

Xeroderma pigmentosum is a genetic disorder that occurs due to atypical gene changes (mutations).

You inherit these gene mutations from one or both of your parents.

Your health care provider may suspect you have xeroderma pigmentosum based on your symptoms.

They use a blood test to confirm the diagnosis of xeroderma pigmentosum. A blood test checks for gene mutations that cause xeroderma pigmentosum.

### Treatment

There is no cure for Xeroderma pigmentosum. But your health care provider may suggest you treatment options to reduce your symptoms, including:

Eye drops: Your doctor may prescribe lubricating eye drops to reduce inflammation in your cornea.

Hearing aids: If you have an increased risk of hearing loss, your doctor may order hearing aids. In some cases, they may recommend cochlear implants.

Surgery: If skin cancer develops, surgery may be needed to remove it. Or surgery can cure certain eye diseases, such as drooping eyelids (ptosis) or corneal problems. In severe cases, you may be given a cornea transplant.

Vitamin D supplements: Many people get adequate amounts of vitamin D through a combination of sunlight and diet. Since people with Xeroderma pigmentosum need to avoid the sun, they may need a vitamin D supplement.

### Checks

If you have xeroderma pigmentosum, there are a few things you should take to manage your overall health, including:

Sun Protection: Protecting your skin from ultraviolet rays is the most important step in managing HP. Wear sunscreen clothing, including long sleeves, pants, gloves, and hats. People should also wear sunglasses that block ultraviolet rays and wear broad-spectrum sunscreen with SPF 35+ daily. Experts also recommend using an ultraviolet light meter to find and avoid the areas around you with the highest levels of ultraviolet rays.



**Eye exams:** An ophthalmologist (ophthalmologist) will look for any changes that could affect your vision, such as drooping eyelids or tumors in your eyes. As directed by your doctor, you should see an ophthalmologist at least once a year or so.

**Skin exams:** You should see a skin specialist (dermatologist) every six to twelve months or more as directed by your doctor. A dermatologist looks for signs of cancerous or precancerous tumors. In between dermatology visits, you should examine your skin at least once a month to check for any changes in skin lesions. A dermatologist should immediately examine for any new lesions or tumors.

**Neurological Support:** People with Xeroderma pigmentosum should undergo regular brain, spine, and nerve tests with a specialist (neurologist) to check their reflexes and hearing. If your health care provider notices any changes in these functions, they may recommend treatments to slow or stop progression.

Life expectancy of xeroderma pigmentosum can vary. Many people with this disease have a shorter life expectancy due to developing skin cancer at a very young age. Taking measures to prevent damage from sunlight early in life can increase your chances of living longer.

There is no way to prevent xeroderma pigmentosum. If your doctor suspects that you have a gene mutation that causes xeroderma pigmentosum, you may choose to undergo genetic testing.

A genetic test tells you if you have a gene mutation. A genetic counselor will review the results with you and help you understand the associated risks, including the likelihood of having a child with the chromoderma pigmentosum. Without protection from the sun and other sources of UVR, most people with xeroderma pigmentosum will develop more than one skin cancer in their lifetime. These cancers are most commonly found in parts of the body that are exposed to the sun, including the face, lips, eyelids, eye surface, scalp, and tip of the tongue. Studies show that people with xeroderma pigmentosum may have an increased risk of certain internal cancers, such as brain tumors, thyroid cancer, and blood cancer. In addition, people who smoke cigarettes have a significantly increased risk of lung cancer.

The eyes of people with Xeroderma pigmentosum may be very sensitive to UVR (photophobia). If the eyes are not protected from UVR, they can become bleed, causing irritation, and the front of the eye (cornea) may become opaque. Some people have droopy eyelashes and thinned eyelids, causing them to tilt in or out unnaturally. In addition to the increased risk of cancer on the surface of the eye, xeroderma pigmentosum has been associated with non-cancerous tumors in the eye. Many of these eye anomalies can impair vision.

In addition to skin and eye problems, about 30% of people with xeroderma pigmentosum develop progressive neurological abnormalities. These abnormalities can include hearing loss, poor coordination, difficulty walking, movement problems, loss of intellectual function, difficulty swallowing and speaking, and convulsions. When these neurological problems occur, they get worse over time.

Early menopause may occur in people with xeroderma pigmentosum.

The researchers identified at least eight genetic forms of xeroderma pigmentosum: complement group A (XP-A) through complement group G (XP-G), and variant type (XP-V). The species is distinguished by genetic reasons. All types increase the risk of skin cancer, although some are more likely to be associated with neurological abnormalities than others.



If you or your child has or think you have xeroderma pigmentosum, you can ask your healthcare provider the following:

What can I do to reduce my risk of skin cancer?

What kind of cancer screenings should I have?

What are the early signs of skin cancer?

What does this diagnosis mean for the rest of my family?

Should I consider genetic testing?

What are the chances of having another child with xeroderma pigmentosum?

### Conclusion

Xeroderma pigmentosum (XP) is a rare genetic disorder. The most common symptom is hypersensitivity to ultraviolet rays. Many people also have symptoms that affect their vision and neurological development. If you have xeroderma pigmentosum, it's important to avoid ultraviolet rays as much as possible. Protecting your skin from the sun and getting regular skin cancer screenings will increase your chances of achieving a positive outlook.

People with xeroderma pigmentosum are 10,000 times more likely to develop non-melanoma skin cancer and 2,000 times more likely to develop melanoma skin cancer than people who don't have the disease. Types of skin cancers that can develop include basal cell carcinoma, squamous cell carcinoma, and melanoma. Most often, the first skin cancer occurs in people under the age of 10. Taking into account these data it is useful to start early detection of the disease and the necessary treatment in time, without delay. Take care of your health and that of your loved ones

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