Gene Section
Review

XPC (xeroderma pigmentosum, complementation group C)

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Identity
Other names: XPCC xeroderma pigmentosum, complementation group C
HGNC (Hugo): XPC
Location : 3p25.1

DNA/RNA
Description
17703 bp; 16 exons.

Transcription
3558 b mRNA.

Protein
Description
939 amino acids.

Expression
Ubiquitous.

Localisation
Nuclear.

Function
Involved in the early recognition of DNA damage present in chromatin. Two proteins have been identified and implicated in (one of) the first steps of NER, i.e. the recognition of lesions in the DNA: the XPA gene product and the XPC gene product in complex with HR23B. This XPC-HR23B complex has been implicated in DNA damage recognition, especially the cyclobutane pyrimidine dimers induced by UV-light. XPC cells have low Nucleotide Excision Repair (NER) repair capacity, but the residual repair has been shown to occur specifically in transcribed genes. It is very likely that the XPC-HR23B complex is the principal damage recognition complex i.e. essential for the recognition of DNA lesions in the genome. Binding of XPC-HR23B to a DNA lesion causes local unwinding, so that the XPA protein can bind and the whole repair machinery can be loaded onto the damaged site. The XPC-HR23B complex is only required for global genome repair. In case of transcription coupled repair when an RNA polymerase is stalled at a lesion, the DNA is unwound by the transcription complex and XPA can bind independently of XPC-HR23B complex.

Homology
MGI : Xpc (Nb 103557).

Mutations
Germlinal
19 mutated sites involved in the XP group C syndrome (XPC), 95% of these mutations (non sense, frameshift, deletion or splice site mutations) give rise to truncated proteins indicating that the XPC gene is not essential for viability.

Implicated in
Xeroderma pigmentosum XPC
Disease
Predisposition to skin cancer: early skin tumours.

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