Bardet-Biedl Syndrome 1 Protein (BBS1) Human Recombinant Protein

PRODUCT OVERVIEW

Product Name	Bardet-Biedl syndrome 1 protein (BBS1), Recombinant Protein
Full Product Name	Recombinant Human Bardet-Biedl syndrome 1 protein (BBS1)
Product Gene Name	BBS1 recombinant protein
Product Synonym Gene Name	BBS1
Sequence	AAASSSDSDA CGAESNEANS KWLDAHYDPM ANIHTFSACL ALADLHGDGE YKLVVGDLGP GGQQPRLKVL KGPLVMTESP LPALPAAAAT FLMEQHEPRT PALALASGPC VYVYKNLRPY FKFSLPQLPP NPLEQDLWNQ AKEDRIDPLT LKEMLESIRE TAEEPLSIQS LRFLQLELSE MEAFVNQHKS NSIKRQTVIT TMTTLKKNLA DEDAVSCLVL GTENKELLVL DPEAFTILAK MSLPSVPVFL EVSGQFDVEF RLAAACRNGN IYILRRDSKH PKYCIELSAQ PVGLIRVHKV LVVGSTQDSL HGFTHKGKKL WTVQMPAAIL TMNLLEQHSR GLQAVMAGLA NGEVRIYRDK ALLNVIHTPD AVTSLCFGRY GREDNTLIMT TRGGGLIIKI LKRTAVFVEG GSEVGPPPAQ AMKLNVPRKT RLYVDQTLRE REAGTAMHRA FQTDLYLLRL RAARAYLQAL ESSLSPLSTT AREPLKLHAV VQGLGPTFKL TLHLQNTSTT RPVLGLLVCF LYNEALYSLP RAFFKVPLLV PGLNYPLETF VESLSNKGIS DIIKVLVLRE GQSAPLLSAH VNMPGSEGLA AA
Host	E Coli or Yeast or Baculovirus or Mammalian Cell
Form/Format	Lyophilized or liquid (Format to be determined during the manufacturing process)
Purity/Purification	Greater or equal to 85% purity as determined by SDS-PAGE. (lot specific)
Preparation and Storage	Store at -20 degrees C. For long-term storage, store at -20 degrees C or -80 degrees C. Store working aliquots at 4 degrees C for up to one week. Repeated freezing and thawing is not recommended.
Molecular Weight	49,006 Da
Sequence Positions	2-593aa; Full Length of Mature Protein
Tag Information	This protein contains an N-terminal tag and may also contain a C-terminal tag. Tag types are determined by various factors including tag-protein stability, please inquire for tag information.
Sterility	Sterile filter available upon request.
Endotoxin	Low endotoxin available upon request.
OMIM	209900
3D Structure	ModBase 3D Structure for Q8NFJ9
Species	Human
ISO Certification	Manufactured in an ISO 9001:2015 Certified Laboratory.
Related Product Information for BBS1 recombinant protein	Mutations in this gene have been observed in patients with the major form (type 1) of Bardet-Biedl syndrome. The encoded protein may play a role in eye, limb, cardiac and reproductive system development.
Other Notes	Small volumes of BBS1 recombinant protein vial(s) may occasionally become entrapped in the seal of the product vial during shipment and storage. If necessary, briefly centrifuge the vial on a tabletop centrifuge to dislodge any liquid in the container`s cap. Certain products may require to ship with dry ice and additional dry ice fee may apply.
Searchable Terms for BBS1 purchase	MBS1354426 is a recombinant protein targeting BBS1. Also known as Recombinant Human Bardet-Biedl syndrome 1 protein (BBS1). Host: E Coli or Yeast or Baculovirus or Mammalian Cell.

ADDITIONAL DETAILS

NCBI/Uniprot data below describe general gene information for BBS1. It may not necessarily be applicable to this product.

NCBI GI #	28395045
NCBI GeneID	582
NCBI Accession #	NP_078925.3
UniProt Primary Accession #	Q8NFJ9
UniProt Secondary Accession #	Q32MM9; Q32MN0; Q96SN4
UniProt Related Accession #	Q8NFJ9
GenBank Nucleotide #	NM_024649.4
NCBI Official Full Name	Bardet-Biedl syndrome 1 protein
NCBI Official Synonym Full Names	Bardet-Biedl syndrome 1
NCBI Official Symbol	BBS1
NCBI Official Synonym Symbols	BBS2L2
UniProt Protein Name	Bardet-Biedl syndrome 1 protein
UniProt Synonym Protein Names	BBS2-like protein 2
Protein Family	Bardet-Biedl syndrome 1 protein
UniProt Gene Name	BBS1
UniProt Synonym Gene Names	BBS2L2
NCBI Protein Information	Bardet-Biedl syndrome 1 protein
UniProt Comments for BBS1	The BBSome complex is thought to function as a coat complex required for sorting of specific membrane proteins to the primary cilia. The BBSome complex is required for ciliogenesis but is dispensable for centriolar satellite function. This ciliogenic function is mediated in part by the Rab8 GDP/GTP exchange factor, which localizes to the basal body and contacts the BBSome. Rab8(GTP) enters the primary cilium and promotes extension of the ciliary membrane. Firstly the BBSome associates with the ciliary membrane and binds to RAB3IP/Rabin8, the guanosyl exchange factor (GEF) for Rab8 and then the Rab8-GTP localizes to the cilium and promotes docking and fusion of carrier vesicles to the base of the ciliary membrane. The BBSome complex, together with the LTZL1, controls SMO ciliary trafficking and contributes to the sonic hedgehog (SHH) pathway regulation. Required for proper BBSome complex assembly and its ciliary localization (PubMed:17574030, PubMed:22072986). Plays a role in olfactory cilium biogenesis/maintenance and trafficking ().
Research Articles on BBS1	**1.** Importantly, one Japanese and one Omani families carried compound biallelic mutations in two distinct genes (TMEM67/RPGRIP1L and TMEM138/BBS1, respectively).
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