

## Recombinant Human BBS4 Protein, GST-tagged

Cat. No. BBS4-108H 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** Wheat Germ

**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.

**Molecular Mass** 82.83 kDa

 Tel: 1-631-559-9269 1-516-512-3133

 Email: [info@creative-biomart.com](mailto:info@creative-biomart.com)  Fax: 1-631-938-8127

 45-1 Ramsey Road, Shirley, NY 11967, USA

**AA Sequence**

MAEERVATRTQFPVSTESQKPRQKKAPEFFPILEKQNWLIHLHYIRKDYEACKAVIKEQ  
 LQETQGLCEYAIYVQALIFRLEGNIQESLELFQTCAVLSPQSADNLKQVARSLFLLGK  
 HKAAIEVYNEAAKLNQKDWEISHNLGVCYIYLKQFNKAQDQLHNALNLRHDLTYIML  
 GKIHLLEGDLDKAIEVYKKAVEFSPENTELLTTLGLLYLQLGIYQKAFEHLGNALTYDP  
 TNYKAILAAGSMMQTHGDFDVALTKYRVVACAVPESPLWNNIGMCFFGKKKYVAAI  
 SCLKRANYLAPFDWKILYNLGLVHLTMQQYASAFHFLSAAINFQPKMGELYMLLAVA  
 LTNLEDTENAKRAYAEAVHLDKCNPLVNLNYAVLLYNQGEKKNALVQYQEMERKVS  
 LLKDSSLEFDSEMVEMAQKLGAALQVGEALVWTKPKVDPKSKHQTSTSTSKPASFQ  
 QPLGSNQALGQAMSSAAAYRTLPSGAGGTSQFTKPPSLPEPEPAVESSPTETSEQ  
 IREK

**Notes**

Best use within three months from the date of receipt of this protein.

**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](#)

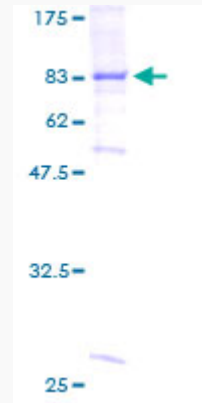
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