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# Gene Section
**Review**

# XPA (xeroderma pigmentosum, complementation group A)

**Anne Stary, 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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## Identity

**Other names:** XPAC xeroderma pigmentosum, complementation group A

**HGNC (Hugo) :** XPA

**Location :** 9q22.3-9q22.3

## DNA/RNA

### Description

Human xeroderma pigmentosum group A 25kbp, six exons, 2 polyadenylation signals.

### Transcription

1377 b mRNA; suggestion of 1 major transcript; promoter G+C rich (73%); one CAAT box and no TATA box.

## Protein

### Description

273 amino acids, 31 kDa. DNA excision repair protein. The functional domain for damaged DNA recognition contains a zinc-finger motif with 4 cysteine residues : Cys-X2-Cys-X17-Cys-X2-Cys motif and a glutamic acid cluster encoded by Exon 2. The nuclear localization signal is located in Exon 1.

### Expression

Ubiquitous.

### Localisation

Nuclear.

### Function

Initiates DNA repair by binding to damaged sites with various affinities, depending upon the chemical structure of the lesion.

Two proteins have been identified and implicated in (one of) the first steps of Nucleotide Excision Repair (NER), i.e. the recognition of lesions in the DNA: the XPA gene product and the XPC gene product. Cells from XPA patients are extremely sensitive to UV and have very low nucleotide excision repair activity. In vitro the XPA protein binds preferentially to damaged DNA compared to nondamaged DNA. The XPA protein binds to replication protein A (RPA) which enhances the affinity of XPA for damaged DNA and is essential for NER. The XPA protein has been shown to bind to ERCC1 and TFIIH. It is possible that the complex XPA/RPA may tell to the repair machinery which strand contained the damage and therefore should be eliminated.

### Homology

Xpac (FlyBase ID) ; Xpa (MGI).

## Mutations

### Germinal

13 nucleotide substitutions and 5 small insertion/deletion in patients.

# Implicated in

## *Xeroderma pigmentosum XPA*

**Disease**

Predisposition to skin cancer: early skin tumours (basal cell carcinoma, squamous cell carcinoma and melanoma); early internal tumours.

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