

Gene Section

Mini Review

ERCC3 (Excision repair cross-complementing rodent repair deficiency, complementation group 3)

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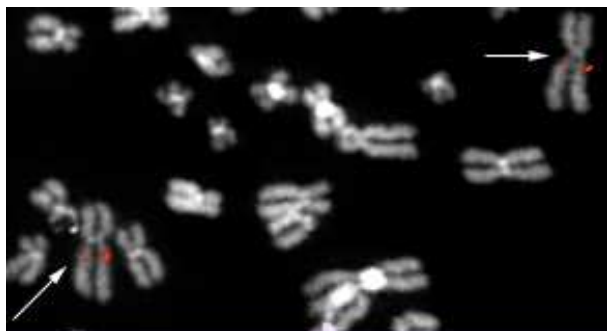
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Identity

Other names: ERCC-3 (Excision repair cross-complementing rodent repair deficiency, complementation group 3); XPB; XPBC

HGNC (Hugo): ERCC3

Location: 2q21



XPB (2q21) - Courtesy Mariano Rocchi, Resources for Molecular Cytogenetics.

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 TFIID involved in both Nucleotide Excision Repair (NER) and the initiation of RNA polymerase II. Indeed, TFIID fulfills a dual role in transcription initiation and NER and the role of TFIID in NER might closely mimic its role in the transcription initiation process. In transcription initiation TFIID is thought to be involved in unwinding of the promoter site to allowing promoter clearance. In the NER process TFIID 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

ERCC3/XPB

Disease

Xeroderma pigmentosum and Cockayne syndrome in the same patient or Trichothiodystrophy. Early skin cancers.

References

Weeda G, van Ham RC, Masurel R, Westerveld A, Odijk H, de Wit J, Bootsma D, van der Eb AJ, Hoeijmakers JH. Molecular cloning and biological characterization of the human excision repair gene ERCC-3. *Mol Cell Biol.* 1990 Jun;10(6):2570-81

Weeda G, van Ham RC, Vermeulen W, Bootsma D, van der Eb AJ, Hoeijmakers JH. A presumed DNA helicase encoded by ERCC-3 is involved in the human repair disorders xeroderma pigmentosum and Cockayne's syndrome. *Cell.* 1990 Aug 24;62(4):777-91

Weeda G, Ma L, van Ham RC, Bootsma D, van der Eb AJ, Hoeijmakers JH. Characterization of the mouse homolog of the XPBC/ERCC-3 gene implicated in xeroderma pigmentosum and Cockayne's syndrome. *Carcinogenesis.* 1991 Dec;12(12):2361-8

Weeda G, Ma LB, van Ham RC, van der Eb AJ, Hoeijmakers JH. Structure and expression of the human XPBC/ERCC-3 gene involved in DNA repair disorders xeroderma pigmentosum and Cockayne's syndrome. *Nucleic Acids Res.* 1991 Nov 25;19(22):6301-8

Weeda G, Wiegant J, van der Ploeg M, Geurts van Kessel AH, van der Eb AJ, Hoeijmakers JH. Localization of the xeroderma pigmentosum group B-correcting gene ERCC3 to human chromosome 2q21. *Genomics.* 1991 Aug;10(4):1035-40

Ma L, Weeda G, Jochemsen AG, Bootsma D, Hoeijmakers JH, van der Eb AJ. Molecular and functional analysis of the XPBC/ERCC-3 promoter: transcription activity is dependent on the integrity of an Sp1-binding site. *Nucleic Acids Res.* 1992 Jan 25;20(2):217-24

Mounkes LC, Jones RS, Liang BC, Gelbart W, Fuller MT. A *Drosophila* model for xeroderma pigmentosum and Cockayne's syndrome: haywire encodes the fly homolog of ERCC3, a human excision repair gene. *Cell.* 1992 Dec 11;71(6):925-37

Schaeffer L, Roy R, Humbert S, Moncollin V, Vermeulen W, Hoeijmakers JH, Chambon P, Egly JM. DNA repair helicase: a component of BTF2 (TFIIH) basic transcription factor. *Science.* 1993 Apr 2;260(5104):58-63

Drapkin R, Reardon JT, Ansari A, Huang JC, Zawel L, Ahn K, Sancar A, Reinberg D. Dual role of TFIIH in DNA excision repair and in transcription by RNA polymerase II. *Nature.* 1994 Apr 21;368(6473):769-72

Humbert S, van Vuuren H, Lutz Y, Hoeijmakers JH, Egly JM, Moncollin V. p44 and p34 subunits of the BTF2/TFIIH transcription factor have homologies with SSL1, a yeast protein involved in DNA repair. *EMBO J.* 1994 May 15;13(10):2393-8

Ma L, Westbroek A, Jochemsen AG, Weeda G, Bosch A, Bootsma D, Hoeijmakers JH, van der Eb AJ. Mutational analysis of ERCC3, which is involved in DNA repair and transcription initiation: identification of domains essential for the DNA repair function. *Mol Cell Biol.* 1994 Jun;14(6):4126-34

Schaeffer L, Moncollin V, Roy R, Staub A, Mezzina M, Sarasin A, Weeda G, Hoeijmakers JH, Egly JM. The ERCC2/DNA repair protein is associated with the class II BTF2/TFIIH transcription factor. *EMBO J.* 1994 May 15;13(10):2388-92

van Vuuren AJ, Vermeulen W, Ma L, Weeda G, Appeldoorn E, Jaspers NG, van der Eb AJ, Bootsma D, Hoeijmakers JH, Humbert S. Correction of xeroderma pigmentosum repair defect by basal transcription factor BTF2 (TFIIH). *EMBO J.* 1994 Apr 1;13(7):1645-53

Dabholkar MD, Berger MS, Vionnet JA, Overton L, Thompson C, Bostick-Bruton F, Yu JJ, Silber JR, Reed E. Comparative analyses of relative ERCC3 and ERCC6 mRNA levels in gliomas and adjacent non-neoplastic brain. *Mol Carcinog.* 1996 Sep;17(1):1-7

Hwang JR, Moncollin V, Vermeulen W, Seroz T, van Vuuren H, Hoeijmakers JH, Egly JM. A 3' → 5' XPB helicase defect in repair/transcription factor TFIIH of xeroderma pigmentosum group B affects both DNA repair and transcription. *J Biol Chem.* 1996 Jul 5;271(27):15898-904

Léveillard T, Andera L, Bissonnette N, Schaeffer L, Bracco L, Egly JM, Wasylyk B. Functional interactions between p53 and the TFIIH complex are affected by tumour-associated mutations. *EMBO J.* 1996 Apr 1;15(7):1615-24

Qadri I, Conaway JW, Conaway RC, Schaack J, Siddiqui A. Hepatitis B virus transactivator protein, HBx, associates with the components of TFIIH and stimulates the DNA helicase activity of TFIIH. *Proc Natl Acad Sci U S A.* 1996 Oct 1;93(20):10578-83

Wang XW, Vermeulen W, Coursen JD, Gibson M, Lupold SE, Forrester K, Xu G, Elmore L, Yeh H, Hoeijmakers JH, Harris CC. The XPB and XPD DNA helicases are components of the p53-mediated apoptosis pathway. *Genes Dev.* 1996 May 15;10(10):1219-32

Yankulov KY, Pandes M, McCracken S, Bouchard D, Bentley DL. TFIIH functions in regulating transcriptional elongation by RNA polymerase II in *Xenopus* oocytes. *Mol Cell Biol.* 1996 Jul;16(7):3291-9

Dianov GL, Houle JF, Iyer N, Bohr VA, Friedberg EC. Reduced RNA polymerase II transcription in extracts of cockayne syndrome and xeroderma pigmentosum/Cockayne syndrome cells. *Nucleic Acids Res.* 1997 Sep 15;25(18):3636-42

Tu Y, Bates S, Pfeifer GP. Sequence-specific and domain-specific DNA repair in xeroderma pigmentosum and Cockayne syndrome cells. *J Biol Chem.* 1997 Aug 15;272(33):20747-55

Weeda G, Eveno E, Donker I, Vermeulen W, Chevallier-Lagente O, Taïeb A, Stary A, Hoeijmakers JH, Mezzina M, Sarasin A. A mutation in the XPB/ERCC3 DNA repair transcription gene, associated with trichothiodystrophy. *Am J Hum Genet.* 1997 Feb;60(2):320-9

Weeda G, Rossignol M, Fraser RA, Winkler GS, Vermeulen W, van 't Veer LJ, Ma L, Hoeijmakers JH, Egly JM. The XPB subunit of repair/transcription factor TFIIH directly interacts with SUG1, a subunit of the 26S proteasome and putative transcription factor. *Nucleic Acids Res.* 1997 Jun 15;25(12):2274-83

Zeng L, Quilliet X, Chevallier-Lagente O, Eveno E, Sarasin A, Mezzina M. Retrovirus-mediated gene transfer corrects DNA repair defect of xeroderma pigmentosum cells of

complementation groups A, B and C. *Gene Ther.* 1997 Oct;4(10):1077-84

de Boer J, de Wit J, van Steeg H, Berg RJ, Morreau H, Visser P, Lehmann AR, Duran M, Hoeijmakers JH, Weeda G. A mouse model for the basal transcription/DNA repair syndrome trichothiodystrophy. *Mol Cell.* 1998 Jun;1(7):981-90

Ribeiro DT, Machado CR, Costa RM, Praekelt UM, Van Sluys MA, Menck CF. Cloning of a cDNA from *Arabidopsis thaliana* homologous to the human XPB gene. *Gene.* 1998 Feb 27;208(2):207-13

Tantin D. RNA polymerase II elongation complexes containing the Cockayne syndrome group B protein interact with a molecular complex containing the transcription factor IIH components xeroderma pigmentosum B and p62. *J Biol Chem.* 1998 Oct 23;273(43):27794-9

Coin F, Bergmann E, Tremeau-Bravard A, Egly JM. Mutations in XPB and XPD helicases found in xeroderma pigmentosum patients impair the transcription function of TFIIH. *EMBO J.* 1999 Mar 1;18(5):1357-66

Maru Y, Kobayashi T, Tanaka K, Shibuya M. BCR binds to the xeroderma pigmentosum group B protein. *Biochem Biophys Res Commun.* 1999 Jul 5;260(2):309-12

Moreland RJ, Tirode F, Yan Q, Conaway JW, Egly JM, Conaway RC. A role for the TFIIH XPB DNA helicase in promoter escape by RNA polymerase II. *J Biol Chem.* 1999 Aug 6;274(32):22127-30

Mounkes LC, Fuller MT. Molecular characterization of mutant alleles of the DNA repair/basal transcription factor haywire/ERCC3 in *Drosophila*. *Genetics.* 1999 May;152(1):291-7

Riou L, Zeng L, Chevallier-Lagente O, Stary A, Nikaido O, Taïeb A, Weeda G, Mezzina M, Sarasin A. The relative expression of mutated XPB genes results in xeroderma pigmentosum/Cockayne's syndrome or trichothiodystrophy cellular phenotypes. *Hum Mol Genet.* 1999 Jun;8(6):1125-33

Takeda N, Shibuya M, Maru Y. The BCR-ABL oncoprotein potentially interacts with the xeroderma pigmentosum group B protein. *Proc Natl Acad Sci U S A.* 1999 Jan 5;96(1):203-7

Tirode F, Busso D, Coin F, Egly JM. Reconstitution of the transcription factor TFIIH: assignment of functions for the three enzymatic subunits, XPB, XPD, and cdk7. *Mol Cell.* 1999 Jan;3(1):87-95

Butkiewicz D, Rusin M, Harris CC, Chorazy M. Identification of four single nucleotide polymorphisms in DNA repair genes: XPA and XPB (ERCC3) in Polish population. *Hum Mutat.* 2000 Jun;15(6):577-8

Douziech M, Coin F, Chipoulet JM, Arai Y, Ohkuma Y, Egly JM, Coulombe B. Mechanism of promoter melting by the xeroderma pigmentosum complementation group B helicase of transcription factor IIH revealed by protein-DNA photo-cross-linking. *Mol Cell Biol.* 2000 Nov;20(21):8168-77

Schultz P, Fribourg S, Poterszman A, Mallouh V, Moras D, Egly JM. Molecular structure of human TFIIH. *Cell.* 2000 Sep 1;102(5):599-607

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