



# XERODERMA PIGMENTOSUM

## WHAT ARE THE AIMS OF THIS LEAFLET?

This leaflet has been written to help you understand more about xeroderma pigmentosum (XP). It explains what it is, what causes it, what can be done about it and where you can find more information.

## WHAT IS XERODERMA PIGMENTOSUM?

XP is a very rare condition with about 120 affected patients living in the UK. XP can affect both males and females, and all ethnic groups.

With XP the skin cells are unable to repair themselves correctly after exposure to ultraviolet radiation (UVR), which is present in all forms of daylight and some artificial light. Without this repair mechanism working correctly a person is much more likely to develop skin cancers. XP can be divided into different types known as complementation groups XP-A, B, C, D, E, F, G and Variant. There are slight clinical differences between the groups; with around 30% developing neurological problems such as hearing loss, balance problems or learning difficulties.

## WHAT CAUSES XERODERMA PIGMENTOSUM?

XP is genetic; a person is born with it. Even though it might not be diagnosed until a person is older, when symptoms develop, they would have had it since birth, and they will always have it.

## IS XERODERMA PIGMENTOSUM HEREDITARY?

Yes. XP is an autosomal recessive inherited condition. This means for a person to be affected by it, they need to have inherited 2

copies of the faulty gene. One gene from their mother and one from their father. If a person has only one copy of the faulty gene, they do not have XP, but are known as a carrier. Genetic counselling and advice are available for individuals affected by XP and for parents of affected children (<https://www.nhs.uk/conditions/genetic-and-genomic-testing/>).

## WHAT ARE THE SYMPTOMS OF XERODERMA PIGMENTOSUM?

A person can have some or all of the following symptoms:

- Hypersensitivity to daylight causing sunburn even on an overcast and cloudy day. This sunburn can often take up to 10 days or longer to get better which is a lot longer than would be expected in someone not affected by XP.
- Sore or red eyes especially on a bright day (photophobia)
- Freckling on skin that is frequently exposed to daylight. This freckling, known as lentiginos, is often reported from an early age (under the age of 2) and is unrelated to skin colour.
- Dry skin and early onset of skin aging.
- Skin cancers and/or eye cancers from an early age (as young as 4 years old). Multiple cases and different types of skin cancer in the same person.
- Neurological problems such as hearing loss, poor balance and



learning difficulties, or loss of thinking (cognitive) skills that a person used to have.

## HOW IS XERODERMA PIGMENTOSUM DIAGNOSED?

If XP is suspected, a small skin biopsy can be taken from a non-sun exposed area of skin, often the buttock, by a doctor or nurse. The biopsy area is numbed by injecting a local anaesthetic into the skin and then removing a small piece of skin. This is then sent to the diagnostic laboratory to be tested.

The XP complementation group (what kind of XP you have) can be diagnosed with a blood test which is sent to a genetic laboratory.

These results can take several months as they are very complicated laboratory tests.

## CAN XERODERMA PIGMENTOSUM BE CURED?

At present, there is no known cure for XP. However, a person can do a lot to help themselves, especially in reducing the chances of skin cancers developing. This is explained in the 'Self-care (what can I do?)' section below.

## HOW CAN XERODERMA PIGMENTOSUM BE TREATED?

It is recommended someone diagnosed with XP should have:

- Frequent, regular skin checks by a dermatologist to look for early signs of any skin cancers. This can reduce the risk of them spreading and causing further harm. If there are any concerning skin areas or lumps and bumps, these will be photographed, biopsied and surgically removed. In some cases, a cream to be applied to the skin may be prescribed which can help treat some early signs of some skin cancers.
- Eye checks, at least once a year, by a consultant ophthalmologist who looks for eye damage associated with UVR. They can advise on how to minimise this damage in the future. They advise on any treatment necessary to already UVR damaged eyes which may involve eye drops or, in some cases, eye surgery.
- The minority of people affected by XP who show signs of balance problems, learning difficulties or a loss of skills they previously had, can benefit from seeing a neurologist (a doctor skilled in problems of the nerves and brain). After a physical examination and taking a detailed history, they may advise on further tests such as an MRI (brain scan), hearing test and/or nerve conduction study (looking at how well nerves and muscles work). These will help them understand the extent of the problem so they can give advice, such as organising help at school to maximise a child's learning potential, hearing aids if required or better footwear to help walking.
- It can sometimes be difficult to understand and adapt to life with XP. It can help to have access to a psychologist where patients can talk about their feelings in a confidential environment. A psychologist may be able to give suggestions as to what may be helpful.
- Protection from UV reduces the Vitamin D levels in the blood as sunlight is needed to help the body produce this vitamin. Vitamin D levels can be checked with a simple blood test either at a GP surgery or hospital and supplements prescribed if required. Low Vitamin D could lead to bone pain and long-term bone problems.



Once a person has been diagnosed with XP, it is suggested they be referred to the National XP service based at St Thomas' Hospital in London. This service is funded by NHS England. They will work in partnership with the patient's referring doctor, giving advice as necessary. If the person is able and willing to travel to the London clinic, they will see a lot of different speciality doctors in one day: e.g. dermatology, neurology, ophthalmology, psychology and genetics. The XP specialist nurse is available to give advice on all matters of protection from UVR. The nurse can visit families in their home, school or workplace if requested.

### SELF-CARE (WHAT CAN I DO?)

Protection from UVR is very important to help reduce any further damage to the skin.

UVR is:

- Part of the electromagnetic spectrum made up of ultraviolet A (UVA), ultraviolet B (UVB) and ultraviolet C (UVC). UVC does not reach the earth's surface.
- Invisible, but is always present at some level from the moment the sun comes up to when it goes down at night.
- Present even when it is cloudy or rainy.
- Higher at altitude, or closer to the equator.
- Reflected off surfaces.
- Known to be responsible for skin cancers, skin aging and skin sunburn.

There is a lot you can do to help yourself:

- **Behaviour.** Think about when it is best to go outside. UVR levels are highest when the sun is directly overhead (11 am to 3 pm). UVR is not present outside at night and therefore no extra precautions would be needed. Understandably

people have to be able to go out during the day but avoiding the highest UVR is advised. Plan in advance how to get to your destination to best avoid UVR.

- **Clothing** should cover the whole body, including long sleeves and long trousers, to protect the arms and legs, and gloves to protect the hands. Tight weave materials let through less UVR, and thick clothing or multiple layers can block UVR completely. Darker coloured materials absorb more UVR. Some manufacturers produce clothing to protect from UVR and this is marked with a 50+ UPF label (a measure of how much UVR a material lets through), these can help, but can be expensive. Some people choose to wear two thinner layers of clothing instead.
- **Hat** with a wide brim that covers the forehead, ears and neck.
- **Glasses** to protect the eyes, with a high UVR protection rating to protect against UVA and UVB wavelengths. These can either have a clear lens or a darkened lens. The style of glasses that wrap around the face or have side covers to limit UVR reaching the eye are suggested.
- **UVR Protective face visor** is very good for full protection of the face. Pictures of this can be found on the Action for XP (<https://www.actionforxp.org/>) below, along with details on how to get one or make one.
- **Sunscreen** should be broad spectrum 50+ sun protective factor (SPF) which protects against UVB as well as UVA. Sunscreens come in a variety of forms, sprays, creams, lotions, gels, roll-on along with different smells and colours/tints. It is important to choose one that a



person will be happy to use all the time. Some are available on prescription for people with XP. Sunscreen should be applied 20 minutes before going out, and should be reapplied 2-3 hourly. Reapply the sunscreen if the cream is removed, e.g. after hand washing. Care should be taken while applying it to ensure no areas of skin are missed and the correct amount is used. Sunscreen is tested for its SPF rating by testing with 2 mg/cm<sup>2</sup>. So if you are using a lotion, this equals six heaped teaspoons of lotion or 35 ml to cover an average adult body. Different formulations vary so check the manufacturer's recommendations. Lips should not be forgotten - use a protection lip-block, with sun protection factor (SPF) 50+.

- **Skin checks.** If you notice anything different on your skin, or a freckle or mole looks like it is changing in some way, make an appointment to see your doctor or local dermatologist to get it checked. The sooner something is noticed and treated the better. Your dermatologist/GP or specialist nurse can teach you what to look for and how to do a skin check.
- **Eye concerns.** If you notice any changes with your eyes or notice any lumps, inform your medical team as soon as possible to get them checked.
- **Environment.** UVR, in particular UVA, can travel through some window glass, although not all. It all depends on how the glass was manufactured which can be difficult to know if it is already in a window. UVR protective film is a transparent film that can be applied to windows to reduce the amount of UVA coming through. It is available from companies that

supply window films and can be up to 99.8% effective at blocking UVA. Some patients may be eligible for a "disabled facilities grant" to help fund this (<https://www.gov.uk/disabled-facilities-grants>) or Action for XP (XP charity) may be able to help. Car windows may also need film protection. Remember an open window will let in UVR. Drawing curtains and blinds is an effective way of reducing UVR.

- **Light bulbs.** In particular, halogen, fluorescent and compact fluorescent bulbs are known to produce some UVA. This risk is relatively low but can be a problem if a bulb is close to a person affected by XP. Bulbs can be covered with protective UVR sleeves or changed to those which produce less UVR such as LEDs.
- **UV meter.** As UVR is invisible it can be hard to know when you are at risk. UV meters are provided by the Action for XP charity to those patients seen in the UK National XP service at Guy and St. Thomas Hospital, London. UV meters can help with a more accurate assessment of the amount of UVR in the environment.

## WHERE CAN I GET MORE INFORMATION ABOUT XERODERMA PIGMENTOSUM?

*Weblinks to support groups:*

XP charity support group

<https://www.actionforxp.org/>

*Weblinks to additional resources:*

The UK National XP service which also provides links to further patient information leaflets

<https://www.guysandstthomas.nhs.uk/xp>



This leaflet aims to provide accurate information about the subject and is a consensus of the views held by representatives of the British Association of Dermatologists: individual patient circumstances may differ, which might alter both the advice and course of therapy given to you by your doctor.

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*This leaflet has been assessed for readability by the British Association of Dermatologists' Patient Information Lay Review Panel*

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