

Xeroderma pigmentosum

This leaflet will answer some of the questions you might have about xeroderma pigmentosum (XP). If you have any further questions, please speak to a doctor or nurse caring for you.

What is XP and why do I have it?

XP is a rare skin condition where the body is not able to repair damage caused by the ultraviolet (UV) part of daylight.

If you have XP you are born with it, even if it is not diagnosed until later in life. It is inherited by a faulty gene passed unknowingly from both parents. It affects both sexes and all ethnic groups.

What are the signs and symptoms?

- Easy sun-burning despite adequate sun protection, unrelated to the colour of your skin
- Freckles from an early age (under two years) at sites exposed to daylight
- Eye sensitivity to bright light (photophobia)
- Skin cancers
- Skin ageing
- Nerve/brain (neurological) problems, such as hearing loss, poor balance, poor memory or learning problems.

People can experience some or all of the above signs and symptoms.

Do I need any tests to confirm the diagnosis?

Yes, a skin biopsy is required to diagnose XP, so a small piece of skin is removed and sent to the laboratory. The biopsy is done under a local anaesthetic – an injection to numb the area of skin that the biopsy will be taken from. A separate leaflet, **Having a skin biopsy**, is available. Please ask if you would like a copy.

Results of the biopsy to identify if you have XP can usually be expected within three months.

If a member of your family has already been diagnosed with XP and you also have some symptoms, diagnosis from a blood sample may be possible. This would need to be discussed with your consultant.

What treatments are available?

XP is a lifelong condition, and there is currently no known cure. However, there are a number of ways in which XP can be managed. For example:

- complete protection is needed from ultraviolet radiation (UVR) to keep skin damage to a minimum
- regular skin checks (usually every three to six months) can detect and treat any signs of skin cancers early
- an eye check (at least once a year) can detect any problems and ensure you are offered treatments, such as eye drops, advice about glasses and biopsies of any suspicious areas in the eye
- Neurological assessment can identify any brain or nerve problems. This will involve a physical examination and sometimes hearing tests, nerve conduction studies and an MRI (magnetic resonance imaging) scan. Early detection of these problems can ensure that the correct support is organised.
- you will be given the opportunity to speak to a psychologist if you feel you need support regarding your XP, and how you are managing with your diagnosis.
- blood tests can check vitamin D levels, so that supplements can be given if required. Most people with XP have a low vitamin D level as a result of protecting their skin from daylight.

What happens if I do not get treatment?

If you do not protect yourself from all UVR, freckling can get worse and the chance of getting skin cancers is much higher. If these cancers are not detected early they can get larger and deeper. This will cause more scarring if we are able to remove the cancer. If not detected early, this can be life threatening, as the cancer may spread to other organs.

If your vitamin D levels are low and you do not take supplements there is a risk that your bones will be weaker. Ask for our leaflet **Vitamin D and xeroderma pigmentosum** if you don't have a copy.

Is there anything I can do to help myself?

Yes, there are lots of things you can do to help yourself.

Complete skin protection from UVR is recommended to reduce damage to the skin. You should stay away from **all** sources of UVR wherever possible. UVR can be found in all types of daylight including sun and shade. UVR can also pass through most window glass and can be found in some types of light bulbs. The XP nurses can give you more information and advice on sources of UVR.

You can also wear UVR protective clothing, such as:

- wide brimmed or legionnaires' hats to cover as much of the head, face and neck as is possible
- full face visors and/or wrap-around sunglasses that conform to EU standards.
- long-sleeved tops
- full length trousers
- gloves
- long hairstyles, which can help protect the ears and the back of your neck.

Closely woven materials are more effective in reducing UVR exposure. To check how close the weave of the material is, hold it up to the light – the less light passing through it, the more protection it will give you. Some clothing is made from ultraviolet protective factor (UPF) treated materials. There is nothing wrong with these, but dense weave materials can be just as effective and are often cheaper. Some patients choose to wear two layers of clothing to completely block their skin from UVR.

Forward planning is important. UVR risk is higher in the summer and in the middle of the day when the sun is at its highest and should be avoided as much as possible. Plan travel so that time spent outside is minimised.

Using sunscreen

- For your skin, use a sunscreen with high (50+) SPF (sun protection factor) and with a high UVA star rating.
- For your lips, use sunscreen or lip screen with a high SPF.

Sunscreen should be applied to all UVR-exposed areas of the body, ensuring nowhere is missed. It should be applied 20 to 30 minutes before any exposure to UVR, and reapplied every two to three hours while exposed to risk. Sunscreen should form a film when first applied – this ensures enough has been used.

Sunscreens are available in different formulations (creams, lotions, sprays, roll-ons). Some are available on prescription. We can help you choose a suitable sunscreen and show you examples of products that are available.

It is often helpful to have a stock of sunscreen in different places so that you always have some available.

If make-up is used, sunscreen should be applied prior to application.

Knowledge of your own condition

It is important that you have a good understanding of your condition so that you can detect any potential problems early. You can do this by:

- observing your own skin for signs of any suspicious skin growth or spots. You may need a family member to help with any areas you cannot see clearly. Any concerns regarding your skin should be reported immediately to either your local dermatologist or the XP team at St Thomas' Hospital.
- being aware of any potential eye problems. If you have any concerns about your eyes, contact either your local ophthalmologist or the XP team at St Thomas' Hospital.

Your environment

Windows

UVR can pass through most window glass, so a UVR protective film should be applied. This is clear and will not affect the visible light in the room. Various companies can supply this at a cost. This cost can sometimes be met by council/housing associations, if applicable, and the education authority if schools/colleges are affected. The XP Support Group may also be able to help with funding (contact details at the end of this leaflet).

Some window glass has an inbuilt UVR filter, which is applied when it is made. This can be found out by contacting the manufacturer or testing with a UV meter.

Drawing curtains or pulling blinds on unprotected windows reduces UVR levels. Open windows let in more UVR, so keep them closed as much as possible.

Light bulbs

Most light bulbs produce some UVR, in particular halogen, fluorescent and compact fluorescent (energy saving) bulbs. In general, the further away from the bulb a person is, the lower the risk. Risk can also be significantly reduced by covering bulbs in a protective cover. For more information, please contact the XP clinical nurse specialist (CNS).

The XP CNS can assess UVR risk using a UV meter and can visit you at home, school, college, university or in your workplace, if required. UV meters can also be purchased if this is something you feel you would use. Details of the meters are available from the XP nurse specialists and the XP Support Group.

Lifestyle

Smoking

Smoking can increase the chances of cancer for anyone with XP so we strongly recommend that you do not smoke. Help and advice regarding stopping smoking are available at www.smokefree.nhs.uk.

Work/school/college/university

It is best to tell your employer or teachers that you have XP and give them some basic information about your condition. They may be able to help with reducing UVR by protecting the work/school environment for you, and structuring your hours to suit your health needs. The XP CNSs are happy to give advice, support and offer visits to assess UVR safety. An additional XP leaflet, **When a child with XP is starting at your school** is available on the XP section of our website (www.gstt.nhs.uk/xp) or you can request a copy from the XP nurses.

The type of job you have may have been influenced by your medical need. Careers advice regarding jobs that provide less exposure to UVR can be obtained from the XP clinical nurse specialists.

Holidays

Holidays do not need to be avoided because of XP, but planning is important. Choosing the destination or time of year may help reduce risk. Ensure you have packed adequate supplies of protective clothing and sunscreen. It may be helpful, if you are planning a trip abroad, to take a recent copy of your clinic letter, in case of any medical problems. Remember to inform your holiday insurance company about your XP diagnosis.

Useful sources of information

You may find it helpful to get in touch with other patients with the same condition as you. If you feel you would benefit from this, please contact the XP clinical nurse specialists or the XP Support Group/Teddington Trust

XP Support Group – offers advice and practical help to anyone affected by XP

t: 01494 456 192

e: info@xpsupportgroup.org.uk

w: www.xpsupportgroup.org.uk

Teddington Trust – offers advice and practical help to anyone affected by XP

t: 01233 645360

e: support@teddingtontrust.com

w: www.teddingtontrust.com

Contact us

If you have any questions or concerns about XP, please contact the XP CNS,
t: 020 7188 6339 or 020 7188 6351, Monday to Friday, 9am to 5pm. Out of hours,
please leave a message on the answer phone with your contact details.

Help and advice regarding stopping smoking, **w:** www.smokefree.nhs.uk.

For more information leaflets on conditions, procedures, treatments and services offered at
our hospitals, please visit www.guysandstthomas.nhs.uk/leaflets

Pharmacy Medicines Helpline

If you have any questions or concerns about your medicines, please speak to the staff caring for
you or call our helpline.

t: 020 7188 8748 9am to 5pm, Monday to Friday

Your comments and concerns

For advice, support or to raise a concern, contact our Patient Advice and Liaison Service
(PALS). To make a complaint, contact the complaints department.

t: 020 7188 8801 (PALS) **e:** pals@gstt.nhs.uk

t: 020 7188 3514 (complaints) **e:** complaints2@gstt.nhs.uk

Language and accessible support services

If you need an interpreter or information about your care in a different language or format,
please get in touch.

t: 020 7188 8815 **e:** languagesupport@gstt.nhs.uk

NHS 111

Offers medical help and advice from fully trained advisers supported by experienced nurses and
paramedics. Available over the phone 24 hours a day.

t: 111

NHS Choices

Provides online information and guidance on all aspects of health and healthcare, to help you
make choices about your health.

w: www.nhs.uk

Get involved and have your say: become a member of the Trust

Members of Guy's and St Thomas' NHS Foundation Trust contribute to the organisation on a
voluntary basis. We count on them for feedback, local knowledge and support. Membership is
free and it is up to you how much you get involved. To find out more, please get in touch.

t: 0800 731 0319 **e:** members@gstt.nhs.uk **w:** www.guysandstthomas.nhs.uk/membership

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