

XPC gene

XPC complex subunit, DNA damage recognition and repair factor

Normal Function

The *XPC* gene provides instructions for making a protein that is involved in repairing damaged DNA. DNA can be damaged by ultraviolet (UV) rays from sunlight and by toxic chemicals, radiation, and unstable molecules called free radicals.

DNA damage occurs frequently, but normal cells are usually able to fix it before it can cause problems. One of the major mechanisms that cells use to fix DNA is known as nucleotide excision repair (NER). The XPC protein starts this repair process by detecting DNA damage. Then a group (complex) of other proteins unwind the section of DNA where the damage has occurred, snip out (excise) the abnormal section, and replace the damaged area with the correct DNA.

Studies suggest that the XPC protein may have additional roles in DNA repair and in other cell activities. Less is known about these proposed functions of the XPC protein.

Health Conditions Related to Genetic Changes

Xeroderma pigmentosum

Many variants (also called mutations) in the *XPC* gene have been found to cause xeroderma pigmentosum. People with this condition have an extreme sensitivity to UV rays from sunlight. As a result, affected individuals have a high risk of sunlight-induced cancer and premature aging.

Variants in the *XPC* gene are the most common cause of this disorder in the United States and Europe. Most *XPC* gene variants prevent the production of any XPC protein. A loss of this protein keeps cells from repairing DNA damage normally. As damage builds up in DNA, cells malfunction and eventually become cancerous or die.

These problems with DNA repair cause people with xeroderma pigmentosum to be extremely sensitive to UV rays. When UV rays damage genes that control cell growth and division, cells can grow too fast and in an uncontrolled way. This uncontrolled cell growth can lead to cancer. In people with xeroderma pigmentosum, cancers occur most frequently in areas of the body that are exposed to the sun, such as the skin and eyes.

Individuals with xeroderma pigmentosum caused by variants in the *XPC* gene may have an increased risk of early menopause compared to the general population. Unlike some of the other forms of xeroderma pigmentosum, when the disorder is caused by variants in the *XPC* gene it is generally not associated with neurological abnormalities (such as delayed development and hearing loss). It is unclear why some people with xeroderma pigmentosum develop neurological abnormalities and others do not.

Other Names for This Gene

- RAD4
- Xeroderma pigmentosum group C-complementing protein
- xeroderma pigmentosum, complementation group C
- XP3
- XPC_HUMAN
- XPCC

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of XPC ([https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=7508\[geneid\]](https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=7508[geneid]))

Scientific Articles on PubMed

- PubMed ([https://pubmed.ncbi.nlm.nih.gov/?term=\(XPC%5BTI%5D\)+AND+\(\(Genes%5BMH%5D\)+OR+\(Genetic+Phenomena%5BMH%5D\)\)+AND+english%5Bla%5D+AND+human%5Bmh%5D\)](https://pubmed.ncbi.nlm.nih.gov/?term=(XPC%5BTI%5D)+AND+((Genes%5BMH%5D)+OR+(Genetic+Phenomena%5BMH%5D))+AND+english%5Bla%5D+AND+human%5Bmh%5D)))

Catalog of Genes and Diseases from OMIM

- XPC COMPLEX SUBUNIT, DNA DAMAGE RECOGNITION AND REPAIR FACTOR; XPC (<https://omim.org/entry/613208>)

Gene and Variant Databases

- NCBI Gene (<https://www.ncbi.nlm.nih.gov/gene/7508>)
- ClinVar ([https://www.ncbi.nlm.nih.gov/clinvar?term=XPC\[gene\]](https://www.ncbi.nlm.nih.gov/clinvar?term=XPC[gene]))

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Genomic Location

The *XPC* gene is found on chromosome 3 (<https://medlineplus.gov/genetics/chromosome/3/>).

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