

ERCC-3 (Excision repair cross-complementing rodent repair deficiency, complementation group 3)

Identity

Other names

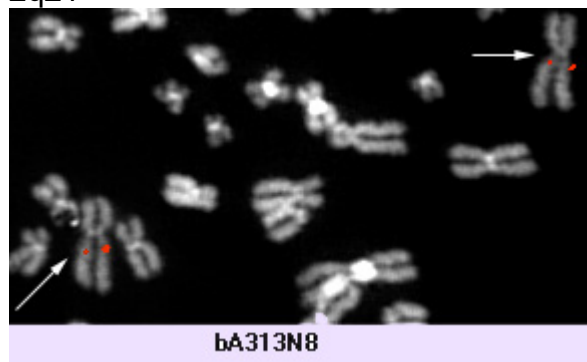
XPB

Hugo Location

XPBC

ERCC3

2q21



[XPB \(2q21\)](#) - Courtesy Mariano Rocchi, [Resources for Molecular Cytogenetics](#). Laboratories willing to validate the probes are welcome : contact rocchi@biologia.uniba.it

DNA/RNA

Description 2751 b mRNA

Protein

Description 782 amino acids

Expression ubiquitous

Localisation nuclear

Function DNA excision repair protein. 3'-5' ATP-dependent helicase activity involved in excision DNA repair and initiation of basal transcription

The XPB protein displays a 3'-5' helicase activity. This protein is a subunit of the basal transcription factor TFIIF involved in both [Nucleotide Excision Repair](#) (NER) and the initiation of RNA polymerase II . Indeed, TFIIF fulfills a dual role in transcription initiation and NER and the role of TFIIF in NER might closely mimic its role in the transcription initiation process. In transcription initiation TFIIF is thought to be involved in unwinding of the promoter site to allowing promoter clearance. In the NER process TFIIF causes unwinding of the lesion-containing region

that has been localized by XPC-HR23B and XPA-RPA, enabling the accumulation of NER proteins around the damaged site.

Among the Xeroderma pigmentosum (XP) patients, XPB patients are extremely rare (only 3 patients known in the world) due to the fact that the XPB gene product is essential for transcription initiation and in all cases, these patients show the double symptoms of Xeroderma pigmentosum and Cockayne syndrome (CS) (see below).

Homology haywire gene (FLYBASE, hay) ; Ercc3 (MGI : 95414)

Mutations

Germinal F99S (T296C) is found in two XPB/CS patients; T119P (A355C) is found in two TTD/XPB patients; FS740 is found in one XPB/CS patient

Implicated in

Entity ERCC3/XPB

Disease [Xeroderma pigmentosum](#) and [Cockayne syndrome](#) in the same patient or [Trichothiodystrophy](#). Early skin cancers

External links

Nomenclature

[Hugo](#)

[ERCC3](#)

[GDB](#)

[ERCC3](#)

[Entrez Gene](#)

[ERCC3 2071](#) excision repair cross-complementing rodent repair deficiency, complementation group 3 (xeroderma pigmentosum group B complementing)

Cards

[Atlas](#)

[XPBID296](#)

[GeneCards](#)

[ERCC3](#)

[Ensembl](#)

[ERCC3](#)

[CancerGene](#)

[ERCC3](#)

[Genatlas](#)

[ERCC3](#)

[GeneLynx](#)

[ERCC3](#)

[eGenome](#)

[ERCC3](#)

[euGene](#)

[2071](#)

Genomic and cartography

[GoldenPath](#)

[ERCC3 - 2q21 chr2:127731096-127767982 - 2q14.3](#) (hg17-May_2004)

[Ensembl](#)

[ERCC3 - 2q14.3 \[CytoView\]](#)

[NCBI](#)

[Genes Cyto](#) [Gene Seq](#) [Map View - NCBI]

[OMIM](#)

[Disease map \[OMIM\]](#)

[HomoloGene](#)

[ERCC3](#)

Gene and transcription

[Genbank](#)

[AY163769](#) [SRS] [AY163769](#) [ENTREZ]

[Genbank](#)

[AK091500](#) [SRS] [AK091500](#) [ENTREZ]

[Genbank](#)

[AK095557](#) [SRS] [AK095557](#) [ENTREZ]

[Genbank](#)

[AK127469](#) [SRS] [AK127469](#) [ENTREZ]

[Genbank](#)

[BC008820](#) [SRS] [BC008820](#) [ENTREZ]

[RefSeq](#)

[NM_000122](#) [SRS] [NM_000122](#) [ENTREZ]

[RefSeq](#)

[NT_086627](#) [SRS] [NT_086627](#) [ENTREZ]

[AceView](#)

[ERCC3](#) AceView - NCBI

[TRASER](#)

[ERCC3](#) Traser - Stanford

[Unigene](#)

[Hs.469872](#) [SRS] [Hs.469872](#) [NCBI] [HS469872](#) [spliceNest]

	Protein : pattern, domain, 3D structure
SwissProt	P19447 [SRS] P19447 [EXPASY] P19447 [INTERPRO]
Interpro	IPR001650 Helicase_C [SRS] IPR001650 Helicase_C [EBI]
Interpro	IPR006935 ResIII [SRS] IPR006935 ResIII [EBI]
Interpro	IPR001161 XPB_DNA_repair [SRS] IPR001161 XPB_DNA_repair [EBI]
CluSTr	P19447
Pfam	PF00271 Helicase_C [SRS] PF00271 Helicase_C [Sanger]
Blocks] pfam00271 [NCBI-CDD]
	P19447
	Polymorphism : SNP, mutations, diseases
OMIM	133510 [map]
GENECLINICS	133510
SNP	ERCC3 [dbSNP-NCBI]
SNP	NM_000122 [SNP-NCI]
SNP	ERCC3 [GeneSNPs - Utah] ERCC3 [SNP - CSHL] ERCC3 [HGBASE - SRS]
	General knowledge
Family Browser	ERCC3 [UCSC Family Browser]
SOURCE	NM_000122
SMD	Hs.469872
SAGE	Hs.469872
Enzyme	3.6.1.- [Enzyme-SRS] 3.6.1.- [Brenda-SRS] 3.6.1.- [KEGG] 3.6.1.- [WIT]
Amigo	function 3' to 5' DNA helicase activity
Amigo	function ATP binding
Amigo	function ATP-dependent DNA helicase activity
Amigo	process DNA topological change
Amigo	function damaged DNA binding
Amigo	function helicase activity
Amigo	function hydrolase activity
Amigo	process induction of apoptosis
Amigo	component nucleus
Amigo	process perception of sound
Amigo	function protein binding
Amigo	process regulation of transcription, DNA-dependent
Amigo	component transcription factor TFIIH complex
Amigo	process transcription from Pol II promoter
Amigo	process transcription-coupled nucleotide-excision repair
BIOCARTA	CARM1 and Regulation of the Estrogen Receptor
BIOCARTA	Chromatin Remodeling by hSWI/SNF ATP-dependent Complexes
BIOCARTA	Nuclear receptors coordinate the activities of chromatin remodeling complexes and coactivators to facilitate initiation of transcription in carcinoma cells
PubGene	ERCC3
	Other databases
	Probes
Probe	Cancer Cytogenetics (Bari)
Probe	ERCC3 Related clones (RZPD - Berlin)
	PubMed
PubMed	33 Pubmed reference(s) in LocusLink
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Medline [2167179](#)

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Contributor(s)

Written 02-2001 Anne Stary and Alain Sarasin

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