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 the sun 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

More than 40 mutations in the *XPC* gene have been found to cause xeroderma pigmentosum. Mutations in this gene are the most common cause of this disorder in the United States and Europe.

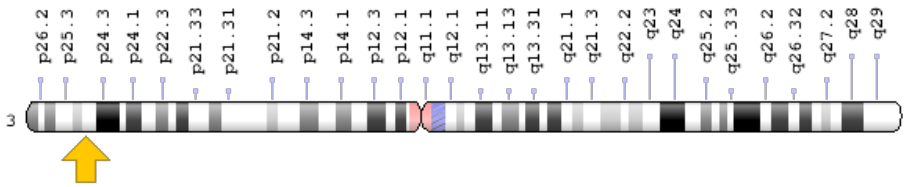
Most *XPC* gene mutations prevent the production of any XPC protein. A loss of this protein keeps cells from repairing DNA damage normally. As a result, abnormalities accumulate in DNA, causing cells to malfunction and eventually to become cancerous or die. These problems with DNA repair cause people with xeroderma pigmentosum to be extremely sensitive to UV rays from sunlight. When UV rays damage genes that control cell growth and division, cells can grow too fast and in an uncontrolled way. As a result, people with xeroderma pigmentosum have a greatly increased risk of developing cancer. These cancers occur most frequently in areas of the body that are exposed to the sun, such as the skin and eyes.

Unlike some of the other forms of xeroderma pigmentosum, when the disorder is caused by mutations 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.

Chromosomal Location

Cytogenetic Location: 3p25.1, which is the short (p) arm of chromosome 3 at position 25.1

Molecular Location: base pairs 14,145,145 to 14,178,672 on chromosome 3 (Homo sapiens Updated Annotation Release 109.20190905, GRCh38.p13) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

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

Additional Information & Resources

Educational Resources

- Cancer Medicine (sixth edition, 2003): Steps in Nucleotide Excision Repair (image) https://www.ncbi.nlm.nih.gov/books/NBK13017/?rendertype=figure&id=A5537
- Molecular Biology of the Cell (fourth edition, 2002): DNA Repair https://www.ncbi.nlm.nih.gov/books/NBK26879/
- Xeroderma Pigmentosum Society, Inc.: DNA Repair Explained in Simple Terms https://www.xps.org/single-post/2015/04/08/DNA-Repair-Explained-in-Simple-Terms

Clinical Information from GeneReviews

- Xeroderma Pigmentosum https://www.ncbi.nlm.nih.gov/books/NBK1397

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28XPC%5BTI%5D%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1800+days%22%5Bdp%5D

Catalog of Genes and Diseases from OMIM

- XPC GENE
  http://omim.org/entry/613208

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
  http://atlasgeneticsoncology.org/Genes/XPCID122.html
- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=XPC%5Bgene%5D
- HGNC Gene Symbol Report
  https://www.genenames.org/data/gene-symbol-report/#!/hgnc_id/HGNC:12816
- Monarch Initiative
  https://monarchinitiative.org/gene/NCBIGene:7508
- NCBI Gene
  https://www.ncbi.nlm.nih.gov/gene/7508
- UniProt
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