

## BLM (Bloom)

### Identity

Hugo [BLM](#)  
Location 15q26.1

### DNA/RNA

Transcription 4.4kb mRNA

### Protein

Description 1417 amino acids; ATP binding in amino acid 689-696; DEAH box in 795-798; two putative nuclear localization signals in the C-term in 1334-1349

Expression accumulates to high levels in S phase of the cell cycle, persists in G2/M and sharply declines in G1. Hyperphosphorylated in mitosis

Localisation nuclear (PML nuclear bodies and nucleolus)

Function 3'-5'DNA helicase; probable role in DNA replication and double-strand break repair

Preferred substrates : G-quadruplex DNA, D-loops structures and X-junctions.

Recombinant protein promotes ATP-dependent branch migration of Holliday junctions, effects, with topoisomerase III?, the resolution of a recombination intermediate containing a double Holliday junction with no flanking sequence exchanges, and possess a strand pairing activity.

Recombinant BLM possess a strand pairing activity.

Participates in a supercomplex of BRCA1 -associated proteins named BASC (BRCA1-Associated genome Surveillance Complex) containing [ATM](#) (defective in [ataxia telangiectasia](#)), [NBS1](#) (defective in [Nijmegen syndrome](#)) and [MRE11](#) (defective in ataxia-telangiectasia-like disorder), [MLH1](#), MSH2 and MSH6, which are involved in human [non-polyposis colorectal cancer](#), RAD50 and DNA replication factor C.

Participates in a complex named BRAFT (BLM, RPA, FA, Topoisomerase III) containing five of the Fanconia Anemia (FA) complementation group proteins ([FANCA](#), [FANCG](#), [FANCC](#), [FANCE](#) and [FANCF](#)).

Interacts physically and/or functionally with [p53](#), 53BP1, [WRN](#), MLH1, RAD51, TRF2, ATR, the largest subunit of CAF-1, ligase IV, FEN1, Mus81, the monoubiquitinated FANCD2 isoform

Is Associated with telomeres and ribosomal DNA repeats.

Is phosphorylated in mitotic cells through the cdc2 pathway, and in response to DNA damaging agents or stalled replication forks.

Homology homologous to RecQ helicases, a subfamily of DEXH box-containing

helicases; in particular, similarity with the four known human members in the RecQ subfamily, human [RecQL](#), human [Wrn](#), the product of the [Werner syndrome](#) gene, and the human [RecQL4](#), involved in the [Rothmund-Thomson syndrome](#), and [RecQL5](#) proteins

## Mutations

**Germinal** five BLM mutations introducing amino acid substitutions and four BLM mutations introducing premature nonsense codons into the coding sequence have been described to date; one BLM mutation consisting in a 6 bp deletion accompanied by a 7 bp insertion at nucleic acid position 2281 is common in patients from Ashkenazi Jewish ancestry, leading to a truncated protein of 739 amino acids in length; two BLM mutations, 631delCAA and 1610insA were detected in Japanese patients.

## Implicated in

**Entity** [Bloom syndrome](#)

**Disease** Bloom syndrome is a chromosome instability syndrome/cancer prone disease (at risk of numerous, early occurring cancers of various types)

**Prognosis** 1/3 of patients are dead at mean age 24 yrs, and the mean age of the 2/3 remaining alive patients is 22 yrs

**Cytogenetics** chromatid/chromosome breaks; triradial and quadriradial figures, highly elevated spontaneous sister chromatid exchange rate

## External links

### Nomenclature

[Hugo](#) [BLM](#)

[GDB](#) [BLM](#)

[Entrez\\_Gene](#) [BLM\\_641](#) Bloom syndrome

### Cards

[Atlas](#) [BLM109](#)

[GeneCards](#) [BLM](#)

[Ensembl](#) [BLM](#)

[CancerGene](#) [BLM](#)

[Genatlas](#) [BLM](#)

[GeneLynx](#) [BLM](#)

[eGenome](#) [BLM](#)

[euGene](#) [641](#)

### Genomic and cartography

[GoldenPath](#) [BLM](#) - [15q26.1](#) [chr15:89061606-89159601](#) + [15q26.1](#) (hg17-May\_2004)

[Ensembl](#) [BLM - 15q26.1 \[CytoView\]](#)

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

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

[HomoloGene](#) [BLM](#)

### Gene and transcription

[Genbank](#) [BC034480](#) [SRS] [BC034480](#) [ENTREZ]

[Genbank](#) [U39817](#) [SRS] [U39817](#) [ENTREZ]

[RefSeq](#) [NM\\_000057](#) [SRS] [NM\\_000057](#) [ENTREZ]

[RefSeq](#) [NT\\_086832](#) [SRS] [NT\\_086832](#) [ENTREZ]

[AceView](#) [BLM](#) AceView - NCBI

[TRASER](#) [BLM](#) Traser - Stanford

[Unigene](#) [Hs.169348](#) [SRS] [Hs.169348](#) [NCBI] [HS169348](#) [spliceNest]

### Protein : pattern, domain, 3D structure

[SwissProt](#) [P54132](#) [SRS] [P54132](#) [EXPASY] [P54132](#) [INTERPRO]

[Prosite](#) [PS00690](#) [DEAH\\_ATP\\_HELICASE](#) [SRS] [PS00690](#)  
[DEAH\\_ATP\\_HELICASE](#) [Expasy]

[Prosite](#) [PS50967](#) [HRDC](#) [SRS] [PS50967](#) [HRDC](#) [Expasy]

[Interpro](#) [IPR001410](#) [DEAD](#) [SRS] [IPR001410](#) [DEAD](#) [EBI]

[Interpro](#) [IPR002464](#) [DEAH\\_box](#) [SRS] [IPR002464](#) [DEAH\\_box](#) [EBI]

[Interpro](#) [IPR001650](#) [Helicase\\_C](#) [SRS] [IPR001650](#) [Helicase\\_C](#) [EBI]

[Interpro](#) [IPR002121](#) [HRDC](#) [SRS] [IPR002121](#) [HRDC](#) [EBI]

[Interpro](#) [IPR004589](#) [RecQ](#) [SRS] [IPR004589](#) [RecQ](#) [EBI]

[CluSTr](#) [P54132](#)

[Pfam](#) [PF00270](#) [DEAD](#) [SRS] [PF00270](#) [DEAD](#) [Sanger] [pfam00270](#) [NCBI-CDD]

[Pfam](#) [PF00271](#) [Helicase\\_C](#) [SRS] [PF00271](#) [Helicase\\_C](#) [Sanger]  
] [pfam00271](#) [NCBI-CDD]

[Pfam](#) [PF00570](#) [HRDC](#) [SRS] [PF00570](#) [HRDC](#) [Sanger] [pfam00570](#) [NCBI-CDD]

[Blocks](#) [P54132](#)

### Polymorphism : SNP, mutations, diseases

[OMIM](#) [604610](#) [[map](#)]

[GENECLINICS](#) [604610](#)

[SNP](#) [BLM](#) [dbSNP-NCBI]

[SNP](#) [NM\\_000057](#) [SNP-NCI]

[SNP](#) [BLM](#) [GeneSNPs - Utah] [BLM](#) [SNP - CSHL] [BLM](#) [HGBASE - SRS]

### General knowledge

[Family Browser](#) [BLM](#) [UCSC Family Browser]

[SOURCE](#) [NM\\_000057](#)

[SMD](#) [Hs.169348](#)

[SAGE](#) [Hs.169348](#)

[Enzyme](#)      [3.6.1.-](#) [ Enzyme-SRS ]   [3.6.1.-](#) [ Brenda-SRS ]   [3.6.1.-](#) [ KEGG ]   [3.6.1.-](#) [ WIT ]

[Amigo](#)      [function|ATP binding](#)

[Amigo](#)      [function|ATP-dependent DNA helicase activity](#)

[Amigo](#)      [function|DNA binding](#)

[Amigo](#)      [process|DNA recombination](#)

[Amigo](#)      [process|DNA repair](#)

[Amigo](#)      [process|DNA replication](#)

[Amigo](#)      [process|antimicrobial humoral response \(sensu Vertebrata\)](#)

[Amigo](#)      [function|hydrolase activity](#)

[Amigo](#)      [component|nucleus](#)

[PubGene](#)      [BLM](#)

**Other databases**

**Probes**

[Probe](#)      [BLM Related clones \(RZPD - Berlin\)](#)

**PubMed**

[PubMed](#)      [32 Pubmed reference\(s\) in LocusLink](#)

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