

Genetic testing for Xeroderma pigmentosum (XP)

Your doctor has requested a blood test for genetic testing in order to find out if you (or your child) have the rare genetic condition called Xeroderma pigmentosum, also known as XP. This leaflet explains more about genetic testing for XP including the benefits, risks and any alternatives.

If you have any further questions, please speak to a doctor or nurse caring for you.

What is genetic testing for XP?

This test is done to confirm if you (or your child) have XP.

This is a unique test. It is available on the NHS and has been specially developed to support the National XP Service.

- You will need to have a simple blood test first.
- The genetics test is performed on your sample of blood.
- The test is extremely complex and takes up to **six months** to be completed.
- The tests may need to be repeated on a new sample of blood if the results are not certain, although this is a very rare occurrence.

How is XP inherited?

XP is a 'recessive genetic condition'.

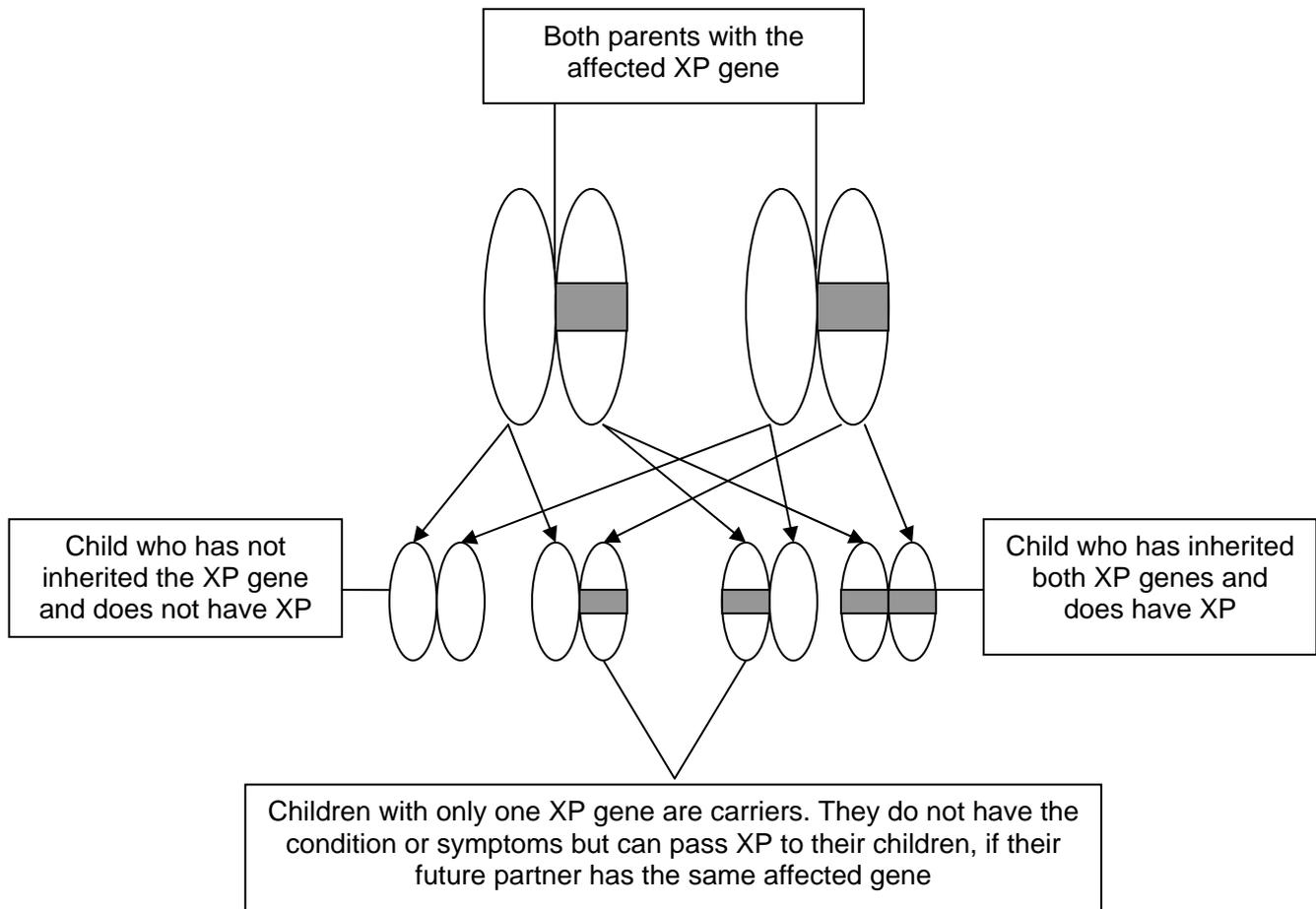
A recessive condition means two copies of a certain gene must be present in order for a condition to develop.

Genes come in pairs. One gene comes from the mother and the other from the father. Recessive inheritance means both the genes from each parent must be affected to cause the disorder. People with only one affected gene in their pair are called carriers and they do not have the condition. However, they can pass the affected gene onto their children.

If you are born to parents that both carry the same affected gene, you have a 1 in 4 chance of inheriting both affected genes and developing the disease, a 1 in 2 chance of being a carrier for the condition, and a 1 in 4 chance of not being affected at all.

XP gene = 

Recessive genetic conditions:



Parents that are closely related to each other, such as first cousins, are more likely to have children with recessive conditions. This is because they are more likely to carry the same faulty genes. This is the case for all recessive conditions, not just XP.

Having this genetic information about yourself may change your life decisions in the future and you may need to think about this carefully. If it is proved that you do have XP you will be offered the opportunity to have genetics counselling.

Giving consent (permission)

We want to involve you in decisions about your care and treatment. If you decide to go ahead, you will be asked to sign a consent form. This states that you agree to have the treatment and you understand what it involves. Parents will need to give consent for their child to be tested.

If you would like more information about our consent process, please speak to a member of staff caring for you.

Will I have a follow-up appointment?

You will be informed by letter of the results once they have been completed. If you **do not** have XP then you will be referred back to the care of your GP or referring doctor. If you **do** have XP then you will be invited to the National XP service. The National XP Service is a rare diseases specialist service funded by NHS England. The service works closely with patients and their local health authorities to provide the best coordinated care for people with XP.

Useful sources of information

For further information please visit: www.geneticseducation.nhs.uk

Contact us

If you have any questions or concerns about your results for genetics testing please contact the XP administrator on 020 7188 0847 (Monday to Friday, 9am to 5pm).

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 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, and to become a member:

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

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