

Gene Section

Review

ERCC4 (xeroderma pigmentosum, complementation group F)

Anne Sary, Alain Sarasin

Laboratory of Genetic Instability and Cancer, UPR2169 CNRS, Institut de Recherches sur le Cancer, 7, rue guy Moquet, BP 8, 94801 Villejuif, France (AS, AS)

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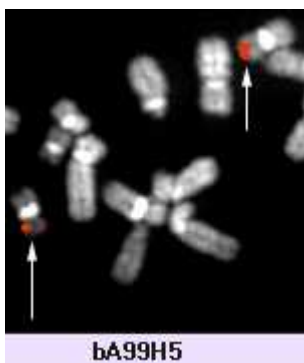
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Identity

Other names: XPF (xeroderma pigmentosum, complementation group F); ERCC4

HGNC (Hugo): ERCC4

Location: 16p13.1-13.2



XPF (16p13) - Courtesy Mariano Rocchi, Resources for Molecular Cytogenetics.

DNA/RNA

Description

28.2 kb.

Transcription

2881 bp =D0 11 exons.

Protein

Description

Xeroderma pigmentosum group F complementing factor; DNA-repair protein complementing XPF cells

905 amino acids; form a stable complex with the ERCC1 protein; The XPF protein and the ERCC1 protein form a complex that exhibits structure specific endonuclease activity that is responsible for the 5' incision during the NER reaction. XPF-ERCC1 also binds to XPA (through ERCC1) and to RPA (through XPF) but not preferentially to damaged DNA. At the site of a lesion Nucleotide Excision Repair (NER) proteins create a DNA bubble structure over a length of approximately 25 nucleotides and the XPG protein incises the damaged DNA strand 0-2 nucleotides 3' to the ssDNA-dsDNA junction. In most studies the 3'-incision made by the XPG protein appeared to be made prior to and independently of the 5'-incision by XPF-ERCC1. XP-F patients have a relatively mild XP phenotype without neurological abnormalities. Cells from XP-F patients are slightly UV-sensitive and exhibit low levels of repair initially after UV-irradiation.

The XPF protein is a single-stranded DNA endonuclease that is also involved in a pathway of recombination repair of DNA interstrand crosslinks.

Homology

Substantial homology with the eucaryotic DNA repair and recombination proteins MEI-9 (*Drosophila melanogaster*), Rad16 (*Saccharomyces cerevisiae*) and Rad1 (*Schizosaccharomyces pombe*).

Mutations

Germinal

9 point mutations; 3 small deletions; 1 small insertion; 1 gross deletion.

Implicated in

Xeroderma pigmentosum, XP group F

Disease

Early skin tumours in XPF patients.

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