DermNet NZ

Authoritative facts about the skin from the New Zealand Dermatological Society Incorporated.

Xeroderma pigmentosum

What is xeroderma pigmentosum?

Xeroderma pigmentosum (XP) is a very rare skin disorder where a person is highly sensitive to sunlight (photosensitive), has premature skin aging and is prone to developing skin cancers. This is caused by a cellular hypersensitivity to ultraviolet (UV) light as a result of a defect in the DNA repair system.

What causes xeroderma pigmentosum?

Xeroderma pigmentosum is an autosomally recessive inherited disease, which means that you have inherited two recessive xeroderma pigmentosum genes (one from each parent). If your parents are only carriers of the xeroderma pigmentosum trait (each have one xeroderma pigmentosum gene and one normal gene), they will not show signs or symptoms of the disease. By having the two xeroderma pigmentosum genes this causes you to have an extreme sensitivity to UV light and as a result experience a range of signs and symptoms of xeroderma pigmentosum. At least seven different gene abnormalities or complementation groups have been described in different families (XPA to XPG) resulting in varying disease severity.

Essentially, the signs and symptoms of xeroderma pigmentosum are a result of an impaired DNA repair system. In people who do not have xeroderma pigmentosum, cell damage from UV light is mended by the DNA repair system. However, people with xeroderma pigmentosum have a defect in this repair system and any damaged cells from UV light remain unrepaired, leading to cancerous cells or cell death.

Who is at risk of xeroderma pigmentosum?

Couples who are each carriers of the xeroderma pigmentosum trait are at greater risk of producing a child with xeroderma pigmentosum. Parents already with a child with xeroderma pigmentosum have a 1 in 4 chance of having another child with xeroderma pigmentosum.

Xeroderma pigmentosum occurs worldwide and affects people of all races. Males and females alike can have the condition.

What are the signs and symptoms of xeroderma pigmentosum?

The disease usually progresses through 3 stages. The first stage occurs around 6 months after birth (skin appears normal at birth) with the following signs:

- Areas exposed to the sun such as the face show a reddening of the skin with scaling and freckling. Irregular dark spots may also begin to appear.
- These skin changes progress tthe neck and lower legs. In severe cases the trunk may be involved.
- Over the winter months these changes may diminish.

Continued sun exposure will lead to the second stage, which is characterised by poikiloderma. This is where there are irregular patches of lightened or darkened skin, a spider web-like collection of blood spots and vessels are seen through the skin, and there is thinning of the skin.

The third stage is the development of solar keratoses and skin cancers. These may occur as early as age 4-5 years and are more prevalent in sun-exposed areas such as the face. The common skin cancers, basal cell

carcinoma, squamous cell carcinoma, and melanoma, occur significantly more often in people with xeroderma pigmentosum. Other complications, including eye and neurological problems are also apparent in patients with xeroderma pigmentosum.

Eye problems occur in nearly 80% of xeroderma pigmentosum patients.

- Eyes become painfully sensitive to the sun (photophobia).
- Eyes easily irritated, bloodshot and clouded. Conjunctivitis may occur.
- Non-cancerous and cancerous growths on the eyes may occur.

Neurological problems occur in about 20% of xeroderma pigmentosum patients.

- These can be mild or severe and include spasticity, poor coordination, developmental delay, deafness, and short stature.
- May develop in late childhood or adolescence. Once they do occur they tend to worsen over time.

How is xeroderma pigmentosum diagnosed?

Usually the disease is detected in early infancy, around 1-2 years.

A child presenting with severe sunburn after their first exposure to sun may be a clue to the diagnosis of xeroderma pigmentosum. Xeroderma pigmentosum can usually be conclusively diagnosed by measuring the DNA repair factor from skin or blood samples.

What is the treatment for xeroderma pigmentosum?

There is no cure for xeroderma pigmentosum. The main goal of treatment is to protect oneself from UV exposure and thus prevent the damaging effects it can have on the skin. Xeroderma pigmentosum patients should follow these general precautionary measures:

- Sun avoidance methods
- - Wear protective clothing (long sleeves and pants, shirts with collars, tightly woven fabrics that don't let light through), hats (wide-brimmed) and eyewear (specifically made to protect from UV rays)
  - Use sunscreens with SPF of 30 or greater: apply to all exposed areas
  - Outdoor activities should be avoided and kept to a minimum if at all necessary
  - Restrict outdoor activities to night time
- Undergo frequent skin examinations by someone who has been taught to recognise signs of skin cancer. Report to your doctor any suspicious spots or growths immediately.
- Examination by a dermatologist at least every 3 to 6 months. Any suspicious growths can be biopsied. Skin cancers are usually excised.
- Frequent eye examinations by an ophthalmologist.
- Yearly testing (through to age 20) for potential neurological problems.

Solar keratoses may be treated by cryotherapy or 5-fluorouracil cream. Some xeroderma pigmentosum patients who have had many skin cancers may be prescribed isotretinoin. This is a vitamin A derivative that may prevent formation of new cancers.

What is the prognosis?

Many patients with xeroderma pigmentosum die at an early age from skin cancers. However, if a person is diagnosed early, does not have severe neurological symptoms or has a mild variant, and takes all the precautionary measures to avoid exposure to UV light, they may survive beyond middle age.

Patients with xeroderma pigmentosum and their families will face many challenges in daily living. Constant educating and reminding of the need to protect oneself from sunlight is paramount to the management of

xeroderma pigmentosum.

Related information

References:

On DermNet NZ:

Other websites:

- Xeroderma Pigmentosum – emedicine dermatology, the online textbook
- Xeroderma Pigmentosum Society, Inc.
- XP Support Group (UK)
- DermIS
- Skin Cancer Scientist Sees Ray of Hope A Conversation with James Cleaver By Jeff Miller, University of California, San Francisco

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If you have any concerns with your skin or its treatment, see a dermatologist for advice.