CONDITION

Xeroderma pigmentosum

Xeroderma pigmentosum is an inherited condition that makes ultraviolet light damage the skin and eyes unusually easily.

WHAT IT IS
Inherited inability to repair ultraviolet light damage

BODY SYSTEM
Skin, eyes, and nervous system

MAIN SYMPTOMS
Severe sun sensitivity, early freckling, eye irritation, skin cancers

WHAT CAUSES IT
Inherited changes in DNA repair genes

HOW IT IS DIAGNOSED
Genetic testing, sometimes specialised DNA repair testing

MAIN TREATMENT
Ultraviolet protection, monitoring, and early treatment of damage

CAN IT BE CURED?
No, inherited DNA repair changes remain

IS IT AN EMERGENCY?
Sometimes, with sudden vision loss or severe eye pain

LONG-TERM OUTLOOK
Lifelong cancer risk reduced by careful ultraviolet protection

Overview

Xeroderma pigmentosum, often shortened to XP, is a rare inherited condition. It affects how the body repairs DNA damaged by ultraviolet, or UV, light. UV light comes mainly from sunlight, but some lamps and other artificial sources also produce it.

In most people, cells repair much of the DNA damage caused by UV exposure. In XP, this repair process does not work properly. Small amounts of UV can therefore cause lasting damage, especially in skin and eye cells.

Signs often begin in infancy or early childhood. A child may develop an unusually severe sunburn after limited exposure. Some children do not burn severely but develop many freckles on sun-exposed skin at a very young age. This early freckling can be an important clue.

Without strong UV protection, damaged cells can become cancerous. People with XP have a high risk of skin cancers and cancers on the surface of the eye. Some forms can also affect hearing, movement, swallowing, or thinking over time.

XP is lifelong and cannot currently be cured. However, careful protection from UV light, frequent checks, and early treatment of abnormal areas can prevent some damage and improve long-term health.

Symptoms

XP does not affect everyone in the same way. Symptoms depend partly on which gene is involved and how much UV exposure has occurred.

Skin signs may include:

Some people with XP do not have dramatic sunburn reactions. A lack of severe burning does not rule out the condition.

The eyelids and front surface of the eyes are also exposed to UV. Possible symptoms include redness, pain, light sensitivity, watering, dry eyes, and inflammation. Eyelashes may become sparse, and the eyelids may turn inward or outward. Clouding or damage to the cornea, the clear front layer of the eye, can affect vision.

A proportion of people develop neurological problems. These may progress slowly and are not caused by sunlight alone. Possible features include hearing loss, poor balance, muscle stiffness, reduced reflexes, problems with coordination, learning difficulties, or trouble speaking and swallowing. Many people with XP never develop significant neurological symptoms.

- Severe or blistering sunburn after brief exposure
- Freckles appearing unusually early on the face and other exposed areas
- Dry, thin, rough, or unevenly coloured skin
- Persistent scaly patches, sores, lumps, or changing moles
- Skin cancers developing much earlier than usual

Causes and risk factors

XP is caused by harmful changes in genes involved in repairing DNA. Most forms affect a process called nucleotide excision repair. This process normally finds and removes sections of

DNA damaged by UV light. XP variant, a related form, affects how cells copy DNA that has already been damaged.

The condition is usually inherited in an autosomal recessive pattern. This means a child inherits one altered copy of the same XP-related gene from each parent. The parents are usually healthy carriers because their other copy works normally.

When both parents carry an altered copy of the same gene, each pregnancy has a chance of resulting in a child with XP. The result of one pregnancy does not change the chance in a later pregnancy.

XP is not caused by anything a parent did during pregnancy. It is not contagious, and it cannot be spread through touch, blood, or shared objects.

UV exposure does not cause the inherited condition. It does, however, cause the cell damage responsible for many of its skin and eye complications. Exposure can come through windows and from certain artificial light sources as well as direct sunlight.

Diagnosis

A clinician may suspect XP from severe sun sensitivity, very early freckling, characteristic skin or eye changes, or a family history. The diagnosis should not be based on appearance alone because other inherited conditions can cause overlapping symptoms.

Genetic testing can confirm the diagnosis by finding harmful changes in both copies of an XP-related gene. It can also identify the specific genetic type. This information may help with planning follow-up and understanding whether neurological problems are more likely.

If genetic results are unclear, a specialist laboratory may test how skin cells respond to UV light or measure their ability to repair UV-related DNA damage. These tests are not available in every hospital.

Assessment often includes a full skin examination and a detailed eye examination. Hearing, development, movement, balance, and swallowing may also be checked when relevant.

Genetic counselling can help families understand test results and inheritance. Once the family's genetic changes are known, carrier testing and testing during a future pregnancy may be possible for relatives who want that information.

Treatment and management

There is currently no treatment that corrects the inherited DNA repair problem throughout the

body. Management focuses on preventing UV damage, finding complications early, and treating them promptly.

UV protection usually involves more than sunscreen. A specialist team can help identify where exposure occurs at home, school, work, during travel, and around artificial lighting. Portable UV meters can be useful because visible brightness does not reliably show how much UV is present.

Protective measures may include tightly woven clothing, gloves, a hat, and a UV-protective face shield or eyewear. UV-filtering films can be fitted to suitable windows. Light sources may need to be tested and replaced or covered when they emit UV. Sunscreen can protect uncovered skin, but it does not replace clothing, shade, or environmental controls.

Regular skin examinations are important. The interval is chosen according to age, previous findings, and cancer history. Suspicious or precancerous areas may be treated with surgery, freezing, creams, or other dermatology procedures. The choice depends on the type, size, depth, and location of the lesion.

Eye care may include UV-blocking glasses, lubricating treatments for dryness, and treatment for eyelid, corneal, or surface changes. Surgery is sometimes needed for a tumour or for damage affecting the eyelids or vision.

Neurological care is tailored to the symptoms. Hearing aids, physical therapy, communication support, nutrition advice, and swallowing assessment may help preserve function and independence.

Strict UV avoidance can lead to low vitamin D. Blood testing and supplements may be considered under clinical guidance. Psychological support, practical help at school or work, and contact with an XP support organisation can also be valuable.

Complications

The main long-term concern is cancer in areas exposed to UV light. XP can lead to basal cell carcinoma, squamous cell carcinoma, and melanoma. These are different types of skin cancer. Cancers may also develop on the eyelids, lips, or surface of the eye.

Repeated UV injury can cause extensive skin changes even when cancer is not present. These may include thinning, scarring, areas of increased or reduced colour, and fragile blood vessels.

Eye complications can include chronic inflammation, corneal damage, reduced vision, and tumours. Changes affecting the eyelids can make it harder to protect and lubricate the eye.

Neurological complications, when they occur, may worsen over years. They can include

progressive hearing loss, difficulty walking, reduced coordination, problems with speech or swallowing, and changes in intellectual function.

Frequent appointments and the need to organise life around UV protection can affect education, work, relationships, sleep, and emotional wellbeing. These effects deserve attention as part of XP care, not only after a crisis develops.

Prevention

XP itself cannot be prevented because it is inherited. Parents do not cause it through food, medicines, activities, or sun exposure during pregnancy.

Genetic counselling can explain reproductive options when the altered gene is known in a family. These options are personal, and testing does not obligate anyone to make a particular choice.

Many complications can be reduced, although not always completely prevented, by limiting UV exposure from the time XP is suspected. Consistent protection of the skin and eyes is central. Regular examinations also allow damaged areas to be treated before they become larger or harder to manage.

Families should not rely only on avoiding obvious sunshine. UV may pass through untreated windows or come from some artificial lights. A specialist assessment can help focus protection on sources that actually produce UV without unnecessarily restricting safe activities.

Living with it and what to expect

XP remains present throughout life. UV sensitivity does not fade with age, so protective routines and medical monitoring continue into adulthood.

Daily life often requires planning. Safer outdoor activity may be possible at night or when UV measurements show that exposure is very low. Homes, vehicles, schools, and workplaces may need UV-filtering measures. Protective clothing and equipment usually become part of the daily routine.

Skin and eye damage is related in part to accumulated UV exposure. Strong protection started early can reduce further damage, but it cannot reverse genetic changes or all previous injury. New suspicious areas may still develop and need treatment.

Some people have repeated skin procedures yet maintain active education, work, family, and social lives. Others face major visual, neurological, or cancer-related complications. The

specific XP gene, past UV exposure, access to protection, and response to complications all influence the course.

Neurological symptoms do not occur in everyone. When they do occur, they may progress gradually rather than appearing all at once. Regular hearing and neurological assessments can identify changes and allow support to begin earlier.

Long-term care is usually shared between dermatology, eye care, genetics, and primary care, with neurology, hearing, rehabilitation, dental, or mental health services added when needed. Routine follow-up remains important even when you feel well because early cancers may be painless.

When to get medical help

Arrange a prompt medical or dermatology assessment for a new or changing mole, a lump, a scaly patch that persists, or a sore that does not heal. Bleeding, crusting, rapid growth, changing colour, or an irregular border also need timely review. These signs do not always mean cancer, but they should not wait until the next distant routine appointment.

Report increasing eye redness, discomfort, light sensitivity, eyelid changes, or a gradual change in vision promptly to an eye specialist.

Seek urgent same-day care for severe eye pain, a sudden loss or major change in vision, or a chemical or UV injury to the eyes.

Call emergency services for a seizure lasting several minutes, sudden severe weakness, loss of consciousness, choking with inability to breathe, or another immediately life-threatening symptom. These are emergency signs regardless of whether someone has XP.

Routine appointments are appropriate for planned skin and eye checks, hearing monitoring, vitamin D review, equipment questions, and gradual changes in mobility or learning.

Common questions

Is xeroderma pigmentosum contagious?

No. XP is caused by inherited gene changes. It cannot spread between people through touch, close contact, sex, blood, or shared belongings.

Can someone with XP ever go outside?

Outdoor activity requires careful planning because even brief UV exposure can cause damage. Some people choose nighttime activities or use UV measurements, protective clothing, face

protection, and other controls. A specialist team can help make a practical plan rather than relying on visible sunlight alone.

Is sunscreen enough for XP?

No. Sunscreen can protect small areas of exposed skin, but it may be missed, rubbed off, or applied unevenly. Clothing, gloves, head and face protection, UV-blocking eyewear, shade, window films, and control of artificial UV sources are also important.

Does everyone with XP develop cancer?

Cancer risk is very high, but an individual outcome cannot be predicted from the diagnosis alone. The genetic type, lifetime UV exposure, quality of protection, and frequency of monitoring all matter. Early identification and treatment can make cancers easier to manage.

Does XP affect life expectancy?

It can shorten life, particularly when aggressive cancers or progressive neurological complications occur. Strong UV protection, close surveillance, and early treatment have improved outcomes, but they do not remove all risk.

Can a person with XP have children?

Yes. XP does not automatically prevent pregnancy or parenthood. The chance that a child will inherit XP depends on the other biological parent's carrier status and the gene involved. Genetic counselling and carrier testing can clarify this before or during pregnancy.

Sources

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