



XERODERMA PIGMENTOSUM (XP)

What are the aims of this leaflet?

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

What is XP?

XP is a very rare condition with about 100 patients living with it in the UK. The genetic material in the skin is unable to repair itself correctly after exposure to ultraviolet radiation (UVR) which is present in all forms of daylight and some artificial light (see section 'light bulbs'). Without this repair mechanism working correctly a person is much more likely to develop skin cancers of all types. XP can be divided into different types known as complementation groups A to G and Variant. There are slight clinical differences between the groups; some are known to develop neurological problems such as hearing loss, balance problems or learning (cognitive) difficulties.

What causes XP?

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

Is XP hereditary?

Yes it is. It is an autosomal recessive condition. This means a person with XP would need to inherit one copy of the XP gene from each parent. The parents are known as carriers as they only carry one copy of the XP gene and therefore do not themselves have XP. Parents often have no idea they carry the XP gene, however once it is known that they do, they are advised that there is a 1 in 4 risk in every further pregnancy of having a child with XP. XP

can affect both sexes and all racial groups. Genetic counselling and advice is available to people with XP and parents of children with XP.

What are the symptoms of XP?

A person can have some or all of the following:

- Easy sunburning even on an overcast and cloudy day. This sunburn can often take up to 10 days or longer to get better.
- Sore or red eyes especially on a bright day (photophobia), watery eyes.
- Freckling (lentiginos) on sun exposed sites. This freckling is often reported from an early age, (under the age of 2) and is unrelated to skin colour.
- Early onset of skin aging, dry skin
- Risk of skin cancers or eye cancers from early childhood or multiple numbers and types of skin cancer in a person.
- History of neurological problems such as hearing loss, poor balance and learning difficulties.

How is XP diagnosed?

If XP is suspected, a small skin biopsy can be taken from a non-sun exposed site, often the buttock. After a person has given their consent, the biopsy site would be made numb by injecting some local anaesthetic into the skin and removing a small piece of skin. This would be sent to the diagnostic laboratory to be tested. Results can take up to 4 months as it is a very complicated laboratory test.

The XP group can then be diagnosed with a blood test which would be sent to a genetic laboratory.

Can XP 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.

How can XP be treated?

It is recommended someone diagnosed with XP should have:

- Skin checks by a dermatologist who looks 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 lesions these can be biopsied and surgically removed. In some cases topical treatments (creams to skin) may be prescribed which can help treat some early signs of skin cancers.

- Eye checks by an ophthalmologist who looks for eye damage associated with UVR. Sometimes eye lubricants are prescribed to treat dryness. Various types of glasses may be suggested to reduce UVR damage and in some cases, surgery may be recommended if there are any suspicious areas, which could indicate eye cancer, although this is rare.
- The minority of people with XP who show signs of balance problems or 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). They would examine the patient and take a detailed history and may also suggest a MRI (brain scan), hearing test and/or nerve conduction study. These will help them understand the extent of the problem so they can give advice. This advice may include things such as organising help at school to maximise a child's learning potential, hearing aids if required, better footwear to help walking.
- It can sometimes be hard to understand and adapt to live with XP; it can help to have access to a psychologist who patients can talk to in confidence about their feelings. They may be able to give suggestions as to what may be helpful to that person.
- If a patient is protecting themselves from UVR then Vitamin D levels in their blood can be low, due to lack of sunlight which is needed to help the body produce this vitamin. Vitamin D levels can be checked with a simple blood test 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 that they are 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 lots 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. They can visit families in their home, school or workplace if requested.

Self care (What can I do?)

There is a lot you can do.

Protection from UVR is very important to help reduce any further damage to your skin. UVR is present at the same time as daylight, from the moment the sun comes over the horizon to when it falls at night. UVR is part of the electromagnetic spectrum but unlike sunlight, it is invisible. Although there is less UVR present when it is cloudy or rainy, it is still there and therefore a person would still need to protect themselves in the following ways all year round, whatever the weather.

Think about when to go outside. UVR levels are highest when the sun is directly overhead. 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 levels would be advised. In addition there are a number of other protective steps recommended.

- **Clothing** to 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. Some manufacturers produce clothing to protect from UVR and this is marked with a 50+ UDF (a measure of how much UVR a material lets through) label, these can help, but can be expensive. Some people opt to wear two thinner layers of clothing instead.
- **Hat** with a wide brim that covers the forehead, ears and neck for example a legionnaire's style hat.
- **Sunglasses** to protect the eyes with a high UVR protection rating to protect against UVA and UVB wavelengths. If a person prefers clear non-tinted glasses these can now be bought with UVA and UVB coating. 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 XP Support Group (XPSG) and Teddington Trust websites 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 in application to ensure no areas are missed and the correct amount is used. Sunscreen is tested for its SPF rating by testing with 2 mg/cm² so if using a lotion, this equates to six heaped teaspoons of lotion or 35 ml to cover an adult body. This can vary if using different formulations so check the manufacturer's recommendations. Lips should not be forgotten, SPF 50+ protection lipblock should also be used.

- **Skin checks.** If you notice anything different on your skin or a freckle or mole looks like it is changing in some way then make an appointment to see your doctor or local dermatologist to get it checked out. 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 a window to reduce 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 disability facilities grant to help fund this (<https://www.gov.uk/disabled-facilities-grants>) or the XP charities may be able to help. Cars may also need this protection. Remember an open window will let in UVR. Drawing curtains and blinds is an effective way of reducing UVR but makes a room dark.
- **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 with XP. Bulbs can be covered with protective UVR sleeves or changed to ones that produce less UVR such as LEDs.

- **UV meter.** As UVR is invisible it can be hard to know when you are at risk. A UV meter, available to those seen in the UK National XP service, can help with a more accurate assessment of the amount of UVR in the environment.

Where can I get more information about XP?

www.gstt.nhs.uk/xp The UK National XP service which also provides links to further patient information leaflets

<http://xpsupportgroup.org.uk/> XP support group working in partnership with <http://www.teddingtontrust.com/>

For details of source materials used please contact the Clinical Standards Unit (clinicalstandards@bad.org.uk).

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.

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

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