

What else can cause harm besides ultraviolet light ?

Laboratory tests indicate that sunlight is the major DNA damaging agent to the cells of XP patients. However, tobacco smoke and some drugs (such as psoralens, used with ultraviolet light for treating psoriasis) can cause similar DNA damage. People with XP should avoid exposure to tobacco smoke and should not use tobacco products because they are probably at greater risk for developing lung cancer.

Psychosocial aspects of XP

Persons with XP and their families face many challenges in daily living. This disease has many long-term physical, emotional, social, and economic consequences. Skin changes, including cancers at an early age and other physical problems, may affect school experiences, employment opportunities, recreational activities, and social relationships. Coping with chronic illness and disability is very difficult. Some people have problems with health insurance or finances while others may feel anxious or depressed. Persons with XP need a great deal of support from family, friends, and their communities to provide encouragement, build confidence, and give hope.

A well protected person with XP



Xeroderma Pigmentosum Society is a non-profit organization that has recently formed a society in South Africa. We have branches in the UK, Germany and the USA. Our aim is to make more people aware of the dangers of the sun in South Africa, and what symptoms to look for, as well as to help and build a support group for those with the disease, and similar conditions. The XP society will be supporting families of patients and try to raise the much needed funds for distribution of flyers and research to make people more aware of this disease. We also aim to have our own night camps in South Africa so that the patients can experience the joys of holidays as well as to be able to interact with other patients with the same disease. These patients cannot live normal lives as they are not allowed in the sun at all and can only go out at night. Children with Xeroderma Pigmentosum (XP) can only play outdoors safely after nightfall. They have been called midnight children, and children of the dark.

- Other disorders associated with defective DNA repair
 - Ataxia-Telangiectasia
 - Bloom Syndrome
 - Cockayne Syndrome
 - Fanconi Anemia
 - Trichothiodystrophy (TTD)
 - Other disorders characterized by light sensitivity
 - Drug-induced photosensitivity
 - Erythropoietic Protoporphyrin (EPP)
 - Lupus (30% of cases)
 - Polymorphous Light Eruption (PLE)
 - Porphyria (general)

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XERODERMA PIGMENTOSUM

XP SUPPORT GROUP



**Supporting those with
Xeroderma Pigmentosum
and other UV and light
sensitive conditions.**

What is Xeroderma Pigmentosum ?

Xeroderma Pigmentosum (XP) is a rare genetic disorder that causes extreme sensitivity to the sun's ultraviolet rays. Unless patients with XP are protected from the sunlight, their skin and eyes may be severely damaged. This damage may lead to cancers of the skin and eye. **XP has been identified in every genetic group all over the world.** There are about 1000 cases world wide. Many people with XP will die at a early age from skin cancer if they are untreated and unprotected from sunlight. The two most common causes of death for XP victims are metastatic malignant melanoma and squamous cell carcinoma. However if a person is diagnosed early, has no severe neurological problems, is protected from ultraviolet light, and followed carefully for early cancer detection, a normal lifespan is possible. A reduced life span is to be expected, but there are great differences among patients with XP.

Xeroderma pigmentosum, or XP, is an autosomal recessive genetic disorder of DNA repair in which the ability to repair damage caused by ultraviolet (UV) light is deficient. This disorder leads to multiple basal cell carcinomas and other skin malignancies at a young age. In severe cases, it is necessary to avoid sunlight completely. .

The most common defect in xeroderma pigmentosum is an autosomal recessive genetic defect whereby nucleotide excision repair (NER) enzymes are mutated, leading to a reduction in or elimination of NER. Unrepaired damage can lead to mutations, altering the information of the DNA in individual cells. If mutations affect important genes, such as tumour suppressor genes or proto oncogenes, then this disorder may lead to cancer.



What happens next?

People with XP will experience extremely severe sunburn after very short exposure to the sun. This sunburn can last up to several weeks.

In children, this serious sunburn usually occurs during their first exposure to the sun, and can be the main clue to the diagnosis of XP. However, some people with XP will experience different degrees of sunburn, which can result in unusual skin changes appearing over time. This in turn can delay the diagnosis of XP.

In XP, the skin changes caused by the sun damage often begin in infancy and almost always before the age of 20.

Why does it happen ?

Two factors combine to cause XP. Firstly a person inherits traits from each parent which when combined, lead to an unusual sensitivity to the damaging effects of ultraviolet light. Secondly, exposure to the sun, which contains ultraviolet light, leads to changes in the skin and eyes.

Can XP be treated ?

There is no cure for XP, However, there is much that can be done to prevent and treat some of the problems caused by this devastating disease.

- protection from ultraviolet light
- frequent skin and eye examinations
- prompt removal of cancerous tissue
- neurological examination
- psychosocial care.

Prognosis:

Fewer than 40% of individuals with the disease survive beyond age 20 years. Individuals with milder disease may survive beyond middle age.

What are the signs and symptoms of xeroderma pigmentosum ?

Some of the most common symptoms are:

An unusually severe sunburn after a short sun exposure. The sunburn may last for several weeks. The sunburn usually occurs during a child's first sun exposure.

- **development of many freckles at an early age.**
- **Irregular dark spots.**
- **Thin skin.**
- **Excessive dryness.**
- **Rough-surfaced growths (solar keratoses), and skin cancers.**
- **Eyes that are painfully sensitive to the sun and may easily become irritated, bloodshot, and clouded.**
- **Blistering or freckling on minimum sun exposure. Premature aging of skin, lips, eyes, mouth and tongue**

How is Xeroderma Pigmentosum diagnosed ?

Usually Xeroderma Pigmentosum is detected between the ages of 1 and 2 years old.

XP can usually be diagnosed in the laboratory by measuring the DNA repair defect. This test is performed on skin or blood obtained from the patient. Soon, only a few cells may be needed to make this diagnosis.