



THE AUSTRALASIAN COLLEGE
OF DERMATOLOGISTS

Xeroderma pigmentosum

What is it?

Xeroderma pigmentosa (XP) is a rare genetic condition which is characterised by extreme sun sensitivity. It manifests as skin pigmentation changes, sunburns and an increased risk of skin cancers. Approximately one quarter of affected individuals will have associated neurological manifestations.

What causes it?

It is caused by a mutation on one of eight genes. It is autosomal recessive, meaning two copies of the defective gene are needed to develop the disease. If both copies are present, the disease occurs 100% of the time.

Who gets it?

XP occurs in all ethnic and sex groups.

How is it diagnosed?

In some individuals, XP may present as a reddening in the first few weeks of life due to an exaggerated sunburn response. In other individuals, they will often develop freckle-like pigmentation on sun-exposed areas such as the forehead and cheeks in early childhood, usually before the age of 2 years.

If XP is suspected, you or your family member may have a skin biopsy performed and referred to a genetics clinic and offered a blood test. This blood test looks to see if the faulty gene is present and can help confirm the disease.

What are the other associated signs and symptoms of the disease?

Other associated skin findings are hypo- and hyperpigmentation, warty growths and accelerated photoaging. The eyes may also be affected.

In 25% of individuals, neurological signs will be present such as reduced deep tendon reflexes, progressive hearing loss, and difficulty remembering things, balance impairment and acquired decrease in size of the brain (microcephaly).

Skin cancers

Individuals with XP are at a significantly increased risk of non-melanoma and melanoma skin cancer.

How is it managed?

Strict lifetime sunlight avoidance is key to reducing the risk of associated sequelae.

Individuals with XP need to take precautions to minimise any sun exposure, including:

- Wearing SPF 50+ and sun-protective clothing such as long sleeves, wide-brimmed hat and wraparound sunglasses.
- Ensuring access to SPF 50+ at all times.
- Avoiding outdoor occupations or recreational activities.
- Avoiding sun exposure through car or building windows, either through drawing curtains and/or applying UV protective tinting to windows.
- Using lightbulbs which emit lower levels of UV.
- Avoiding going outdoors during the middle of the day, and planning travel and activities in periods of low sunlight.

Awareness of your own skin is very important if you suffer from XP, to detect skin cancers early.

It is also worse in people who smoke, and therefore smoking cessation is crucial.

Individuals with XP must also take a vitamin D supplement, as they will be avoiding the sun.

Further information and links to support networks can be found on the Xeroderma Pigmentosum Society, Inc website: <https://www.xps.org/>

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