

Recombinant Human BBS4 Protein, Myc/DDK-tagged, C13 and N15-labeled

Cat. No. BBS4-3959H **Lot. No.** (See product label)

SPECIFICATION

Product Overview	BBS4 MS Standard C13 and N15-labeled recombinant protein (NP_149017) with a C-terminal MYC/DDK tag, was expressed in HEK293 cells.
Species	Human
Source	HEK293
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 cognitive disability. 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 protein's 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 archaebacteria and in human forms a multi-protein "BBSome" complex with seven other BBS proteins. Alternate splicing results in multiple transcript variants.
Molecular Mass	58.3 kDa
AA Sequence	MAEERVATRTQFPVSTESQKPRQKKAPEFFILEKQNWLIHLHYIRKDYEACKAVIKEQ

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LQETQGLCEYAIYVQALIFRLEGNIQESLELFQTCAVLSPQSADNLKQVARSLFLLGK
 HKAAIEVYNEAAKLNQKDWEISHNLGVCYIYLKQFNKAQDQLHNALNLNRHDLTYIML
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 TNYKAILAAGSMMQTHGDFDVALTKYRVVACAVPESPLWNNIGMCFFGKKKYVAAI
 SCLKRANYLAPFDWKILYNLGLVHMTMQQYASAFHFLSAAINFQPKMGELYMLLAVA
 LTNLEDTENAKRAYAEAVHLDKCNPLVNLNYAVLLYNQGEKKNALVQYQEMEKKVS
 LLKDSSLEFDSEMVEMAQKLGAALQVGEALVWTKPVKDPKSKHQTSTSTSKPASFQ
 QPLGSNQALGQAMSSAAAYRTLPSGAGGTSQFTKPPSLPLEPEPAVESSPTETSEQ
 IREKTRTRPLEQKLISEEDLAANDILDYKDDDDKV

Purity > 80% as determined by SDS-PAGE and Coomassie blue staining

Stability Stable for 3 months from receipt of products under proper storage and handling conditions.

Storage Store at -80 centigrade. Avoid repeated freeze-thaw cycles.

Concentration 50 µg/mL as determined by BCA

Storage Buffer 100 mM glycine, 25 mM Tris-HCl, pH 7.3.

GENE INFORMATION

Gene Name BBS4 Bardet-Biedl syndrome 4 [Homo sapiens (human)]

Official Symbol BBS4

Synonyms BBS4; Bardet-Biedl syndrome 4; Bardet-Biedl syndrome 4 protein;

Gene ID 585

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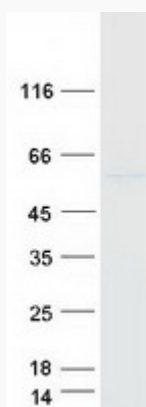
mRNA Refseq NM_033028

Protein Refseq NP_149017

MIM 600374

UniProt ID Q96RK4

SDS-PAGE



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