Recombinant Human BLM Protein

Cat. No. BLM-34H **Lot. No.** (See product label)

SPECIFICATION

Product Overview	Recombinant human BLM protein without tag was expressed in E. coli.
Species	Human
Source	E.coli
Description	The Bloom syndrome is an autosomal recessive disorder characterized by growth deficiency, microcephaly and immunodeficiency among others. It is caused by homozygous or compound heterozygous mutation in the gene encoding DNA helicase RecQ protein on chromosome 15q26. This Bloom-associated helicase unwinds a variety of DNA substrates including Holliday junction, and is involved in several pathways contributing to the maintenance of genome stability. Identification of pathogenic Bloom variants is required for heterozygote testing in at-risk families.
Form	Lyophilized
Molecular Mass	75 kDa
AA Sequence	MSRNLKHERFQSLSFPHTKEMMKIFHKKFGLHNFRTNQLEAINAALLGEDCFILMPT GGGKSLCYQLPACVSPGVTVVISPLRSLIVDQVQKLTSLDIPATYLTGDKTDSEATNIY LQLSKKDPIIKLLYVTPEKICASNRLISTLENLYERKLLARFVIDEAHCVSQWGHDFRQ DYKRMNMLRQKFPSVPVMALTATANPRVQKDILTQLKILRPQVFSMSFNRHNLKYYV LPKKPKKVAFDCLEWIRKHHPYDSGIIYCLSRRECDTMADTLQRDGLAALAYHAGLS DSARDEVQQKWINQDGCQVICATIAFGMGIDKPDVRFVIHASLPKSVEGYYQESGRA GRDGEISHCLLFYTYHDVTRLKRLIMMEKDGNHHTRETHFNNLYSMVHYCENITECR

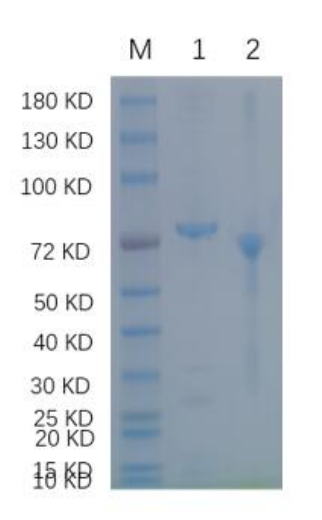
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	RIQLLAYFGENGFNPDFCKKHPDVSCDNCCKTKDYKTRDVTDDVKSIVRFVQEHSS SQGMRNIKHVGPSGRFTMNMLVDIFLGSKSAKIQSGIFGKGSAYSRHNAERLFKKLIL DKILDEDLYINANDQAIAYVMLGNKAQTVLNGNLKVDFMETENSSSVKKQKALVAKV SQREEMVKKCLGELTEVCKSLGKVFGVHYFNIFNTVTLKKLAESLSSDPEVLLQIDGV TEDKLEKYGAEVISVLQKYSEWTSPAEDSSPG
Purity	> 90% by SDS-PAGE
Storage	Store it under sterile conditions at -20 to -80 centigrade. It is recommended that the protein be aliquoted for optimal storage. Avoid repeated freeze-thaw cycles.
Storage Buffer	Lyophilized from 50mM Tris, 500mM NaCl, pH 7.5, 5% Trehalose, 5% Manitol

GENE INFORMATION

Gene Name	BLM BLM RecQ like helicase [Homo sapiens (human)]
Official Symbol	BLM
Synonyms	BLM; BLM RecQ like helicase; BS; RECQ2; RECQL2; RECQL3; MGRISCE1; recQ-like DNA helicase BLM; Bloom syndrome RecQ like helicase; Bloom syndrome, RecQ helicase-like; DNA helicase, RecQ-like type 2; bloom syndrome protein; recQ protein-like 3; EC 3.6.4.12
Gene ID	641
mRNA Refseq	NM_000057
Protein Refseq	NP_000048
MIM	604610

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UniProt ID	P54132
Reducing 12% SDS-PAGE analysis profile of purified BLM	 1. BLM 2. BSA

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