

Recombinant Full Length Human BBS4 Protein, GST-tagged

Cat. No. BBS4-1586HF Lot. No. (See product label)

SPECIFICATION

Product Overview Human BBS4 full-length ORF (AAH27624, 1 a.a. - 519 a.a.) recombinant protein with GST-tag at N-terminal.

Species Human

Source In Vitro Cell Free System

ProteinLength 519 amino acids

Description This gene is a member of the Bardet-Biedl syndrome (BBS) gene family. Bardet-Biedl syndrome is an autosomal recessive disorder characterized by severe pigmentary retinopathy, obesity, polydactyly, renal malformation and mental retardation. The proteins encoded by BBS gene family members are structurally diverse. The similar phenotypes exhibited by mutations in BBS gene family members are likely due to the proteins shared roles in cilia formation and function. Many BBS proteins localize to the basal bodies, ciliary axonemes, and pericentriolar regions of cells. BBS proteins may also be involved in intracellular trafficking via microtubule-related transport. The protein encoded by this gene has sequence similarity to O-linked N-acetylglucosamine (O-GlcNAc) transferases in plants and archaeobacteria and in human forms a multi-protein "BBSome" complex with six other BBS proteins. Alternative splice variants have been described but their predicted protein products have not been experimentally verified.

Form 50 mM Tris-HCl, 10 mM reduced Glutathione, pH=8.0 in the elution buffer.

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Molecular Mass	82.83 kDa
AA Sequence	<p>MAEERVATRT QFPVSTESQK PRQKKAPEFP ILEKQNWLIH LHYIRKDYE CKAVIKEQLQ ETQGLCEYAI YVQALIFRLE GNIQESLELF QTCAVLSPQS ADNLKQVARS LFLGKHKAA IEVYNEAAKL NQKDWEISHN LGVCYIYLKQ FNKAQDQLHN ALNLRHDLT YIMLGKIHL EGDLDKAEV YKKAVEFSPE NTELLTTLGL LYLQLGIYQK AFEHLGNALT YDPTNYKAIL AAGSMMQTHG DFDVALTKYR VVACAVPESP PLWNNIGMCF FGKKKYVAI SCLKRANYLA PFDWKILYNL GLVHLTMQQY ASAFHFLSAA INFQPKMGEL YMLLAVALTN LEDTENAKRA YAEAVHDKC NPLVNLNYAV LLYNQGEKKN ALVQYQEMER KVSLKDNSS LEFDSEMVEM AQKLGAAALQV GEALVWTKPV KDPKSKHQT STSKPASFQQ PLGSNQALGQ AMSSAAAYRT LPSGAGGTSQ FTKPPSLPLE PEPAVESSPT ETSEQIREK</p>
Storage	Store at -80 centigrade. Aliquot to avoid repeated freezing and thawing.
Storage Buffer	50 mM Tris-HCl, 10 mM reduced Glutathione, pH=8.0 in the elution buffer.
GENE INFORMATION	
Gene Name	BBS4 Bardet-Biedl syndrome 4 [Homo sapiens]
Official Symbol	BBS4
Synonyms	BBS4; Bardet-Biedl syndrome 4; Bardet-Biedl syndrome 4 protein
Gene ID	585
mRNA Refseq	NM_001252678
Protein Refseq	NP_001239607

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MIM 600374

UniProt ID Q96RK4

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