

Recombinant Human BBS7 Protein, MYC/DDK-tagged

Cat. No. BBS7-2377H **Lot. No.** (See product label)

SPECIFICATION

Product Overview	Recombinant human BBS7 protein, fused to MYC/DDK-tagged at C-terminus, was expressed in HEK293
Species	Human
Source	HEK293
Description	This gene encodes one of eight proteins that form the BBSome complex containing BBS1, BBS2, BBS4, BBS5, BBS7, BBS8, BBS9 and BBIP10. The BBSome complex is believed to recruit Rab8(GTP) to the primary cilium and promote ciliogenesis. The BBSome complex assembly is mediated by a complex composed of three chaperonin-like BBS proteins (BBS6, BBS10, and BBS12) and CCT/TRiC family chaperonins. Mutations in this gene are implicated in Bardet-Biedl syndrome, a genetic disorder whose symptoms include obesity, retinal degeneration, polydactyly and nephropathy; however, mutations in this gene and the BBS8 gene are thought to play a minor role and mutations in chaperonin-like BBS genes are found to be a major contributor to disease development in a multiethnic Bardet-Biedl syndrome patient population. Two transcript variants encoding distinct isoforms have been identified for this gene.[provided by RefSeq, Oct 2014].
Form	25 mM Tris.HCl, pH 7.3, 100 mM glycine, 10% glycerol.
Molecular Mass	75.3 kDa
Purity	> 80% as determined by SDS-PAGE and Coomassie blue staining

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Concentration >50 ug/mL as determined by microplate BCA method

GENE INFORMATION

Gene Name BBS7 Bardet-Biedl syndrome 7 [Homo sapiens]

Official Symbol BBS7

Synonyms BBS2L1; BBS2L1

Gene ID 55212

mRNA Refseq NM_018190

Protein Refseq NP_060660

MIM 607590

UniProt ID Q8IWX6

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